Diagnostic Imaging
Pediatrics

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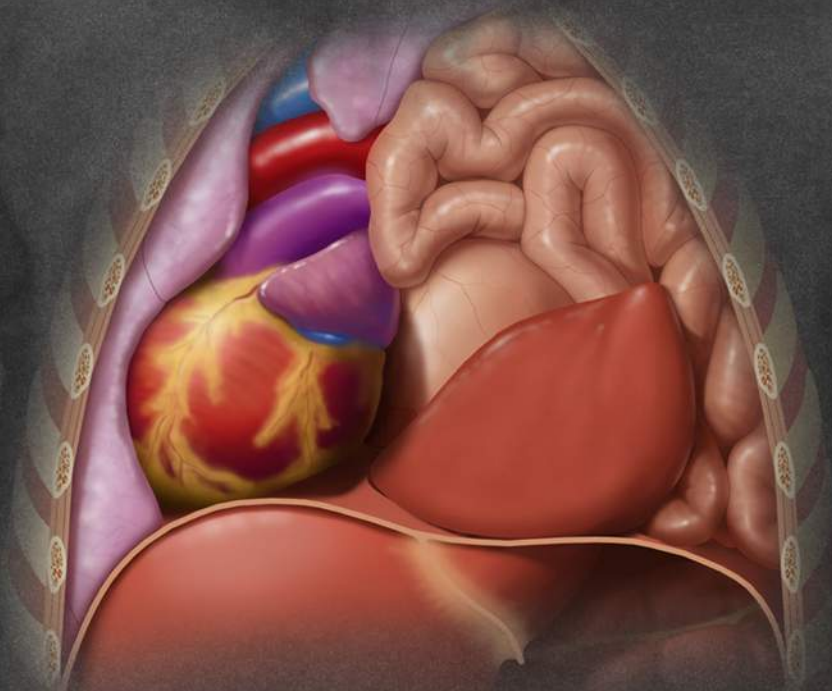

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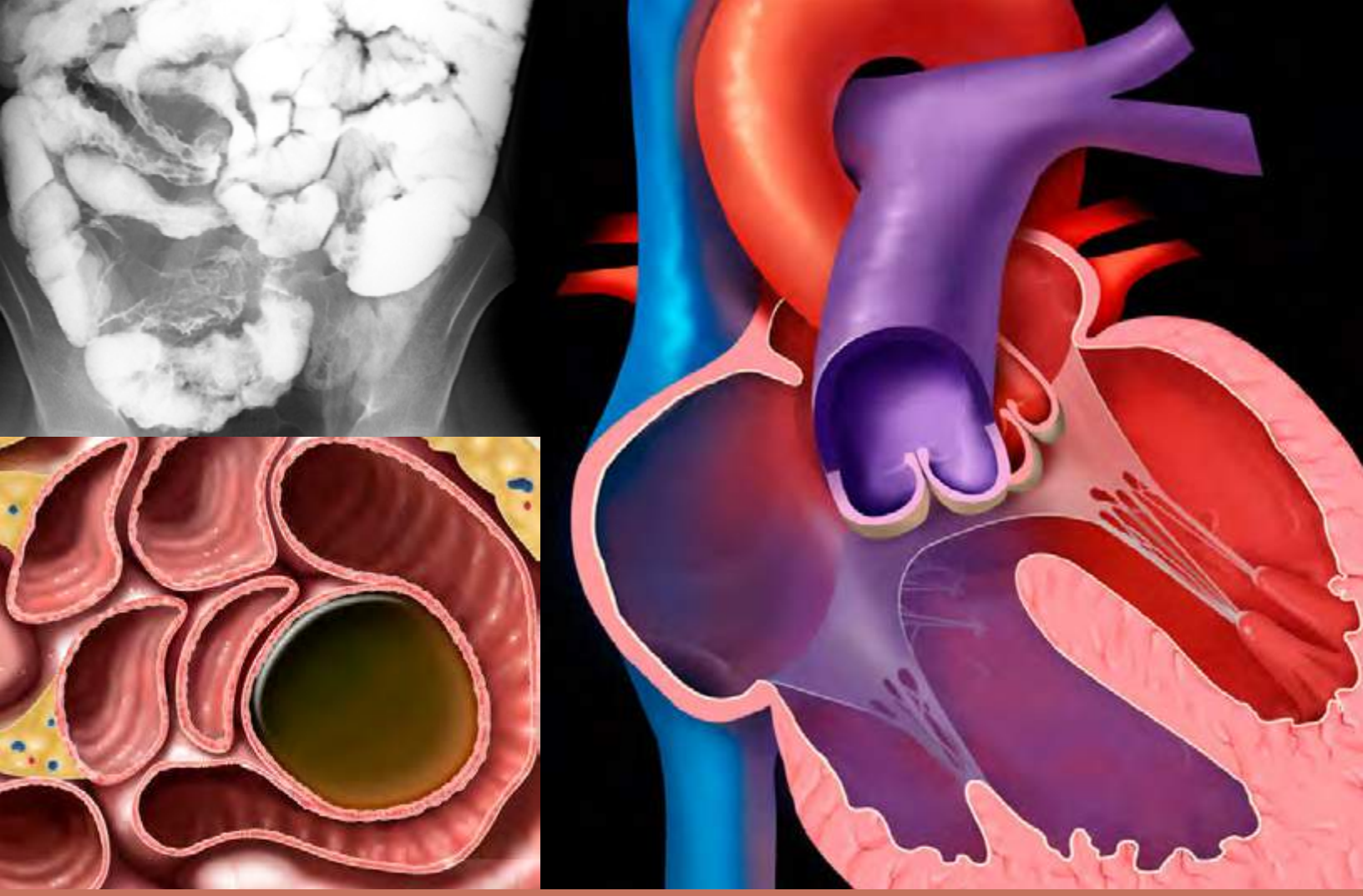

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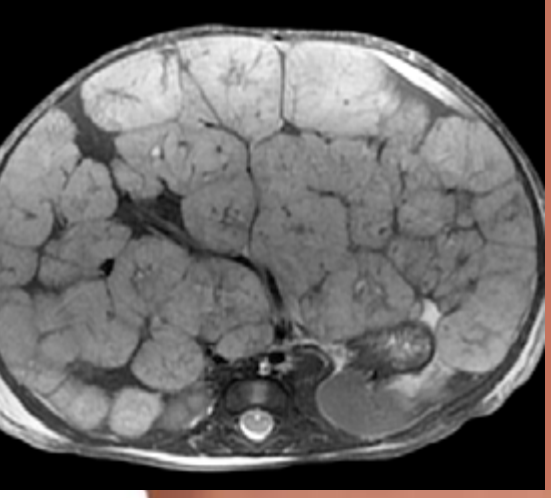

Diagnostic Imaging

Pediatrics

THIRD EDITION

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Dedications

To Nan—the love of my life—I am bound to you through and through. Thank you so much for your support. This would not have been possible without you.

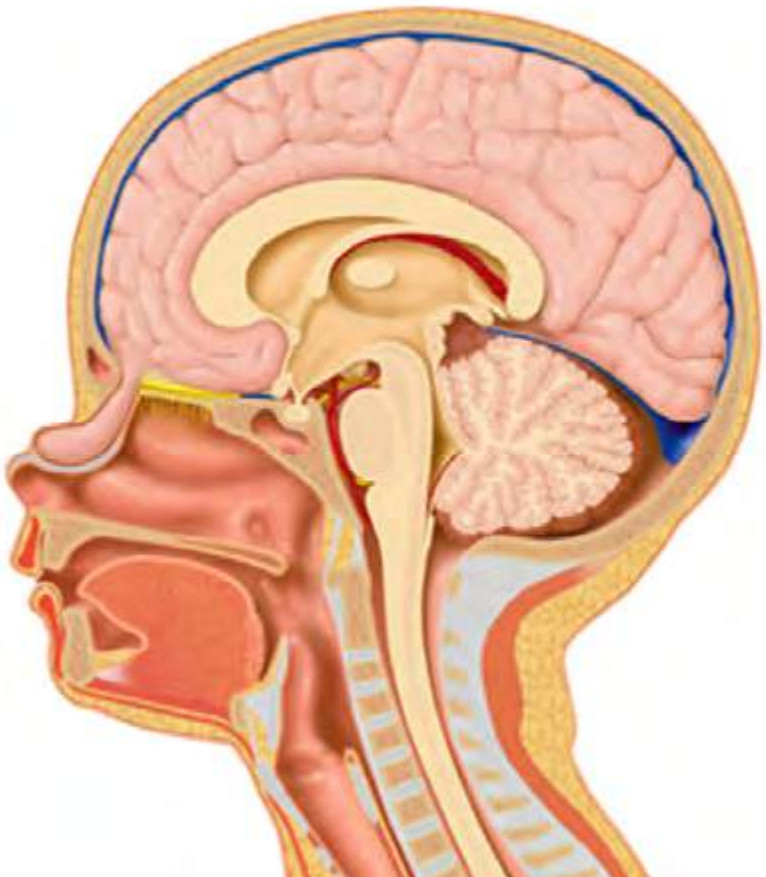
To Cora and Nate—I am so grateful to be your father. May I have taught you at least a fraction of the things you have taught me.

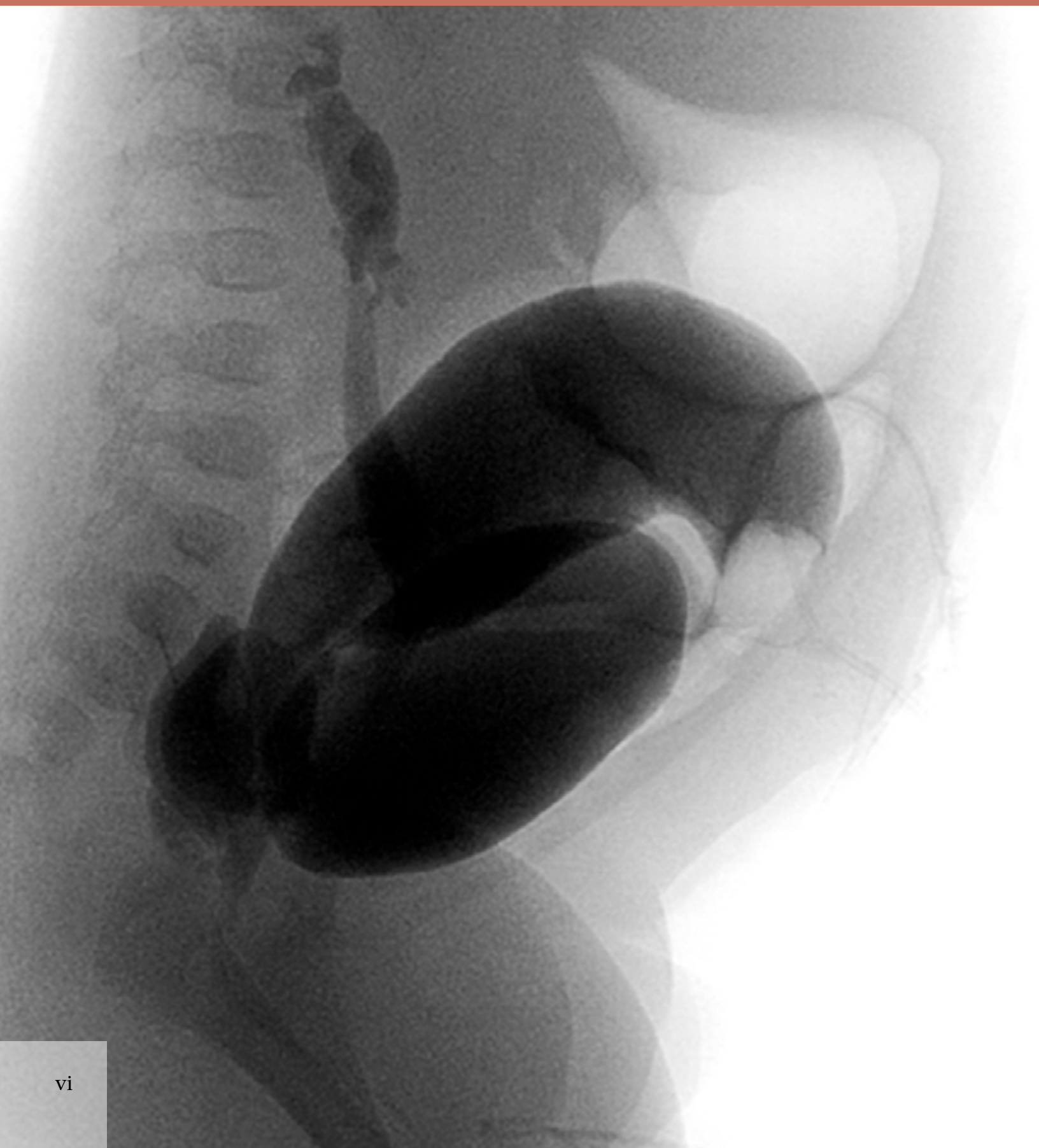
To my parents—thank you for your lifelong sacrifices and encouragement.

To my teachers—your enthusiasm and engagement with your patients and trainees have touched so many. May this text be another conduit of your passion and mercy.

To my patients and their families—let the physician not forget that none of this work matters apart from you. Your fight inspires daily.

ACM





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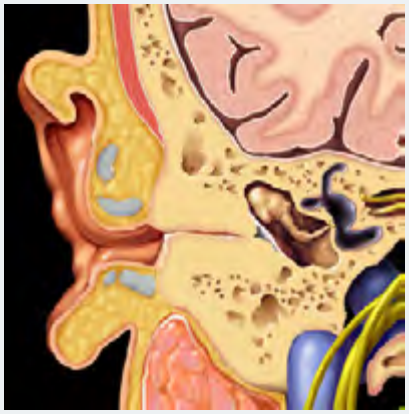

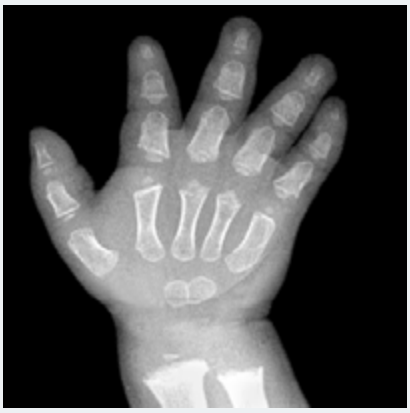

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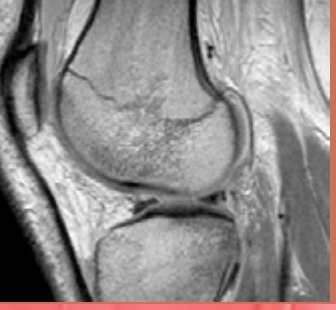

Preface

Welcome to the third edition of *Diagnostic Imaging: Pediatrics*. As in all of medicine, there has been substantial evolution in the understanding of pediatric disorders in the last five years, particularly with regards to molecular genetics, therapeutic options, clinical outcomes, & imaging manifestations of various diseases. In some cases, this has resulted in revised classifications & treatment pathways. Technical advances & refined research have also led to modified diagnostic algorithms. Our pediatrics team has been excited to update this text accordingly, meticulously revising the previous edition and adding over 70 new topics and more than 2,000 new print & online images.

It has been my goal, however, not just to update the text with the latest research available, but also to maximize the teaching impact of this work. I have desired to create a text from which anyone can learn. That is, one that is simultaneously rich with relevant details and accessible to all levels of training, from the medical student to the experienced attending physician. We have specifically sought to optimize the Key Facts sections for the quick review of information critical to each diagnosis. Additionally, we have enhanced the "Imaging Recommendations" and "Diagnostic Checklist" segments of most chapters to enrich the "viewbox teaching" for each topic (with the author imparting to you a few supplementary or summary pearls of wisdom by which to make the correct diagnosis).

I offer many thanks to the authors of this text—I am honored to have led such a distinguished group of colleagues on this journey. Many of the contributors are well-established experts in their pediatric subspecialties. Others have an intense interest and impressive awareness of their fields given an earlier career stage. But each author was hand-selected for his or her distinct knowledge base, clinical and academic experience, and passion for teaching.

I would also like to thank the fantastic staff at Elsevier (especially Nina!) for their guidance, support, and seemingly tireless efforts that have helped bring to life my vision for this book.

I must also give special thanks to my family, whose patience, encouragement, and love enabled the creation of this text.

And thank you, reader, for choosing this product—may it daily enhance your care of children no matter the location or type of your practice.

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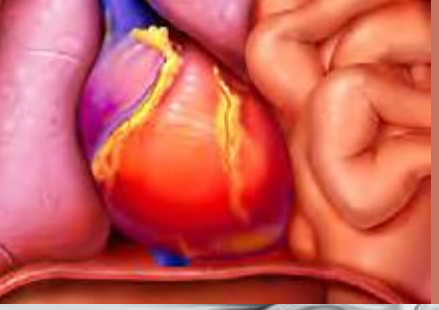

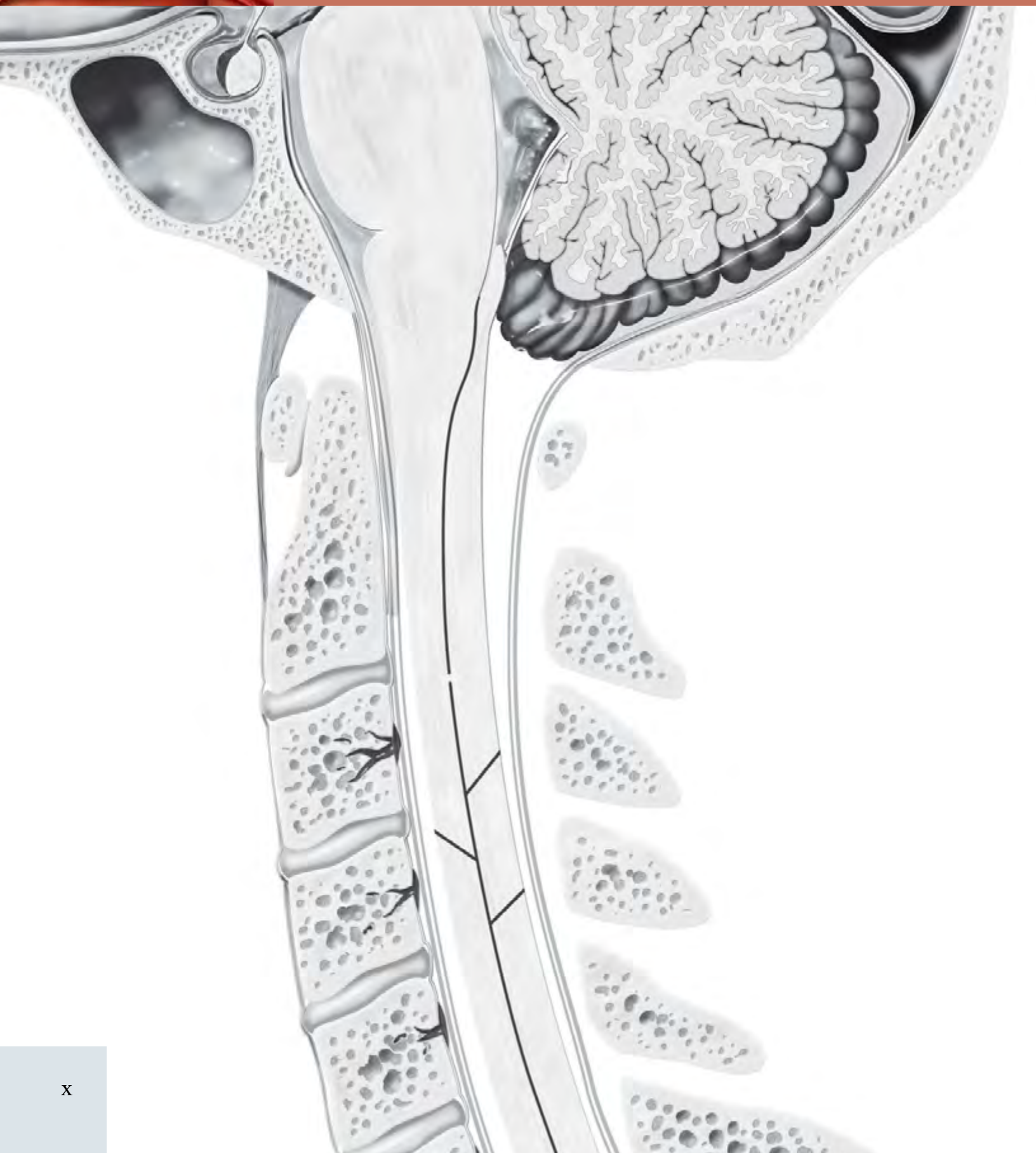

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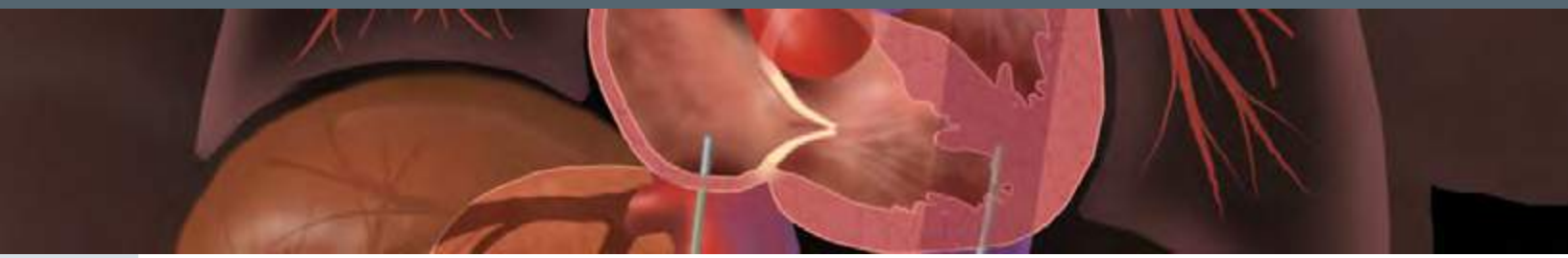

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SYNDROMES WITH CRANIOFACIAL INVOLVEMENT

NECK MASSES

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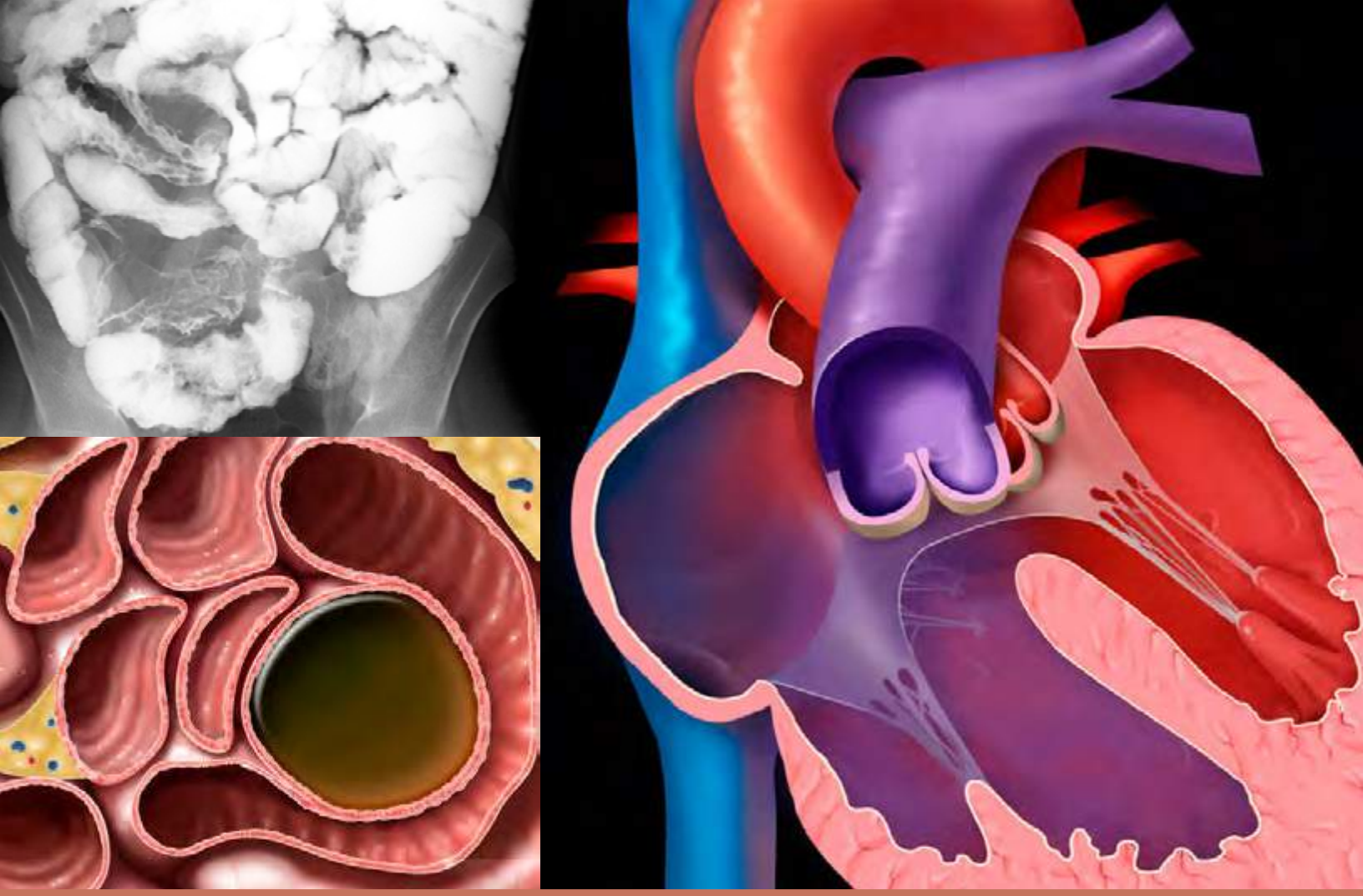

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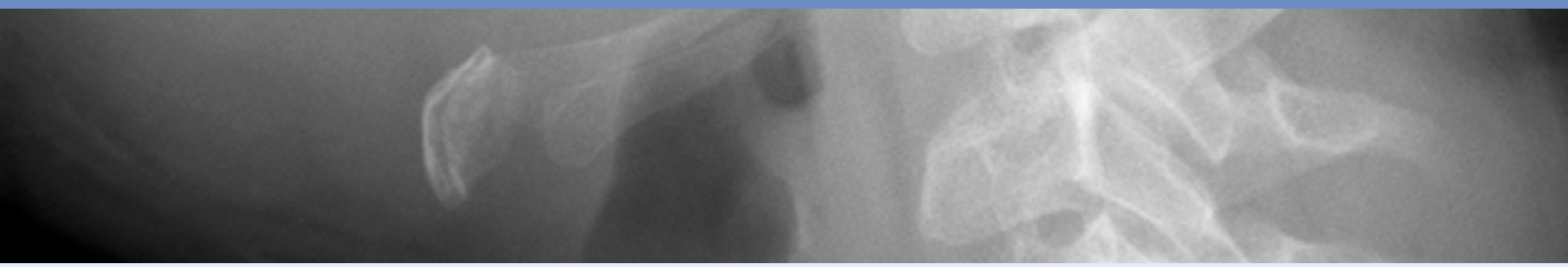
SECTION 1

Airway



Introduction

Anatomically & functionally, the pediatric airway can be divided into upper & lower segments at the glottis (larynx) or large & small airways at the transition from the cartilage-containing bronchi proximally to the distal airways that lack supporting cartilage. The superimposed disease processes may be extrinsic or intrinsic, & they may manifest as acute or chronic airway compromise at a variety of ages. The general categories of diseases listed below are not always distinct, with some processes affecting multiple levels or presenting later in childhood despite an underlying congenital issue.

Anatomic Considerations

The oral cavity is the portion of the airway superior to the tongue & anterior to the soft palate. The nasopharynx is the portion of the airway superior to the soft palate & anterior to the adenoids. The oropharynx extends between the soft palate & the tip of the epiglottis. The hypopharynx extends inferior to this level & includes the remainder of the pharynx above the glottis & esophagus.

Retropharyngeal soft tissues: The retropharyngeal soft tissues should not exceed the thickness of 1 vertebral body width when the soft tissues are measured somewhere between the level of the adenoids superiorly & epiglottis inferiorly. Below the epiglottis, the esophagus is also present, & the soft tissues are normally thicker. "Pseudothickening" of the retropharyngeal soft tissues can occur in young children when there is poor inspiration, or if the neck is not fully extended.

Normal upper airway motion: On cine images, the upper airway of a normal sleeping child is relatively stationary. The walls of the upper airway should not move by more than several mm. The normal airway never demonstrates intermittent complete collapse.

Epiglottis: The normal epiglottis has very thin borders. Thickening results in a thumbprint appearance. The "omega" epiglottis is a term for a normal variant that occurs at imaging when the epiglottis is viewed obliquely & the left & right sides of the cylindrical epiglottis do not overlap perfectly.

Aryepiglottic folds: These are mucosal folds that extend from the epiglottis superiorly to the arytenoid cartilages posteroinferiorly. On the lateral view, they should appear thin & flat or convex inferiorly. With inflammation of the epiglottis, they become markedly thickened & convex superiorly.

Subglottic trachea: On the frontal view, the subglottic trachea should have symmetric lateral convexities ("shoulders"). With abnormal inflammation in this area, the convexities become concave (i.e., the shoulders are lost). This loss of the shoulders has been likened to a church steeple.

On radiography, the trachea should be consistent in diameter for its entire length & well visualized on frontal & lateral views. The normal left aortic arch should gently push the trachea toward the right & mildly indent the trachea on the left.

On axial cross-sectional imaging, the intrathoracic airway should be round or oval in configuration (& slightly greater in AP diameter). The posterior aspect of the trachea is noncartilaginous & may have a linear or flat appearance, especially in expiration. A very small & round trachea is suggestive of complete tracheal rings. A trachea that is flattened (small anterior to posterior diameter) is indicative of extrinsic compression or tracheomalacia.

Congenital Airway Obstructions

Within a short timeframe, the newborn makes the conversion from complete placental support as a fetus to extracting sustenance from the surrounding environment, including oxygen in the air. A normal caliber & reliably patent conduit (airway) from the nostrils to the alveoli is required for this transition. The lack of such a conduit will prevent a successful changeover & require emergent intervention in the delivery suite. Critical anomalies that compromise this conduit are often detected prenatally by secondary findings of a large extrinsic cervical mass (such as a teratoma) or abnormally dilated lungs with diaphragm eversion (as seen in CHAOS). In these circumstances, careful planning for airway management in the delivery suite is required, often utilizing the ex utero intrapartum therapy (EXIT) procedure that allows the airway to be secured while the newborn is maintained on placental circulation.

Most congenital airway anomalies are not immediately fatal & only manifest at times of feeding or stress. Lesions involving the nasal airway commonly present during feedings in the newborn period as young infants are obligate nose breathers. Typical such anomalies include choanal atresia, pyriform aperture stenosis, & bilateral nasolacrimal duct mucoceles.

Infectious Causes of Airway Compromise

This differential diagnosis can be narrowed by age of presentation & includes croup (mean age: 1 year), epiglottitis (postvaccine era mean age: 14.6 years), exudative tracheitis (mean age: 6-10 years), & retropharyngeal abscess (mean age: 6-12 months).

Noninfectious Intrinsic or Intraluminal Obstructions

A foreign body should be considered in any child who has the new onset of airway symptoms, particularly after an episode of choking. Secondary manifestations of air-trapping or atelectasis may be the only radiographic clue to an airway blockage if the foreign body is not radiopaque compared to the surrounding soft tissues. While expiratory or decubitus views may be helpful, the clinician should have a low threshold for CT or bronchoscopy in the correct setting.

Infantile hemangiomas of the airway are most commonly subglottic & present with stridor & asymmetric airway narrowing. CECT (which is rarely performed) will show eccentric, lobular, vigorously enhancing tissue partially effacing the airway.

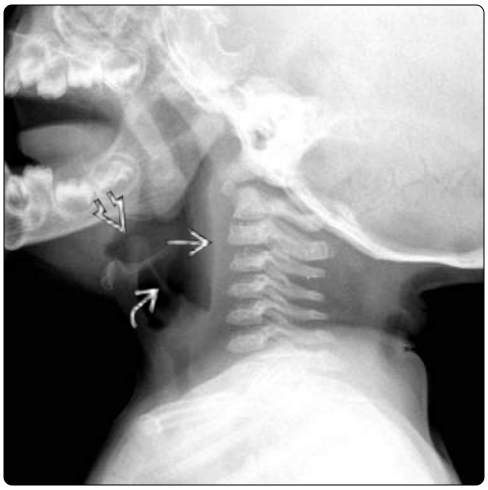
Tracheobronchomalacia (or abnormal collapsibility of the airway) is common & may be 1° or 2° (i.e., associated with extrinsic anomalies). Rings of complete cartilage that result in a round small caliber trachea are often associated with abnormal branching patterns & a pulmonary sling.

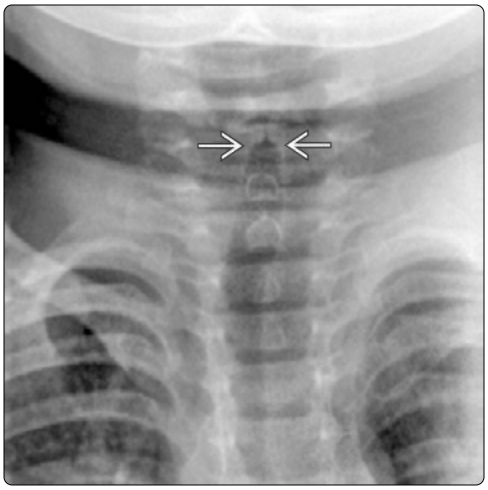
Extrinsic Compression of Lower Airway

This differential diagnosis includes vascular rings, midline descending aorta, thoracic deformity, & mediastinal masses.

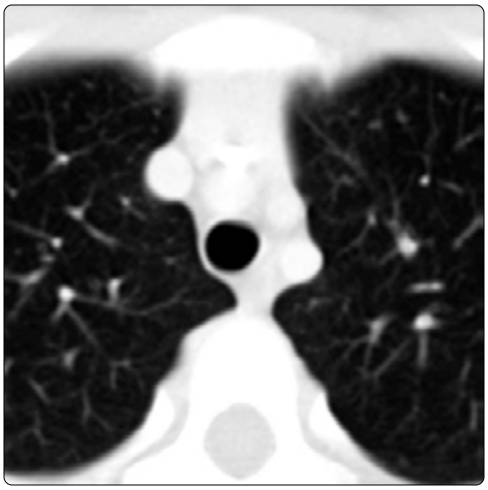
Obstructive Sleep Apnea (OSA)

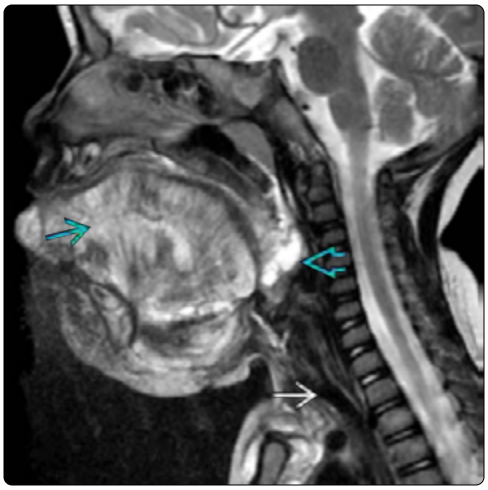
Most children with OSA are otherwise normal & have enlarged adenoid & palatine tonsils. Subgroups of children with more complex anatomic & dynamic (i.e., increased airway collapsibility) issues may benefit from MR sleep study evaluation, as may children with recurrent OSA after surgery.



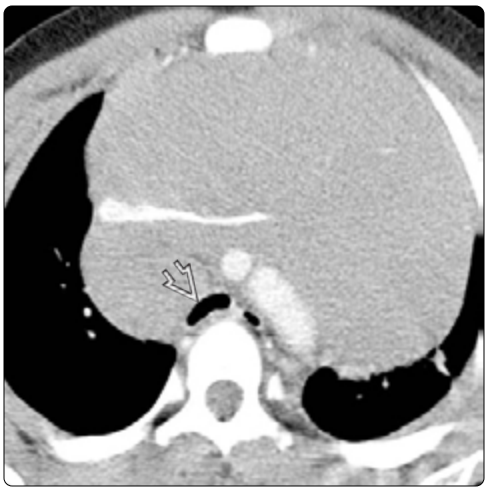


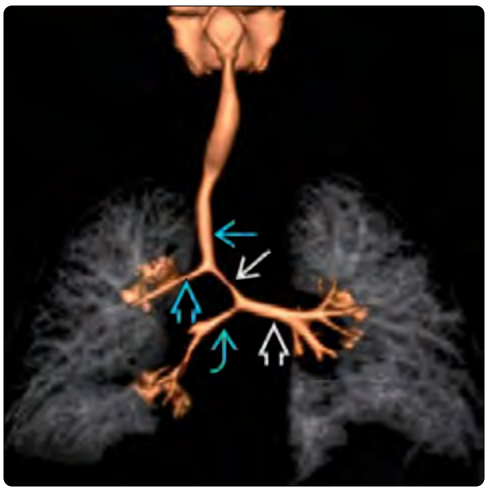
(Left) *Lateral radiograph of a normal airway shows a "thin" & well-defined epiglottis. Note the normal retropharyngeal soft tissue width & thin aryepiglottic folds.* **(Right)** *AP radiograph of a normal airway shows normal "shoulders" or lateral convexities in the subglottic region.*





(Left) *Axial CECT of a normal trachea shows a normal round morphology of the tracheal lumen. The posterior wall may flatten slightly with expiration.* **(Right)** *Sagittal T2 MR in a 17-month-old patient with an extensive facial lymphatic malformation shows marked infiltration of the tongue, floor of mouth tissues, & soft palate by the lesion, resulting in complete effacement of the oral cavity, oropharynx, & upper hypopharynx. A tracheostomy is partially visualized.*





(Left) *Axial CECT in a child with lymphoma shows a large mediastinal mass with a flattened appearance of the trachea (typical of extrinsic compression).* **(Right)** *Anterior view of a 3D NECT of the airways shows a round, narrow caliber distal trachea, typical of complete cartilage rings. There is an isolated right upper lobe bronchus arising from the trachea and leaving a narrowed intermediate left bronchus, which then gives rise to the left main bronchus & a right bridging bronchus.*

Expiratory Buckling of Trachea

KEY FACTS

TERMINOLOGY

- Intermittent normal change in transverse & craniocaudal configuration of trachea in infants during expiration

IMAGING

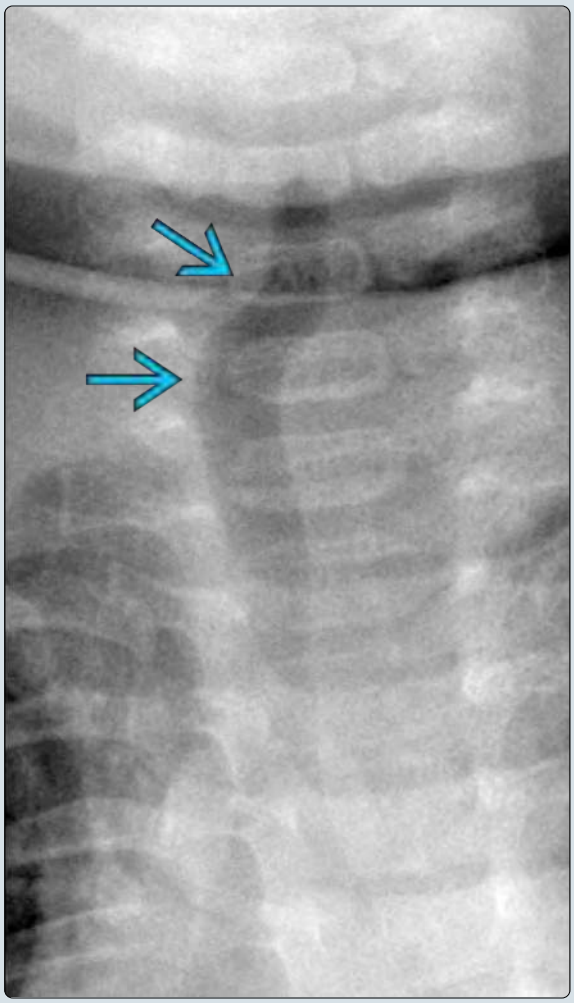
- On AP view, trachea is normally straight vertically in older children & adults throughout respiratory cycle
- In infants, trachea is normally straight during inspiration but changes with expiration
  - Focal shortening, crinkle, bend, or curve at/above thoracic inlet without caliber change
  - Directed toward right in patients with left aortic arch
    - Buckling toward left suggests right aortic arch
  - Trachea becomes straight again with inspiration
- No need to repeat radiograph

TOP DIFFERENTIAL DIAGNOSES

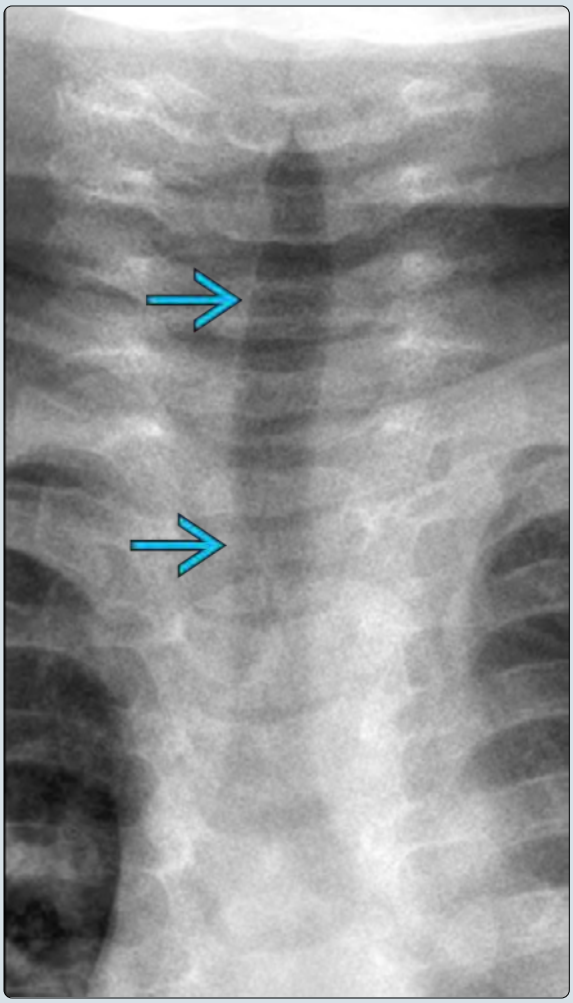
- Croup: Symmetric narrowing of subglottic trachea in young child with characteristic "barky" cough
- Infantile hemangioma: Persistent asymmetric tracheal narrowing by intraluminal benign vascular neoplasm
  - Often associated with cutaneous infantile hemangioma in "beard" distribution
- Tracheomalacia: Abnormal dynamic tracheal collapse in anterior to posterior dimension (not transverse)
  - Static lateral view may show caliber narrowing; dynamic change confirms tracheomalacia rather than fixed stenosis or compression
- Compression by extrinsic mass or aberrant vessel
  - Persistent focal airway narrowing with deviation away from mass/vessel; mass may enlarge mediastinum

CLINICAL ISSUES

- Incidental finding on chest or airway radiographs if morphology & patient age correct
- Does not cause symptoms that lead to imaging



AP radiograph of the airway in an 8-month-old patient shows the typical configuration of expiratory tracheal buckling; the trachea at & just above the thoracic inlet demonstrates a focal bend toward the right ⇨ but does not demonstrate narrowing.



AP radiograph in an 8-month-old patient during inspiration shows a trachea that is relatively straight vertically ⇨.

Pseudothickening of Retropharyngeal Tissues

KEY FACTS

TERMINOLOGY

- Transient thickening of normal retropharyngeal soft tissues of infant on lateral airway radiograph
  - "Swelling" with expiration or poor extension
  - Resolution with inspiration & adequate extension
- Contributing factors to this appearance include
  - Relatively short necks of infants & young children which lead to poor positioning for airway radiographs
  - Relatively long expiratory component of crying also challenges acquisition during maximal inspiration

IMAGING

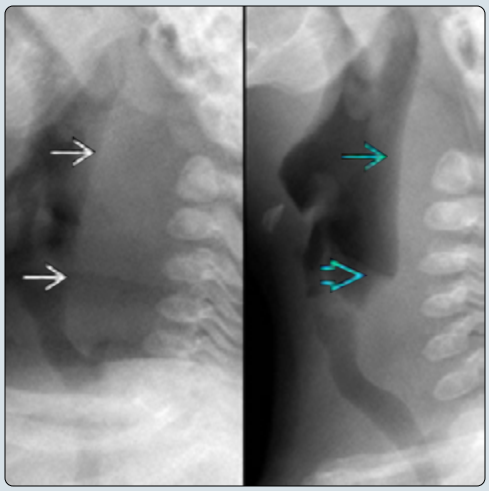
- Generalized thickening/bulging of prevertebral soft tissues
  - ± retention of normal "step-off" at junction of hypopharynx & cervical esophagus
    - Persistent "step-off" favors pseudothickening over true retropharyngeal pathology
- Resolves on repeat lateral radiograph with improved inspiratory timing of exposure & ↑ neck extension
- Observation of dynamic airway changes under fluoroscopy can confirm intermittent thickening & resolution
  - Use "last image capture/image hold" for documentation

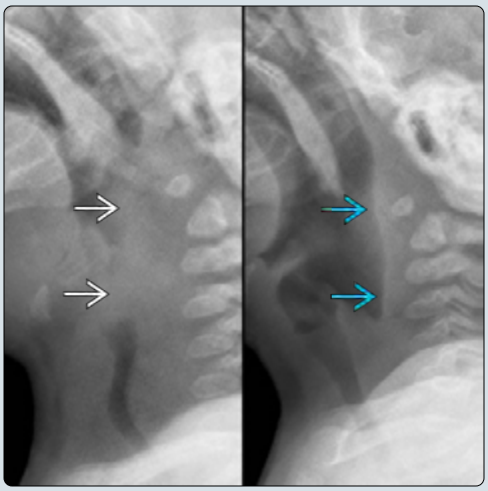
TOP DIFFERENTIAL DIAGNOSES

- Retropharyngeal cellulitis/abscess
  - Convex generalized bulging of prevertebral soft tissues persists despite inspiration & neck extension
  - Often lose normal "step-off" at hypopharyngeal-esophageal junction
- Cervical spine pathology
  - Trauma, inflammation/infection, or neoplasm → prevertebral soft tissue swelling
  - ± radiographically visible bony abnormality

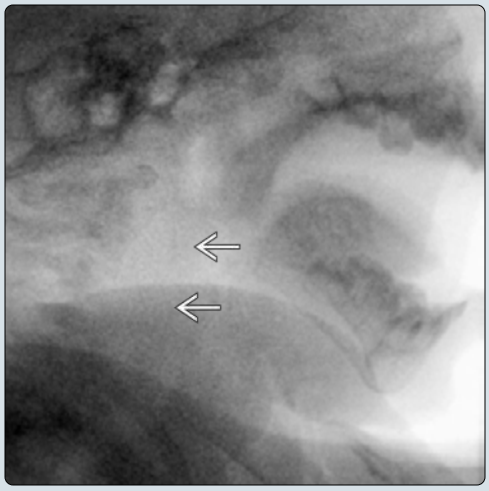
CLINICAL ISSUES

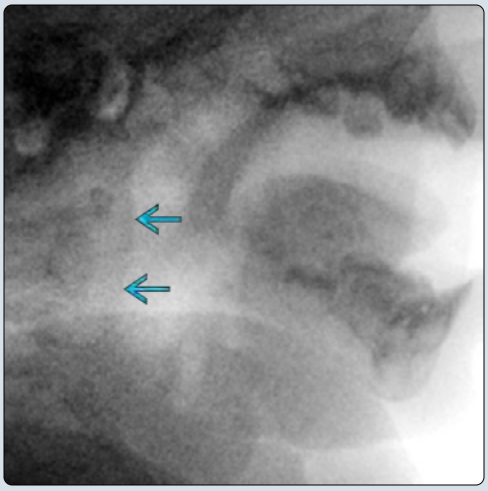
- Uncommon in children > 2 years of age
- Unlike true pathology, pseudothickening does not cause characteristic signs/symptoms





(Left) *Lateral airway radiographs in an infant show pseudothickening of the prevertebral soft tissues initially ➡ with resolution upon improved neck extension ⇨. Note the normal "step-off" at the hypopharyngeal-esophageal junction ⇨ on the 2nd image.* **(Right)** *Lateral airway radiographs in a 4 month old show dynamic protrusion ➡ & collapse ⇨ of the retropharyngeal tissues between expiration (left) & inspiration (right). The 2nd image confirms a normal thickness & morphology of the prevertebral tissues.*





(Left) *Lateral fluoroscopic "image hold" in an 11 month old with stridor & suggested retropharyngeal thickening on preceding radiographs (not shown) demonstrates bulging of the prevertebral soft tissues ➡ during expiration.* **(Right)** *Lateral fluoroscopic "image hold" in the same patient during inspiration shows normal collapse of the prevertebral soft tissues ⇨, confirming pseudothickening on the initial image.*

Congenital Nasal Pyriform Aperture Stenosis

KEY FACTS

TERMINOLOGY

- Congenital narrowing of anterior bony nasal passageway [pyriform aperture (PA)]

IMAGING

- Best tool: Bone CT in axial & coronal planes
  - Medial deviation of anterior maxillae with thickening & convergence of nasal processes
    - PA axial width < 11 mm in term infant is diagnostic
  - Triangle-shaped hard palate on axial images
  - Abnormal maxillary dentition: Solitary median maxillary central incisor (SMMCI) in 75%

TOP DIFFERENTIAL DIAGNOSES

- Nasolacrimal duct mucoceles
- Choanal stenosis/atresia

PATHOLOGY

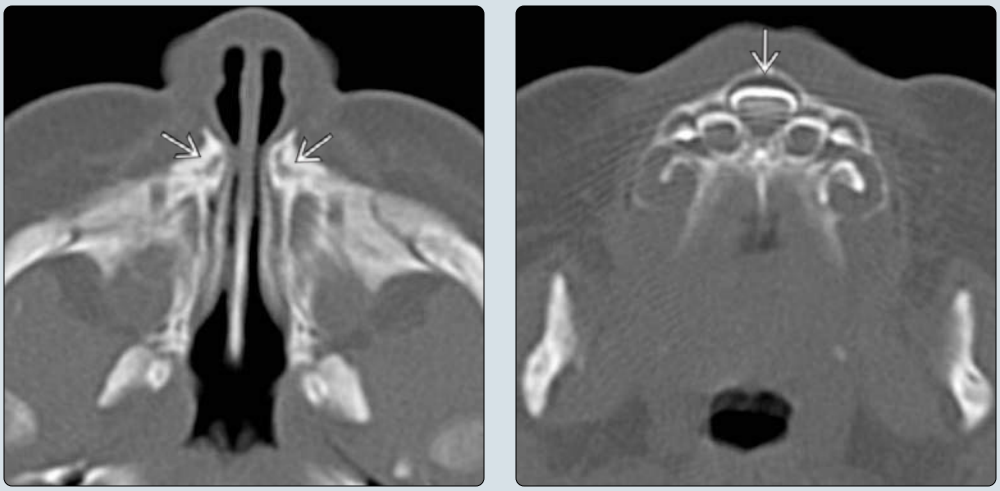
- Congenital nasal pyriform aperture stenosis (CNPAS) without SMMCI: Almost always isolated
- SMMCI in 75% of CNPAS
  - Associated with holoprosencephaly, pituitary-adrenal axis dysfunction, microcephaly, many other findings

CLINICAL ISSUES

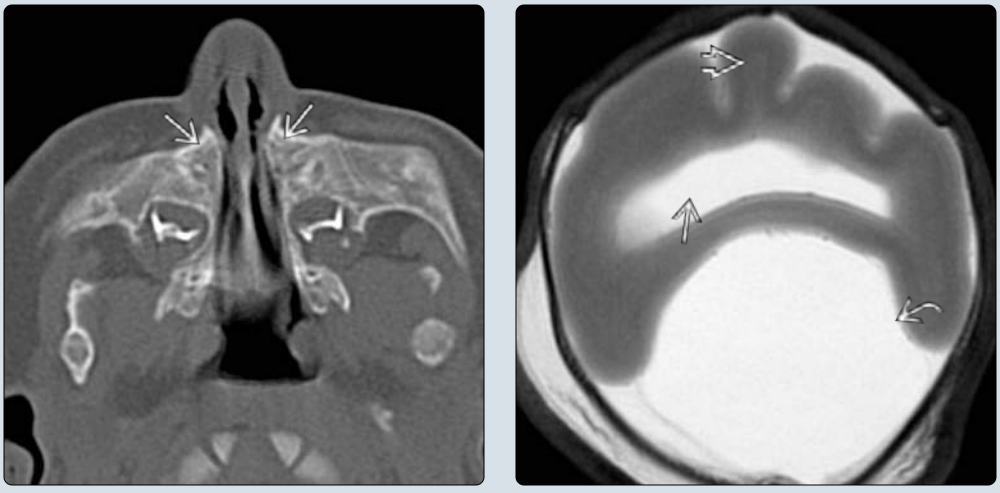
- Respiratory distress in newborn/infant
  - Symptoms more pronounced with feeding
  - Breathing problems may be triggered by URI
  - Narrow nasal inlet on clinical exam
- Can mimic choanal atresia/stenosis clinically
  - CNPAS 1/5 to 1/3 as common
- Nasal cavity eventually grows; mild cases may improve
- Surgery for persistent respiratory difficulties & poor weight gain; PA width < 5.7 mm may predict surgical need

DIAGNOSTIC CHECKLIST

- Bone CT to confirm/characterize bony narrowing & identify dental/palatal abnormalities
- Brain MR if SMMCI to exclude midline brain anomalies



(Left) *Axial bone CT in a newborn shows the typical features of congenital nasal pyriform aperture stenosis. There is overgrowth of the anterior maxillae ➡ with marked narrowing of the pyriform aperture/nasal inlet.* **(Right)** *Axial bone CT at the level of the anterior maxilla in the same patient shows a solitary median maxillary central incisor (or "megaincisor") ➡. This is a common associated finding in children with congenital pyriform aperture stenosis, with or without midline intracranial abnormalities.*



(Left) *Axial bone CT in a newborn with respiratory distress shows thickening of the anterior & medial aspects of the maxillae ➡ causing pyriform aperture stenosis. This child did not have an associated solitary median maxillary central incisor or intracranial anomaly.* **(Right)** *Axial T2 brain MR necropsy image in a child with a solitary median maxillary incisor (not shown) reveals a monoventricle ➡, absence of frontal lobe cleavage ➡, & a large dorsal midline cyst ➡, findings that are all typical of alobar holoprosencephaly.*

TERMINOLOGY

Abbreviations

- Congenital nasal pyriform aperture stenosis (CNPAS)

Definitions

- Pyriform aperture (PA): Single triangular bony opening of anterior skull to nasal passages
  - Anterior nasal passages separated by septal cartilage
- CNPAS: Narrowing of single bony opening by characteristic bilateral maxillary bony anomalies

IMAGING

General Features

- Best diagnostic clue
  - Medialization & thickening of anterior maxillae with narrowing of anterior nasal airway
- Size
  - PA size in CNPAS
    - Axial width < 11 mm in term infant diagnostic (normal: 13.4-15.6 mm)
    - Area: 0.2-0.4 cm² (normal: 0.7-1.1 cm²)

CT Findings

- Bone CT
  - Narrowed bony nasal inlet
    - Medial deviation of lateral walls of PA (anterior maxillae) ± thickened & converging bony nasal (frontal) processes
  - Triangle-shaped hard palate on axial images
    - Bony ridge along oral surface of hard palate on coronal images
  - ± abnormal maxillary dentition
    - Fused or malaligned central & lateral incisors
    - Solitary median maxillary central incisor (SMMCI) in 75%
  - Posterior choanae normal in caliber

Imaging Recommendations

- Best imaging tool
  - Bone CT to confirm/characterize bony narrowing & identify dental/palatal abnormalities
  - Brain MR recommended in cases of SMMCI to exclude midline brain anomalies
- Protocol advice
  - Bone CT with axial & coronal reformatted images
    - Cover from tips of incisors through nasal cavity
    - Contrast unnecessary

DIFFERENTIAL DIAGNOSIS

Nasolacrimal Duct Mucoceles

- Obstruction of distal nasolacrimal ducts by bulging cysts at inferior meatus that narrow anterior nasal cavity
- Bony aperture normal

Choanal Stenosis/Atresia

- Narrow or occluded posterior nasal passage: Membranous, osseous, or mixed
- Anterior nasal passage normal in caliber

PATHOLOGY

General Features

- Etiology
  - 2 theories of pathogenesis
    - Deficiency of primary palate derived from midline mesodermal tissue
      - Embryologically, medial maxillary swelling forms structures of primary palate, including 4 incisors
      - Primary palatal deficiency → narrowed anterior nasal cavity, abnormal incisors, & triangular palate
      - Mesoderm thought to have inductive effect on forebrain, hence association of SMMCI syndrome with holoprosencephaly
    - Overgrowth or dysplasia of nasal processes of maxilla
- Associated abnormalities
  - CNPAS without SMMCI almost always isolated
  - SMMCI syndrome (75% of CNPAS cases) with variable presence of
    - Semilobar or alobar holoprosencephaly
    - Endocrine dysfunction of pituitary-adrenal axis → short stature, ambiguous genitalia
    - Microcephaly & intellectual disability

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Respiratory distress, especially with feeding (as young infants are "obligate nasal breathers")
    - May be triggered by upper respiratory infection that further compromises narrowed airway
    - Can mimic choanal atresia/stenosis
  - Narrow nasal inlet on clinical exam

Demographics

- Age
  - Newborns or infants in 1st few months of life
- Epidemiology
  - Congenital airway obstruction affects 1 in 5,000 infants
    - Majority due to choanal atresia
    - CNPAS 1/5 to 1/3 as common as choanal atresia

Treatment

- Mild cases may be treated conservatively with special feeding techniques as nasal cavity eventually grows
- Surgical intervention in patients with persistent respiratory difficulty & poor weight gain
  - PA width < 5.7 mm in neonate may correlate with need for surgical intervention
  - Resection of anteromedial maxillae ± anterior aspect of inferior turbinates + reconstruction anterior nasal orifice

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Nasolacrimal Duct Mucocele

KEY FACTS

TERMINOLOGY

- Synonym: Congenital dacryocystocele

IMAGING

- Well-defined, cystic medial canthal mass in continuity with enlarged nasolacrimal duct (NLD) in newborn
  - Unilateral or bilateral
- Absent or minimal wall enhancement (unless infected)
- Coronal/sagittal reformatted images show continuity of proximal cyst at lacrimal sac with distal inferior meatus cyst through dilated NLD

TOP DIFFERENTIAL DIAGNOSES

- Orbital dermoid & epidermoid
  - Lateral > medial canthus
- Acquired dacryocystocele
  - Typically posttraumatic, usually adults

PATHOLOGY

- Tears & mucus accumulate in NLD with imperforate Hasner membrane (i.e., distal duct obstruction)
- Most common abnormality of infant lacrimal apparatus

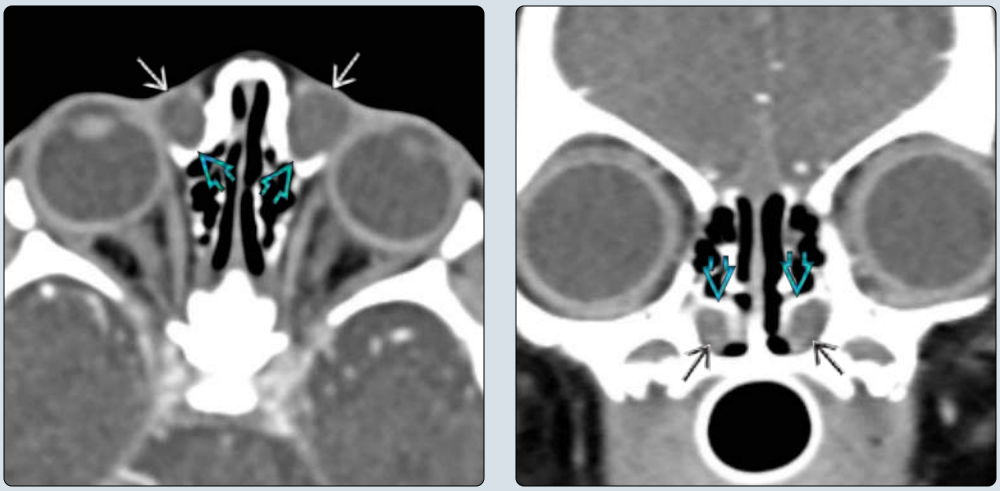
CLINICAL ISSUES

- Proximal cyst: Small, round, bluish, medial canthal mass identified at birth or shortly thereafter; ± cellulitis
- Distal cyst: Nasal airway obstruction with respiratory distress if bilateral (especially during feeding)

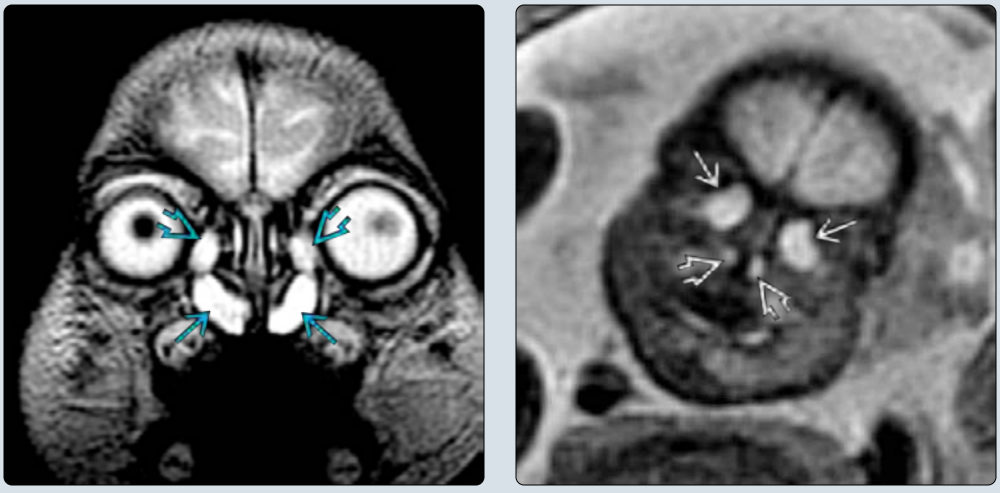
DIAGNOSTIC CHECKLIST

- Cross-sectional imaging evaluates extent of lesion along lacrimal apparatus & excludes other sinonasal causes of respiratory distress in newborn
- Comment on full extent of lesion from medial canthus to inferior meatus
- Exclude contralateral lesion

(Left) *Axial CECT in a 4 day old with bluish bilateral medial orbital swelling & left purulent drainage shows bilateral lacrimal sac enlargement ➡. Note also the bilateral lacrimal sac fossae splaying ⇨. (Right) Coronal CECT in the same patient demonstrates the typical locations of the distal intranasal components of nasolacrimal duct mucoceles ⇨ inferior to the inferior turbinates ⇨.*



(Left) *Coronal T2 MR in an infant shows hyperintense nasolacrimal duct mucoceles extending from the dilated lacrimal sacs proximally ⇨ to protrude inferomedially from the inferior nasolacrimal ducts ⇨. (Right) Coronal SSFSE T2 MR in a 2nd-trimester fetus demonstrates bilateral lacrimal sac enlargement ➡ with distal extension into each inferior meatus ➡, a typical appearance of nasolacrimal duct mucoceles.*



TERMINOLOGY

Synonyms

- Congenital dacryocystocele

Definitions

- Nasolacrimal duct (NLD) mucocele: Cystic dilation of nasolacrimal apparatus secondary to obstruction of NLD
- Canthus: Corner of eye where eyelids meet

IMAGING

General Features

- Best diagnostic clue
  - Well-defined, cystic, medial canthal mass in continuity with enlarged NLD in newborn
- Location
  - From lacrimal sac at medial canthus to distal aspect of NLD at inferior meatus
  - Unilateral or bilateral

CT Findings

- Hypodense, thin-walled cyst at medial canthus ± bulging cystic component at inferior meatus
  - Cysts communicate through enlarged NLD
- Minimal wall enhancement normally; thick rim enhancement ± fluid/debris level if infected

MR Findings

- T1-hypointense/T2-hyperintense, well-circumscribed mass(es)
- Signal intensity varies with protein content &/or infection
- Minimal wall enhancement normally
- If inflamed/infected → thick rim of enhancement with surrounding poorly defined soft tissue stranding

Imaging Recommendations

- Best imaging tool
  - Thin-section bone CT
  - ± contrast (for better soft tissue characterization)

DIFFERENTIAL DIAGNOSIS

Orbital Dermoid & Epidermoid

- Lateral > medial canthus
- Tethered at suture: Frontozygomatic > nasolacrimal
- 50% show fat density/intensity with thin rim enhancement

Dacryocystocele

- Acquired lacrimal sac cyst from trauma, other processes
- Typically in adults with history of prior regional trauma

PATHOLOGY

General Features

- Etiology
  - Tears & mucus accumulate in NLD due to distal imperforate Hasner membrane (i.e., distal duct obstruction)
    - Most membranes perforate during vaginal delivery or normal breathing & crying at birth
  - Nasolacrimal sac distension &/or anatomic variation compresses canalicular system → trapdoor nasolacrimal sac obstruction
  - If bacteria enter distended sac → dacryocystitis ± cellulitis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Small, round, bluish, medial canthal mass identified at or shortly after birth = distended lacrimal sac
  - Nasal airway obstruction with respiratory distress (especially during feeding) with bilateral nasal components
    - Obligate nose breathers during infancy
- Other signs/symptoms
  - Tearing & crusting at medial canthus, preseptal cellulitis, dacryocystitis
  - Small NLD mucoceles may be identified incidentally on brain MR imaging in infants

Demographics

- Age
  - Infancy: 4 days to 10 weeks typically
- Gender
  - M < F (1:3)
- Epidemiology
  - Most common abnormality of infant lacrimal apparatus

Natural History & Prognosis

- 90% of simple distal NLD obstructions (or congenital dacryostenoses) resolve spontaneously by age 1
- Only 50% of patients recognized on prenatal MR ultimately have postnatal symptoms
- Intervention recommended before infection occurs to prevent nasal airway obstruction, dacryocystitis, & permanent sequelae

Treatment

- Daily manual massage ± prophylactic antibiotics
  - Manual massage inappropriate if NLD mucocele infected or causing airway obstruction
- 10% require probing with irrigation ± Silastic stent placement
- If endonasal component & no response to above → endoscopic resection with marsupialization
- Prognosis excellent with adequate initial treatment
- Theoretical risk of nasolacrimal apparatus scarring, amblyopia, & permanent canthal asymmetry if left untreated

DIAGNOSTIC CHECKLIST

Consider

- Cross-sectional imaging evaluates extent of lesion along lacrimal apparatus & excludes other sinonasal causes of respiratory distress in newborn

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Choanal Atresia

KEY FACTS

TERMINOLOGY

- Congenital obstruction of posterior nasal aperture(s)

IMAGING

- Unilateral or bilateral osseous narrowing of posterior nasal cavity with complete obstruction by associated membrane or bony plate
  - Thickening of vomer
  - Medial bowing of posterior maxilla(e)
  - ± air-fluid level in obstructed nasal cavity
- Unilateral in up to 75% (right > left)
- Bilateral in up to 25%
  - 75% of bilateral cases have other anomalies

TOP DIFFERENTIAL DIAGNOSES

- Choanal stenosis
- Pyriform aperture stenosis
- Nasolacrimal duct mucocele

PATHOLOGY

- Choanal atresia is most common congenital abnormality of nasal cavity
- Choanal atresia types
  - Mixed bony & membranous atresia in up to 70%
  - Purely bony atresia in up to 30%

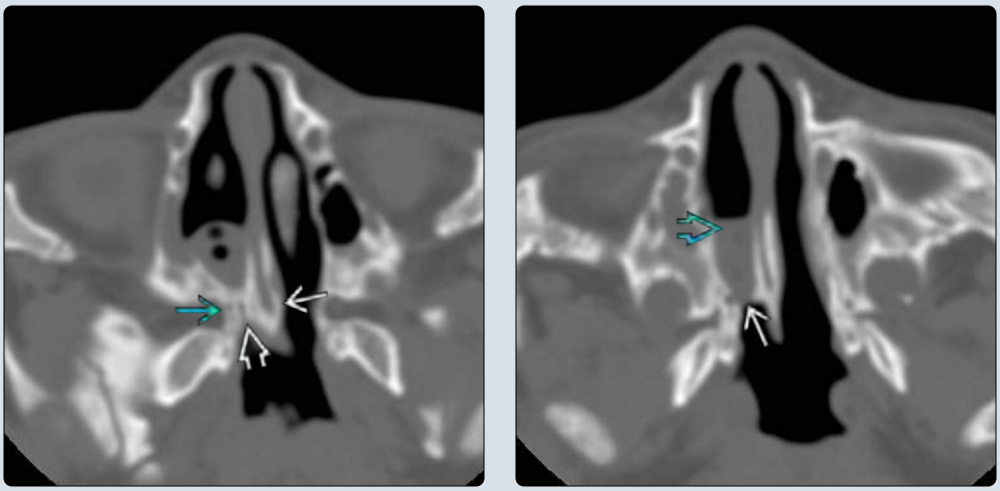
CLINICAL ISSUES

- Typical presentations include
  - Bilateral choanal atresia: Significant respiratory distress in newborn (due to "obligate nasal breather" status)
  - Unilateral choanal atresia: Chronic, purulent unilateral rhinorrhea with mild airway obstruction in older child

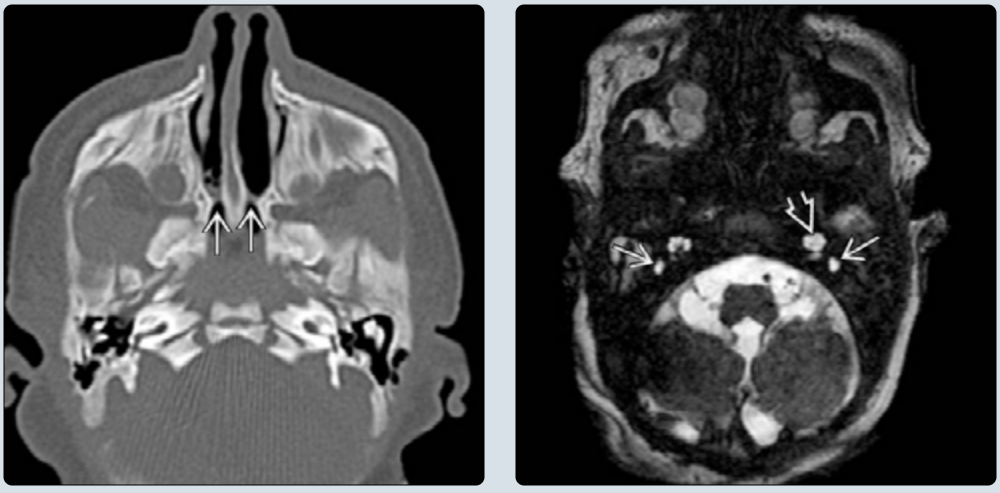
DIAGNOSTIC CHECKLIST

- Respiratory distress & suspected nasal obstruction in newborn should be evaluated with thin-section bone CT

(Left) *Axial NECT (in bone windows) through the upper choanae in a child shows a complete osseous right choanal obstruction secondary to fusion of an enlarged vomer to the thickened, medially positioned posterior maxilla.* **(Right)** *Axial NECT (in bone windows) in the same patient shows a component of membranous atresia at the narrowed inferior aspect of the choana. Note also the retained right nasal cavity secretions secondary to choanal obstruction.*



(Left) *Axial bone CT in a child with CHARGE syndrome demonstrates bilateral choanal obstructions secondary to linear membranes extending between the thickened vomer & each medially positioned posterior maxilla, typical of a mixed choanal atresia.* **(Right)** *Axial volume 3D FIESTA MR in the same child shows typical inner ear anomalies of CHARGE syndrome, including small/dysplastic vestibules, absent semicircular canals, & left cochlear dysplasia.*



TERMINOLOGY

Definitions

- Congenital obstruction of posterior nasal apertures
  - Choana: Junction of posterior nasal cavity & nasopharynx
  - Choanal atresia: Lack of communication between nasal cavity & nasopharynx

IMAGING

General Features

- Best diagnostic clue
  - Bony narrowing of posterior nasal cavity with membranous &/or osseous obstruction of choana
- Location
  - Unilateral in ~ 75% (right > left), bilateral in ~ 25%
- Size
  - Newborn choanal opening abnormal if < 0.34-cm wide
  - Newborn vomer abnormal if > 0.23-cm thick
- Morphology
  - Medial bowing of posterior maxilla (lateral nasal wall) & pterygoid plate
  - Large/thickened vomer
  - Bony narrowing ± soft tissue membrane/plug or bony plate obstructing choana
    - Mixed bony & membranous atresia in up to 70%
    - Purely bony atresia in up to 30%

CT Findings

- Bone CT
  - Choanal narrowing by medially bowed posterior maxilla & thickened vomer
  - Narrow gap between maxilla & vomer bridged by continuous bony plate or membrane
    - Membranous atresia may be thin/strand-like or thick/plug-like
  - Air-fluid level frequently present in obstructed nasal passage
    - Nasal cavity may also be filled with soft tissue, hypertrophied inferior turbinates

Imaging Recommendations

- Best imaging tool
  - High-resolution unenhanced bone CT
- Protocol advice
  - Suction secretions from nasal cavity prior to scanning
  - Axial images angled 5° cephalad to palate
    - If angle too great, region of choanae at level of skull base creates false appearance of choanal atresia
  - Edge enhancement bone kernel helps delineate bone margins in partially ossified skull base
  - Multiplanar reformations as needed
    - Sagittal usually best plane for this entity
  - 3D reconstructions may be helpful for clinical decision making & surgical planning

DIFFERENTIAL DIAGNOSIS

Choanal Stenosis

- Posterior nasal airway narrowed (not completely occluded)

Pyriform Aperture Stenosis

- Narrowed anterior inferior nasal passage(s)
- Thickened anteromedial maxilla(e)
- ± single central megaincisor
- Must evaluate brain for holoprosencephaly

Nasolacrimal Duct Mucocele

- Bilobed cystic mass extending from medial orbital nasolacrimal fossa to inferior meatus

PATHOLOGY

General Features

- Associated abnormalities
  - Syndromes common in bilateral atresia (up to 75%)
    - CHARGE syndrome
  - Unilateral choanal atresia more likely to be isolated

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Bilateral choanal atresia: Respiratory distress in newborn
    - Infants breathe through nose ("obligate nasal breathers") up to 6 months of age
    - Aggravated by feeding, relieved by crying
  - Unilateral choanal atresia or stenosis: Chronic, purulent, unilateral rhinorrhea in older child

Treatment

- Establish oral airway immediately to ensure proper breathing
- Membranous atresia may be perforated upon passage of nasogastric tube
- Surgical treatment effective for alleviating respiratory symptoms

DIAGNOSTIC CHECKLIST

Consider

- Once airway established, respiratory distress with suspected nasal obstruction in newborn should be evaluated with thin-section bone CT

Image Interpretation Pearls

- Determine if choanal atresia unilateral or bilateral
- Look for associated anomalies in head & neck

Reporting Tips

- Describe choanal atresia as
  - Unilateral or bilateral
  - Mixed membranous/bony or purely bony
    - Comment on thickness of atretic bone plate

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3. Slovis TL et al: Choanal atresia: precise CT evaluation. Radiology. 155(2):345-8, 1985

Congenital High Airway Obstruction Sequence

KEY FACTS

TERMINOLOGY

- Rare congenital anomaly of airway with complete, intrinsic laryngeal &/or tracheal obstruction
- Results in dysfunctional & hyperexpanded lungs, everted hemidiaphragms, & cardiac/venous compression with ascites/hydrops

IMAGING

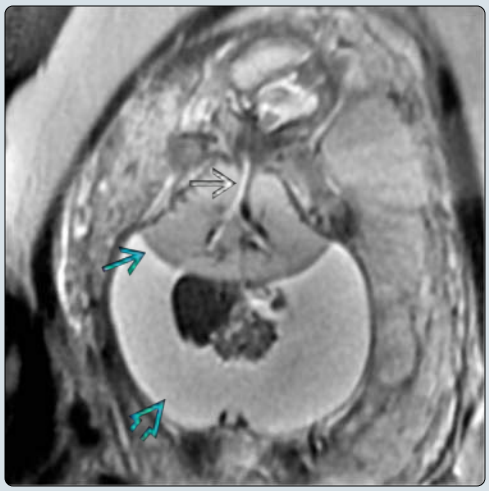
- Diffusely enlarged lungs, flattened/everted diaphragm
  - Prenatal imaging: Echogenic (US) or T2 hyperintense (MR) lungs with dilated, fluid-filled trachea inferior to obstructing lesion, ± polyhydramnios
  - Site of obstruction appears as persistent short or long segment of absent airway fluid at or below glottis
- Centralized, compressed heart
- Limited motion of abnormal diaphragm
- Abdominal distention with large volume of ascites
- Findings lessened with stenosis, membrane perforation, or fistula (as each enables airway decompression)

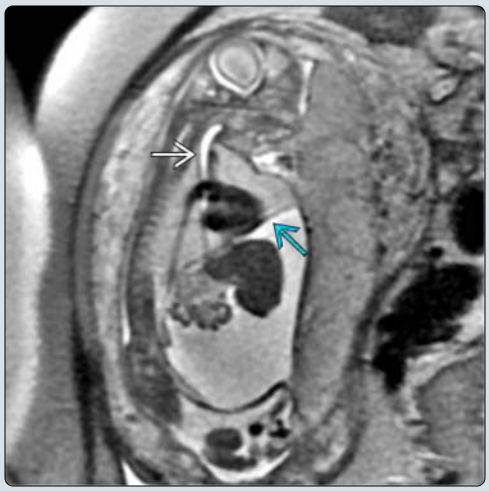
PATHOLOGY

- Most commonly due to laryngeal membrane or atresia
  - Obstruction prevents clearance of fluid from lungs
  - ↑ tracheal & lung pressures cause hyperexpansion & maldevelopment
- 50% have additional anomalies

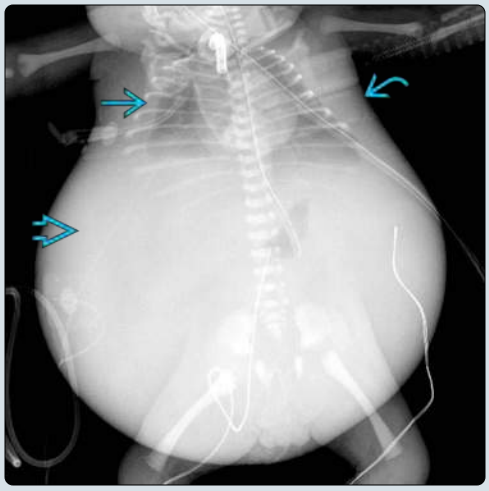
CLINICAL ISSUES

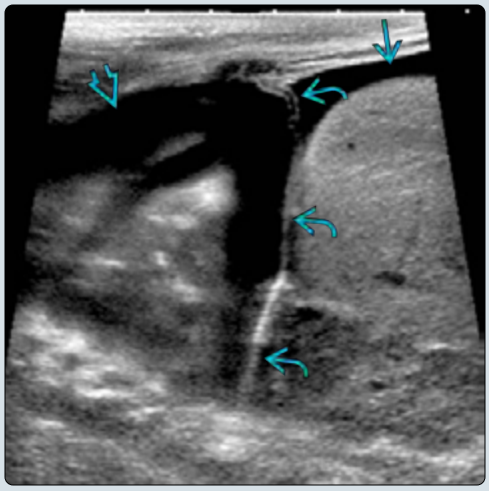
- Presentations
  - In utero: Large, bilateral solid lung masses + ascites
  - At birth: Respiratory distress, aphonia, & failed intubation
    - Lethal at delivery without prenatal detection
- Ex utero intrapartum therapy (EXIT) procedure: Controlled delivery with airway secured via tracheostomy while placental circulation maintained
  - Improves survival at delivery
  - Respiratory function remains poor
  - Survival > 1 year ~ 20%, with high morbidity





(Left) *Coronal SSFSE T2 MR in a fetus at 20 weeks gestation shows dilated, fluid-distended bronchi & trachea up to the larynx. The hemidiaphragms are everted, & the lungs show diffuse high signal intensity. There is a large volume of ascites.* **(Right)** *Sagittal SSFSE T2 MR in the same patient again shows the dilated, fluid-filled trachea (with obstructing membrane at the larynx), enlarged & high signal intensity lungs with flattened diaphragm, & ascites in this patient with CHAOS. The oligohydramnios was due to renal anomalies.*





(Left) *Coronal AP radiograph of a newborn with CHAOS shows marked abdominal distention (due to ascites), a pleural effusion, & chest wall edema, consistent with hydrops. The hemidiaphragms are flattened. A tracheostomy was placed during an EXIT procedure.* **(Right)** *Right parasagittal ultrasound of the lower thorax/upper abdomen in the same patient shows an abnormal, everted, & lax diaphragm, which did not move during the exam. The pleural effusion & ascites are noted.*

KEY FACTS

TERMINOLOGY

- Rare, highly lethal anomaly with absence of majority of trachea from subglottis to main bronchi

IMAGING

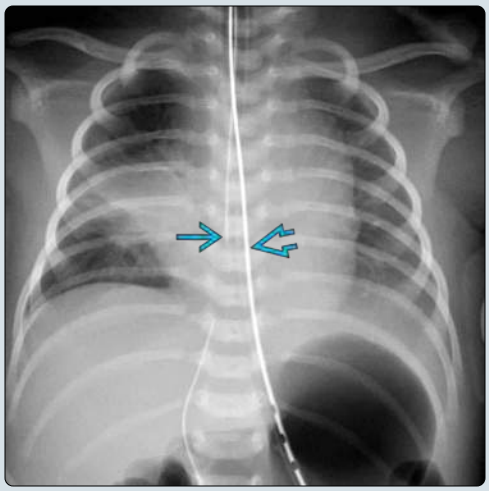
- Uncommon detection prenatally, as majority of cases have fistula from residual lower airway to esophagus
  - Allows decompression of otherwise obstructed lungs
  - May only demonstrate polyhydramnios in utero
    - If airway anomaly suspected, MR performed due to superior in utero airway evaluation vs. ultrasound
  - Minority of cases have no fistula, presenting as CHAOS
    - Lacks dilated, fluid-filled trachea of CHAOS
- Postnatally, temporary ventilation occurs via fistula
  - Low lung volumes with patchy opacities of atelectasis or aspiration
  - Low origin, horizontally oriented main bronchi
  - Endotracheal tube (ETT) in midline esophagus, possibly below expected location of carina
  - Nasogastric tube (NGT) may curve into main bronchi

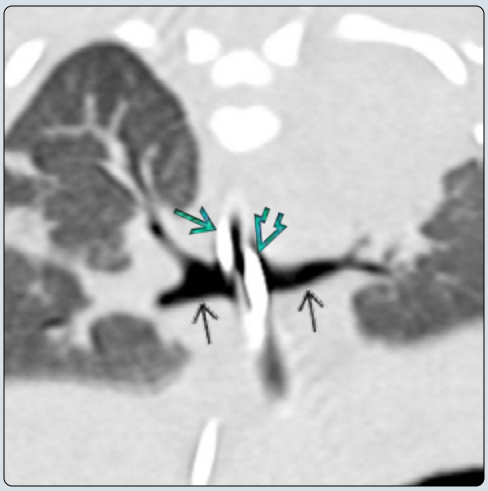
PATHOLOGY

- Classification by Floyd (most widely used): Types I-III
  - I: Absent proximal trachea; short distal trachea + TEF
  - II: Absent trachea; residual carina with main bronchi ± bronchoesophageal fistula
  - III: Absent trachea & carina; main bronchi arise from esophagus
- Associated anomalies in up to 94%

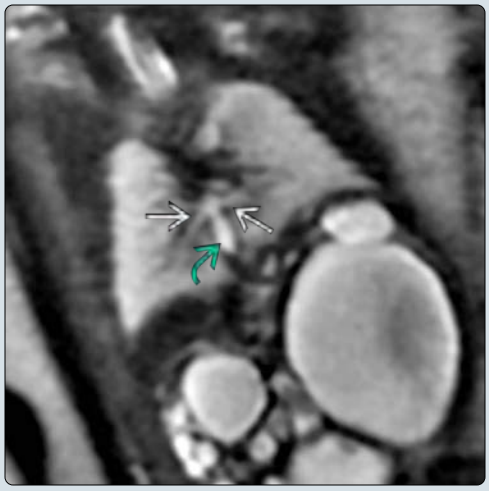
CLINICAL ISSUES

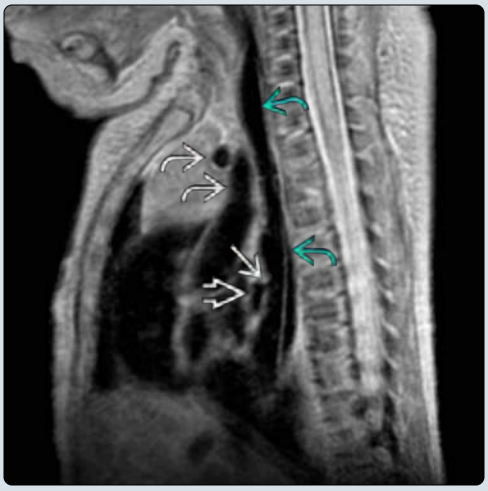
- Inability to conduct air at delivery → aphonia & insufficient ventilation; intubation difficult
- Highly lethal due to lack of sustainable conduit for air transit from glottis to lungs
- Long-term tracheal replacement options sparse
  - Rarely successful reconstruction with esophagus
  - Bioengineered graft material may be future therapy





(Left) *AP chest radiograph in a 34-week gestation newborn with cyanosis, aphonia, & poor ventilation at delivery shows the tip of the endotracheal tube (ETT) in the thoracic midline below the expected level of the carina. The ETT follows a similar course to the nasogastric tube (NGT).* **(Right)** *Coronal CECT in the same patient shows a horizontal orientation of low-lying main bronchi that connect to the esophagus. Note that both the NGT & low ETT lie in the esophagus in this patient with complete tracheal agenesis.*





(Left) *Coronal T2 SSFSE MR in a 31-week gestation fetus shows an abnormal connection of the bilateral main bronchi to the esophagus. No trachea was identified. This fetus had numerous anomalies, including radial ray, genitourinary, gastrointestinal, & cardiac.* **(Right)** *Sagittal PD MR in a 34-week gestation newborn shows a fistula from the carina to the esophagus. Vessels are seen anterior to the esophagus without a discernible trachea, consistent with tracheal agenesis.*

Epiglottitis

KEY FACTS

TERMINOLOGY

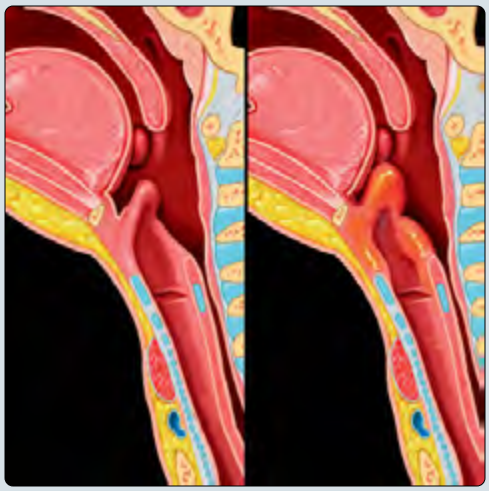
- Airway obstruction secondary to inflammation of epiglottis & surrounding tissues

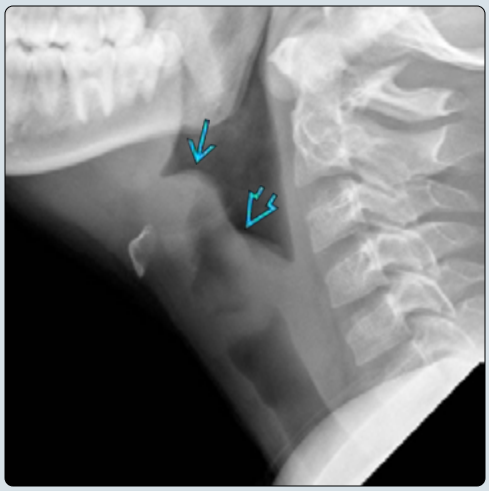
IMAGING

- Frontal & lateral radiographs only obtained in stable patients with questionable diagnosis
  - Child should be kept upright & comfortable
  - Patient may drool due to difficulty handling oral secretions: Should not be agitated or placed supine
- Lateral radiograph
  - Marked thickening of epiglottis
  - Thickening of aryepiglottic folds
    - Extend from epiglottis anterosuperiorly to arytenoid cartilages posteroinferiorly
    - Normally thin & convex inferiorly
    - May become thickened & convex superiorly
    - Swelling of these folds → actual airway obstruction
- Frontal radiograph
  - Only lateral radiograph should be obtained when epiglottitis highly suspected
  - ± symmetric subglottic tracheal narrowing, similar to that seen in croup
  - Swelling of epiglottis & aryepiglottic folds often not seen on frontal view; may be visualized through foramen magnum
- CT does not play routine role in diagnosing epiglottitis

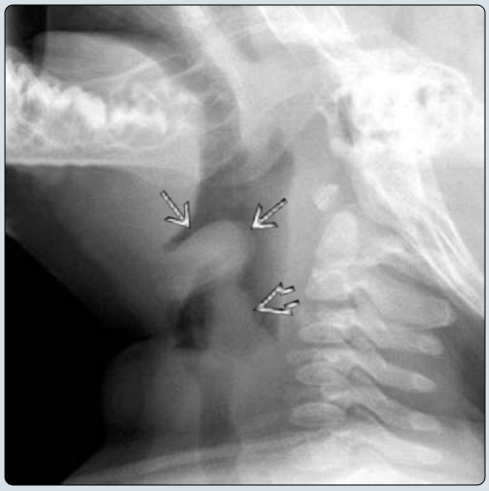
CLINICAL ISSUES

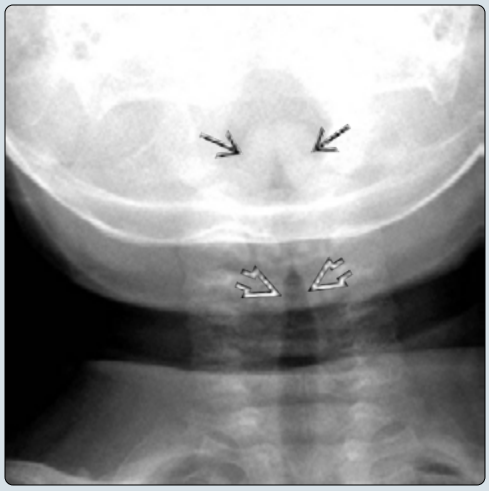
- Marked ↓ in incidence in children since vaccine for *Haemophilus influenzae* introduced
  - Mean age in children has shifted from 3.5 years to 14.6 years
  - Now more common in adults than children
- Life threatening if untreated
- Direct laryngoscopy & bronchoscopy with intubation performed in operating room with otolaryngology present
- Steroids & broad-spectrum IV antibiotic therapy





(Left) *Sagittal graphics shows epiglottitis (right) as compared with a normal epiglottis (left). The epiglottis & aryepiglottic folds are swollen & diffusely enlarged (right).* **(Right)** *Lateral radiograph of the airway in a 13 year old with lymphoma shows thickening of the epiglottis & aryepiglottic folds, consistent with epiglottitis.*





(Left) *Lateral radiograph in a 6 month old shows marked thickening of the epiglottis & the aryepiglottic folds, typical of epiglottitis.* **(Right)** *AP radiograph of the same child shows thickening of the epiglottis & aryepiglottic folds seen through the skull base/foramen magnum. There is mild subglottic narrowing, which can be seen in varying degrees with epiglottitis.*

TERMINOLOGY

Synonyms

- Supraglottitis

Definitions

- Airway obstruction secondary to inflammation of epiglottis & surrounding tissues

IMAGING

General Features

- Best diagnostic clue
  - Classic imaging appearance: Lateral radiograph shows enlargement/thickening of epiglottis & aryepiglottic folds
    - Epiglottis often appears like thumbprint (or frontal perspective of thumb with nail en face to x-ray beam)
  - Not to be confused with Ω epiglottis: Normal variant when epiglottis imaged obliquely
    - May appear like hitchhiker thumb (or lateral perspective of thumb with nail tangential to x-ray beam)
    - Epiglottis maintains sharp interfaces with well-defined central posterior border; no thickening of aryepiglottic folds
- Location
  - Serious, life-threatening inflammation & swelling of epiglottis & surrounding tissues (i.e., aryepiglottic folds)
- Morphology
  - Swelling of epiglottitis → thumbprint appearance on lateral radiograph

Radiographic Findings

- Lateral radiograph
  - Marked thickening of epiglottis
  - Aryepiglottic folds
    - Extend from epiglottis anterosuperiorly to arytenoid cartilages posteroinferiorly
    - Normally thin & convex inferiorly, outlined by air
    - May become thickened & convex superiorly
    - Swelling of these folds causes actual airway obstruction
  - ± nonspecific "ballooning" (air distention) of hypopharynx
- Frontal radiograph
  - Only lateral radiograph obtained if epiglottitis highly suspected
    - Supine positioning of ill-patient could lead to airway occlusion
  - ± symmetric subglottic narrowing, similar to that seen in croup
  - Swelling of epiglottis & aryepiglottic folds may not be seen on frontal view
    - May see swollen epiglottis through skull base

CT Findings

- CECT
  - CT has no role in diagnosing epiglottitis
  - If obtained (occasionally for other reasons), will show edematous, enlarged epiglottis with involvement of aryepiglottic folds
    - Epiglottis slightly lower in attenuation when compared with other soft tissue
  - In rare cases, may see phlegmonous collection within adjacent soft tissues
  - May be helpful in evaluating for complications such as deep neck space infection/abscess
    - Very rare in pediatric population as opposed to adult population where it can be seen in 2-29% of cases

Imaging Recommendations

- Best imaging tool
  - Due to serious, life-threatening airway emergency, unstable patients with classic clinical presentation undergo direct laryngoscopy & bronchoscopy with intubation in operating room by otolaryngology as indicated
  - Only lateral radiograph should be obtained in cases suspecting epiglottis
- Protocol advice
  - Child should be upright & comfortable
  - Patient may drool due to difficulty handling oral secretions; patient should not be agitated or placed supine
  - Patient with suspected epiglottitis should be accompanied by physician with readily available supportive equipment to secure airway if necessary
  - Obtaining lateral radiograph should never interfere with securing airway given potential for rapidly fatal outcome

DIFFERENTIAL DIAGNOSIS

Ω Epiglottis (Normal Variant)

- Artificially widened appearance of obliquely imaged normal epiglottis
  - Left & right sides of epiglottis being imaged adjacent to each other
- May appear as hitchhiker thumb, but epiglottis maintains sharp margins & well-defined central posterior border
- Lacks thickening of aryepiglottic folds

Croup

- Most common acute airway condition of children
- Benign, self-limited condition with "barky" cough in patients < 3 years of age
- Symmetric subglottic tracheal narrowing (steeple sign)

Exudative Tracheitis

- Children typically older than those with croup
- Intraluminal filling defects (membranes), tracheal wall plaque-like irregularity, poorly defined tracheal margins, asymmetric subglottic narrowing

Retropharyngeal Abscess

- Pyogenic infection of retropharyngeal space
- Persistent thickening of retropharyngeal soft tissues despite inspiration & neck extension on lateral view
  - Loss of normal "step-off" at junction of hypopharynx & esophagus

Enlarged Lingual Tonsils

- Rounded mass bulging from posterior tongue base, potentially filling vallecula & displacing epiglottis

Vallecular Mass

- Most commonly cyst but rarely sarcoma
- Fills vallecula, potentially displacing or effacing epiglottis depending on exact site of origin

PATHOLOGY

General Features

- Etiology
  - Most common agent remains *Haemophilus influenzae*
    - Dramatic changes in incidence, etiology, & patient demographics since *H. influenzae* (HiB) vaccine introduction
    - More cases of epiglottitis resulting from other bacterial, viral, or combined viral-bacterial infections now seen since introduction of HiB vaccination
      - Other organisms include group A β-hemolytic *Streptococcus*, *Staphylococcus aureus*, *Klebsiella pneumoniae*, *Moraxella catarrhalis*, *Pseudomonas* species, *Candida albicans*, *Pasteurella multocida*, & *Neisseria* species
      - Bacterial superinfections of preceding viral infections such as herpes simplex, parainfluenzae, varicella-zoster, & Epstein-Barr
  - Can also rarely occur from noninfectious etiologies such as angioneurotic edema, trauma, Stevens-Johnson syndrome, caustic ingestion, & bee stings

Gross Pathologic & Surgical Features

- Marked inflammation & edema of epiglottis & aryepiglottic folds
- Complete airway obstruction may occur at any time

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abrupt onset of stridor (usually inspiratory), often associated with dysphagia
- Other signs/symptoms
  - High fever, sore throat, dysphonia, "hot potato voice," hoarseness, & drooling
  - Patients have toxic appearance with fever
  - Patients described as anxious & uncomfortable
  - ↑ respiratory distress when recumbent
  - May have characteristic "tripod position" (sitting up with neck extended & leaning forward with jaw thrust out to maximize laryngeal opening)
  - Viral prodrome & cough more likely with croup

Demographics

- Age
  - Marked ↓ in incidence in children since HiB vaccine introduced
    - 0.6-0.8 cases /100,000 persons immunized
    - Vaccine effectiveness 98%
  - Mean age in children has shifted from 3.5 years to 14.6 years since introduction of HiB vaccine
    - Significantly older than children with croup (mean age: 1 year)
  - Adult incidence has remained steady (although relatively uncommon) post vaccine
  - Now more common in adults than children (mean age: 40 years)
- Gender
  - M:F = 1:1

Natural History & Prognosis

- Life-threatening disease often requiring emergent intubation
  - Mortality 0.89%, majority adults
- Intubation period usually short (2-3 days)

Treatment

- Emergent tracheal intubation to relieve/prevent airway obstruction & respiratory failure
  - Has evolved from tracheotomy to direct laryngoscopy & bronchoscopy with intubation performed in operating room with otolaryngology present
- Steroids & broad-spectrum IV antibiotic therapy

DIAGNOSTIC CHECKLIST

Consider

- Frontal & lateral radiographs should be obtained only in stable patients with questionable diagnosis
  - Patient should be quickly returned to emergency department
- Patient should remain in comfortable position, typically upright

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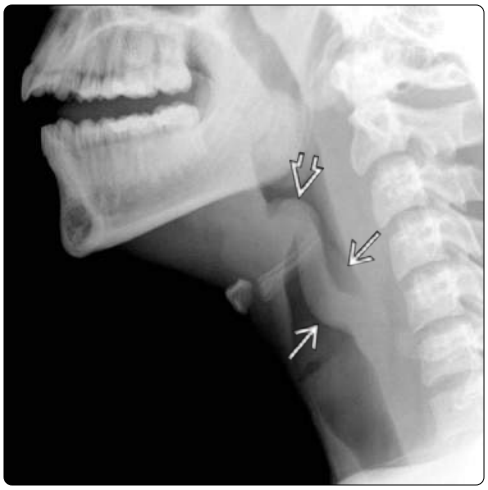
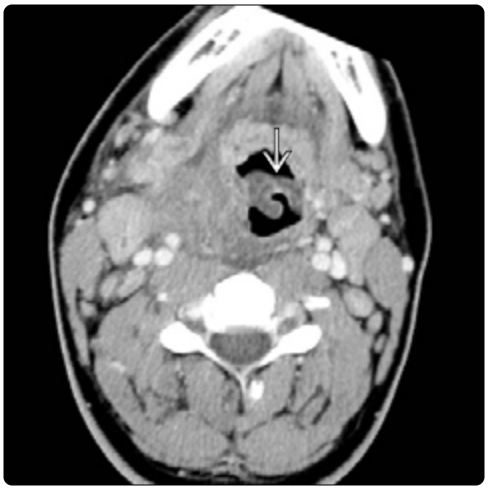
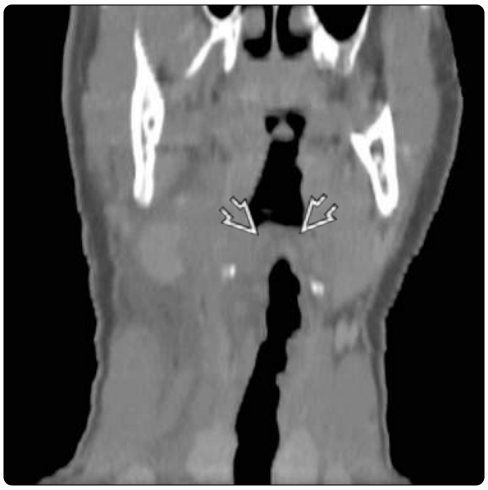
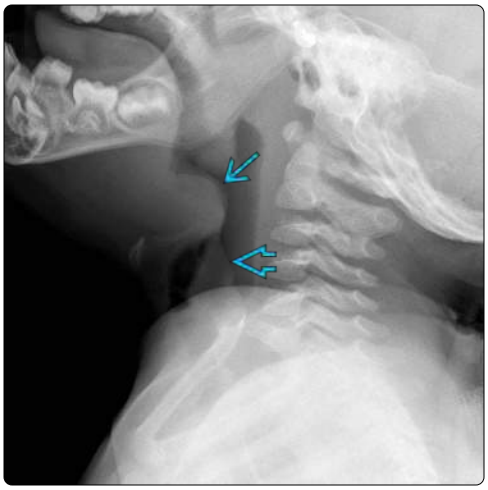


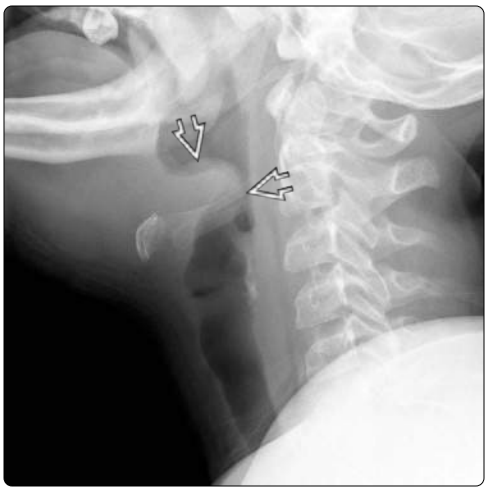


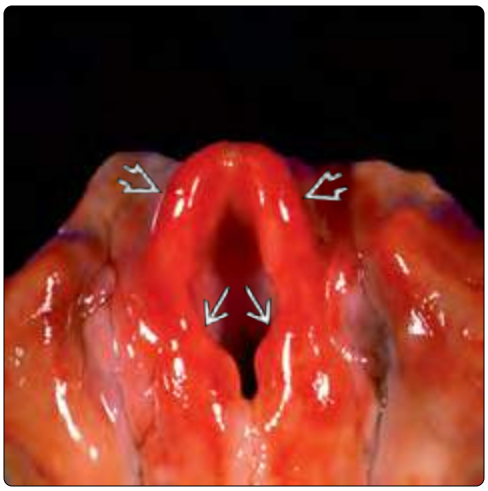
(Left) *Lateral radiograph shows marked thickening of the epiglottis & aryepiglottic folds.* **(Right)** *Axial CECT at the level of the hypopharynx shows an enlarged & low-attenuation, edematous epiglottis.*





(Left) *Coronal CECT reformatted image shows enlarged aryepiglottic folds.* **(Right)** *Lateral airway radiograph in an 18 month old with epiglottitis shows a markedly enlarged epiglottis with poorly defined margins. The right aryepiglottic fold was thickened at laryngoscopy. The patient was subsequently intubated.*





(Left) *Lateral airway radiograph shows epiglottitis as a markedly swollen & poorly defined epiglottis that has a thumbprint appearance in a patient with congenital insensitivity to pain.* **(Right)** *Gross pathology shows an inflamed & edematous epiglottis with swollen aryepiglottic folds. It is the swelling of the aryepiglottic folds that leads to airway obstruction.*

Croup

KEY FACTS

TERMINOLOGY

- Benign self-limited viral inflammation of upper airway
- Symmetric subglottic edema results in stridor & characteristic "barky" cough

IMAGING

- Radiographs used to exclude more serious causes of stridor (rather than diagnosing croup)
- Frontal view: Often more revealing than lateral view
  - Gradual symmetric tapering of subglottic trachea from inferior to superior
    - "Steeple," "pencil tip," or "inverted V" configuration
    - Loss of normal "shoulders" (focal lateral convexities) of subglottic trachea secondary to edema
- Lateral view: Best for excluding other diagnoses
  - Relatively mild narrowing of AP dimension
  - Haziness with loss of subglottic tracheal wall definition
  - ± hypopharyngeal overdistention

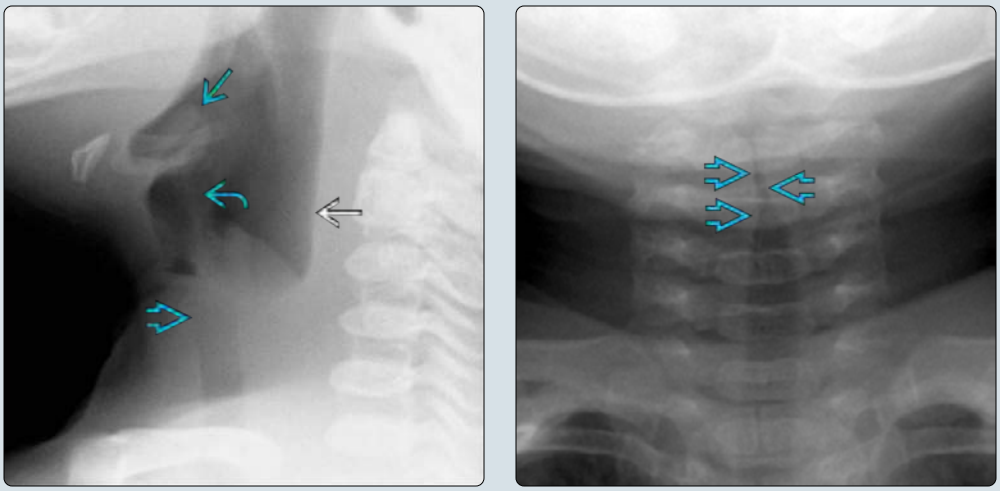
TOP DIFFERENTIAL DIAGNOSES

- Foreign body
- Epiglottitis
- Exudative tracheitis
- Angioedema
- Infantile hemangioma
- Iatrogenic subglottic stenosis

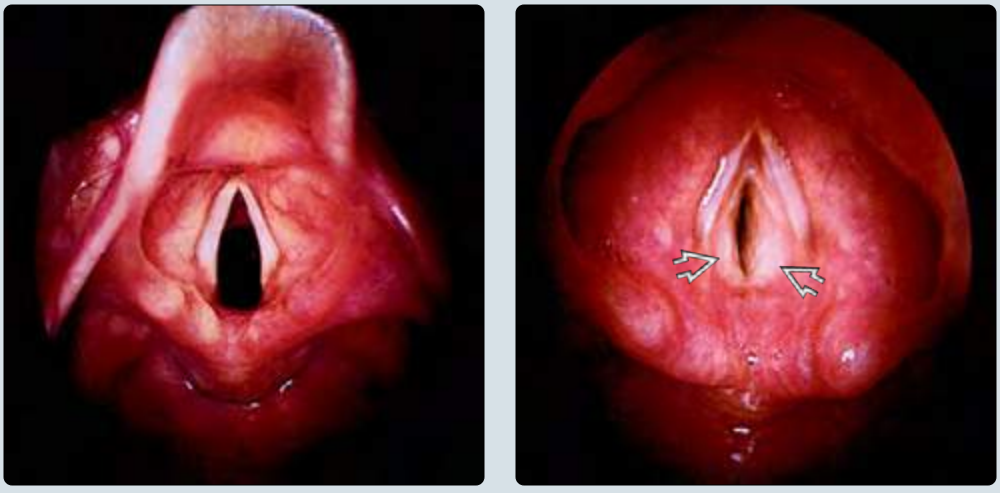
CLINICAL ISSUES

- Acute clinical syndrome characterized by "barky" or "seal-like" ("croupy") cough, inspiratory stridor, hoarseness
  - Age range: 6 months to 3 years; peak age: 1 year
- ± prodrome of low-grade fever, mild cough, rhinorrhea
- Affected child usually well otherwise
- Most cases successfully treated with corticosteroids ± nebulized epinephrine with < 4 hours of observation
- Recurrent episodes or atypical age suggest alternate diagnosis

(Left) *Lateral radiograph in a 9-month-old infant with stridor shows haziness of the subglottic airway. Overdistention (ballooning) of the hypopharynx is noted. The epiglottis & aryepiglottic folds are normal.* **(Right)** *AP radiograph in the same patient shows symmetric narrowing of the subglottic trachea, typical of croup. The loss of the normal abrupt subglottic/glottic shouldering plus gradual tapering of the subglottic airway lumen from inferior to superior is referred to as the steeple sign.*



(Left) *Endoscopic photograph shows a normal appearance of the subglottic airway. The subglottis is widely patent such that the mucosa is actually hidden beneath the vocal cords.* **(Right)** *Endoscopic photograph in a child with viral croup shows edematous subglottic mucosa, which is visualized through the vocal cords. There is marked narrowing of the subglottic airway lumen, predominantly in the transverse dimension.*



TERMINOLOGY

Synonyms

- Acute laryngotracheitis

Definitions

- Croup: Self-limited viral inflammation of subglottic trachea causing stridor & characteristic cough
- Acute laryngotracheobronchitis: Croup + lower airway involvement
- Spasmodic croup: Recurrent episodes, typically without viral prodrome or fever
- Atypical croup: Recurrent episodes or croup outside expected age range

IMAGING

General Features

- Best diagnostic clue
  - Symmetric subglottic airway narrowing with gradual tapering from inferior to superior on anteroposterior (AP)/frontal radiograph
  - Loss of normal focal "shoulders" of subglottis/glottis
- Morphology
  - Normal
    - Uniform caliber of cervical & thoracic trachea from subglottis to carina
    - Normal "shoulders" at subglottis/glottis: Symmetric, focal convex/angular lateral margins of airway
      - More horizontal than vertical
  - Croup shows gradual tapering of subglottic trachea from inferior to superior
    - Affected vertical length variable; most commonly upper 1/2-1/3 of cervical trachea
    - Edema effaces normal subglottic "shoulders"

Radiographic Findings

- Radiography
  - AP/frontal view
    - "Steeple," "pencil tip," or "inverted V" configuration of subglottic trachea
      - Loss of normal "shoulders" (lateral convexities) of subglottic trachea secondary to subglottic edema
      - Narrowing extends inferior to pyriform sinuses
  - Lateral view
    - Mild narrowing of subglottic trachea in AP dimension
      - In contrast to moderate to marked narrowing of transverse dimension on frontal view
    - Haziness with poor definition of subglottic tracheal walls
    - ± hypopharyngeal overdistention
      - Hypopharynx may be collapsed with distension of lower cervical trachea on expiratory image
    - Normal epiglottis, aryepiglottic folds, retropharyngeal soft tissues
    - No foreign body

Imaging Recommendations

- Best imaging tool
  - Diagnosis of croup primarily clinical, not imaging-based
    - Radiographs primarily used to exclude more serious causes of stridor
  - Frontal radiograph most useful view to confirm croup
  - Lateral radiograph helps exclude other diagnoses
- Protocol advice
  - Ensure that neck is extended with adequate inspiration on lateral view
    - Decreases crowding of airway structures that may simulate disease in young child
  - Avoid image acquisition while child swallows

DIFFERENTIAL DIAGNOSIS

Foreign Body

- Minority of foreign bodies radiopaque
  - May appear as soft tissue density but with straight, irregular, or pointed margins
- Can lodge virtually anywhere in airway from nasal/oral cavities to bronchi
  - Right main bronchus most common lower airway site
    - Causes asymmetric lung aeration on chest radiographs
  - Tracheal foreign bodies uncommon
- Symptoms depend on location of object
- Esophageal foreign bodies may also cause edema of adjacent trachea, especially in subacute/chronic setting

Epiglottitis

- Typically occurs in older children
  - Historical mean age (pre-Hib vaccine) = 3 years
  - Now teenagers more common
- Severe, life-threatening condition
- Marked enlargement of epiglottis & aryepiglottic folds
- May cause symmetric subglottic narrowing on frontal view

Exudative Tracheitis

- Toxic-appearing child, typically 6-10 years old
- Intraluminal filling defects (pseudomembranes)
- Plaque-like irregularity &/or poor definition of tracheal walls
- Asymmetric subglottic narrowing

Thermal Injury

- History of smoke inhalation, burns

Angioedema

- Rapid swelling of facial soft tissues & upper airway
  - ± itching, pain, hives
- May be due to allergic reaction or hereditary angioedema

Infantile Hemangioma

- Asymmetric airway narrowing with focal convex mass bulging from tracheal wall into lumen
  - Often not visible radiographically
- Associated with cutaneous infantile hemangiomas of face/neck in "beard" distribution
- Develops in 1st few weeks of life & grows rapidly over 6-12 months before beginning slow spontaneous regression

Iatrogenic Subglottic Stenosis

- History of prolonged intubation
- Predisposes to recurrent croup-like episodes

PATHOLOGY

General Features

- Etiology
  - Benign, self-limited condition secondary to viral illness
  - Infectious agents
    - Most cases: Parainfluenza viruses types 1-3
    - Less frequently: Rhinovirus, enterovirus, respiratory syncytial virus, influenza, measles; bacterial forms uncommon (consider *Corynebacterium diphtheriae* & *Mycoplasma pneumoniae*)
  - Leads to inflammation & edema of subglottic airway
    - Redundant mucosa predisposes to edema & narrowing
      - Loose mucosal attachment of conus elasticus
    - Swelling of vocal cords → hoarseness
    - Characteristic (but not specific) "barking/barky" cough results from inflammation of larynx & trachea
    - Inspiratory stridor due to proportionately small subglottic trachea in young children
      - Same viral infections & edema do not compromise adult-sized airway
- Associated abnormalities
  - With atypical or spasmodic croup
    - 20-64% incidence of large airway lesions: Subglottic hemangioma, stenosis, laryngeal cleft, tracheomalacia, laryngomalacia, papillomatosis, laryngeal web, or vocal cord paralysis
    - Additional common disorders in this group: Gastroesophageal reflux, asthma, sleep-disordered breathing, allergies, chronic cough, prematurity

Staging, Grading, & Classification

- Clinical staging
  - Mild: Stridor at rest or when agitated
  - Moderate: Stridor + mild tachypnea, mild retractions
  - Severe: Stridor + respiratory distress, severe retractions, ± altered mental status

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute clinical syndrome characterized by "barking" or "seal-like" ("croupy") cough, inspiratory stridor, hoarseness, respiratory distress
- Other signs/symptoms
  - ± prodrome of low-grade fever, mild cough, rhinorrhea
  - Affected child usually well otherwise & able to manage secretions
  - More severe cases: Intercostal retractions, tachypnea, pallor, cyanosis, tachycardia, altered mental status
  - Symptoms worse at night or with agitation
  - May occur with other symptoms of lower respiratory tract infection (wheezing, cough, etc.)

Demographics

- Age
  - Range: 6 months to 3 years; peak: 1 year
  - If > 3 years, consider other acute causes of stridor
    - Mean age of atypical croup: 2.7-4.8 years
  - If < 6 months, consider predisposing abnormality
- Gender
  - M:F = 3:2
- Epidemiology
  - Most common cause of acute upper airway obstruction in young children
    - Affects 5% of children by age 2 years
    - Affects 3% of children per year
  - Seasonal occurrence with viral disease
    - Most prevalent in fall-winter

Natural History & Prognosis

- Benign, self-limited disease
- 75% of mild cases resolve within 3 days
- 11% of mild & 49% of moderate cases of croup worsen
- 53% of severe cases require endotracheal intubation
- Overall: 8% hospitalized, 1% admitted to intensive care unit
- 5% return to emergency department < 1 week after initial evaluation
  - Consider other causes with recurrence, persistence

Treatment

- Most frequently managed supportively as outpatient
  - Mild croup: Systemic or nebulized corticosteroids, 2-hour observation
  - Moderate croup: Above + nebulized epinephrine, 4-hour observation
  - Severe croup: Above (can repeat epinephrine), admission to hospital
- Parents reassured, instructed to monitor for worsening course
- Endoscopy/bronchoscopy rarely needed if
  - Foreign body suspected
  - Superimposed exudative tracheitis suggested by clinical & imaging features
    - Requires membrane stripping, IV antibiotics, ± intubation
  - Assistance required for intubation in severe croup

DIAGNOSTIC CHECKLIST

Consider

- Alternate diagnosis with recurrent episodes or atypical age

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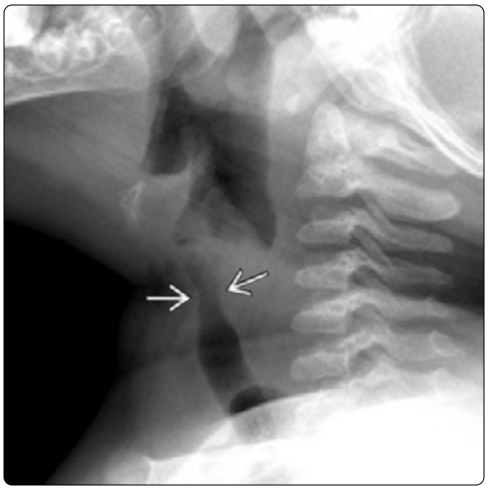
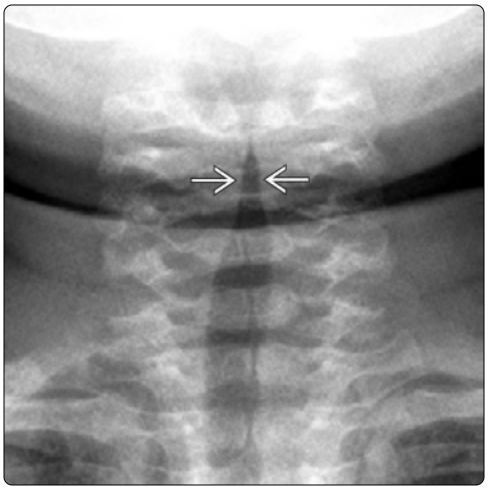
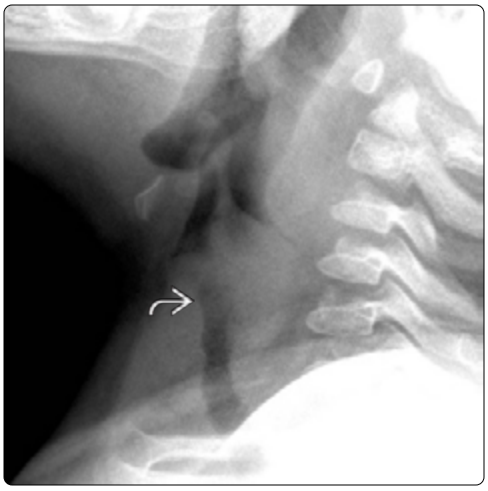
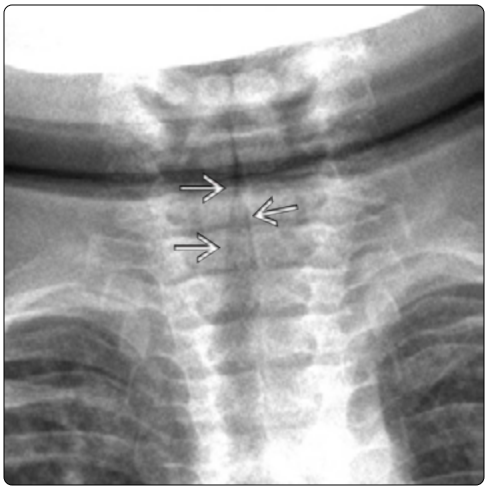


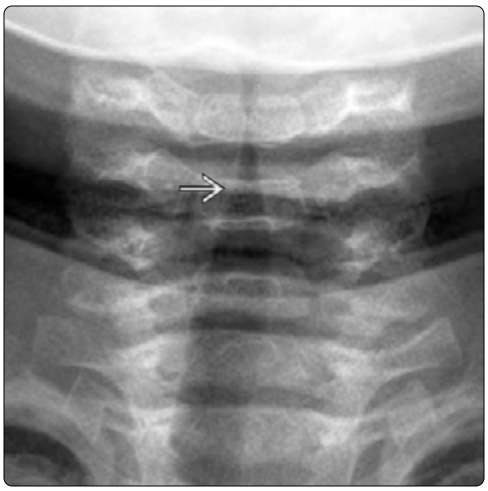


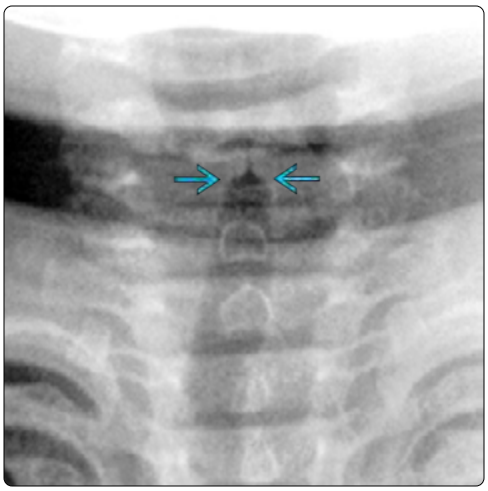
(Left) *Lateral radiograph in a 7-month-old infant with stridor shows symmetric narrowing of the subglottic airway ➡. (Right) AP radiograph of the same infant shows the steepled appearance ➡ of symmetric subglottic airway narrowing that is typical of viral croup.*





(Left) *Lateral radiograph in an 8-month-old infant with fever & a "barky" cough shows poor definition & haziness of the subglottic airway ➡, a common finding in viral croup.* **(Right)** *AP radiograph in the same patient shows severe narrowing of the subglottic trachea ➡, which gradually tapers from inferior to superior.*





(Left) *AP radiograph in a 16-month-old child with stridor shows symmetric subglottic airway narrowing ➡, typical of viral croup.* **(Right)** *AP radiograph in a normal child shows the typical shouldering ⇨ of the subglottic airway, which is more focal & horizontal than the long, gradual, vertical "steepling" seen in croup.*

Exudative Tracheitis

KEY FACTS

TERMINOLOGY

- Synonyms: Bacterial tracheitis, membranous or pseudomembranous croup, membranous laryngotracheobronchitis
  - Membranous croup: Confusing term overlapping much more common benign entity of viral croup
- Definition: Purulent infection of trachea
  - Results in thick, adherent, exudative plaques along tracheal walls
  - Plaques can slough, leading to airway obstruction
- Controversial disease
  - While many cases with significant morbidity & mortality have been confirmed, questions remain concerning overdiagnosis/overtreatment at certain centers

IMAGING

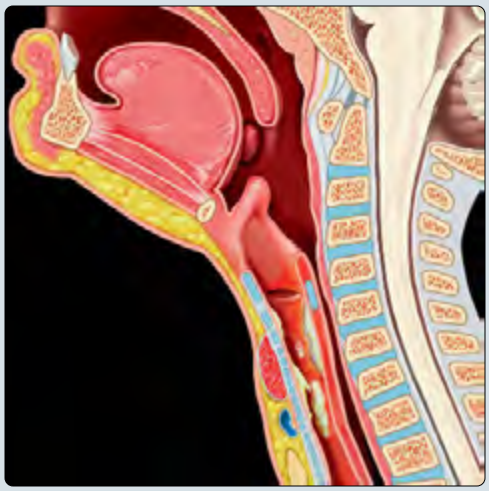
- Characteristic radiographic findings
  - Thin or thick linear or irregular soft tissue filling defects within airway (visualized pseudomembranes)
  - Loss of smooth well-defined parallel tracheal walls plus nodular plaque-like irregularity of walls
  - Hazy or indistinct tracheal air column
  - Symmetric or asymmetric subglottic narrowing in acutely ill child older than typically seen with viral croup

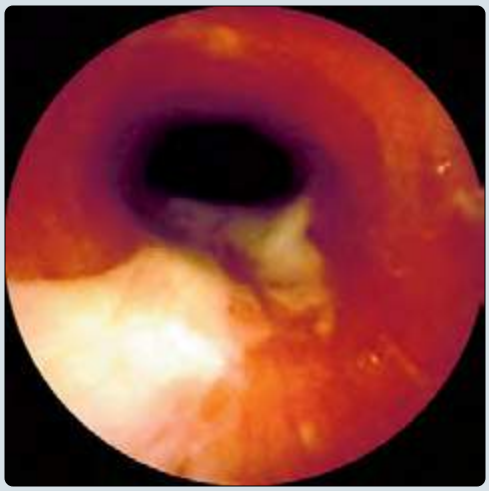
TOP DIFFERENTIAL DIAGNOSES

- Epiglottitis
- Croup
- Retropharyngeal abscess
- Upper airway obstruction by foreign body

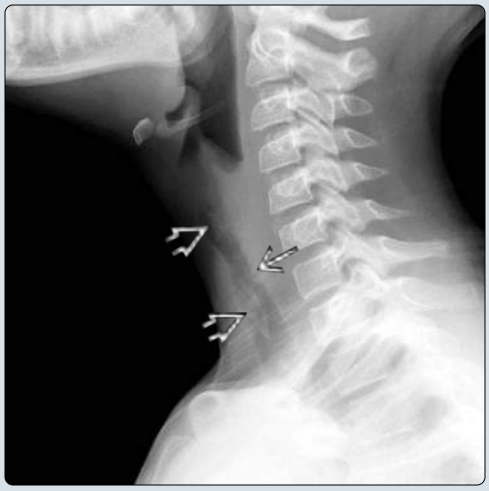
CLINICAL ISSUES

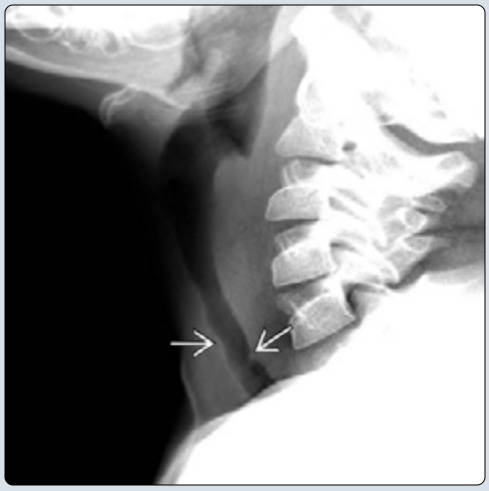
- High-grade fever, cough, severe stridor, rapid onset (2-10 hours) of respiratory distress; often after viral prodrome
- Peak age: 3-8 years (older than classic viral croup)
- Aggressive treatment to prevent airway obstruction, death
  - Flexible laryngoscopy → rigid bronchoscopy, removal of pseudomembranes ± intubation, IV antibiotics





(Left) *Graphic shows inflammation of the trachea with the formation of inflammatory plaques & pseudomembranes along the tracheal walls. These plaques may detach from the tracheal wall & occlude the airway.* **(Right)** *Photograph from bronchoscopic visualization of the trachea in a child with bacterial tracheitis shows multiple purulent exudative plaques along the tracheal walls.*





(Left) *Lateral airway radiograph shows multiple irregular intraluminal filling defects ➡ as well as tracheal wall irregularity ➡ & moderate luminal narrowing, consistent with exudative tracheitis.* **(Right)** *Lateral airway radiograph in a 5-year-old boy with fever, throat pain, & cough shows mild generalized tracheal narrowing with subtle lobular & linear filling defects ➡ concerning for plaques/pseudomembranes. Exudative tracheitis was confirmed at laryngoscopy.*

TERMINOLOGY

Synonyms

- Bacterial tracheitis, membranous or pseudomembranous croup, membranous laryngotracheobronchitis
  - Membranous croup: Confusing term overlapping much more common benign entity of viral croup

Definitions

- Purulent infection of trachea
  - Results in thick, adherent, exudative plaques along tracheal walls
  - Plaques can slough & obstruct airway
- Controversial disease
  - More frequent in certain medical centers; rarely seen in other centers of similar climate
  - No known cases of death in community without reaching medical center (as would be expected with life-threatening disease)
  - While many cases with significant morbidity/mortality have been confirmed, questions remain concerning overdiagnosis/overtreatment at certain centers

IMAGING

General Features

- Best diagnostic clue
  - Radiography demonstrates plaque-like irregularity of tracheal walls &/or linear filling defects (pseudomembranes) within tracheal lumen
- Location
  - Purulent infection of trachea, which may extend to involve larynx & bronchi

Radiographic Findings

- Radiography
  - Linear filling defects within subglottic airway
  - Plaque-like irregularity with loss of well-defined, smooth parallel tracheal wall contours (candle dripping sign)
    - Mucus can mimic plaques but should clear on repeat images after patient coughs
      - Coughing may be contraindicated if clinical suspicion high (due to risk of dislodging pseudomembranes)
  - Hazy, indistinct tracheal air column
  - Symmetric or asymmetric subglottic narrowing in child older than typically seen with croup

CT Findings

- Not routinely used in diagnosis
- Tracheal filling defects may be detected on lung window images obtained for other clinical suspicions

MR Findings

- Not used in diagnosis

Imaging Recommendations

- Best imaging tool
  - Frontal + lateral airway radiographs

DIFFERENTIAL DIAGNOSIS

Epiglottitis

- Severe, life-threatening condition
- Marked enlargement of epiglottis & aryepiglottic folds
- May cause symmetric subglottic narrowing on frontal view, similar in appearance to croup (but not as isolated finding)
- Fever, sore throat, drooling, respiratory distress in recumbent position
- Peak age now shifted to teenage years
  - Previously 3.5 years prior to *Haemophilus influenzae* vaccine

Croup

- Benign, self-limited condition
- Most common upper airway disease encountered in children
- Symmetric subglottic narrowing (steepling)
- Characteristic cough (barking, seal-like)
- Younger age (mean: 1 year) than exudative tracheitis

Retropharyngeal Abscess

- True thickening of retropharyngeal soft tissues that does not change with inspiration
- CECT confirms rim-enhancing fluid collection with surrounding edema & adenopathy
- Fever, sore throat, toxic-appearing, leukocytosis
- Most often < 6 years of age

Upper Airway Foreign Body

- Rarely radiodense
- May be seen as soft tissue filling defect in oropharynx/hypopharynx
- ± recent choking episode

PATHOLOGY

General Features

- Favored to represent bacterial superinfection following compromise of respiratory mucosa by viral illness rather than isolated primary bacterial infection
- *Staphylococcus aureus* remains most common etiology
  - *H. influenzae*, *Streptococcus* species, *Moraxella catarrhalis, Pseudomonas aeruginosa* less commonly implicated
  - Polymicrobial infection often present, supporting etiology of secondary infection
- Viral cultures also frequently positive (up to 65%), which may be due to preceding illness
  - Influenza A most common

Gross Pathologic & Surgical Features

- Endoscopy shows laryngeal & subglottic edema/erythema, ulcerations, copious secretions with adherent pseudomembrane formation
- Exudative plaques can slough & lead to obstruction of airway (much like historic airway obstructions with *Corynebacterium diphtheriae*)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms

- High-grade fever, cough, severe stridor, rapid onset (2-10 hours) of respiratory distress
  - Cough may be barky like viral croup
- Usually preceded by several-day history of viral upper respiratory tract infection, low-grade fever, cough

- Other signs/symptoms
  - Initial descriptions reported patients as severely toxic in appearance
  - More recently encountered patients not always severely ill
  - Affected children typically able to handle oral secretions & tolerate supine position, unlike epiglottitis
- Clinical profile
  - Preexisting intubation or tracheostomy may predispose to bacterial tracheitis
    - Presentations more indolent
      - ↑ purulent secretions requiring more frequent suctioning of airway
      - ↓ oxygen saturations
    - Most commonly develops ~ 4 days after intubation with 2-11% incidence
      - Less predictable, lower incidence with longstanding tracheostomy
    - May be precursor to pneumonia

Demographics

- Age
  - 3 months to 14 years; peak age of 3-8 years with mean age of 5 years
    - Much older than patients who present with classic viral croup
- Gender
  - No predilection
- Epidemiology
  - Incidence: 4-8 per 1 million children
  - Has replaced epiglottitis as most common life-threatening acute inflammatory airway disease
    - Due to development of *Haemophilus influenzae* vaccine, which has decreased epiglottitis frequency

Natural History & Prognosis

- Systemic complications include
  - Septic shock
  - Pulmonary edema
  - Acute respiratory distress syndrome
- Mortality historically 20-40%
  - ↓ due to more aggressive work-up, therapy

Treatment

- If exudative tracheitis suspected clinically/radiographically, child usually evaluated with flexible laryngoscopy
- If tracheal pseudomembranes/plaques confirmed with direct visualization, patient undergoes rigid bronchoscopy with stripping of pseudomembranes ± intubation
  - Multiple debridements usually not required
  - Intubation lasts 2-7 days
    - Less frequently required in older children
      - Close monitoring still necessary
- IV antibiotics may be transitioned to oral antibiotics once culture sensitivities return
- ± corticosteroids, racemic epinephrine to reduce airway edema
  - Efficacy not proven

DIAGNOSTIC CHECKLIST

Consider

- Exudative tracheitis may present
  - With rapid deterioration after typical viral croup manifestations in young child
  - In older child with croup-type symptoms

Image Interpretation Pearls

- Luminal linear filling defects (pseudomembranes) + irregularity/poor definition of tracheal walls (plaques): Very suggestive of diagnosis
- Repeat radiographs after patient coughs if adherent mucus suspected as cause of tracheal filling defects/nodularity
- Coughing may be contraindicated if clinical suspicion high (due to risk of dislodging pseudomembranes)

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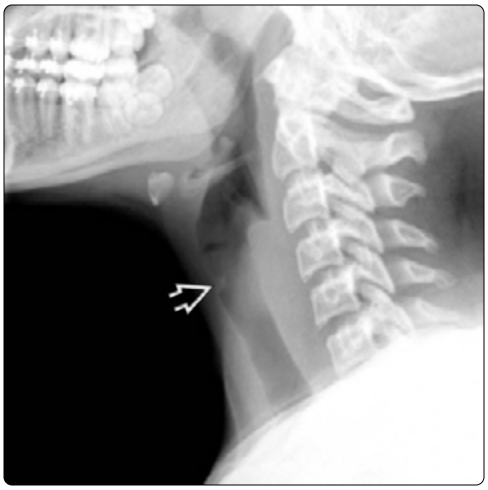
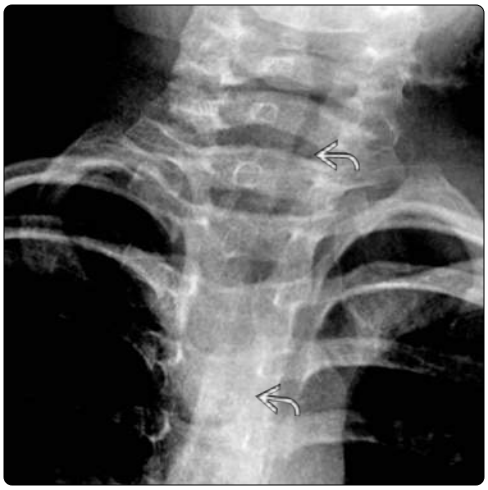
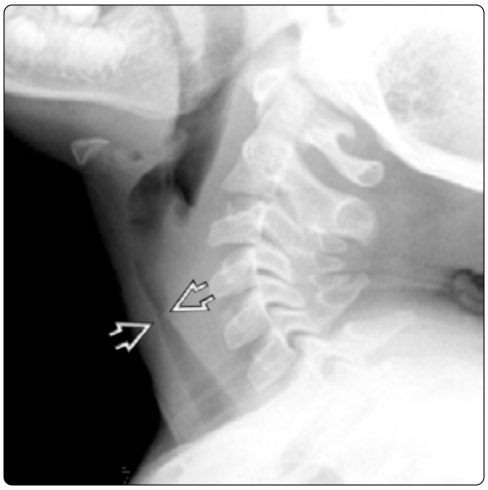
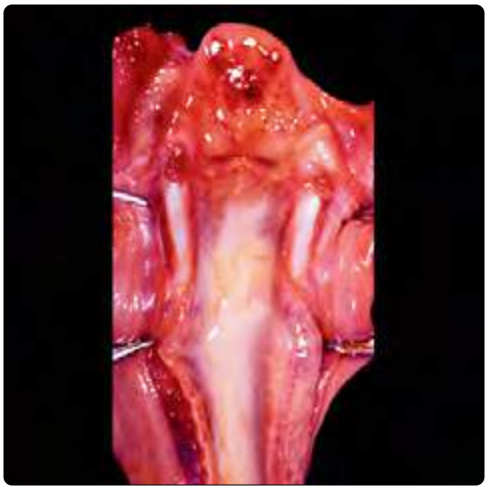


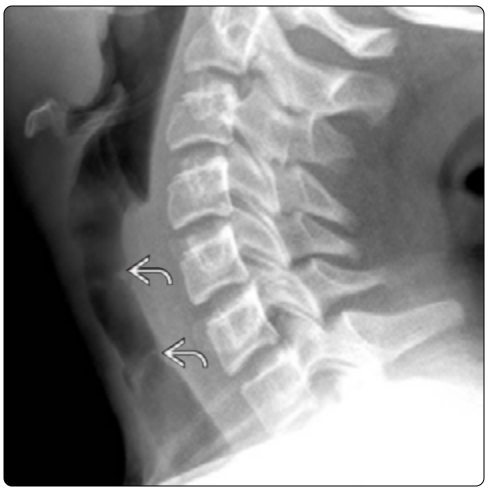


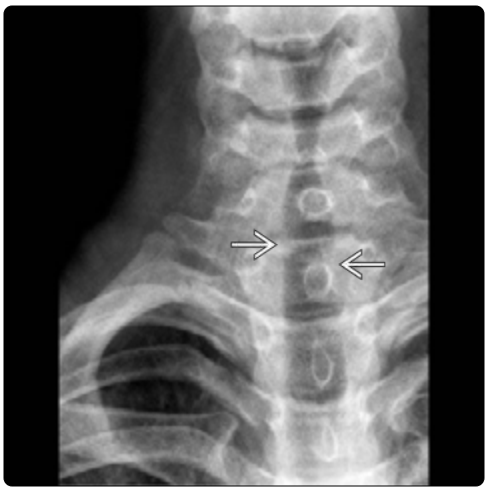
(Left) *Lateral radiograph in a young boy shows a plaque-like excrescence protruding into the subglottic airway lumen from the anterior tracheal wall.* **(Right)** *AP airway radiograph in a 4-year-old girl with fever & rapidly progressive respiratory distress shows poor definition of the tracheal wall with adjacent luminal filling defects. Extensive pseudomembranes were found at rigid bronchoscopy.*





(Left) *Lateral radiograph in a 9-year-old boy shows moderate narrowing with an associated plaque in the cervical trachea, consistent with exudative tracheitis.* **(Right)** *Gross pathology image from a deceased patient shows exudative material along the entire subglottic trachea. Airway compromise & obstruction may result from sloughing of this exudative material.*





(Left) *Lateral airway radiograph in a 12-year-old girl with fever, throat pain, & difficulty breathing shows several linear luminal filling defects along sites of wall lobularity, suspicious for pseudomembranes.* **(Right)** *AP radiograph in the same patient shows similar findings with lobular filling defects along the tracheal walls. Laryngoscopy confirmed exudative tracheitis.*

Retropharyngeal Space Abscess

KEY FACTS

TERMINOLOGY

- Extranodal purulent fluid collection in retropharyngeal space (RPS)

IMAGING

- Lateral radiograph: Wide prevertebral distance with loss of normal contours at hypopharynx-esophagus interface
- CECT best tool for rapid characterization & evaluation of extent/complications
  - RPS distended by defined, ovoid, rim-enhancing low-density collection with convex anterior margin
  - Complications include airway compromise, jugular vein thrombosis/thrombophlebitis, mediastinal extension/mediastinitis, internal carotid artery (ICA) pseudoaneurysm (rare, suggests methicillin-resistant *Staphylococcus aureus*)

TOP DIFFERENTIAL DIAGNOSES

- Pseudothickening of retropharyngeal soft tissues
- Retropharyngeal space edema
- Necrotic/suppurative adenopathy in RPS
- Lymphatic malformation

PATHOLOGY

- Most common etiology: Rupture of suppurative RPS lymph node → abscess

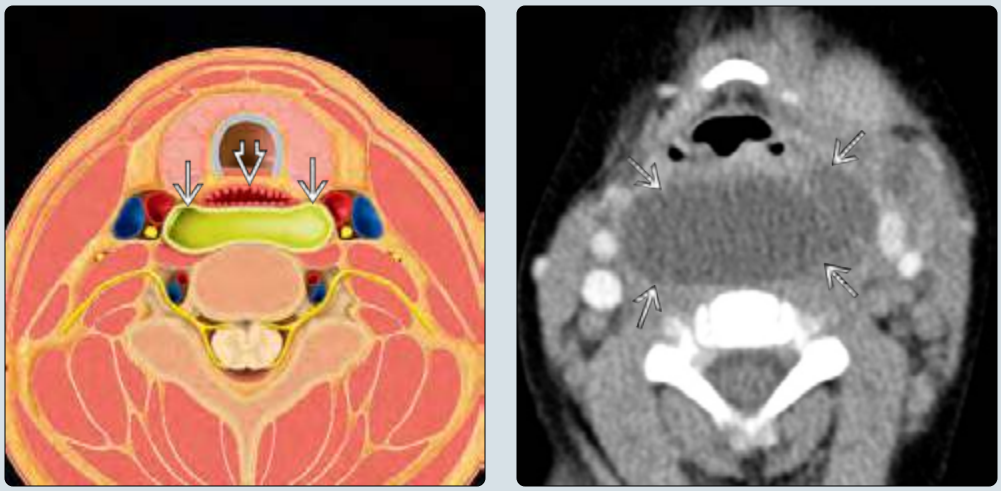
CLINICAL ISSUES

- Dysphagia, sore throat, poor oral intake, dehydration
- Toxic patient: Fever, chills, ↑ WBC & ESR
- Most < 6 years old; increasing incidence in adults

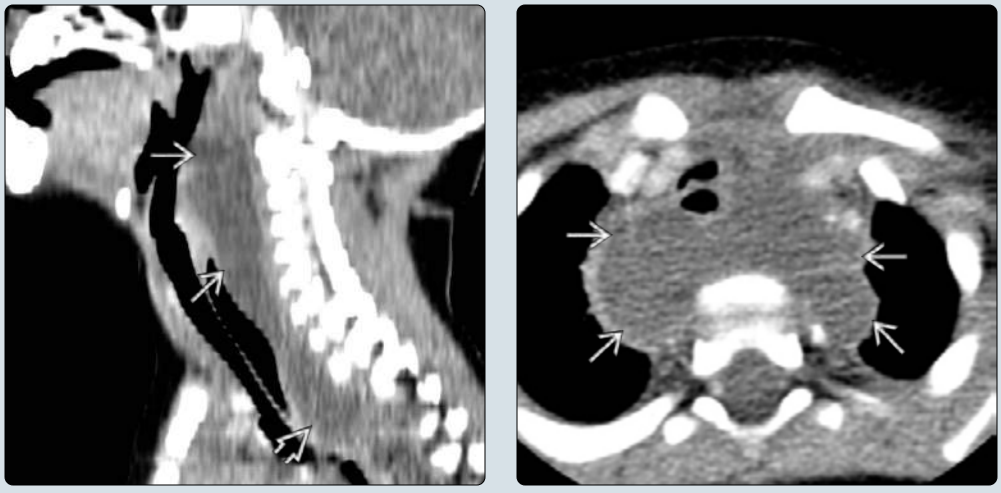
DIAGNOSTIC CHECKLIST

- Distinction from RPS edema sometimes difficult
  - Oval-shaped collection with convex anterior margin & rim enhancement suggests drainable abscess
    - Rim enhancement may be minimal if
      - Early in disease course
      - CECT performed with early/arterial timing

(Left) *Axial graphic illustrates the location & typical contour of a retropharyngeal space (RPS) abscess displacing the cervical esophagus anteriorly & flattening the prevertebral muscles.* **(Right)** *Axial CECT in a 10 month old with a 5-day history of febrile illness reveals a large, low-density, ovoid collection distending the RPS with anterior displacement of the pharynx & splaying of the carotid sheaths. There is minimal enhancement of the collection wall.*



(Left) *Sagittal reformatted CECT in the same infant reveals an abscess displacing the pharynx & esophagus anteriorly & extending inferiorly to involve the superior mediastinum.* **(Right)** *Axial CECT reveals the inferior extension of this methicillin-resistant Staphylococcus aureus (MRSA) abscess to the superior mediastinum. This fluid was drained through a cervical approach to the collection. There was no stridor or other sign of airway compromise despite a displaced & narrowed airway.*



TERMINOLOGY

Definitions

- Retropharyngeal space (RPS): Midline space posterior to pharyngeal mucosa & cervical esophagus from skull base to T3 vertebral level in mediastinum
- RPS abscess: Extranodal purulent fluid collection in RPS

IMAGING

General Features

- Best diagnostic clue
  - Midline rim-enhancing RPS fluid collection with mass effect in toxic-appearing patient
- Location
  - Posterior to pharynx & anterior to prevertebral muscles
- Size
  - Variable: May extend from skull base to mediastinum
- Morphology
  - Defined collection, not just infiltrative edema
  - Axial plane: Oval shape with convex anterior margin

Radiographic Findings

- Lateral view critical
  - Normal prevertebral soft tissue thickness
    - C2 (at hypopharynx): ≤ 7 mm at any age
    - C6 (at cervical esophagus): ≤ 14 mm if < 15 years, ≤ 22 mm in adults
  - In children: Must perform lateral radiograph during inspiration & with neck extension
    - Neck flexion & expiration often cause pseudothickening of prevertebral soft tissues in young children
    - Lateral fluoroscopy can distinguish persistent true thickening vs. dynamic pseudothickening
- With RPS abscess, lateral view shows widened/thickened prevertebral soft tissues
  - Convex anterior bowing with loss of normal step-off at interface of hypopharynx & esophagus
  - Limited utility for defining extent of collection & differentiating cellulitis/phlegmon from abscess
  - RPS gas rare but diagnostic of abscess (if no trauma)

CT Findings

- CECT
  - RPS markedly distended by defined fluid collection with enhancing wall
    - In early stages, enhancement may be subtle
    - Thick enhancing wall suggests mature abscess
    - Prevertebral muscles may also appear edematous
    - Gas rarely present
  - Assess for complications
    - Airway compromise
    - Internal jugular vein (IJV) frequently compressed/effaced, rarely thrombosed
    - Internal carotid artery (ICA) frequently narrowed (spasm)
      - Pseudoaneurysm rare; suggests methicillin-resistant *Staphylococcus aureus* (MRSA)
    - Mediastinal extension

Ultrasonographic Findings

- Not able to assess full craniocaudal or deep extent of disease
- Limited by operator experience & patient tolerance
- May help evaluate jugular vein patency

Imaging Recommendations

- Best imaging tool
  - CECT
- Protocol advice
  - CT from skull base to carina
  - Contrast loading (split) bolus &/or non-CTA technique improves soft tissue contrast resolution given rapid acquisition times of CT
    - ↑ sensitivity for collections, rim enhancement

DIFFERENTIAL DIAGNOSIS

Pseudothickening of Retropharyngeal Soft Tissues

- Common radiographic mimic of retropharyngeal pathology in young children
- Adequate extension & inspiration or fluoroscopy will confirm as transient finding

Retropharyngeal Space Edema

- Poorly defined, elongated, homogeneous fluid infiltration of prevertebral soft tissues without rim enhancement
- In axial plane: Bow tie shape with concave anterior margin
- RPS vessels may traverse infiltrated tissues
- Drainage not required
- Etiologies include
  - Regional inflammation
    - Pharyngitis, tonsillitis, longus colli tendinitis
  - Venous or lymphatic obstruction
    - IJV thrombosis or resection, radiation

Suppurative Adenopathy in Retropharyngeal Space

- Centrally hypodense/necrotic lymph node in lateral RPS with adjacent cellulitis
- May lead to pus formation in reactive node (suppuration): Intranodal abscess
- May progress to extranodal RPS abscess with inadequate medical therapy

Lymphatic Malformation

- Uni- or multilocular, transspatial, cystic neck mass with thin, nonenhancing wall (unless infected)
- Typically involves anterior & lateral neck

PATHOLOGY

General Features

- Etiology
  - Head & neck infection (pharyngitis, tonsillitis) seeds RPS lymph node
    - Reactive node → suppurative intranodal abscess → nodal rupture → RPS abscess
    - Most common organisms: *Staphylococcus aureus*, *Haemophilus*, *Streptococcus*
  - Pharyngeal penetration by foreign body
    - Child running with object in mouth
      - Lollipop/sucker, toothbrush, stick, toy

- Ventral spread of discitis/osteomyelitis & prevertebral infection
  - Uncommon in children
  - Pyogenic or tuberculous
- Mediastinal abscess spreading cranially
  - Esophageal rupture & mediastinitis with danger space (DS) abscess

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Dysphagia, sore throat, poor oral intake, dehydration
  - Septic patient: Fever, chills, elevated WBC & ESR
- Other signs/symptoms
  - Posterior pharyngeal wall edema or bulge
  - Reactive cervical adenopathy
- Clinical profile
  - Toxic-appearing child with marked neck pain & limited movement, especially in extension
  - Uncommonly presents with airway compromise (stridor)

Demographics

- Age
  - Most often children < 6 years old
  - Increasing frequency in adult population
    - Immunocompromised states: Diabetes, HIV, alcoholism, malignancy
    - Discitis/osteomyelitis with perivertebral infection
    - Trauma with foreign body impaction
    - Following anterior cervical spine surgery
- Gender
  - M:F = 2:1
- Epidemiology
  - ↑ frequency over last decade
  - ↓ incidence of abscess when infection detected & treated in earlier cellulitic stage

Natural History & Prognosis

- Prognosis generally excellent with early diagnosis & aggressive management
- Complications may result from infection spread
  - Narrowing of pharyngeal lumen → airway compromise & stridor
  - Inferior spread via DS to mediastinum → mediastinitis
    - Up to 50% mortality (much less in infants)
      - ↓ mortality rate in recent years with aggressive initial & repeat surgical intervention
    - MRSA frequent
  - Carotid space involvement
    - Jugular vein thrombosis or thrombophlebitis
    - Narrowing of ICA caliber often found; neurological sequelae infrequent
    - Rarely ICA pseudoaneurysm &/or rupture; described with MRSA infection
  - Perivertebral space abscess may lead to epidural abscess
  - Aspiration pneumonia
  - Grisel syndrome rare
    - Inflammatory, nontraumatic atlantoaxial subluxation
    - Distension or loosening of atlantoaxial ligaments after head & neck inflammation

Treatment

- Early ENT consultation
- IV antibiotics, airway management, fluid resuscitation
- Surgical intervention (incision & drainage) for
  - Significant or complex abscess
  - Lack of improvement/worsening with IV antibiotics

DIAGNOSTIC CHECKLIST

Consider

- Lateral radiograph: Often 1st-line screening tool
- Study of choice: CECT
  - Determine RPS vs. perivertebral space
  - Distinguish RPS abscess from edema
  - Evaluate full craniocaudal extent
  - Evaluate for vascular/airway complications
  - Consider contrast prebolus/split bolus or venous phase imaging to maximize wall enhancement

Image Interpretation Pearls

- Distinction from RPS edema sometimes difficult
  - Convex anterior margin or oval-shaped collection suggests abscess
  - Rim enhancement suggests abscess
    - May be minimal early in disease
    - May not be evident due to early arterial scanning
- Important to evaluate full extent of abscess & presence of complications
- ENT consultation imperative

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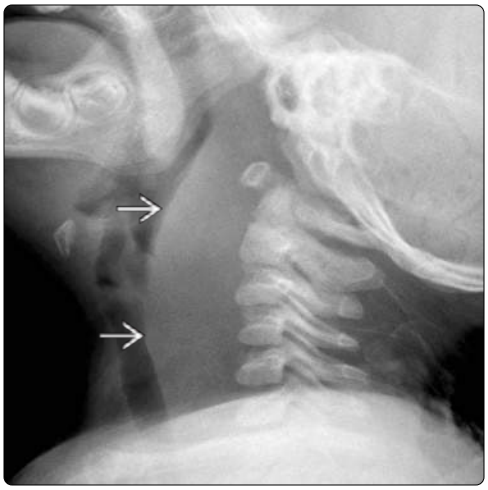
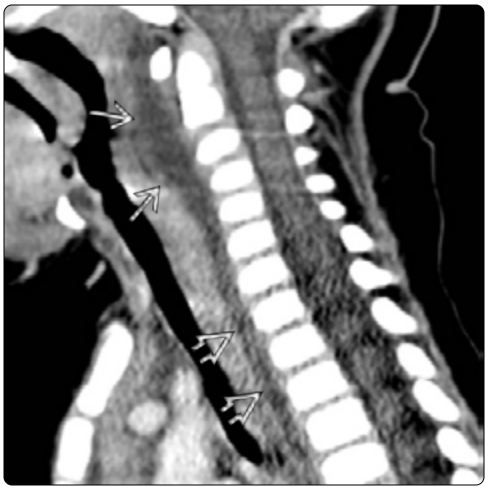
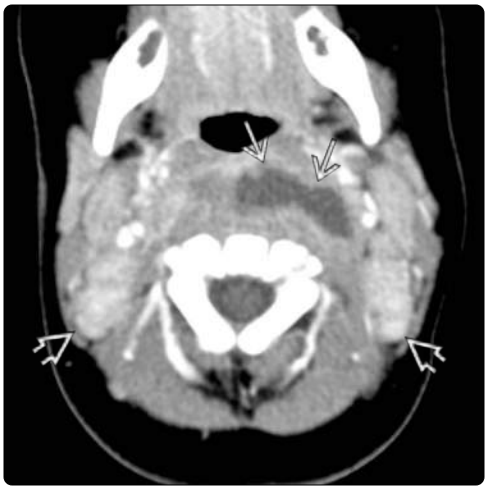
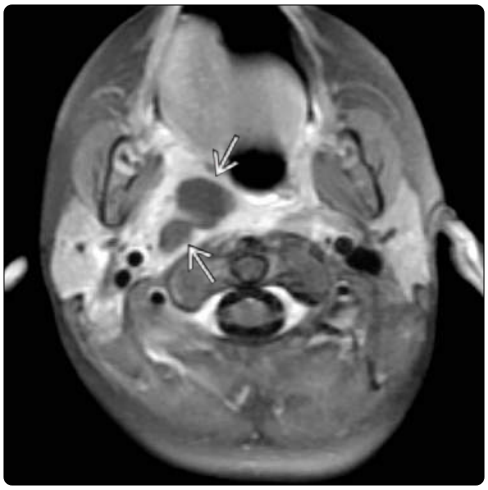


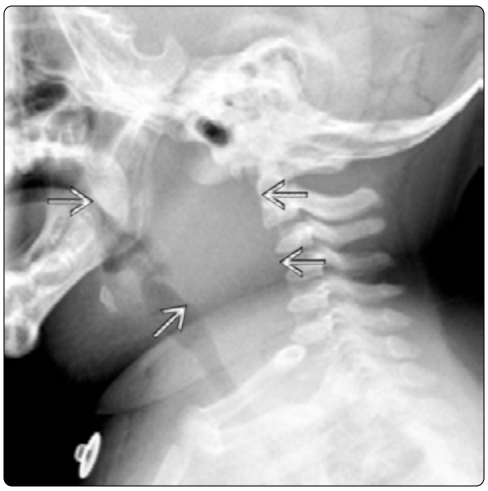


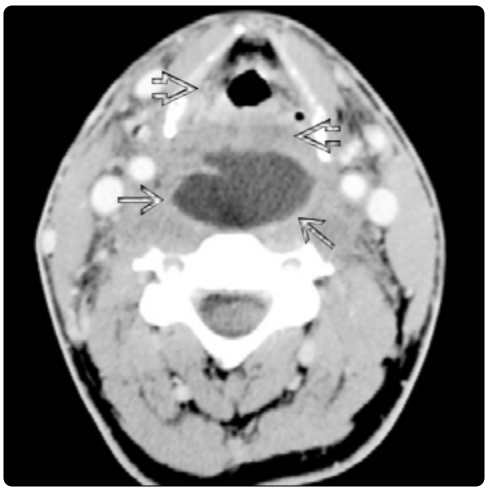
(Left) *Lateral radiograph in a 12-month-old boy with sepsis shows significant thickening of the prevertebral soft tissues ⇨. The normal step-off at the pharyngeal-esophageal junction has been effaced.* **(Right)** *Sagittal reformatted CECT in the same child clearly shows the cause of the prominent soft tissues to be a convex anterior RPS abscess ⇨ with extension of fluid into the posterior mediastinum ⇨.*





(Left) *Axial CECT in a 5-month-old boy shows an irregularly shaped RPS fluid collection ⇨ in the midline & left paramidline soft tissues, most likely an extranodal spread of a suppurative left lateral RPS lymph node. Notice the bilateral nonsuppurative cervical adenopathy ⇨.* **(Right)** *Axial T1 C+ FS MR in a child allergic to iodinated contrast demonstrates a well-defined abscess ⇨ in the right lateral RPS with significant surrounding contrast enhancement.*





(Left) *Lateral radiograph in a 7-month-old child with fever shows marked thickening of the prevertebral soft tissues ⇨ with ventral displacement of the pharynx & larynx. There is loss of the normal step-off at the pharyngeal-esophageal junction.* **(Right)** *Axial CECT in the same patient shows a faintly enhancing rim ⇨ to a large RPS, which displaces the hypopharynx & larynx anteriorly ⇨.*

Enlarged Adenoid Tonsils

KEY FACTS

TERMINOLOGY

- Adenoid tonsils (AT) lie along posterior wall of nasopharynx
- Enlargement may be chronic & idiopathic or acute/subacute with pathology (as from infection or neoplasm)
- Chronically enlarged AT can lead to obstructive sleep apnea (OSA) by obstructing posterior nasopharynx
- AT regrowth is common cause of recurrent/persistent OSA following palatine tonsillectomy & adenoidectomy (T&A)
  - Visualization of AT on such studies = recurrence

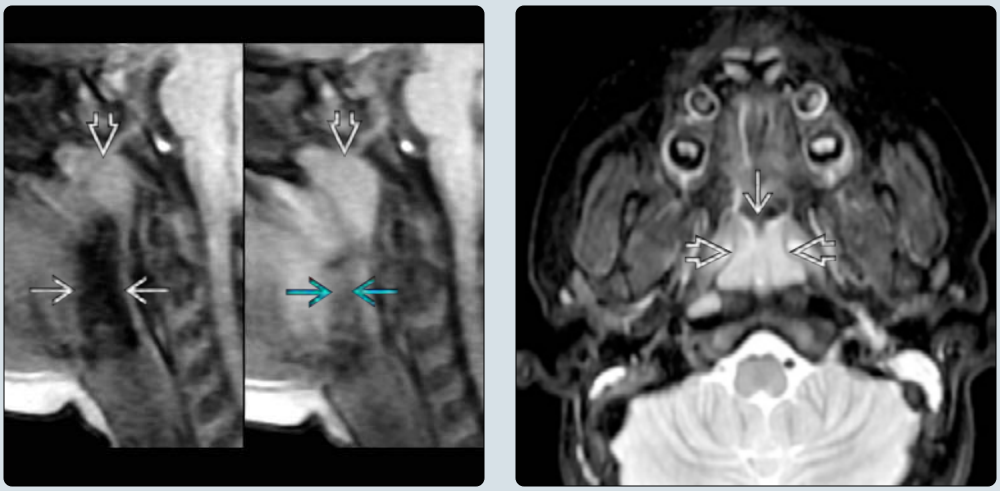
IMAGING

- Growth of normal adenoids without history of T&A
  - Absent at birth
  - Rarely visible radiographically prior to 6 months of life
  - Proliferate rapidly during infancy
  - Reach maximal size by 2-10 years of age
  - Progressively ↓ in size during 2nd decade of life
- Enlarged &/or recurrent AT
  - Radiographs
    - Soft tissue fullness of nasopharyngeal posterior wall
      - > 12-mm thickness
    - Encroachment on posterior nasopharyngeal airway
  - MR imaging with cine sequences during respiratory cycle
    - T2 FS/STIR: High-signal AT tissue
      - V-shaped defect in midportion of anterior adenoid surface on axial images status post T&A
    - Cine: Intermittent collapse of posterior nasopharynx ± secondary collapse of retroglossal airway

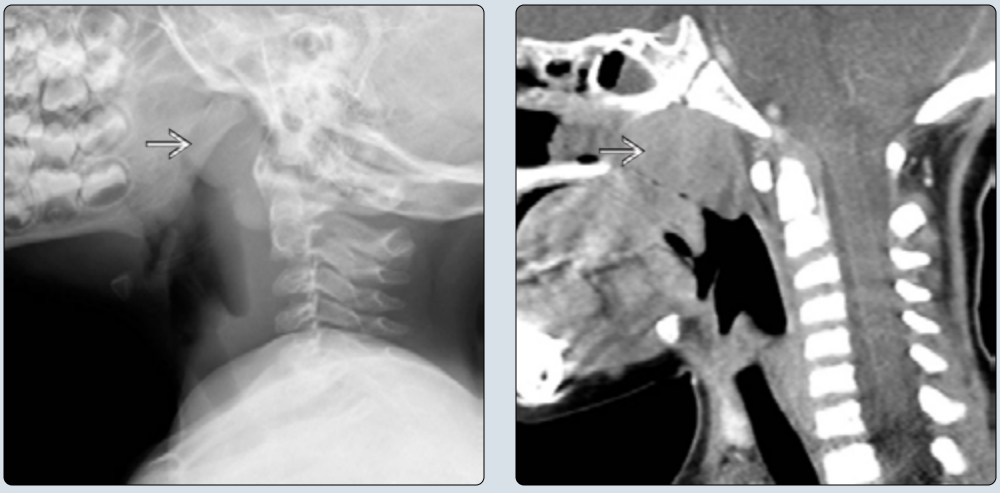
CLINICAL ISSUES

- OSA in 3% of children, any age
  - OSA may manifest as snoring (± pauses, gasps), neurocognitive impairment, daytime sleepiness, pulmonary or systemic hypertension, failure to thrive
  - Treatment: Adenoidectomy; repeat for recurrence
- Other pathologic causes of adenoid enlargement (acute to chronic, without OSA): Tonsillitis/pharyngitis, neoplasm

(Left) *Sagittal MR cine images during expiration (left) & inspiration (right) show enlarged adenoid tonsils with effacement of the nasopharynx. The retroglossal airway is patent during expiration but collapses during inspiration due to the negative pressure generated by the more cephalad obstruction.* **(Right)** *Axial STIR MR in the same patient after adenoidectomy shows recurrent adenoid tonsils. Note the anterior midline defect of the adenoids, a typical postoperative appearance.*



(Left) *Lateral radiograph in a 4 year old with fever & difficulty swallowing shows marked enlargement of the adenoid tonsils due to streptococcal pharyngitis.* **(Right)** *Sagittal midline CECT in the same patient shows marked enlargement of the adenoid tonsils with homogeneous enhancement. No abscess was identified in this patient with streptococcal pharyngitis.*



TERMINOLOGY

Definitions

- Adenoid tonsils (AT): Exposed lymphoid tissue at posterior wall of nasopharynx
- Enlargement may be chronic & idiopathic or acute/subacute with pathology (as from infection or neoplasm)
- When AT chronically enlarged, may lead to obstructive sleep apnea (OSA) by obstructing posterior nasopharynx

IMAGING

General Features

- Location
  - Posterior nasopharyngeal wall anterior to clivus, C1, & dens of C2
- Size
  - Normal adenoid growth [without history of tonsillectomy & adenoidectomy (T&A)]
    - Absent at birth
    - Rarely visible radiographically < 6 months of life
    - Proliferate rapidly during infancy
    - Reach maximal size by 2-10 years of age
    - Progressive ↓ in size during 2nd decade of life
  - Recurrence of enlarged adenoids (after T&A)
    - Oblique AP dimension (depth) > 12 mm
      - Measured perpendicular to anterior clivus
- Morphology
  - Lateral/sagittal images: Triangular ± bulging anterior convexity
  - Axial images: Smooth anterior margin
  - Recurrent tissue (after T&A) shows V-shaped defect in midportion of anterior adenoids on axial images
    - Adenoidectomy performed centrally (to avoid damage to ostiomeatal complex), often leaving residual tissue laterally
    - AT regrow from lateral to medial

Radiographic Findings

- Soft tissue fullness of nasopharyngeal posterior wall
  - > 12-mm thickness on lateral view
- Loss of lucent nasopharyngeal airway at posterosuperior soft palate

CT Findings

- CECT shows homogeneous enhancement unless necrosis present due to infection or tumor

MR Findings

- Most MR sleep studies performed for persistent/recurrent OSA after previous T&A
  - Visualization of AT on such studies = recurrence
- T2 FS or STIR
  - Sagittal & axial planes best for AT
    - High-signal AT tissue against dark background
- MR cine sequences during respiratory cycle
  - Intermittent collapse of
    - Posterior nasopharynx (primary phenomenon)
    - Retroglossal airway (secondary phenomenon)
      - Secondary to negative pressure during inspiration
      - Generated by more cephalad obstruction by adenoids

Imaging Recommendations

- Best imaging tool
  - For previously healthy child with suspected upper airway pathology → lateral radiograph of upper airway
  - For child with persistent/recurrent OSA despite T&A → MR sleep study with cine

DIFFERENTIAL DIAGNOSIS

Soft Tissue Mass in Nasopharynx

- Foreign body
- Retention cyst
- Tornwaldt cyst
- Antrochoanal polyp
- Infection (cellulitis, retropharyngeal abscess)
- Trauma (edema, hematoma)
- Neoplasm
- Cephalocele

Common Causes of Obstructive Sleep Apnea

- Enlarged palatine tonsils
- Enlarged lingual tonsils
- Glossoptosis
- Hypopharyngeal collapse
- Enlarged soft palate

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - OSA (chronic): Snoring (± pauses, gasps), neurocognitive impairment, daytime sleepiness, pulmonary or systemic hypertension, failure to thrive
  - Neoplasm (subacute/chronic): Nasal stuffiness, asymmetric fullness, cervical adenopathy
  - Tonsillitis/pharyngitis (acute): Fever, sore throat, preceding viral prodrome, cervical adenopathy

Demographics

- Snoring in 12% & OSA in 3% of children, any age
  - Various predisposing factors: Obesity, Down syndrome, neuromuscular disorders, craniofacial anomalies

Treatment

- OSA requires adenoidectomy; repeat for recurrence
  - 2-4% readmission rate, often for hemorrhage
- If neoplasm suspected, requires biopsy plus further staging prior to therapy
- Uncomplicated tonsillitis/pharyngitis treated with antibiotics

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Enlarged Palatine Tonsils

KEY FACTS

TERMINOLOGY

- Palatine tonsils: Paired, discrete masses of lymphoid tissue on either side of oropharynx
- Enlargement may be chronic & idiopathic or acute/subacute (as from infection or neoplasm)
- Chronic enlargement may lead to obstructive sleep apnea (OSA)

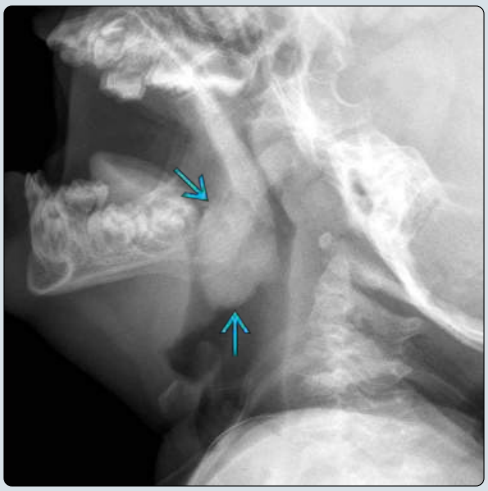
IMAGING

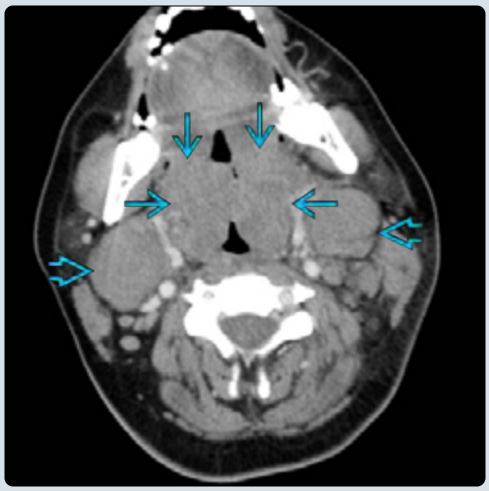
- Clinical evaluation based on
  - Local & systemic symptoms
  - Size, asymmetry, & discoloration of palatine tonsils + presence of cervical adenopathy
    - No clear size measurements established on imaging
    - Enlargement determined by clinically visible percentage of oropharynx occupied by palatine tonsils
- Lateral airway radiograph to evaluate upper airway pathologies (other than palatine tonsils)
  - Palatine tonsils appear as well-circumscribed soft tissue mass superimposed over mandibular angle
- CECT for acute presentations suggesting drainable abscess
  - Symmetric, homogeneously enhancing paired palatine tonsils show variable degrees of airway effacement
  - Asymmetry & heterogeneous enhancement may indicate pathology (e.g., abscess, necrotic tumor)
- MR sleep studies typically reserved for
  - Recurrent OSA despite previous surgery
    - Palatine tonsils do not typically recur
  - Complex syndromes predisposing to multilevel obstruction

CLINICAL ISSUES

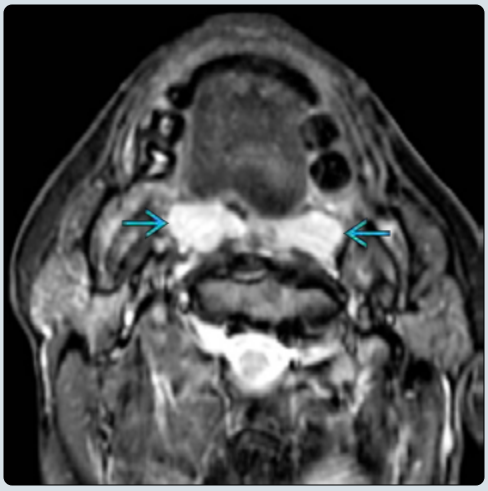
- Most children presenting with OSA are otherwise healthy with enlarged adenoid & palatine tonsils
  - Enlarged palatine tonsils diagnosed on physical exam
  - Imaging limited to lateral airway radiograph

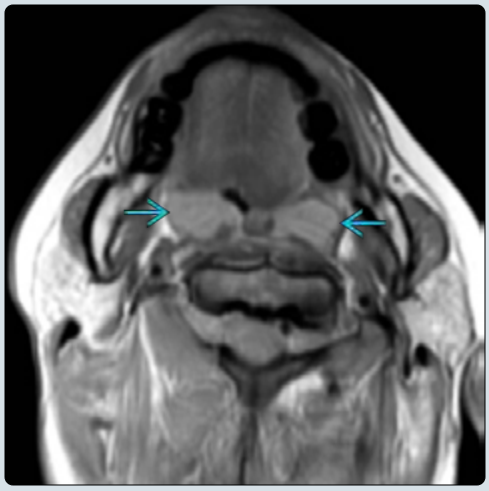
(Left) *Lateral airway radiograph in a 1-year-old child with drooling & decreased oral intake due to pharyngitis shows markedly enlarged palatine tonsils ⇨ outlined by air.* **(Right)** *Axial CECT in a 17-year-old girl with cervical adenopathy ⇨ due to Epstein-Barr virus (EBV) shows marked symmetric enlargement of homogeneously enhancing "kissing" palatine tonsils ⇨ that efface much of the oropharynx.*





(Left) *Axial STIR MR shows paired palatine tonsils ⇨ as symmetric, well-defined, high signal intensity structures in the palatine fossae.* **(Right)** *Axial PD MR in the same patient shows intermediate signal intensity of the palatine tonsils ⇨.*





TERMINOLOGY

Definitions

- Palatine tonsils: Paired, discrete, round to ovoid masses of lymphoid tissue on either side of oropharynx
- Enlargement may be chronic & idiopathic or acute/subacute (as from infection or neoplasm)
- Chronic enlargement may lead to obstructive sleep apnea (OSA)

IMAGING

General Features

- Size
  - Clinical evaluation based on percentage of oropharynx occupied by palatine tonsils
  - No clear size measurements established on imaging
  - Enlarged = high percentage of oropharynx occupied; intermittent obstruction on MR cine images

Radiographic Findings

- Lateral radiograph of soft tissue neck/airway demonstrates well-circumscribed soft tissue mass superimposed over mandibular angle

CT Findings

- CECT: Symmetric, homogeneously enhancing paired structures showing variable degrees of airway effacement
  - Asymmetry may be due to anatomic variation (most common), focal inflammation, or neoplastic infiltration
    - Heterogeneous enhancement may indicate underlying pathology (e.g., abscess, necrotic tumor)

Ultrasonographic Findings

- Transcutaneous approach more practical in child than transoral
- Palatine tonsil lies deep to submandibular gland when high frequency linear transducer positioned under mandible
  - Hypoechoic relative to submandibular gland
  - Tonsils show hypo- & hyperechoic striations normally
    - Striations maintained with uncomplicated tonsillitis
    - Pockets of ↓ echogenicity in tonsil due to edema, necrosis, or abscess
- Surrounding edema with peritonsillar cellulitis
- Hypoechoic avascular collection with surrounding hyperemia at lateral wall of tonsil suggests peritonsillar abscess

MR Findings

- T2 FS/STIR
  - Paired high signal, well-defined ovoid to round structures in palatine fossae
- MR cine during respiratory cycle
  - When significantly enlarged, round palatine tonsils will pulse inferomedially, collide, & intermittently obstruct airway

Imaging Recommendations

- Best imaging tool
  - Lateral airway radiograph in child with suspected upper airway pathology
  - CECT for acute clinical presentations suggesting drainable abscess
  - MR sleep studies typically reserved for
    - Recurrent OSA despite previous tonsillectomy & adenoidectomy or other airway surgery
      - Palatine tonsils not typically present
    - Syndromes with multilevel airway obstruction

DIFFERENTIAL DIAGNOSIS

Soft Tissue Mass in Oropharynx

- Foreign body
- Abscess
- Neoplasm
- Antrochoanal polyp
- Vascular malformation
- Dermoid
- Foregut duplication cyst

Common Causes of OSA

- Enlarged adenoid tonsils
- Enlarged lingual tonsils
- Glossoptosis
- Hypopharyngeal collapse
- Abnormal soft palate

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - OSA: Chronic snoring (± pauses, gasps), neurocognitive impairment, daytime sleepiness, pulmonary or systemic hypertension, failure to thrive
  - Neoplasm (lymphoma): Tonsillar asymmetry, dysphagia, mucosal color alteration, snoring, recurrent tonsillitis, cervical adenopathy
  - Tonsillitis/pharyngitis: Fever, odynophagia, tonsillar erythema & exudate, preceding viral prodrome, tender cervical adenopathy
- Clinical profile
  - Most children with OSA are otherwise healthy with enlarged adenoid & palatine tonsils
    - Enlarged palatine tonsils typically diagnosed clinically
    - Imaging typically limited to lateral neck radiograph
      - Evaluate for adenoid enlargement & nasopharyngeal obstruction

Treatment

- Depends on presentation & etiology
  - OSA requires tonsillectomy
  - Possible lymphoma requires biopsy & staging
  - Uncomplicated tonsillitis/pharyngitis treated with antibiotics; drainage for peritonsillar abscess

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Enlarged Lingual Tonsils

KEY FACTS

TERMINOLOGY

- Chronic idiopathic enlargement of lingual tonsils (lymphoid tissue at base of tongue) contributing to obstruction of retroglossal airway

IMAGING

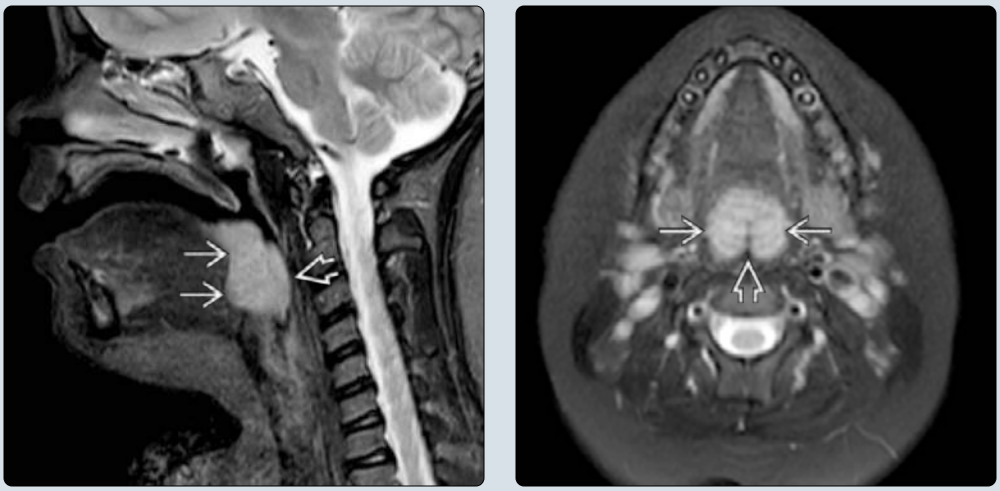
- AP & lateral radiographs initially obtained in child with suspected upper airway pathology
  - Lingual tonsils best seen on lateral view
    - Round or lobulated mass protruding posteriorly from tongue base into vallecula & oropharyngeal/hypopharyngeal airway
    - Markedly enlarged if AP diameter ≥ 10 mm
- MR sleep study for recurrent/complex OSA
  - High T2 FS/STIR signal round, lobulated, or dumbbell-shaped mass protruding posteriorly from tongue base & filling retroglossal airway
    - Normally separate left & right lingual tonsils

TOP DIFFERENTIAL DIAGNOSES

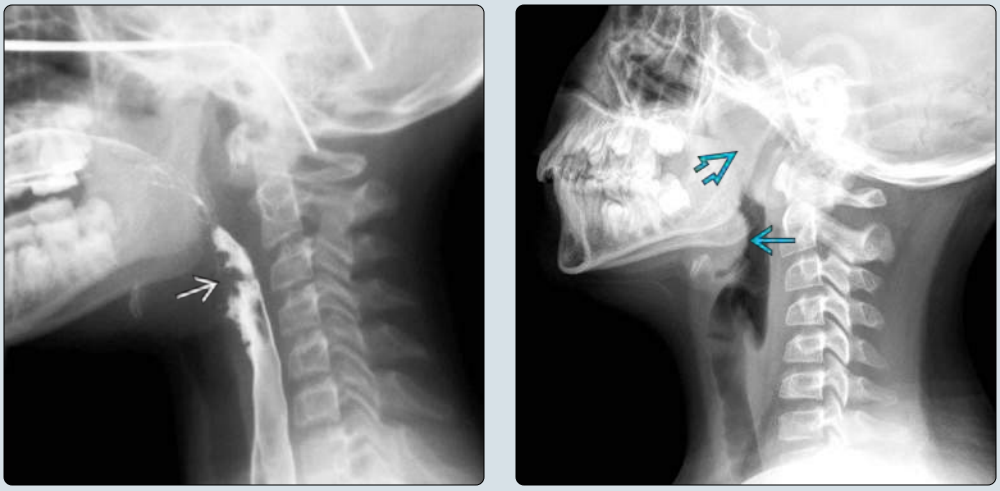
- Other causes of obstructive sleep apnea (OSA)
- Nonidiopathic causes of lingual tonsil hypertrophy
- Other vallecular masses

CLINICAL ISSUES

- OSA presentations: Snoring, poor rest, daytime fatigue
- Enlargement of lingual tonsils occurs most commonly
  - After previous palatine tonsillectomy & adenoidectomy
  - In children with Down syndrome (trisomy 21)
  - In obese children
  - In other systemic processes (e.g., EBV infection, lymphoma) causing enlargement of lymphoid tissue
- If lingual tonsils causative of recurrent OSA, lingual tonsillectomy usually curative



(Left) *Sagittal STIR MR in a child with Down syndrome & other causes of obstructive sleep apnea (OSA) shows enlarged lingual tonsils as a high-signal mass at the base of the tongue. Note the filling & obstruction of the retroglossal airway.* **(Right)** *Axial T2 FS MR in a child with Down syndrome & OSA shows enlarged lingual tonsils as a high-signal mass at the base of the tongue. The enlarged lingual tonsils fill much of the retroglossal airway, leaving only a tiny patent lumen.*



(Left) *Lateral esophagram in a child with Down syndrome shows a fungiform filling defect of the barium column at the posterior aspect of the tongue, consistent with enlarged lingual tonsils.* **(Right)** *Lateral airway radiograph in an 11 year old with snoring shows round, mildly lobulated soft tissue at the tongue base filling the vallecula, consistent with lingual tonsil enlargement. Note the concurrent enlargement of the adenoids.*

KEY FACTS

TERMINOLOGY

- Abnormal posterior motion of tongue during sleep leading to obstructive sleep apnea (OSA)
- Typically associated with underlying hypotonia, macroglossia, &/or micrognathia
- Glossoptosis very rare in otherwise healthy children

IMAGING

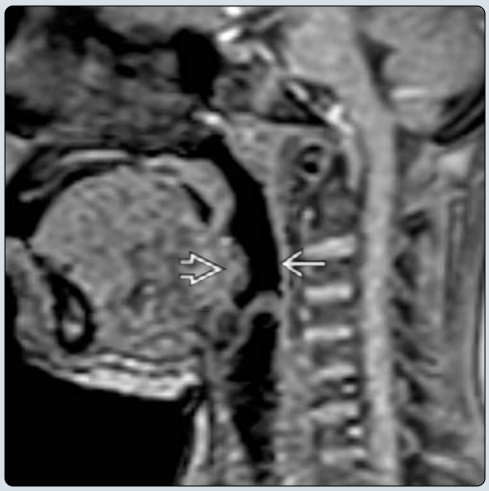
- MR sleep study evaluates airway motion & anatomic abnormalities during real-time respiratory cycle
- Indications for dynamic MR sleep imaging
  - Persistent OSA despite previous tonsillectomy & adenoidectomy (T&A) or other airway surgery
  - Complex OSA with predisposition to obstruction at multiple sites (e.g., Down syndrome)
  - Evaluation of OSA prior to any complex airway surgery
  - OSA & severe obesity
- Characteristic findings of glossoptosis
  - Tongue "falls" posteriorly → posterior border of tongue abuts posterior wall of pharynx → obstruction of retroglossal airway during inspiration
    - Can also abut & displace velum (soft palate) → obstruction of posterior nasopharynx
  - Stationary posterior & lateral walls of hypopharynx
    - In contrast to hypopharyngeal collapse
  - ± macroglossia & fatty infiltration of tongue

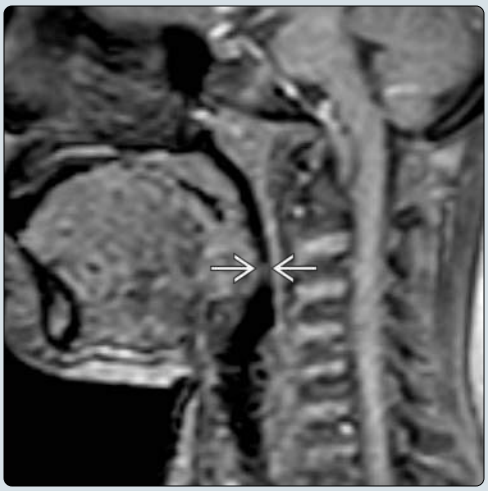
PATHOLOGY

- OSA difficult to manage in Down syndrome: Multiple anatomic abnormalities + ↓ muscle tone
- Findings in Down syndrome & persistent OSA after T&A
  - Glossoptosis (63%), recurrent & enlarged adenoid tonsils (63%), enlarged lingual tonsils (30%), macroglossia (74%)

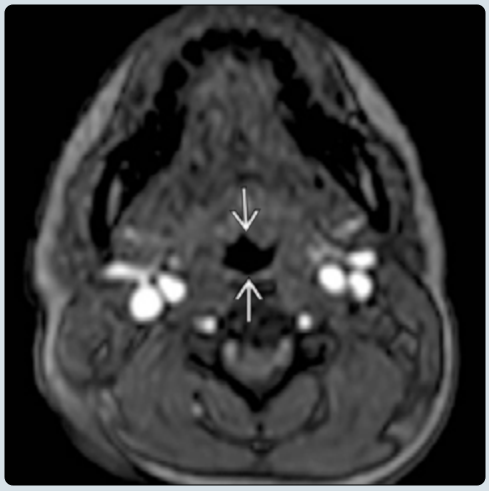
CLINICAL ISSUES

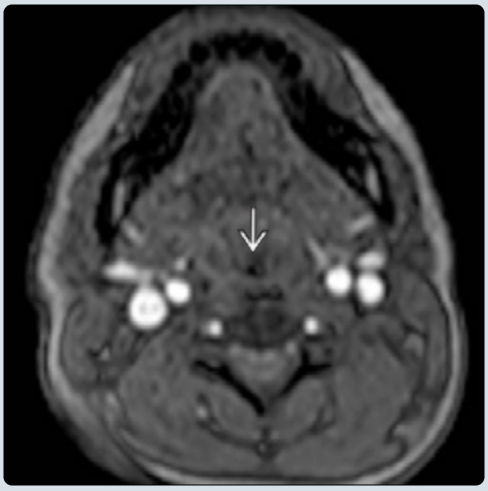
- Positive pressure ventilation (CPAP) treatment of choice
- Various procedures to ↓ tongue bulk &/or reposition tongue





(Left) *Sagittal cine MR in a child with Down syndrome obtained during the expiratory phase of the respiratory cycle shows the retroglossal airway ➡ to be patent despite the large posteriorly positioned tongue. The lingual tonsils ➡ are also enlarged.* **(Right)** *Sagittal cine MR in the same patient, obtained during inspiration, shows the posterior aspect of the tongue & lingual tonsils to have moved posteriorly, nearly obstructing the retroglossal airway ➡.*





(Left) *Axial cine MR obtained during expiration shows the retroglossal airway ➡ to be patent.* **(Right)** *Axial cine MR in the same patient, obtained during inspiration, shows the posterior aspect of the tongue & lingual tonsils to have moved posteriorly, nearly completely obstructing the retroglossal airway ➡.*

Double Aortic Arch

KEY FACTS

TERMINOLOGY

- Congenital aortic arch anomaly related to persistence of both left & right 4th aortic arches

IMAGING

- Chest radiography often suggestive of diagnosis
  - Bilateral tracheal indentations & mid tracheal narrowing
  - Right arch indentation commonly somewhat higher & more substantial than left ("right dominant")
- Esophagram suggestive with characteristic bilateral & posterior indentations
- Cross-sectional imaging (CTA/MR) for definitive diagnosis & characterization
  - Left & right arches arise from ascending aorta, encircle trachea & esophagus, & join to form descending aorta
  - Each arch gives rise to 1 ventral carotid & 1 dorsal subclavian artery (4 artery sign on axial slice)
  - Right arch commonly larger, more superior, & more posterior extending than left (70% of cases)
    - Left descending aorta in these cases

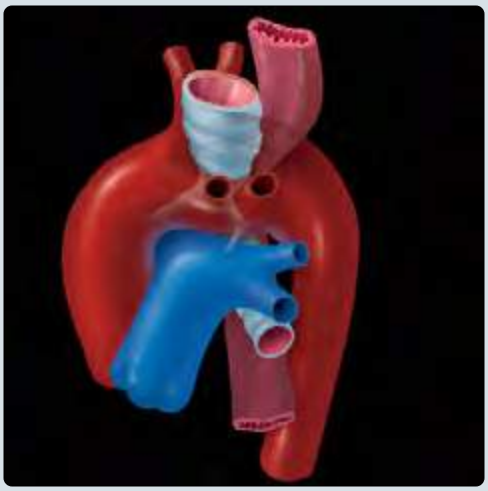
PATHOLOGY

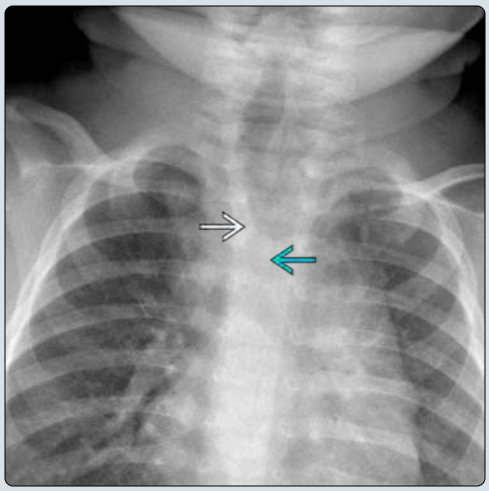
- Typically isolated; congenital intracardiac disease in 20%
- Dominant right arch, left descending aorta: 70-75%
- Dominant left arch, right descending aorta: 15-20%
- Arches equal in size: 5-10%
- Smaller of 2 arches may be partially atretic

CLINICAL ISSUES

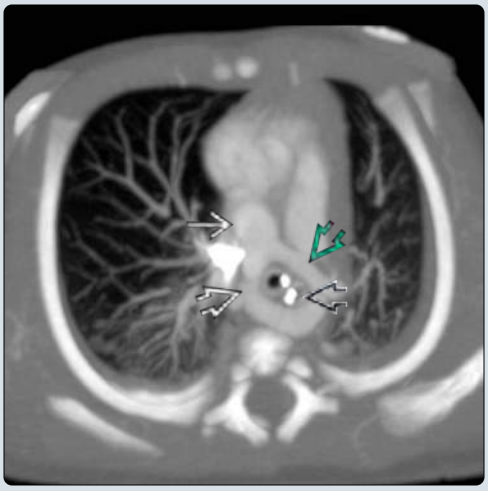
- Most common symptomatic vascular ring (55%)
- Often presents < 3 years of age with stridor, wheezing, & choking that worsens with feeding
- Surgery: Left (usually) thoracotomy with division of smaller arch, atretic segment, & ligamentum arteriosum with mobilization of trachea & esophagus
  - Up to 11% require 2nd operation to relieve persistent airway symptoms

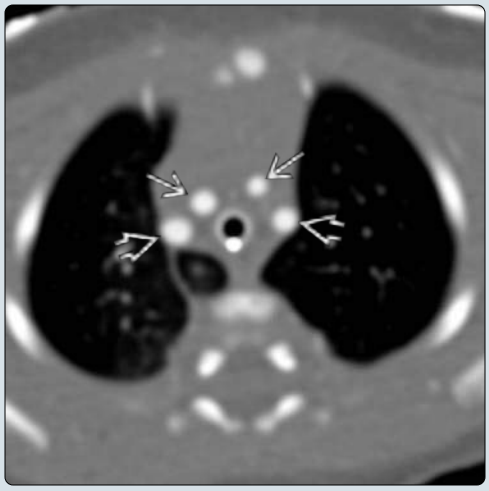
(Left) *Oblique graphic shows a double aortic arch anomaly with a complete vascular ring encircling & compressing the trachea & esophagus.* **(Right)** *AP radiograph of a 2 month old with biphasic stridor shows impressions on the trachea from both the right & left, with the right impression ➡ being higher than the left ⇨. Radiography can be highly suggestive of a double aortic arch, though the airway morphology is often overlooked.*





(Left) *Axial CTA MIP image shows the ascending aorta ➡ dividing into the right ➡ & left ⇨ arches, which extend along either side of the trachea. The right arch is dominant, as is typically seen. Both arches converge to form the descending aorta to the left of the thoracic spine. The trachea & esophagus ⇨ are both surrounded & compressed by the arches.* **(Right)** *Axial CTA just above both aortic arches shows separate ostia of both carotid ➡ & subclavian ➡ arteries (the 4 artery sign), typical of a double aortic arch.*





TERMINOLOGY

Abbreviations

- Double aortic arch (DAA)

Definitions

- Congenital aortic arch anomaly related to persistence of both left & right 4th aortic arches

IMAGING

General Features

- Best diagnostic clue
  - Radiography: Characteristic indentations of bilateral tracheal walls
  - Cross-sectional imaging: Ascending aorta splits anterior to trachea with confluence of discrete right & left arches posterior to esophagus
- Location
  - Right arch commonly more superior & posterior than left
  - Right arch typically runs behind esophagus to join left arch to form left-sided descending aorta
- Size
  - Right arch commonly larger than left
- Morphology
  - Part of smaller arch (usually left) may be atretic
    - Patent portions remain connected by fibrous band, completing compressive ring around trachea & esophagus

Radiographic Findings

- Chest radiography often suggestive of DAA
  - Symmetric mediastinal soft tissue on either side of trachea (or right > left)
    - Typically greater on left with normal left arch
  - Trachea deviated by dominant arch or in abnormal midline position
    - Normally, trachea slightly deviated to right by left arch
  - Bilateral tracheal indentations with mid tracheal narrowing in 47%
    - Right arch indentation commonly higher & larger than left
  - Lateral view shows generalized tracheal narrowing ± anterior indentation
  - Symmetric aeration of lungs (no unilateral air-trapping)

Fluoroscopic Findings

- Frontal view: Bilateral indentations on contrast-filled upper esophagus, often at different levels
- Lateral view: Rounded posterior indentation

CT Findings

- CTA
  - Ascending aorta divides anteriorly into discrete right & left arches → arches encircle trachea & esophagus → arches rejoin posteriorly to form descending aorta
    - Can cause severe mid tracheal compression
    - Smaller of 2 arches may be partially atretic
      - If atresia occurs between left carotid & subclavian arteries (instead of more common atresia beyond left subclavian artery), look for superior-pointing descending aortic diverticulum to confirm atresia
      - Distorted subclavian artery sign: Proximal subclavian ipsilateral to atresia shows abnormal inferior & posterior course
  - 4 artery sign: Symmetric take-off of 4 separate aortic branches on axial image at thoracic inlet
    - 2 ventral carotid & 2 dorsal subclavian arteries
    - 1 carotid & 1 subclavian arise from each arch

MR Findings

- Findings similar to those obtained with CTA
  - Lacks airway detail of CTA
- Can be depicted with variety of conventional, flow-related, & postcontrast techniques

Ultrasonographic Findings

- Prenatal diagnosis: Trident sign of 2 arches + ductus arteriosus on transverse 3-vessel-trachea view

Echocardiographic Findings

- Suprasternal notch view most helpful: 2 separate aortic arches, each giving rise to separate carotid & subclavian arteries (with no common brachiocephalic trunk)
- Does not adequately show airway & descending aorta
- Excludes intracardiac pathologies

Angiographic Findings

- Rarely required with use of cross-sectional imaging

Imaging Recommendations

- Best imaging tool
  - Radiography remains primary diagnostic test
    - Virtually all patients receive chest radiography 1st for respiratory symptoms
      - 25% can be normal in appearance
  - Esophagram rarely obviates need for cross-sectional imaging when arch anomaly suspected clinically & radiographically
    - However, many arch anomalies diagnosed 1st by oral contrast studies performed for feeding difficulties
  - Cross-sectional imaging (CTA or MRA) performed to confirm diagnosis & define anatomic variations for presurgical planning
- Protocol advice
  - Multidetector CTA faster to perform than MR
    - Generally no need for anesthesia/intubation
  - CT shows airway details somewhat better than MR
  - Multiplanar & volume-rendered images useful for characterization & communication with referring providers
    - Particularly helpful for understanding vascular ring relationship to airway

DIFFERENTIAL DIAGNOSIS

Right Arch With Aberrant Left Subclavian Artery

- Diverticulum of Kommerell points anteriorly toward pulmonary artery

Left Pulmonary Artery Sling

- Compression on anterior aspect of esophagus & posterior aspect of trachea on lateral radiograph or esophagram
- Often associated with tracheomalacia or complete cartilaginous tracheal rings (~ 2/3 of cases)

- Pulmonary sling only vascular ring to be associated with asymmetric aeration

Innominate Artery Compression Syndrome

- Compression on anterior aspect of trachea without esophageal compression

Nonvascular Masses

- Variety of benign & malignant solid or cystic soft tissue masses can compress & deviate trachea

PATHOLOGY

General Features

- Etiology
  - Related to embryological persistence of right & left 4th aortic arches
- Genetics
  - 22q11 deletion in ~ 25% of all arch anomalies without intracardiac disease
    - Partial arch atresia more likely in this setting than 2 patent arches
- Associated abnormalities
  - Typically isolated lesion without associated abnormalities
    - 20% have congenital heart disease (tetralogy of Fallot, ventricular septal defect, coarctation, patent ductus arteriosus, transposition of great arteries, truncus arteriosus)
  - Underlying tracheomalacia frequently present
- Pathophysiology
  - Variable airway & esophageal compression by vascular ring
    - Usually severe enough to present early in life
  - No hemodynamic sequelae unless associated with congenital heart disease

Gross Pathologic & Surgical Features

- True complete vascular ring with encircled trachea & esophagus
  - Dominant right arch, left descending aorta: 70-75%
  - Dominant left arch, right descending aorta: 15-20%
  - Arches equal in size: 5-10%
  - Smaller of 2 arches may be partially atretic

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 0-3 years old presenting with stridor, wheezing, & choking that worsens with feeding
  - Apnea or life-threatening respiratory arrest requiring resuscitation as initial presentation in 5-10%
- Other signs/symptoms
  - Apneic attacks, noisy breathing, "seal bark" cough
  - Feeding difficulty

Demographics

- Age
  - Typically presents with respiratory symptoms soon after birth
- Epidemiology
  - Most common symptomatic vascular ring anomaly (55%)

Treatment

- Left (usually) thoracotomy with division of smaller arch, atretic segment, & ligamentum arteriosum with mobilization of trachea & esophagus
  - Postoperative complications: Chylothorax, vocal cord paralysis, transient hypertension
  - Rare complication occurring pre- or postoperatively: Aortoesophageal fistula with severe hemorrhage
- Most common persistent symptom postoperatively: Stridor in 30%
  - Persistent airway symptoms most commonly due to
    - Tracheal stenosis
    - Tracheobronchomalacia
    - Persistent extrinsic airway compression
      - Caused by midline/circumflex descending aorta or previously ligated arch
- Up to 11% of patients require 2nd operation to relieve airway symptoms
  - Aortopexy or other vascular suspension procedures
  - Cartilaginous tracheal ring resection followed by airway reconstruction

DIAGNOSTIC CHECKLIST

Consider

- Look for atretic segment of double arch anomaly
  - Unopacified on CTA or MRA
  - No flow void on spin echo MR
- Smaller arch by cross-sectional imaging determines side of thoracotomy

Image Interpretation Pearls

- 4 artery sign on axial CTA/MR slice at thoracic inlet

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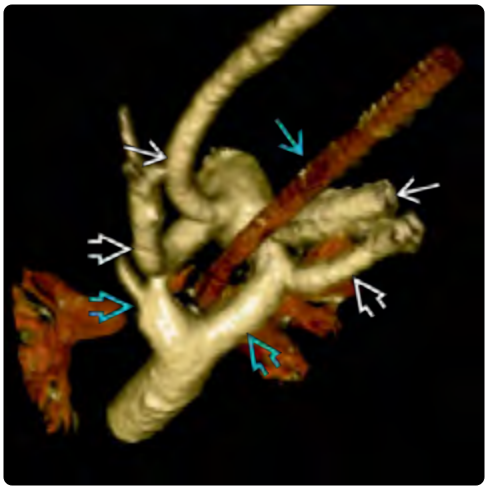
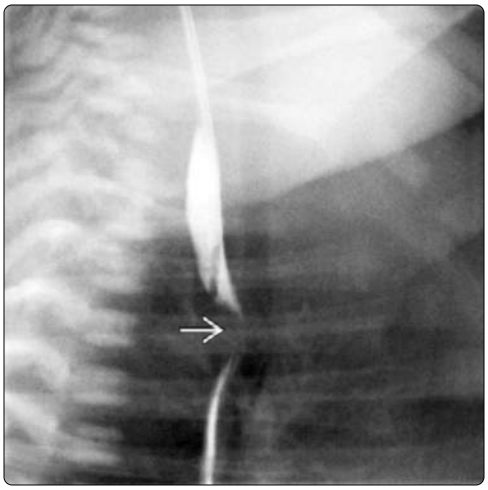
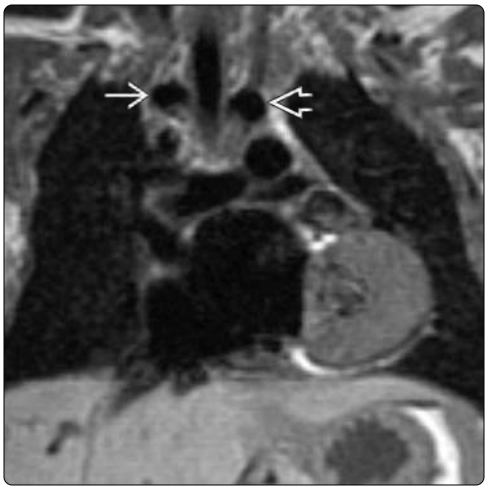
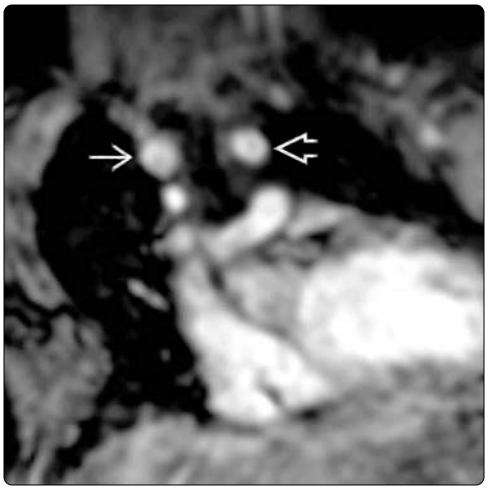


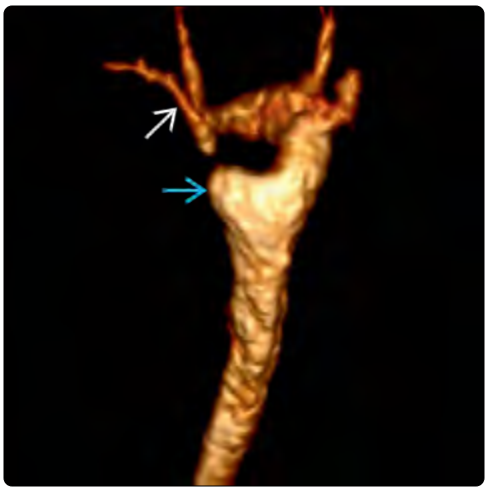


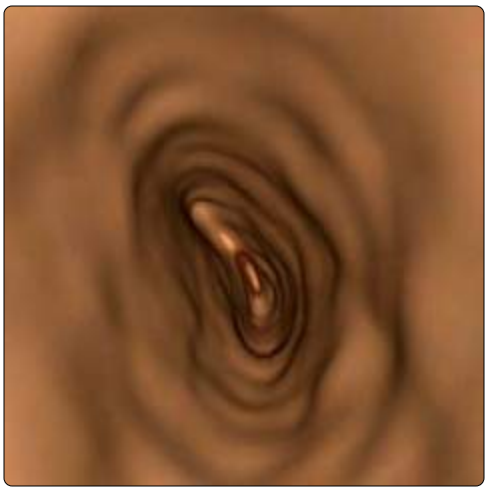
(Left) *Volume-rendered CTA of a double aortic arch & trachea viewed from a posterior & superior oblique perspective shows encirclement of the trachea ⇨ by bilateral aortic arches ⇨. Note the separate origins of the 4 aortic branches with 2 ventral carotid arteries ➡ & 2 dorsal subclavian arteries ➡.* **(Right)** *Lateral esophagram shows a round posterior indentation ➡ on the column of contrast in the esophagus, compatible with either a double aortic arch or an aberrant subclavian artery.*





(Left) *Coronal T1 MR shows symmetric right ➡ & left ➡ arch flow voids in a patient with a double aortic arch.* **(Right)** *Coronal MR cine velocity encoded phase contrast single image, in the same patient, shows a relatively symmetric appearance to the arches. Blood flow measurements showed the right arch ➡ to be dominant with 60% of flow vs. 40% in the left arch ⇨.*





(Left) *Posterior view of a CTA 3D reformation of a double aortic arch shows an atretic segment between the left subclavian artery ➡ & the descending aorta "diverticulum" ⇨ (which represents the distal left aortic arch stump). The atretic segment is a fibrous band that completes the vascular ring.* **(Right)** *Intraluminal superior to inferior view from a CT virtual bronchoscopy in a patient with a double aortic arch shows generalized tracheal narrowing.*

Pulmonary Sling

KEY FACTS

TERMINOLOGY

- Definition: Left branch pulmonary artery originates from posterior aspect of proximal right branch pulmonary artery & forms sling around right & posterior distal tracheal walls as it passes leftward between trachea & esophagus
- Synonym: Left pulmonary artery sling (LPAS)

IMAGING

- Essentially only vascular ring to course between trachea & esophagus
  - Compresses posterior trachea & anterior esophagus
  - Lateral chest radiograph: Round opacity between distal trachea & esophagus
  - Lateral esophagram: Anterior indentation on esophagus
- Multidetector CT angiography preferred over MR to confirm diagnosis & delineate anatomy prior to surgery
  - Rapid acquisition of CTA avoids intubation
  - Exquisite 3D reconstructions of airway & vessels

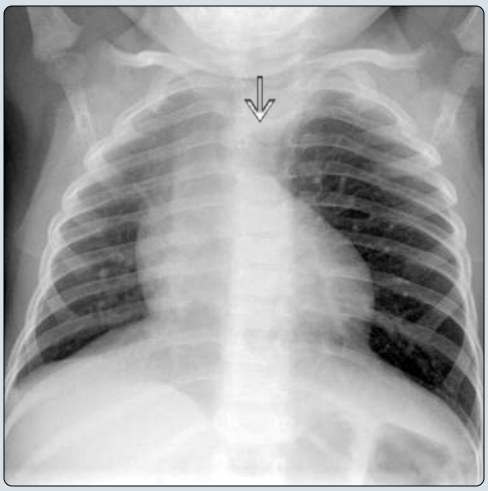
PATHOLOGY

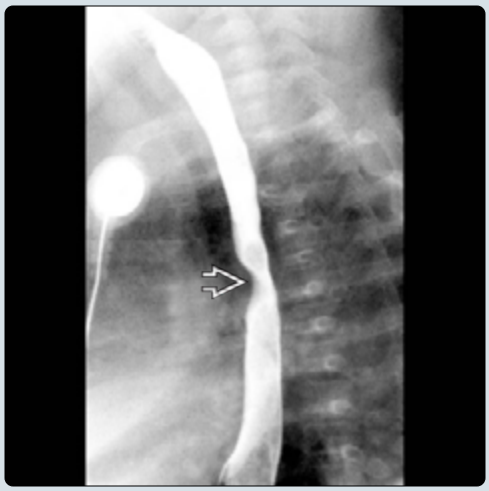
- Type I LPAS: Carina in normal location at T4-T5 with either normal eparterial bronchus (type IA) or tracheal bronchus to right upper lobe (type IB) → hyperinflation of right lung
- Type II LPAS: Low carina at T6 with diffuse stenosis of intermediate left bronchus by complete cartilaginous rings at multiple levels (ring-sling complex) → bilateral or left lung hyperinflation
- Many associated airway, pulmonary, & cardiac anomalies

CLINICAL ISSUES

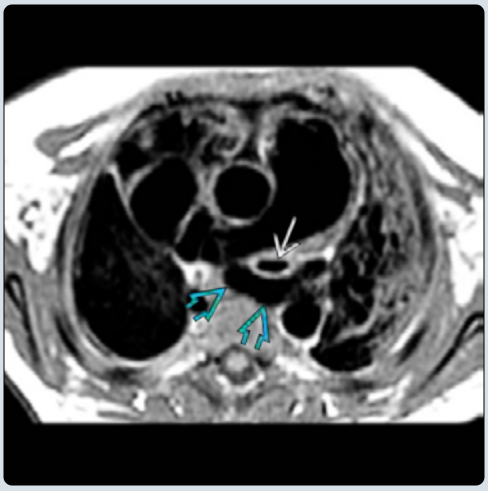
- Typically presents in neonatal period
  - Severe stridor, hypoxia, ventilator dependency
- Surgical repair
  - Division of LPA from its anomalous origin with implantation to its normal origin (main pulmonary artery)
  - Tracheobronchial reconstruction required for complete cartilaginous rings or other associated tracheobronchial malformations (types IIA & IIB)

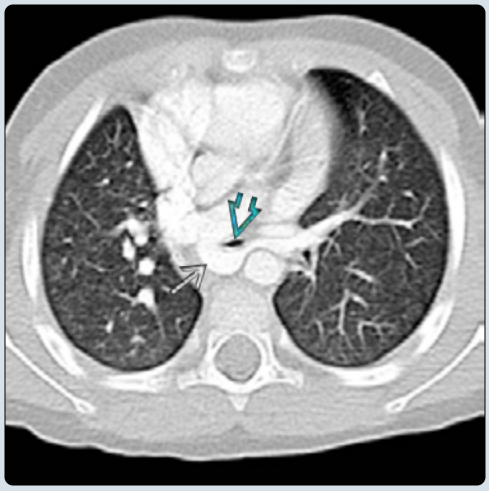
(Left) *AP radiograph in a patient with a left pulmonary artery sling (LPAS) shows asymmetric hyperinflation of the left lung with rightward mediastinal shift. There is an inapparent (poorly visualized) distal trachea.* **(Right)** *Lateral esophagram shows an anterior impression on the esophageal contrast column from the aberrant left pulmonary artery (sling), a classic finding on fluoroscopy.*





(Left) *Axial T1 MR shows a LPAS encircling & compressing the distal trachea.* **(Right)** *Axial CTA (in lung windows & algorithm) shows an aberrant left pulmonary artery coursing posterior to the airway. Note the distal tracheal narrowing in this child with associated complete tracheal rings. Also note the mild hyperinflation of the left lung compared to the right.*





TERMINOLOGY

Abbreviations

- Pulmonary artery sling (PAS)
- Left pulmonary artery sling (LPAS)

Synonyms

- Aberrant left pulmonary artery

Definitions

- Left pulmonary artery (LPA) originates from proximal right pulmonary artery (RPA) & wraps around right & posterior walls of distal trachea (forming sling) as it passes leftward between trachea & esophagus

IMAGING

General Features

- Best diagnostic clue
  - Classic imaging appearance: Asymmetric lung inflation, leftward displacement of narrowed distal trachea, & focal anterior impression on midesophagus
  - Only vascular ring to course between trachea & esophagus (indenting posterior tracheal & anterior esophageal margins)
- Morphology
  - Classified by associated abnormal bronchial branching patterns
    - Type I: Carina in normal location at T4-T5 with either normal eparterial bronchus (type IA) or tracheal bronchus to right upper lobe (type IB) → right lung hyperinflation
    - Type II: Low carina at T6 with diffuse stenosis of intermediate left bronchus (ILB) by complete cartilaginous rings at multiple levels (ring-sling complex) → bilateral or left lung hyperinflation
      - Type IIA: Initial bifurcation at T4 into right upper lobe bronchus & ILB which then bifurcates at T6 into bridging right lower lobe bronchus & left main bronchus
      - Type IIB: Absent or abortive right upper lobe bronchus

Radiographic Findings

- Radiography
  - Lateral view most helpful
    - Round opacity between distal trachea & esophagus
    - Posterior impression on distal trachea or carina
    - Distal trachea or right main bronchus bowed anteriorly
  - Frontal view
    - Low position of left hilum
    - Leftward displacement of narrowed distal trachea

Fluoroscopic Findings

- Esophagram: Only vascular ring causing anterior indentation on esophagus
  - Posterior tracheal indentation at same level

CT Findings

- CTA
  - LPA arises from posterior RPA & courses to left behind distal trachea, forming sling
    - LPA passes between trachea & esophagus
  - Tracheal stenosis or compression can be severe
    - Tracheal rings result in round narrow trachea on axial images

MR Findings

- Spin-echo T1WI shows dark tracheal & vascular lumens
- Contrast-enhanced angiographic techniques provide 3D rendering

Echocardiographic Findings

- Absence of normal pulmonary artery bifurcation
- Anomalous origin of left pulmonary artery from proximal posterior right pulmonary artery
- ± other associated cardiac anomalies

Imaging Recommendations

- Best imaging tool
  - Multidetector CT angiography preferred to confirm diagnosis & delineate anatomy prior to surgery (particularly in critically ill infants with tenuous airway)
    - Rapid acquisition avoids intubation & risks of airway manipulation
    - Exquisite resolution & detail obtained with 3D reconstructions of airway & vessels
      - 3D reconstruction of trachea delineates length, severity, & nature of tracheal narrowing
- Protocol advice
  - Optimize intravascular contrast bolus timing
  - Paired inspiratory/expiratory or dynamic images can demonstrate contribution of tracheobronchomalacia to airway narrowing

DIFFERENTIAL DIAGNOSIS

Middle Mediastinal Mass

- Lymphadenopathy
- Bronchogenic cyst
- Esophageal duplication cyst

Primary Bronchial Malformation

- Congenital lobar overinflation
- Bronchial atresia
- Tracheobronchomalacia
- Complete cartilaginous ring

Midline Descending Aorta-Carina Compression Syndrome

- Descending aorta immediately anterior to spine, leading to "crowding" of mediastinum
  - Posterior compression on carina or left main bronchus
- May be isolated or associated with right lung hypoplasia, arch anomalies

Right Arch With Aberrant Left Subclavian Artery

- Rarely courses anterior to esophagus
- More superior position that LPAS

PATHOLOGY

General Features

- Etiology
  - Embryology

  - Agenesis or obliteration of left 6th aortic arch, which normally forms LPA
  - Arterial supply of left lung via persistent primitive artery originating from RPA
- Pathophysiology
  - Severe stridor secondary to
    - Compression of distal trachea, carina, & main bronchi by sling, resulting in asymmetric inflation of lungs (obstructive hyperinflation > atelectasis)
    - Associated tracheobronchomalacia
    - Associated intrinsic airway narrowing (from complete cartilaginous rings)
  - Hemodynamics determined by associated cardiac anomalies
  - Pulmonary hypertension from severe hypoxia
- Genetics
  - No specific genetic defect identified
- Associated abnormalities
  - Intrinsic tracheobronchial anomalies: Complete rings (in 2/3), tracheomalacia, branching anomalies, tracheoesophageal fistula
    - Main bronchi have abnormal horizontal course (inverted T) with abnormal branching patterns to upper & lower lobes (types IIA & IIB)
  - Other congenital lung anomalies: Right lung hypogenesis or agenesis, horseshoe lung
  - Congenital heart disease: Aortic arch anomalies, ventricular septal defect (10%), atrial septal defect (20%), patent ductus arteriosus (25%), single ventricle, tetralogy of Fallot, partial anomalous pulmonary venous return, persistent left superior vena cava (20%)
  - Others: Imperforate anus, Hirschsprung disease, absent gallbladder, biliary atresia, Meckel diverticulum

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Severe stridor, hypoxia, ventilator dependency
- Other signs/symptoms
  - Noisy breathing, "seal bark" cough, apneic spells, recurrent pulmonary infections early in life

Demographics

- Age
  - Typically presents in neonatal period

Natural History & Prognosis

- Type II less favorable than other vascular rings due to associated anomalies (60-80%)

Treatment

- Surgical division of LPA from its anomalous origin with implantation to its normal origin (main pulmonary artery)
- Tracheobronchial reconstruction if complete cartilaginous rings or other associated tracheobronchial malformations present (types IIA & IIB)

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Anterior indentation on esophagus = LPAS
  - Essentially only vascular ring to course between trachea & esophagus
- Round small caliber distal trachea on axial images = complete cartilaginous tracheal rings

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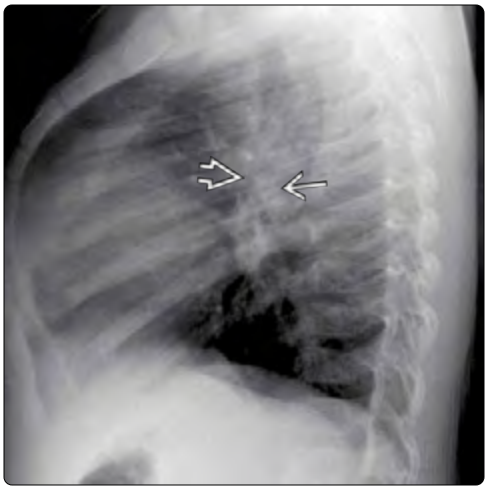
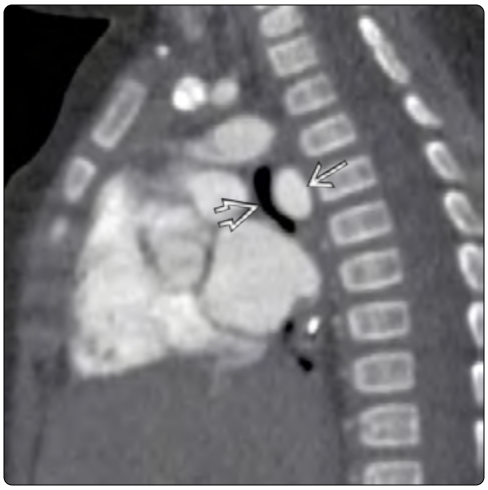
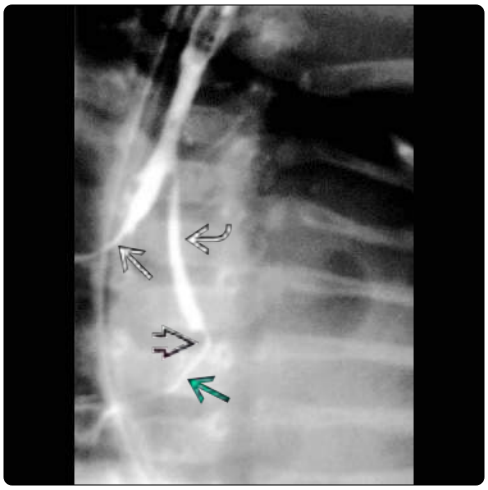
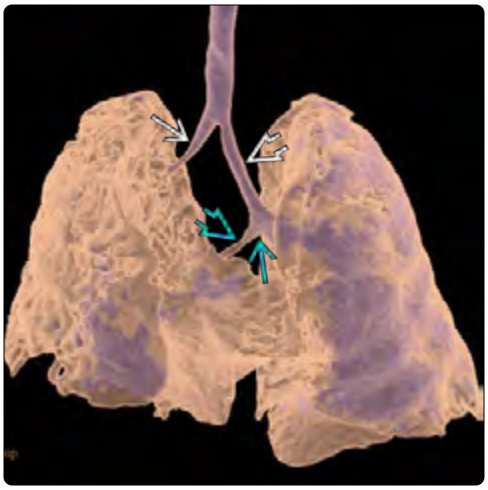


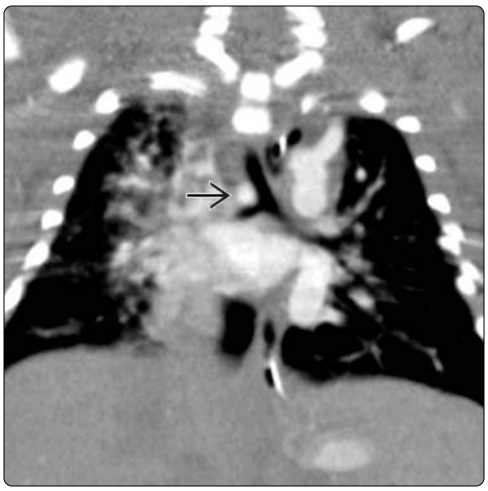


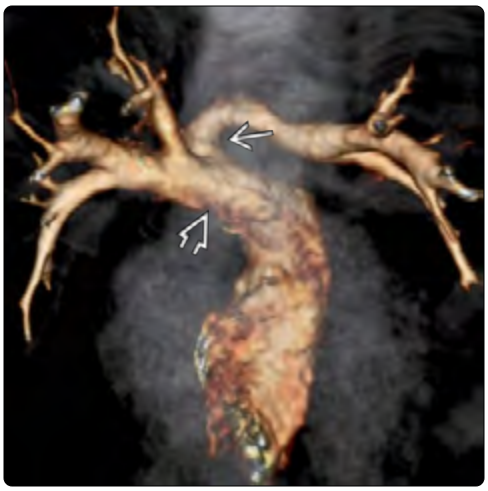
(Left) *Lateral radiograph in a child with a LPAS shows a round opacity ➡ that is directly posterior to the distal trachea & corresponds to an aberrant left pulmonary artery that narrows & anteriorly bows the trachea ➡. Note the bowing of the sternum & flattening of the hemidiaphragms, suggesting bilateral hyperinflation.* **(Right)** *Sagittal CECT in an infant with a LPAS shows the aberrant left pulmonary artery ➡ directly posterior to the narrowed distal trachea (which is bowed anteriorly ➡).*





(Left) *Bronchogram shows the LPAS type IIA bronchial branching pattern with a high bifurcation into the right upper lobe ➡ & a diffusely narrowed intermediate left bronchus ➡. Distally, there is a low LPAS indentation ➡ & a bridging right bronchus ➡.* **(Right)** *In this volume-rendered anterior view CT image of an infant with type IIA LPAS, note the right upper lobe tracheal bronchus ➡, intermediate left bronchus ➡, low carina ➡, & bridging right bronchus ➡.*





(Left) *Coronal CTA shows an aberrant left pulmonary artery (LPA) ➡ that courses over the right main bronchus with the carina at T4, compatible with a type IA pulmonary artery sling.* **(Right)** *CTA 3D volume rendering shows the aberrant LPA ➡ arising from the posterior aspect of the right pulmonary artery (RPA) ➡.*

Innominate Artery Compression Syndrome

KEY FACTS

TERMINOLOGY

- Symptomatic tracheal compression secondary to innominate artery traversing anterior to trachea

IMAGING

- Anterior impression on trachea from innominate artery → tracheal narrowing in AP dimension on any imaging study
  - Tracheal narrowing confined to level at which innominate artery crosses anterior to trachea
- Detect tracheomalacia (excessive dynamic airway collapse) at level of innominate artery crossing by dynamic imaging on CT or MR
  - > 50-75% tracheal narrowing on dynamic imaging in symptomatic patient

TOP DIFFERENTIAL DIAGNOSES

- Mediastinal mass lesion causing tracheal compression
- Aberrant thyroid or ectopic thymus
- Vascular anomalies such as vascular ring

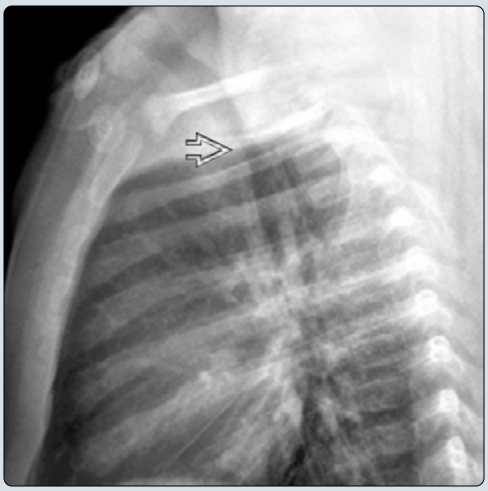
PATHOLOGY

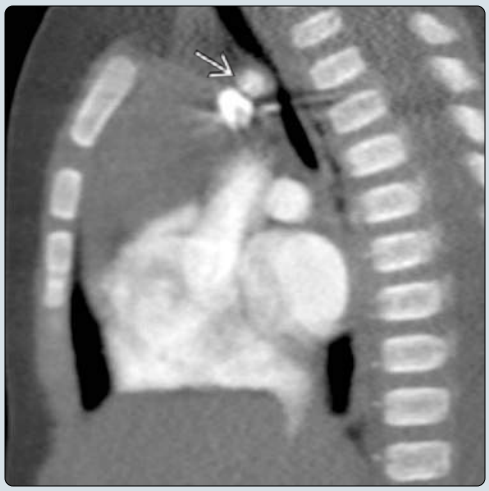
- Infantile trachea relatively flaccid, lacking rigidity
- Normal innominate artery arises from left of trachea & courses across it anteriorly
  - Anomalous innominate artery syndrome is misnomer

CLINICAL ISSUES

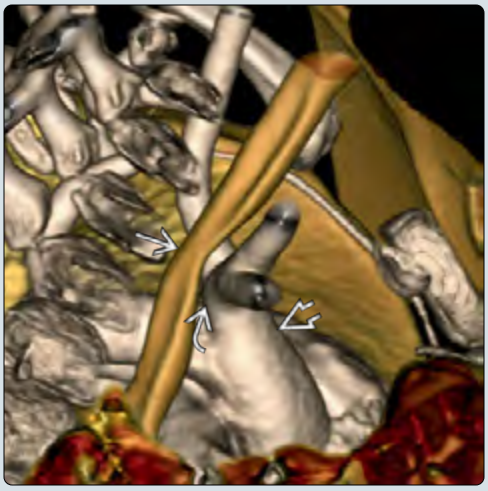
- Presentations include stridor, apnea, dyspnea
  - History of recurrent bronchopulmonary infections in some patients with innominate artery compression syndrome
- Many asymptomatic children will have mild anterior compression (normal variant) on lateral radiographs & cross-sectional imaging studies
- Compression & resultant symptoms may ↓ over time as child grows

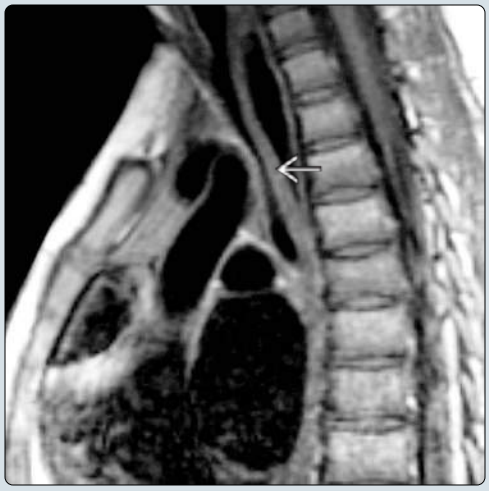
(Left) *Lateral chest radiograph shows significant narrowing of the trachea by an anterior impression just below the level of the sternal notch, compatible with innominate artery compression syndrome.* **(Right)** *Sagittal CTA in the same patient shows the relationship of the innominate artery to the trachea as it creates the anterior impression that was also seen on the lateral radiograph of the chest.*





(Left) *Right lateral view of a vessel & airway 3D CTA reconstruction in a 7-month-old child demonstrates the aortic arch with the innominate artery crossing anterior to the trachea & causing compression.* **(Right)** *Sagittal T1 MR shows the innominate artery immediately anterior to the trachea & causing discrete anterior tracheal compression as it ascends rightward & superiorly.*





TERMINOLOGY

Synonyms

- Anomalous innominate artery

Definitions

- Symptomatic tracheal compression secondary to innominate artery crossing anterior to trachea
  - Related to lack of rigidity in infantile trachea

IMAGING

General Features

- Best diagnostic clue
  - Anterior impression on trachea from innominate artery on any imaging study, typically at thoracic inlet

Radiographic Findings

- Radiography
  - On lateral radiography, anterior tracheal impression may be present with lack of air-trapping on frontal projection

CT Findings

- CECT
  - Tracheal narrowing confined to level of innominate artery crossing
  - Can evaluate other etiologies in patients with tracheal compression on bronchoscopy
- CTA
  - 3D post processing shows relationship of vessels to trachea, along with virtual bronchoscopy views
  - Dynamic imaging can be performed to detect excessive dynamic airway narrowing (> 50-75% tracheal compression when compared to normal trachea)

MR Findings

- Tracheal narrowing as innominate artery crosses anteriorly

Other Modality Findings

- Endoscopy shows fixed, pulsatile compression from anterior aspect of trachea in most cases

Imaging Recommendations

- Best imaging tool
  - CT/MR defines degree of tracheal compression by innominate artery & excludes other causes
- Protocol advice
  - Endotracheal tube or positive pressure respiratory support can stent trachea & mask obstruction
  - CT evaluates vessels & relationships to airway; performance of dynamic imaging is plus
  - MR demonstrates anatomy on axial/sagittal imaging; dynamic airway imaging provides information on tracheomalacia

DIFFERENTIAL DIAGNOSIS

Masses Compressing Trachea

- Mediastinal bronchogenic cyst, duplication cyst, neurofibroma, lymphoma, thymic cyst, & teratoma

Normal Structures in Atypical Position

- Aberrant thyroid or ectopic thymic tissue

Vascular Anomalies

- Right aortic arch + aberrant left subclavian artery, double aortic arch, & aneurysmal dilation of aorta or innominate artery

Lymphatic Malformation

- Mediastinal extentension causing airway compression/infiltration

PATHOLOGY

General Features

- Etiology
  - Narrowing of trachea at thoracic inlet due to compression by innominate artery
    - Infantile trachea relatively flaccid; → crossing innominate artery accentuates tracheal narrowing

Gross Pathologic & Surgical Features

- Anterior wall of trachea demonstrates abnormal cartilage

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Presentations include stridor, apnea, dyspnea

Natural History & Prognosis

- Symptoms may ↓ over time as child grows

Treatment

- Most children treated conservatively & outgrow disease
- Surgical approach is adopted in some
  - With repeated hospitalizations due to respiratory infections
  - Symptomatic patients who fail conservative management
  - Aortopexy or reimplantation of innominate artery origin

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Tracheal compression caused by innominate artery traversing anterior to trachea at thoracic inlet
- Typically detected on cross-sectional imaging either incidentally or in patients with symptoms
- Associated tracheomalacia at site of crossing innominate artery can be elicited on dynamic airway imaging by CT or MR

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Right Arch With Aberrant Left Subclavian Artery

KEY FACTS

IMAGING

- Right-sided aortic arch (RAA) with aberrant origin of left subclavian artery (ALSCA) from Kommerell diverticulum
- Lateral radiograph: Anterior tracheal bowing due to aberrant retroesophageal vessel
- Barium esophagram: Posterior indentation by aberrant vessel
  - Large posterior indentation: Diverticulum of Kommerell
- CTA or MRA with 3D reconstructions now preferred modality for diagnosis prior to surgical intervention
  - Dynamic airway imaging can evaluate tracheomalacia at level of vascular ring

PATHOLOGY

- Related to embryologic persistence of right 4th aortic arch
- Left ductus persists as ligamentum arteriosum, which completes vascular ring
- ALSCA is typically retroesophageal; rarely it may lie anterior to esophagus (15%) or anterior to trachea (5%)

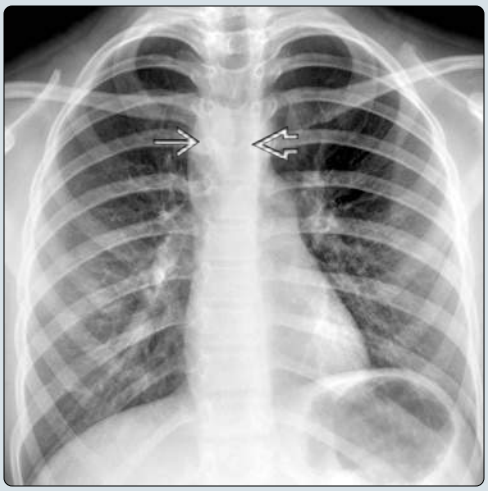
CLINICAL ISSUES

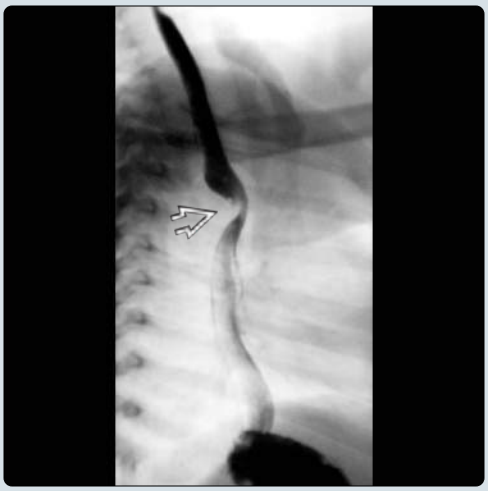
- Child presents with symptoms of stridor, apnea, cyanosis, recurrent respiratory infection, or chronic cough
  - Often referred for chest radiograph & ultimately cross-sectional imaging

DIAGNOSTIC CHECKLIST

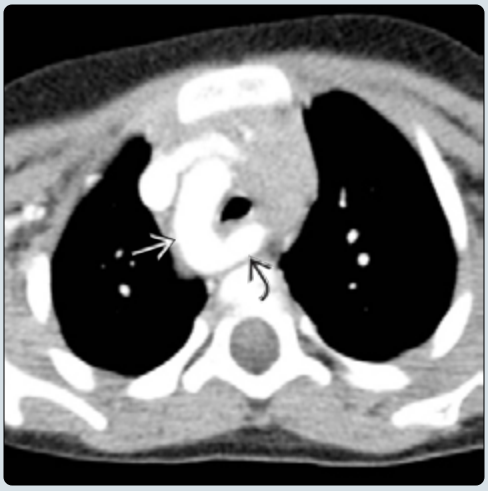
- RAA on radiograph in symptomatic child & absence of known underlying congenital heart disease (CHD) → perform further cross-sectional imaging
- RAA with airway compression & ALSCA on esophagram → perform cross-sectional imaging
- RAA, mirror image branching pattern → evaluate for underlying CHD

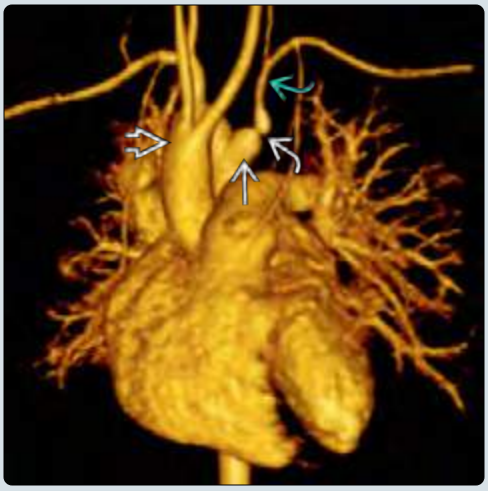
(Left) *PA radiograph of the chest shows a right aortic arch ➡ causing an impression on the right side of the trachea & displacing the trachea to the left ➡. This is an initial clue to diagnosing a right aortic arch with aberrant left subclavian artery (RAA-ALSCA) & is often overlooked.* **(Right)** *Lateral esophagram in an infant shows a large posterior indentation ➡ on the esophagus due to a RAA-ALSCA with a diverticulum of Kommerell.*





(Left) *Axial CECT shows a RAA ➡ with an ALSCA ⇨ arising from the dorsal aorta & coursing posterior to the trachea. This RAA, in concert with a tight left ligamentum arteriosum (which is usually not visible), creates a complete vascular ring.* **(Right)** *3D volume-rendered CTA (anterior projection) shows a RAA ➡ with an ALSCA ⇨ originating from a diverticulum of Kommerell ➡. There is stenosis ⇨ at the origin of the ALSCA from the diverticulum.*





TERMINOLOGY

Definitions

- Aortic arch lies to right of trachea [right aortic arch (RAA)] & aberrant left subclavian artery (ALSCA) originates from proximal descending aorta to course behind esophagus

IMAGING

Radiographic Findings

- Frontal view: Round soft tissue density along right side of trachea resulting in focal right-sided indentation & leftward tracheal deviation
- Lateral view: Anterior bowing of trachea

Fluoroscopic Findings

- Barium esophagram
  - Frontal view: Oblique filling defect at proximal 1/3 of esophagus coursing from right inferior to left superior
  - Lateral view: Large posterior indentation of esophagus caused by diverticulum of Kommerell

CT Findings

- Right-sided aortic arch with retroesophageal ALSCA arising from bulbous diverticulum of Kommerell from proximal descending thoracic aorta
- Mass effect & airway narrowing at level of vascular ring (caused by left-sided ligamentum arteriosum)
- Reformatted 2D planes & 3D reconstructions nicely demonstrate effect on airway

MR Findings

- Black blood, white blood, or postcontrast MRA show abnormal branching pattern of vessels
- Black blood images show effect on tracheobronchial tree

Echocardiographic Findings

- Defines RAA & branching pattern along with origin of ALSCA

Imaging Recommendations

- Best imaging tool
  - CTA with 3D reconstruction
    - Advantages: Fast, generally no need for sedation/anesthesia, dynamic airway imaging for concurrent tracheobronchomalacia
    - Disadvantages: Radiation dose, use of IV iodinated contrast, & correct timing of contrast bolus
  - MRA
    - Advantages: No radiation, multiplanar technique
    - Disadvantages: May need sedation, need IV gadolinium contrast, & long exam time

DIFFERENTIAL DIAGNOSIS

Right Aortic Arch With Mirror Image Branching

- High association with cyanotic congenital heart disease (CHD), such as tetralogy of Fallot & truncus arteriosus

Double Aortic Arch With Atretic Left Arch

- Left arch often atretic; left ligamentum arteriosum along with dominant right arch completes vascular ring
- Inferior tenting of left common carotid artery & 4-pronged branching pattern of arches at thoracic inlet

Left Aortic Arch With Aberrant Right Subclavian Artery

- Typically isolated & incidental abnormality (without airway compression)

PATHOLOGY

General Features

- Genetics
  - Right arch often associated with 22q11 deletion
- Embryology/anatomy
  - Related to embryologic persistence of right 4th aortic arch
  - Retroesophageal (Kommerell) diverticulum: Remnant of embryonic left 4th dorsal aortic arch; connects to left ductus ligament
  - Right ductus obliterated & left ductus persists as ligamentum arteriosum, which completes vascular ring

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Stridor, apnea, cyanosis, recurrent respiratory infection, chronic cough, & rarely dysphagia (lusoria) due to posterior esophageal compression
  - May be incidental finding on esophagram

Demographics

- Epidemiology
  - Aberrant subclavian artery most common congenital anomaly of aortic arch
    - RAA-ALSCA: 0.1% of asymptomatic population
    - LAA-ARSCA: 0.5% of asymptomatic population

Natural History & Prognosis

- Prognosis generally good after division of vascular ring
- Symptoms may persist from tracheomalacia & residual stenosis after vascular ring repair

Treatment

- RAA-ALSCA with constricting (symptomatic) left ligamentum arteriosum: Division of ligamentum via left thoracotomy
  - ± aortopexy if associated with midline descending aorta

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Infantile Hemangioma, Airway

KEY FACTS

TERMINOLOGY

- Benign vascular neoplasm of capillaries in infant
- Arises in many locations; airway occurrence potentially life threatening

IMAGING

- Asymmetric subglottic tracheal narrowing in young child
  - May be transglottic, rarely distal
- Avidly enhancing submucosal mass on CT/MR
  - Unilateral > bilateral or circumferential

TOP DIFFERENTIAL DIAGNOSES

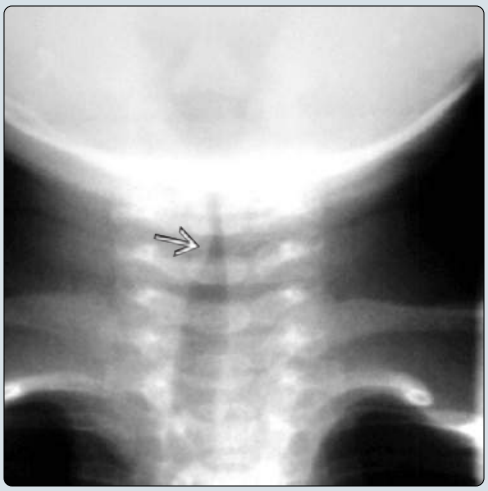
- Congenital subglottic tracheal stenosis
- Iatrogenic subglottic tracheal stenosis
- Croup
- Tracheomalacia
- Exudative tracheitis
- Papillomatosis

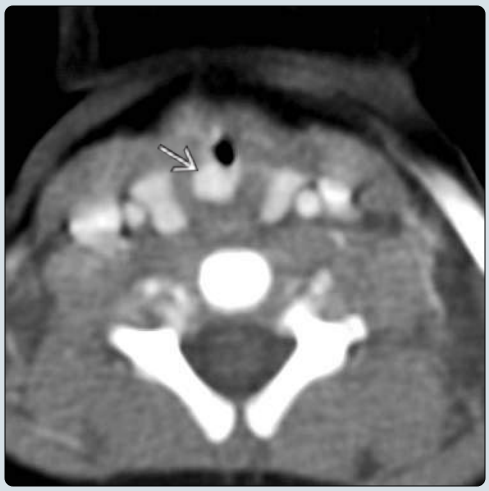
PATHOLOGY

- Predictable life cycle of infantile hemangioma
  - Proliferative phase: Appears days-weeks after birth & grows rapidly for ~ 4-12 months
  - Involuting phase: Slow gradual regression over next several years
  - Involuted phase: Static minimal residua of lesion
- Tissue stains positive for GLUT1 in all phases
- Often occurs with PHACES association

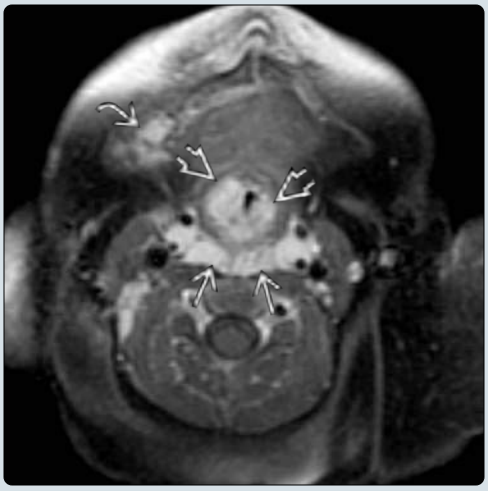
CLINICAL ISSUES

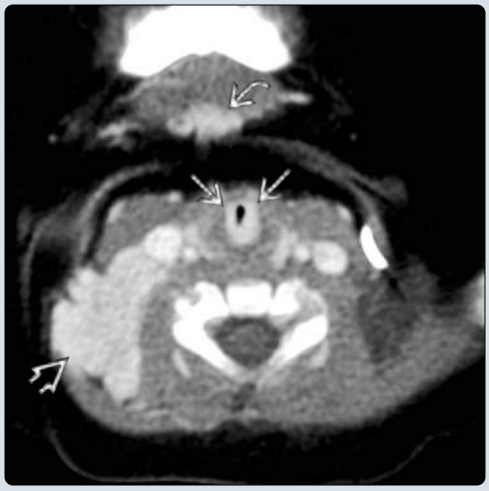
- Inspiratory stridor in infants < 6 months of age
  - Most lesions progressively obstruct airway during proliferative phase, requiring treatment
- Cutaneous hemangiomas in 50%
  - Classically in "beard distribution"
- Treatment
  - Most common: Conservative monitoring or propranolol
  - Others: Corticosteroids, laser therapy, surgical excision





(Left) *AP radiograph shows asymmetric subglottic tracheal narrowing in a 2-week-old infant presenting with stridor. Asymmetric narrowing is always concerning for a hemangioma, as opposed to symmetric subglottic narrowing that is typical of croup.* **(Right)** *Axial CECT in the same patient shows a well-defined, avidly enhancing infantile hemangioma along the dorsolateral aspect of the subglottic airway.*





(Left) *Axial T1 C+ FS MR in a child with PHACES association demonstrates multiple enhancing infantile hemangiomas in the retropharyngeal space, surrounding the subglottic trachea, & involving the right submental space.* **(Right)** *Axial CECT in an 11-week-old girl demonstrates a circumferential, well-defined subglottic infantile hemangioma. Note the additional posterior cervical space & submental infantile hemangiomas, which should raise suspicion for PHACES association.*

TERMINOLOGY

Definitions

- Benign vascular neoplasm of capillaries in infant
- Arises in many locations; airway occurrence potentially life threatening

IMAGING

General Features

- Best diagnostic clue
  - Asymmetric subglottic narrowing in young child
  - Avidly enhancing submucosal mass on CT/MR
- Location
  - Subglottic > > transglottic > distal trachea

Radiographic Findings

- Asymmetric subglottic tracheal narrowing on AP radiograph

CT Findings

- Usually solitary, avidly enhancing lobular subglottic mass

MR Findings

- Intermediate T1, high T2, avid enhancement

Imaging Recommendations

- Best imaging tool
  - CECT helps define mass & tracheal effacement
    - Adjust CT technique for pediatric patient
  - MR more sensitive but sedation usually needed

DIFFERENTIAL DIAGNOSIS

Congenital Subglottic Tracheal Stenosis

- Symmetric circumferential tracheal narrowing
  - Complete tracheal rings: Small, round trachea on axial images
  - ± pulmonary sling or congenital heart disease

Iatrogenic Subglottic Tracheal Stenosis

- Prior history of intubation or tracheostomy

Croup

- Acute symmetric subglottic tracheal narrowing, typically of viral etiology
- Characteristic "barky" or "seal-like" cough
- Most common in children 6 months to 3 years of age

Tracheomalacia

- Abnormal dynamic collapse of intrathoracic trachea

Exudative Tracheitis

- Intraluminal filling defects/inflammatory exudates
- Children usually older than those with viral croup

Airway Papillomatosis

- Nodules of larynx > trachea & bronchi > lungs

PATHOLOGY

General Features

- Etiology
  - Benign vascular neoplasm (not malformation) of proliferating endothelial cells
- Associated abnormalities
  - Cutaneous infantile hemangiomas in 50%, classically in "beard distribution"
  - **PHACES** association: **P**osterior fossa brain malformations, **h**emangiomas of face, **a**rterial anomalies, **c**ardiac anomalies, **e**ye abnormalities, **s**ternal clefts or **s**upraumbilical raphe
    - 7% have subglottic hemangiomas

Staging, Grading, & Classification

- Mulliken & Glowacki classification of vascular anomalies: Neoplasms vs. malformations
  - Reflects cellular kinetics & clinical behavior

Microscopic Features

- GLUT1 immunohistochemical marker: Positive in all phases of infantile hemangioma
  - Not positive in other less common hemangiomas

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Inspiratory stridor in infant < 6 months of age
- Other signs/symptoms
  - Cutaneous hemangiomas in 50%
  - Hoarseness or abnormal cry

Natural History & Prognosis

- Benign condition but may have morbidity/mortality
  - Complications: Bleeding, ulceration, airway obstruction
    - Not coagulopathy
- Predictable life cycle of all infantile hemangiomas
  - Proliferative phase: Lesion appears days-weeks after birth & grows rapidly for ~ 4-12 months
    - Majority have progressive airway obstruction during proliferation, requiring treatment
  - Involuting phase: Slow gradual regression of lesion over next several years
    - Symptoms resolve with involution
  - Involuted phase: Static through remaining life with minimal residual lesion
- Diagnosis made at endoscopy

Treatment

- Conservative monitoring in limited circumstances
- Propranolol: 1st-line treatment in recent years due to rapid response & low rate of side effects
- Systemic corticosteroids: Prior 1st-line treatment
- "Laser" therapy
- Laryngotracheoplasty: Direct excision of small masses, usually not required with current medical therapies
- Combination of therapies used in 75% of children

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Tracheobronchomalacia

KEY FACTS

TERMINOLOGY

- Excessive expiratory collapse of trachea &/or bronchi
  - May be purely intrinsic or in association with longstanding extrinsic compression

IMAGING

- Dynamic 4D MDCT (real-time model of airway during respiration) vs. static inspiratory & expiratory CT scans
  - Multiplanar reconstructions & 3D volume rendering of airway helpful
  - IV contrast useful to look for contributory adjacent mass or aberrant vessel
- > 50% ↓ in cross-sectional area of tracheal &/or bronchial lumen during expiration or coughing
  - Correlates well with bronchoscopy
  - Relatively underdiagnosed as imaging is routinely performed only at end inspiration
- ↑ frequency & severity of air trapping

TOP DIFFERENTIAL DIAGNOSES

- Difficult to control asthma
- Foreign body aspiration
- Extrinsic compression
- Complete tracheal rings

PATHOLOGY

- Primary (congenital): Impaired cartilage maturation
- Secondary (acquired): Otherwise normal cartilage degenerates following infection, chronic inflammation, intubation, trauma, or longstanding extrinsic compression

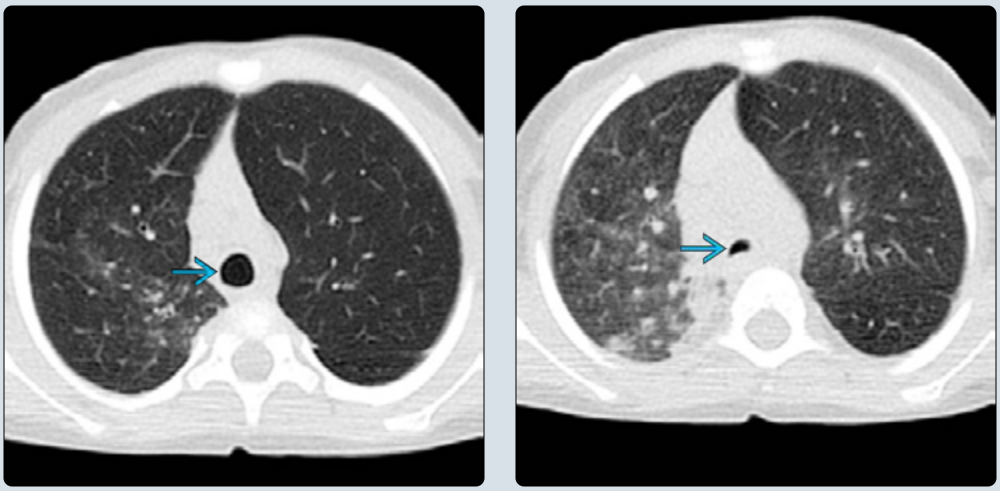
CLINICAL ISSUES

- Presentations: Expiratory stridor, dyspnea, cough, sputum production, acute life threatening events (ALTEs)

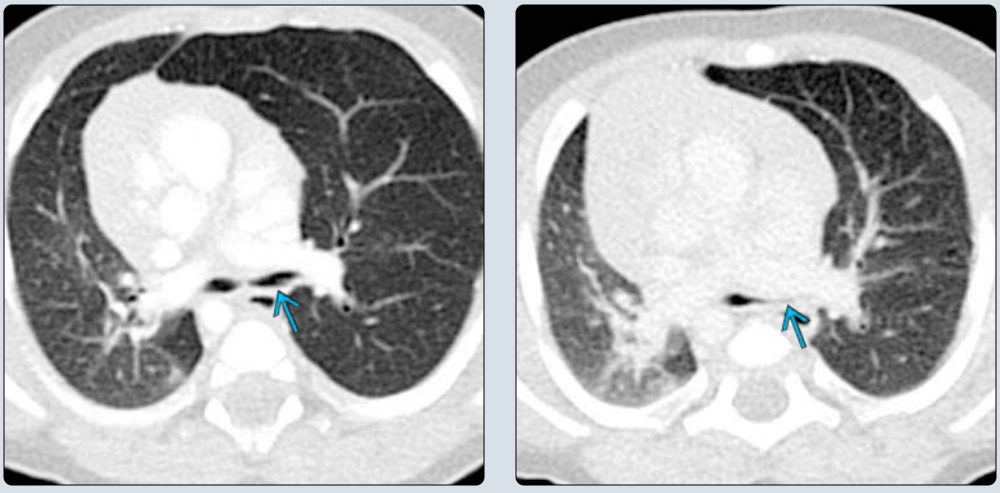
DIAGNOSTIC CHECKLIST

- Expiratory phase of imaging necessary when evaluating for tracheobronchomalacia

(Left) *Inspiratory axial HRCT shows a normal, nearly rounded appearance of the trachea. The posterior aspect is often nearly flat as it lacks supportive cartilage. The caliber of the trachea should be uniform from the glottis to the carina & relatively unchanging between inspiration & expiration.* **(Right)** *Expiratory axial HRCT in the same patient shows marked narrowing of the trachea, a substantial change from the inspiratory image that is consistent with tracheomalacia.*



(Left) *Inspiratory axial HRCT with contrast shows a patent left main bronchus.* **(Right)** *Expiratory axial HRCT in the same patient shows interval collapse of the left main bronchus, consistent with bronchomalacia. No adjacent mass or abnormal vessel was noted on soft tissue windows (not shown).*



TERMINOLOGY

Synonyms

- Excessive dynamic airway collapse

Definitions

- Excessive expiratory collapse of trachea or bronchi
  - May be purely intrinsic or in association with longstanding extrinsic compression

IMAGING

General Features

- Best diagnostic clue
  - > 50% ↓ in cross-sectional area of tracheal &/or bronchial lumen during expiration or coughing
    - Correlates well with bronchoscopy, which remains gold standard for diagnosis

Radiographic Findings

- Airway radiography
  - Only 62% sensitive for tracheomalacia

CT Findings

- > 50% ↓ in cross-sectional area of tracheobronchial lumen during expiration found to correlate well with bronchoscopy
- ↑ frequency & severity of air trapping compared to children without tracheomalacia
- Adjacent mass or abnormal vessel (causing extrinsic compression) may contribute to collapse

MR Findings

- Cine MR: Increasingly feasible technique for assessment of tracheobronchomalacia

Imaging Recommendations

- Best imaging tool
  - CT: Dynamic airway (cine or 4D) MDCT vs. paired static inspiratory-expiratory CT
- Protocol advice
  - Intravenous contrast helpful to define vascular anatomy
  - Expiratory phase necessary to evaluate dynamic large airway disease
  - Multiplanar reconstructions & 3D volume rendering helpful

DIFFERENTIAL DIAGNOSIS

Difficult to Control Asthma

- No large airway caliber change with expiration

Foreign Body Aspiration

- Air trapping on expiratory or ipsilateral decubitus images
- Intraluminal filling defect of airway
- ± classic history of choking episode with persistent symptoms

Extrinsic Compression

- Cystic or solid mass
- Aberrant or aneurysmal vessel, vascular ring
- Chronically dilated proximal esophageal pouch due to congenital esophageal atresia

Complete Tracheal Rings

- Completely round, small-caliber trachea
- Unchanging between inspiration & expiration
- May be associated with pulmonary sling

PATHOLOGY

General Features

- Etiology
  - Weakness of airway wall/supporting cartilage

Staging, Grading, & Classification

- Primary (congenital): Impaired cartilage maturation
- Secondary (acquired): Otherwise normal cartilage degenerates following infection, chronic inflammation, intubation, trauma, or longstanding extrinsic compression
  - More common than primary

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Expiratory stridor, dyspnea, cough, sputum production, acute life-threatening events (ALTEs)

Demographics

- Epidemiology
  - Exact incidence unknown
  - Found in up to 15% of infants & 30% of children < 3 years old who have undergone bronchoscopy for respiratory distress

Treatment

- Conservative therapy in mild cases; may improve with age
- Continuous positive airway pressure (CPAP) in moderate cases
- Aortopexy
  - Most common initial operative approach
- Anterior tracheal suspension
  - Relief of airway symptoms in most severe & refractory cases
- Intraluminal tracheal stenting rarely employed

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SECTION 2

Chest

Introduction

Chest radiography is one of the most frequently performed pediatric imaging studies. The radiographic appearance of many conditions is contingent on the age & history of the patient. Additionally, there is often overlap in the radiographic appearances of very different clinical entities. Therefore, arriving at the correct diagnosis often depends on obtaining an adequate history, reviewing prior imaging, & effectively communicating with the referring clinician(s).

Pulmonary Developmental Abnormalities

With the current use of prenatal ultrasound & MR, most developmental pulmonary abnormalities have already been imaged prior to the 1st newborn chest radiograph. This has substantially improved the perinatal care for high-risk infants, particularly given that information predicting pulmonary hypoplasia & persistent pulmonary hypertension can be extracted from these exams (which allows improved planning for the delivery suite). In such lesions, postnatal imaging may be used to confirm the diagnosis & is helpful for surgical planning, particularly given that varying degrees of spontaneous regression have been reported with some entities in late gestation. Postnatal CT for prenatally diagnosed pulmonary masses should be performed as a CT angiogram because lesions can be of mixed etiology (i.e., a hybrid lesion); it is critical for the surgeons to know the vascular supply of such lesions prior to resection.

Examples

Congenital pulmonary airway malformation (CPAM) is a multicystic pulmonary mass with variable amounts of air/fluid in the lesion after birth. The mass is often seen prenatally. Postnatal CTA is always warranted, even if the radiograph appears normal.

Bronchopulmonary sequestration (BPS) is a solid lower lobe lesion that has systemic arterial supply. It is crucial to extend the CTA through the top of the kidneys to exclude an origin of the supplying artery from the upper abdominal aorta.

Components of both CPAM & BPS are present simultaneously in **hybrid lesions**.

Bronchogenic cyst is a fluid density or soft tissue density mass in the mediastinum or medial lung. These cysts can present with respiratory distress or dysphagia.

Congenital lobar overinflation presents with a hyperlucent & hyperexpanded lobe or segment in a neonate or infant. The most frequent locations are the left upper lobe > right middle lobe > right upper lobe. Do not mistake these lesions for a pneumothorax.

Bronchial atresia presents with a characteristic-appearing round or branching, hilar or pulmonary mass ("finger-in-glove") with a hyperinflated lung distally. These lesions may be diagnosed at any age.

Congenital diaphragmatic hernia is not a primary pulmonary developmental anomaly but may appear similar to CPAM or sequestration. Congenital diaphragmatic hernia contains variable abdominal contents (stomach, small/large bowel, liver, spleen). Use abnormal support device positions as clues to this diagnosis.

Acquired Neonatal Lung Disease

Clinical history is often key in diagnosing neonatal lung disease. For example, the diagnostic dilemma of differentiating meconium aspiration from neonatal pneumonia can be solved with a brief history or a phone call to the NICU. At the very least, the radiologist should be provided with the gestational age, as this single data point often significantly narrows the differential diagnosis.

Premature Infant

- **Surfactant deficiency disease** is characterized by low lung volumes & diffuse hazy granular opacities; pleural effusions are uncommon in this setting
- Due to low pulmonary compliance & positive-pressure ventilation, patients with surfactant deficiency disease are at risk for developing **pulmonary interstitial emphysema** (PIE); characterized by bubbly or linear-branching lucencies, it can be difficult to differentiate PIE from the bubbly lucencies of air bronchograms & cysts; look for linear lucencies extending to the lung periphery in PIE; remember that air in the interstitium prevents gas exchange, similar to a pneumothorax
- **Bronchopulmonary dysplasia** (or chronic lung disease of prematurity) is characterized by hyperinflation with coarse reticular opacities & intervening lucencies (typically with > 28 days of oxygen/ventilator support); these patients are at increased risk for pulmonary infections (e.g., RSV) in first 2 years of life

Term Infant

- **Neonatal pneumonia** classically presents as patchy asymmetric perihilar opacities, often with pleural effusions; however, the reality is that neonatal pneumonia can mimic virtually any other neonatal lung disease; thus clinicians are highly conservative in treating for pneumonia until proven otherwise
- **Meconium aspiration** typically shows radiating, coarse rope-like bilateral lung opacities with hyperinflation (& sometimes small pleural effusions); these patients are at risk for pneumothorax; the clinical course is highly variable; adequate history is key as clinicians will already know whether or not meconium was present at birth
- **Transient tachypnea of the newborn** (TTN), or retained fetal lung fluid, presents with streaky perihilar or granular pulmonary opacities; infants with TTN tend to be less seriously ill; imaging findings resolve in 24-48 hours
- **Chylothorax** should be considered in infants who present with pleural fluid, especially in the setting of a lymphatic anomaly, Turner syndrome, or Noonan syndrome
- It should be remembered that congenital heart disease can mimic neonatal lung disease

Pediatric Catheters & Tubes

The most common indication for ordering a NICU chest radiograph is to evaluate the positions of various indwelling support devices (such as vascular catheters, airway tubes, & enteric tubes). Neonatologists are especially meticulous in this regard, as many of them have witnessed catastrophes or near-catastrophes associated with line/tube malpositions. The ubiquitous use of lines & tubes & the low frequency of complications make it disconcertingly easy to overlook line/tube malposition. Rather than simply reporting "lines & tubes unchanged," forcing oneself to report the position of

each support device increases the likelihood of detecting malposition before a complication occurs.

Examples

The ideal position for the **umbilical arterial catheter** tip is between the T6 & T10 levels. The ideal position for an **umbilical venous catheter** (UVC) is at or just below the right atrium/inferior vena cava junction. Remember that a UVC that has not reached the ductus venosus should generally not be used for an infusate, as it will be dispersed into the liver.

Esophageal intubation classically presents with hypoinflated lungs plus gaseous distention of the esophagus & stomach.

ECMO may be performed with either arteriovenous or venovenous cannulae. Central great vessel catheters may rarely be used, particularly in the setting of complex heart disease.

Peripherally inserted central catheters are associated with rare but potentially catastrophic complications, particularly if left to dwell in the right atrium.

Common Pediatric Pulmonary Infections

"Rule out pneumonia" is one of the most common indications for outpatient pediatric imaging. The appearance of pediatric airway disease can be variable. The radiologist who finds himself or herself struggling with the diagnosis of airway disease should keep in mind that most pediatricians rely more heavily on clinical findings than on the radiologist's report to diagnose bronchiolitis or asthma. Clinicians are often more interested to know whether or not there are findings of bacterial pneumonia or other explanations for the patient's symptoms.

Examples

Viral infection is shown by perihilar peribronchial opacities & hyperinflation, often with intermixed atelectasis.

Round pneumonia has a well-defined mass-like appearance, typically occurring in patients under age 8 (due to poorly developed collateral pathways). If it is paraspinous in location, follow-up radiography is advised to ensure resolution of the pneumonia (thereby excluding paraspinal masses such as neuroblastoma).

Management of **parapneumonic effusion** & **empyema** should be based on clinical findings, serial radiography, & ultrasound rather than CT.

Fungal pneumonia in immunocompromised patients classically presents as nodules, potentially with a halo of ground-glass opacity (that suggests hemorrhage).

Papillomatosis may present as multiple laryngeal, tracheal, &/or pulmonary nodules; the latter may cavitate.

Pediatric Mediastinal Masses

Simply differentiating a normal from abnormal thymus can be difficult in infants & young children. However, a few key clues usually lead to confident radiographic differentiation. Cross-sectional imaging, starting with CT, usually allows the radiologist to diagnose the etiology of a true mediastinal mass with reasonable confidence.

Examples

The **normal thymus** can be large in children up to 5 years of age. Look for an undulating contour, the sail or wave sign, & overlying pulmonary vascular markings. There should be no mass effect on the trachea. Ultrasound can be useful to confirm thymic tissue if there is doubt radiographically.

Lymphoma often presents as a bulky anterior mediastinal mass without Ca^{2+}. Mass effect on the trachea & vascular structures is typical.

Mediastinal germ cell tumors present as anterior mediastinal masses, classically with fat & Ca^{2+} (teratoma).

Neuroblastoma typically presents as a posterior mediastinal mass. Look closely for Ca^{2+} as well as bony changes (with posterior rib splaying or erosion being particularly suggestive).

Noninfectious Pediatric Lung Masses

Metastases are by far the most common pulmonary malignancies in the pediatric age group. Primary pulmonary neoplasms are rare in children.

Examples

Lung metastases are most common with osteosarcoma, Ewing sarcoma, hepatoblastoma, soft tissue sarcomas, Wilms tumor, testicular tumors, & thyroid tumors.

Pleuropulmonary blastoma typically presents as a large heterogeneous cystic &/or solid mass. Though much less common than CPAM, it can be impossible to differentiate these entities.

Bronchial obstruction may be due to a foreign body, intrinsic airway mass, or extrinsic compression. Look for asymmetric inflation on bilateral decubitus radiographs.

Pediatric Chest Trauma

A very important element of chest radiography is to evaluate for evidence of child abuse, especially in infants & young children. The radiologist is obligated to immediately contact the referring clinician if there are any suspicious findings of nonaccidental trauma to ensure that the case is appropriately investigated & the child is appropriately protected.

Examples

Multiple **rib fractures** in a child < 3 years old are usually due to child abuse. Rib fractures in abuse most often occur posteriorly but may be lateral & anterior.

Pulmonary contusion appears as a nonspecific opacity in the setting of trauma, often with a thin zone of peripheral lung sparing. The presence of an air-filled or fluid-filled cavity suggests pulmonary laceration.

Aortic injury more often occurs in older children & teenagers, manifesting as in adults (with obscuration of the aorta, mediastinal widening, pleural capping, & pleural fluid on radiography).

Pediatric Diffuse Lung Disease

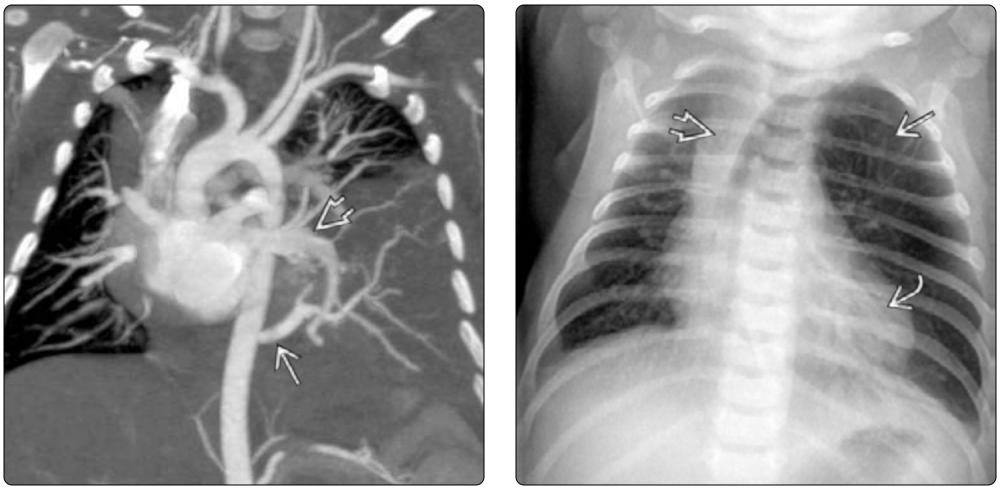
Diffuse lung disease is rare. A few etiologies have a characteristic appearance that may suggest a single diagnosis with reasonable confidence.

Examples

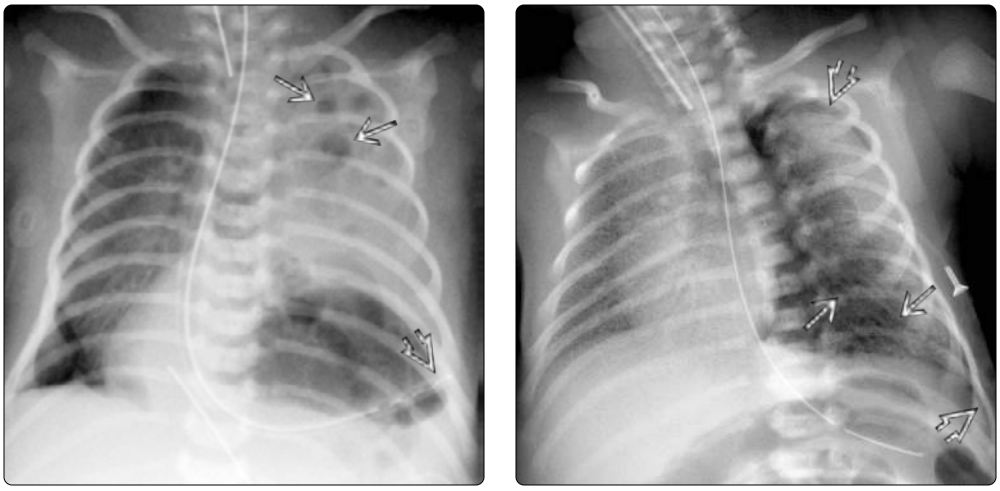
Neuroendocrine cell hyperplasia of infancy (NEHI) has a characteristic HRCT appearance of central & lingular + right middle lobe geographic ground-glass opacities.

Lymphangioleiomyomatosis presents in young women with tuberous sclerosis as numerous bilateral thin-walled cysts.

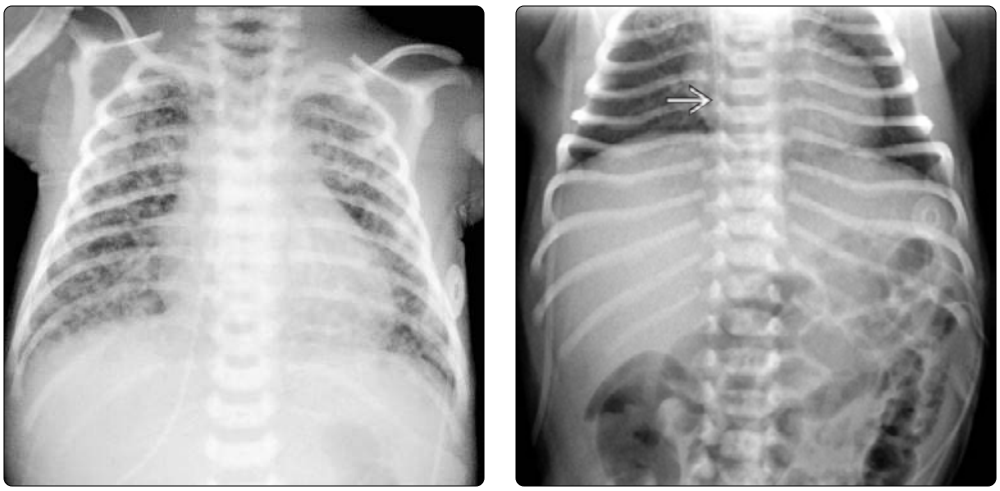
(Left) *Coronal chest CTA MIP image in a 2-month-old infant with a prenatally diagnosed congenital lung lesion shows both the systemic arterial supply ➡ & the pulmonary venous drainage ➡ of the bronchopulmonary sequestration.* **(Right)** *AP chest radiograph in a 2-month-old infant with congenital lobar overinflation shows hyperexpansion of the left upper lobe ➡ with mass effect on the thymus ➡ & compressive atelectasis of the left lower lobe ➡.*

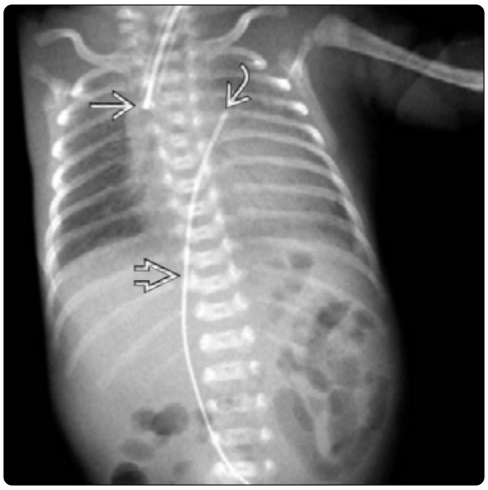


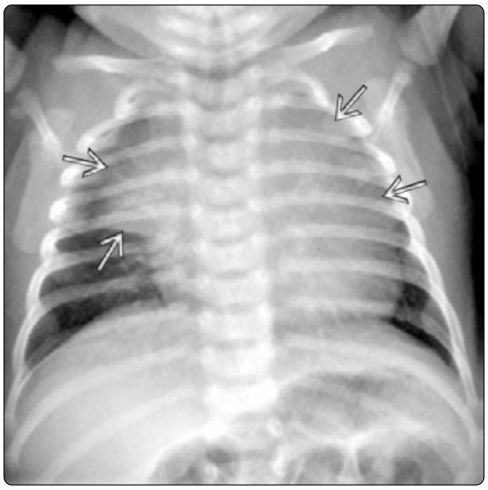
(Left) *AP chest radiograph of a neonate with a left congenital diaphragmatic hernia shows several gas-containing bowel loops ➡ in the left upper hemithorax. The stomach is also in the chest, as confirmed by the enteric tube ➡.* **(Right)** *AP chest radiograph of a premature neonate shows fine granular opacities throughout the right lung, consistent with surfactant deficiency. Note the pulmonary interstitial emphysema ➡ & left pneumothorax ➡ as a result of positive pressure ventilation & ↓ lung compliance.*



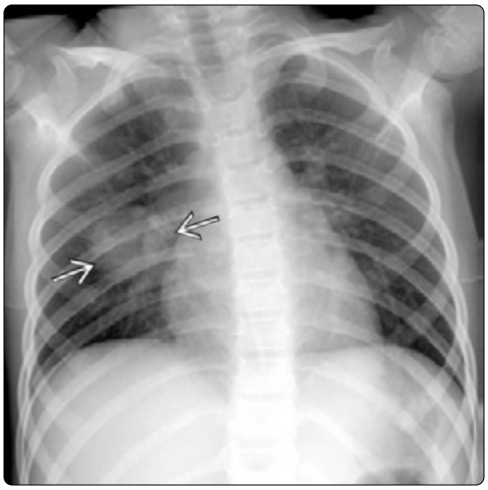
(Left) *AP chest radiograph shows hyperinflation with patchy airspace, & interstitial opacities in this postterm neonate with meconium aspiration.* **(Right)** *AP abdominal radiograph shows a 7-day-old boy with abdominal distension. An upper extremity peripherally inserted central catheter (PICC) tip ➡ is in the right atrium. Soon after this radiograph, the catheter perforated the myocardium, causing life-threatening tamponade. Unlike larger bore central venous catheters, PICCs should not reside in the right atrium.*

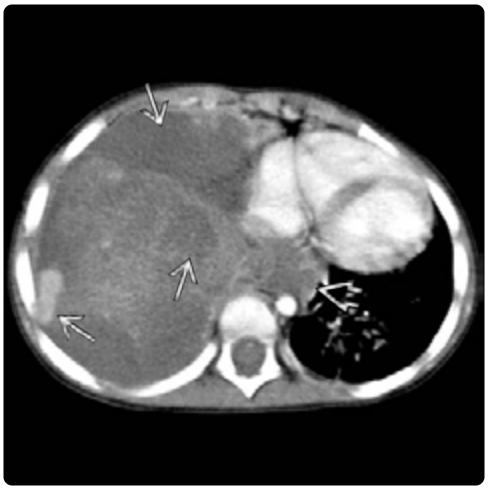




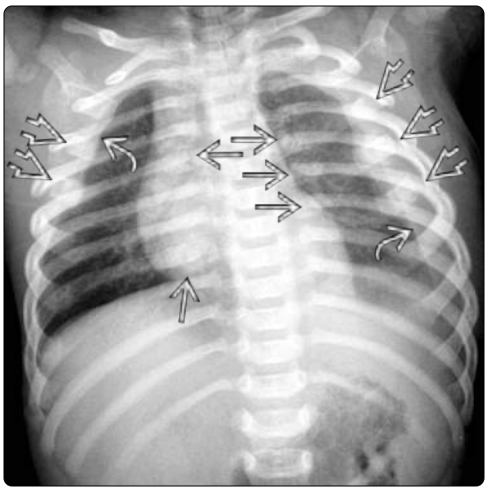


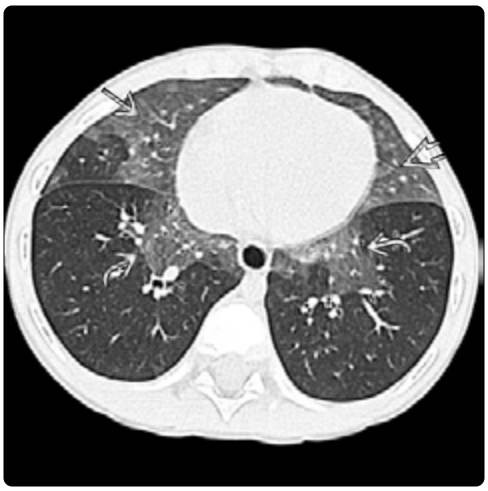
(Left) *AP radiograph following endotracheal tube & umbilical venous catheter (UVC) placement shows abnormal position of the UVC extending into the left upper pulmonary vein (having crossed a patent foramen ovale to the left atrium).* **(Right)** *AP chest radiograph of a 1-month-old boy shows a large but normal thymus that could be mistaken for a mediastinal mass. The shape, mild undulation, location, lack of mass effect, & age are normal for thymus. If unsure, US can confirm thymic tissue.*





(Left) *AP chest radiograph of a 5-year-old child with cough & fever shows a round opacity with air bronchograms in the right lung, consistent with a round pneumonia.* **(Right)** *Axial CECT performed in a 2-year-old child with wheezing shows a heterogeneous mass filling & expanding the right hemithorax. The mass proved to be a pleuropulmonary blastoma.*





(Left) *AP chest radiograph of a 6-month-old boy who had been physically abused shows multiple posterior & anterior rib fractures with bilateral pleural effusions/pleural thickening.* **(Right)** *Axial HRCT in a 13-month-old child with dyspnea, crackles, & failure to thrive shows a characteristic distribution of ground-glass opacities in the middle lobe, lingula, & paramediastinal lower lobes, typical of neuroendocrine cell hyperplasia of infancy (NEHI).*

Normal Thymus

KEY FACTS

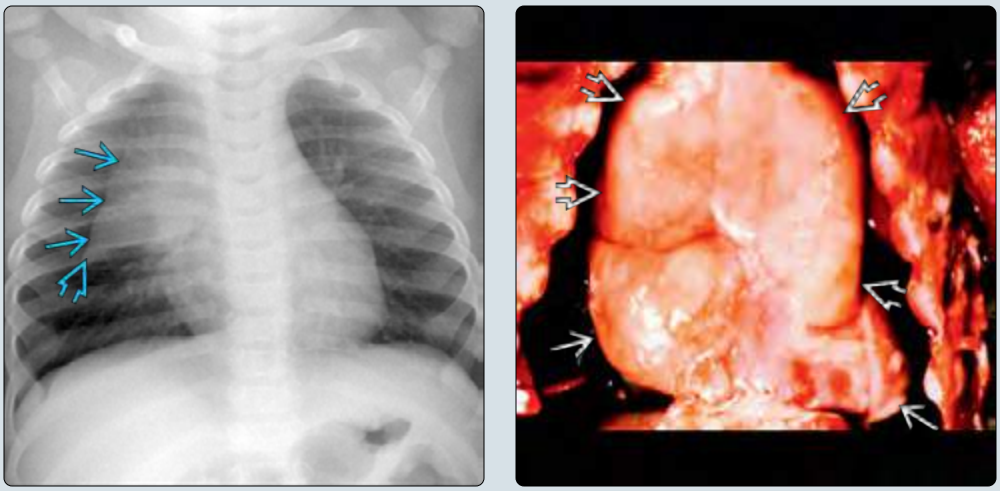
TERMINOLOGY

- Normal organ of anterior superior mediastinum involved in production of T cells
  - Appears prominent in many young children

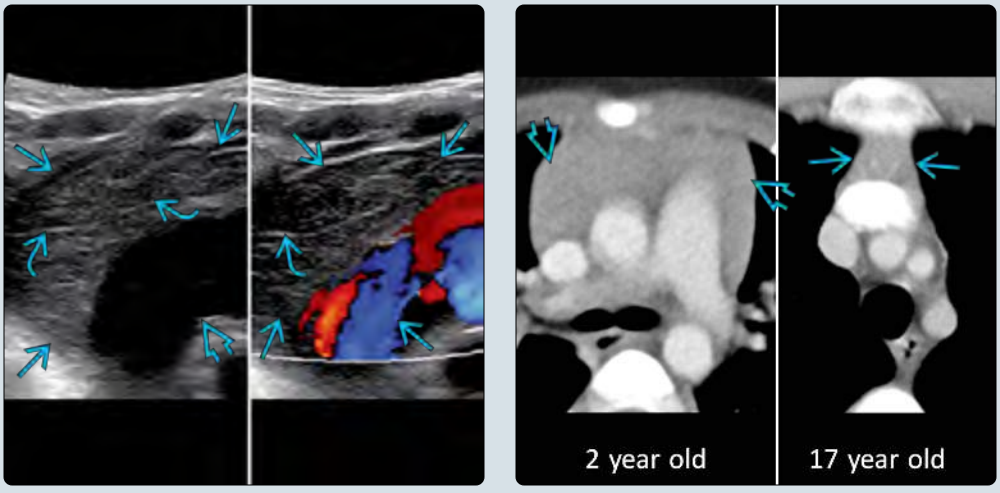
IMAGING

- Normal thymus variable in size, but ↓ with age
  - Moderate to large on chest x-ray up to 5 years of age
  - ↓ in relative size by end of 1st decade
  - No "mass" should be present in 2nd decade
- Normal thymus variable in contour
  - Smooth curvilinear borders ± undulation with ribs
- Normal thymus variable in shape & symmetry
  - Bilobed with convex margins in young children vs. concave margins in older children
  - Sail sign: Lateral triangular extension (right > left)
- Homogeneous consistency without CA^{2+} or cysts
  - Slightly lucent: Vessels seen through thymus
- No mass effect on adjacent structures
- Normal variant locations: Cervical & retrocaval extensions
  - Can be confused with lymphadenopathy or mass
  - Keys to diagnosing normal variant thymic extensions
    - Continuous with & similar imaging appearance (on US, CT, MR) to normally located thymus
      - Ultrasound best tool for confirmation in children
- Aberrant, ectopic thymic tissue may occur in inferior lateral neck or thyroid, ± thymic tissue in normal location
- Thymic rebound
  - Thymic volume can ↓ by > 40% with chemotherapy & other stresses
  - Volume ↑ or rebound to original (or larger) size upon stress cessation, potentially mimicking recurrent mass
    - Differentiation of recurrent lymphoma vs. thymic rebound aided by PET/CT or GRE & DWI MR
- Absent thymic visualization on lateral neonatal chest radiograph occurs with ectopia, DiGeorge syndrome, severe combined immunodeficiency, or prior sternotomy for congenital heart disease

(Left) *AP chest radiograph in a 6-month-old girl shows a normal prominent thymus with a triangular configuration extending into the right hemithorax (sail sign). The normal thymus is lucent & "soft" with undulations at the ribs & intercostal spaces.* **(Right)** *Gross pathology (with the anterior chest wall removed) from the autopsy of a child who died of SIDS shows a normal but prominent thymus as compared to the heart. (Courtesy J. Strife, MD.)*



(Left) *Longitudinal grayscale (left) & color Doppler (right) US in a 2 day old show a normal thymus. Punctate & linear (dot-dash pattern) echogenic foci are present throughout the hypoechoic parenchyma with no increased vascularity. The great vessels are posteroinferior to the thymus.* **(Right)** *Axial chest CECT images are shown in 2 boys of different ages. In the 2 year old, the normal thymus is prominent with convex borders. In the 17 year old, the normal thymus is small & relatively triangular in shape with concave borders.*



TERMINOLOGY

Synonyms

- Thymic sail sign

Definitions

- Normal organ of anterior superior mediastinum that appears prominent in many young children
  - Involved in production of T cells
- Thymos is Greek for warty excrescence

IMAGING

General Features

- Location
  - Normal location: Anterior superior mediastinum
    - Variable distribution of tissue (right vs. left)
  - Normal variant extensions of thymic tissue
    - Superior cervical extension
      - Tissue reaches superiorly in midline above thoracic inlet, anterior to airway
      - 65% of all children (> 80% ages 0-4 years)
      - Can mimic thoracic inlet or lower cervical mass
    - Retrocaval extension
      - Posterior extension of thymus between superior vena cava & great arteries
      - May displace adjacent structures (airway, vessels)
      - Can mimic mediastinal mass or right upper lobe collapse
    - Keys to diagnosing as normal variant thymic tissue extensions (rather than adenopathy or other mass)
      - Connects to normal thymus
      - Shows identical internal characteristics to normal thymic parenchyma by imaging (US, CT, MR)
  - Aberrant, ectopic thymic tissue can be found discontinuous from (or in absence of) normal thymus
    - Aberrant cervical thymus
      - Arrest of normal embryologic descent along thymopharyngeal duct, anywhere from mandibular angle → thoracic inlet
      - Typically asymptomatic lateral cervical mass of normal thymic tissue
      - Retropharyngeal thymus may lead to airway compromise
      - Shows identical internal characteristics to normal thymic parenchyma by imaging (US, CT, MR)
    - Ectopic intrathyroidal thymic tissue
      - Prevalence inversely associated with age
      - Typically detected incidentally on ultrasound & CT
      - Irregular hypoechoic intrathyroidal mass with punctate internal hyperechoic foci similar to normal thymus
      - With typical appearance, can be monitored with follow-up ultrasound until involution
  - Absence of normal retrosternal thymic tissue on lateral neonatal chest radiograph could suggest
    - Ectopic thymic tissue
    - DiGeorge syndrome
    - Severe combined immunodeficiency
    - Prior sternotomy for congenital heart disease
- Size
  - Variable, even among children of same age & size
    - May be large up until 5 years of age
    - ↓ in relative size by end of 1st decade
  - Thymic rebound
    - Thymic volume can ↓ by > 40% with chemotherapy or other stresses
    - Rebound of thymic volume occurs with cessation of stress, up to 50% > original size
      - Growing mediastinal mass in lymphoma patient after therapy could represent thymic rebound or recurrent lymphoma
      - Thymic rebound shows typical thymic characteristics
      - Recurrent lymphoma typically more heterogeneous, nodular with lobulated contours
      - Overlap may necessitate additional imaging for differentiation: PET/CT or gradient-echo, & diffusion MR
- Morphology
  - Contour
    - Normal: Curvilinear smooth borders, undulates with ribs
    - Abnormal: Irregular, lobulated, poorly defined
  - Shape
    - Bilobed ± triangular or quadrilateral configuration
    - Large, with convex borders in young children
    - Relatively small with concave margins in older children

Radiographic Findings

- Radiography
  - Normal size variable
    - ↓ in relative size by end of 1st decade
    - Quite large on chest x-ray up to 5 years of age
    - Should not have prominent "mass" during 2nd decade
  - Normal shape variable
    - Sail sign: Triangular extension laterally (right > left)
    - Not to be confused with spinnaker sail sign: Thymus lifted off heart by gas collection of pneumomediastinum
  - Homogeneous soft tissue density; no CA^{2+} or lucencies
    - Slightly lucent: Vessels seen through thymus
  - Does not displace/compress airway or vessels
  - Drapes over cardiac silhouette; may make heart look prominent

Fluoroscopic Findings

- Fluoroscopy used historically to differentiate normal, prominent thymus from abnormal mass
  - Normal: "Soft," changing contour with respirations
  - Abnormal: "Harder," less compliant with respirations

Ultrasonographic Findings

- Current high-frequency transducers show predominantly hypoechoic, uniformly granular parenchyma with echogenic punctate & linear septa (dot-dash pattern)
  - Hyperechoic to costal/sternal cartilage
  - Hypoechoic to skeletal muscle

CT Findings

- Homogeneous soft tissue attenuation
  - No calcified or low-attenuation foci
  - Attenuation ↓ with age due to fatty replacement

- Smooth borders, not irregular
- Shape on axial imaging
  - Young children: Large quadrilateral with convex borders
  - Teenagers: Smaller triangle with concave borders
- Does not displace/compress airway or vessels
- Associated findings (such as pericardial or pleural effusion or pulmonary disease) favor pathology

MR Findings

- T1WI
  - Homogeneous signal intensity, typically isointense to skeletal muscle
  - Becomes brighter (due to fat) in later teen, adult years
- T1WI C+ FS
  - Homogeneously dark with little enhancement
- In- & opposed-phase T1 GRE
  - Can help differentiate thymic rebound vs. lymphoma
    - Children > 16 years of age: Thymic rebound ↓ in signal intensity on opposed-phase images due to microscopic fat in normal thymus
    - Children < 16 years: Less helpful, as thymus has less fatty replacement & may not drop signal
- T2WI FS/STIR
  - Homogeneously intermediate to bright signal intensity
- DWI/ADC
  - Also used to differentiate thymic rebound vs. lymphoma
    - Thymic rebound less likely to show restricted diffusion & has higher ADC values vs. malignancies

Nuclear Medicine Findings

- FDG PET
  - Mildly FDG avid, much less than tumor
  - Postchemotherapy thymic rebound shows ↑ FDG avidity, making differentiation from recurrent lymphoma more difficult
  - SUVmax < 3.4 suggests rebound while SUVmax > 4 suggests recurrent tumor

Imaging Recommendations

- If chest radiograph demonstrates unusual anterior/superior mediastinal prominence that is questionably normal for age, go to ultrasound

DIFFERENTIAL DIAGNOSIS

Lymphoma or Leukemia

- Most common pathologic anterior mediastinal mass in children, typically in older children/teenagers
- Confluent or nearly confluent nodular masses with mild heterogeneity
- Definite compression/displacement of vessels & airway
- Frequently causes pleural effusions

Germ Cell Tumor (Teratoma)

- Heterogeneous with CA^{2+} & fat attenuation

Thymic Cyst

- Bilobed cyst coursing through thoracic inlet
- Left lateral neck most common
- Typically along carotid sheath

Langerhans Cell Histiocytosis (LCH)

- Systemic LCH can manifest as thymic mass with CA^{2+} or areas of low attenuation
- Associated findings: Lung, hepatic, or bone lesions

Neuroblastoma

- Posterior mediastinal mass (paraspinal)
- May splay/erode ribs, vertebral elements

Lymphatic Malformation

- Transspatial, multicystic mass involving variable portions of neck, chest wall, mediastinum
- Numerous thin septations
- Cysts often contain fluid-fluid levels due to hemorrhage

Congenital Pulmonary Airway Malformation

- Solid-appearing (microcystic) vs. gas & fluid containing (macrocystic) mass
- Not midline

Foregut Duplication Cyst

- Well-circumscribed cyst of middle or posterior mediastinum or hilum

Thymoma

- Very uncommon in young children

PATHOLOGY

General Features

- Prominent normal thymus relative to thoracic size in early childhood
- Smaller relative to thorax by end of 1st decade, though largest actual thymic size occurs in teenage years

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prominent normal thymus: Asymptomatic
  - Ectopic retropharyngeal thymus rarely causes airway compromise

Demographics

- M:F = 4:1 for prominent normal thymus

DIAGNOSTIC CHECKLIST

Consider

- Ultrasound best tool to confirm normal thymic tissue in young child with atypical size, shape, or location radiographically

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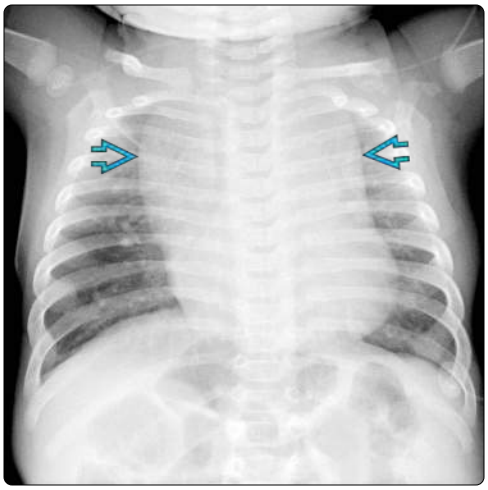
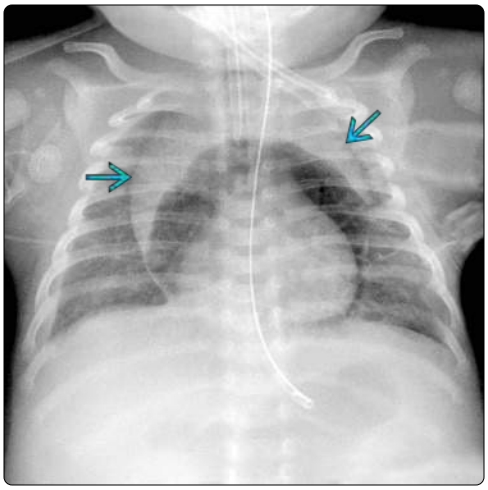
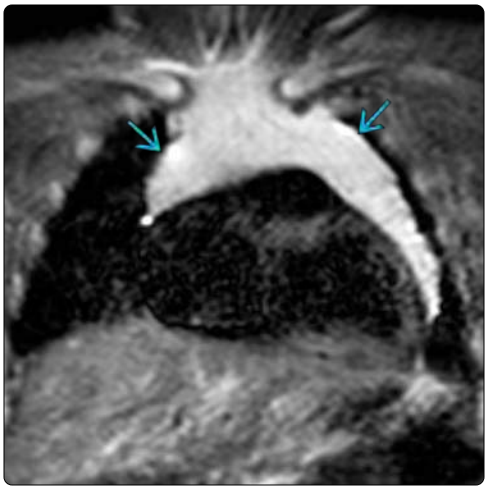
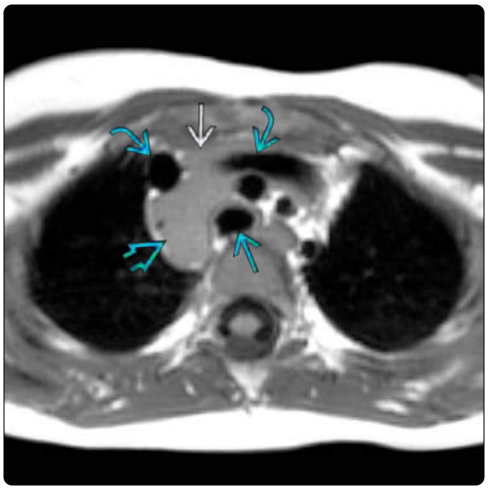
4. Costa NS et al: Superior cervical extension of the thymus: a normal finding that should not be mistaken for a mass. Radiology. 256(1):238-42, 2010
5. Nasseri F et al: Clinical and radiologic review of the normal and abnormal thymus: pearls and pitfalls. Radiographics. 30(2):413-28, 2010



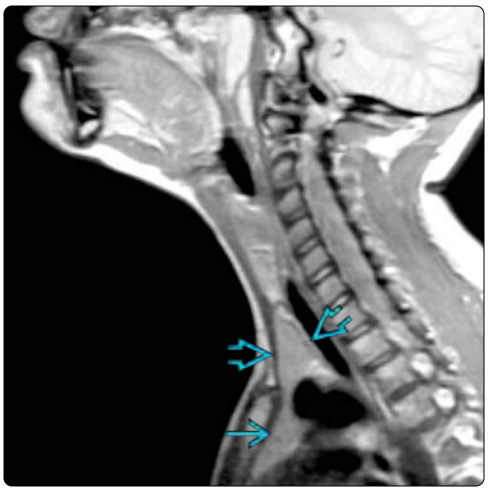


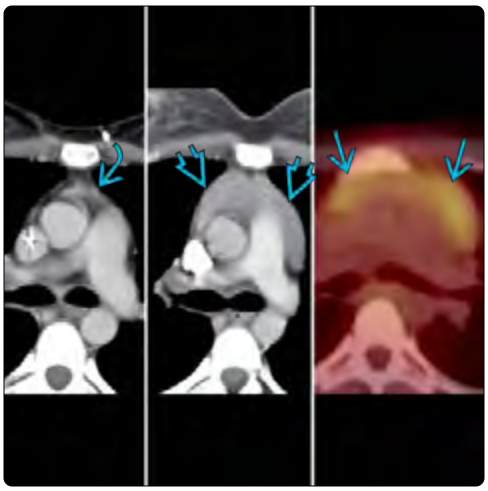
(Left) *AP radiograph in a neonate shows prominence of the superior mediastinum from left to right. This appearance of smooth convex margins is consistent with a normal thymus in a patient of this age.* **(Right)** *AP radiograph of a child in a neonatal intensive care unit shows a spinnaker sail sign, indicative of pneumomediastinum, with a lucent gas collection lifting the thymus off of the cardiac silhouette. The spinnaker sail sign should not be confused with the normal thymic sail sign.*





(Left) *Coronal STIR MR in a 4-month-old patient shows a common configuration of a normal thymus draped over the heart. The thymus is homogeneously hyperintense.* **(Right)** *Axial T1 MR shows a normal variant retrocaval thymus. Lobular soft tissue, isointense in signal to the normally located thymus, extends posteriorly between the trachea & brachiocephalic veins. This appearance should not be mistaken for a mass.*





(Left) *Sagittal PD MR shows superior midline extension of the thymus above the thoracic inlet. Note the continuity of this tissue with the retrosternal thymus.* **(Right)** *CECT in a 16-year-old girl with lymphoma is shown. During chemotherapy (left), her thymus is atrophic. CECT after completion of treatment (middle) shows rebound hyperplasia of the thymus with convex but smooth borders. FDG PET/CT (right) at this time shows increased FDG avidity with an SUVmax < 3.4, typical of rebound.*

Palpable Normal Variants of Chest Wall

KEY FACTS

TERMINOLOGY

- Commonly occurring normal variations of anterior chest wall that may be mistaken for pathology
- Typically isolated variant morphologies of costal cartilages, ribs, &/or sternum
  - Prominent asymmetric convexity or thickness of costal cartilage; may be isolated or due to bifid rib
  - Tilted sternum

IMAGING

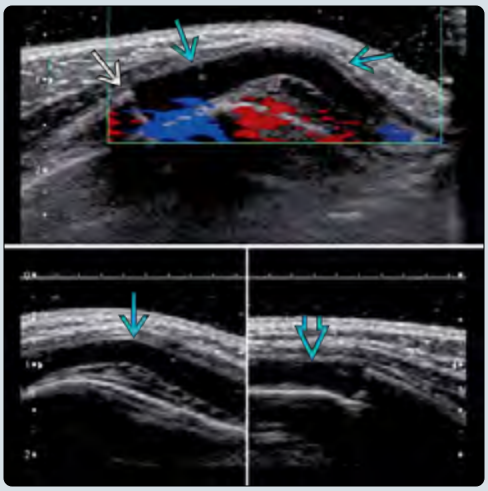
- Ultrasound best initial study for palpable mass unless bony abnormality primarily suspected
- Radiographs best for initial bone evaluation
  - May show bifid rib or bony sternal anomaly
  - Purely cartilaginous variants will not be seen
  - Help exclude other significant processes
    - Bone destruction by malignancy, scoliosis

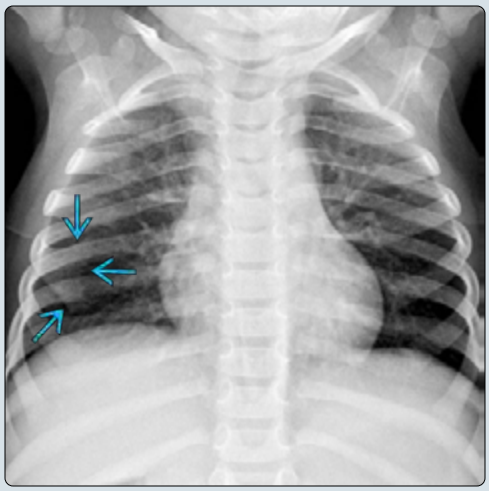
TOP DIFFERENTIAL DIAGNOSES

- Vascular anomaly
- Soft tissue or bone sarcoma
- Osteochondroma
- Pectus excavatum or pectus carinatum
- Scoliosis

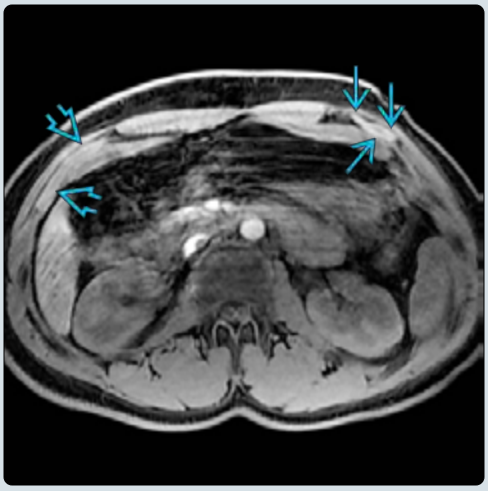
CLINICAL ISSUES

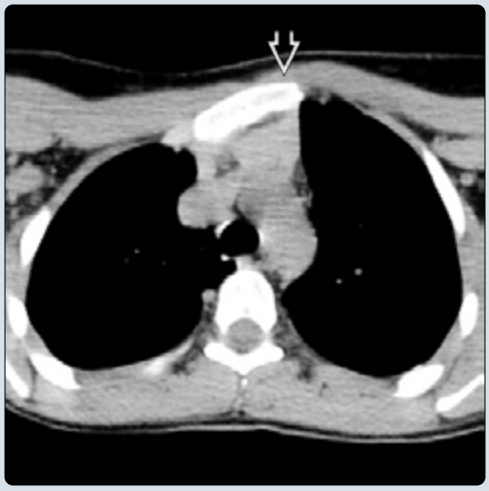
- Asymptomatic palpable "mass" detected by patient, parent, or clinician; usually painless
  - History often erroneously suggests finding as newly or rapidly developed
  - Recent trauma to region may bring palpable abnormality to attention
- Typically isolated finding of little consequence
  - ~ 33% of children imaged for other causes have minor variations in chest wall configuration
  - Isolated bifid rib in 0.15-3.4% of population





(Left) *Transverse color Doppler (top) & grayscale (bottom) ultrasounds of a 2 year old with an asymptomatic palpable abnormality of the right chest wall show an avascular, nearly anechoic anteriorly protuberant costal cartilage that is continuous with the bony anterior rib. The normal comparison left side is shown at the same level.* **(Right)** *AP chest radiograph in the same 2 year old shows a bifid configuration of the right 5th rib at this same level, a normal variant.*





(Left) *Axial T1 GRE FS MR in a teenager with a left lower anterior chest wall palpable abnormality shows focal protuberance & undulation of a costal cartilage on the left, a normal variant. The normal right side is noted for comparison.* **(Right)** *Axial NECT in a patient with a palpable "mass" shows tilting of the sternum with the left margin being more anterior in location as compared to the right.*

TERMINOLOGY

Definitions

- Commonly occurring normal variations of anterior chest wall that may be mistaken for pathology
  - Usually due to variant morphologies of otherwise normal costal cartilages, ribs, &/or sternum

IMAGING

General Features

- Morphology
  - Prominent asymmetric convexity or thickness of costal cartilage; may be isolated or due to bifid rib
  - Tilted sternum
    - More anterior edge of sternum palpated as hard mass on physical examination
    - Frequently associated with prominent asymmetric convexity of costal cartilage

Radiographic Findings

- May show bifid rib or bony sternal anomaly
- Purely cartilaginous variants will not be seen
- Help exclude other significant processes
  - Bone destruction by malignancy
  - Underlying scoliosis

MR Findings

- Excellent soft tissue contrast & variety of sequences allow for characterization of palpable soft tissue or bony mass (whether normal variation or pathology)

Ultrasonographic Findings

- Best 1st-line modality due to
  - Lack of ionizing radiation
  - Lack of need for sedation
  - Excellent superficial spatial resolution
  - Ease of comparison to normal contralateral side
- Easily demonstrates relationship of palpable abnormality to
  - Mildly lobular echogenic subcutaneous fat
  - Striated hypoechoic muscle
  - Nearly anechoic cartilage
  - Echogenic cortical bone with posterior acoustic shadowing

Imaging Recommendations

- Best imaging tool
  - Ultrasound best initial study for palpable mass unless bony abnormality suspected
  - Radiographs best for initial bone evaluation

DIFFERENTIAL DIAGNOSIS

Vascular Anomaly

- Slow flow vascular malformation: Well-circumscribed or infiltrative soft tissue mass with thin septations & fluid-fluid levels
  - Venous malformation: Channels with phleboliths & gradual patchy enhancement
  - Lymphatic malformation: Cystic compartments with rim/septal enhancement only
- High flow vascular neoplasm
  - Infantile hemangioma
    - Well-circumscribed lobulated high flow mass with many low-resistance arterial waveforms (Doppler US) & early diffuse enhancement (MR)

Soft Tissue or Bone Sarcoma

- Firm, often well-circumscribed chest wall mass ± rib destruction, pleural effusion
- Variable enhancement

Osteochondroma

- May present with hard palpable mass, local symptoms of compression/irritation, or stalk fracture
- Characteristic radiographic appearance of flowing corticomedullary continuity between osteochondroma & parent bone

Pectus Excavatum or Pectus Carinatum

- Midline sternal anomalies of concavity (former) or convexity (latter)
- May be isolated or found with many syndromes
- May have associated rib protrusions & sternal tilting

Scoliosis

- Rotary component of spinal curvature leads to protrusion of anterior chest wall on side of curve concavity

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic palpable "mass" detected by patient, parent, or clinician; usually painless
  - History often erroneously suggests finding as newly or rapidly developed
    - Recent trauma to region may bring palpable abnormality to attention

Demographics

- ~ 33% of children imaged for other causes have minor variations in chest wall configuration
- Isolated bifid rib in 0.15-3.4% of population

Natural History & Prognosis

- Typically isolated finding of little consequence

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Congenital Pulmonary Airway Malformation

KEY FACTS

TERMINOLOGY

- Heterogeneous group of cystic & noncystic lung lesions resulting from early airway maldevelopment
- CPAM term now recommended over CCAM as lesions may not be cystic or adenomatoid

IMAGING

- Multicystic lung mass with variable amounts of air & fluid; no Ca^{2+} or rib abnormalities
- By imaging, categorized into large cyst, small cyst, & microcystic/solid subtypes; lesions often mixed
- Radiograph remains initial postnatal study
  - Prenatally detected lesion may be radiographically occult in asymptomatic patient due to recognized phenomenon of partial or complete in utero regression
- CTA for evaluation of residual lesion & surgical planning
  - Identification of systemic feeding artery of "hybrid lesion" (combined CPAM & bronchopulmonary sequestration) critical

TOP DIFFERENTIAL DIAGNOSES

- Bronchopulmonary sequestration (BPS)
- Congenital diaphragmatic hernia
- Bronchogenic cyst
- Pleuropulmonary blastoma
- Congenital lobar overinflation

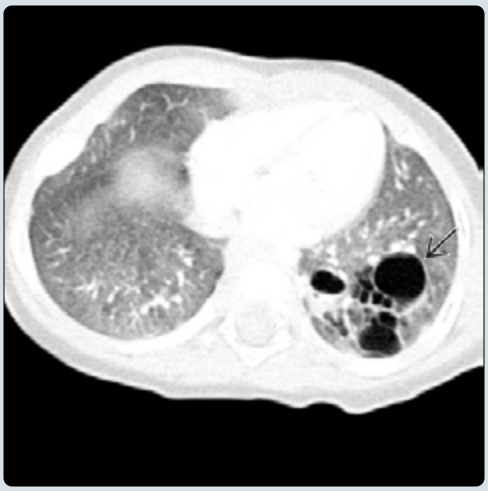
CLINICAL ISSUES

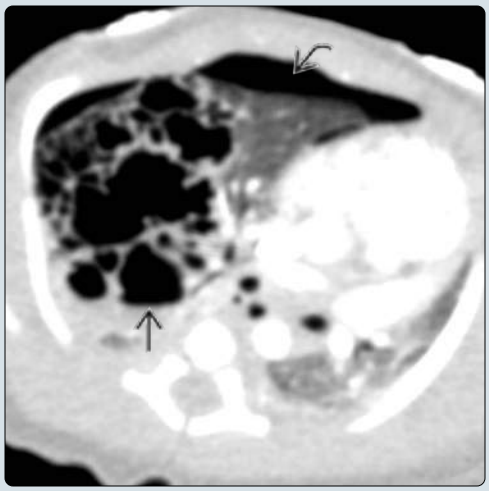
- Overall survival > 95%
- ~ 25% with respiratory distress at birth
  - All symptomatic CPAMs resected
  - Asymptomatic lesions: Expectant vs. elective resection
    - Risks include recurrent infections, underlying malignancy, future malignant degeneration

DIAGNOSTIC CHECKLIST

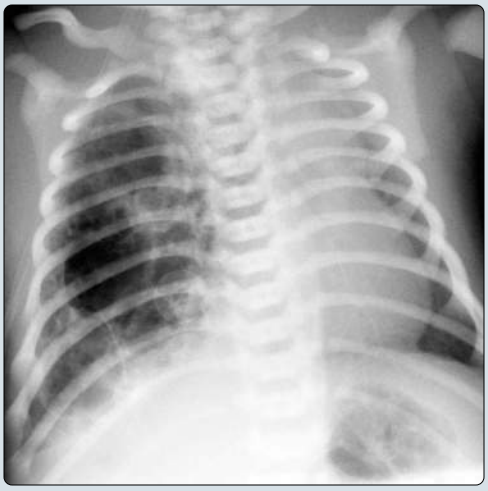
- Low-dose postnatal chest CTA must include celiac axis to look for extrathoracic origin of systemic arterial supply to BPS component of "hybrid lesion"

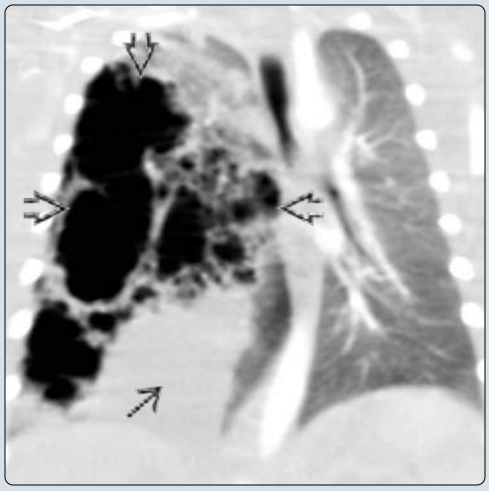
(Left) *Axial chest CTA of a newborn (in lung windows) shows a multicystic lesion in the left lower lobe. The cysts are of varying size, typical of a congenital pulmonary airway malformation (CPAM).* **(Right)** *Axial CTA of a newborn shows a large right lower lobe, predominately macrocystic CPAM. Note the air-fluid level in one of the cysts as well as the accompanying pneumothorax.*





(Left) *Frontal newborn chest radiograph shows a large multiseptated cystic lesion in the right lower lobe causing right to left mediastinal shift.* **(Right)** *Coronal CTA of the same patient shows that, in addition to the large multiseptated macrocystic lesion, there are solid components in the right lower lobe. This was found to be a mixed type 1 & 3 CPAM upon surgical resection.*





TERMINOLOGY

Abbreviations

- Congenital pulmonary airway malformation (CPAM)

Synonyms

- Congenital cystic adenomatoid malformation
  - Antiquated term as lesions may not be cystic or adenomatoid

Definitions

- Heterogeneous group of cystic & noncystic lung lesions resulting from early airway maldevelopment

IMAGING

General Features

- Best diagnostic clue
  - Multicystic lung mass in newborn
    - Fluid-filled cysts at birth, gradually filling with air
- Location
  - Intrapulmonary without lobar predilection
- Size
  - 5 histopathological types (Stocker classification) with varying sizes of cysts reflecting level of inciting airway abnormality & dictating imaging appearance
    - Type 0 (< 2%): Small lungs with acinar dysgenesis (incompatible with life)
    - Type 1 (65%): 1 or more large (3-10 cm) cysts ± smaller cysts blending with normal lung
    - Type 2 (10-15%): Numerous small (0.5-2.0 cm) cysts
    - Type 3 (5-8%): Microcysts (appearing solid) with marked ↑ size of lobe or lung
    - Type 4 (10-15%): Large peripheral cysts with mass effect
  - By imaging, generally categorized into large cyst, small cyst, & microcystic/solid types; may be of mixed types
- Morphology
  - Quite variable: Round, lobulated, amorphous, lobar

Radiographic Findings

- Multicystic lung mass with variable amounts of fluid & air
  - Solid radiographic appearance may be due to
    - Microcystic type 3
    - Initially fluid-filled larger cysts (types 1, 2, 4)
- Mass effect most common with types 3 & 4
- Pneumothorax may occur, especially with type 4
- No discrete rib abnormalities
- May be normal despite prenatal diagnosis of lesion
  - Complete vs. partial regression in many cases
  - Occult lesions may be found by CT

CT Findings

- CTA
  - May show partial or complete regression of prenatally detected lesion
  - Performed to determine extent of residual lesion & evaluate for systemic feeding vessel prior to surgery
    - Systemic arterial supply classically suggests component of bronchopulmonary sequestration (BPS) ("hybrid lesion" with CPAM)
    - Origin of arterial supply critical for surgical planning
      - Must cover through celiac axis to exclude extrathoracic origin of artery
  - Mass ranges from solid/microcystic to macrocystic
    - Components of each may be present
    - Cysts of variable size; contain air &/or fluid
      - Intermixed microcysts may appear as air trapping
      - Margins with normal lung may be difficult to define
  - No Ca^{2+} or rib abnormalities
  - May cause hyperinflation or atelectasis of adjacent lung

MR Findings

- May be used in prenatal evaluation of
  - Nature of lung lesion
  - Hypoplasia of remaining lungs
    - Lung volume calculations may be helpful
  - Mass effect on mediastinal structures
  - Presence of additional anomalies
  - Associated hydrops
- T2 SSFSE most useful sequence
  - Solid/microcystic lesions almost always higher in signal intensity than remaining lungs
    - Lower lesion signal may be due to regression
  - Cystic lesions of fluid signal intensity

Ultrasonographic Findings

- Typically limited to prenatal evaluation
  - Solid-appearing echogenic mass relative to normal lung
  - Typically stabilizes or ↓ in size > 26 weeks gestation
    - Often regresses or "resolves" by late 3rd trimester
      - False-negative rate of 40% compared to postnatal CT
  - Discrete anechoic cysts of variable size
  - ± pleural effusion, skin edema, other findings of hydrops
    - CPAM volume ratio (CVR) > 1.6 → 80% develop hydrops
  - Systemic arterial supply on Doppler → component of BPS ("hybrid lesion")

DIFFERENTIAL DIAGNOSIS

Bronchopulmonary Sequestration

- Left > right lower lobes
- Solid-appearing; only becomes air-filled after infection
- Systemic arterial supply on fetal US/MR or infant CTA
- Mixed CPAM/sequestration ("hybrid") lesions common
  - More likely to have discrete cysts

Congenital Diaphragmatic Hernia

- Appears radiographically as multicystic, air-containing mass
  - "Cysts" often uniform in size & morphology
- Expected abdominal courses of support devices altered
  - Nasogastric tube with stomach herniation
  - Umbilical venous catheter with liver herniation
- Paucity of abdominal bowel gas (due to herniation)

Bronchogenic Cyst

- Round, typically mediastinal or perihilar, fluid-density mass
- May become air-filled if infected

Pleuropulmonary Blastoma

- Very rare neoplasm of young children; typically not discovered in prenatal/neonatal periods

- Type I: Air-filled cystic pleuropulmonary blastoma (PPB) overlaps CPAM appearance
  - Large cyst, solid soft tissue component, multilobar or bilateral involvement, & gene mutation make PPB more likely
    - *DICER1* mutation: ± concurrent cystic nephroma

Cavitary Necrosis/Abscess Complicating Pneumonia

- Ill children, typically not neonates
- Cavitary necrosis typically surrounded by consolidated lung
- Temporal factors favoring cavitary necrosis: Prior normal chest radiograph, lesion progression during illness, lesion resolution after illness

Congenital Lobar Overinflation

- Progressive lobar overexpansion in fetus/neonate
- Preserved pulmonary markings without discrete cyst walls
- Rarely occurs in lower lobes

Persistent Pulmonary Interstitial Emphysema

- Focal cyst may develop with otherwise typical appearance of barotrauma-induced pulmonary interstitial emphysema
  - Regional small bubbly interstitial lucencies in background of granular airspace opacities of surfactant deficiency

PATHOLOGY

General Features

- Etiology
  - Congenital lung mass of various cell origins
    - Type 0: Tracheobronchial; type 1: Bronchial/bronchiolar; type 2: Bronchiolar; type 3: Bronchiolar/alveolar; type 4: Distal acinar
  - Lesion development poorly understood; related to varying degrees & levels of early airway obstruction
    - Lesions in communication with bronchial tree at birth
  - Hyperplastic, hamartomatous, & neoplastic elements within different lesions
    - Type 4 difficult to differentiate from PPB by imaging & histopathology
- Associated abnormalities
  - Other congenital anomalies in type 2 lesions (50%)

Microscopic Features

- Large cysts: Lined by respiratory epithelium
- Small cysts: Closely apposed, bronchiole-like structures with variable intervening lung parenchyma
- Microcystic lesion: Adenomatoid appearance with randomly scattered, bronchiolar/alveolar, duct-like structures

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prenatal diagnosis ± symptoms at birth
  - Respiratory distress in newborn (~ 25%)
    - Depends on size of lesion, mediastinal shift, hydrops
  - Recurrent lung infections in older child

Demographics

- Age
  - Most commonly detected prenatally or during infancy
  - Can present at any age
- Epidemiology
  - Incidence: 1 per 15,000-25,000 live births

Natural History & Prognosis

- Overall survival > 95%
- ~ 13% require neonatal respiratory support
- Risks of unresected CPAM
  - Recurrent infections
  - Future malignant degeneration
    - Bronchioloalveolar carcinoma, rhabdomyosarcoma
  - Small risk that lesion actually represents PPB
- Prenatally detected CPAM may grow until 26 weeks gestation followed by varying degrees of regression
  - ~ 20% become sonographically undetectable in 3rd trimester but with false-negative rate of 40%
  - 10% (typically microcystic with low initial CVR) show complete regression by postnatal CT &/or pathology

Treatment

- Prenatally detected lesions followed with ultrasound/MR
  - Smaller lesions without hydrops: Close follow-up ± corticosteroids
  - Larger lesions at risk for/with hydrops: Corticosteroids, consider fetal interventions
    - Aspiration/shunting of dominant cyst
    - Resection of lesion
- Symptomatic CPAM after delivery → surgical resection
- Asymptomatic CPAM management controversial
  - Most advocate elective resection due to risks of infection & malignancy
  - Fewer complications if surgery performed prior to development of symptoms
    - ~ 2/3 of expectantly managed patients ultimately become symptomatic

DIAGNOSTIC CHECKLIST

Consider

- Most true multicystic pulmonary masses containing air in neonatal period represent CPAM

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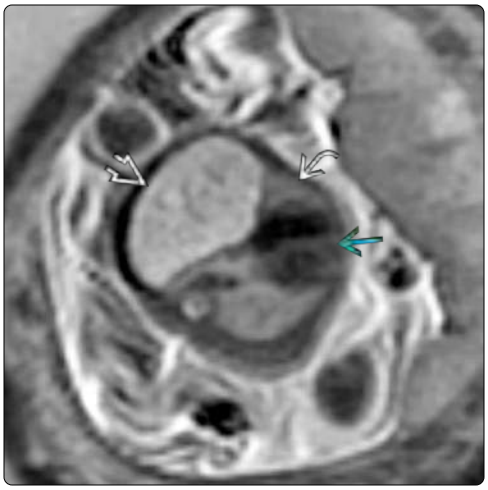
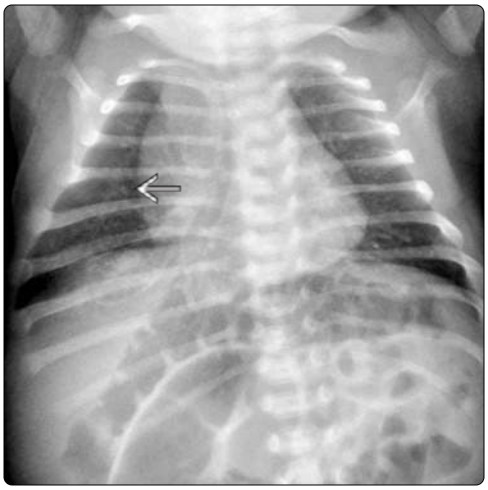
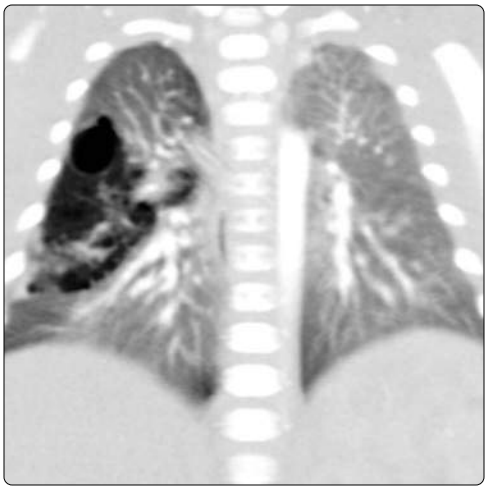
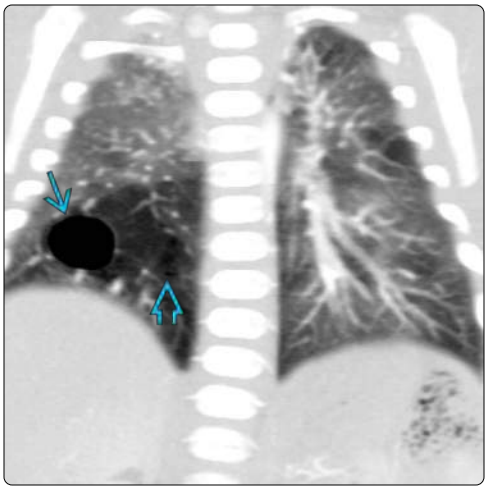


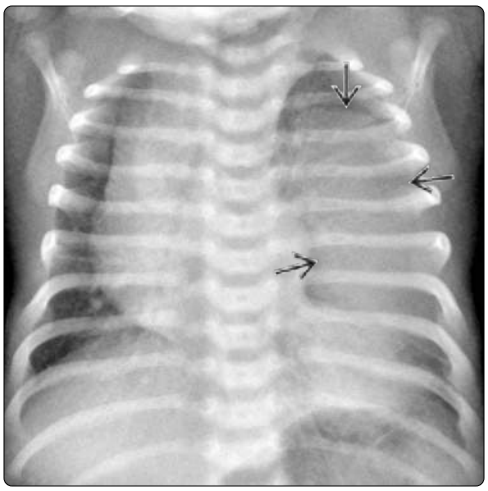


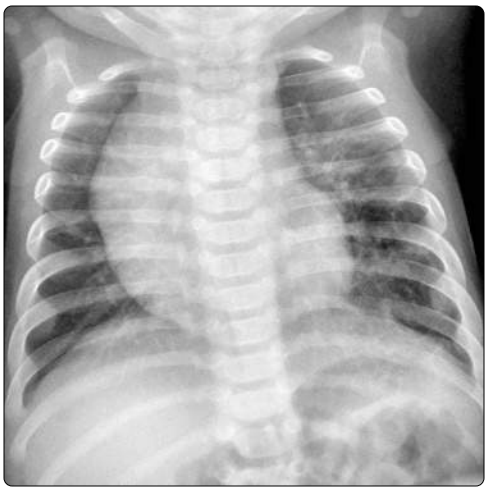
(Left) *Axial T2 SSFSE MR in a 23-week gestation fetus shows a large homogeneously hyperintense lesion occupying the majority of the right hemithorax & compressing the residual normal right lung. The heart is displaced into the left chest.* **(Right)** *Newborn AP chest radiograph of the same patient shows interval regression of the lesion with only mild architectural distortion identified in the right lung. The heart now has a normal position.*





(Left) *Coronal CTA in the same patient shows a residual, predominately hyperlucent right lung lesion consisting of cysts of variable size. The lesion splays normal pulmonary vessels. A mixed type 1 & type 2 CPAM was confirmed upon resection.* **(Right)** *Coronal CTA in a neonate shows a hyperlucent right lower lobe CPAM. There is a single large macrocyst laterally in the lesion. Other smaller cysts are intermixed with microcysts &/or air trapping.*





(Left) *AP chest radiograph in a 3-week-old boy shows a large dense mass of the left lung causing left to right mediastinal shift.* **(Right)** *Follow-up AP chest radiograph in the same infant 6 weeks later shows that the large left lower lobe lesion has become aerated, typical of a CPAM.*

Bronchopulmonary Sequestration

KEY FACTS

TERMINOLOGY

- Congenital focus of abnormal lung that does not connect to bronchial tree or pulmonary arteries
- Divided into intralobar (75%) & extralobar (25%) types
  - Intralobar shares pleural investment with normal lung & usually has pulmonary venous drainage
  - Extralobar has separate pleural investment from lung & usually has systemic venous drainage

IMAGING

- Lower lobe (left > right) opacity persisting over time
  - Mass may occur in mediastinum or in/below diaphragm
- Systemic arterial supply characteristic
  - Identification important for diagnosis, surgical planning
    - Postnatal CTA recommended for all congenital lung lesions (even with 3rd-trimester regression)
  - Arterial supply may be subdiaphragmatic in origin even if BPS supradiaphragmatic
    - Chest CTA must cover through celiac axis
- Often occurs as "hybrid lesion" with congenital pulmonary airway malformation (CPAM)

TOP DIFFERENTIAL DIAGNOSES

- Depends on location & imaging features
  - Chronic lung opacity
  - Anomalous systemic blood flow to lung
  - Focal chest mass
  - Solid suprarenal mass

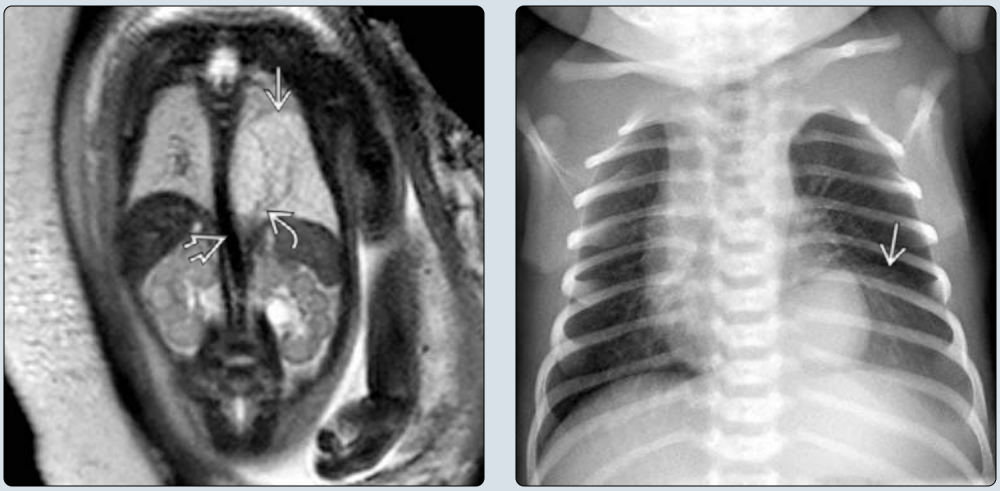
CLINICAL ISSUES

- Intralobar typically presents as isolated anomaly in older children/adults with recurrent pneumonia
- Extralobar typically detected in prenatal/neonatal period, often with additional congenital anomalies
- Symptomatic BPS resected; management of asymptomatic lesions controversial

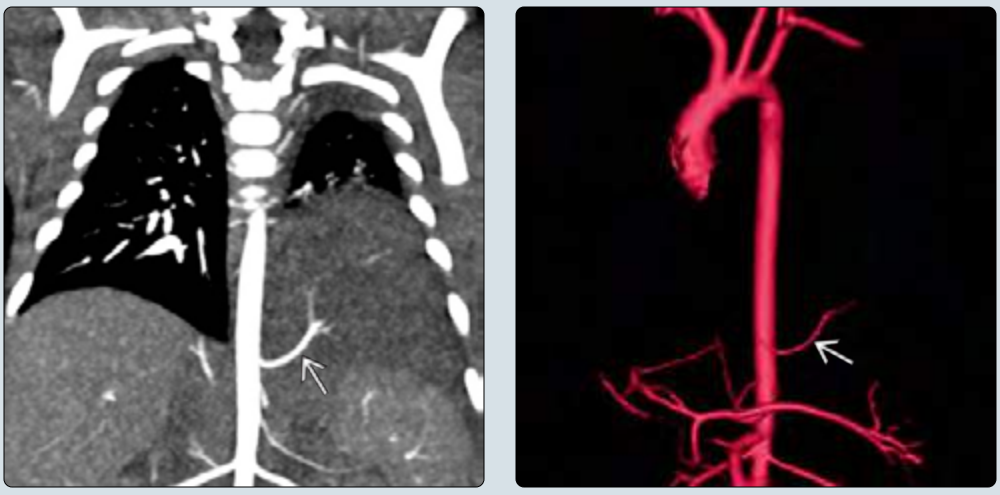
DIAGNOSTIC CHECKLIST

- Consider BPS with recurrent lower lobe pneumonia

(Left) *Coronal T2 SSFSE MR of a 31-weeks gestation fetus shows a large, homogeneously hyperintense left lower lobe mass ➡ displacing the left upper lobe & aorta ➡. Flow voids ➡ are seen extending into the mass from the aorta.* **(Right)** *AP radiograph of the same patient 5 days after birth shows mild decreased size of the left lower lobe mass ➡.*



(Left) *Coronal CT angiogram of the same patient 11 days later shows an arterial feeding vessel ➡ extending into the solid mass from the upper abdominal aorta, confirming systemic arterial supply in a BPS. Upon resection, the mass contained components of both BPS & CPAM, consistent with "hybrid lesion."* **(Right)** *3D coronal oblique reconstruction from the same CT angiogram shows the feeding vessel ➡ relationship to other upper abdominal arterial origins.*



TERMINOLOGY

Definitions

- Bronchopulmonary sequestration (BPS): Dysplastic nonfunctional lung tissue with no connection to bronchi or pulmonary arteries
- Intralobar (75%) vs. extralobar (25%) types
  - Intralobar sequestration (ILS) typically has pulmonary venous drainage without separate pleural investment
  - Extralobar sequestration (ELS) typically has systemic venous drainage with separate pleural investment

IMAGING

General Features

- Best diagnostic clue
  - Lower lobe opacity that persists over time
  - Systemic arterial supply to lesion
- Location
  - 98% in lower lobes (left > right)
  - Up to 15% of ELS occurs below diaphragm
  - Rarely in diaphragm, mediastinum
- Systemic feeding artery usually from descending aorta
  - Identification important for diagnosis, surgical planning
- ± elements of other congenital lung lesions, especially congenital pulmonary airway malformation (CPAM)
  - 17-42% of congenital lung lesions "hybrid" or "complex"
- May be difficult to differentiate ILS vs. ELS by imaging
  - May not affect surgical management

Radiographic Findings

- Radiography
  - Persistent lower lobe opacity over time
  - May contain gas from infection, bronchial fistula (in ILS), collateral flow, or CPAM components ("hybrid lesion")
  - ELS may present as subdiaphragmatic mass displacing adjacent interfaces, such as paraspinal stripes

CT Findings

- CTA
  - Recommended for all congenital lung lesions to evaluate systemic vascular supply prior to surgery
    - Descending thoracic aorta most common source of feeding vessel
    - Other sources: Abdominal aorta; celiac, splenic, intercostal, subclavian, coronary arteries
      - Rarely aberrant pulmonary artery
    - Arterial supply may be subdiaphragmatic in origin even if sequestration is supradiaphragmatic
    - 3D reconstruction helpful for surgical planning
  - Discrete solid mass ± cysts vs. opacification of lower lobe parenchyma
  - ± gas in lesion with infection, fistula, collateral alveolar communication, or hybrid lesion with CPAM
  - ELS may torse, thrombose, or hemorrhage, making diagnosis more difficult

MR Findings

- Fetal MR often used for prenatal lung lesion assessment
  - Not typically used postnatally unless neuroblastoma suspected
- Most frequently: Homogeneous, T2-hyperintense, solid-appearing mass; ± cystic components in hybrid lesions
- Feeding artery visualized as flow void on SE/FSE sequences; may be bright on some GRE sequences

Ultrasonographic Findings

- Postnatal ultrasound
  - Abnormal lung may provide acoustic window
  - Doppler ultrasound may demonstrate feeding vessel
- Prenatal ultrasound
  - Homogeneous echogenic lung mass ± cystic components in hybrid lesions
  - May see systemic feeding artery with Doppler

Imaging Recommendations

- Best imaging tool
  - Prenatal imaging
    - US excellent first-line study for detection/characterization; may not need fetal MR
  - Postnatal imaging
    - When BPS suspected prenatally, obtain postnatal CTA for feeding vessel detection prior to surgery
- Protocol advice
  - Extend chest CTA through celiac axis to detect subdiaphragmatic arterial supply

DIFFERENTIAL DIAGNOSIS

Chronic Lung Opacity

- Chronic or recurrent pneumonia
  - Typically no systemic arterial supply
- Chronic bronchial obstruction
  - Aspirated foreign body
  - Endobronchial lesion (e.g., carcinoid)

Anomalous Systemic Blood Flow to Lung

- Hypogenetic lung syndrome (scimitar)
  - Hypoplasia of right lung & pulmonary artery
  - Partial anomalous pulmonary venous return
  - Systemic arterial supply of right lung base ± BPS
- Chronic inflammation with hypertrophied bronchial arteries
  - Cystic fibrosis, chronic pneumonia
- Pulmonary artery atresia
  - Multiple aortopulmonary collateral arteries (MAPCAs)
- Bronchial atresia may have systemic supply
  - Overlaps with ILS
- Isolated aberrant artery without lung abnormality

Focal Chest Mass

- CPAM
  - Often has cystic components filling with air postnatally
  - No systemic arterial supply unless hybrid lesion
  - May be seen in middle, upper lobes
- Bronchogenic cyst
  - Mediastinal or central pulmonary location
  - Well-defined cyst without feeding artery
  - May compress bronchus leading to overinflation lesion (mimicking BPS prenatally)
- Round pneumonia
  - Most common round lung opacity in young children
    - Usually < 8 years of age
  - Should resolve/improve with antibiotics

- Pleuropulmonary blastoma (rare)
  - Type I indistinguishable from benign lung cysts
  - Large thoracic soft tissue mass (types II & III)
- Neuroblastoma
  - Ovoid paraspinal mass displacing paraspinal stripe(s)
  - ± rib splaying, erosion
- Lymphatic malformation
  - Multicystic mass crossing multiple soft tissue planes
  - Often involves upper mediastinum, neck, chest wall

Solid Suprarenal Mass

- Neuroblastoma
  - Often calcifies & engulfs normal vessels
- Adrenal hemorrhage
  - No central vascularity
  - ↓ or resolves over time

PATHOLOGY

General Features

- Abnormal dysplastic lung tissue lacking normal/functional tracheobronchial connection
- Development likely related to caudal accessory lung bud with abnormal vascular supply from foregut
- Underlying airway obstruction likely contributes to maldevelopment
  - Foci of bronchial atresia found in most congenital lung lesions including BPS (ELS > ILS)
- Aberrant feeding artery in all cases
  - Usually systemic (5% from pulmonary artery)
  - Single supplying artery in 80%; 2 or more in 20%
  - May not be visualized by imaging
- Extralobar sequestration
  - Always congenital
  - Often associated with other anomalies (65%)
    - Congenital diaphragmatic hernia, cardiac abnormalities, pulmonary hypoplasia, foregut duplication cysts
  - Separate pleural investment; usually has systemic venous drainage
- Intralobar sequestration
  - Not all lesions congenital; some may develop from chronic postnatal inflammation
  - Infection may lead to bronchial fistula
  - Not associated with other anomalies
  - Shares pleural investment with normal lung; usually drains into pulmonary venous system

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - ELS: Often asymptomatic postnatally
    - May cause neonatal respiratory distress from mass effect or adjacent pulmonary hypoplasia
    - Rarely becomes infected
  - ILS: Recurrent pneumonia symptoms in older child/adult
- Other signs/symptoms
  - ELS: May torse, infarct
    - Prenatally can develop hydrothorax, hydrops
    - Postnatally may cause chest/abdominal pain
  - ILS: Chest pain, hemoptysis; incidental in 15%

Demographics

- Age
  - ELS: Usually detected in prenatal/neonatal period
  - ILS: Classically, late childhood/early adulthood presentation; 50% > 20 years old
- Gender
  - ILS: M = F
  - ELS: M:F = 3-4: 1

Natural History & Prognosis

- High rate (up to 67%) of spontaneous in utero regression (partial or complete) of prenatally detected echogenic lung lesions
  - ~ 50% of prenatally detected BPS lesions

Treatment

- Surgical resection in symptomatic cases
  - Reports of preoperative vs. isolated embolization of feeding artery (with variable success as primary therapy)
- Management of asymptomatic cases controversial
  - Elective surgical resection vs. monitoring
    - Easier surgery prior to infection
  - If no feeding artery by imaging, may be excised for concern of other lesions
- High mortality if ELS torsion leads to hydrops in utero
  - High success rates of minimally invasive in utero laser ablation of feeding vessel → ↓ size of mass & effusion with resolution of hydrops → up to 100% survival

DIAGNOSTIC CHECKLIST

Consider

- Raise BPS possibility with recurrent lower lobe pneumonia &/or paraspinal mass
- Postnatal CTA should be performed for prenatally detected lung lesions (even with 3rd-trimester regression)

Reporting Tips

- Imaging differentiation of ELS vs. ILS may not be possible
- Describe size, location, vascular supply/drainage, aeration, involvement of adjacent structures, associated anomalies
  - Critical to define feeding vessel origin prior to surgery

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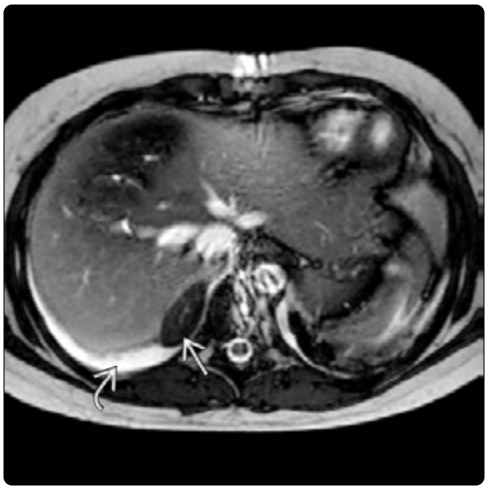
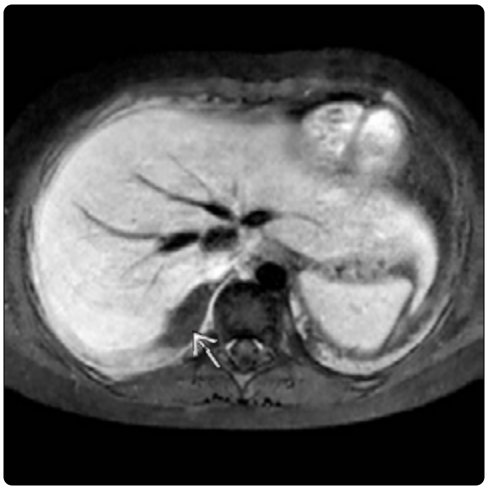
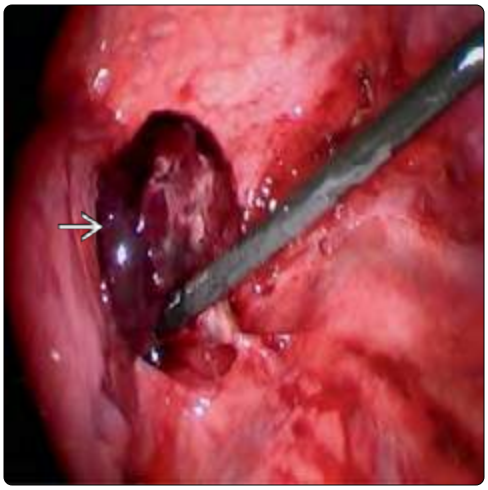
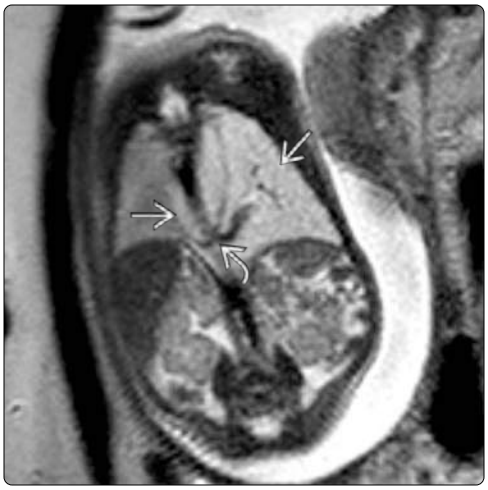


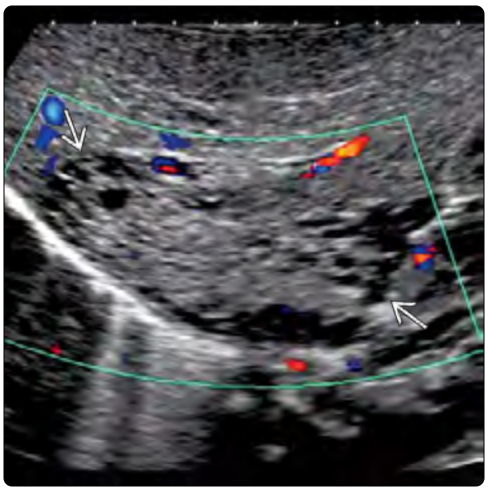


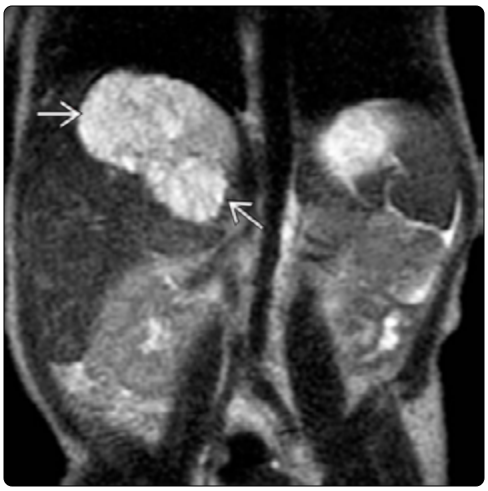
(Left) *Axial SSFP MR in a 10-year-old patient with right abdominal pain & fever shows an ovoid, hypointense mass ➡ medially in the right lower hemithorax with an adjacent pleural effusion ➡.* **(Right)** *Axial T1 C+ FS MR in the same patient shows no enhancement of the mass ➡.*





(Left) *Thoracoscopy of the right hemithorax in the same patient shows a small, hemorrhagic mass ➡ in the pleural space. Histology revealed a torsed & infarcted extralobar BPS.* **(Right)** *Coronal T2 SSFSE MR in a 27-weeks gestation fetus shows a large, uniformly hyperintense, solid left lower lobe lesion ➡ with a large, trifurcating feeding vessel ➡ arising from the thoracic aorta, consistent with BPS.*





(Left) *Right upper quadrant color Doppler US in a newborn with a prenatally diagnosed suprarenal vs. hepatic mass shows an ovoid mixed solid & cystic lesion ➡ with no significant internal vascularity or feeding vessel.* **(Right)** *Coronal T2 MR SSFSE in the same patient shows a heterogeneously hyperintense, lobular mass ➡ along the posterior liver. Upon resection, a retroperitoneal extralobar BPS was confirmed with CPAM components ("hybrid lesion"). No feeding vessel was ever seen on imaging or surgical exploration.*

Bronchogenic Cyst

KEY FACTS

TERMINOLOGY

- Congenital mass in family of foregut duplication cysts: Bronchogenic cysts (BC), enteric cysts, neurenteric cysts
- Result from abnormal ventral budding of tracheobronchial tree between 26th & 40th days of gestation

IMAGING

- Well-defined, round to ovoid, unilocular, thin-walled cyst with smooth margins
- Mediastinal location (usually middle) > > lung (medial 1/3 of lower lobes) > neck, abdomen, heart, subcutaneous tissues
  - Typically subcarinal, paratracheal, or hilar
- May compress airway → adjacent atelectasis or air-trapping
- Fluid filled; gas in cyst uncommon, may be due to infection or rare communication with airway
- CECT: Variable attenuation of cyst contents due to protein, hemorrhage, or (rarely) calcium; minimal enhancement of thin wall
- MR: Variable T1 signal; T2 FS or STIR imaging maximizes conspicuity of high fluid signal in cyst

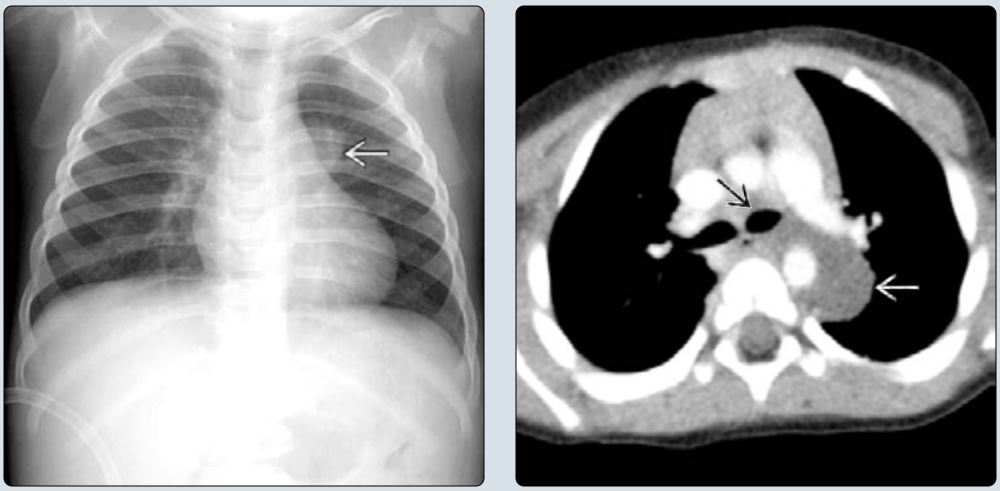
TOP DIFFERENTIAL DIAGNOSES

- Esophageal duplication cyst
- Round pneumonia
- Neuroblastoma
- Lymphadenopathy
- Lymphatic malformation
- Congenital pulmonary airway malformation
- Bronchopulmonary sequestration

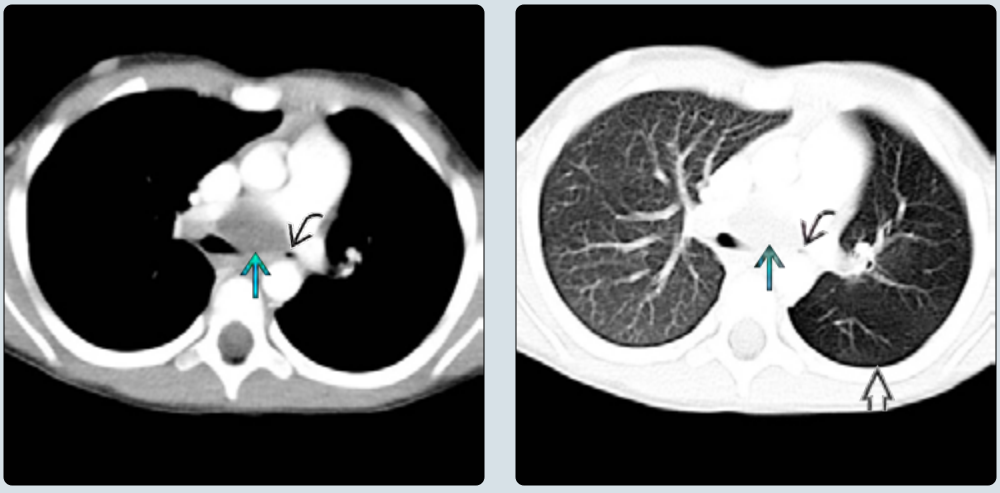
CLINICAL ISSUES

- Common presentations
  - In infants: Respiratory distress
  - In older children: Chest pain, dysphagia, recurrent infections
  - May be asymptomatic in older children & adults
- Definitive treatment: Surgical resection

(Left) *Chest radiograph of a 14-month-old boy reveals an ovoid left perihilar mass.* **(Right)** *Axial CECT of the same patient shows a well-circumscribed, homogeneous, fluid-attenuation mass partially encasing the aorta. This bronchogenic cyst extends posteriorly from the margin of the left main bronchus to the left paraspinal region. There is mild mass effect on the left main bronchus without significant narrowing.*



(Left) *Axial CECT of a 5-year-old girl with recurrent left lower lobe pneumonia shows a 2-cm, well-circumscribed, fluid-attenuation mass in a subcarinal location. There is marked narrowing of the left main bronchus.* **(Right)** *Axial lung window image from the same CECT shows diffuse hyperlucency of the left lung, indicating postobstructive air-trapping due to extrinsic compression of the left main bronchus by the bronchogenic cyst (with the cyst being poorly visualized on this setting).*



TERMINOLOGY

Definitions

- Congenital mass in family of foregut duplication cysts: Bronchogenic cysts (BC), enteric cysts, neurenteric cysts
- BC result from abnormal ventral budding of tracheobronchial tree between 26th & 40th days of gestation

IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed, round, middle mediastinal mass of soft tissue or water density
- Location
  - Mediastinal: 85%
    - 79% middle, 17% posterior, 3% anterior
    - Typically subcarinal, paratracheal, or hilar
  - Extramediastinal (usually pulmonary)
    - Majority in medial 1/3 of lower lobes
  - Rare occurrence in diaphragm, retroperitoneum, base of tongue, suprasternal or presternal soft tissues, pericardium or myocardium, subcutaneous tissues
- Size
  - 1-11 cm; average: 4.8 cm
- Morphology
  - Well-defined ovoid or round mass with smooth or slightly lobulated borders
  - Almost always unilocular
  - Communication with airway uncommon; occurs more frequently in pulmonary BC after infection
    - Air in mediastinal BC suggests ongoing infection
  - May compress airway or esophagus, leading to
    - Air-trapping (hyperinflation) or atelectasis
    - Dysphagia & vomiting

Radiographic Findings

- Radiography
  - Well-defined spherical mass with smooth borders
  - Soft tissue/fluid density
  - May be occult or subtle on radiography
  - Compression of adjacent bronchus may lead to hyperinflation or atelectasis

Fluoroscopic Findings

- Extrinsic mass may be found incidentally on upper GI series, compressing/distorting contrast-filled esophagus

CT Findings

- CECT
  - Sharply marginated mass with smooth borders
  - Cyst contents variable: Water or proteinaceous fluid, air
    - Mediastinal: 50% water attenuation, 50% soft tissue attenuation
    - Lung: Many lesions at least partially air filled
  - Ca^{2+} uncommon; may occur in wall or as layering calcium oxalate in cyst contents
  - Typically shows non- or minimally enhancing thin wall
    - More prominent wall enhancement & thickening may be seen with infection
    - Solid nodular components favor neoplasm
  - No central enhancement

MR Findings

- T1WI
  - Well-circumscribed, thin-walled cystic mass
    - Variable internal signal, often hyperintense to cerebrospinal fluid (CSF) (due to proteinaceous or hemorrhagic fluid)
- T2WI FS
  - Central contents have fluid signal intensity, similar to or brighter than CSF
    - Intermediate or dark T2 signal intensity could be due to hemorrhage or (rarely) Ca^{2+}
- DWI
  - Uncomplicated cyst should not restrict diffusion
- T1WI C+ FS
  - No central enhancement (but may be bright prior to contrast)
  - Thin wall may mildly enhance
  - Thicker enhancing wall implies infection

Ultrasonographic Findings

- Grayscale ultrasound
  - Well-circumscribed, round mass with posterior acoustic enhancement
  - Thin wall with variable echogenicity of internal contents
- Color Doppler
  - No internal vascularity
  - With inflammation, thickened cyst wall & surrounding tissues may be hyperemic
  - Can show relationship of BC to adjacent vessels

Imaging Recommendations

- Best imaging tool
  - If mass location allows adequate soft tissue acoustic window, ultrasound may demonstrate cystic nature of lesion without radiation or sedation
  - MR provides more comprehensive evaluation of lesion to exclude other diagnoses & determine full extent
    - Lung parenchyma & small airways not as well visualized as on CT
- Protocol advice
  - T2 FS or STIR MR to maximize contrast of cyst fluid against surrounding tissues
  - Consider additional MR sequences
    - Flow-sensitive sequence to exclude aneurysm/varix, show exact relationship to mediastinal/hilar vessels
    - Precontrast T1 FS in single plane for subtraction from postcontrast sequence
      - Mere addition of FS to sequences can change intrinsic signal characteristics in cystic lesions without true enhancement
      - Therefore, subtraction provides most accurate assessment to confirm lack of lesion enhancement

DIFFERENTIAL DIAGNOSIS

Esophageal Duplication Cyst

- Identical imaging to BC, though duplication cyst may have thicker wall (as smooth muscle always present)

Neurenteric Cyst

- Cyst communicates with spinal canal
- Associated vertebral anomaly in ~ 50%

Round Pneumonia

- Well-circumscribed lung opacity in child < 8 years of age
- Air bronchograms confirm airspace disease
- No mass effect on adjacent structures
- Follow-up radiographs will show resolution after antibiotics

Neuroblastoma

- Moderately enhancing solid paraspinal mass
- Intermediate T2 signal intensity; frequently calcified
- ± adjacent rib & vertebral changes, intraspinal extension

Lymphadenopathy

- Generally multifocal, ovoid, solid appearing
- Necrotic lymph nodes
  - Usually have associated pulmonary findings of infection (e.g., histoplasmosis or tuberculosis)
  - Thick, irregular rim of enhancement
  - Other nearby nodes usually enlarged but not necrotic

Lymphatic Malformation

- Multilobulated, multiseptated cystic mass
- Infiltrates across multiple soft tissue compartments
  - Neck, chest wall, upper extremity
- Surrounds & displaces adjacent structures
- Macrocysts may contain fluid-fluid levels from hemorrhage

Congenital Pulmonary Airway Malformation

- Heterogeneous, multicystic lesion with architectural distortion of lung parenchyma
- Individual cysts in lesion vary widely in size
- Initially fluid-filled at birth with subsequent aeration

Bronchopulmonary Sequestration

- Heterogeneous cystic &/or solid mass
- Borders tend to be less well defined
- Systemic feeding vessel from aorta pathognomonic

PATHOLOGY

General Features

- Etiology
  - Congenital lesions that develop from abnormal budding of ventral foregut
    - Early budding results in mediastinal cysts
    - Later budding results in lung parenchymal cysts
- Associated abnormalities
  - Occasional association with other congenital cardiopulmonary malformations
    - Bronchopulmonary sequestration (BPS), lobar overinflation, diaphragmatic hernia
    - Pericardial defect, congenital heart disease

Gross Pathologic & Surgical Features

- Well circumscribed
- Almost 50% have stalk connecting lesion to trachea/carina, pleura, or esophagus
- Typically lacks communication with airway
- Contents usually thick or gelatinous fluid

Microscopic Features

- Lined by ciliated respiratory epithelium
- May contain bronchial glands, smooth muscle, bronchial cartilage, or (rarely) gastric mucosa

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Infants: Respiratory distress
  - Older children: Chest pain, dysphagia, recurrent infections
  - Symptoms more common in mediastinal lesions
- Other signs/symptoms
  - Asymptomatic cysts may be discovered incidentally in older children & adults
    - Up to 45% develop symptoms eventually
  - May be detected in utero ± mass effect
    - Can compromise airway, lead to overinflation of adjacent lung
  - Cervical & subcutaneous lesions frequently present as palpable masses

Demographics

- Age
  - May present at any age but usually in childhood
- Gender
  - No sex predilection
- Epidemiology
  - Frequency unknown due to asymptomatic population

Natural History & Prognosis

- Propensity for infection
- Rare case reports of malignancy arising in lesions

Treatment

- Surgical resection recommended in children
- Conservative treatment in adults or children considered to be of high surgical risk

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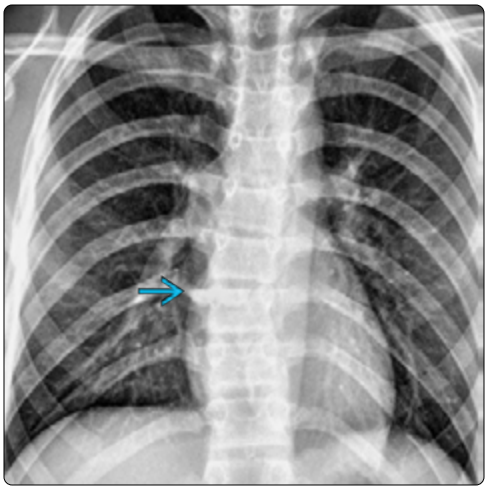
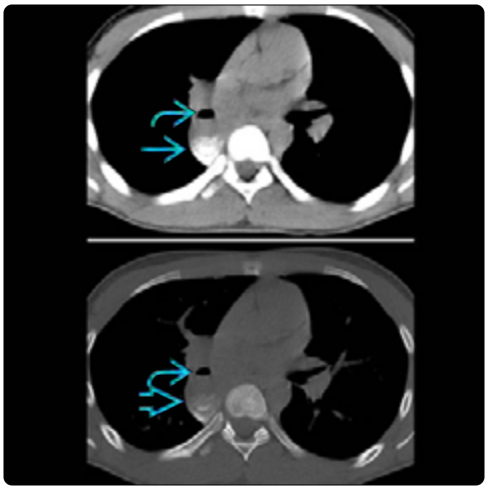
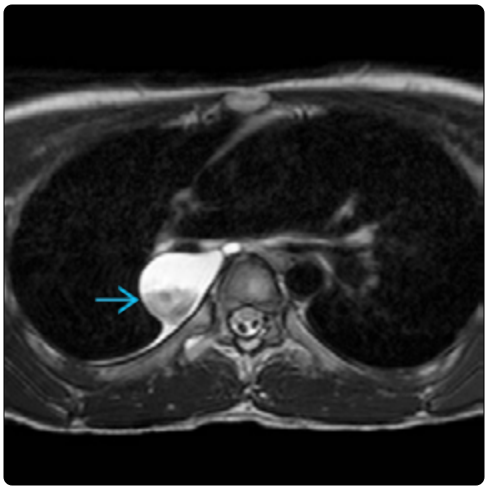
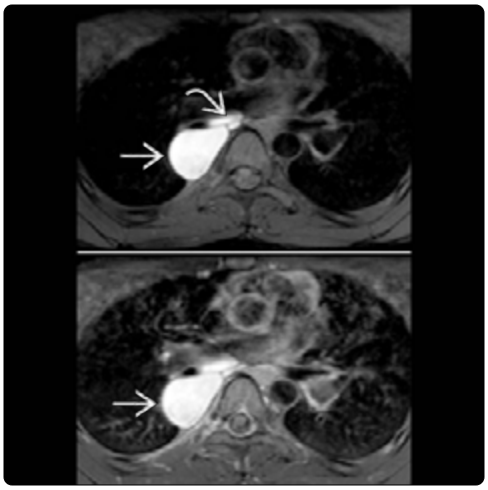


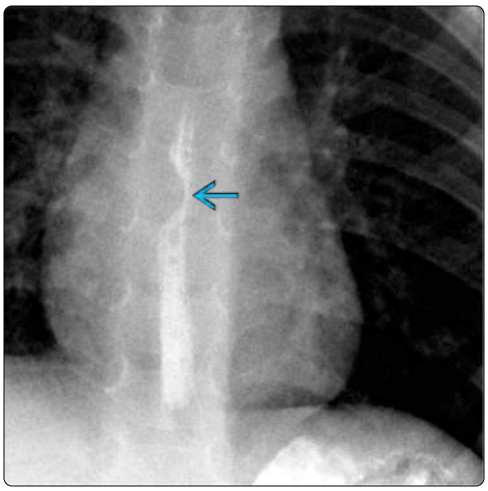


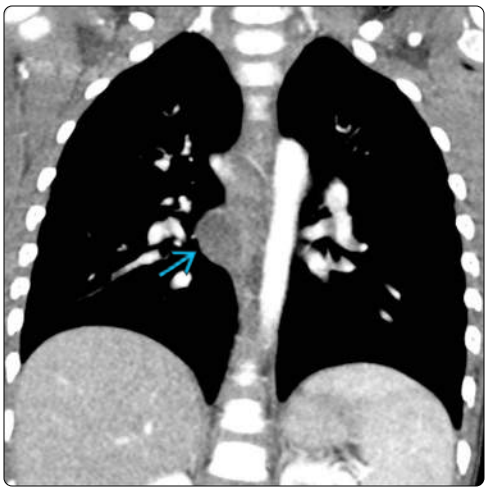
(Left) *Upright frontal radiograph in a 14-year-old boy with suspected scoliosis shows calcifications layering dependently in an otherwise occult lesion.* **(Right)** *Axial NECT images in the same patient show dependent positioning of the CA²⁺ within the otherwise fluid-attenuating mass (seen here in both soft tissue & bone windows). The mass is intimately related to the bronchus intermedius. Of note, CA²⁺ are uncommon in bronchogenic cysts.*





(Left) *Axial T2 MR in the same patient shows dependently layering low signal intensity material (corresponding to the known CA²⁺) within the rounded mass.* **(Right)** *Axial T1 FS MR images in the same patient before (top) & after (bottom) contrast administration show no significant change in the mass. Note the intrinsically bright material within the mass, likely due to complex proteinaceous fluid. Small foci of the lesion extend posterior to the pulmonary artery. A bronchogenic cyst was confirmed upon resection.*





(Left) *Frontal esophagram image from a 3-year-old patient with choking episodes shows a round, relatively lucent mass extrinsically compressing the contrast-filled mid esophagus from the right. The lesion was occult radiographically.* **(Right)** *Coronal CECT in the same patient shows the round mass to be of fluid-attenuation with minimal rim enhancement. This bronchogenic cyst was directly apposed to the bronchus intermedius.*

Congenital Lobar Overinflation

KEY FACTS

TERMINOLOGY

- Congenital abnormality of lower airways that results in progressive overexpansion of pulmonary lobe
- Considered final, common pathway of various disruptions of bronchopulmonary development
- Synonyms: Congenital lobar emphysema, congenital lobar hyperinflation, infantile lobar emphysema

IMAGING

- Hyperlucent & hyperexpanded lobe in neonate
- Lobar predilection: Left upper lobe > right middle lobe > right upper lobe > > lower lobes
- Mass effect: Compression of adjacent lung, mediastinal shift; persistent hyperinflation in ipsilateral decubitus position
- Diagnosis typically made by chest radiography
- CTA performed to confirm diagnosis, elucidate cause, exclude other causes of hyperlucent lung, define extent of disease

TOP DIFFERENTIAL DIAGNOSES

- Pneumothorax
- Congenital pulmonary airway malformation
- Bronchial atresia

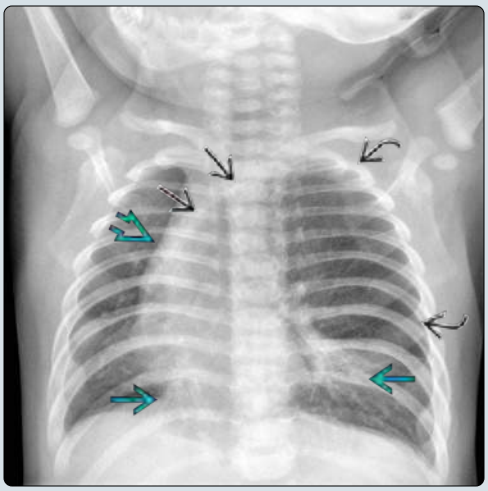
PATHOLOGY

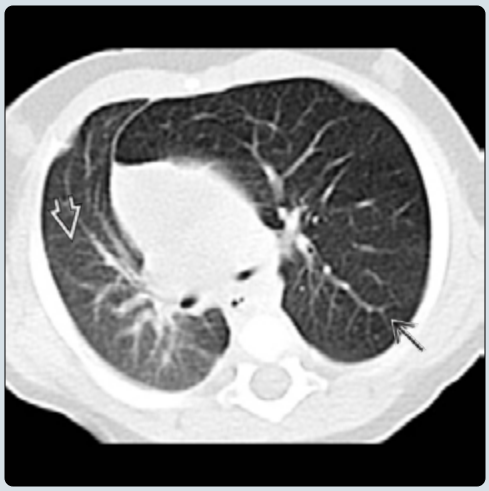
- Cause found in ~ 50% of cases
  - Wall abnormality, lumen obstruction, extrinsic compression

CLINICAL ISSUES

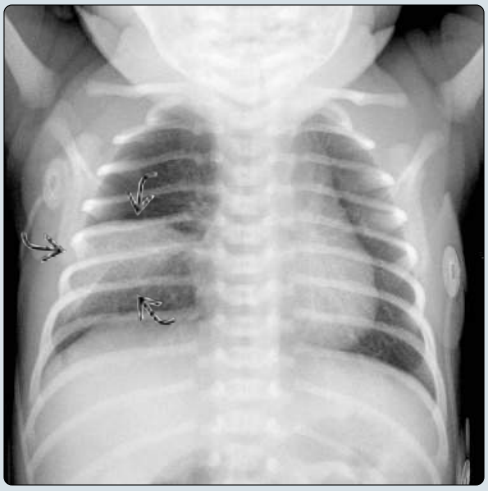
- Respiratory distress in neonatal period
- May cause progressive respiratory distress → can be fatal if not resected
- Treatment
  - Bronchoscopy to exclude endobronchial lesion
  - Surgical lobectomy

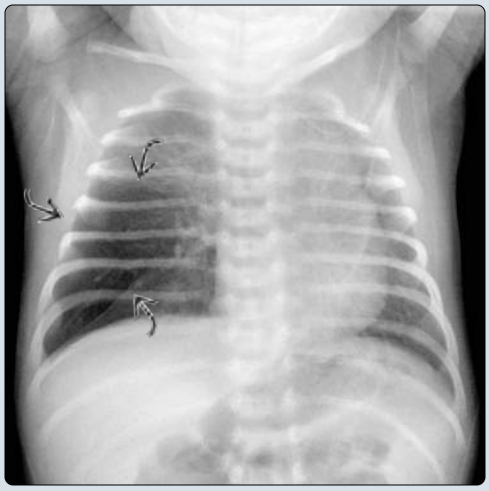
(Left) *Frontal radiograph of an infant with chronic wheezing shows an abnormally lucent left upper lobe that is so hyperexpanded that it herniates across the midline. Note the mass effect causing mediastinal shift & lower lobe atelectasis.* **(Right)** *Axial NECT from the same patient reveals smaller vessels in the affected hyperexpanded & hyperlucent left upper lobe with compressive atelectasis in the right lung.*





(Left) *Chest radiograph obtained in the 1st few hours of life demonstrates segmental airspace opacification of the right middle lobe. The opacity reflects retained fetal fluid filling alveoli in the affected segment.* **(Right)** *A follow-up radiograph the next day shows that the same segment is now gas filled, quite lucent, & hyperexpanded. The retained fetal fluid has been resorbed with the findings now reflecting air-trapping.*





TERMINOLOGY

Abbreviations

- Congenital lobar overinflation (CLO)

Synonyms

- Congenital lobar emphysema, congenital lobar hyperinflation, infantile lobar emphysema

Definitions

- Congenital abnormality of lower airways that results in progressive overexpansion of pulmonary lobe
- Considered final, common pathway of various disruptions of bronchopulmonary development
  - 50% idiopathic
  - 25% due to intrinsic cartilaginous abnormality
  - 25% due to extrinsic compression

IMAGING

General Features

- Best diagnostic clue
  - Neonate with overexpanded, lucent pulmonary lobe
    - Especially left upper lobe
    - May be fluid-filled in 1st few hours after birth
- Location
  - Lobar predilection
    - Left upper lobe: 40%
    - Right middle lobe: 35%
    - Right upper lobe: 20%
    - Lower lobes: 5%
      - May involve ≥ 1 lobe or only segment

Radiographic Findings

- Radiography
  - Classic: Radiodense lobe becomes progressively hyperlucent & hyperexpanded
    - May have reticular pattern as fluid clears via distended lymphatics
  - Classic appearance uncommon
    - CLO usually detected as hyperexpanded lobe
  - Pulmonary vessels may appear attenuated & uniformly splayed
  - Mass effect
    - Compression of ipsilateral lung
    - Mediastinal shift with possible tracheal deviation & compression of contralateral lung
    - Deviation of anterior junction line
    - Occasional rib separation & diaphragm depression
    - Persistent hyperinflation in ipsilateral decubitus position

CT Findings

- CTA
  - IV contrast used to evaluate vascular structures & aid in evaluating other abnormalities (e.g., sequestration, congenital pulmonary airway malformation, bronchogenic cyst)
  - Hyperinflation with uniform hyperlucency of affected lung parenchyma (reflecting distended alveoli)
  - Attenuated vessels: Smaller & more widely spaced than those in adjacent lung
    - Vessels not focally splayed as by cystic mass

MR Findings

- Fetal MR
  - Diffusely high T2 signal of expanded lobe
  - Mass effect with compression of ipsilateral remaining lung & mediastinal deviation
  - Location of lesion helpful, but mass effect can make localizing lesion difficult

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal ultrasound: Echogenic upper lobe mass
  - Postnatal ultrasound: Typically only used if lobe still fluid filled

Nuclear Medicine Findings

- V/Q scan
  - Ventilation initially ↓, then isotope retained in affected lobe
  - Matching ↓ perfusion in same distribution

Imaging Recommendations

- Best imaging tool
  - Diagnosis typically made by serial chest radiographs
  - CTA performed to
    - Confirm diagnosis
    - Exclude other neonatal hyperlucent lung lesions
    - Evaluate for causal obstructing mass
    - Define extent of disease

DIFFERENTIAL DIAGNOSIS

Pneumothorax

- Pleural line or other clues confirming abnormal gas collection of pleural space
  - No lung markings beyond pleural line
  - Lobes collapse, not expand

Persistent Pulmonary Interstitial Emphysema

- In rare cases, persistent pulmonary interstitial emphysema can present as expanding hyperlucent mass
- Pulmonary vessels & bronchi surrounded by air

Pulmonary Hypoplasia

- Affected lung is small
- Ipsilateral bronchus is small or absent
- No air-trapping

Swyer-James Syndrome

- Presents later in childhood after viral infection
- Often affects > 1 lobe

Bronchial Atresia

- Central tubular, rounded, or branching mass

Congenital Pulmonary Airway Malformation

- Variable-sized, gas-filled cysts

PATHOLOGY

General Features

- Etiology
  - Cause found in ~ 50% of cases

- Most likely related to bronchial obstruction with ball valve phenomenon
  - Air enters involved region but has difficulty leaving → progressive hyperinflation
- Wall abnormality
  - Deficient, immature, or dysplastic bronchial cartilage
  - Redundant bronchial mucosal folds
  - Stenotic or kinked bronchus
- Lumen obstruction
  - Inspissated mucus plug
  - Mucosal web or fold
- Extrinsic compression
  - Pectus excavatum
  - Foregut duplication cyst
  - Vascular anomaly
    - Pulmonary arterial sling
    - Tetralogy of Fallot with absent pulmonary valve
    - Patent ductus arteriosus
    - Pulmonic stenosis
    - Dilated superior vena cava with anomalous pulmonary venous return
- Associated abnormalities
  - ~ 15% have cardiac anomalies
  - May also have renal, GI, & MSK abnormalities

Staging, Grading, & Classification

- 2 forms
  - Hypoalveolar → normal or fewer alveoli than expected
  - Polyalveolar → more alveoli than expected

Gross Pathologic & Surgical Features

- Hyperexpanded lobe is rounded
- Sponge-like appearance
- Resected lobe does not deflate
  - Compressed ipsilateral lobe will reinflate & eventually compensatorily hyperinflate

Microscopic Features

- Dilated alveoli
- Alveolar walls are thinned but intact
  - Not true emphysema (hence effort to abandon congenital lobar emphysema nomenclature)
- Defective cartilage in bronchial walls seen in < 50%

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Respiratory distress in neonatal period
  - May be progressive
- Other signs/symptoms
  - Asymmetry of movement of chest with respiration
  - Use of accessory muscles of respiration
  - Decreased breath sounds on affected side
  - Hyperresonant hemithorax

Demographics

- Age
  - May be diagnosed in utero
  - Majority become symptomatic in neonatal period & infancy
    - 50% present in first 4 weeks; 75% in first 6 months
  - May present with symptoms later in childhood or may be incidental finding
- Gender
  - M:F = 1.8:1.0
- Ethnicity
  - More common in Caucasians

Natural History & Prognosis

- Fluid seen in neonatal period is removed by lymphatics & capillary reabsorption
- Lobe subsequently filled with air by collateral air drift
- May cause progressive respiratory distress as lobe continues to expand & cause lung compression → can be fatal if not resected
- Some patients have minor symptoms & can be observed
- Over time, involved lobe becomes small with diminished markings
- No increased risk of malignancy or infection

Treatment

- Bronchoscopy to exclude endobronchial lesion
- Electively intubate opposite lung
- Place patient in ipsilateral decubitus position
- Surgical lobectomy
  - May be needed emergently if progressive hyperinflation occurs → can be life threatening
- Conservative treatment has been advocated for patients with minimal symptoms

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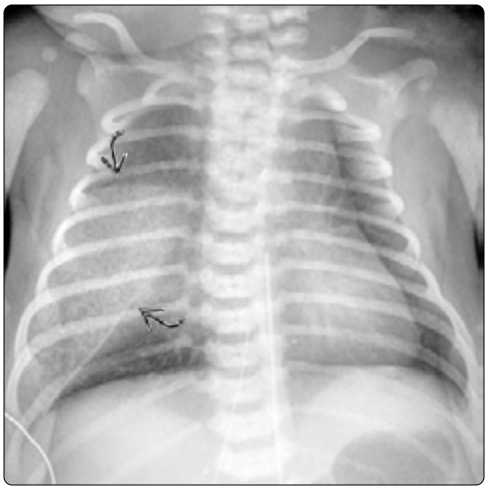
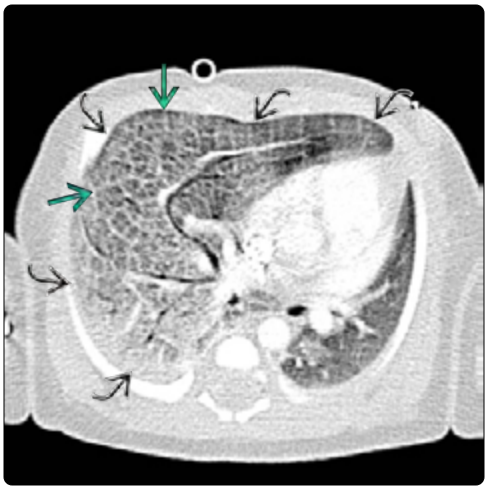
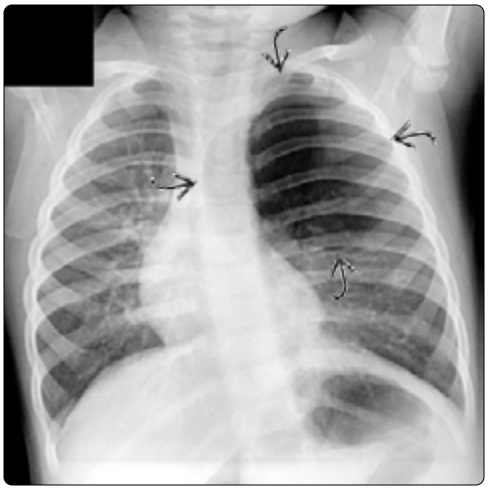
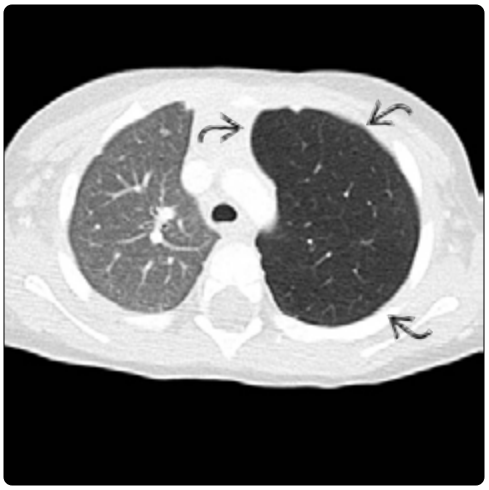


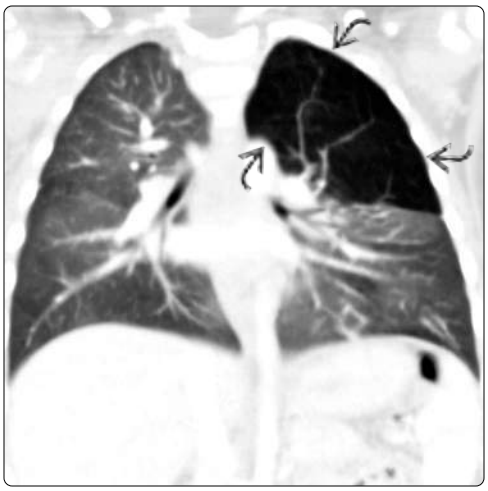


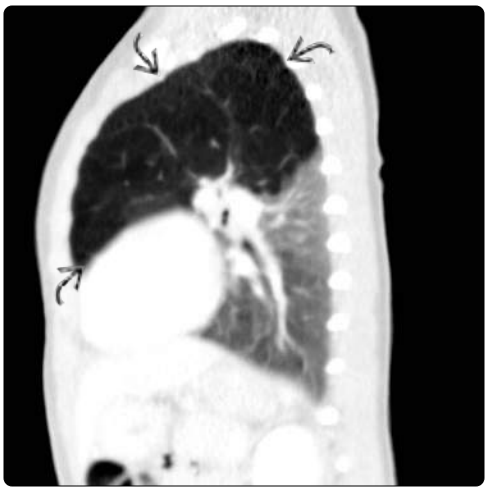
(Left) *Chest radiograph of congenital lobar overinflation (CLO) during the period when fetal fluid has not fully been resorbed, but air-trapping has already begun, shows a picture of lobar hyperexpansion with distinct reticular interstitial markings.* **(Right)** *Axial CECT in the same patient shows how the right middle lobe is hyperexpanded & herniates across the midline. The radiographic reticular pattern resulted from fluid engorgement of lymphatics causing interlobular septal thickening.*





(Left) *AP radiograph shows marked lucency & mass effect from air-trapping in the left upper lobe.* **(Right)** *Axial CECT from the same patient shows that the affected lobe is overexpanded & lower in attenuation compared to the normal right lung. Also note the relative oligemia within the left upper lobe.*





(Left) *Coronal CECT from the same patient again demonstrates relative overexpansion, hyperlucency, & oligemia.* **(Right)** *The same findings of left upper lobe overexpansion, hyperlucency, & oligemia are shown on this sagittal CECT.*

Bronchial Atresia

KEY FACTS

TERMINOLOGY

- Focal interruption of bronchus → distal mucoid impaction (bronchocele/mucocele) & hyperinflation of distal lung
  - ± hyperplasia of distal lung

IMAGING

- Classic appearance: Ovoid/round or tubular branching mass (finger-in-glove sign) with hyperinflated lung distally
- Neonates & infants may present with atelectatic, fluid-filled segment that gradually fills with air
- Protocol: CTA with multiplanar reconstructions
  - Fluid-attenuation round or tubular branching mass
  - Hyperinflated distal lung + ↓ vascularity
  - No systemic feeding artery
  - Expiratory images helpful to evaluate air-trapping
- May occur in any lobe/segment/subsegment

TOP DIFFERENTIAL DIAGNOSES

- Congenital lobar overinflation (CLO)
- Congenital pulmonary airway malformation (CPAM)
- Bronchogenic cyst
- Bronchopulmonary sequestration (BPS)
- Allergic bronchopulmonary aspergillosis
- Scimitar syndrome (hypogenetic lung syndrome)

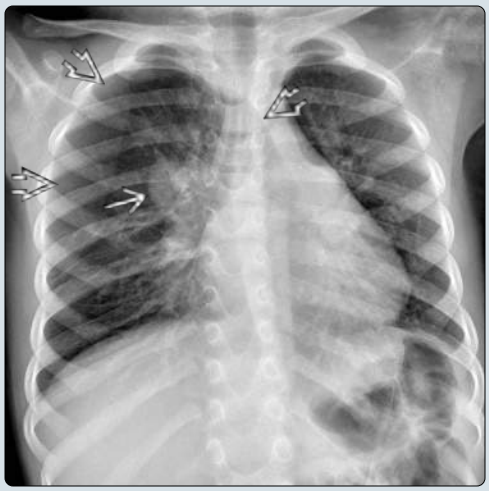
PATHOLOGY

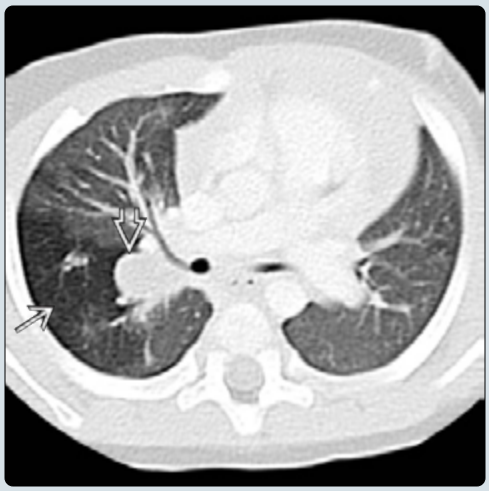
- Within spectrum of other congenital lung lesions (CLO, CPAM, BPS, bronchogenic cyst)
  - Components of atretic bronchi frequently found in other congenital lung lesions
- Aeration of distal lung occurs through collateral air drift

CLINICAL ISSUES

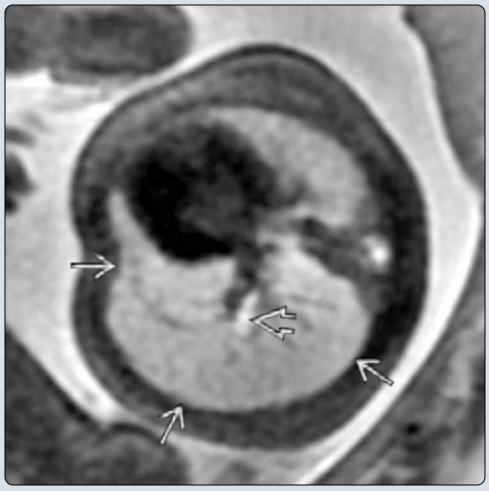
- Diagnosed at any age
  - Older children/adults (majority) often asymptomatic
  - May present with recurrent respiratory tract infections, chronic cough, dyspnea, wheezing in early childhood
- Surgical resection usually reserved for recurrent infections

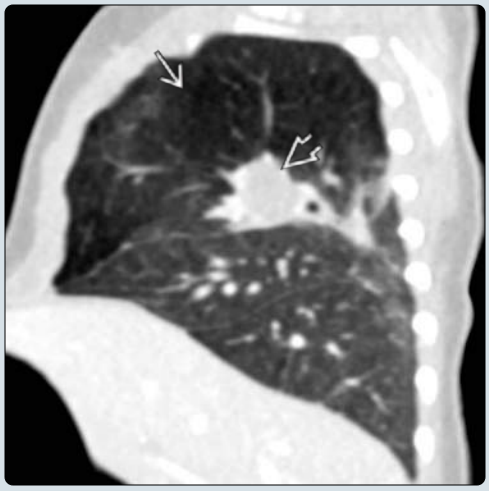
(Left) *AP radiograph in a 3 year old with a cough & diminished breath sounds shows a round perihilar mass-like opacity ➡ due to a dilated, atretic right upper lobe (RUL) segmental bronchus. Note the relatively lucent & hyperexpanded RUL ➡.* **(Right)** *Axial CECT (in lung windows) in the same patient confirms the findings of segmental RUL bronchial atresia. Note the RUL hyperinflation ➡ due to collateral air drift beyond the obstructed & dilated RUL bronchus (bronchocele) ➡.*





(Left) *Axial SSFSE T2 fetal MR shows hyperintense signal & expansion of the RUL ➡. Note the discrete hyperintense tubular structure in the right hilum ➡ representing the fluid-distended atretic RUL bronchus.* **(Right)** *Sagittal CECT after birth in the same patient shows the fluid-distended atretic right upper lobe bronchus ➡ & the hyperinflated right upper lobe ➡.*





TERMINOLOGY

Definitions

- Focal interruption of main, lobar, segmental, or subsegmental bronchus with associated mucoid impaction (bronchocele/mucocele) & hyperinflation of distal lung
  - ± hyperplasia of distal lung depending on level of bronchial obstruction

IMAGING

General Features

- Best diagnostic clue
  - Round or tubular branching mass (finger-in-glove sign) + hyperinflated distal lung

Radiographic Findings

- Neonates & infants
  - Presentation more variable than in children & adults
  - Lobe or segment may be atelectatic or fluid-filled initially → gradually fills with air
  - Hyperinflation less frequent than in children & adults
- Children & adults
  - Radiating, branching, tubular opacity (mucoid impaction of dilated airway) distal to atretic bronchus
  - More central perihilar round or ovoid mass (bronchocele)
  - Distal lung hyperinflated with ↓ vascularity

CT Findings

- CTA
  - Fluid-attenuation round or tubular branching mass
  - Hyperinflated distal lung + ↓ vascularity
    - Expiratory images confirm air-trapping

MR Findings

- Fetal MR
  - Discrete & uniformly T2 hyperintense solid lung mass with varying degrees of mass effect
  - Dilated fluid-filled bronchus in medial/central aspect of lesion helps differentiate from other lesions

Ultrasonographic Findings

- Obstetric ultrasound
  - Homogeneously hyperechoic segment or lobe
  - May see dilated, fluid-filled bronchi

Imaging Recommendations

- Best imaging tool
  - CTA confirms that tubular opacity represents impacted atretic bronchus & not vessel
    - Expiratory images can confirm air-trapping

DIFFERENTIAL DIAGNOSIS

Congenital Lobar Overinflation (CLO)

- Overlapping appearance; no mass/bronchocele in CLO
- Usually involves entire lobe with progressive hyperinflation

Congenital Pulmonary Airway Malformation (CPAM)

- CPAM often heterogeneous due to macroscopic cysts with distortion/splaying of residual lung
- No associated bronchial dilation/impaction

Bronchogenic Cyst

- Discrete round (nonbranching) fluid-filled cyst in central lung or mediastinum

Bronchopulmonary Sequestration

- Systemic arterial supply, usually from aorta
- Usually in left lower lobe
- May present beyond infancy with recurrent infections

Allergic Bronchopulmonary Aspergillosis

- FInger-in-glove sign often present
- Frequently bilateral with widespread bronchiectasis

Scimitar Syndrome (Hypogenetic Lung Syndrome)

- Hypoplastic lung with partial anomalous pulmonary venous drainage
  - Typically right lower lobe with curvilinear opacity (vein) directed toward medial right hemidiaphragm

Slow-Growing Endobronchial Tumor

- Mass usually smaller than mucoid impaction
- Distal lung usually atelectatic, not hyperinflated

PATHOLOGY

General Features

- Etiology
  - Occurs between 5th-15th weeks of gestation, though exact mechanism unclear
    - Intrauterine interruption of arterial supply to bronchus → scarring of primitive bronchus
    - Disconnection of primitive bronchial cells at tip of bronchial bud with continued normal growth distally
  - Aeration of distal lung through collateral air drift
- Associated abnormalities
  - Components of atretic bronchi frequently found in other congenital lung lesions

CLINICAL ISSUES

Presentation

- Congenital lesion but may be diagnosed at any age
  - Older children/adults (majority) often asymptomatic
  - Infancy, early childhood (< 3 years): Recurrent respiratory tract infections, chronic cough, dyspnea, wheezing
  - Uncommon main bronchus atresia can be lethal in utero

Treatment

- Surgical resection typically reserved for patients with repeated infections

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Esophageal Atresia and Tracheoesophageal Fistula

KEY FACTS

TERMINOLOGY

- Atresia: Congenital occlusion of upper esophagus
- Fistula: Anomalous congenital connection(s) from esophagus to trachea

IMAGING

- 5 major anatomic variations of esophageal atresia (EA)-tracheoesophageal fistula (EA-TEF)
- Fistula level variable depending on type of EA-TEF
  - Most commonly above/near carina
- Atretic segments variable in length
  - Gap often long in EA without TEF
- Radiographs
  - Air-distended upper esophageal pouch
  - Enteric tube tip near thoracic inlet in pouch
  - EA with TEF: Gas in stomach & bowel
  - EA without TEF: Gasless stomach & bowel
- Limited indications for preoperative esophagram (except isolated TEF)
- Postoperative esophagram
  - Esophageal anastomotic leak, anastomotic stricture, recurrent/additional TEF, esophageal dysmotility, gastroesophageal reflux

TOP DIFFERENTIAL DIAGNOSES

- Tube malposition
- Laryngotracheal cleft
- Chronic respiratory issues (mimicking H-type TEF)
- Esophageal strictures of various etiologies
- Extrinsic esophageal compression

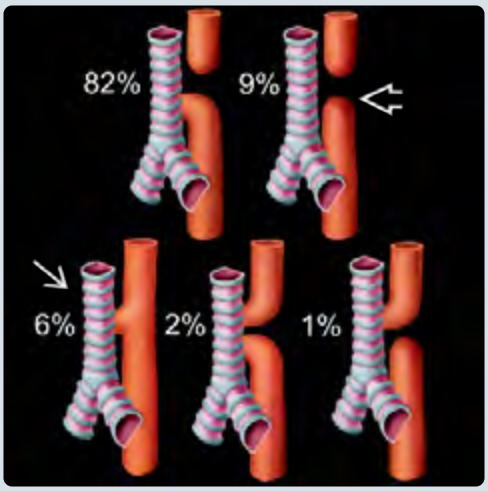
PATHOLOGY

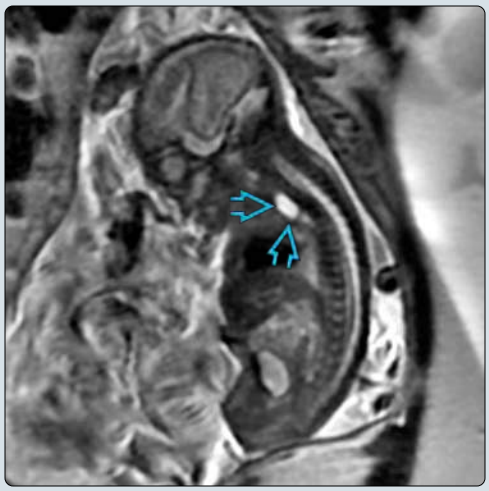
- 47-75% have associated anomalies

CLINICAL ISSUES

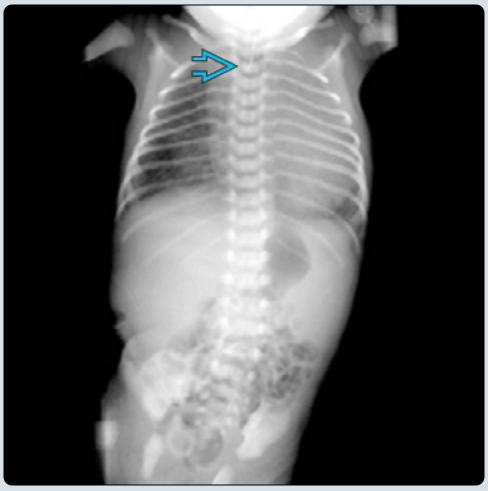
- Presentation: Excessive oral secretions, cyanosis, choking, coughing during feeding
- Postsurgical survival: 75-95% (depends on associated cardiac anomalies, birth weight)

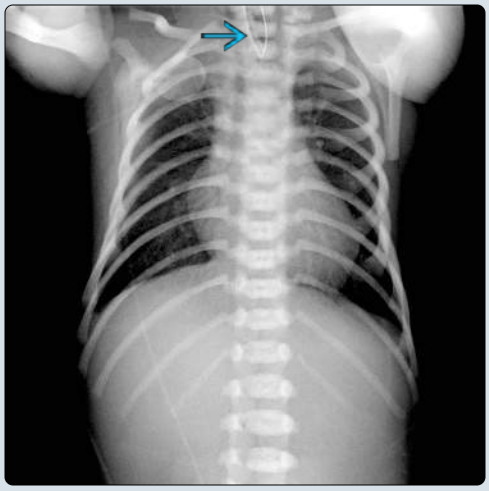
(Left) *Graphic shows the 5 main types of esophageal atresia with tracheoesophageal fistula (EA-TEF), including the isolated TEF without EA (H-type fistula) ⇨ & the isolated EA without TEF ⇨.* **(Right)** *Sagittal T2 SSFSE MR of a 26-week gestation fetus imaged for polyhydramnios shows a collection of fluid in the region of the upper esophagus ⇨ that is suggestive of an atretic esophageal pouch, which was confirmed during surgery in the newborn period.*





(Left) *AP radiograph of a premature newborn male with a history of not handling secretions shows a nasogastric tube (NGT) tip ⇨ overlying the thoracic inlet due to EA. The bowel gas confirms the presence of a distal TEF. Cardiomegaly suggests congenital heart disease.* **(Right)** *AP chest radiograph in a newborn shows a NGT coiled at the thoracic inlet ⇨ suggesting EA. Note the lack of gas in the upper abdomen, which is typical of EA without a distal TEF.*





TERMINOLOGY

Definitions

- Esophageal atresia (EA): Congenital occlusion of upper esophagus
- Tracheoesophageal fistula (TEF): Single (less commonly multiple) anomalous congenital connection(s) from esophagus to trachea

IMAGING

General Features

- Best diagnostic clue
  - Enteric tube tip in gas-distended proximal esophageal pouch on newborn chest radiograph
- Location
  - Fistula level variable depending on type of EA-TEF
    - Most commonly above/near carina
    - Higher levels with multiple fistulas
    - Rarely esophagobronchial fistula
  - Atresia usually above junction of superior & middle 1/3 of esophagus
- Size
  - TEF usually small
  - Atretic segments of esophagus variable in length
    - Gap often long in EA without TEF
- Morphology
  - 5 major anatomic variations of EA-TEF

Radiographic Findings

- Radiography
  - Intermittently air-distended, dilated upper esophageal pouch
  - Enteric tube tip near thoracic inlet in pouch
  - Gas in stomach & bowel: EA with TEF
  - Gasless stomach & bowel: EA without TEF
  - Displaced, bowed, narrowed trachea due to dilated esophageal pouch & tracheomalacia
  - Signs of other congenital anomalies
    - Cardiomegaly, abnormal pulmonary vascularity, vertebral anomalies, dilated bowel
  - Side of aortic arch important for surgical repair (thoracotomy performed opposite side of arch)

Fluoroscopic Findings

- Rarely needed for diagnosis of EA (clinical + radiographic diagnosis)
  - Confirm blind-ending pouch: Inject air into nasogastric tube (NGT)
- Contrast esophagram useful preoperatively for
  - H-type TEF without EA (clinical, radiographic diagnosis less clear in this type)
    - Oblique fistula from esophagus craniad to trachea (looks like "N")
  - EA without TEF (often long gap)
    - Often 2-stage repair if long gap
    - Preoperative measurement of gap
      - Surgical bougie into upper pouch ± contrast
      - Surgical bougie into lower pouch ± contrast via gastrostomy
      - Measure gap prior to 2nd surgery
- Esophagram/upper GI after EA-TEF repair (using low-osmolality water-soluble contrast)
  - Esophageal anastomotic leak (early complication)
  - Recurrent or additional TEF
  - Esophageal dysmotility in nearly 100%
  - Gastroesophageal reflux (GER)
  - Malrotation
  - Esophageal anastomotic stricture (delayed complication)

Ultrasonographic Findings

- Prenatal ultrasound: Nonvisualization of fetal stomach, intermittent filling of proximal esophageal pouch, polyhydramnios
  - Low-detection rate of EA by US
  - MR may ↑ upper pouch visualization

MR Findings

- Fetal: Fluid signal in intermittently distended esophageal pouch ± decompressed stomach, associated anomalies, polyhydramnios

Imaging Recommendations

- Best imaging tool
  - Radiographs ± air injection of NGT for EA
  - Esophagram for suspected isolated TEF or postoperative complication
- Protocol advice
  - Esophagram/upper GI for isolated (H-type) TEF
    - Lateral position for H type
    - Barium by mouth in most cases (tube rarely needed)
    - If contrast injected, 5 or 8 French feeding tube sideports at mid to distal trachea
    - Pulsed fluoroscopy continuously in lateral position coned from pharynx to below carina
    - No exposures until convinced: Normal vs. fistula
      - ± exposures in multiple positions
    - Do not mistake aspiration for missed fistula
    - Image stomach to duodenojejunal junction to exclude malrotation, if patient stable
  - Postoperative esophagram: Evaluate for leak or other complication
    - At least 1 scout image (± other positioning)
    - By mouth vs. tube injection of low-osmolality nonionic water-soluble contrast in orthogonal views
      - If by tube, position new tube above anastomosis (rather than pulling critical catheter, currently in stomach, back across anastomosis)
      - Limited distention of pouch actually needed to detect leak
    - If no leak with water-soluble contrast, consider using barium to ↑ detection of subtle leaks
    - Rereview images at workstation after study
      - May miss subtle leak on real-time imaging

DIFFERENTIAL DIAGNOSIS

Tube Malposition

- Traumatic pharyngeal perforation with NGT into mediastinum
  - Inject air or low-osmolality water-soluble contrast via NGT to differentiate air-filled pharyngeal pouch vs. pneumomediastinum

Laryngotracheal Cleft

- High fistulous connection, variable length

Chronic Respiratory Issues (Mimicking Occult H-Type TEF)

- GER with aspiration
- Bronchial foreign body
- Cystic fibrosis
- Infected congenital lung lesion

Esophageal Strictures

- Caustic injury, eosinophilic esophagitis, prior foreign body, epidermolysis bullosa, mediastinal radiation

Extrinsic Esophageal Compression

- Vascular rings
- Mediastinal masses

PATHOLOGY

General Features

- Etiology
  - Likely faulty division of foregut into trachea & esophagus in 1st month of gestation
- Genetics
  - Chromosomal anomalies in 2-10%
    - Trisomy 18, trisomy 21 most common
- Associated abnormalities
  - 47-75% have associated anomalies
    - Musculoskeletal: 14-24%
    - Cardiovascular: 11-55%
    - Gastrointestinal: 8-27%
    - Genitourinary: 8-21%
    - Craniofacial: 10%
    - Neurologic: 3-15%
    - Pulmonary: 2-6%
    - VACTERL (vertebral, anal, cardiac, tracheoesophageal, renal, limb anomalies): 10-20%

Staging, Grading, & Classification

- Many classifications by letters or numbers
  - Best to be descriptive rather than using letters/numbers for types
- Most widely cited surgical classification (Gross)
  - Type A: EA with no TEF (7-9%)
  - Type B: EA with proximal TEF (1%)
  - Type C: EA with distal TEF (82-86%)
  - Type D: EA with proximal & distal TEF (2%)
  - Type E: Isolated (H-type) TEF (no EA) (4-6%)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Excessive oral secretions, cyanosis, choking, coughing during 1st attempts at feeding
- Other signs/symptoms
  - Enteric tube fails to reach stomach
  - Recurrent pneumonia, dysphagia (H-type TEF)

Demographics

- Age
  - Newborn or early child (isolated H type)
  - 20-35% premature
- Epidemiology
  - 1:3,000 live births

Natural History & Prognosis

- Postoperative survival: 75-95% (depends on associated cardiac anomalies, birth weight)

Treatment

- Surgical
  - Bronchoscopy, esophagoscopy to visualize fistula(s) + extrapleural ligation of fistula + anastomosis of esophageal segments
  - If EA without TEF, esophageal ends often far apart
    - 1st surgery: Place gastrostomy tube
    - 2nd surgery months later: Connect esophagus after growth
    - Ultralong gap requires conduit
      - Colonic interposition vs. gastric pull-up/tube
    - Foker procedure: Actively stretches esophagus over time → primary anastomosis
- Early postsurgical complications
  - Anastomotic leak (up to 15%; most small leaks resolve spontaneously)
  - Additional TEF not seen initially
- Longer term postsurgical issues
  - Anastomotic stricture (18-50%)
    - ↑ with ↑ EA gap length, anastomotic tension, GER
    - Treated with repeated balloon or bougie dilations
  - Recurrent TEF (up to 10% of cases)
  - Esophageal dysmotility (nearly 100%)
  - GER; esophagitis (51%), Barrett esophagus (6%)
    - Surgical treatment (Nissen) if medical therapy fails
  - Respiratory infections
  - Tracheomalacia (10-20%)

DIAGNOSTIC CHECKLIST

Consider

- Is there distal bowel gas?
- Are there associated anomalies?

Image Interpretation Pearls

- Always rereview postoperative fluoroscopic studies at workstation for subtle leak or fistula not seen real time

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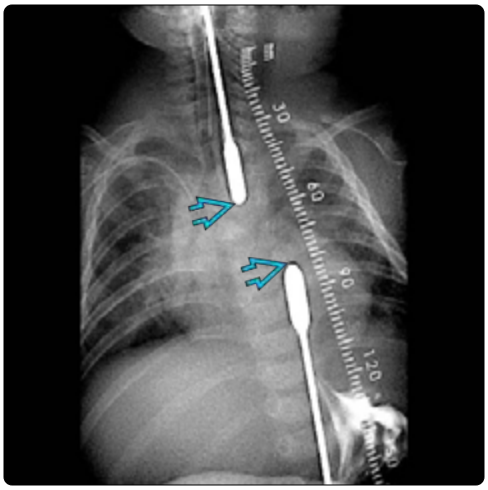
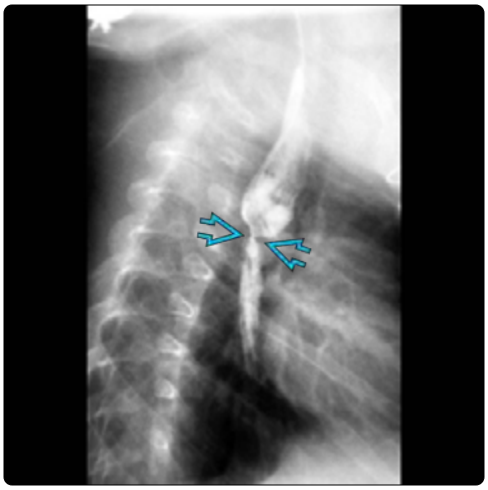
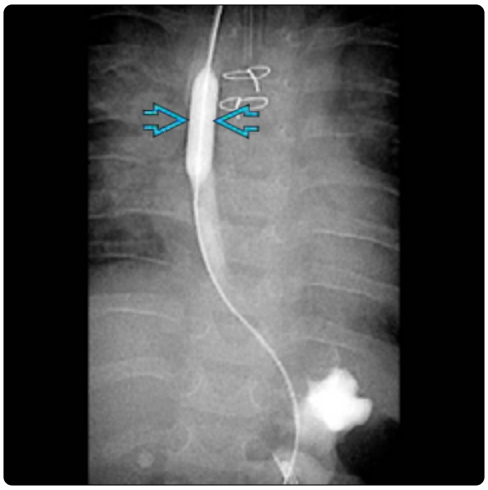
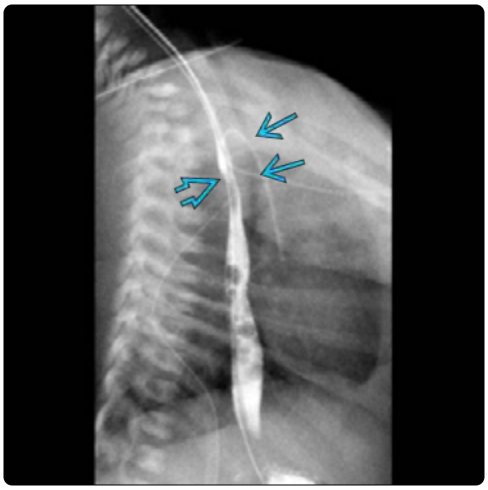


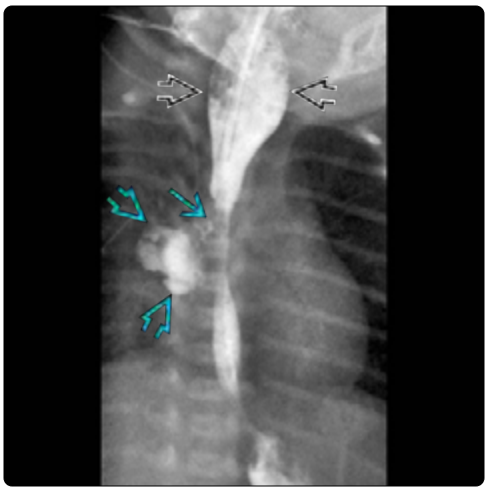


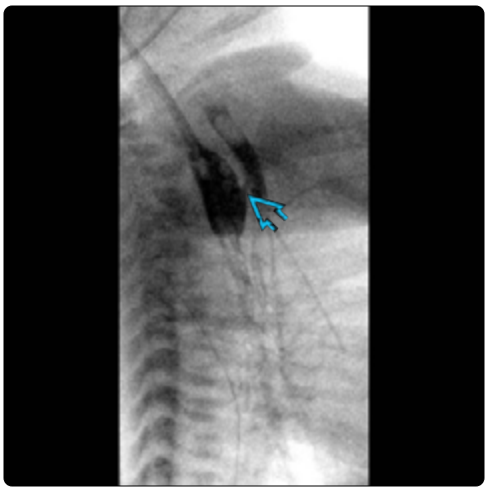
(Left) *Esophagram in an EA patient without TEF shows a long gap atresia, as measured by placing surgical bougies orally (into the upper pouch) & through the gastrostomy (into the distal atretic segment). A Foker procedure was then performed over several weeks to stretch the esophagus.* **(Right)** *Lateral esophagram in the same patient (who underwent a primary anastomosis of the esophagus in the interval) shows an anastomotic narrowing after repair. No contrast leak was seen.*





(Left) *An esophagram with balloon dilation was performed on the same patient to relieve the anastomotic stricture.* **(Right)** *Lateral esophagram performed in a neonate status post EA-TEF repair shows prompt flow through the anastomosis without a leak. There is tracheomalacia in the vicinity of the atresia, a common finding in patients with EA. Note that the study was performed via an additional 5 French feeding tube above the anastomosis (as not to pull back the critical NGT across the surgical site).*





(Left) *Postoperative esophagram in a neonate status post EA-TEF repair shows an extrapleural leak from a small tract next to the anastomosis. The upper esophageal dilation is likely related to residual in utero dilation rather than a postoperative obstruction.* **(Right)** *Postoperative esophagram on a neonate after EA-TEF repair shows no leak; however an additional, unsuspected proximal TEF was detected ~ 10 mm above the anastomotic site (the rare Gross type D, occurring in only 2% of all EA).*

Congenital Diaphragmatic Hernia

KEY FACTS

TERMINOLOGY

- Herniation of abdominal contents into chest via congenital defect in diaphragm, most commonly posterior (Bochdalek)

IMAGING

- Best clue: Bubbly, round, or tubular, relatively uniform air-filled lucencies in chest displacing mediastinum
- Intrathoracic herniated contents may include stomach, small & large bowel, liver, gallbladder, spleen
  - Results in paucity of bowel gas in abdomen
- Support devices distorted in relatively specific ways
  - Nasogastric tube due to stomach herniation
  - Umbilical venous catheter due to liver herniation
- Side of congenital diaphragmatic hernia (CDH): Left 85%, right 13%, bilateral 2%

TOP DIFFERENTIAL DIAGNOSES

- Congenital pulmonary airway malformation
- Diaphragmatic eventration
- Congenital lobar overinflation
- Bronchopulmonary sequestration

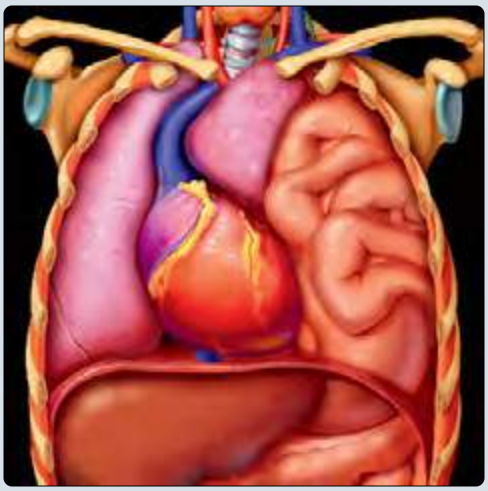
CLINICAL ISSUES

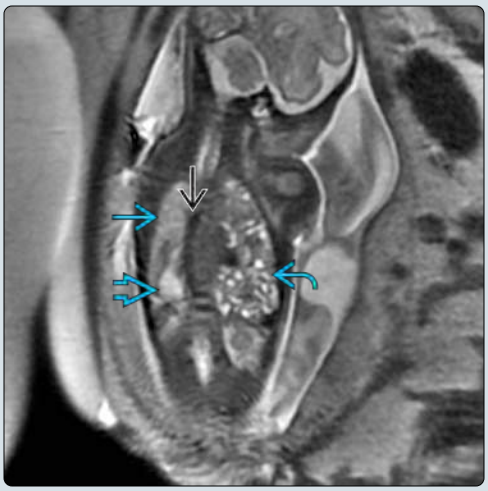
- Most detected prenatally; respiratory distress at birth in majority; delayed presentation uncommon
- Immediate initiation of supportive postnatal care until patient can tolerate surgical repair (primary vs. patch)
- Prognosis most related to severity of pulmonary hypoplasia, pulmonary arterial hypertension, congenital heart disease, & other anomalies
  - Various fetal US & MR parameters used to guide prognosis & postnatal management
- Survival ↑ if diagnosis known prenatally & patient delivers at high-volume center
- Increasing awareness of long-term morbidity in survivors

DIAGNOSTIC CHECKLIST

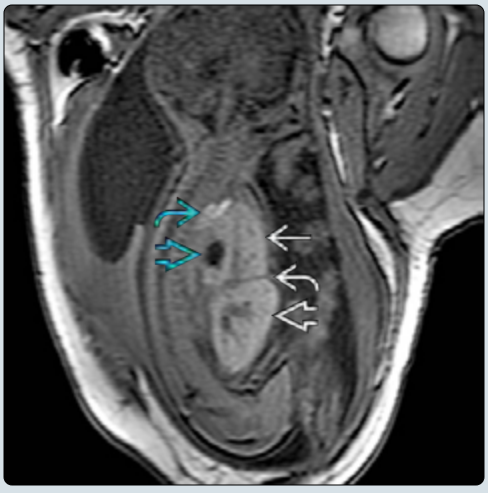
- Postnatal chest imaging of CDH beyond radiography typically unnecessary

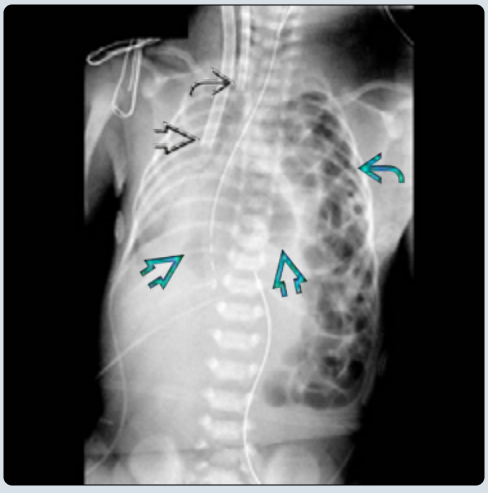
(Left) *Graphic shows a large posterior defect in the left hemidiaphragm with partial herniation of the stomach & bowel into the left hemithorax. There is rightward mediastinal shift with compression & hypoplasia of both the ipsilateral & contralateral lungs.* **(Right)** *Coronal SSFSE T2 MR of a 32-week gestation fetus shows herniated bowel loops filling the left hemithorax. The hypoplastic right lung overlies the aorta & herniated stomach, which are shifted into the right hemithorax.*





(Left) *Sagittal T1 MR in the same fetus shows a herniated left hepatic lobe separated from the remaining intraabdominal right hepatic lobe by residual anterior diaphragm. Note the herniated stomach & meconium-containing bowel.* **(Right)** *AP chest x-ray of the same patient after delivery shows gas-filled bowel filling the left hemithorax. The herniated stomach has an organoaxial rotation & overlies the herniated left hepatic lobe. Note the arterial & venous ECMO cannulae.*





TERMINOLOGY

Definitions

- Congenital diaphragmatic hernia (CDH): Herniation of abdominal contents into chest via developmental defect in diaphragm
  - Bochdalek hernia (70-95%): Posterior
  - Morgagni hernia (2-28%): Anterior
  - Central tendon hernia (2-7%)

IMAGING

General Features

- Best diagnostic clue
  - Bubbly, round, & tubular lucencies in neonatal hemithorax with contralateral mediastinal shift
- Location
  - Left 85%, right 13%, bilateral 2%
- Size
  - Degrees of diaphragmatic deficiency & herniated contents variable
    - May contain stomach, small & large bowel, liver, gallbladder, spleen

Radiographic Findings

- Depend on hernia contents & presence vs. absence of air in herniated bowel
  - Uniform soft tissue density of herniated solid organs
  - Herniated bowel may appear solid prior to progression of swallowed air
    - Upon air entering bowel, loops appear relatively uniform in size with round & tubular morphologies
      - Degree of gaseous distention changes day to day
    - Herniated gas-filled stomach larger than bowel & often abnormally rotated
- Mediastinal shift away from hernia
- Low lung volumes from hypoplasia
  - Ipsilateral lung more severe than contralateral
- Abnormal positions of support devices
  - Esophageal portion of nasogastric (NG) tube: Contralateral shift & convexity
    - NG tube often descends into upper abdomen then back into left hemithorax ("reverse J")
    - NG tube tip may lodge at gastroesophageal junction
  - Umbilical venous catheter (UVC) shifted toward side of hernia (if "liver up"); may not advance to right atrium
    - If UVC extends to right atrium, catheter typically convex toward hernia
- ↓ intraabdominal bowel gas
- Postoperative appearance
  - Resolution of herniated contents
  - Pleural fluid in 28% (typically chylothorax), eventually resolves
  - Ex vacuo negative pressure pneumothorax: Hypoplastic lung does not fill space initially
  - Gradual expansion of hypoplastic lungs
  - Repaired hemidiaphragm often angular or laterally sloped

Ultrasonographic Findings

- Prenatal ultrasound
  - Absent crescentic hypoechoic hemidiaphragm
  - Mixed echogenicity mass in lower hemithorax with mediastinal shift into contralateral hemithorax
    - Bowel & liver difficult to differentiate from lung lesion
      - Look for displacement of gallbladder & hepatic vessels
    - Herniated fluid-filled stomach helpful to confirm CDH
  - Lung:head ratio (LHR): Critical prognostic parameter in left CDH
    - Predictor of pulmonary hypoplasia based on residual contralateral lung
    - LHR = Area of right lung at level of heart 4-chamber view divided by head circumference
      - LHR < 1: Poor prognosis
  - ± polyhydramnios

MR Findings

- Fetal MR
  - T2 SSFSE
    - Serpentine fluid-filled structures (small bowel loops) fill most of hemithorax
    - Stomach usually herniated with abnormal rotation
    - Partial herniation of low signal intensity colon
    - ± herniation of low signal intensity liver, spleen
    - Contralateral mediastinal shift
    - ↓ total, observed/expected, & predicted lung volumes
      - Lungs of lower signal intensity than normal
      - Residual ipsilateral lung much smaller than contralateral & typically displaced superomedially
    - "Sac type" hernia has better prognosis
      - Cap of lung evenly distributed over hernia apex
    - Paucity of normal bowel in abdomen
  - T1 GRE
    - Helps localize intermediate signal intensity liver & high signal intensity meconium of small & large bowel against low signal intensity lungs

Imaging Recommendations

- Best imaging tool
  - Prenatal US & MR provide diagnostic & prognostic information
  - Postnatal radiograph to confirm support devices

DIFFERENTIAL DIAGNOSIS

Congenital Pulmonary Airway Malformation

- Macrocystic type appears as multicystic, air-containing mass
  - Cysts typically not uniform in size
  - After gas replaces fluid in cysts (in 1st few days after birth), congenital pulmonary airway malformation (CPAM) appearance typically static
    - CPAM more likely to have air-fluid levels than CDH
- Support device positions less altered by CPAM than CDH

Diaphragmatic Eventration

- Focal muscle aplasia of diaphragm without free protrusion of bowel into thorax
  - Abdominal contents contained by sac of peritoneum, diaphragm tendon, & parietal pleura

Congenital Lobar Overinflation

- Progressive overdistention of single lobe

- Rarely involves lower lobes

Bronchopulmonary Sequestration

- Solid mass near diaphragm with systemic arterial supply

PATHOLOGY

General Features

- Etiology
  - Normal diaphragm development (4th-12th weeks of gestation)
    - Lateral pleuroperitoneal folds fuse with septum transversum
    - Migrating myogenic precursors reach primitive diaphragm & proliferate
    - Error in any portion of process may lead to CDH
  - Underlying cause of defective development unknown in ~ 70-80%
    - Various chromosomal anomalies (2-35%)
      - Trisomy 18 most common (2-5%); Down syndrome (trisomy 21) most likely to have Morgagni type
    - Known specific genetic mutations (< 10%)
- Associated abnormalities
  - Isolated in ~ 50%
  - Cardiac malformations in 20-40%: Hypoplastic left heart syndrome, tetralogy of Fallot, transposition of great vessels, double-outlet right ventricle, & ventricular septal defect
  - Variety of other system anomalies described

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Diagnosis known prenatally in 60-80%
  - Respiratory distress at birth
- Other signs/symptoms
  - Presence of intrathoracic bowel sounds
  - Absence of ipsilateral breath sounds
  - Scaphoid abdomen
  - < 3% present after 30 days (100% survival)
    - Gastrointestinal symptoms more likely than respiratory

Demographics

- Gender
  - M:F = 1.5:1.0
- Epidemiology
  - 1 in 2,000-3,000 live births

Natural History & Prognosis

- Prognosis related to severity of pulmonary hypoplasia, pulmonary hypertension, & other anomalies (especially heart disease)
  - Most common fetal calculations used for prognosis
    - Lung volumes: Total, observed/expected, & percent predicted by MR; LHR by US
    - Volume of thorax occupied by herniated liver (independent of lung volumes) by MR
    - Pulmonary artery diameters: Modified McGoon Index, Persistent Pulmonary Hypertension Index by MR; various parameters by echocardiography
- Mortality rates variable: 10-68%
  - ↑ survival reported at single high-volume centers
  - Worse with specific syndromes, cardiac anomalies, esophageal atresia, bilateral hernias, large defects/hemidiaphragm agenesis, extracorporeal membrane oxygenation (ECMO) requirement, higher pulmonary support requirement at 30 days of life
- Long-term morbidity: Impaired lung function + recurrent respiratory infections, neurocognitive & language delays, failure to thrive, chest wall deformities, scoliosis

Treatment

- Varying degrees of supportive care required until definitive surgery able to be tolerated
  - Ventilation/oxygenation
    - Immediate intubation with gentle ventilation
    - ± ECMO
  - Pulmonary hypertension: Inhaled nitric oxide or IV sildenafil
- Surgery: Primary closure of small defects vs. patch repair for larger; timing debated
  - Complications include chylothorax, abdominal compartment syndrome, recurrent hernia; malrotation of midgut expected but does not carry typical risk of volvulus
- Fetal tracheal occlusion therapy: Temporary balloon occlusion of trachea in utero (to stimulate lung growth)
  - Still under study at limited centers

DIAGNOSTIC CHECKLIST

Consider

- Diagnosis usually apparent on early radiographs; other postnatal chest imaging modalities typically unnecessary

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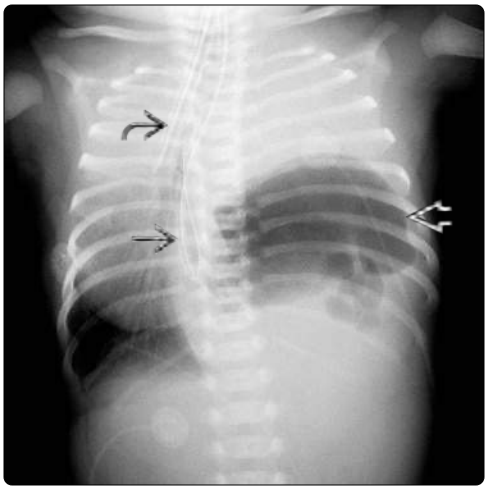
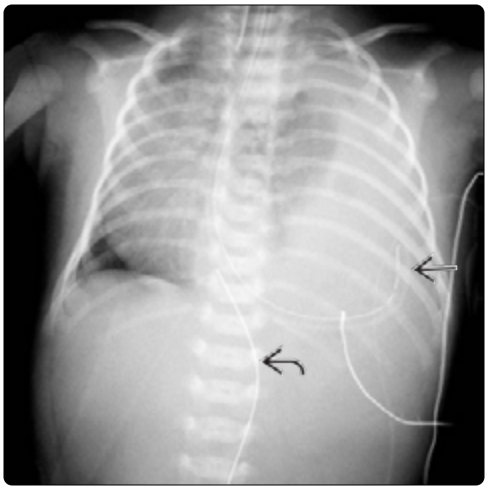
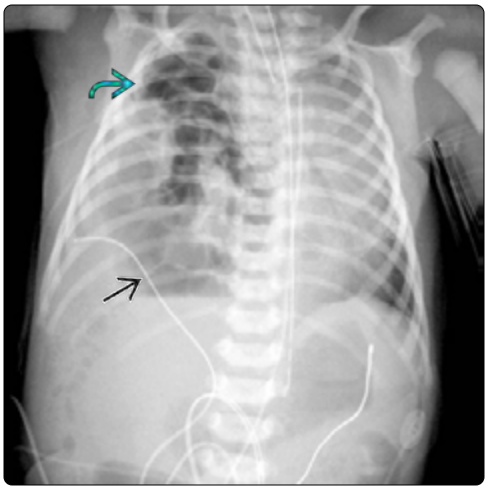
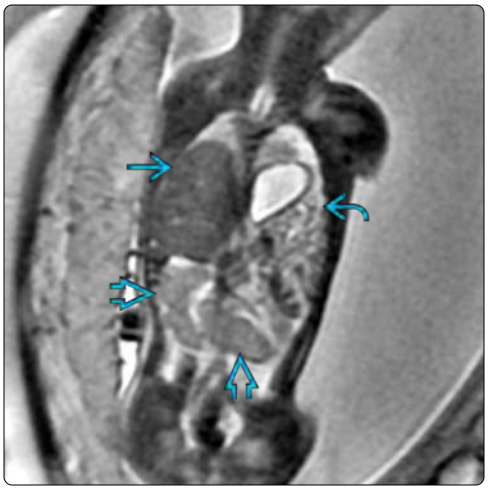


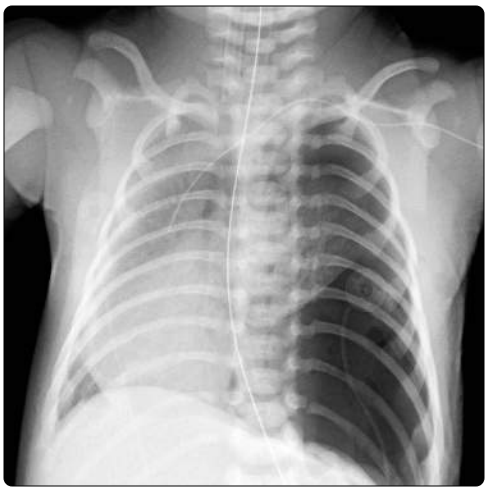


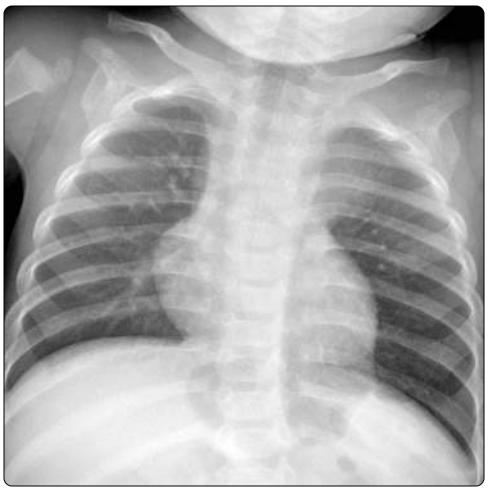
(Left) *Chest x-ray shows a newborn with a left congenital diaphragmatic hernia (CDH). The nasogastric (NG) tube ⇨ is coiled in the distal esophagus. The heart & endotracheal tube ⇨ are shifted to the right. The stomach ⇨ is in the left lower chest.* **(Right)** *Subsequent AP chest x-ray of the same patient shows the NG tube tip ⇨ decompressing the herniated stomach. An umbilical venous catheter (UVC) shows convexity toward the left ⇨ due to partial liver herniation. Decompressed bowel fills the left chest.*





(Left) *AP radiograph shows herniated bowel ⇨ filling the right hemithorax. The heart & support devices are shifted to the left. The UVC ⇨ is malpositioned in the herniated right hepatic lobe.* **(Right)** *Coronal SSFSE T2 MR in a 29-week gestation fetus shows bilateral diaphragmatic hernias with liver filling the right hemithorax ⇨ & stomach/bowel filling the left hemithorax ⇨. There is polyhydramnios + an incompletely visualized horseshoe kidney ⇨ in this fetus with Cornelia de Lange syndrome.*





(Left) *AP chest radiograph in a patient after repair of a left-sided CDH shows a large ex vacuo left pneumothorax. The heart, peripherally inserted central catheter (PICC), & NG tube are still shifted to the right. However, a chest tube should not be placed, as this is a postsurgical, negative-pressure pneumothorax.* **(Right)** *AP chest radiograph of the same patient 8 months later shows resolution of the pneumothorax & mediastinal shift. The hypoplastic left lung is now mildly hyperexpanded & hyperlucent.*

Surfactant Deficiency Disease

KEY FACTS

TERMINOLOGY

- Surfactant deficiency disease (SDD) favored term
  - a.k.a. respiratory distress syndrome & hyaline membrane disease (archaic)
- Common lung disease occurring in premature infants due to lack of surfactant
- Microatelectasis & abnormal pulmonary compliance are hallmarks of disease

IMAGING

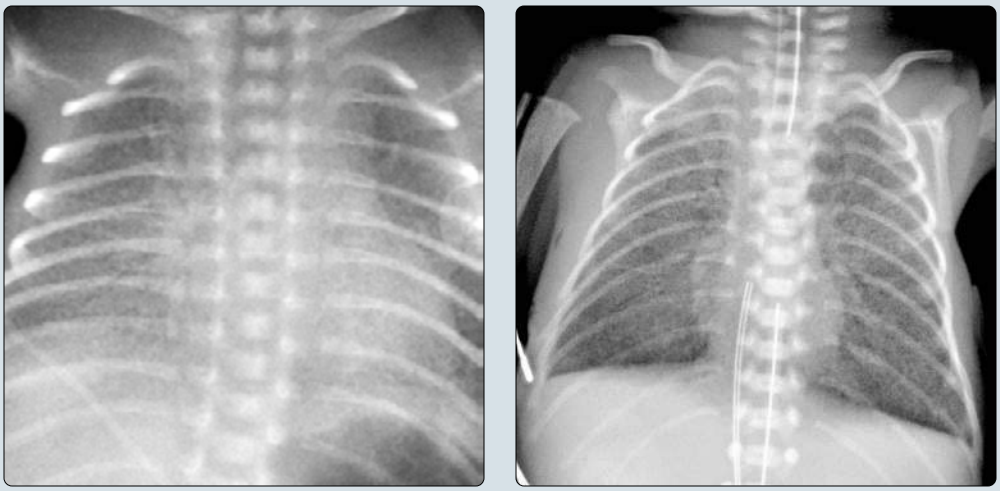
- Premature infants < 32-weeks gestation at risk
- Initial findings are low lung volumes & diffuse reticular granular opacities
- High incidence of patent ductus arteriosus, which causes pulmonary edema ("whiteout" of lungs with cardiomegaly)
- Bronchopulmonary dysplasia eventually occurs in 17-55% of premature infants

PATHOLOGY

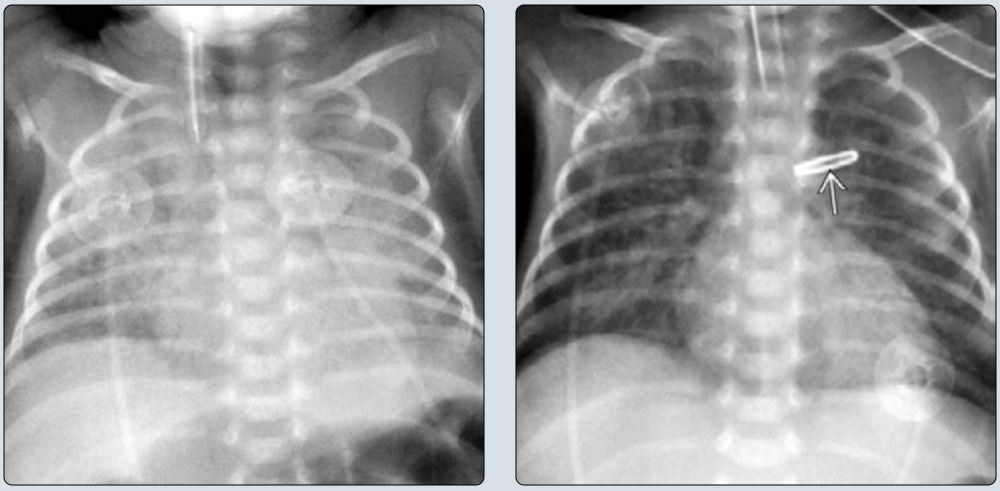
- Surfactant normally coats alveoli & ↓ surface tension, allowing alveoli to stay open
  - Prematurity-related SDD: Immature type II pneumocytes cannot produce sufficient surfactant
  - Secondary SDD: Surfactant deactivation from meconium aspiration or infection
  - Primary SDD: Dysfunctional surfactant due to abnormalities of 1 of several gene products (especially SFTPB, SFTPC, ABCA3, TTF-1)

CLINICAL ISSUES

- Most common cause of death in live newborn infants
- Acute complications: Alveolar rupture with barotrauma (pneumothorax, pneumomediastinum, pulmonary interstitial emphysema)



(Left) *Frontal radiograph of the chest in a premature patient with surfactant deficiency disease (SDD) shows pulmonary hypoventilation & granular densities bilaterally.* **(Right)** *AP radiograph of the chest shows diffuse granular opacities with marked hyperinflation of the lungs. Although classically lung volumes are decreased in patients with SDD, artificial ventilation can make them hyperinflated, especially after surfactant administration.*



(Left) *AP radiograph of the chest in a 1 week old with SDD shows diffuse opacification of the lungs. This patient had a patent ductus arteriosus (PDA) that had reversed its direction of flow & was now causing pulmonary edema.* **(Right)** *AP radiograph of the chest in the same patient shows increased aeration of the lungs after PDA ligation (with a surgical clip ➡ now in place).*

TERMINOLOGY

Abbreviations

- Surfactant deficiency disease (SDD)

Synonyms

- Lung disease of prematurity
- Respiratory distress syndrome
- Hyaline membrane disease (archaic)

Definitions

- Prematurity-related SDD (most common usage)
  - Surfactant deficiency in premature newborns
  - Lungs begin producing surfactant at about 24 weeks gestational age (GA); adequate amounts are usually present by 36 weeks GA
  - Lack of sufficient surfactant causes diffuse microatelectasis & abnormal pulmonary compliance
- Secondary SDD (less common usage)
  - Meconium aspiration or pneumonia can cause surfactant deactivation
  - Results are similar to those seen in prematurity, but usually heterogeneous & superimposed on other findings
- Primary SDD (least common usage)
  - Rare genetic causes of surfactant dysfunction
  - Abnormal gene products adversely affect composition &/or function of surfactant
  - Most well-known causes: Surfactant protein-B, surfactant protein-C, ABCA3, & TTF-1 deficiencies
  - Often termed "surfactant dysfunction disorders"

IMAGING

General Features

- Best diagnostic clue
  - Initial findings are low lung volumes & diffuse granular opacities
    - Air bronchograms & poor lung expansion
    - Cardiac size is normal
  - Subsequent imaging shows significant bilateral lung disease
    - Localized areas of atelectasis
    - Barotrauma leads to pneumothorax, pneumomediastinum, pulmonary interstitial emphysema (PIE)
    - Complications of patent ductus arteriosus (PDA)
    - Bronchopulmonary dysplasia (BPD) or chronic lung disease of premature infant
- Age
  - Premature infants < 32 weeks GA

Radiographic Findings

- Radiography
  - Initial features
    - Low lung volumes secondary to microcollapse of innumerable alveoli
    - Diffuse granular opacities represent collapsed alveoli interspersed with open alveoli (microatelectasis)
    - Air bronchograms demonstrate patent bronchi in abnormal lung
    - Pleural effusions very uncommon
    - Potential acute complications include PIE, pneumomediastinum, pneumothorax, superimposed pneumonia, pulmonary hemorrhage
  - Features after surfactant administration
    - Clearing of granular opacities & ↑ lung volumes
    - May have asymmetric or partial response
  - Findings after several days
    - Intubation & ventilatory support change imaging appearance
    - High incidence of PDA, which may cause pulmonary edema ("whiteout" of lungs with cardiomegaly)
  - BPD develops in 17-55% of premature infants
    - Chronic lung disease characterized by coarse interstitial opacities with focal areas of atelectasis & hyperinflation vs. diffuse hyperinflation

Other Modality Findings

- Premature infants have many associated diseases
  - Intracranial ultrasound used to assess for hemorrhage
  - Abdominal films for necrotizing enterocolitis

Imaging Recommendations

- Best imaging tool
  - Chest radiograph with comparison to prior imaging

DIFFERENTIAL DIAGNOSIS

Congenital Heart Disease

- Echocardiography is gold standard for diagnosis
- PDA is common in infants > 1,000 g
  - Usually closed with prostaglandin inhibitor
  - Transcatheter occlusion vs. surgery if contraindication present

Group B Streptococcal Pneumonia

- Very common in neonates, especially premature infants
- Acquired during birth (25% of women colonized)
- Bilateral granular opacities & low lung volumes
- Pleural effusion common (67%): Only imaging finding that helps differentiate from SDD

Meconium Aspiration Syndrome

- Term infants
- Rope-like densities radiating from hila
- Usually high lung volumes
- High incidence of barotrauma

Transient Tachypnea of Newborn

- Findings similar to pulmonary edema but resolve by 24-48 hours
- Prominent interstitial markings with normal heart size
- Pleural effusions may be present

PATHOLOGY

General Features

- Etiology
  - Immature type II pneumocytes cannot produce surfactant
  - Surfactant normally coats alveoli & ↓ surface tension, allowing alveoli to stay open & improving lung compliance
  - Deficiency of surfactant results in alveolar atelectasis

- ↓ lung compliance is associated with interstitial edema
- Secondary surfactant insufficiency occurs due to
  - Meconium aspiration pneumonia
  - Pulmonary infections
  - Intrapartum asphyxia
- Genetics
  - Prematurity-related SDD
    - Mothers who deliver premature infants are more likely to have subsequent premature infants
    - Premature infants have many associated abnormalities
  - Uncommon primary (genetic) SDD
    - Several genetic mutations associated with dysfunction of key surfactant components/regulators
      - Surfactant protein-B, surfactant protein-C, ABCA3, & TTF-1 deficiencies

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - History of prematurity
  - Respiratory distress

Demographics

- Age
  - Disease of premature infants (< 36 weeks GA, < 2.5 kg)
    - 50% of premature infants will have SDD
  - Infants < 27 weeks GA have higher incidence of sequelae
- Ethnicity
  - All races affected worldwide
- Epidemiology
  - M > F
  - Most common cause of death in live newborn infants
  - Occurs in 40,000 infants each year in USA
  - Common in all causes of premature labor & delivery
  - More common in infants of diabetic mothers

Natural History & Prognosis

- Acute complications & associations
  - Alveolar rupture with pneumothorax, pneumomediastinum, PIE
  - Sepsis & pulmonary infections
  - PDA with shunting & wide pulse pressure
  - Pulmonary hemorrhage
  - Apnea
  - Necrotizing enterocolitis ± perforation
  - Intracranial hemorrhage, periventricular leukomalacia
- Chronic complications
  - BPD defined as ≥ 28 days oxygen (O_2) dependence in premature infant
    - Assessed by O_2 challenge at 36 weeks postmenstrual age
  - Retinopathy of prematurity
  - ↑ incidence of sudden death
  - Gastroesophageal reflux
  - Neurologic impairment in 10-70% (depending on GA)
  - Family psychodynamic complications
    - 2-year risk of parental divorce/separation is 2x higher compared to parents of term newborns
    - Maternal depression/anxiety rates are 5x higher than in mothers of term newborns

Treatment

- Prenatal prevention by treatment of mother
  - Efforts to delay delivery, allowing fetus to mature
  - Maternal steroid administration: Steroids will cross placenta & ↑ surfactant
- Surfactant administration
  - Can be given in nebulized or aerosol forms
  - Injected into trachea via endotracheal tube or via catheter inserted into trachea
  - May be given prophylactically or after symptoms develop
  - Improves oxygenation & ventilator setting requirements; ↓ rates of barotrauma, intracranial hemorrhage, BPD, & death
  - ↑ risk of PDA & pulmonary hemorrhage
- Mechanical ventilation with positive end-expiratory pressure
- High-frequency oscillatory ventilation
- Meet special needs of premature patient
  - Respiratory support
  - Monitor temperature, prevent hypothermia
  - Treat metabolic acidosis
  - Intravenous access with fluid & caloric needs

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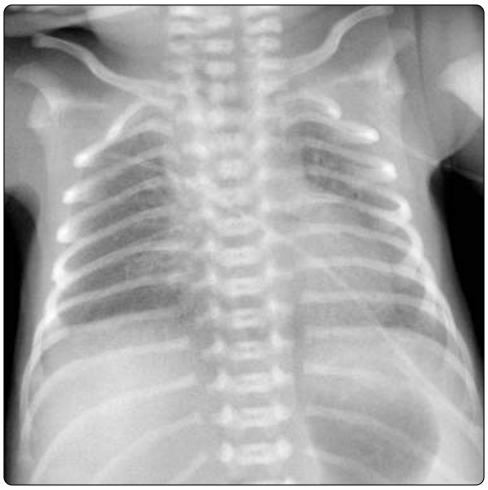
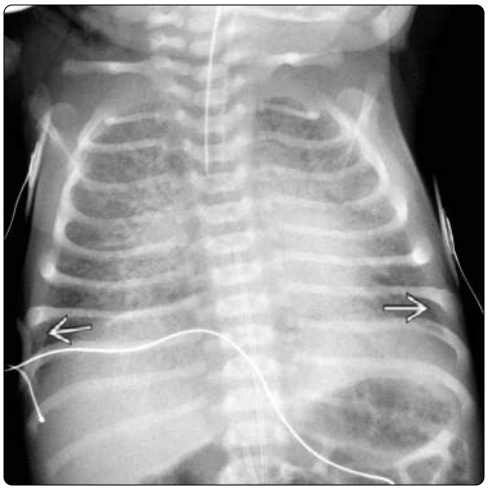
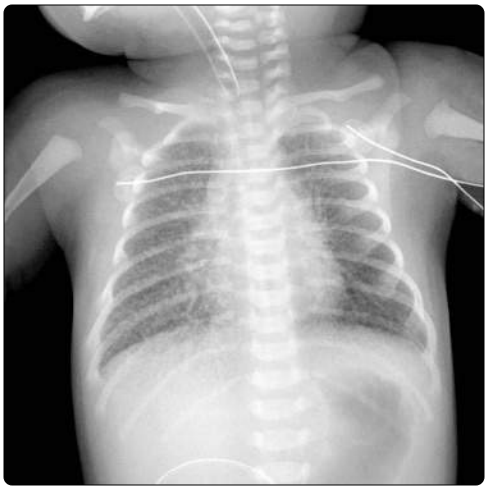
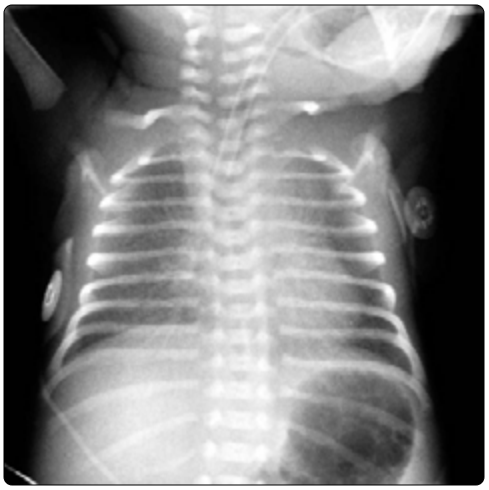


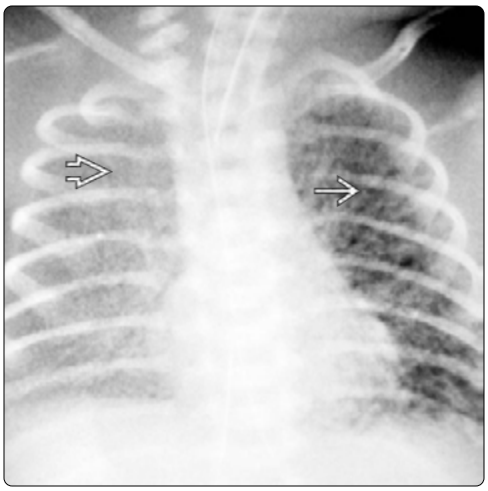


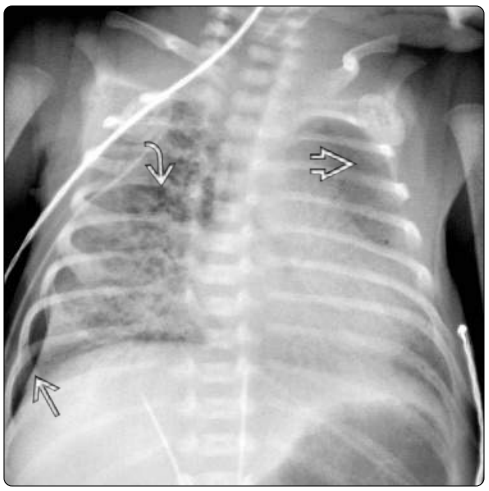
(Left) *AP radiograph of the chest in a premature infant shows low lung volumes with increased diffuse hazy opacity consistent with respiratory distress syndrome (RDS) or SDD.* **(Right)** *AP radiograph shows the chest in an infant with diffuse hazy opacity bilaterally & bilateral pleural effusions. This patient had an obstructed-type total anomalous venous return. Pulmonary edema & other diffuse pulmonary disease can look similar to RDS.*





(Left) *AP radiograph of the chest in a patient with neonatal pneumonia shows diffuse granular opacity that can look similar to the typical RDS patient.* **(Right)** *AP radiograph of the chest in a premature infant with diffuse granular opacities is consistent with SDD.*





(Left) *Frontal radiograph of the chest in a premature infant shows typical granular opacities on the right in a patient with SDD. There are linear lucencies seen on the left, consistent with pulmonary interstitial emphysema.* **(Right)** *Frontal radiograph of the chest in a premature infant shows typical granular opacities on the left in a patient with SDD. Linear lucencies are seen on the right, consistent with pulmonary interstitial emphysema, with a small right pneumothorax.*

Neonatal Pneumonia

KEY FACTS

TERMINOLOGY

- Pneumonia in first 28 days of life
- Early onset: Typically presents within 48 hours
- Late onset: Presents in 2nd-4th weeks of life

IMAGING

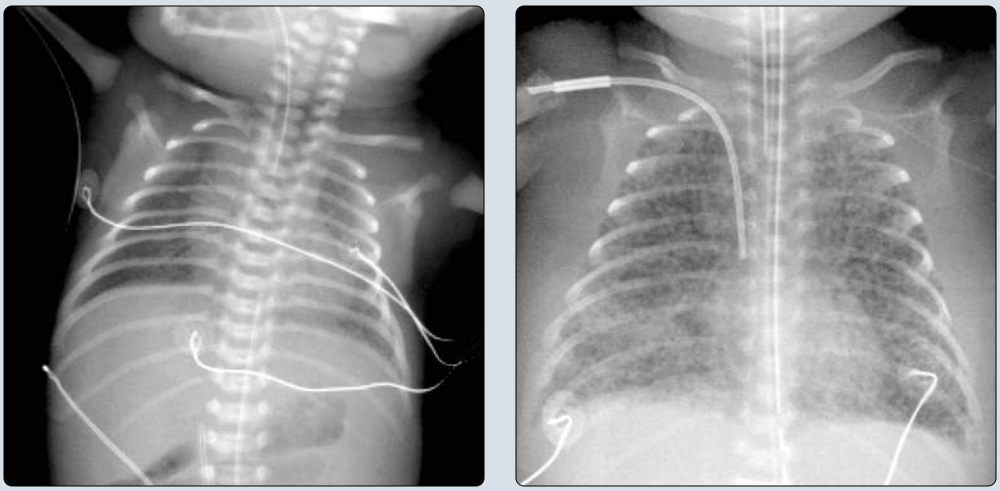
- Radiographs
  - Low lung volumes & granular opacities similar to surfactant deficiency (but pleural effusion in up to 67%)
  - Confluent > patchy alveolar or reticular opacities; may be perihilar
  - Complications: Pneumothorax, pneumomediastinum, pulmonary interstitial emphysema
- Ultrasound
  - Pleural line abnormality
  - Hepatization of parenchyma with air bronchograms
  - > 3 B-lines (areas of white lung)
  - Disappearance of "lung sliding"
  - Pulsation of lung synchronized with heart

PATHOLOGY

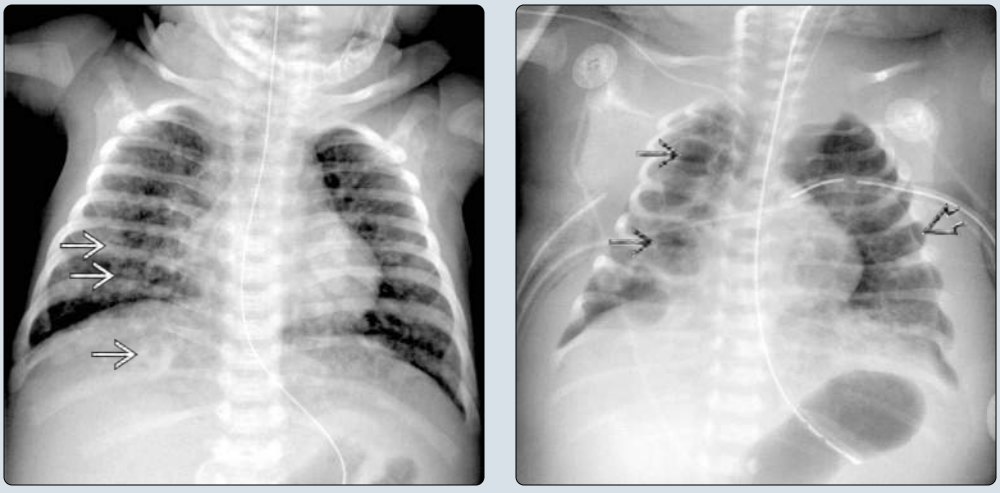
- Bacterial pathogens most common in early & late onset
- Most common causes of early-onset pneumonia: Group B *Streptococcus* (GBS) (developed countries), *Escherichia coli* (developing countries)
- Transmission
  - Early onset: Transplacental, aspiration of infected amniotic fluid, maternal systemic infection
  - Late onset: Contaminated/colonized equipment or individuals

CLINICAL ISSUES

- Typical presentations: Respiratory distress, sepsis
- Prevention: Universal screening at 35- to 37-weeks gestation for maternal GBS colonization
  - Intrapartum antibiotic prophylaxis with penicillin
- Treatment of neonate
  - Early onset: Empiric ampicillin + gentamicin
  - Late onset: Empiric vancomycin + aminoglycoside



(Left) *AP radiograph of the chest in a neonate with group B streptococcal pneumonia shows diffuse, bilateral hazy opacities with low lung volumes. This appearance is very similar to patients with surfactant deficiency disease.* **(Right)** *AP radiograph of the chest in a patient with neonatal pneumonia shows increased lung volumes with diffuse nodular pulmonary opacities. Anasarca is also noted.*



(Left) *AP radiograph in a neonate with tuberculosis shows multiple cavitating nodules in the right lung.* **(Right)** *AP radiograph in a patient with neonatal pneumonia shows a left pneumothorax & multiple bilateral pneumatoceles, complications that can be seen with neonatal pneumonia.*

TERMINOLOGY

Definitions

- Pneumonia in first 28 days of life
- Early onset: First 48 hours of life; some define as < 7 days
- Late onset: 2nd-4th weeks of life

IMAGING

Radiographic Findings

- Radiography
  - Group B *Streptococcus* (GBS)
    - Low lung volumes + granular opacities similar to surfactant deficiency
    - Pleural effusion in up to 67%
  - Other appearances
    - Confluent/patchy alveolar or reticular opacities
    - Regions of hyperinflation
  - Complications: Pneumothorax, pneumomediastinum, pulmonary interstitial emphysema

Ultrasonographic Findings

- Absent, irregular, disrupted, or coarse pleural line
- Parenchymal hepatization & air bronchograms (punctiform or linear hyperechogenicities)
- > 3 B-lines or areas of white lung
- Disappearance of "lung sliding"
- Pulsation of lung synchronized with heart rate

Imaging Recommendations

- Best imaging tool
  - Chest radiography
  - ↑ use of ultrasound, but role still debated
    - Metaanalysis shows sensitivity of 96% & specificity of 93% for pneumonia in children of all ages
      - Similar findings also seen in surfactant deficiency

DIFFERENTIAL DIAGNOSIS

Surfactant Deficiency Disease

- Premature infants < 32-weeks gestation
- Mimics GBS infection but no effusion

Congenital Pulmonary Airway Malformation

- Mixed cystic &/or solid mass

Meconium Aspiration Syndrome

- History of meconium staining with respiratory distress

Transient Tachypnea of Newborn

- Occurs in term infants delivered by C-section
- Interstitial pattern with effusions
- Resolves within 48 hours

Congenital Heart Disease

- Abnormal cardiac silhouette & pulmonary vascularity ± edema
- Echocardiography for diagnosis

PATHOLOGY

General Features

- Etiology
  - GBS most common cause of early-onset neonatal pneumonia/sepsis in developed countries
    - β-hemolysin (pore-forming cytolysin) → ↑ epithelial cell permeability → alveolar edema & hemorrhage
    - β-hemolysin inactivated by surfactant → premature infants at ↑ risk
  - *Escherichia coli* most common cause of early-onset neonatal pneumonia in developing countries
  - Numerous other bacterial, viral, & fungal etiologies

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - World Health Organization does not distinguish between neonatal pneumonia & sepsis
  - Respiratory distress: Rapid/noisy/difficult breathing, respiratory rate > 60, chest retractions, cough, grunting
  - Sepsis: Lethargy, poor feeding, poor reflexes, temperature instability, abdominal distension
  - Abnormal WBC with > 20% bands, ↑ CRP, ↑ ESR
  - Positive cultures

Demographics

- Epidemiology
  - Early onset transmission: Transplacental, aspiration of infected amniotic fluid, maternal systemic infection
  - Late-onset transmission: Contaminated/colonized equipment or individuals
  - GBS is leading infectious cause of infant morbidity & mortality in USA (0.34-0.37 cases/1,000 live births)

Natural History & Prognosis

- Found in up to 38% of deaths occurring in first 48 hours
- GBS case fatality rate up to 30% if ≤ 33-weeks gestation, 2-3% if full term
- Can be associated with intraventricular hemorrhage, neurological damage, & developmental delay

Treatment

- GBS screening at 35- to 37-weeks gestation; intrapartum antibiotic prophylaxis with penicillin if positive
- Neonatal antibiotic regimen otherwise depends on pathogen, early- vs. late-onset, antimicrobial susceptibility

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Consider if effusion accompanies radiographic findings otherwise suggestive of surfactant deficiency

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Meconium Aspiration Syndrome

KEY FACTS

TERMINOLOGY

- Meconium aspiration syndrome (MAS): Respiratory distress after aspiration of meconium-stained amniotic fluid
  - Causes ↓ lung compliance & hypoxia ± pulmonary hypertension & air leak syndrome

IMAGING

- Coarse, thick, "rope-like" linear & nodular perihilar opacities
- Patchy, hazy opacities of atelectasis & pneumonitis
- Generalized hyperinflation
- ± pleural effusion
- ± air leak: Pneumomediastinum, pneumothorax, pulmonary interstitial emphysema

TOP DIFFERENTIAL DIAGNOSES

- Congenital heart disease, neonatal pneumonia, transient tachypnea of newborn, surfactant deficiency disease, pulmonary hypoplasia

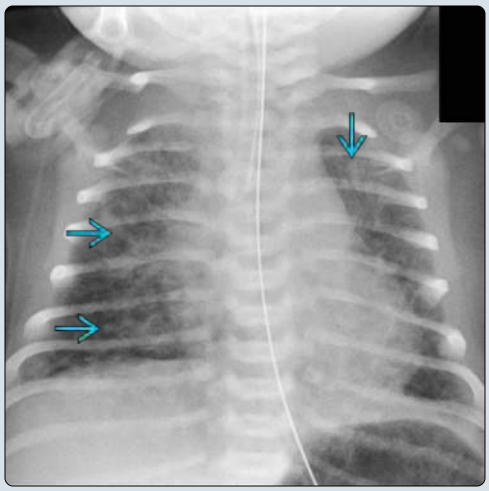
PATHOLOGY

- Aspirated meconium causes injury by several mechanisms
  - Mechanical obstruction of small airways → air-trapping, air-leak complications
  - Chemical pneumonitis of airways & parenchyma
  - Surfactant inactivation → diffuse atelectasis
  - Pulmonary vasoconstriction → persistent pulmonary hypertension

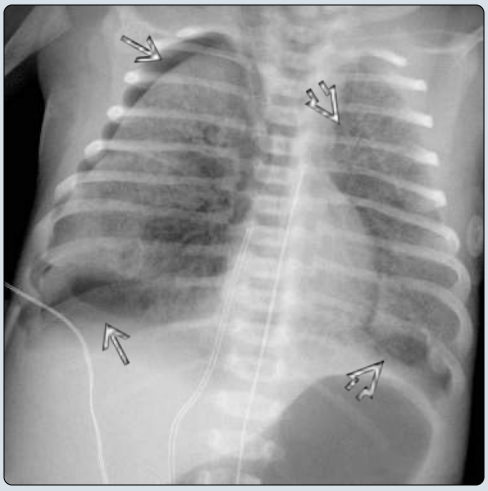
CLINICAL ISSUES

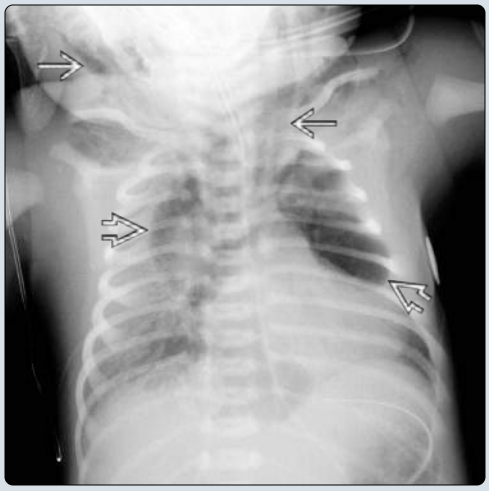
- Disease of term & postterm neonates
- Meconium staining of amniotic fluid occurs in infants with in utero or intrapartum hypoxia or stress
  - 4-12% with meconium staining develop MAS
- Meconium-stained & distressed infant suctioned immediately ± intubation
- ECMO for severe pulmonary hypertension
- Mortality 7-12%; chronic lung disease 2.5%

(Left) *Graphic demonstrates asymmetric areas of hyperinflation & atelectasis as well as rope-like perihilar densities of aspirated meconium & inflamed airways.* **(Right)** *AP chest radiograph in a 41-week gestation newborn with meconium staining of amniotic fluid & respiratory distress shows coarse, reticular, rope-like perihilar opacities, typical of meconium aspiration. There are patchy asymmetric regions of increased hazy lung opacity bilaterally.*



(Left) *AP radiograph of the chest in a full-term infant with meconium aspiration demonstrates right greater than left pneumothoraces with bilateral lung hyperinflation & diffuse opacification. Pneumothorax is a common complication in these patients.* **(Right)** *AP radiograph of a full-term patient with witnessed meconium aspiration shows a large pneumomediastinum. Note the linear lucencies of subcutaneous emphysema tracking into the neck bilaterally.*





TERMINOLOGY

Abbreviations

- Meconium aspiration syndrome (MAS)

Definitions

- Respiratory distress in setting of aspiration of meconium-stained amniotic fluid
  - MAS causes ↓ lung compliance & hypoxia ± pulmonary hypertension & air leak syndrome
  - Severe MAS: Requires mechanical ventilation

IMAGING

General Features

- Best diagnostic clue
  - Coarse, linear-nodular, rope-like peribronchial opacities bilaterally + hyperinflation in term infant
  - Usually clear history of meconium-stained amniotic fluid ± visualization of meconium at/below vocal cords
- Location
  - Bilateral disease, often in middle 2/3 of lung
  - Frequently asymmetric
- Morphology
  - Segmental hyperinflation from air-trapping with focal areas of atelectasis

Radiographic Findings

- Radiography
  - Hyperinflated lungs, though often asymmetric
  - Linear, nodular, rope-like densities radiating from hila
  - Patchy, hazy opacities of atelectasis & pneumonitis
  - ± pleural effusion
  - ± air leak: Pneumothorax (10-40%), pneumomediastinum, pulmonary interstitial emphysema (PIE)

Ultrasonographic Findings

- Lung ultrasound increasingly used at bedside of ill children [point of care ultrasound (POCUS)]
  - Findings relate to artifacts from interaction of sound with air & interstitial fluid
  - Can help predict need for mechanical ventilation
- MAS findings include: Sonographic B-lines (interstitium), consolidation, bronchograms, atelectasis, airway inflammation, pleural effusion
  - Uneven & changing distribution over clinical course

CT Findings

- HRCT
  - May be obtained if course atypical (to exclude other causes)
    - Nodular ground-glass opacities surrounding airways
    - ± dot-dash sign of air surrounding vessels in PIE
  - Occasionally obtained for chronic disease
    - Scarring, air-trapping, cystic change

Imaging Recommendations

- Best imaging tool
  - Chest radiograph

DIFFERENTIAL DIAGNOSIS

Congenital Heart Disease

- Clues to underlying congenital heart disease include
  - Abnormal heart size or configuration
  - Right aortic arch or heterotaxy
  - ↑ or ↓ pulmonary vascular flow
- Echocardiography remains standard for diagnosis

Neonatal Pneumonia

- Patchy asymmetric perihilar densities & hyperinflation
- Common to see pleural effusion
- Usually no history of meconium aspiration

Transient Tachypnea of Newborn

- Occurs secondary to delayed clearance of fetal pulmonary fluid (often in cesarean section)
- ± pleural effusion or fissural thickening
- Benign, self-limited course over 24-72 hours

Surfactant Deficiency Disease

- Diffuse granular pulmonary opacities in premature infant with low lung volumes
- Pleural effusions uncommon

Pulmonary Hypoplasia

- In utero lung development impaired by
  - ↓ available space (e.g., diaphragmatic hernia, congenital lung lesion, or skeletal dysplasia)
  - Longstanding oligohydramnios with ↓ fluid entering developing lungs (e.g., severe bilateral renal disease, bladder outlet obstruction)
- Pneumothoraces common
- Bell-shaped thorax in severe cases

PATHOLOGY

General Features

- Etiology
  - Intrauterine or intrapartum meconium aspiration
    - Fetal distress &/or hypoxia near or beyond term → passage of meconium into amniotic fluid, which may enter lungs
    - Risk factors: Delivery ≥ 41 weeks, placental insufficiency, maternal hypertension, preeclampsia, maternal drug use, acute tocolysis, oligohydramnios, abnormal fetal heart tracings, low Apgar score, low umbilical cord pH
  - Meconium alters amniotic fluid, causing
    - Placental inflammation
    - ↑ risk of bacterial infection & neonatal sepsis
    - Potentially severe damage to umbilical cord, leading to vasoconstriction ± fetal distress with asphyxia
      - Cord damage associated with lower Apgar scores & lower arterial pH
    - Fetal skin irritation
  - Aspirated meconium causes injury by several mechanisms
    - Mechanical obstruction of small airways → air-trapping, air leak complications
      - Pneumothorax, pneumomediastinum, PIE
    - Chemical pneumonitis of airways & parenchyma

        - Mild surrounding lung inflammation vs. none (57% in autopsy series)
        - More diffuse pneumonitis may be secondarily incited by inflammatory mediators
    - Surfactant inactivation with stripping of surfactant from alveolar surface (causing diffuse atelectasis)
    - Pulmonary vasoconstriction leading to persistent pulmonary hypertension
        - Leading cause of death in MAS

Gross Pathologic & Surgical Features

- Meconium: Tenacious, thick, viscous material of fetal bowel
    - Green-black substance of mucus, vernix, epithelial cells, lanugo, fatty acids, pancreatic enzymes, bile
    - Normal passage of meconium occurs in 1st 24 hours after birth
    - Thick meconium more likely to cause MAS than thin

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
    - Meconium staining of amniotic fluid at birth
    - Respiratory distress
        - Cyanosis, nasal flaring, tachypnea, grunting, intercostal retractions
- Other signs/symptoms
    - Laryngoscopy shows meconium staining on vocal cords
    - Green urine due to excretion of bile acids
    - Overdistention of chest from bilateral hyperinflation
    - Metabolic acidosis from perinatal stress
    - Respiratory acidosis from parenchymal disease & pulmonary hypertension
    - May be accompanied by inappropriate secretion of antidiuretic hormone (SIADH) or acute renal failure
    - Cyanosis with right-to-left ductus arteriosus shunting

Demographics

- Epidemiology
    - 1-6.5:1,000 live births annually in USA
    - Occurs in term or postterm infants with in utero or intrapartum hypoxia or stress
    - Meconium staining of amniotic fluid occurs in
        - 5% of preterm births
        - 25% of term births
        - 23-52% of postterm births
    - 4-12% with meconium staining develop MAS
        - 20-30% of MAS patients have meconium visualized below vocal cords
    - MAS accounts for 10% of neonatal respiratory distress

Natural History & Prognosis

- Complications
    - Meconium injury & hypoxia contribute to high pulmonary vascular resistance
    - Severe pulmonary hypertension occurs in 10% → persistent fetal circulation (right-to-left shunting across patent ductus arteriosus)
    - Anoxic brain injury → cerebral palsy, developmental delay, seizures
    - Subcutaneous fat necrosis
- Outcome depends on degree of aspiration & distress
    - Some recover within 72 hours
    - Up to 25% may develop air leak complications
    - Mortality reports range from 1-38%; likely on order of 7-12% currently
    - Chronic lung disease with abnormal pulmonary function, cough, wheezing, &/or hyperinflation in minority (2.5%)

Treatment

- Before delivery
    - Improved maternal care to ↓ risk of meconium aspiration
        - Labor induction by 41 weeks
    - Amnioinfusion of fluid dilutes meconium but controversial
- At delivery
    - Routine intrapartum suctioning of airway not beneficial
    - Meconium-stained & distressed infant suctioned immediately
- After delivery
    - Supplemental oxygen
    - Mechanical ventilation required in 33-70%
    - Bronchoalveolar lavage in 44%
    - Replacement of surfactant
        - ↓ length of stay, length of mechanical ventilation, & need for ECMO but not mortality
    - Inhaled nitrous oxide for pulmonary vasodilation
    - Helium-oxygen mixture (heliox) ventilation may be used
    - Steroids to ↓ inflammation of chemical pneumonitis
    - Antibiotics
        - Limited data suggests no benefit if not septic
- ECMO (extracorporeal membrane oxygenation)
    - For severe pulmonary hypertension
    - Delivers oxygen to blood via external oxygenator, ↓ barotrauma

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Relevant history essential for accurate interpretation of all neonatal chest disease, particularly MAS
    - Term or postterm infant
    - Meconium staining of amniotic fluid at delivery

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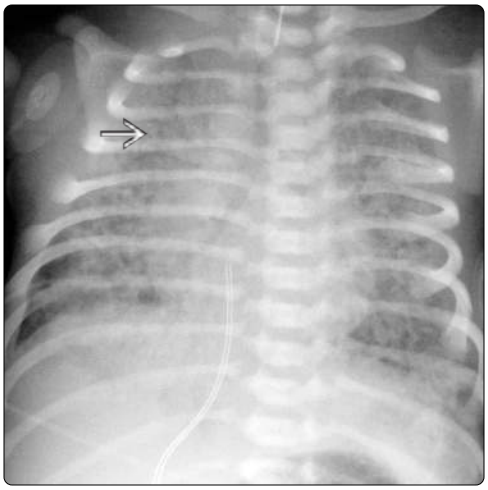
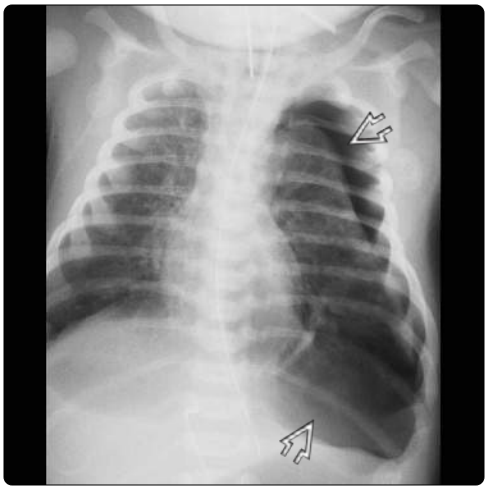
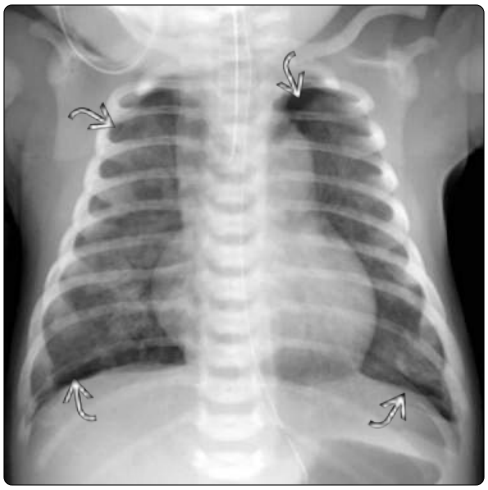
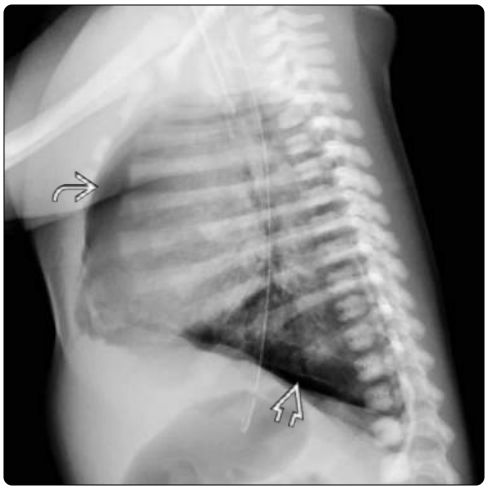


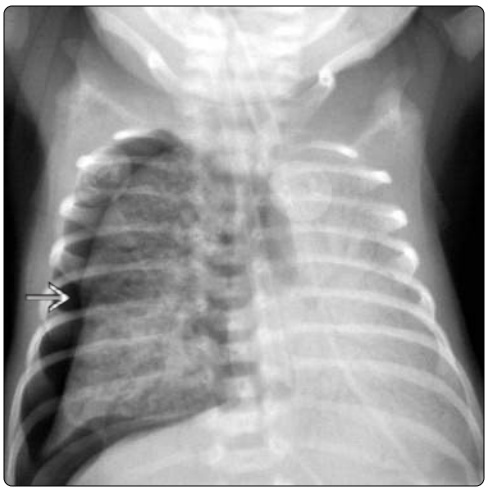


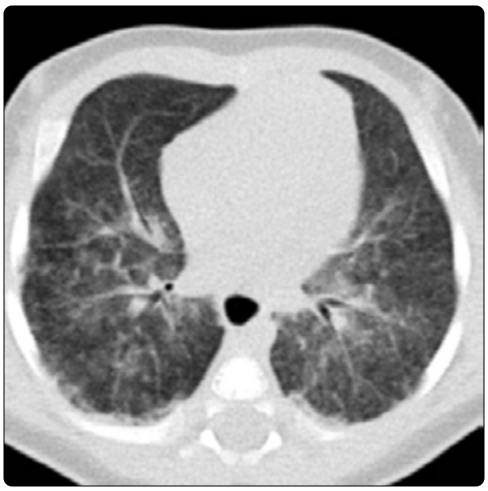
(Left) *AP radiograph in a patient with meconium aspiration shows bilateral patchy opacities & mild hyperinflation. Notice the coarse, rope-like interstitial densities in the hilar region, which are typical in patients with meconium aspiration.* **(Right)** *AP chest radiograph in a term newborn with meconium aspiration shows a large left pneumothorax. The lungs remain relatively hyperinflated, consistent with air trapping & diminished compliance.*





(Left) *AP chest radiograph in a newborn with meconium aspiration shows bilateral pneumothoraces with heterogeneous opacification & hyperexpansion of the bilateral lungs.* **(Right)** *Lateral radiograph in the same patient shows the lucent pneumothoraces tracking to anterior & subpulmonic locations. The hemidiaphragms are flattened, but the lungs remain hyperexpanded due to air-trapping with decreased compliance.*





(Left) *AP radiograph of the chest in a neonate with meconium aspiration shows a right pneumothorax with mediastinal shift to the left. The lungs are diffusely opacified.* **(Right)** *Axial HRCT in a 3-week-old full-term infant with witnessed meconium aspiration & continued tachypnea shows a predominantly posterior distribution of nodular, ground-glass opacities due to pneumonitis.*

Transient Tachypnea of Newborn

KEY FACTS

TERMINOLOGY

- Transient tachypnea of newborn (TTN), a.k.a. wet lung disease, retained fetal fluid
- Usually occurs after cesarean section due to lack of normal thoracic compression during vaginal delivery
- Caused by delayed evacuation of fetal lung fluid that creates engorgement of pulmonary lymphatics & capillaries

IMAGING

- Patients usually not intubated
- Findings similar to pulmonary edema
  - Diffuse, bilateral, & often symmetric ↑ lung markings
  - ± pleural effusion
- Lungs become normal within 24-48 hours
- Diagnosis of exclusion
  - Not associated with any chronic condition or lung disease

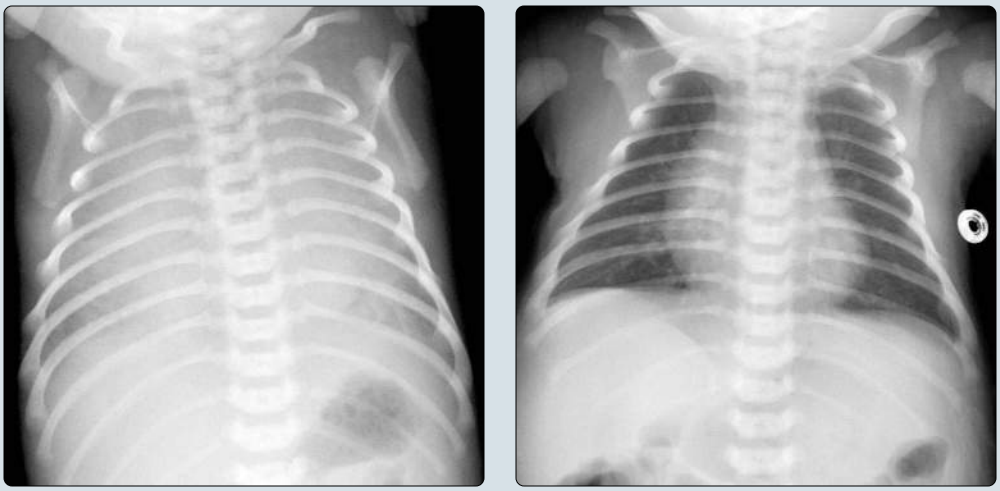
CLINICAL ISSUES

- Uncommon in premature infants
- More common in cesarean section infants
- Infants usually improve rapidly & are normal on follow-up
  - Occasionally need oxygen for several hours
  - Respiratory symptoms usually disappear by 3 days
- Some fluid restriction may be helpful

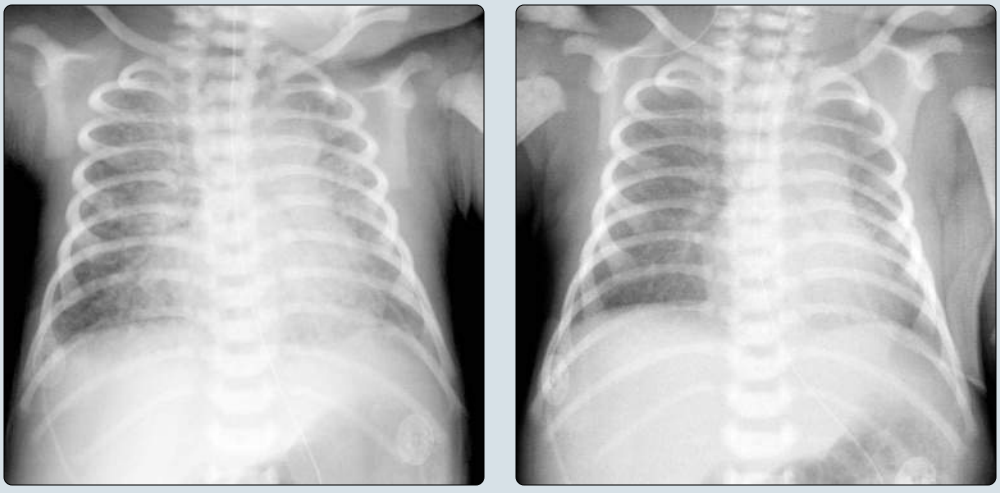
DIAGNOSTIC CHECKLIST

- Normal to ↑ lung volumes, coarse streaky opacities, pleural fluid, fluid in fissure
- Diagnosis of exclusion
  - No chorioamnionitis, maternal infection, sepsis
  - No meconium staining of amniotic fluid
  - No premature rupture of membranes

(Left) *AP radiograph of the chest in a full-term infant with tachypnea demonstrates diffuse bilateral hazy opacities. The findings & symptoms resolved, & the diagnosis of TTN was made.* **(Right)** *AP radiograph of the chest shows interval clearing of the previously seen diffuse opacification of the lungs. TTN is a diagnosis of exclusion & cannot be confidently diagnosed radiographically unless the findings resolve.*



(Left) *AP radiograph of the chest in a full-term infant with tachypnea shows diffuse nodular opacities throughout the lungs.* **(Right)** *AP radiograph of the chest in the same patient shows resolution of previously seen diffuse nodular opacities consistent with TTN.*



TERMINOLOGY

Abbreviations

- Transient tachypnea of newborn (TTN)

Synonyms

- Wet lung disease, retained fetal fluid

Definitions

- Transient tachypnea occurs when there is delayed evacuation of fetal lung fluid, causing engorgement of pulmonary lymphatics & capillaries
  - Usually term infants
  - More common (3x) in those born by cesarean section
    - Normal thoracic compression that occurs during vaginal delivery is bypassed by cesarean section
  - Lack of normal breathing may occur with sedated infants

IMAGING

General Features

- Best diagnostic clue
  - Prominent pattern of interstitial edema on chest radiograph
  - History of cesarean section
  - No signs of infection

Imaging Recommendations

- Best imaging tool
  - Chest radiographs in first 24-48 hours of life
    - Patients usually not intubated
    - No or minimal cardiomegaly
    - Normal to ↑ lung volumes
    - Findings similar to pulmonary edema
      - Prominent interstitial markings
      - Fluid in fissures
      - ± pleural effusion
    - Lungs become normal within 24-48 hours
    - No consolidation or other focal lung lesion
  - Imaging occasionally necessary to exclude other causes
    - Echocardiography to exclude congenital heart disease
    - Intracranial ultrasound to exclude high-flow shunt
    - Chest ultrasound or CT rarely helpful
- Protocol advice
  - Chest radiograph
  - Clinical history is helpful

DIFFERENTIAL DIAGNOSIS

Congenital Heart Disease

- Echocardiogram is gold standard for making diagnosis
- Obstructed total anomalous pulmonary venous return (TAPVR)
  - Normal heart size & interstitial edema
  - Cyanosis, acidosis
- Left-sided heart obstruction
  - Hypoplastic left heart
    - Most common
    - Radiograph may be normal initially
  - Severe coarctation
  - Aortic stenosis
  - Aortic interruption
  - Endocardial fibroelastosis
- Cardiac arrhythmias
  - Supraventricular tachycardia most common
  - Congenital heart block

Meconium Aspiration Syndrome

- Term infant
- Rope-like perihilar markings
- Hyperinflation

Congenital Lymphangiectasia

- Very rare, presents with persistent tachypnea
- Persistent interstitial pattern ± pleural effusions

Intrathoracic Causes of Tachypnea

- Neonatal pneumonia
  - *Escherichia coli* or group B *Streptococcus*
  - Tuberculosis pneumonia in endemic areas
  - Before delivery: Transplacental
    - Syphilis, herpes
  - During delivery
    - *E. coli*, group B *Streptococcus*, *Chlamydia*, herpes
  - After delivery
    - 30% premature infants colonized with *Candida*
    - *Chlamydia* pneumonia has delayed appearance
    - Staphylococcal, *Pseudomonas* pneumonia
- Neonatal chest masses
  - Congenital pulmonary airway malformation
    - Focal, space-occupying lesion
    - May be cystic, solid, or mixed
    - May cause mediastinal shift, effusion
  - Congenital diaphragmatic hernia
    - Multicystic mass (gas-filled bowel loops) with mediastinal shift
    - May have solid components (liver, compressed bowel)
    - Abnormal position of stomach, spleen or liver, bowel
    - Usually diagnosed in utero
  - Congenital lobar hyperexpansion
    - Solid-appearing in first 24-48 hours of life
    - Lucent or cystic mass after fluid clears
    - May shift mediastinum
  - Duplication cysts
- Congenital malformations
  - Lung hypoplasia or agenesis
    - Primary
    - Skeletal dysplasia
      - Asphyxiating thoracic dystrophy
      - Thanatophoric dwarfism
      - Osteogenesis imperfecta
    - Oligohydramnios
      - Bladder outlet obstruction
      - Bilateral renal anomalies
  - Tracheoesophageal fistula ± esophageal atresia

Iatragenic

- Diaphragmatic paralysis
- Tracheal stenosis

Extrathoracic Causes of Tachypnea

- Hematologic abnormalities
  - Polycythemia or severe anemia

- Arterial/venous malformations
  - Vein of Galen malformation
  - Congenital hepatic hemangioma
- Airway obstruction
  - Choanal atresia
  - Nasal pyriform aperture stenosis
  - Vocal cord paralysis
- CNS abnormalities
  - Cerebral anoxia or depression
  - Birth trauma or cord injury

Systemic Causes of Respiratory Distress

- Sepsis
- Electrolyte abnormalities
- Severe acidosis, hypothermia
- Hypovolemia

Neuromuscular Causes

- Werdnig-Hoffmann disease
- Muscular dystrophy

Intraabdominal Abnormalities

- Pneumoperitoneum
- Abdominal distension

PATHOLOGY

General Features

- Etiology
  - During fetal life, lungs expanded with ultrafiltrate of fetal fluid
  - During & after birth, lung fluid is replaced with air
    - Chest is normally compressed & fluid expelled during vaginal delivery: Thoracic squeeze
    - Pulmonary capillaries & lymphatics remove remaining fluid
  - Most infants with TTN are healthy & normal within 48 hours
- Associated abnormalities
  - Usually healthy infants with no other anomalies

Gross Pathologic & Surgical Features

- Not associated with any mortality or morbidity

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Mild to moderate respiratory distress
  - Frequent history of cesarean section delivery
  - Tachypnea occurs early after birth
    - Respiratory rates may exceed 60/minute
  - Expiratory grunting, chest retractions, nasal flaring
  - Occasional cyanosis which is changed by minimal oxygen
  - Typically do not require intubation
- Other signs/symptoms
  - Usually healthy, large full-term infants
  - Infants usually improve rapidly & are normal

Demographics

- Age
  - Newborns in first 48 hours of life
    - Tend to be term infants
    - Uncommon in premature infants
- Gender
  - More frequently in male patients
- Epidemiology
  - Incidence is 5-10 per 1,000 live births
  - More common in cesarean section infants
    - Incidence has ↑ as cesarean sections have ↑
  - Sibling with TTN increases risk

Natural History & Prognosis

- Mild to moderate respiratory distress within 6 hours of birth
  - Occasionally need oxygen for several hours
- Relatively benign clinical course
- Radiographic resolution usually by 24-48 hours
- Respiratory symptoms disappear usually by 3 days
- In healthy asymptomatic infant, follow-up films not necessary

Treatment

- Exclude other causes of tachypnea in term newborn
- Normal support of infant
  - Oxygen for mild cyanosis, normal feeding
- Some advocate IV furosemide, but controversial

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- ↑ lung volumes, coarse streaky opacities, pleural fluid, fluid in fissure
- Diagnosis of exclusion
  - No chorioamnionitis, maternal infection, sepsis
  - No meconium staining of amniotic fluid
  - No premature rupture of membranes

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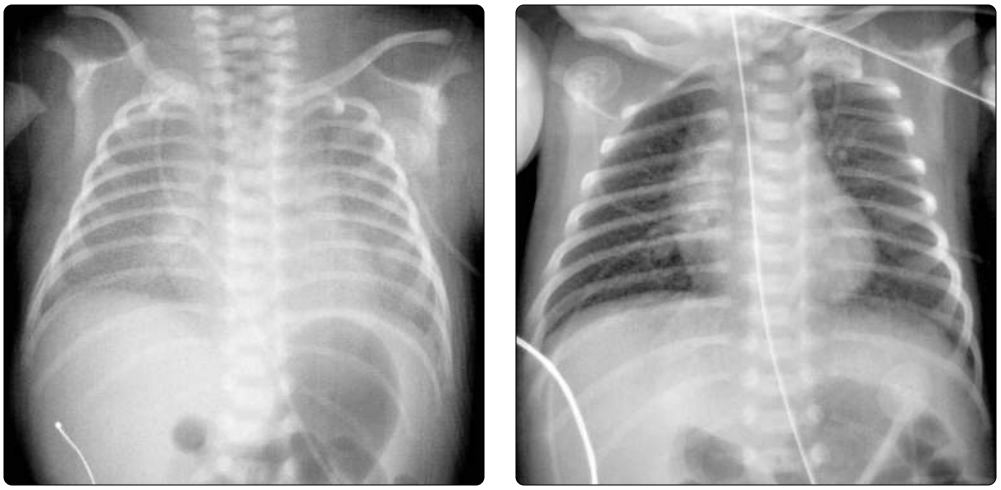
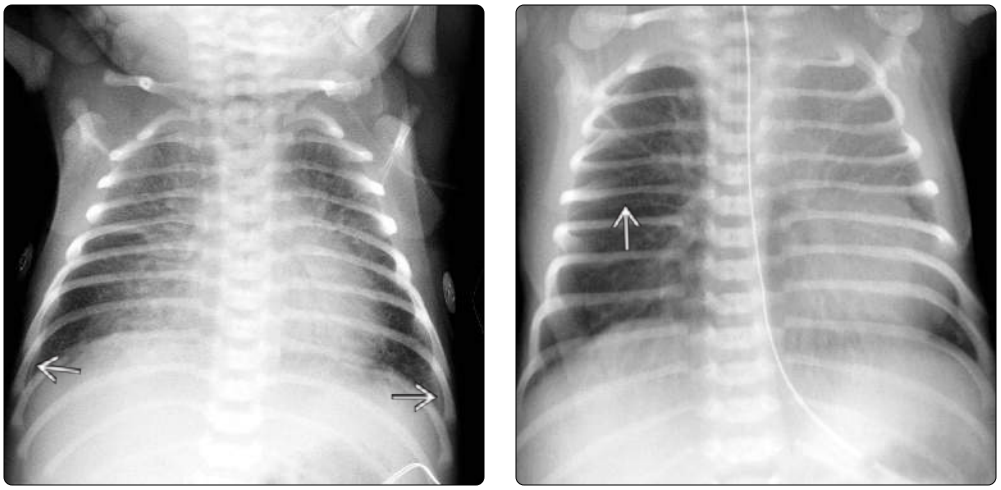
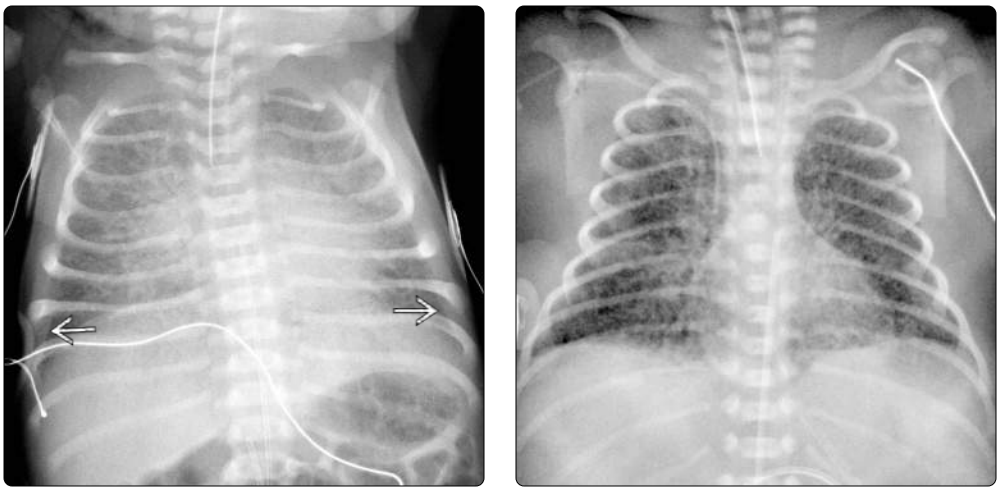


(Left) *AP radiograph of the chest shows hypoinflation with diffuse hazy opacity in a patient with tachypnea.* **(Right)** *AP radiograph of the chest shows interval resolution of the previously seen diffuse hazy opacities with increased aeration & inflation of the lungs in this patient with TTN.*



(Left) *AP radiograph of the chest in a full-term newborn patient with tachypnea shows small bilateral pleural effusions & perihilar opacity.* **(Right)** *AP radiograph of the chest in a 35-weeks gestation newborn with transient tachypnea demonstrates mild perihilar opacity with a small amount of fluid in the minor fissure .*



(Left) *AP radiograph of the chest in a full-term newborn with congenital heart disease shows bilateral diffuse pulmonary edema with bilateral pleural effusions . Notice how similar this appearance can be to patients with TTN.* **(Right)** *AP radiograph in a full-term infant with neonatal pneumonia shows diffuse bilateral nodular opacities. Notice how similar this can appear compared to patients with TTN.*

Pulmonary Interstitial Emphysema

KEY FACTS

TERMINOLOGY

- Pulmonary interstitial emphysema (PIE): Air within pulmonary interstitium & lymphatics
- Usually secondary to barotrauma of positive pressure mechanical ventilation in setting of prematurity, low birth weight, & surfactant deficiency disease (SDD)
  - Not always on mechanical ventilation

IMAGING

- Best clue: New "bubbly" cystic or linear lucencies within lung of intubated premature infant
- Can be limited to 1 lobe vs. bilateral & symmetric
- Small interstitial lucencies > > large focal/multifocal cysts
- Precursor to pneumothorax or pneumomediastinum
- PIE may rarely endure to form large air-filled cystic mass: "Persistent PIE"
  - CT shows pulmonary vessels as linear (dash) or round (dot) densities within gas collections

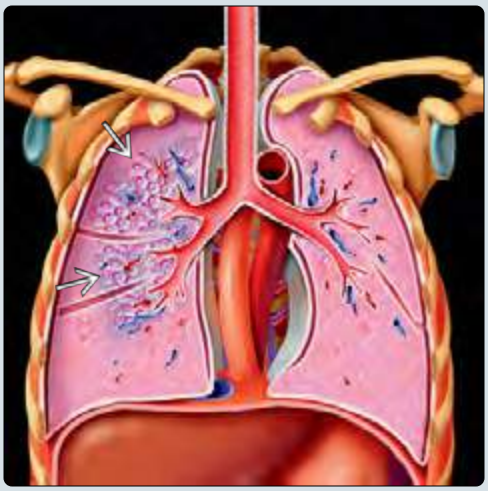
TOP DIFFERENTIAL DIAGNOSES

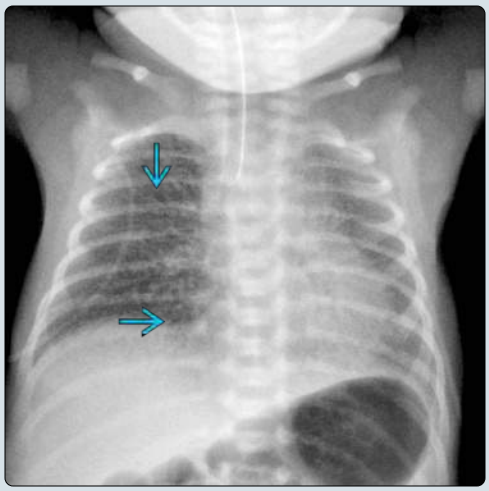
- Surfactant deficiency disease
- Air bronchograms in diffuse lung disease
- Chronic lung disease of prematurity
- Congenital lobar overinflation
- Congenital pulmonary airway malformation
- Pleuropulmonary blastoma

CLINICAL ISSUES

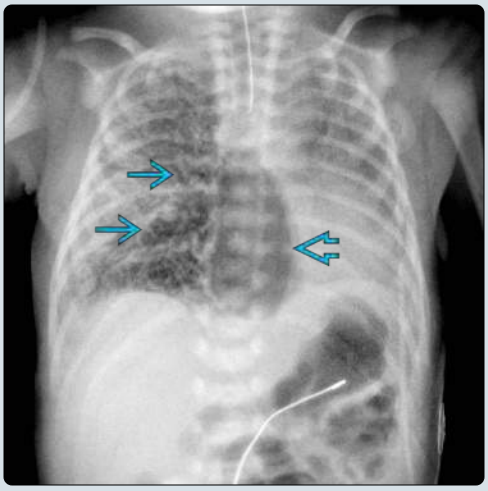
- 2-3% of neonatal ICU patients
- 20-30% of premature neonates with SDD
- Often asymptomatic, detected on neonatal ICU radiographs
- Usually occurs during first 10 days of life
- Usually transient with adequate therapy
- Primary therapy: ↓ mean airway pressure by switching from conventional positive pressure to high-frequency ventilation

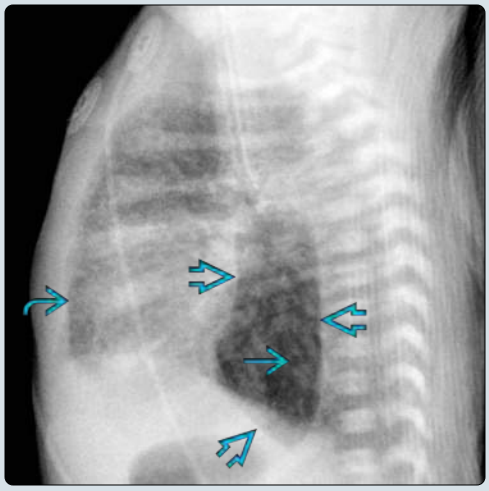
(Left) *Coronal graphic depicts round & linear foci of gas in the right lung parenchyma secondary to air escaping into the pulmonary interstitium.* **(Right)** *AP chest radiograph in an intubated, 2 day old, former 31-weeks premature infant with surfactant deficiency disease (SDD) shows typical diffuse granular pulmonary opacities. There are mild scattered linear & bubbly lucencies of pulmonary interstitial emphysema (PIE) in the right lung.*





(Left) *AP chest radiograph 24 hours later in the same patient shows more extensive bubbly & linear lucencies throughout the right lung, typical of PIE. A large inferomedial gas collection has also developed, shifting the heart leftward.* **(Right)** *Cross-table lateral radiograph of the same patient shows localized infraazygous posterior pneumomediastinum. Lucencies from the right lung PIE are superimposed on the collection. Note that there is no free gas in the nondependent chest to suggest a pneumothorax.*





TERMINOLOGY

Abbreviations

- Pulmonary interstitial emphysema (PIE)

Definitions

- Air within pulmonary interstitium & lymphatics
- Most commonly secondary to barotrauma of positive pressure mechanical ventilation in setting of prematurity, very or extremely low birth weight, & surfactant deficiency disease (SDD)
  - Part of "neonatal air leak syndrome": Air escapes tracheobronchial tree to reside in other spaces
    - Also includes pneumomediastinum, pneumothorax, pneumopericardium, pneumoperitoneum, pneumatocele, subcutaneous emphysema, air embolism
- May occur in neonate with pulmonary hypoplasia on mechanical ventilation or in setting of meconium aspiration or sepsis
- Rarely occurs in older patients with underlying lung disease or Valsalva leading to alveolar rupture
  - Stem cell transplant with graft vs. host disease & bronchiolitis obliterans

IMAGING

General Features

- Best diagnostic clue
  - New "bubbly" cystic or linear lucencies within lung of intubated premature infant
- Location
  - Extent of involvement may range from single lobe to widespread bilateral PIE
- Size
  - Ranges from small interstitial lucencies to large focal/multifocal cysts

Radiographic Findings

- Radiography
  - "Bubbly" cystic or linear lucencies in lung parenchyma
  - Lucencies typically uniform in size
  - Often radiate from hilum
  - Involved lung usually noncompliant, though this may be due to underlying SDD rather than superimposed PIE
  - Typically transient: May last days to weeks
    - Precursor to other complications: Pneumothorax, pneumomediastinum
  - PIE may rarely endure to form large air-filled cyst ("persistent PIE")
    - May act as mass lesion, compressing other thoracic structures to cause worsening respiratory distress
    - Most commonly affects left upper lobe

CT Findings

- Not routinely obtained to evaluate PIE
- May be utilized to evaluate persistent PIE & differentiate it from other neonatal lucent lung masses
  - Round or linear lucencies in lung interstitium; lucencies do not follow normal bronchial anatomy & may widen peripherally (unlike air bronchograms)
  - With prominent cysts of persistent PIE, pulmonary vascular branches are seen as linear (dash) or round (dot) soft tissue densities within gas collections
    - Characteristic dot-dash pattern in 82% of patients with persistent PIE

Imaging Recommendations

- Best imaging tool
  - Frontal radiograph of chest

DIFFERENTIAL DIAGNOSIS

Surfactant Deficiency Disease

- Often underlies PIE
- Uneven distribution of exogenous surfactant (for treatment of disease) may result in partial clearing of collapsed alveoli amidst granular & hazy opacities
  - Pattern of alternating distended & collapsed acini may mimic PIE

Air Bronchograms in Diffuse Lung Disease

- More common in lower lobes
- Follow normal bronchial course
- May be seen with pulmonary edema, hemorrhage, neonatal infections, surfactant dysfunction/deficiency

Chronic Lung Disease of Prematurity

- "Bubbly" lucencies of developing bronchopulmonary dysplasia (BPD) can appear similar to PIE
  - On CT, vessels not engulfed by interstitial air (unlike PIE)
- BPD seen at > 1 month of age
- Timing of onset: PIE abrupt, BPD gradual

Congenital Lobar Overinflation

- Found in perinatal period; often worsens after birth
- May present as lobar opacity on initial radiographs with gradual clearing & increasing expansion of entire lobe
  - Lobar architecture preserved but uniformly displayed without discrete cysts

Congenital Pulmonary Airway Malformation

- Lung mass of cystic lucencies of varying size
- Transition from fluid-filled to air-filled mass after birth

Pleuropulmonary Blastoma

- Rare tumor of young children (but uncommon in perinatal period)
- May be cystic or solid or mixed

PATHOLOGY

General Features

- ↑ alveolar pressure with barotrauma → disruption of alveolar basement membrane → air enters pulmonary interstitium
- Air distributes along bronchovascular structures, appearing on radiography as radiolucent "bubbly" cystic & linear foci
  - Compresses alveoli, impairing gas exchange, compliance, & possibly blood flow
- Air may further dissect, leading to pneumothorax or pneumomediastinum
- Cysts may enlarge due to ball-valve mechanism

Microscopic Features

- With persistent PIE
  - Multinucleated giant cells in interstitium in reaction to prolonged air trapping
  - Irregular cyst walls of fibrous tissue lined by histiocytes & giant cells

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often asymptomatic & detected incidentally on routine neonatal intensive care unit (NICU) radiographs
    - Precursor to other complications: Pneumothorax, pneumomediastinum
  - Impaired gas exchange
- Other signs/symptoms
  - Difficulty with ventilation secondary to pneumothorax

Demographics

- Age
  - Premature infants, typically < 10 days old
  - Rarely occurs in
    - Neonates with other lung diseases (pulmonary hypoplasia, meconium aspiration, sepsis)
    - Older children due to
      - Underlying lung disease
      - Valsalva with alveolar rupture
- Gender
  - No significant gender difference
- Ethnicity
  - No significant race predilection
- Epidemiology
  - 2-3% of neonatal ICU patients
  - 20-30% of premature neonates with SDD
    - Incidence ↓ with use of surfactant in appropriate population
  - Patients on positive pressure mechanical ventilation most at risk
    - Rarely develops in patients not on ventilator

Natural History & Prognosis

- Usually transient with adequate therapy
- Rarely develops into persistent PIE
- Complications: Pneumothorax, pneumomediastinum, ↓ pulmonary compliance, air embolus, chronic lung disease of prematurity (CLD/BPD)
- ↑ rates of intraventricular hemorrhage

Treatment

- Switch from conventional to high-frequency oscillatory ventilation or high-frequency jet ventilation
  - Lower mean airway pressures
- ↑ frequency of clinical & radiographic monitoring for complications such as pneumothorax
- Heliox with nitric oxide may be useful
- Steroids may be useful
- Additional options in persistent PIE
  - Selective intubation of uninvolved lung
  - Temporary balloon blockage of main bronchus of affected side
  - Decubitus positioning with affected side down
  - Surgical resection of large cystic foci reserved for unmanageable respiratory distress
    - ~ 53% of patients with persistent PIE in one series

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Age of patient & rapidity of development help differentiate PIE from BPD

Reporting Tips

- Early treatment may help avoid complications

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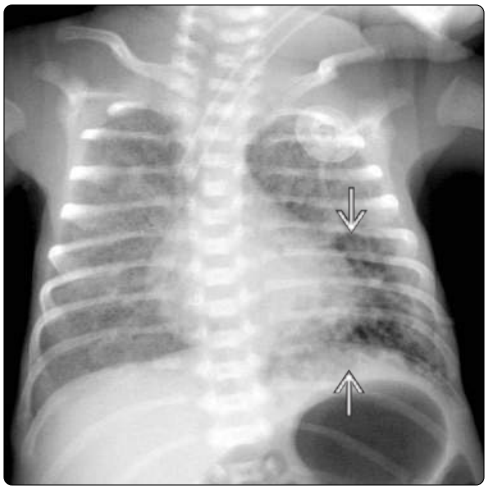
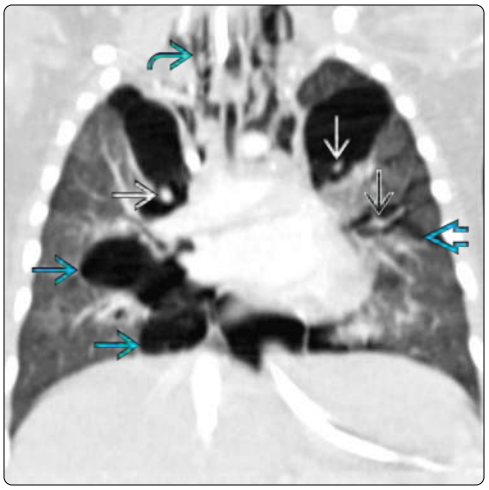
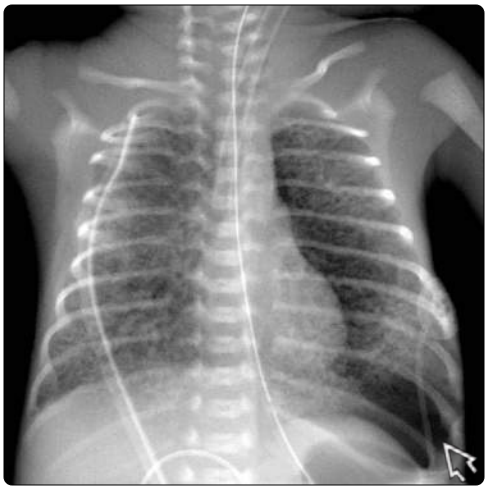
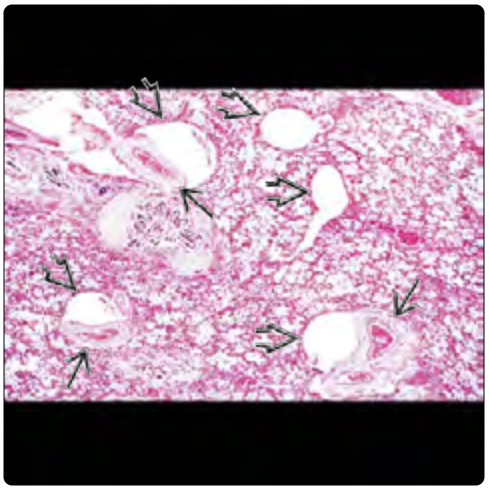


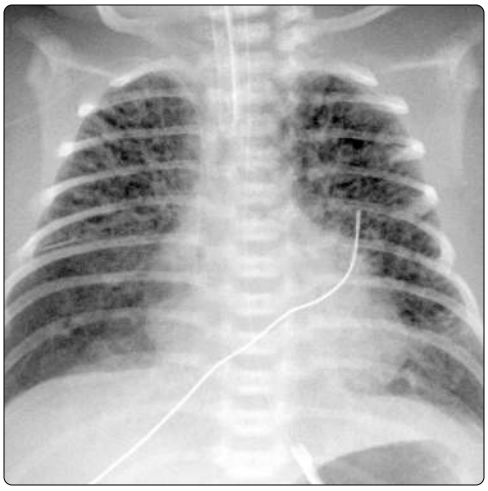


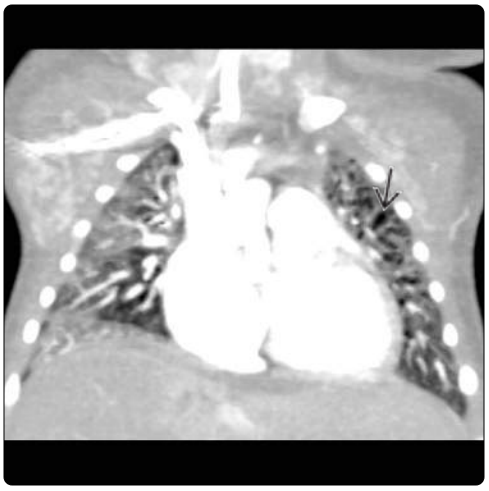
(Left) *AP radiograph shows cystic & linear lucencies from PIE in a patient with SDD. Note that only the left lower lobe is involved. The distribution of PIE can range from a single lobe to widespread bilateral involvement.* **(Right)** *Coronal CECT in a 6 month old with tracheal rings & PIE shows cystic & elongated gas collections within the pulmonary interstitium outlining several arteries & veins. Note the extension of pneumomediastinum into the neck.*





(Left) *AP chest radiograph shows diffuse bilateral PIE, worse on the left, with a subpulmonic left pneumothorax. Note the depression of the left hemidiaphragm, consistent with tension. The lack of associated lung collapse & mediastinal shift is likely due to lung noncompliance in the setting of SDD & PIE.* **(Right)** *Microscopic H&E image shows PIE as gas collections in the pulmonary interstitium/lymphatics. The foci of gas abut & surround the bronchial arteries.*





(Left) *AP chest radiograph shows bilateral upper lobe PIE in a premature infant with hazy pulmonary opacities due to SDD. Note the relative sparing of the lower lobes.* **(Right)** *Coronal CECT of the chest demonstrates linear lucencies in the interstitium of the left upper lobe (superimposed on a background of SDD). The lucencies in PIE do not follow the anatomy of a bronchus & may get wider near the periphery of the lung, helping to differentiate PIE from air bronchograms.*

Neonatal Pneumothorax

KEY FACTS

IMAGING

- Neonatal chest radiographs typically obtained supine
  - Clues to pneumothorax diagnosis on supine study
    - Large, hyperlucent hemithorax
    - Lucency cloaking diaphragm, mediastinum, &/or lung
    - Medial stripe sign: Lucency along mediastinum
    - Deep sulcus sign: Well-defined costophrenic sulcus
    - Gas-filled pleural sac herniated across midline
    - Visualization of anterior junction line if bilateral
  - Cross-table lateral view shows air anterior to lung
  - Edge enhancement on PACS may improve visibility
- Ultrasound: Absence of lung sliding & comet-tail artifact

TOP DIFFERENTIAL DIAGNOSES

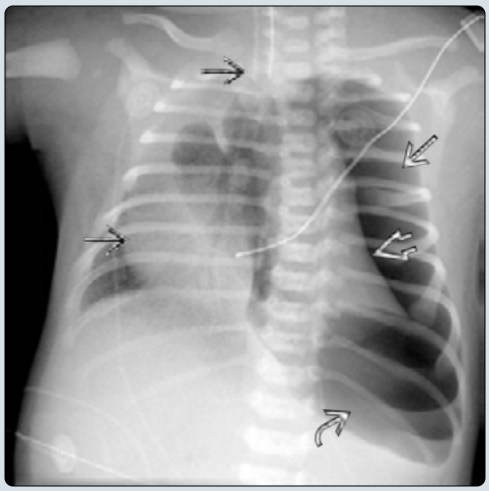
- Pneumomediastinum
- Air-filled mass
- Artifact
- Recent surgical evacuation of hemithorax

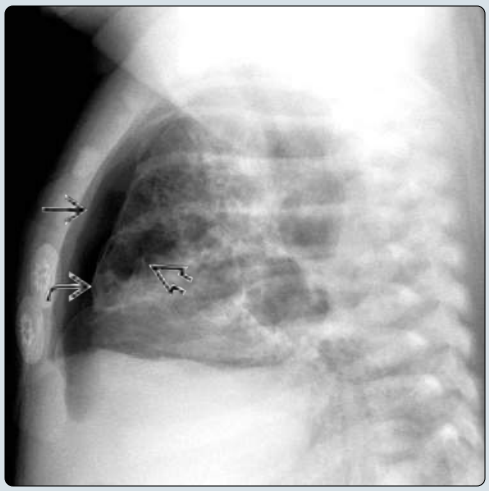
PATHOLOGY

- Over distention & rupture of alveoli
  - Directly into pleural cavity → pneumothorax
  - Into lung interstitium first → interstitial emphysema
- Risk factors: Low birth weight, premature or postmature gestation, male gender, surfactant deficiency disease, meconium aspiration, pulmonary hypoplasia, resuscitation at birth, mechanical ventilation, macrosomia

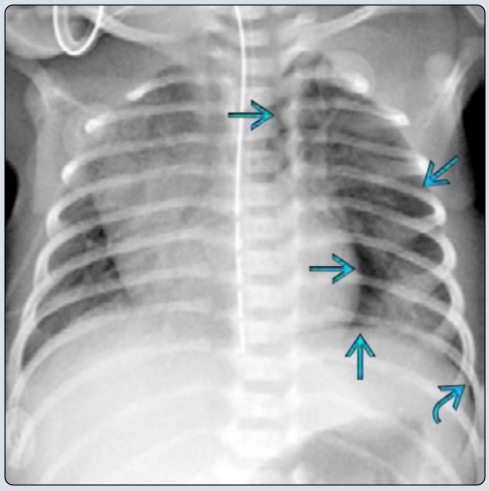
CLINICAL ISSUES

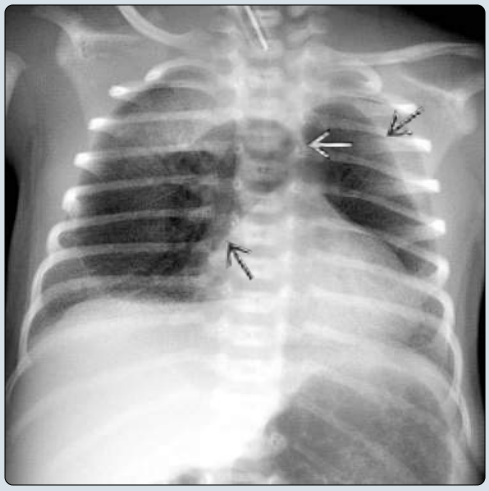
- Common signs/symptoms: Ipsilateral ↓ breath sounds, respiratory distress, cyanosis, retractions, grunting, nasal flaring, chest asymmetry (↑ on affected side), hypercapnia, hypoxemia; tension pneumothorax → hypotension, shock
- Treatment
  - Expectant/supportive management in asymptomatic patients or those with mild disease
  - Needle aspiration or chest tube drainage if symptomatic





(Left) *Supine chest radiograph of a 2-day-old term neonate in respiratory distress shows a lucent left hemithorax, collapse of the left lung, depression of the diaphragm, & rightward mediastinal shift, consistent with a left tension pneumothorax. Findings were immediately communicated.* **(Right)** *Cross-table lateral view of the chest in a 1-day-old full-term girl shows air anterior to the lung, consistent with a pneumothorax. Cystic lucencies relate to her known congenital pulmonary airway malformation.*





(Left) *Supine chest radiograph of a 2-day-old neonate born at 34 weeks shows lucency "cloaking" the left hemithorax with a well-defined left costophrenic angle as compared to the right. This appearance was due to a moderate left pneumothorax that caused mediastinal shift to the right.* **(Right)** *Supine chest radiograph of a 2-day-old preterm neonate with surfactant deficiency disease shows lucencies outlining the anterior junction line, consistent with bilateral pneumothoraces.*

TERMINOLOGY

Definitions

- Air in pleural space between visceral & parietal pleura

IMAGING

Radiographic Findings

- Neonatal chest radiograph typically obtained with patient in supine position
- Air collects anterior & medial → pleural line often not seen
- Clues to pneumothorax in supine position
  - Large, hyperlucent hemithorax
  - Lucency cloaking diaphragm, mediastinum, &/or lung
    - Thymus may be compressed → "pseudomass"
  - Medial stripe sign: Lucency along mediastinal edge
  - Deep sulcus sign: Well-defined costophrenic sulcus
  - Gas-filled pleural sac herniated across midline
    - Appearance similar to pneumomediastinum
  - Bilateral anterior pneumothoraces displace thymus & allow visualization of anterior junction line
    - Anterior junction line usually not seen in neonates
    - Uneven tension → support devices displaced away from side of greater tension
- Cross-table lateral view
  - Intrapleural air lucency anterior to lung & mediastinum
  - Minimizes neonate manipulation vs. decubitus view
- Tension pneumothorax
  - Large pneumothorax with lung collapse
    - Diseased "stiff" lungs may not collapse despite tension
  - Flattened or everted hemidiaphragm
  - Bulging intercostal spaces
  - Mediastinal shift toward/into contralateral hemithorax

Ultrasonographic Findings

- Absence of lung sliding & comet-tail artifact

Imaging Recommendations

- With equivocal supine chest radiograph + clinical concern, obtain cross-table lateral view
- Edge enhancement on PACS may improve visibility

DIFFERENTIAL DIAGNOSIS

Pneumomediastinum

- Air confined to upper retrosternal space
- Mediastinal shift unlikely
- Lifting of thymus: Spinnaker sail sign

Artifact

- Skin fold
- Materials external to patient
- Mach effect

Air-Filled Mass

- Congenital pulmonary airway malformation (CPAM)
- Congenital lobar emphysema
- Congenital diaphragmatic hernia (CDH)

Recent Surgical Evacuation of Hemithorax

- Early radiographs after CDH repair or large CPAM resection demonstrate negative pressure ex vacuo pneumothorax
- Fills with fluid; ultimately ↓ in size due to lung growth

PATHOLOGY

General Features

- Uneven alveolar ventilation, air-trapping, ↑ transpulmonary pressure → alveolar overdistension → rupture into pleural cavity → pneumothorax
  - Rupture may first occur into interstitium (pulmonary interstitial emphysema) with ultimate dissection to mediastinum or pleural cavity
- Risk factors: Low birth weight, premature or postmature gestation, male gender, surfactant deficiency disease, meconium aspiration, pulmonary hypoplasia, resuscitation at birth, mechanical ventilation, macrosomia

CLINICAL ISSUES

Presentation

- Timing of pneumothorax presentation
  - Infants > 2,500 g: Median of 5.5 hours after birth
  - Infants < 2,500 g: Median of 2nd day of life
- Most common signs/symptoms: Ipsilateral ↓ breath sounds, respiratory distress, cyanosis, retractions, grunting, nasal flaring, chest asymmetry (larger on affected side), hypercapnia, hypoxemia
- Asymptomatic pneumothoraces more common in non-low birth weight neonates
- Tension pneumothorax may result in hypotension, shock

Natural History & Prognosis

- Incidence: 1% of term births, 6-7% of preterm births
  - ↑ morbidity & mortality in preterm infants
- High-frequency positive pressure ventilation may ↓ incidence compared to conventional mechanical ventilation

Treatment

- Expectant/supportive management in asymptomatic patients & those with mild disease
- Needle aspiration or chest tube drainage if symptomatic

DIAGNOSTIC CHECKLIST

Reporting Tips

- Unexpected or tension pneumothoraces should be communicated to clinical team immediately

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KEY FACTS

TERMINOLOGY

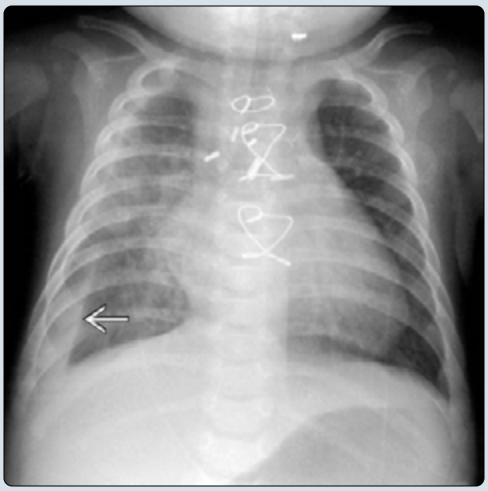
- Lymphatic fluid in pleural space secondary to congenital or acquired conditions, including obstruction, congenital anomaly, increased pressure, impaired drainage, or trauma to thoracic duct

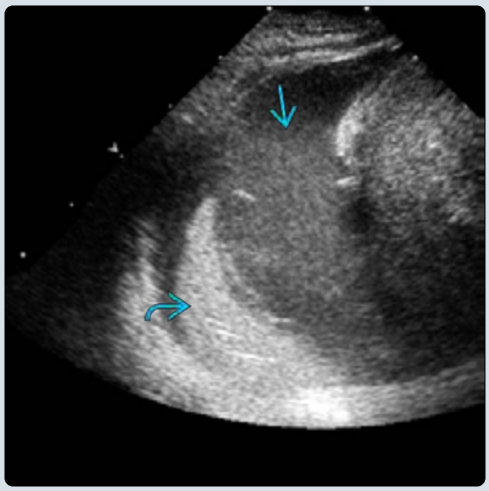
IMAGING

- US, CT, MR may be done to discover underlying cause of chylothorax & evaluate treatment options
  - Persistent pleural effusion following cardiac surgery
  - Congenital chylothorax from Turner or Noonan syndrome
  - Various associated lymphatic disorders, including conducting channel anomalies, generalized lymphatic anomaly, Gorham-Stout disease, discrete cystic lymphatic malformations, & pulmonary lymphangiectasia
- Lymphangiogram (conventional, nuclear, or MR): Used to investigate suspected central conducting channel anomalies
  - Disorders (including atresia, obstruction, disruption, or dysmotility) may show abrupt halt to visualization of normal lymphatic pathways, collaterals, delayed transit, &/or abnormal accumulations

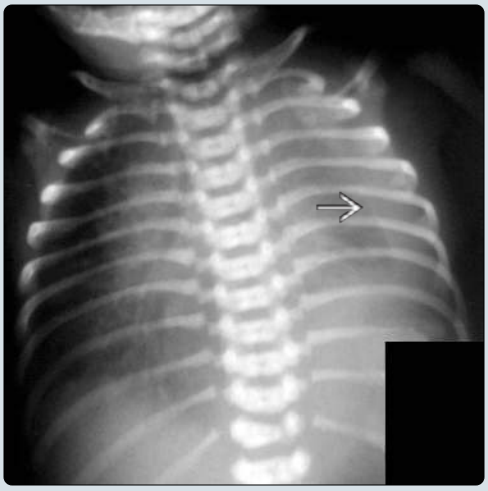
CLINICAL ISSUES

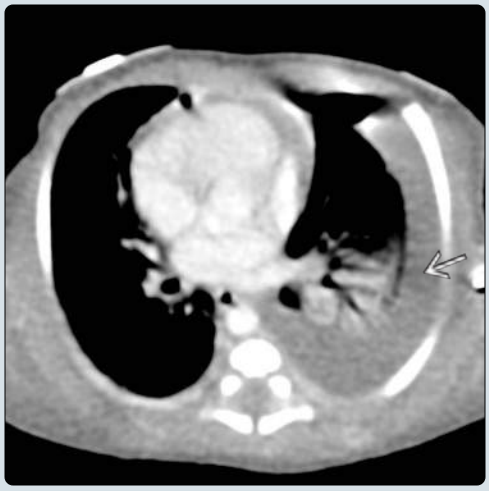
- Symptoms may include tachypnea & dyspnea, generalized edema, chylous ascites, immunosuppression, protein-losing enteropathy (if gut involved)
- Treatment options include: Fat-restricted diet, rapamycin/sirolimus, thoracentesis or draining procedure, thoracic duct ligation or embolization, microsurgical thoracic duct repair, pleurodesis
  - Reports of cure with ethiodized oil lymphangiography
- Thoracic duct injury may resolve spontaneously in 50%
- Poorer prognosis with syndromes, multiple sites of chylous fluid accumulations, prematurity





(Left) *AP radiograph in a 4-month-old infant status post surgery for congenital heart disease shows a moderate right chylothorax from a thoracic duct injury. It is not possible to assess the complexity of fluid on radiography.* **(Right)** *Transverse US of the right chest in the same 4-month-old patient status post heart surgery & thoracic duct injury shows a large, mildly echogenic pleural effusion. Notice the compressed echogenic right lung surrounded by the effusion.*





(Left) *AP radiograph shows a large congenital left chylous effusion in a patient with Turner syndrome.* **(Right)** *Axial CECT shows a large left pleural effusion in a patient with Turner syndrome from a congenital chylothorax. Congenital chylothorax is more commonly seen on the left than on the right.*

TERMINOLOGY

Definitions

- Lymphatic fluid in pleural space secondary to congenital or acquired conditions, including obstruction, congenital anomaly, increased pressure, impaired drainage, or trauma to thoracic duct

IMAGING

General Features

- Best diagnostic clue
  - Isolated unilateral pleural effusion in newborn: May be due to birth trauma
  - Persistent pleural effusion following cardiac surgical repairs: Left thoracotomy procedures, Glenn & Fontan procedures; fluid accumulation occurs after feedings resumed
  - Newborn with congenital heart disease: Turner syndrome, Noonan syndrome
  - Lymphangiectasia in newborn with bilateral interstitial edema, anasarca, protein-losing enteropathy, & fluid accumulations in body cavities: Likely due to conducting channel disorder
  - Macroscopic lymphatic malformation of neck & chest
  - Bone disease with chylothorax: Gorham-Stout disease ("vanishing bone disease"), generalized lymphatic anomaly (GLA)

Radiographic Findings

- Persistent nonspecific pleural effusion
  - Left-sided fluid more common
  - Associated disorders dictate other findings

Ultrasonographic Findings

- Anechoic or mildly echoic fluid without septations

CT Findings

- CECT
  - Nondescript pleural fluid
  - Lymphangiectasia demonstrates diffuse pulmonary interstitial thickening
  - Poorly defined paraspinal soft tissue infiltration from abnormal lymphatics
  - Macro- &/or microcystic lymphatic malformation may infiltrate mediastinum & pleura
    - ± bone lesions
      - Nonprogressive, well-defined cysts without cortical destruction in GLA
      - Progressive localized destruction of multiple adjacent bones by microcystic lymphatic malformation in Gorham-Stout disease
  - ± solid mediastinal mass obstructing vessels
  - ± visceral (especially splenic) cystic lymphatic malformations

Lymphangiogram

- Used to investigate suspected central conducting channel anomalies
  - Chylous accumulations &/or generalized edema without other explanations
- Can be performed with nuclear medicine, MR, or conventional fluoroscopy
- Depending on specific question & modality being employed, specific agent injected into dermal or nodal lymphatics of proximal or distal lower extremity
- Extremity & body subsequently imaged over 10-60 minutes while agent travels proximally through central lymphatic system to cisterna chyli & thoracic duct & ultimately empties into left subclavian vein
- Disorders (including atresia, disruption, dysmotility) may show abrupt halt to visualization of normal tiny channels
  - ± visualization or adjacent collaterals, leak into adjacent cavity, or localized extraluminal accumulation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Tachypnea & dyspnea
  - Classic symptoms of pleural effusion
    - ↓ breath sounds, usually with cough
- Other signs/symptoms
  - Generalized edema, chylous ascites, immunosuppression, protein-losing enteropathy (if gut involved)

Natural History & Prognosis

- Depends on etiology
- Thoracic duct injury may resolve spontaneously in 50%
- Poorer prognosis with syndromes, multiple sites of chylous fluid accumulations, prematurity

Treatment

- Conservative or medical treatment
  - ↓ chyle production
    - Fat-restricted oral diet
    - Total parenteral nutrition
  - Rapamycin/sirolimus of benefit with some lymphatic disorders, not clearly shown in conducting channel anomalies
  - Octreotide
- Interventions may be indicated with chyle leak > 1 L/day for 5 days or persistent leak for > 2 weeks
  - Thoracentesis or draining procedure of pleural space
  - Thoracic duct ligation or embolization
  - Reports of cure with ethiodized oil lymphangiography
  - Microsurgical thoracic duct repair
  - Pleurodesis
- Treat underlying cause of chylothorax if possible

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Bronchopulmonary Dysplasia

KEY FACTS

TERMINOLOGY

- Current definition
  - Chronic lung disease of premature infants born at < 32 weeks gestation
  - O_2 dependency for at least 28 days
  - Failure of O_2 challenge at 36 weeks postmenstrual age
  - Chest radiograph abnormalities no longer required
- Old BPD
  - Larger, later preterm infants with prolonged mechanical ventilation & O_2 therapy
- New BPD
  - More diffuse but overall milder disease of earlier, smaller preterm infants

IMAGING

- Classic old BPD appearance
  - Heterogeneous, hyperinflated lung parenchyma
  - Patchy, small round lucencies separated by coarse, reticular, & band-like opacities
- New BPD
  - Lung appearance may be nearly normal early with progressive diffuse hazy opacification
  - Severe surfactant deficiency disease patients may follow old imaging patterns
- Similar chronic chest CT findings of old & new BPD
  - Peripheral abnormalities may predominate
  - Cystic/emphysematous change
  - Subpleural cysts & triangular opacities
  - Linear, reticular opacities + parenchymal bands
  - Foci of air-trapping on expiratory images

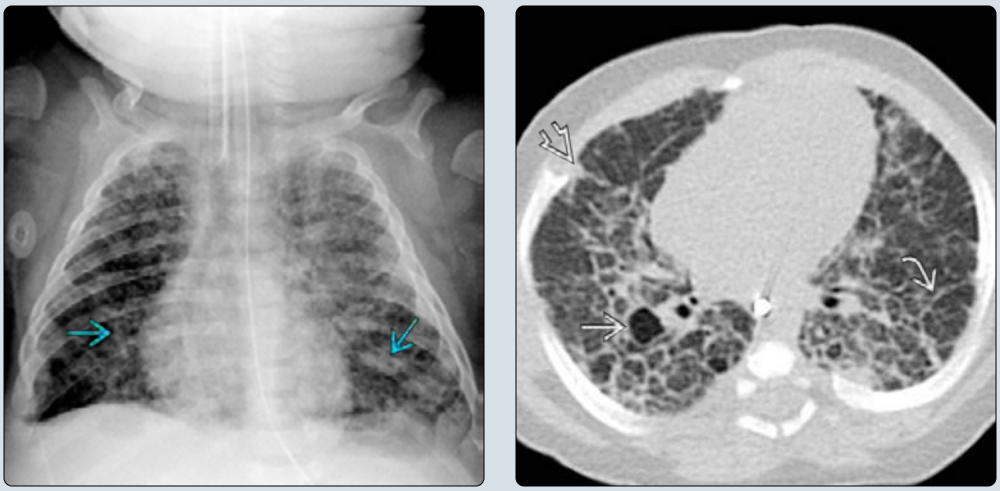
PATHOLOGY

- Strongest predictors: Prematurity & low birth weight

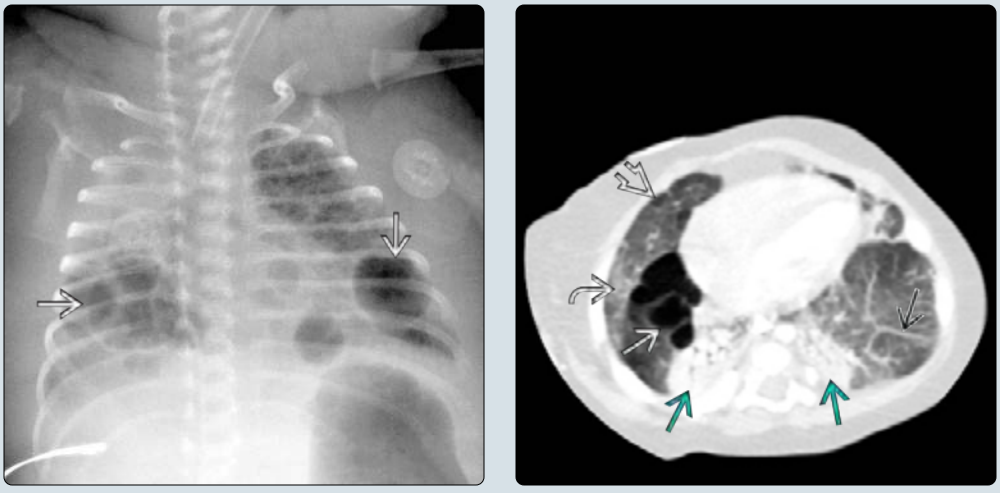
CLINICAL ISSUES

- Most common chronic pulmonary disease of infancy
- Potentially lifelong complications

(Left) *AP radiograph in a 4-month-old former 24-week gestation premature infant with bronchopulmonary dysplasia (BPD) shows mild bilateral hyperexpansion with generally coarsened lung markings. There are patchy foci of hazy parenchymal opacity intermixed with foci of hyperlucency due to air trapping.* **(Right)** *Axial NECT in the same patient shows coarse reticular markings with parenchymal bands, triangular subpleural opacities, & scattered cysts.*



(Left) *AP radiograph of the chest in a neonate with chronic lung disease shows development of bilateral pneumatoceles superimposed on regions of hyperinflation & atelectasis. The background parenchymal markings are coarsened.* **(Right)** *Axial CECT demonstrates pneumatocele formation in a premature patient with chronic lung disease. Findings include patchy ground-glass opacities, bibasilar consolidations, parenchymal bands, & foci of air trapping.*



TERMINOLOGY

Abbreviations

- Bronchopulmonary dysplasia (BPD), chronic lung disease (CLD) of prematurity

Definitions

- Old definition (1979): 28 days of oxygen (O_2) exposure + characteristic changes on radiographs
- New NIH consensus definition (2001): Chronic disease of premature infants born at < 32 weeks gestation with O_2 dependency for at least 28 days
  - Assessed at 36 weeks postmenstrual age (PMA)
  - Chest radiograph abnormalities no longer required
- Physiologic modification (2004): Specific O_2 challenge at 36 weeks PMA; has lead to more standardized diagnosis
- Classic or old BPD of early descriptions: Larger, later preterm infants with prolonged mechanical ventilation & O_2 therapy leading to inflammation & fibrosis
- New BPD: More diffuse but overall milder disease of earlier, smaller preterm infants in setting of antenatal steroids, postnatal surfactant administration, & reduced ventilator/O_2 intensity yielding less inflammation & fibrosis

IMAGING

General Features

- Best diagnostic clue
  - Varying degrees of hyperinflation, cysts, hazy opacities, & coarse linear opacities diffusely in premature infant

Radiographic Findings

- Classic old BPD
  - Characteristic appearance of established BPD
    - Heterogeneous lung parenchyma with patchy focal lucencies separated by coarse, reticular, & band-like opacities of fibrosis, atelectasis
  - More opacities in upper lobes vs. hyperinflation at bases
  - Lateral radiograph may show relatively narrow AP diameter of chest for degree of hyperinflation (differing from bronchiolitis & asthma)
  - Classic stages (develop over 1 month)
    - I: Diffuse, granular opacities of surfactant deficiency disease (SDD)
    - II: Complete opacification
    - III: Rounded, small lucencies + irregular opacities
    - IV: Hyperinflation, enlarging lucencies, linear bands
- Current new BPD
  - Lung appearance may be nearly normal early
  - Progressive diffuse hazy opacification that may ultimately lead to cystic foci with air-trapping, parenchymal bands

CT Findings

- HRCT
  - Overlapping chest CT findings between old & new BPD
    - Findings evolve over time with trend of improvement
  - Peripheral abnormalities may predominate
  - Cystic emphysematous change in 1st months; mosaic attenuation more likely after age 2 years
  - Subpleural cysts & triangular opacities
  - Linear, reticular opacities
  - Architectural distortion
  - Foci of air-trapping on expiratory images
  - Positive correlation of CT findings with pulmonary function

MR Findings

- GRE cine imaging to evaluate cardiac RV function, which may be ↓ in patients with BPD

Ultrasonographic Findings

- Echocardiography to evaluate RV function

Nuclear Medicine Findings

- V/Q scan: Ventilation/perfusion mismatch

Imaging Recommendations

- Protocol advice
  - CT optimized with anesthesia or controlled ventilation

DIFFERENTIAL DIAGNOSIS

Pulmonary Interstitial Emphysema

- Air in interstitium of premature neonate, usually on positive pressure ventilation
- Cystic & linear lucencies, often in background of SDD
  - May create unique dot-dash appearance of interstitial air collections surrounding pulmonary vessels
- Usually rapid onset in 1st 10 days of life
- Typically transient with switch to high-frequency ventilation

Meconium Aspiration

- Aspiration of meconium-stained amniotic fluid → toxic pneumonitis
- Usually in post-mature infants ± distress in utero
- Rapid onset of radiographic changes
- Coarse perihilar opacities with hyperinflation ± pneumothorax, pneumomediastinum

Neonatal Pneumonia

- Most common organisms: Group B β-hemolytic *Streptococcus* & *Escherichia coli*
- Often bilateral & diffuse, but may be unilateral
- Pleural effusion common, unlike BPD

Congenital Pulmonary Airway Malformation

- Often fluid-filled multicystic mass at birth with progressive aeration due to airway communication
- Typically involves 1 lobe
- Smaller lesions may present later in life

Congenital Lobar Overinflation

- Opacified lobe with progressive aeration & expansion
- Asymptomatic until significant compression of adjacent lung &/or mediastinum occurs

PATHOLOGY

General Features

- Etiology
  - Strongest predictors: Prematurity & low birth weight
    - BPD incidence ↑ with earlier gestational ages & lower weights

- Definite risk factors: Fetal growth restriction, mechanical ventilation, higher levels of inspired supplemental oxygen, postnatal sepsis
- Not definite (or controversial) risk factors: Lack of antenatal corticosteroids during preterm labor, chorioamnionitis, patent ductus arteriosus, gastroesophageal reflux
- Many other possible associations

- Genetics
  - No known genetic predisposition

Microscopic Features

- Old BPD
  - Airway injury, inflammation, & metaplasia
  - Smooth muscle hypertrophy of airways & vessels
  - Patchy atelectasis, emphysema, & septal fibrosis
- New BPD
  - Uniform inflation, less heterogeneity
  - ↓ fibrosis, inflammation
  - Impaired vascular & alveolar growth
    - Diminished number of alveoli, which appear large & simplified
    - Dysmorphic vessels, abnormal capillary distribution

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Sustained O_2 requirement in premature neonate
  - Many new BPD patients have mild respiratory disease initially followed by progressive deterioration
  - Patients with severe SDD may progress like old BPD

Demographics

- Age
  - Premature infants < 32 weeks gestation at birth
- Gender
  - M:F = 2-5:1
- Epidemiology
  - Most common chronic pulmonary disease of infancy
  - No significant change in incidence of BPD since initial description
    - Due to ↑ survival of very premature infants
  - BPD develops in
    - 25% of infants weighing < 1,500 g at birth, 50% of those weighing < 1,000 g
    - 68% born at 22-26 weeks gestation
    - 10,000-15,000 infants in USA annually

Natural History & Prognosis

- Earlier detection of lung function abnormalities portends worse prognosis, often with lifelong pulmonary problems
- ↑ risk of pulmonary infections in 1st 2 years of life with ↑ morbidity & mortality, especially with respiratory syncytial virus (RSV)
- > 50% hospital readmission rate during infancy
- Pulmonary hypertension in up to 25% of BPD patients
  - Up to 48% 2-year mortality in this group
- Slowly improving pulmonary function with fewer respiratory infections later in childhood
- Abnormal pulmonary function with ↑ airway hyperreactivity & obstruction may persist into adulthood
  - Clinically similar to asthma: Wheezing, coughing, dyspnea, exercise-intolerance
- ↑ risk of emphysema as young adult
- Beyond lung function
  - Neurodevelopmental delay
  - Growth failure

Treatment

- Many conflicting results in BPD prevention/therapy trials
- For prevention
  - Prenatal administration of steroids to mother
  - Exogenous surfactant administration
  - Gentle ventilation (pressure limited, volume targeted)
  - Low rather than high inspired oxygen concentrations
  - Closure of patent ductus arteriosus
  - Inhaled nitric oxide
  - Vitamin A
  - Caffeine
- Limited roles for pharmacologic intervention during developing or established BPD: Inhaled vs. systemic steroids, diuretics, bronchodilators, nitric oxide, sildenafil

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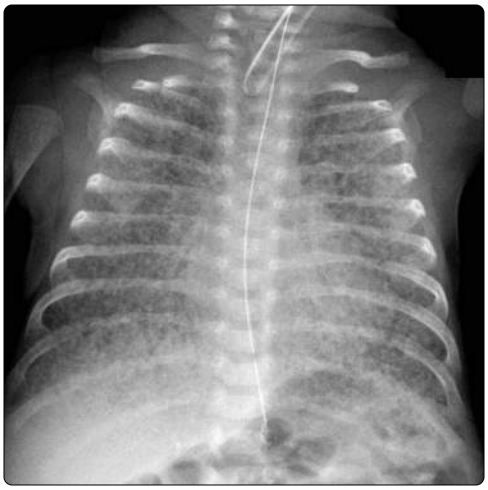
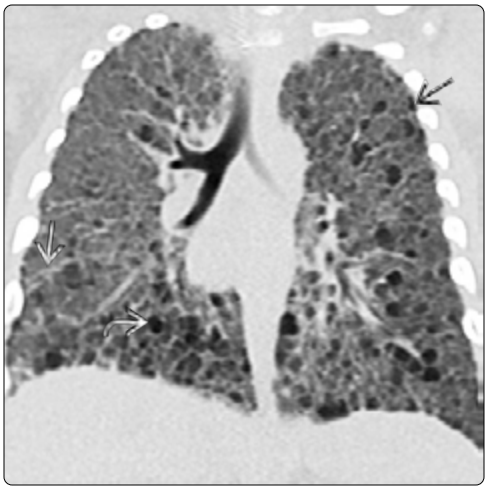
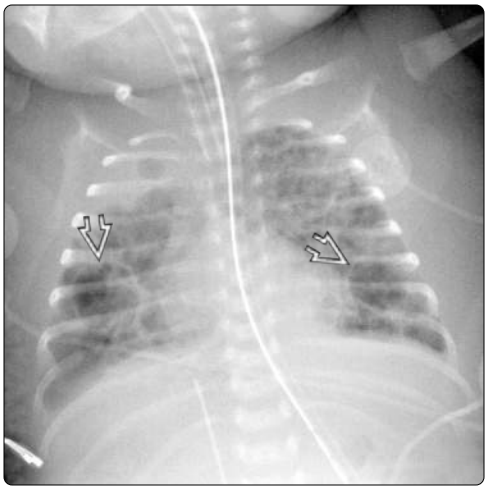
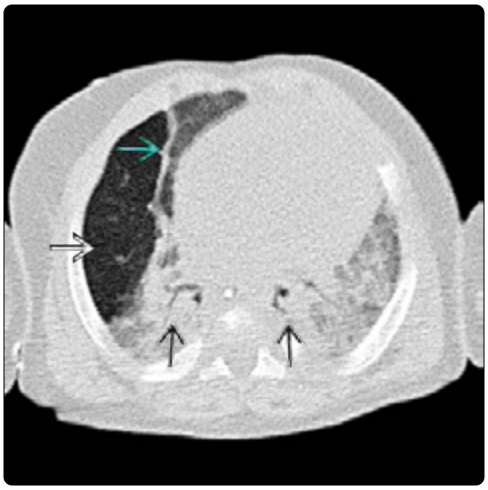


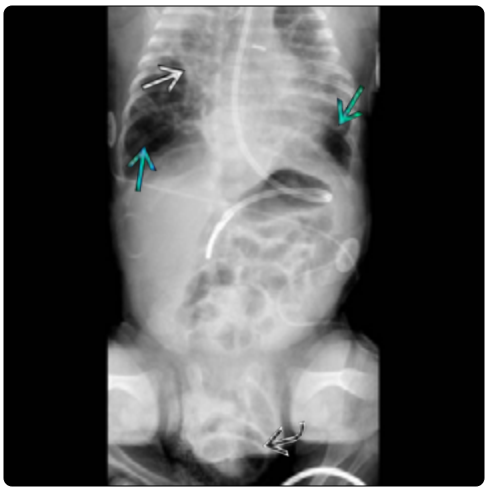


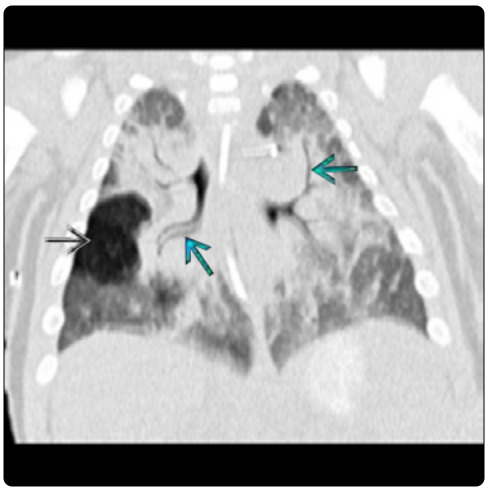
(Left) *AP radiograph in a 5-month-old former 26-week gestation premature infant with BPD shows generalized pulmonary hyperexpansion with diffuse hazy opacities & coarse interstitial markings.* **(Right)** *Coronal NECT during inspiration in an infant with BPD shows numerous parenchymal & subpleural cysts with generalized ground-glass opacities & parenchymal bands.*





(Left) *AP radiograph in a premature patient with chronic lung disease shows bilateral pneumatoceles in the setting of coarse linear & patchy hazy opacities.* **(Right)** *Axial NECT shows air trapping in the right mid lung with a coarse interstitial band & bibasilar consolidations in a patient with chronic lung disease of prematurity.*





(Left) *AP radiograph of the chest & abdomen in a patient with BPD shows coarse interstitial markings with hyperinflation of the lower lobes & a left inguinal hernia containing bowel. Hernias are common & often secondary to chronic hyperinflation of the lungs causing increased intraabdominal pressure.* **(Right)** *Coronal CECT in an infant with BPD shows air trapping in the right lower lobe & perihilar consolidations with prominent air bronchograms.*

Umbilical Catheter Positions and Complications

KEY FACTS

TERMINOLOGY

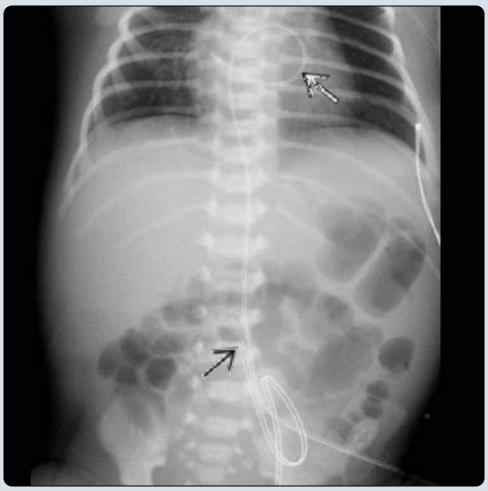
- Umbilical venous catheter (UVC)
  - Normal course: Umbilical vein → umbilical recess → left portal vein → ductus venosus → middle or left hepatic vein → inferior vena cava (IVC) → right atrium (RA)
  - Indications: Central venous access in ill/premature neonate for fluids/medications, total parenteral nutrition (TPN), exchange transfusion, venous pressure monitoring
- Umbilical arterial catheter (UAC)
  - Normal course: Umbilical artery → internal iliac artery → common iliac artery → aorta
  - Indications: Frequent blood sampling, monitoring of arterial pressures, angiography, exchange transfusion

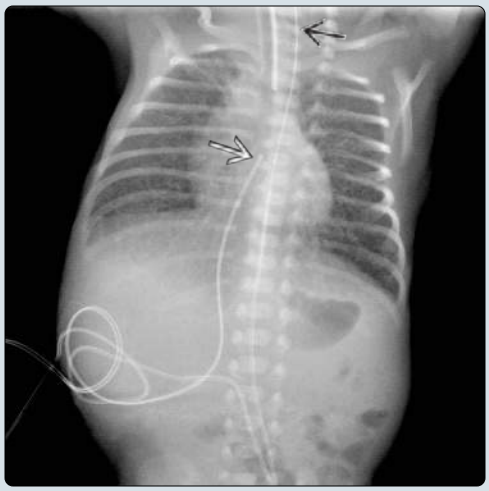
IMAGING

- UVC tip optimal location: IVC-RA junction
  - At/just above diaphragm, usually T8-T9
- UAC tip optimal location
  - Ideal high line: Descending thoracic aorta (T6-T10)
  - Acceptable low line: Distal abdominal aorta (L3-L4)
- Complications include
  - UVC or UAC: Malposition, thrombosis, infection
  - UVC: Hepatic laceration, hematoma, TPN extravasation (intrahepatic or intraperitoneal), arrhythmia, atrial perforation, pericardial effusion
    - Hepatic extravasation may mimic mass
  - UAC: Aneurysm/pseudoaneurysm

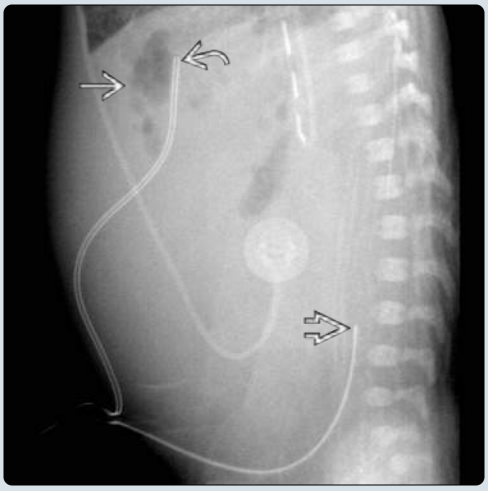
DIAGNOSTIC CHECKLIST

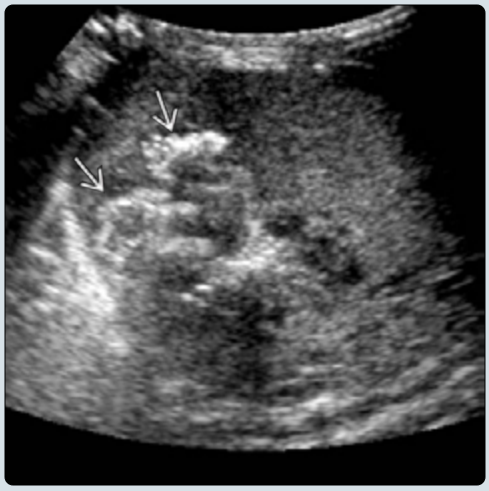
- Correct position must be confirmed prior to use
- ~ umbilical catheter position most quickly assessed by AP chest/abdomen radiograph
  - Cross-table lateral view can provide additional detail
- Ultrasound most reliable way to confirm appropriate UVC position initially
  - Also useful if vascular anatomy distorted by anomaly
- Ultrasound best modality to assess complications





(Left) *AP radiograph shows a UVC coiled over the left atrium (LA) ➡ after traversing the right atrium (RA) & a patent foramen ovale (PFO). It is unclear if the UVC has crossed the PFO a 2nd time back into the RA or lies at an LA pulmonary vein ostium. There is an acceptable low position of the UAC tip ⇨ at the top of L4.* **(Right)** *AP radiograph shows malposition of a UAC ⇨ into the left common carotid artery, though the tip is not included on this image. A malpositioned UVC tip ➡ courses toward the LA across a PFO.*





(Left) *Cross-table lateral radiograph shows an abnormal vertical course of a UVC ➡ in the liver with an associated parenchymal gas & fluid collection ➡. The UAC tip ➡ lies at the lower abdominal aorta.* **(Right)** *Longitudinal ultrasound of the liver in the same patient shows a heterogeneous hepatic collection containing echogenic foci of gas ➡. The UVC tip was seen in an extravascular location just inferior to this collection. The gas may be from recent infusion or superimposed infection.*

TERMINOLOGY

Definitions

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  - Indications: Central venous access in ill/premature neonate for fluids/medications, total parenteral nutrition (TPN), exchange transfusion, venous pressure monitoring
- Umbilical arterial catheter (UAC)
  - Normal course: Umbilical artery → internal iliac artery → common iliac artery → aorta
  - Indications: Frequent blood sampling, monitoring of arterial pressures, angiography, exchange transfusion

IMAGING

Radiographic Findings

- UVC tip optimal location: IVC-RA junction
  - Best radiographic correlate controversial
    - At/just above diaphragm; vertebral levels variable, ~ T7-T11 (most recommend T8-T9)
  - Small amount of gas may be visualized near tip (normal)
- UAC tip optimal location
  - Ideal high line: Descending thoracic aorta, T6-T10
    - T5 or above: Risk to aortic arch branches
    - T12-L1: Risk to abdominal aortic branches
  - Acceptable low line: Distal abdominal aorta, L3-L4
- Incorrect position in > 50% of 1st attempts
  - Common UVC malposition
    - Low (below T10): Umbilical vein or recess
    - Wrong vessel: Portal branch (left or right) or main portal, splenic, or superior mesenteric vein
    - High: RA, superior vena cava (SVC); right ventricle (via tricuspid valve); left atrium (via foramen ovale), pulmonary vein
  - Common UAC malposition
    - Low: Internal iliac artery or branches (e.g., superior gluteal, inferior gluteal, pudendal)
    - Wrong vessel: Celiac, superior mesenteric artery (SMA), renal arteries
    - High: Aortic arch or branch artery; pulmonary artery (via ductus arteriosis)
- Correctly positioned catheter may be shifted or distorted with diaphragmatic hernia, large thoracic/abdominal mass, or congenital heart disease

Ultrasonographic Findings

- Can be used to confirm catheter position during routine placement or if vascular anatomy distorted by congenital anomaly
- Normal catheter: Parallel echogenic lines surrounded by anechoic flowing blood in vessel lumen
- Thrombus (UVC or UAC): Lobular echogenic material surrounding catheter, often at tip; may occlude vessel lumen, propagate away from tip, or embolize
- Hepatic extravasation/hematoma/abscess (UVC): May mimic mass
  - Multilobulated/multiseptated collection of variable echogenicity & heterogeneity
    - Usually hyperechoic rim, hypoechoic center
  - UVC may be in cystic mass
  - With removal of catheter, collection regresses & calcifies
- TPN ascites (UVC): Hypoechoic peritoneal fluid ± debris
- Aneurysm/pseudoaneurysm (UAC): Focal dilation or irregular outpouching of artery with turbulent flow on Doppler

PATHOLOGY

General Features

- Malposition
  - UVC tip proximal to ductus venosus or in portal vein
    - Instillation of hypertonic or vasoactive fluid into portal veins results in hepatic rather than systemic infusion
      - May cause portal vein thrombus, hepatic necrosis, TPN abscess, portobiliary fistula
    - Hepatic laceration, hematoma
  - UVC malposition in heart/lung
    - Arrhythmia, thrombotic endocarditis, atrial wall perforation with pericardial effusion ± cardiac tamponade, hemothorax
- Thrombosis
  - May become infected, leading to septic emboli &/or mycotic aneurysm/pseudoaneurysm
  - UVC-associated thrombosis (13-30% of UVCs)
    - Portal & hepatic veins at highest risk
  - UAC-associated thrombosis
    - SMA: Gut ischemia, necrotizing enterocolitis
    - Renal arteries: Hypertension, kidney injury
    - Aortic thrombosis more likely with low line
      - Limb ischemia, limb growth impairment
- Infection
  - UVC placement highest risk factor for neonatal sepsis

CLINICAL ISSUES

Treatment

- In general, reposition or remove malpositioned catheter
- Hepatic collections may require drainage
- Ascites may require paracentesis
- Infection necessitates antibiotic therapy
- Incidental UVC or UAC thrombus may be observed (frequent spontaneous resolution)
- Symptomatic or propagating UVC or UAC thrombus: Heparinization ± thrombolytics

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Esophageal Intubation

KEY FACTS

TERMINOLOGY

- Inadvertent placement of endotracheal tube (ETT) in esophagus

IMAGING

- Radiography
  - Esophagus normally lies posterior & left of trachea
  - Frontal view
    - Malpositioned ETT overlies esophageal air column, typically left of trachea
      - May directly overlie enteric tube
      - ± deviation of trachea to right
    - Malpositioned ETT may extend caudal to carina
    - Gaseous distention of stomach &/or esophagus
  - Lateral view
    - ETT overlies esophageal air column posterior to trachea
  - 25° right posterior oblique view with head turned right
    - Presents esophagus & trachea relationship en face
    - Facilitates visualization of esophagus to left of trachea
- Ultrasound
  - Point-of-care confirmation of ETT placement
  - Advantages: No radiation exposure, rapid detection of position in real-time at bedside, less handling of patient, potential for early surfactant delivery
  - Disadvantages: User dependent, lack of widespread availability
  - Technique & findings
    - High-frequency linear transducer transversely oriented on anterior neck just above suprasternal notch
    - Distortion of glottis & trachea not observed with esophageal intubation
    - Lateral & posteriorly positioned esophagus stented open by tube with hyperechoic curvilinear anterior wall & posterior acoustic shadowing
  - Esophageal intubation sensitivity 93%, specificity 97% in recent metaanalysis

TOP DIFFERENTIAL DIAGNOSES

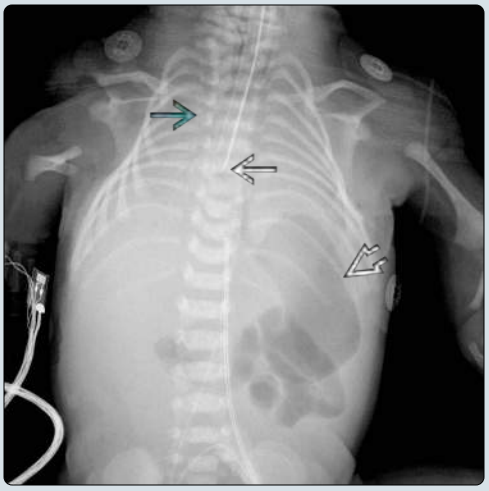
- Tracheal intubation
  - ETT projects over tracheal air column
  - No significant esophageal distention
- Hypopharyngeal intubation
  - Ineffective ventilation
  - Gastric & esophageal distention
- Right main bronchus intubation
  - Left lung atelectasis
  - Right upper lobe atelectasis if ETT tip distal to right upper lobe bronchus
- Tracheal perforation
  - Pneumomediastinum or pneumothorax
  - Aberrant lateral deviation of tube
- Tracheal intubation of tracheoesophageal fistula
  - Tip should be positioned distal to fistula
  - Tip may enter fistula, cause esophageal distention

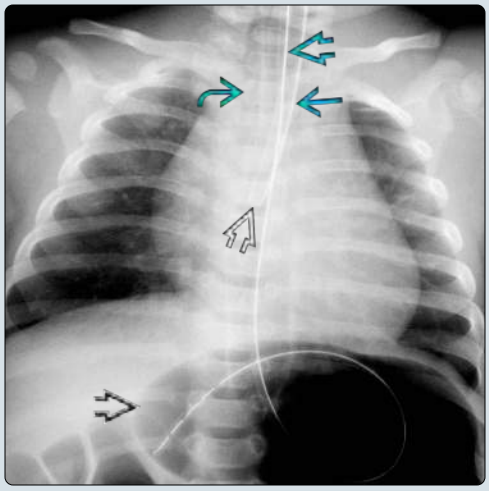
CLINICAL ISSUES

- American College of Emergency Physicians policy for verification of endotracheal intubation
  - Confirm placement with: Direct visualization of tube passing through vocal cords, auscultation of chest & epigastrium, bilateral symmetric chest rise, fogging of ETT with respiration, pulse oximetry, chest radiography, capnography (most accurate)
- Complications of esophageal intubation
  - Hypoxemia, regurgitation, aspiration, cardiac dysrhythmia, death
- Esophageal intubation frequency in pediatrics
  - 28% of all intubation adverse events
  - Immediately recognized in 4.1% of intubations
  - Delayed recognition in 0.19% of intubations
    - Radiology may be 1st to recognize malposition

DIAGNOSTIC CHECKLIST

- Esophageal intubation is emergent finding, potentially fatal
- Immediate communication to clinical team required





(Left) *AP chest radiograph in a newborn with respiratory distress shows the endotracheal tube (ETT) tip extending beyond the level of the carina. The lungs are markedly hypoinflated, & the stomach & small bowel are distended with gas, consistent with esophageal intubation.* **(Right)** *AP chest radiograph in a 4 month old shows an esophageal intubation. An ETT & an enteric tube project over an esophageal air column to the left of the trachea. The ETT tip lies caudal to the carina. The stomach is distended.*

KEY FACTS

TERMINOLOGY

- Extracorporeal membrane oxygenation (ECMO): Modified pulmonary or cardiopulmonary bypass circuit
- Deoxygenated blood removed from venous system → oxygenated → returned to circulation via venous system [venovenous ECMO (VV-ECMO)] or arterial system [venoarterial ECMO (VA-ECMO)]

IMAGING

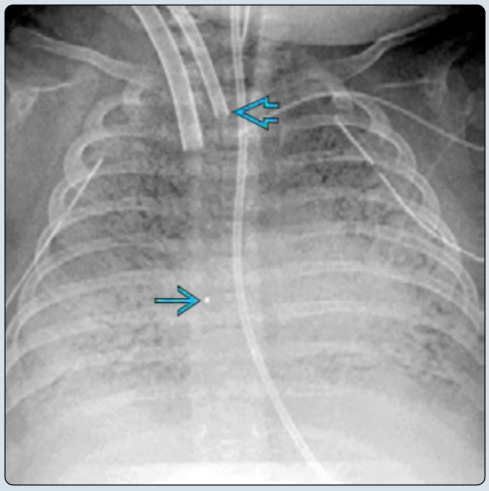
- **Radiographs** to monitor catheter positions, complications
  - Most common VV-ECMO circuit
    - Internal jugular vein (dual- or triple-lumen catheter)
    - 1 or 2 lumina for venous extraction
    - 1 lumen for venous return of oxygenated blood
    - Tip in right atrium (RA) at posterior 8th or 9th rib
  - Most common VA-ECMO circuit
    - Internal jugular vein - common carotid artery (CCA)
      - Venous tip in RA at posterior 8th or 9th rib
      - Arterial tip in CCA origin at 2nd or 3rd posterior rib
  - Other circuits include
    - Femoro-femoral: VA-ECMO or VV-ECMO
    - RA-aorta/pulmonary artery: VA-ECMO
  - Right-sided vessels favored for cannulation due to more direct central route
    - Requires sacrifice/ligation of vessels
  - Expected chest & abdominal radiographic findings
    - Generalized dense pulmonary opacification
      - Does not correlate with severity of lung disease
      - Related to low "rest" ventilator settings & systemic inflammatory response syndrome from bypass
    - Paucity of bowel gas
- **Ultrasound** to evaluate for intracranial hemorrhage or ischemia, thoracic & abdominal fluid collections
  - Enlargement of subarachnoid CSF spaces
    - May resolve after ECMO
- **CT** used for suspicion of ECMO complication, underlying thoracic disease, &/or delay in clinical improvement
  - Can help identify & further characterize intracranial lesions beyond head US
  - High proportion of clinically significant findings requiring intervention in appropriate setting: Up to 84%
- Imaging protocols vary by institution
  - Typically include head US & chest radiographs before & after ECMO initiated & at specified intervals

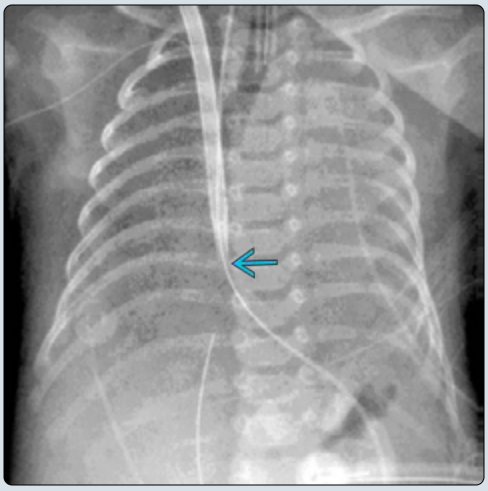
PATHOLOGY

- Mechanical complications
  - General: Thrombus in circuit
    - Prevention requires adequate anticoagulation
  - Venous cannula too distal: Obstruction of peripheral venous return, poor venous extraction due to blocked sideholes
  - Venous cannula too proximal: Sideholes outside lumen or dislodged → ↑ risk air of emboli, hemorrhage
  - Arterial cannula too distal: Aortic flow obstruction, ↑ left ventricle afterload
  - Arterial cannula too proximal: Inadvertent dislodgement → ↑ risk of hemorrhage
- Clinical complications
  - Intracranial hemorrhage & ischemia most common
    - 85% of intracranial hemorrhages in 1st 72 hours
      - Common intracranial hemorrhage locations: Intraparenchymal (64%), posterior fossa (27%)
    - Risk of ischemia ↑ with duration of ECMO
  - Other complications: Lung consolidation, infarction, necrosis ± cavitation, barotrauma, hemothorax, pulmonary emboli; intra/retroperitoneal hemorrhage, adrenal hemorrhage, solid organ infarct

CLINICAL ISSUES

- Indications for ECMO: Causes of severe reversible cardiac &/or respiratory failure, including meconium aspiration, congenital diaphragmatic hernia, pulmonary hypertension, sepsis, cardiomyopathy, preoperative stabilization of congenital heart disease
- Survival to discharge: 33-41%





(Left) *AP chest radiograph in a 4 month old with sepsis shows a venous ECMO cannula radiopaque marker projecting over the right atrium at T8. The arterial catheter projects over the common carotid artery origin at T2-T3. The pulmonary opacities & interstitial emphysema relate to underlying pulmonary disease.* **(Right)** *AP chest radiograph of a 2 month old on venovenous ECMO shows a dual-lumen catheter with its tip projecting over the right atrium at the posterior 8th right rib. There is expected lung whiteout.*

PICCs

KEY FACTS

TERMINOLOGY

- Vascular line inserted percutaneously into peripheral vein with tip residing in central vein

IMAGING

- Optimal upper extremity tip position: Lower 1/3 of superior vena cava (SVC), near SVC-right atrial (RA) junction
  - SVC-RA junction: ~ 2 vertebral bodies + disc spaces ("vertebral units") below carina
  - Approximate SVC boundaries: Superior at right tracheobronchial angle, inferior just below right superior cardiac border
  - SVC always to right of trachea
    - Descent left of trachea suggests venous or arterial malposition or possibly left-sided SVC
      - Confirmation with cross-sectional imaging or angiography required prior to peripherally inserted central catheter (PICC) use
  - Tip most cranial with arm in straight 90° abduction
  - Tip appears caudal with supine position & poor inspiration
- Optimal lower extremity PICC position: IVC-RA junction
  - IVC-RA junction: ~ T8 or T9, close to level of diaphragm
  - Consider ascending lumber vein location with kink or bend, zig zag paraspinal course, or failure of left approach PICC to cross midline at L5

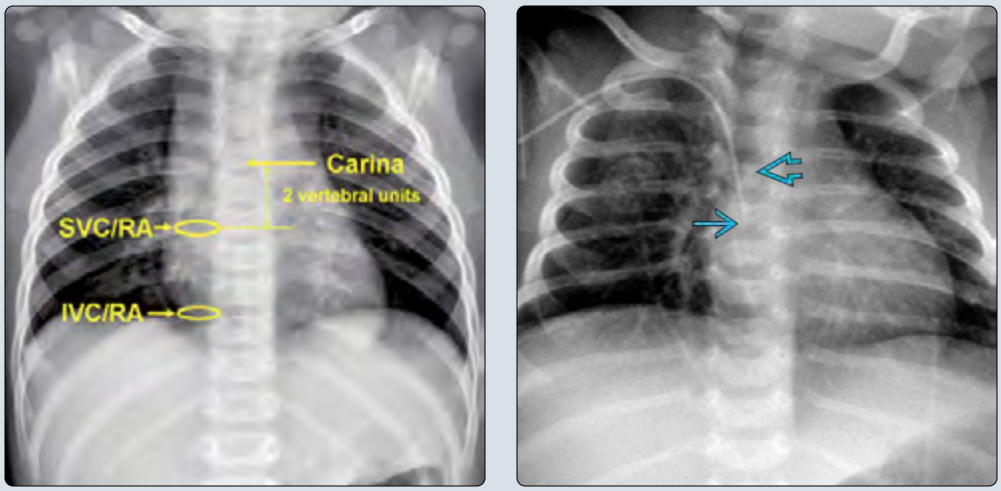
PATHOLOGY

- Risk of complications 8x greater if tip noncentral
- Complications include: Thrombosis, thrombophlebitis, infection, sepsis, pulmonary embolus, mechanical malfunction, extravasation, arrhythmia, myocardial perforation, arterial placement with distal tissue necrosis

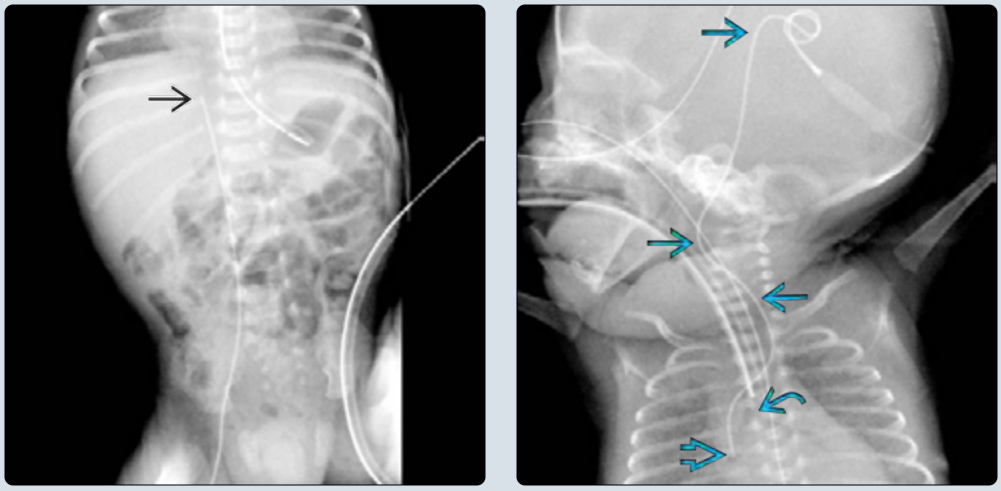
CLINICAL ISSUES

- Indicated for long-term central venous access (1-8 weeks): Antibiotics, TPN, repeated transfusions, short course of chemotherapy, frequent venous sampling, central venous pressure measurements, tenuous venous access

(Left) *AP chest radiograph depicts commonly used radiographic definitions of the superior vena cava (SVC)-right atrium (RA) & inferior vena cava (IVC)-RA junctions.* **(Right)** *Supine chest radiograph in a 3 month old with meningitis shows a right upper extremity peripherally inserted central catheter (PICC) tip ⇨ projecting ~ 2 vertebral bodies below the carina ⇨ in the region of the SVC-RA junction.*



(Left) *AP abdominal radiograph shows a right lower extremity PICC tip ⇨ in an appropriate position in the IVC above the level of the renal veins & below the right atrium.* **(Right)** *Supine chest radiograph of a premature infant demonstrates a scalp PICC ⇨ with its tip ⇨ projecting ~ 2 vertebral units below the carina ⇨ in the expected region of the SVC-RA junction.*



TERMINOLOGY

Definitions

- Peripherally inserted central catheter (PICC): Vascular line inserted percutaneously into peripheral vein with tip residing in central vein

IMAGING

General Features

- Location
  - Most common insertion sites
    - Upper extremity veins: Basilic, cephalic, brachial
    - Lower extremity veins: Saphenous; occasionally deep veins used, especially in newborns
    - Scalp veins used if extremity access difficult
  - Tip considered central if in
    - Right atrium (RA): Not recommended due to risks of perforation & arrhythmia (specific to PICCs)
    - Superior vena cava (SVC); inferior vena cava (IVC) above renal veins
    - Brachiocephalic veins (controversial)
  - Central tip position allows for effective infusion with appropriate hemodilution given greater blood flow

Radiographic Findings

- Optimal upper extremity PICC position: Lower 1/3 of SVC, near SVC-RA junction
  - Definition of SVC on AP chest radiograph controversial as use of landmarks varies
    - Best estimation of SVC-RA junction: 2 vertebral units (bodies + disc spaces) below level of carina
    - Superior SVC boundary: Right tracheobronchial angle
    - Inferior SVC boundary: Below right superior cardiac border
  - SVC remains right of trachea, even on rotated image
- Optimal lower extremity PICC position: IVC close to IVC-RA junction, above renal veins
  - Landmarks on supine radiograph
    - IVC-RA junction: ~ T8 or T9, close to level of diaphragm
    - Renal vein entrance to IVC: ~ L1
    - Common iliac vein confluence at IVC: ~ L5
- Malposition examples
  - Arterial: PICC descends left of trachea/midline (upper approach) or ascends left of midline (lower approach)
  - Internal jugular vein: Tip directed cephalad into neck
  - Left SVC: PICC descends left of trachea, typically from left upper extremity approach
    - Acceptable except in rare circumstances of left SVC draining into left atrium
    - Must be confirmed with prior or subsequent cross-sectional imaging or angiography before use
  - Ascending lumbar veins
    - Arise from common iliac veins at L5-S1
    - Left approach PICC fails to cross midline at L5
    - Bend or kink at L4-L5 with paraspinal zig zag course
    - Marked posterior deviation at L4-S1 on lateral view

Imaging Recommendations

- Protocol advice
  - If position unclear, inject contrast during fluoroscopy
    - If arterial position being questioned, retract tip distal to neck vessel origins prior to injection

PATHOLOGY

General Features

- Complications: 1-19 per 1,000 catheter days
  - Infection, sepsis (↑ with PICC duration)
  - Arterial placement can lead to distal tissue necrosis
  - Pulmonary embolus (air, thrombus, catheter fragment)
  - Mechanical malfunction (catheter damage, extravasation or leakage, unplanned catheter removal)
  - Thrombosis & thrombophlebitis
  - Arrhythmia or myocardial perforation if tip in RA
  - Spinal cord injury if tip in lumbar vein
- Risk of complications 8x greater if tip noncentral
  - ↓ vein diameter → ↓ blood flow/hemodilution → ↑ turbulence + prolonged intimal contact of infusates → ↑ risk of endothelial injury

CLINICAL ISSUES

Indications

- Need for intermediate or long-term central venous access (1-8 weeks): Antibiotics, TPN, repeated transfusions, short course of chemotherapy, frequent venous sampling, central venous pressure measurement
- Tenuous venous access otherwise

Not Indicated For

- Injection of IV contrast (unless specifically rated to withstand high pressures/flow rates)
- Longer chemotherapy courses (port catheter preferred)
- Rapid fluid resuscitation

DIAGNOSTIC CHECKLIST

Consider

- Each institution should develop standard PICC placement/verification protocols, nomenclature, & communication criteria
- PICC position varies with respiratory motion, arm position

Reporting Tips

- Avoid phrase "cavoatrial junction" (as this could represent either SVC-RA or IVC-RA junction)

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Viral Chest Infection

KEY FACTS

TERMINOLOGY

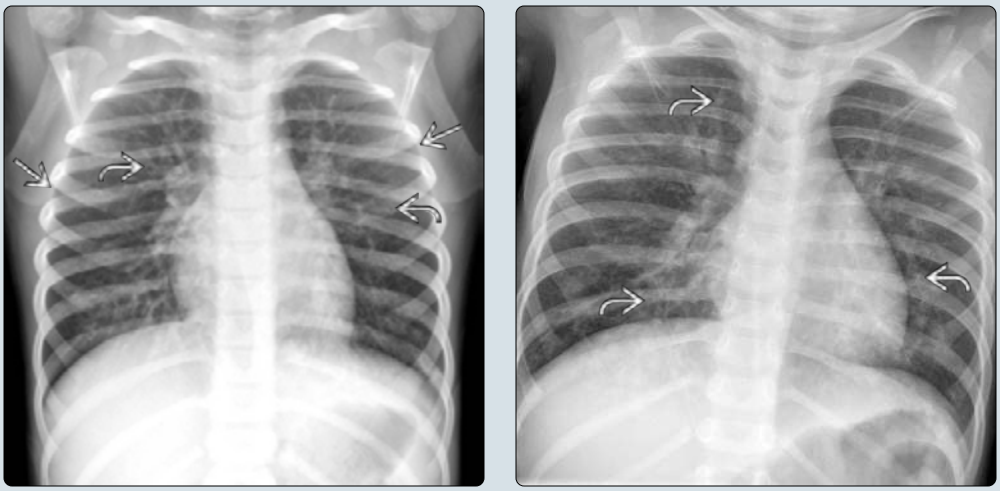
- Viral infection may involve airways &/or lung parenchyma (alveoli, interstitium)
- Bronchiolitis: Acute inflammation & necrosis of epithelial cells lining small airways with ↑ mucus production
  - Classically < 2 years of age
- Other terms: Viral pneumonia, lower respiratory tract infection, peribronchial pneumonia

IMAGING

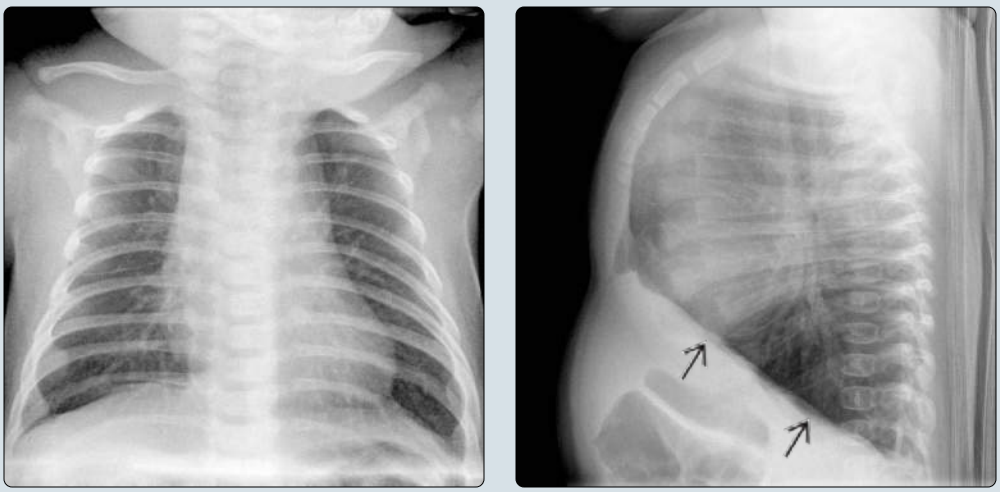
- Primary goal of chest radiography: Differentiate viral airway infection from bacterial pneumonia (which requires antibiotics)
  - 92% negative predictive value for bacterial pneumonia
- Best imaging clues for viral airway infection
  - ↑ peribronchial markings
    - Radiating linear rope-like or "dirty" perihilar opacities
    - "Doughnuts" of circumferentially thickened bronchial walls (viewed in cross section)
  - Hyperinflation: Depression of hemidiaphragms with downward sloping on lateral view; ↑ AP chest diameter on lateral view; ± convex bulging of lungs between ribs
  - Subsegmental atelectasis, possibly multifocal
  - Lack of focal/lobar consolidation or pleural effusion
- Best imaging clues for viral parenchymal involvement
  - Interstitial, nodular, or patchy ground-glass opacities

CLINICAL ISSUES

- Pathogens detected in hospitalized pediatric pneumonia patients: Viral 73%, bacterial 15%
- Most common viral etiologies differ by age
  - RSV < 2 years, rhinovirus > 2 years
- Treatment
  - Antibiotics for concomitant bacterial infection
  - Nebulized hypertonic saline in hospitalized infants may shorten length of stay
  - Antiviral therapy for influenza cases



(Left) *AP radiograph in a wheezing child shows typical findings of viral airways disease. There are mildly increased perihilar markings with an increased number of "doughnuts" (thickened bronchial walls viewed in cross section) ➡. The lungs show convex bulging between ribs ➡, typical of hyperinflation.* **(Right)** *AP radiograph shows viral airways disease with increased perihilar markings of bronchial wall edema ➡. There is no focal lung consolidation or pleural effusion.*



(Left) *AP radiograph in a wheezing child shows hyperinflated lungs with increased rope-like perihilar markings, consistent with viral airways disease. There is no focal lung consolidation.* **(Right)** *Lateral radiograph in the same patient shows marked hyperinflation with flattening of the hemidiaphragms ➡ (much more evident than on the corresponding frontal view) & widening of the AP diameter of the chest. Also note the increased markings radiating from the hila.*

TERMINOLOGY

Definitions

- Terminology varies, can be confusing
- Bronchiolitis (as defined by American Academy of Pediatrics): Viral lower respiratory tract infection in infants with "acute inflammation, edema, & necrosis of epithelial cells lining small airways," & ↑ mucus production
- Lower respiratory tract infection may describe findings identical to bronchiolitis in patients ≥ 2 years old but may also refer to any infection of lower airways & parenchyma
- Viral pneumonia may refer to viral infection of lung parenchyma ± airways infection
- Peribronchial pneumonia sometimes used to differentiate airways infection from parenchymal involvement
- Lower airways disease includes viral airways infection as well as asthma

IMAGING

General Features

- Best diagnostic clue
  - ↑ Peribronchial markings with symmetric hyperinflation

Radiographic Findings

- Primary goal: Differentiate viral airways infection from bacterial pneumonia
- Small airways viral infection
  - Lack of focal dense geographic, round, or "fluffy" lung consolidation (hallmark of bacterial infection)
  - ↑ peribronchial markings/↑ number of visible bronchi
    - Symmetric, coarse linear markings radiating from hila
    - ↑ thickness of bronchial walls appearing as doughnuts in cross section
    - Central lungs may appear dirty or busy
    - Hila may appear prominent on lateral view
    - Somewhat subjective finding
  - Hyperinflation
    - Hyperlucency
    - Depression of diaphragm > 10 posterior ribs or > 6 anterior ribs
    - Flattening/downward sloping hemidiaphragms (best seen on lateral view)
    - ↑ AP chest diameter (in infants, chest wider than tall on lateral view)
    - ± convex bulging of lungs between ribs
    - May not be seen > 2 years old: Bronchi larger, less prone to narrowing or obstruction with inflammatory change
  - Subsegmental atelectasis
    - Wedge-shaped or triangular opacity, often narrow
      - Most common in mid or lower lung
      - Often multifocal
    - Commonly misinterpreted as suspicious for bacterial pneumonia
- Parenchymal involvement
  - Interstitial nodular or patchy opacities
  - Lobar consolidation rarely occurs
  - Associated findings of airways infection
- Mild hilar lymphadenopathy
  - Not alarming in this setting (unlike adults)
- Gas-distended bowel loops in upper abdomen, typically due to air swallowing

CT Findings

- Not used primarily to make diagnosis of viral disease but may be obtained in patient with unclear clinical/imaging presentation
- Small airways viral infection
  - Prominent, ill-defined hila & peribronchial markings radiating into lung
  - Bronchial & bronchiolar wall thickening
  - Mosaic attenuation: Patchy heterogeneity of attenuation due to hypoventilation & air-trapping from obstruction/narrowing
  - Ground-glass opacities
  - Tree-in-bud nodules
- Parenchymal involvement
  - Interlobular septal thickening
  - Patchy, ill-defined consolidation
  - Nodules, micronodules
  - ± findings of small airways infection
- Mild hilar lymphadenopathy

Imaging Recommendations

- Best imaging tool
  - Chest radiographs (frontal & lateral)
    - Performance in identifying/excluding bacterial pneumonia
      - Positive predictive value: 30%
      - Negative predictive value (NPV): 92%
    - Goal: Antibiotic therapy for all children with bacterial pneumonia while minimizing unnecessary antibiotic administration (for isolated viral infection)
      - High NPV of chest radiograph helpful
    - Difficult to differentiate viral from bacterial pneumonia (i.e., lung parenchymal infection) on imaging
      - One study showed lobar infiltrate in 15% of exclusively viral pneumonia & exclusively interstitial infiltrate in 28% with bacterial pneumonia

DIFFERENTIAL DIAGNOSIS

Bacterial Pneumonia

- Focal/lobar lung consolidation > interstitial infiltrate
  - Confluent geographic, round, or fluffy opacities
- Lack of ↑ peribronchial markings, hyperinflation
- Pleural effusions more common with bacterial infection

Asthma

- ↑ peribronchial markings & hyperinflation
  - Inflammation of small airways common to asthma & viral disease
- Primary asthma diagnosis difficult to establish in young children

Bronchial Foreign Body

- May present with wheezing very similar to viral disease
- Asymmetric hyperinflation characteristic
  - Affected lung volume static throughout respiratory cycle
- Foreign body often radiographically occult

Left-to-Right Cardiovascular Shunts

- In infants, left-to-right shunts may have similar appearance
  - ↑ pulmonary arterial flow may mimic ↑ peribronchial markings
  - ± hyperinflation
- Shunts have associated cardiomegaly

Miliary Tuberculosis

- Diffuse small nodules, thickened interlobular septa
- Lymphadenopathy, may be low in attenuation
- History of sick contact with TB
- Very uncommon in young children

PATHOLOGY

General Features

- Etiology
  - Acute inflammation & necrosis of epithelial cells lining small airways with ↑ mucus production
  - ↓ caliber of small, relatively compliant airway lumina significantly ↑ resistance to airflow in infants
  - Occlusion of airways results in hyperinflation & foci of subsegmental atelectasis
  - Parenchymal findings vary: Interstitial pneumonitis with lymphocytic infiltration, infection of alveolar epithelium, diffuse alveolar damage, desquamation of pneumocytes, hyaline-membrane formation, alveolar hemorrhage
  - Typical pathogens
    - Bronchiolitis: RSV > rhinovirus, adenovirus, influenza > coronavirus, human metapneumovirus (hMPV), parainfluenza
      - Multiple viruses in up to 25%
    - Hospitalization-inducing community acquired pneumonia: RSV, rhinovirus > hMPV, adenovirus, *Mycoplasma pneumoniae*, parainfluenza, influenza, coronavirus, *Streptococcus pneumoniae*, *Staphylococcus aureus*, *Streptococcus pyogenes*

CLINICAL ISSUES

Presentation

- Signs & symptoms
  - Bronchiolitis: Rhinorrhea, cough, tachypnea, wheezing, rales, ↑ respiratory effort (grunting, nasal flaring, intercostal/subcostal retractions)
  - Community-acquired pneumonia: Cough, fever, anorexia, dyspnea, wheezing (up to 62%)
- Difficult to differentiate bacterial from viral parenchymal infection based on physical exam or laboratory tests
  - Procalcitonin level may have diagnostic role: NPV of 95% for bacterial infection if < 2 ng/ml

Demographics

- Age
  - Typical, striking radiographic findings of viral disease more often in young children (< 5 years of age)
- Epidemiology
  - Bronchiolitis: Most common cause of hospitalization in 1st year of life
  - Viruses cause majority of chest infections in preschool children (4 months-5 years of age)
    - < 2 years old: RSV most common
      - Typical in late autumn, winter
    - > 2 years old: Rhinovirus most common
      - Typical in autumn, spring
  - Pneumonia-related hospitalization from multicenter prospective study
    - Greatest in children < 5 years old
    - Viral in 73%, bacterial in 15%
  - Predisposing conditions for more severe viral chest infections: Age < 6 months, prematurity, congenital heart disease, immunosuppression/immunodeficiency, asthma

Natural History & Prognosis

- Resolution of symptoms over time, typically days to weeks

Treatment

- Antibiotics for concomitant bacterial infection only (not isolated viral infection)
- Bronchiolitis
  - Nebulized hypertonic saline in infants may shorten hospitalization by increasing mucociliary clearance
  - Oxygen supplementation if saturations < 90%
  - Palivizumab (RSV prophylaxis, not vaccine): Given in 1st year of life to infants born before 29 weeks gestation or infants with hemodynamically significant heart disease or chronic lung disease of prematurity
  - Routine use of albuterol & nebulized epinephrine not recommended
  - Corticosteroid use not supported
- Clinical pneumonia with viral pathogen
  - Hospitalization if hypoxemia or respiratory distress
  - Antiviral therapy for influenza cases

DIAGNOSTIC CHECKLIST

Consider

- Consider aspirated foreign body if lung volumes asymmetric

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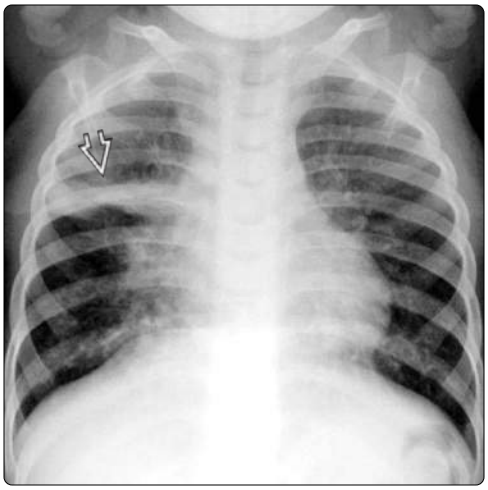
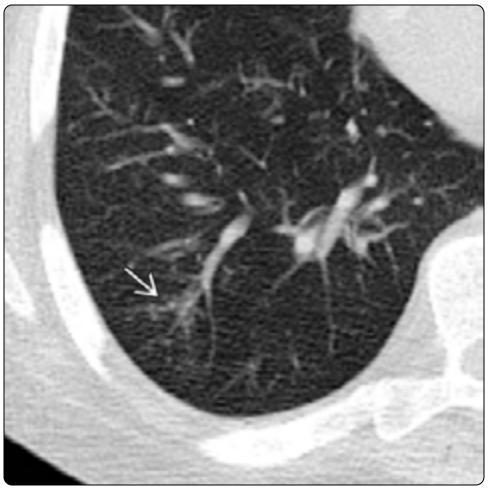
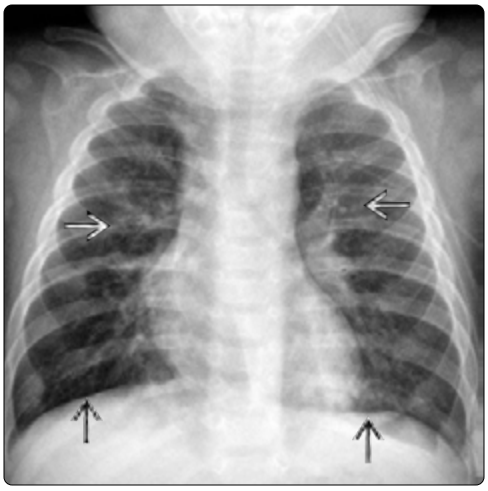
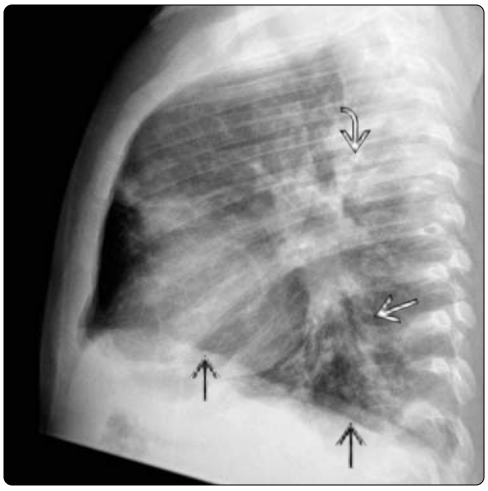


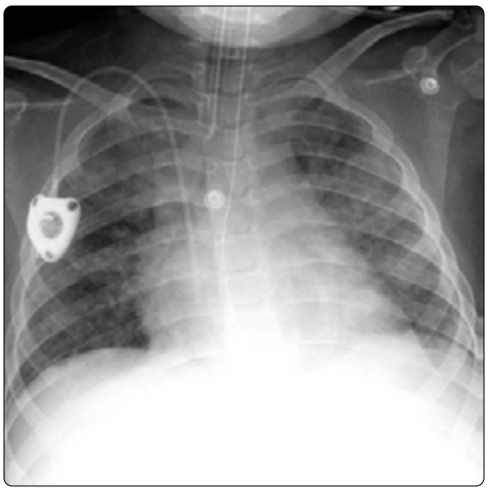


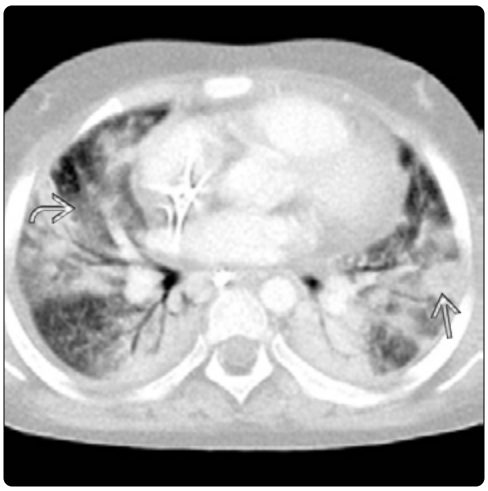
(Left) *AP radiograph in a child with viral illness shows symmetric hyperinflation, increased peribronchial markings, & a linear band of atelectasis in the right upper lobe.* **(Right)** *Axial CECT from a 25-year-old man with known influenza A infection shows tree-in-bud opacities in the right lower lobe, consistent with impaction of inflamed airways (bronchioles) by mucus or cells.*





(Left) *Frontal chest radiograph of a 5-month-old patient with cough & difficulty breathing shows bilateral "dirty" perihilar peribronchial opacities & lung hyperinflation with diaphragm depression, consistent with bronchiolitis.* **(Right)** *Lateral radiograph from the same patient shows linear opacities extending from the hila, thickened bronchial walls, & downward sloping of the flattened hemidiaphragms. Note the widened AP dimension of the chest.*





(Left) *AP chest radiograph of a 7-year-old girl with influenza A infection shows patchy bilateral pulmonary opacities superimposed on bronchial wall thickening, consistent with viral parenchymal & airways disease.* **(Right)** *Axial CECT from the same patient shows bilateral patchy, consolidative & ground-glass opacities, which can be seen with influenza A chest infection.*

Round Pneumonia

KEY FACTS

TERMINOLOGY

- Bacterial lung infection with very round, well-defined appearance on chest radiography; simulates mass lesion
- Majority seen in patients < 8 years of age

IMAGING

- Well-circumscribed round opacity ± air bronchograms
- Most common posteriorly in lower lobe superior segments
- No mass effect on or invasion of adjacent tissues
  - No mediastinal or vascular distortion
  - No splaying or erosion of ribs
- Margins of round lung "mass" classically create acute angles with mediastinum or chest wall but can be obtuse

TOP DIFFERENTIAL DIAGNOSES

- Bronchogenic cyst
- Neuroblastoma
- Congenital pulmonary airway malformation
- Bronchopulmonary sequestration

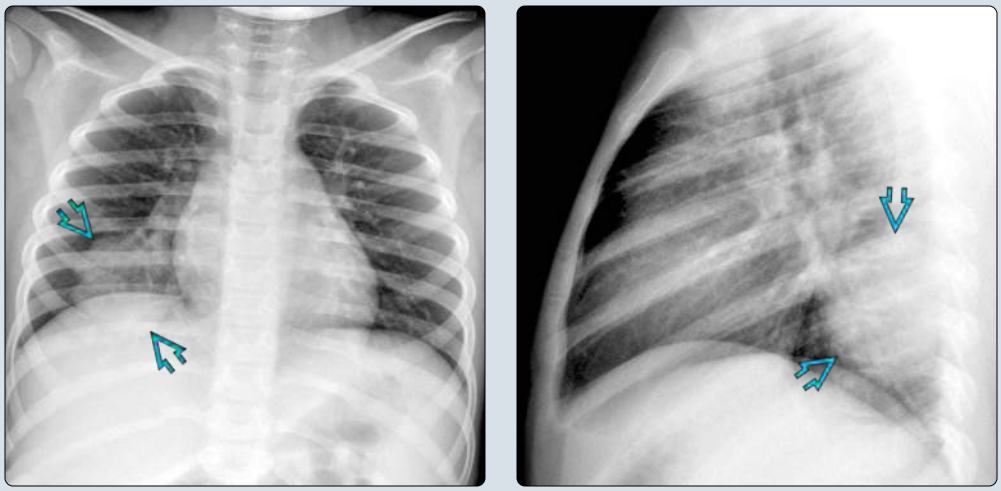
PATHOLOGY

- Collateral pathways of air circulation in lung not well developed until ~ 8 years of age
  - Channels of Lambert, pores of Kohn
- Spread of bacterial infection through lung therefore hindered in young children, predisposing to round appearance
- Typically occurs with *Streptococcus pneumoniae* infection

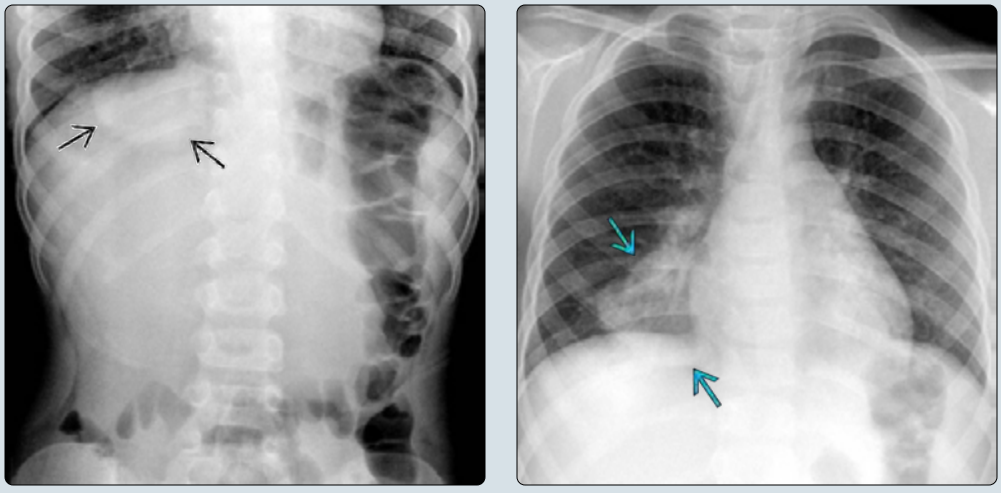
DIAGNOSTIC CHECKLIST

- Round lung opacity in child < 8 years of age → strongly consider round pneumonia
- With classic symptoms of pneumonia (cough, fever) in this age range, other masses do not need to be excluded
- If any doubt of diagnosis, consider
  - Targeted US or CT through lesion
  - Follow-up radiograph after completion of antibiotic course
    - Resolution of "mass" excludes other etiologies

(Left) *AP radiograph in a young child with cough & fever shows a round, mildly lobulated, well-circumscribed density in the medial aspect of the right lower lobe.* **(Right)** *Lateral radiograph in the same patient confirms that the round "mass" is located posteriorly in the right lower lobe. Note that the lesion makes acute angles with the posterior chest wall, consistent with a pulmonary origin. These findings are typical of a round pneumonia.*



(Left) *Abdominal radiograph of a 7-year-old boy with fever & vomiting shows a round opacity projecting over the right hemidiaphragm, suggesting a right lower lobe round pneumonia.* **(Right)** *Subsequent chest radiograph in the same patient clearly demonstrates a lobulated, well-circumscribed right lower lobe opacity, consistent with a round pneumonia. Note that the adjacent ribs are normal.*



TERMINOLOGY

Definitions

- Bacterial lung infection with very round, well-defined appearance on chest radiography; simulates mass lesion
- Majority seen in patients < 8 years of age
- Typically occurs with *Streptococcus pneumoniae* infection

IMAGING

General Features

- Best diagnostic clue
  - Round lung opacity with well-defined borders in febrile child < 8 years of age
- Location
  - Most common posteriorly in superior segments of lower lobes
  - No peripheral or central predisposition
  - Single focus most common (98%)
    - Multiple (2 or 3) foci can rarely occur
- Size
  - Ranges from 1-7 cm
  - Varies in size depending on timing of imaging relative to pneumonia development
  - May evolve to typical lobar pneumonia (no longer round)
  - With adequate treatment, usually resolves/clears (rather than progressing to lobar consolidation)
    - Resolution on follow-up imaging in 95%
- Morphology
  - Well-defined borders in 70%

Radiographic Findings

- Radiography
  - Round lung opacity typically marginated by clear lung
    - Paraspinal pneumonia may have acute or obtuse angle borders with posterior mediastinum/spine
  - Respects lobar anatomy without crossing fissures
  - Does not exert mass effect or invade adjacent tissues
    - No osseous changes in adjacent ribs (such as splaying or erosion) or spine
  - Air bronchograms help confirm airspace disease
  - Pleural effusion uncommon
  - Rarely progresses to lobar pneumonia

CT Findings

- NECT
  - Not advocated for cases of suspected round pneumonia
    - May be obtained if mass lesion of primary concern
    - Abdominal CT may show round pneumonia in lower lobe as cause of abdominal pain
  - Homogeneous round opacity ± air bronchograms
  - Respects lobar anatomy, does not cross fissures
  - No changes in adjacent bones
- CECT
  - Normal pulmonary vessels course through consolidated lung without mass effect
  - No enhancing rim or wall
  - No systemic arterial supply from descending aorta (as seen with bronchopulmonary sequestration)
  - Central cavity or fluid level favors alternative diagnosis

MR Findings

- Not utilized for primary suspicion of round pneumonia
- May be encountered if MR performed to further work-up round paraspinal mass suspected to be neuroblastoma
- Round pneumonia appears as moderately high (but not fluid bright) T2 signal intensity lesion within pulmonary parenchyma ± air bronchograms
- With contrast, enhancing nondisplaced vessels course through mildly enhancing, otherwise homogeneous "mass"

Ultrasonographic Findings

- Increasingly used for diagnosis of childhood pneumonia
  - Sensitivity 96%, specificity 93% overall
    - May be lower if round pneumonia completely surrounded by aerated lung
- Abnormality of normally echogenic pleural line: Irregular, coarse, interrupted, or absent
- "Hepatization" of subpleural parenchyma with branching mobile hyperechogenicities (air bronchograms)
- Interstitial involvement: > 3 B-lines (type of comet-tail artifact in which vertical echogenic line extends from pleural surface to edge of FOV)

Imaging Recommendations

- Pediatric Clinical Practice Guidelines for chest radiography in pneumonia
  - Not necessary to confirm suspected community-acquired pneumonia (CAP) in patients well enough to be treated as outpatients; exceptions include
    - Hypoxemia
    - Respiratory distress
    - Failed antibiotic therapy
  - Chest radiographs (frontal & lateral) should be obtained in all patients hospitalized for management of CAP
- If child < 8 years old has symptoms of pneumonia + round opacity on chest radiograph, additional imaging (such as CT) not necessary
  - Look carefully at adjacent bones
- Follow-up radiograph after antibiotic therapy may be helpful to document resolution of round opacity
  - Particularly helpful for paraspinal round pneumonia that may mimic neuroblastoma
- > 8 years old: Consider evaluation for other pathologies with CECT or MR

DIFFERENTIAL DIAGNOSIS

Bronchogenic Cyst

- Round, well-defined soft tissue mass
- Most common in perihilar regions
- ± mass effect on adjacent structures
  - Airway compression may cause air trapping/atelectasis
- Only contains air or air-fluid levels if infected
- CECT: Water attenuation mass without air bronchograms; ± rim enhancement if infected

Neuroblastoma

- Round or elongated posterior mediastinal/paraspinal mass
- Obtuse angle borders with mediastinum
  - May only widen paraspinal stripe without focal bulge
- Rib erosion or splaying common
- Ca^{2+} in majority by CT

Congenital Pulmonary Airway Malformation

- Round/lobulated parenchymal mass distorting adjacent lung & vessels
- Most commonly presents in utero or during newborn period
- Macrocystic type may temporarily appear solid after birth due to retained fluid
  - Quickly fills with air due to communication with airway

Bronchopulmonary Sequestration

- Lobulated or triangular parenchymal mass
- Most common in lower lobes
- Typically congenital, presenting in perinatal period; may present as recurrent pneumonia (always in same location) later in life
  - Round pneumonia almost never recurs in same location
- Characteristic systemic feeding artery arises from descending thoracic or abdominal aorta
  - May be seen prenatally or on postnatal CTA

Lung Abscess

- Round cavity containing air-fluid level
- Surrounded by irregular or poorly defined consolidation

Metastatic Disease

- Typically smaller multifocal nodules
- Rarely found incidentally in children

Pleural Pseudocyst

- Partially circumscribed focal fluid collection within fissure
- Very uncommon as isolated finding in children without preceding history of thoracic process

Chest Wall Ewing Sarcoma

- Partially circumscribed round mass with definite bony destruction (lysis, permeation)
- Associated pleural effusions frequent
- Typically in children > 8 years of age

PATHOLOGY

General Features

- Etiology
  - Collateral pathways of air circulation not well developed until ~ 8 years of age
    - Channels of Lambert & pores of Kohn
  - Lack of well-developed collateral circulation thought to hinder spread of bacterial infection, predisposing to round appearance on radiography

Gross Pathologic & Surgical Features

- Exudative opacification of pulmonary airspaces related to bacterial infection
  - *S. pneumoniae* most common pathogen

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Cough & fever
- Other signs/symptoms
  - Abdominal pain, malaise, anorexia

Demographics

- Age
  - 75% < 8 years old; 90% < 12 years old; mean age: 5 years
  - Cases rarely reported in older children & adults
    - Sometimes leads to biopsy
    - Antibiotics & short-term follow-up may be considered if round pneumonia diagnosis not clear

Natural History & Prognosis

- With appropriate antibiotic therapy, symptoms & opacity should resolve over days-weeks
  - Resolution of "mass" on follow-up radiograph after completion of antibiotic course confidently excludes other etiologies
- With antibiotic resistance, may progress to lobar pneumonia ± complications

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Round lung opacity in child < 8 years of age → strongly consider round pneumonia
  - With classic symptoms of pneumonia at presentation, other causes of radiographic mass do not need to be excluded with imaging
- > 8 years old, consider other pathologies & further work-up

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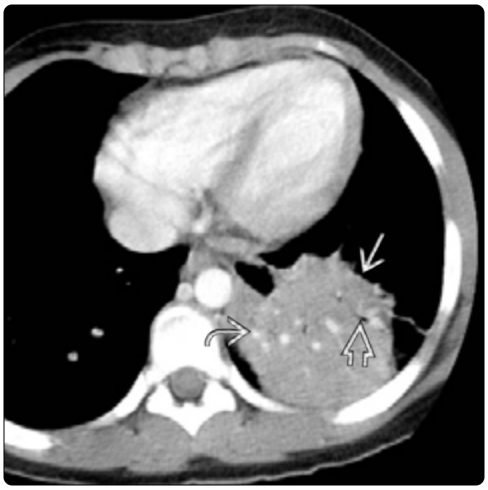
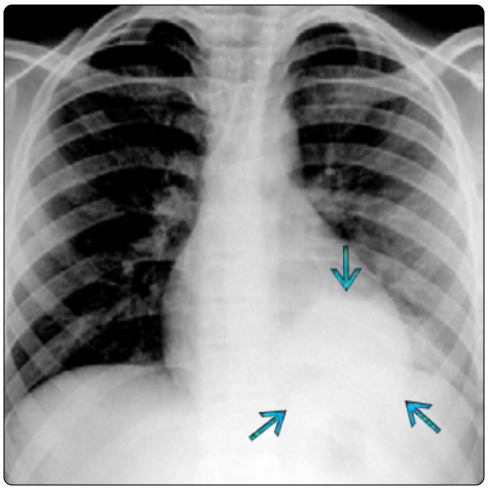
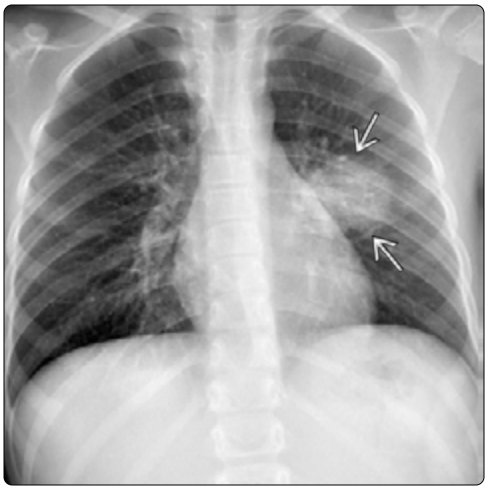
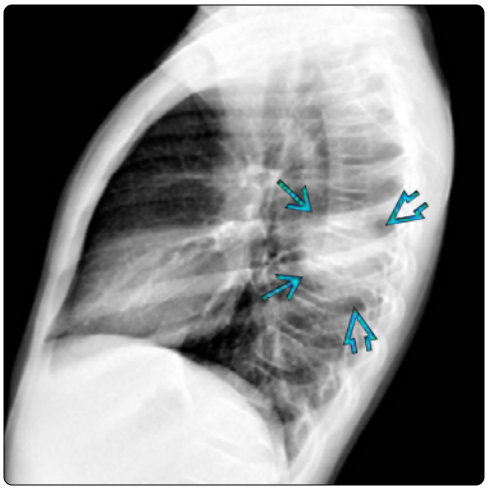


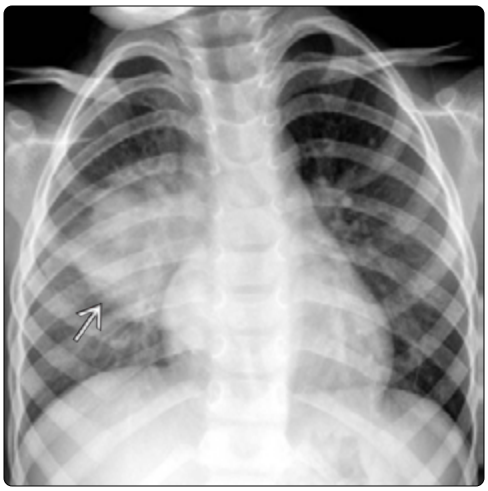


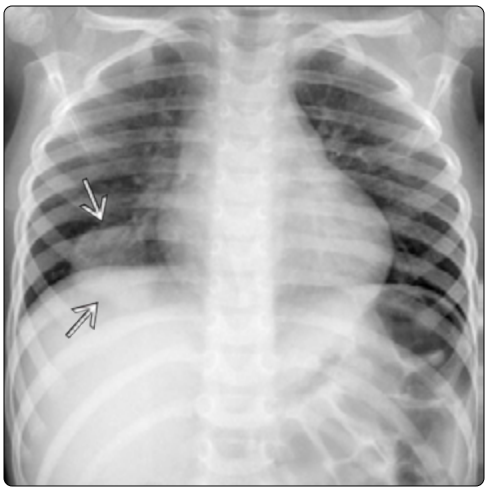
(Left) *Axial CECT of the abdomen in a 12-year-old boy with left upper quadrant abdominal pain shows a round focus of consolidation in the left lower lobe. The presence of internal air bronchograms, lack of distorted/splayed traversing vessels, & otherwise homogeneous appearance of the lesion confirms a round pneumonia.* **(Right)** *Subsequent PA chest radiograph performed on the same day in the same patient further demonstrates the round margins of the left lower lobe pneumonia.*





(Left) *PA chest radiograph of a 6-year-old girl with a cough demonstrates a round opacity projecting over the left hilum, suggesting a round pneumonia.* **(Right)** *Lateral chest radiograph from the same patient localizes the round opacity to the superior segment of the left lower lobe. The acute (rather than obtuse) angles at the interface of the opacity with the posterior chest wall are suggestive of its origin within the lung parenchyma.*





(Left) *PA chest radiograph from a 4-year-old boy with cough, fever, & chest pain shows a large, round, right lower lobe opacity. A follow-up radiograph performed approximately 2 weeks later (after antibiotic therapy) was normal. The findings & clinical course are consistent with a round pneumonia.* **(Right)** *Frontal chest radiograph of a 3-year-old girl with cough & fever demonstrates a well-defined, right lower lobe round opacity, most consistent with a round pneumonia.*

Parapneumonic Effusion and Empyema

KEY FACTS

TERMINOLOGY

- Pleural effusions classified as transudative or exudative
- Parapneumonic effusions are exudative secondary to adjacent lung infection & ↑ capillary permeability

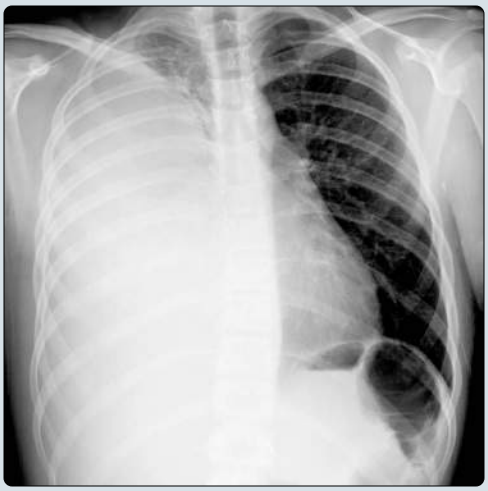
IMAGING

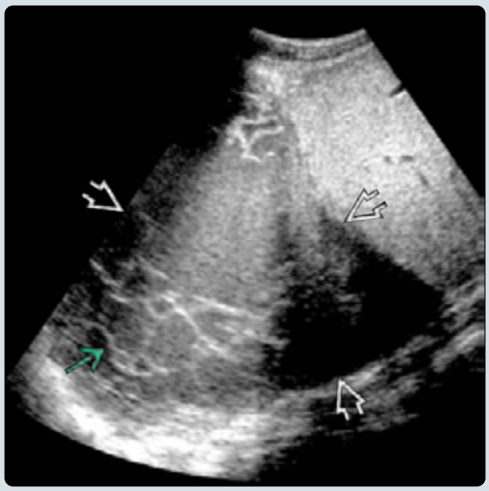
- Upright chest radiograph
  - Flattened & elevated hemidiaphragm, lateral shift of diaphragm apex, gastric bubble > 1.5 cm from diaphragm secondary to subpulmonic fluid
  - Blunted posterior costophrenic angle (~ 50 mL)
  - Blunted lateral costophrenic angle (~ 200 mL)
  - Hemidiaphragm inversion (> 2,000 mL)
- Supine chest radiograph may require up to 500 mL
  - Homogeneous vs. gradation of hazy/dense opacification of hemithorax ± pleural cap, mass effect
- CECT: Parietal pleural enhancement & thickening, thickening of extrapleural space, & chest wall edema seen with both transudative & exudative effusions in children
- US: Effusion appears anechoic, echoic, or mixed with floating/swirling/undulating echoes
  - Floating fibrin strands attached to pleural surface, septations, &/or pleural rind/thickening; immobile lung suggests entrapment by pleural rind
  - Loculation: Nonshifting fluid with position change
- Imaging recommendations
  - US if pleural disease suspected on chest radiograph
  - CECT if persistent/progressive illness despite treatment

CLINICAL ISSUES

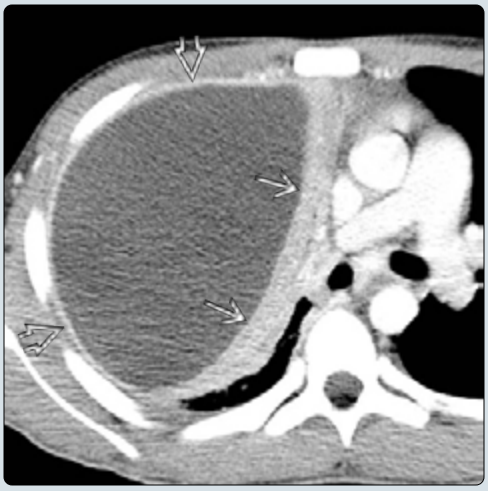
- Majority of children make complete clinical recovery
- American Pediatric Surgical Association recommendations
  - Antibiotics
  - Chest tube drainage if effusion of large volume, loculated, or with worsening/persistent symptoms
  - If empyema: Chest tube + tissue plasminogen activator; video-assisted thoracoscopic surgery if no clinical improvement & pleural disease persists on imaging

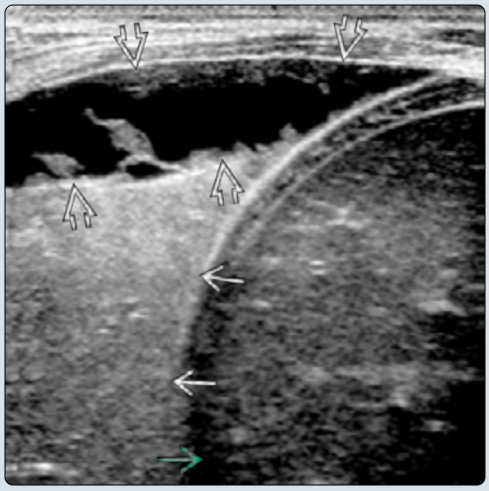
(Left) *PA radiograph of a child with a right pneumonia shows opacification of the majority of the right hemithorax with mediastinal shift to the left.* **(Right)** *Sagittal US of the right hemithorax in the same patient shows a large pleural effusion ➡ with internal debris & multiple echogenic septa ⇨, consistent with a fibrinopurulent parapneumonic effusion.*





(Left) *Axial CECT in the same patient shows the large pleural effusion compressing the right lung ➡. While the rind of enhancement ➡ suggests complexity, the septations demonstrated on the previous US are not visualized on this CT examination.* **(Right)** *Longitudinal US of the right lung in a patient with pneumonia shows a right pleural effusion ➡ with multiple areas of echogenic debris & septations. Note the consolidated, echogenic lung ➡. The liver ⇨ is shown inferiorly.*





TERMINOLOGY

Definitions

- Pleural effusions classified as transudative or exudative
  - Transudative: Due to hydrostatic & oncotic imbalances
  - Exudative: Due to ↑ capillary permeability
- Parapneumonic effusions are exudative
  - Stages of progression: Exudative (simple) → fibrinopurulent (complicated) → organized
  - Empyema is fibrinopurulent parapneumonic effusion

IMAGING

Radiographic Findings

- Pneumonia + adjacent pleural effusion
- Sequence of accumulation on upright image
  - Flattened & elevated hemidiaphragm, lateral shift of diaphragm apex, gastric bubble > 1.5 cm from diaphragm secondary to subpulmonic fluid
  - Blunted posterior costophrenic angle (~ 50 mL)
  - Blunted lateral costophrenic angle (~ 200 mL)
  - Hemidiaphragm inversion (> 2,000 mL)
- Supine images: Sensitivity 70%, may require up to 500 mL
  - Homogeneous vs. gradation of hazy/dense opacification of hemithorax ± pleural cap, mass effect
- Loculation suggested by lenticular shape or nonshifting fluid on decubitus views, but better characterized on US
- Fissural accumulation may present as thickening or pseudotumor in minor fissure

CT Findings

- Parietal pleural thickening & enhancement, thickened extrapleural space, & chest wall edema seen with both transudative & exudative effusions in children
- Inferior to US at demonstrating fibrin strands or septations
- Loculation inferred if air in collection separates into bubbles rather than single air-fluid level

Ultrasonographic Findings

- Effusion appears anechoic, echoic, or mixed with floating/swirling/undulating echoes
- Fibrin deposition: Floating/flapping strands attached to pleural surface, septations, pleural rind/thickening; lack of lung mobility suggests pleural rind entrapping lung
- Loculation: Nonshifting fluid with position change

Imaging Recommendations

- US if suspected pleural space disease on chest radiograph
  - Must scan dependent & nondependent pleural cavity
- CECT if persistent/progressive illness despite treatment (to evaluate for malpositioned chest tube, necrosis, abscess, purulent pericarditis)

DIFFERENTIAL DIAGNOSIS

Malignant Pleural Effusion

- Lymphoma, Ewing sarcoma, pleuropulmonary blastoma
- Look for nodularity/mass &/or rib destruction

Chylothorax

- Birth trauma, lymphangiectasia, lymphatic malformation

Lung Abscess

- Walled-off collection with pneumonia & progressive illness

PATHOLOGY

General Features

- Transudate vs. exudate classification based on fluid analysis
- Light's criteria for exudate (≥ 1 required): Pleural fluid to serum protein ratio > 0.5 or LDH ratio > 0.6; pleural fluid LDH > 2/3 upper limit of normal serum level
- If exudative, then empyema suggested by: pH < 7.2, lactate dehydrogenase (LDH) > 1,000 U, glucose < 40 mg/dL or < 25% blood glucose, positive Gram stain or positive culture, > 10,000 WBC/uL, loculations on imaging
- Common causes include *Streptococcus pneumoniae* & methicillin-resistant *Staphylococcus aureus*
- Test for tuberculosis if lymphocytic exudate
- Stages of parapneumonic effusion progression
  - Precollection: Pleuritis, inflammation
  - Exudative (simple): Free fluid with low white cell count
  - Fibrinopurulent (complicated): Deposition of fibrin & purulent material
  - Organized: Thick pleural peel, which may entrap lung

CLINICAL ISSUES

Presentation

- Fever, cough, chest pain, dyspnea, tachypnea, splinting to affected side, respiratory distress if large volume

Demographics

- Complicates 28-53% of pediatric pneumonias

Natural History & Prognosis

- Most pediatric patients make complete clinical recovery with almost normal radiograph in 3-6 months
- Pediatric mortality < 3% in 1 study (in contrast to adults, with mortality reported up to 20%)

Treatment

- American Pediatric Surgical Association recommendations
  - Antibiotics
  - Fluid evacuation if effusion of large volume, loculated, or with worsening/persistent symptoms
  - < 14F chest tube recommended; single thoracentesis can be used for older children with free fluid
  - If empyema: 12F chest tube + 3 doses of intrapleural tissue plasminogen activator → video-assisted thoracoscopic surgery if no clinical improvement & pleural disease persists on imaging

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Pneumonia With Cavitary Necrosis

KEY FACTS

TERMINOLOGY

- Complication of bacterial pneumonia where dominant focus of necrosis develops in consolidated lung, resulting in variable number(s) of thin-walled cysts
- Synonyms: Necrotizing pneumonia, pulmonary gangrene
  - Abscess has thick, well-defined wall & is more commonly seen in immunocompromised children

IMAGING

- Not identified radiographically until tissue breakdown leads to communication of cavity with aerated lung/airways
- CECT shows lack of normal lung architecture, ↓ lung enhancement, & thin-walled cysts in midst of consolidation
- No clear role for percutaneous drainage in cases of cavitary necrosis in immunocompetent children

TOP DIFFERENTIAL DIAGNOSES

- Congenital pulmonary airway malformation
- Lung abscess
- Bronchopulmonary sequestration
- Bronchogenic cyst

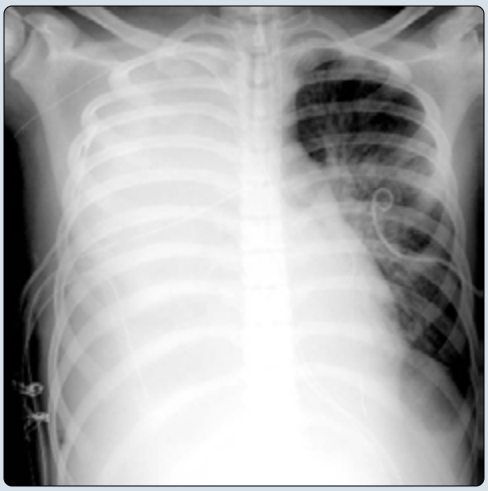
PATHOLOGY

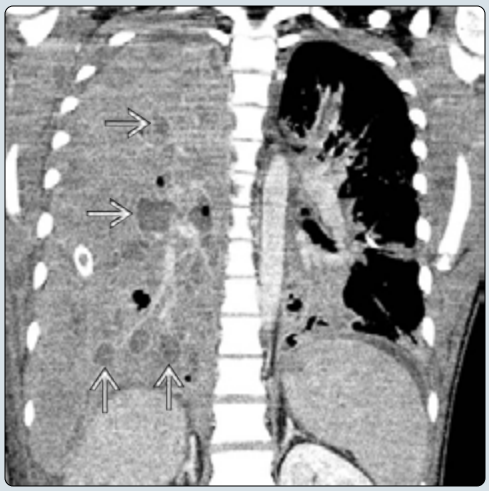
- Increasingly identified as pediatric pneumonia complication
- *Streptococcus pneumoniae* most common cause
  - Other organisms: MSSA, MRSA, other *Staphylococcus* & *Streptococcus* species, *Pseudomonas*, *Fusobacterium*
- May lead to bronchopleural fistula & development of pneumothorax

CLINICAL ISSUES

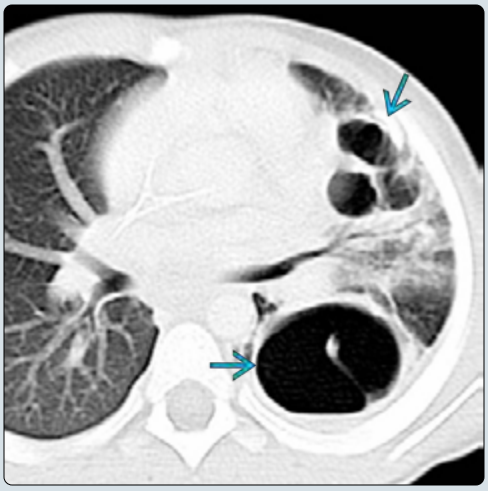
- Progressive symptoms (fever, respiratory distress, sepsis) in pediatric pneumonia patient despite appropriate medical management suggests complication (such as cavitary necrosis)
- Patients with cavitary necrosis tend to be intensely ill (ICU)
- Most do recover with antibiotics & nonsurgical management

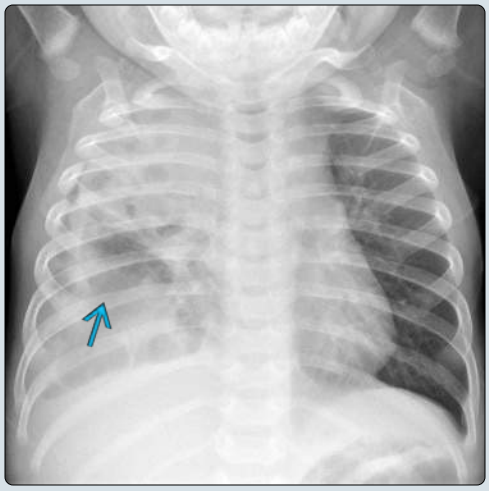
(Left) *AP chest radiograph in a child with rapidly progressive symptoms shows complete opacification of the right hemithorax & partial opacification of the medial left lower lobe (LLL). There are bilateral chest tubes in place.* **(Right)** *Coronal CECT in the same patient shows heterogeneous opacification of the entire right lung with multiple thin-walled, fluid-containing cystic foci, consistent with cavitary necrosis. Also note the LLL opacification. There are no significant pleural effusions.*





(Left) *Axial CECT shows multiple thin-walled, air-containing cysts with surrounding consolidation in the LLL. This appearance is very similar to a congenital pulmonary airway malformation. However, follow-up imaging after antibiotics showed the lesions to resolve over time, most consistent with cavitary necrosis.* **(Right)** *AP radiograph in a young child shows opacification of the right hemithorax with an air-filled cystic focus in the mid lung, suggesting cavitary necrosis.*





TERMINOLOGY

Synonyms

- Necrotizing pneumonia
- Pulmonary gangrene

Definitions

- Complication of bacterial pneumonia where dominant focus of necrosis develops in consolidated lung, resulting in variable number(s) of thin-walled cysts

IMAGING

General Features

- Best diagnostic clue
  - CECT of opacified lung shows loss of normal lung architecture with focally ↓ lung enhancement & development of thin-walled cysts containing air-fluid levels
- Location
  - More common in lower lobes
- Size
  - Variably sized cysts, typically 2-10 cm

Radiographic Findings

- Radiography
  - Cystic lucencies within lung opacified by pneumonia
  - Cavitation not detected radiographically until tissue breakdown leads to communication with aerated lung or bronchi
    - Leads to introduction of air into cavity
  - ± pneumothorax related to bronchopleural fistula
  - Follow-up after antibiotics
    - Progressive ↓ in lung consolidation
    - Cystic lucencies eventually resolve
    - Radiographs obtained > 40 days later: Often normal or showing only minimal linear scarring

CT Findings

- CECT
  - Within area of consolidated lung
    - Loss of normal lung architecture
      - Breakdown of normal bronchogram pattern
    - ↓ enhancement
      - Noncompromised consolidated or atelectatic lung enhances
      - Nonenhancement suggests ischemia or impending necrosis
    - Multiple cystic foci filled with air &/or fluid
      - Not air-filled until communicating with aerated lung/airways
      - Walls of cyst: Thin, often nonenhancing

Ultrasonographic Findings

- Grayscale ultrasound
  - Ill-defined heterogeneously hypoechoic foci in consolidated lung
    - Seen in 24% of cases imaged with both US & CT
- Color Doppler
  - Characteristic appearance of consolidated pneumonia: Tree-like vascularity extending from center to periphery
  - ↓ or absent color Doppler flow within consolidated lung suggests perfusion impairment & correlates with severity of necrosis on CECT

Nonvascular Interventions

- Percutaneous drainage of lung abscesses sometimes advocated, though creation of bronchopleural fistula may be problematic
- No clear role for percutaneous drainage in cases of cavitary necrosis in immunocompetent children

Imaging Recommendations

- Primary imaging modality of pneumonia: Radiographs
  - Insensitive to many complications
- CECT used in minority of pneumonia cases
  - Typically in child who has not responded to appropriate therapy despite lack of clear radiographic explanation
- Follow-up
  - Patients with cavitary necrosis demonstrated on CECT do not typically need follow-up CT to document resolution
  - Radiography obtained after 40 days is typically normal or near normal

DIFFERENTIAL DIAGNOSIS

Infected Congenital Lung Lesion

- Cavitary necrosis can have similar appearance to infected underlying congenital pulmonary airway malformation (CPAM)
- Cavitary necrosis tends to be surrounded by consolidated lung
  - CPAM often surrounded by aerated lung
- Children with cavitary necrosis tend to be more critically ill
- Temporal history most helpful; favor cavitary necrosis if
  - Previously normal chest radiograph
  - Progression of lesion during illness
  - ↓ or resolving lesion after acute illness

Lung Abscess

- Suppurative complications of pneumonia represent spectrum; name given depends upon severity, distribution, & temporal relationship to development of pneumonia
  - Cavitary necrosis: Nonenhancing, poorly defined walls of cysts
  - Lung abscess: Fluid collection with well-defined, thick, enhancing wall
    - Lung abscesses rare in otherwise healthy, immunocompetent children
      - Typically occur in immunocompromised children

Bronchopulmonary Sequestration

- Left > right lower lobe lesion, typically solid
- Does not contain cavities unless superinfected or hybrid lesion with CPAM
- Systemic arterial supply from aorta

Bronchogenic Cyst

- Typically not air-filled but fluid density
- More commonly in perihilar region/mediastinum

PATHOLOGY

General Features

- Etiology
  - Increasingly identified as pediatric pneumonia complication
    - May be due to changing spectrum of causative organisms since introduction of pneumococcal vaccine, ↑ recognition of cavitary necrosis as distinct clinical entity, or ↑ use of CT
  - Causative organisms in large USA retrospective study
    - *Streptococcus pneumoniae* most common, 22%
    - Others: Methicillin-sensitive & -resistant *Staphylococcus aureus*, *Pseudomonas* species, other *Staphylococcus & Streptococcus* species, *Fusobacterium*
- May lead to bronchopleural fistula & development of pneumothorax
  - In 1 large retrospective study, 86% had pleural effusion & 13% developed bronchopleural fistula
    - 100% of cases with bronchopleural fistula previously had chest drain in place > 7 days

Gross Pathologic & Surgical Features

- Lung inflammation → thrombosis of small arterioles with eventual ischemia & necrosis of consolidated lung
  - Tissue breakdown → cavity formation
  - Cavities initially fluid-filled
  - Cavities fill with air when tissue develops communication with aerated lung

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Lack of clinical improvement in pneumonia symptoms despite antibiotic therapy
  - Progressive sepsis
- Clinical profile
  - When children exhibit persistent or progressive symptoms (fever, respiratory distress, sepsis) despite appropriate medical management of pneumonia, suppurative complication (such as cavitary necrosis) usually present

Demographics

- Age
  - May occur in children of all ages
- Epidemiology
  - In mid 1990s, incidence of complicated pneumonias ↑ in children; higher frequency has remained through present time
  - Reason unclear
    - ↑ frequency of antibiotic resistant *Streptococcus pneumoniae*
    - ↑ viral infections (such as influenza A) that damage respiratory mucosa & render host susceptible to multiple infections
    - ↑ in antibiotic resistant bacteria

Natural History & Prognosis

- Patients with cavitary necrosis tend to be intensely ill
- Most do recover with nonsurgical management

Treatment

- Intensive support with IV antibodies
- Parental reassurance
- Surgery reserved for last resort (needed in minority of cases)
  - Surgical therapy may have role for necrotic pneumonia in adults; natural history of recovery without surgical intervention in children should be stressed

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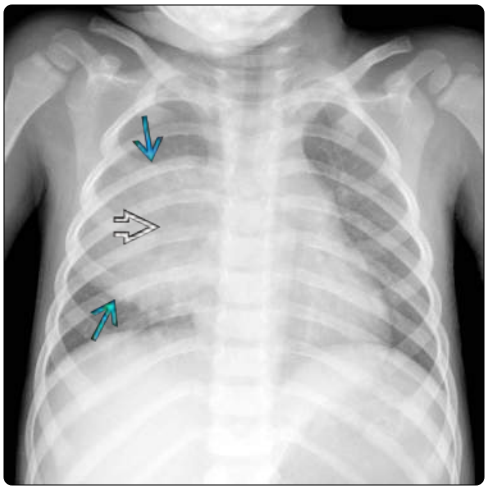
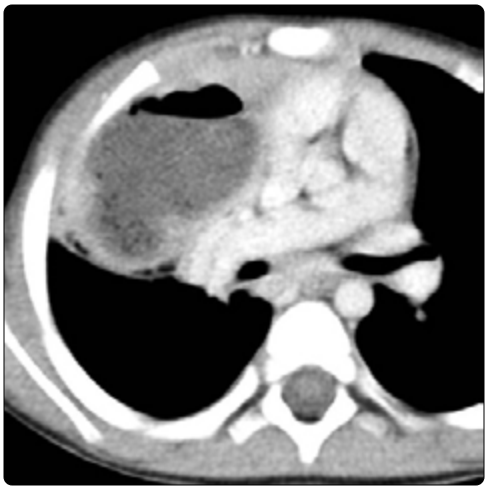
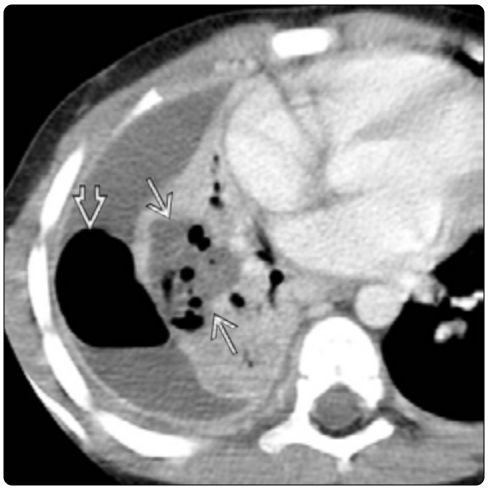
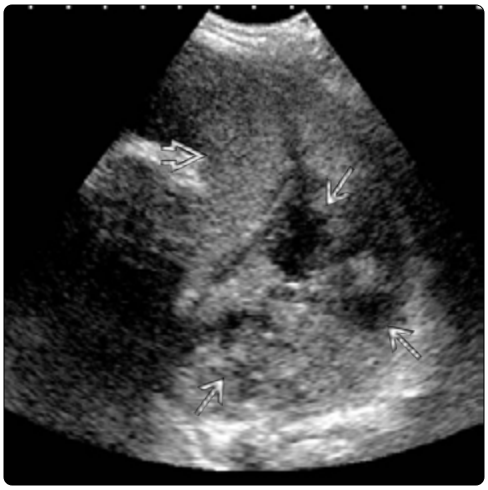


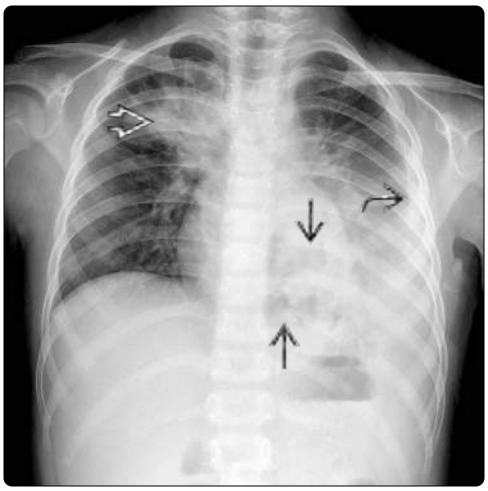


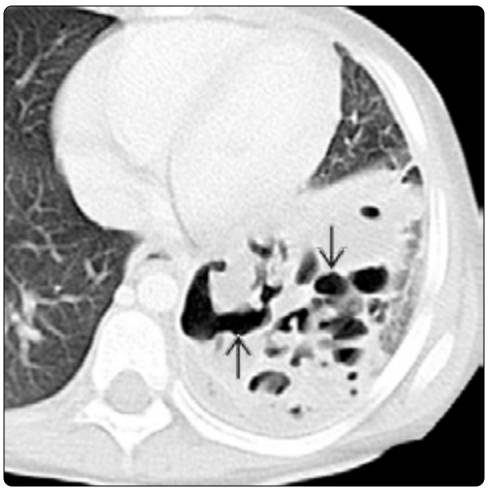
(Left) *AP radiograph in a young child shows a large, rounded opacity ⇨ in the right mid lung. There is faint central lucency ⇨.* **(Right)** *Axial CECT in same patient shows a cavity with an air-fluid level & surrounding opacified lung. CECT 4 months later (not included) showed a normal lung with complete resolution of the cavity & airspace opacification, indicating that the lesion was most consistent with cavitary necrosis rather than an underlying congenital lesion.*





(Left) *Axial CECT in an ill child shows a consolidated right lower lobe containing a central cavity ⇨ with a nonenhancing wall, consistent with cavitary necrosis. Note the large pleural effusion with both fluid & gas ⇨, suggesting a bronchopleural fistula.* **(Right)** *Transverse lung ultrasound from a 5-year-old girl with a LLL pneumonia & a pleural effusion shows multiple hypoechoic foci ⇨ in the consolidated LLL, concerning for cavitary necrosis. The spleen is partially visualized ⇨.*





(Left) *AP chest radiograph from the same patient demonstrates LLL consolidation with multiple foci of radiolucency ⇨ due to cavitary necrosis. There is also right upper lobe consolidation ⇨ & a left pleural effusion ⇨.* **(Right)** *Axial CECT from the same patient shows consolidation of the LLL with tubular & cystic lucencies ⇨ & loss of the normal lung architecture, consistent with cavitary necrosis.*

Fungal Pneumonia in Immunocompromised Children

KEY FACTS

TERMINOLOGY

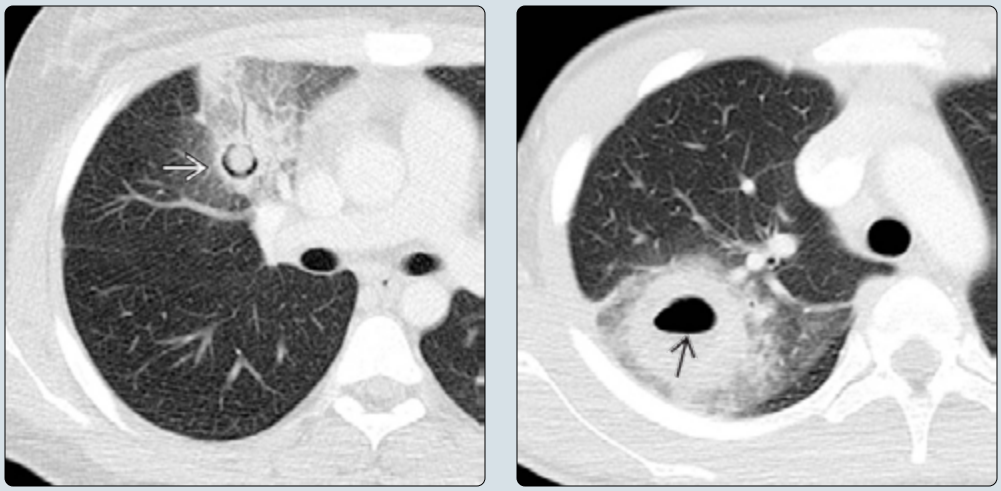
- Causes of immunocompromised states in children
  - Immature immune system: Premature infants
  - Primary immunodeficiency: Chronic granulomatous disease, Job syndrome, severe combined immunodeficiency, Wiskott-Aldrich syndrome, DiGeorge syndrome
  - Acquired immunodeficiency: Immunosuppression, infection, chronic illness

IMAGING

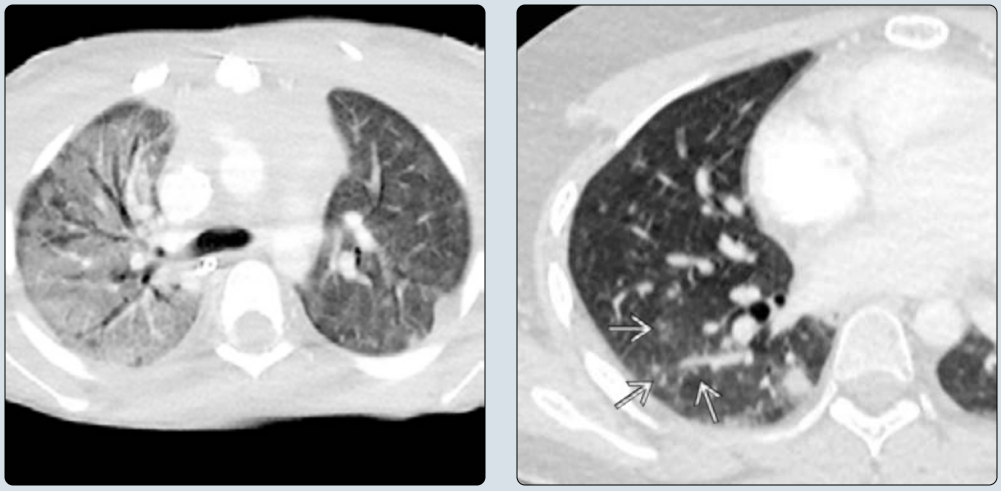
- Best clue: Pneumonia that does not respond to broad-spectrum antibiotics
- *Aspergillus*: Segmental & multifocal consolidation, small pulmonary nodules, & pleural effusion most common
  - Classic findings: Cavitation (~ 25% of patients), nodule with halo sign of surrounding ground-glass attenuation from hemorrhage (11% of patients), air-crescent sign (2% of patients)
- *Candida*: Consolidation, cannot be distinguished from bacterial pneumonia
- *Pneumocystis*: Extensive ground-glass opacity with interlobular septal thickening
- Coccidioidomycosis: Unilateral consolidation, hilar adenopathy, pleural effusion, pulmonary nodules, cavitary lesions, reticulonodular opacity
- *Cryptococcus*: Pulmonary nodules (miliary or large)
- Histoplasmosis
  - Preexisting infection: Calcified pulmonary nodule(s), calcified hilar or right paratracheal lymph nodes
  - Acute infection: Mediastinal or hilar adenopathy, miliary nodules, diffuse consolidation

CLINICAL ISSUES

- Antifungal therapy depends on type of fungus



(Left) *Axial CECT in an adolescent with acute lymphoblastic leukemia & an Aspergillus infection shows a region of ground-glass opacity (GGO) in the right middle lobe. This GGO (which represents hemorrhage) surrounds a cavity in which a crescent of air completely encircles a nondependent focus of necrotic lung, typical of invasive Aspergillus.* **(Right)** *Axial CECT in an adolescent with sickle cell disease & a Candida infection shows a necrotic pneumonia with an abscess that contains an air-fluid level.*



(Left) *Axial CECT in a patient being treated for rhabdomyosarcoma shows diffuse right lung GGO with mild septal thickening. The findings are consistent with Pneumocystis pneumonia.* **(Right)** *Axial CECT in a young adult treated with a bone marrow transplant for acute lymphoblastic leukemia shows multiple small tree-in-bud nodules in the right lower lobe. In an immunocompromised patient, this is concerning for an Aspergillus infection, though it is not specific.*

TERMINOLOGY

Associated Syndromes

- Causes of immunocompromised states in children
  - Immature immune system: Premature infants
  - Primary immunodeficiency: Chronic granulomatous disease, Job syndrome, severe combined immunodeficiency, Wiskott-Aldrich syndrome, DiGeorge syndrome
  - Acquired immunodeficiency
    - Immunosuppression: After transplant or chemotherapy, chronic corticosteroid therapy
    - Infectious: HIV/AIDS
    - Chronic illness: Cystic fibrosis, burns

IMAGING

General Features

- Best diagnostic clue
  - Pneumonia that does not respond to antibiotics
  - Appearance depends on type of fungus & type of immunodeficiency

Radiographic Findings

- *Aspergillus*: Normal early; consolidation & pleural effusion late
  - Cavitary lesions & air-crescent sign classic but uncommon late findings
- *Candida*: Perivascular pulmonary opacities may coalesce into abscess or empyema
- *Pneumocystis*: Diffuse bilateral interstitial or granular opacities
- Coccidioidomycosis: Unilateral consolidation, hilar adenopathy, pleural effusion
- *Cryptococcus*: Miliary nodules, large pulmonary nodules, large areas of consolidation, or lymphadenopathy
- Histoplasmosis
  - Acute infection: Mimics tuberculosis; mediastinal or hilar adenopathy & consolidation
  - Remote infection: Ca^{2+} of pulmonary nodule & lymph nodes

CT Findings

- *Aspergillus*: Segmental & multifocal consolidation, small pulmonary nodules, & pleural effusion most common
  - Classic findings: Cavitation (~ 25% of patients), nodule with halo sign of surrounding ground-glass attenuation (11% of patients), air-crescent sign (2% of patients)
- *Candida*: Consolidation
  - In disseminated disease, may see multiple microabscesses in liver &/or spleen
- *Pneumocystis*: Extensive ground-glass opacity with interlobular septal thickening
- Coccidioidomycosis: Unilateral consolidation, hilar adenopathy, pleural effusion, pulmonary nodules, cavitary lesions, reticulonodular opacity
- *Cryptococcus*: Miliary or large pulmonary nodules
- Histoplasmosis
  - Preexisting infection: Ca^{2+} of pulmonary nodule(s) & hilar or right paratracheal lymph nodes
  - Acute infection: Mediastinal or hilar adenopathy, miliary nodules, diffuse consolidation

Imaging Recommendations

- Best imaging tool
  - Chest radiograph best screening test for infection
  - NECT of chest best for defining extent & characterizing disease

DIFFERENTIAL DIAGNOSIS

Bacterial Pneumonia

- Appearance same as in immunocompetent patients

Viral Pneumonia

- Immunocompromised patients less likely to wheeze & more likely to develop consolidation

Pulmonary Metastases

- Usually appear as discrete, round, solid nodules

CLINICAL ISSUES

Demographics

- Epidemiology
  - *Aspergillus*
    - 5-13% of patients after bone marrow transplant
    - 5-25% of patients after solid organ transplant
    - 10-20% of patients with leukemia who receive intensive chemotherapy
    - Biggest risk for invasive aspergillosis is neutropenia
  - *Candida*
    - Major cause of morbidity & mortality in ICU/NICU
    - Risks: Central venous catheters, invasive interventions, severity of primary illness, treatment with broad-spectrum antibiotics
  - *Pneumocystis*
    - High risk in patients with lymphoid malignancies
  - Coccidioidomycosis
    - Endemic in Southwestern United States
    - Risk for exposure ↑ with storms, high winds, earthquake, & construction
  - *Cryptococcus*
    - Endemic in Southern California
    - Associated with avian guano
  - Histoplasmosis
    - Endemic in Ohio & Mississippi river valleys

Treatment

- Antifungal agent depends on type of fungus

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Papillomatosis

KEY FACTS

TERMINOLOGY

- Recurrent respiratory papillomatosis (RRP): Benign tumors of aerodigestive tract caused by infection with human papillomavirus (HPV)
  - Variable lifelong morbidity; potentially fatal course

IMAGING

- Locations: Junctional sites between respiratory & squamous epithelium
  - Larynx most common site
  - Extralaryngeal spread in ~ 30% of patients
    - Endobronchial spread → pulmonary nodules
      - Lung parenchymal involvement in 3%
- CT appearance
  - Airway: Soft tissue nodules protruding into lumen
  - Lung: Multiple solid or cavitated nodules
    - ± postobstructive atelectasis or pneumonia
    - Malignant degeneration of pulmonary disease in 0.5%
      - Squamous cell carcinoma

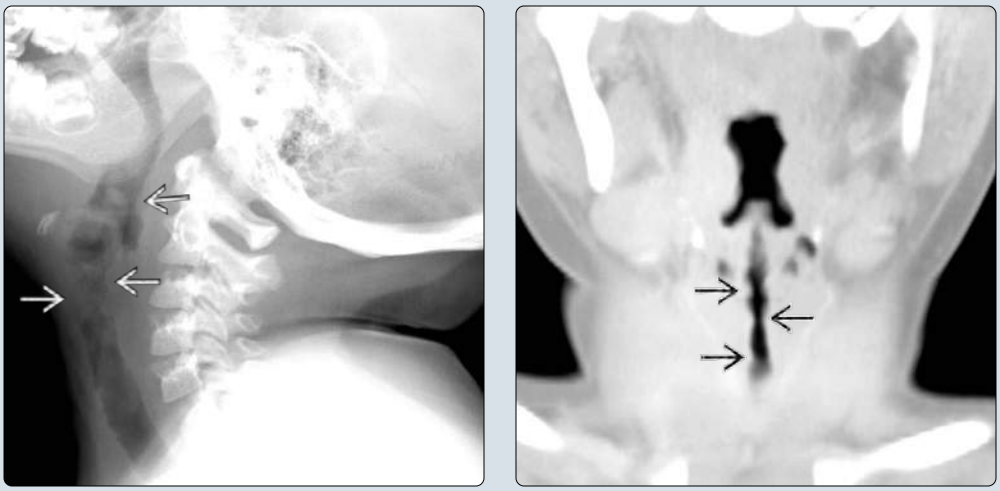
PATHOLOGY

- Perinatal transmission of HPV: Infected mother → child
  - ↑ risk: Vaginal delivery, firstborn, mother < 20 years of age; delivery time > 10 hours doubles risk
- Vast majority caused by HPV-6 & HPV-11

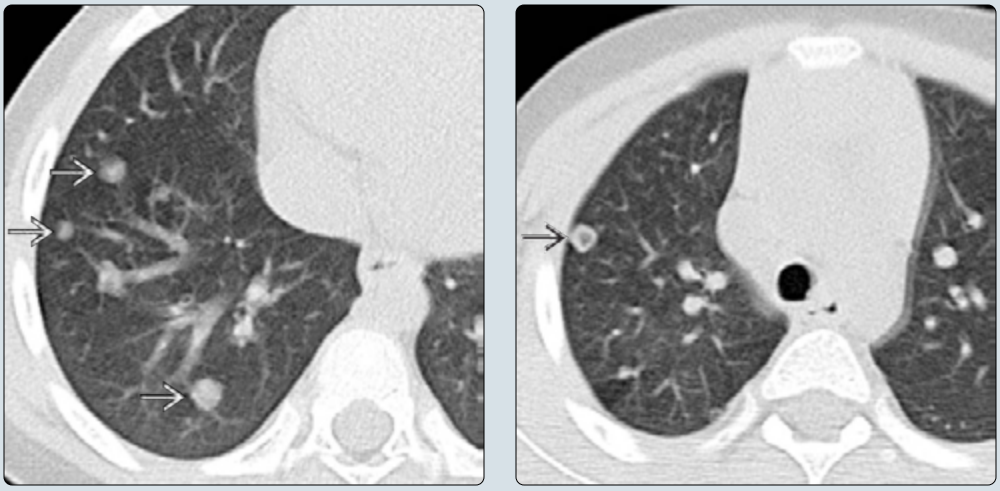
CLINICAL ISSUES

- Most common laryngeal tumor in children
- Most common symptoms: Hoarseness/voice change
- Mean age at diagnosis: ~ 4 years
- Disease remission can occur spontaneously at any stage
- Treatment with resection using powered microdebrider
  - Need for repeated debulking typical
- Tracheostomy needed in 10-15% due to airway obstruction; delayed as long as possible due to ↑ risk of distal spread
- HPV quadrivalent vaccine protects against viruses implicated in RRP & cervical cancer (HPV-6, 11, 16, 18)

(Left) *Lateral radiograph of the airway in a child with recurrent respiratory papillomatosis (RRP) shows multiple nodular filling defects ➡ from papillomas within the pharyngeal, laryngeal, & subglottic airways. RRP most commonly occurs in the larynx.* **(Right)** *Coronal CECT (in lung windows) in the same patient shows narrowing & nodularity ⇨ of the subglottic airway. Patients with RRP usually present with a hoarse voice. Other symptoms include dyspnea, respiratory infections, cough, & stridor.*



(Left) *Axial CT in the same patient shows multiple solid pulmonary nodules ➡ within the right lung. Nearly 30% of patients with RRP have extralaryngeal spread of tumor. Extension to the lung represents the most serious form of the disease.* **(Right)** *Axial CT in the same patient shows a cavitary pulmonary nodule ⇨ in the right lung. Cavitation is a common occurrence in pulmonary RRP. The cavity wall may be thin or thick.*



TERMINOLOGY

Abbreviations

- Recurrent respiratory papillomatosis (RRP)
- Juvenile onset RRP (JO-RRP)

Definitions

- Benign tumors of aerodigestive tract caused by infection with human papillomavirus (HPV)

IMAGING

General Features

- Best diagnostic clue
  - Wart-like growth in larynx
- Location
  - Occurs at junctional sites between respiratory & squamous epithelium
    - Larynx most common
    - Extralaryngeal spread in 30%: Oral cavity, trachea, bronchi, & esophagus
    - Tracheal or central bronchial involvement in 11%
    - Peripheral airway or alveolar involvement in 3%
      - Surgical manipulation of laryngeal papillomas ↑ risk of pulmonary dissemination
- Size
  - 1-2 mm to several centimeters
- Morphology
  - Warty lesions of larynx & tracheobronchial tree
  - Solid or cavitary pulmonary nodules

Radiographic Findings

- Radiography
  - Larynx, trachea, & main bronchi: Multifocal nodular irregularity of airway wall ± luminal narrowing
  - Lung: Multiple solid or cavitated nodules
    - As nodules enlarge, more likely to cavitate
    - Cavities may be thick or thin walled
    - Air-fluid levels suggest superinfection
    - Atelectasis & postobstructive pneumonia uncommon
    - Large mass suggests malignant degeneration

CT Findings

- CECT
  - Soft tissue nodules protruding into airway ± luminal narrowing
  - Pulmonary findings
    - Pulmonary nodules: May have mild enhancement
    - Cavitary lesions: Thin or thick, irregular enhancing walls
    - Malignant degeneration: Suspicious findings include heterogeneous enhancement, enlarging mass, or lymphadenopathy

Nuclear Medicine Findings

- PET/CT
  - May have areas of ↑ FDG uptake with or without malignant degeneration
    - Difficult to distinguish RRP from squamous cell carcinoma
    - May help target specific lesion for biopsy

Imaging Recommendations

- Best imaging tool
  - CT most sensitive to determine extent of disease & to identify complications

DIFFERENTIAL DIAGNOSIS

Subglottic Hemangioma

- Strongly enhancing tracheal nodule on CT
- Usually presents in infancy with stridor
- Lung lesions extremely rare

Wegener Granulomatosis

- Triad of pulmonary, paranasal sinus, & renal disease
- Pulmonary nodules may form thick-walled cavities
- Circumferential subglottic tracheal wall thickening

Metastases

- Variably sized lung nodules in patient with malignancy
- Cavitation uncommon in children

Septic Emboli

- Cavitated pulmonary nodules, often with thick irregular walls & surrounding lung consolidation
- Common causes: Endocarditis & septic thrombophlebitis
  - Lemierre syndrome: Septic thrombophlebitis of internal jugular vein after oropharyngeal infection → septic emboli

Pneumatocele

- Transient thin-walled cyst(s) of variable size
- Usually follows known insult (trauma, infection, hydrocarbon ingestion)

Lymphangioleiomyomatosis

- Thin-walled cysts of variable sizes
- Occurs in women of childbearing age
- May be sporadic or related to tuberous sclerosis complex

Langerhans Cell Histiocytosis

- Nodules &/or cysts, primarily in upper lung zones of older smokers

Invasive Aspergillosis

- Nodules have ground-glass "halo" of hemorrhage
- Immunocompromised host

PATHOLOGY

General Features

- Etiology
  - Perinatal transmission of HPV from infected mother to child
    - Risks: Active condylomata, primigravid mother, prolonged vaginal delivery, prolonged rupture of membranes, & newly acquired infection
    - 1 in 400 children delivered to women with active condylomata develop RRP
    - Still questionable if elective cesarean section protective
  - 95% of cases of RRP caused by HPV-6 & HPV-11
    - HPV-11 more common & causes more severe disease
      - Nearly all patients with malignant degeneration have HPV-11 infection

- Malignant transformation to squamous cell carcinoma in < 1% of RRP

Staging, Grading, & Classification

- Coltera/Derkay staging system
  - Separates aerodigestive tract into 25 subsites
  - Different sites evaluated & scored for severity of disease

Gross Pathologic & Surgical Features

- Pedunculated mass(es) with finger-like projections

Microscopic Features

- Multiple fronds with central fibrovascular core covered by stratified squamous epithelium
- Lung & laryngeal lesions composed of squamous cells; cavities lined with squamous epithelium
- Squamous atypia common even in benign lesions

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Hoarseness/voice change
- Other signs/symptoms
  - Dyspnea, chronic cough, recurrent upper respiratory infections, pneumonia, respiratory distress, dysphagia, & failure to thrive
  - Symptoms often present for ~ 1 year before definitive diagnosis established

Demographics

- Age
  - Bimodal age distribution
    - JO-RRP: Mean age at diagnosis: ~ 4 years
      - Younger age of onset = more severe disease
    - 2nd peak in adults 20-30 years of age = adult-onset RRP (AO-RRP)
      - Etiology most likely sexual transmission of HPV
      - AO-RRP typically less aggressive than in children
- Gender
  - M = F
- Epidemiology
  - Most common laryngeal tumor in children
  - In USA, RRP affects 4.3 per 100,000 children
  - Risk factors
    - Vaginal delivery, firstborn child, & mother < 20 years of age
    - Prolonged delivery time (> 10 hours) doubles risk for development of RRP
    - Active condylomata ↑ risk 231x

Natural History & Prognosis

- Benign disease with varying degrees of lifelong morbidity & potentially fatal course
- Papillomas show variable growth rate
  - Overall, frequent exacerbations typical
- Disease remission can occur spontaneously at any stage
  - Duration of remission variable & unpredictable
  - Laryngeal disease most likely to undergo remission
- Extralaryngeal spread in ~ 30% of patients
  - Tracheobronchial involvement in 11%
    - Tracheostomies associated with significant morbidity
      - Papillomas coalesce at tracheotomy site
      - Distal spread more likely
      - Tracheostomy needed in 10-15% due to airway obstruction; delayed as long as possible
  - Lung parenchymal involvement in 3%
    - Lung nodules grow very slowly, usually measured in decades
    - May cavitate & become secondarily infected
    - May get postobstructive atelectasis & pneumonia
  - Lung cancer (squamous cell carcinoma) in 0.5%
    - High suspicion needs to be maintained
    - Malignant degeneration more common in longstanding disease with history of prior irradiation & smoking
    - Look for increasing size of nodule on imaging
- Death due to large airway obstruction or respiratory failure

Treatment

- Current standard of care: Surgical resection using powered microdebrider
  - Need for repeated debulking typical
- Laser ablation now less commonly used
  - Virus may be aerosolized during ablation
    - ↑ risk of transbronchial spread to lungs
    - Risk to operating room personnel
- Medical therapies used as adjunct to surgery
  - Interferon & antiviral agents (cidofovir & acyclovir) may slow growth but not curative
  - 3-carbinol, retinoic acid, & photodynamic therapy have variable effect
- HPV quadrivalent vaccine protects against viruses implicated in RRP & cervical cancer (HPV-6, 11, 16, 18)

DIAGNOSTIC CHECKLIST

Consider

- Laryngeal mass or soft tissue nodules of airway should raise suspicion for RRP

Image Interpretation Pearls

- Review parenchyma & airways in soft tissue & lung windows on multiplanar CT reconstructions

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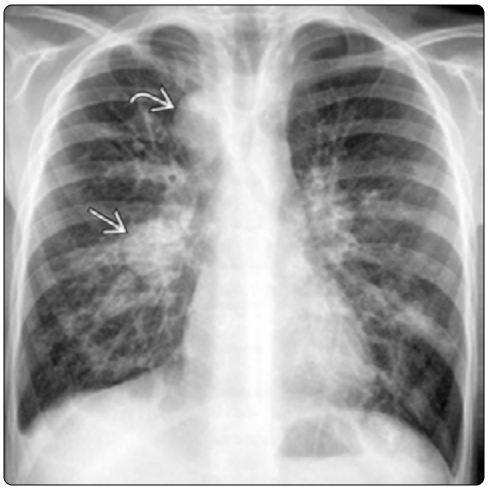
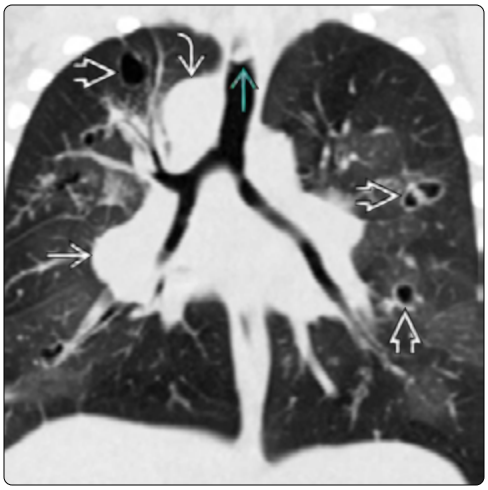
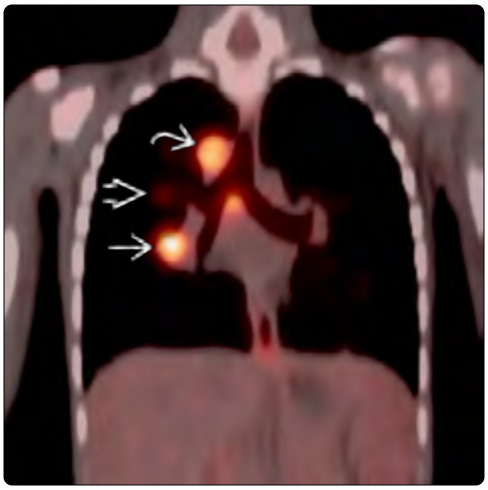
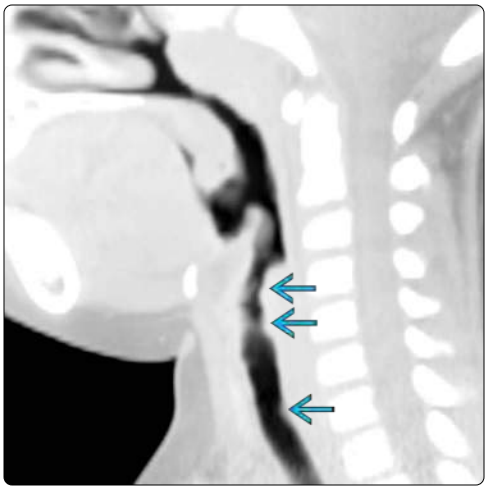


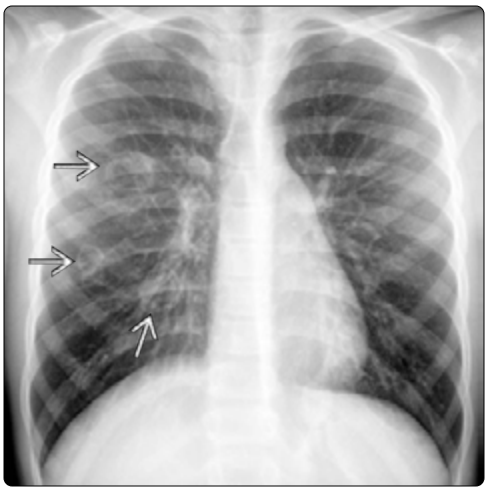


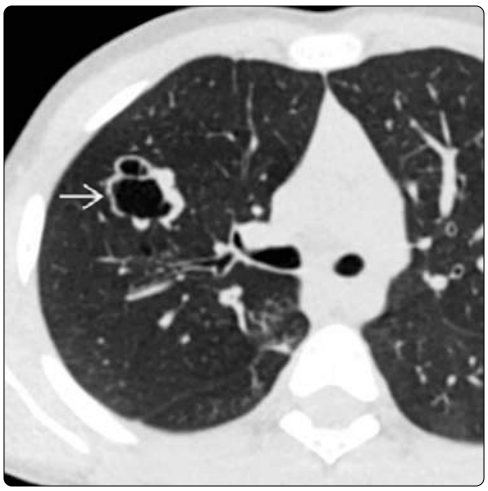
(Left) *PA chest radiograph in an adolescent with a history of RRP shows right hilar & right paratracheal masses. In patients with an enlarging mass & a history of RRP, squamous cell carcinoma should be suspected.* **(Right)** *Coronal CT in the same patient shows the right hilar & right paratracheal masses. In addition, there are multiple cavitary nodules in both lungs. Papillomas are also present in the trachea.*





(Left) *Coronal PET/CT in the same patient shows FDG uptake within the right hilar & right paratracheal masses. There is also mild uptake within a nodule in the right lung. PET/CT may have difficulty in distinguishing squamous cell carcinoma from papillomas as they both may take up the radiopharmaceutical. PET can potentially direct biopsy in this setting.* **(Right)** *Sagittal CT of the airway in an adolescent with RRP shows multiple papillomas at, above, & below the level of the glottis.*





(Left) *Chest radiograph in an adolescent with RRP shows multiple cavitary nodules in the right lung.* **(Right)** *Axial CT in the same patient shows a cavitary lesion in the right lung. Cavitary lesions in patients with RRP may have walls that are thin & smooth or thick & irregular (or a combination thereof).*

Pleuropulmonary Blastoma

KEY FACTS

TERMINOLOGY

- Rare malignant embryonal mesenchymal neoplasm of lung & pleura that arises during organ development

IMAGING

- Typically located in peripheral lung adjacent to or involving visceral pleura
- Tumor appearances
  - Type I: Air-filled, multilocular cystic mass
  - Type II: Mixed cystic (air-filled) & solid mass
  - Type III: Soft tissue mass; opacified hemithorax with contralateral mediastinal shift
- Pleural effusion in 77% of type II or III lesions
- Spontaneous pneumothorax at presentation in 30-65%
- Multiple lesions in 35-50%; bilateral in 11-26%
- Metastases to brain, bones, & rarely liver

TOP DIFFERENTIAL DIAGNOSES

- Congenital pulmonary airway malformation
- Congenital lobar overinflation
- Mesenchymal hamartoma of chest wall
- Undifferentiated sarcoma

PATHOLOGY

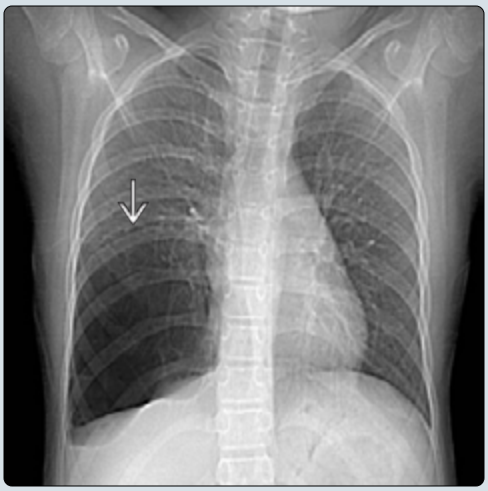
- Clear progression from type I → type II → type III
- *DICER1* mutation: Association of pleuropulmonary blastoma (PPB), multilocular cystic nephroma (MLCN), thyroid & ovarian tumors
  - Germline *DICER1* mutations in 65.5% with PPB
  - 10% with PPB have MLCN

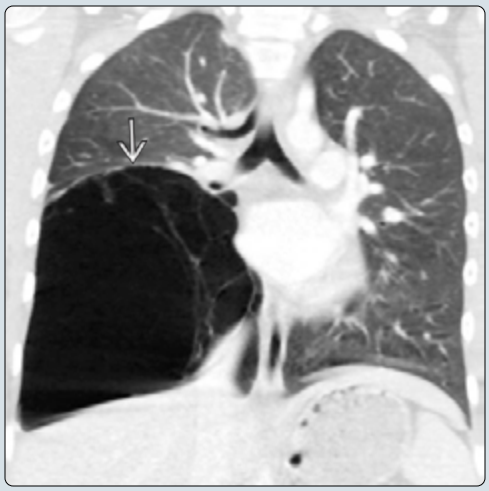
CLINICAL ISSUES

- Most common primary pulmonary malignancy of childhood
- 94% present in children < 6 years of age
- Most common presentation: Respiratory distress

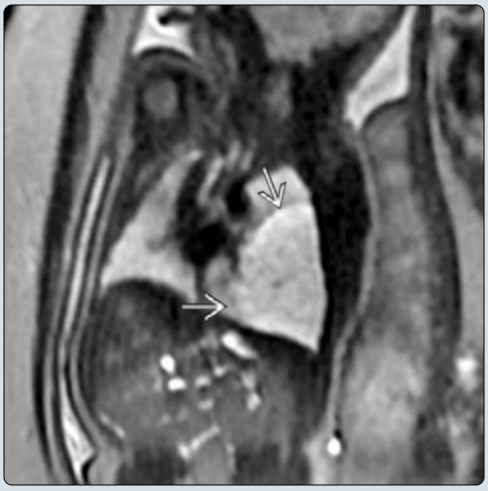
DIAGNOSTIC CHECKLIST

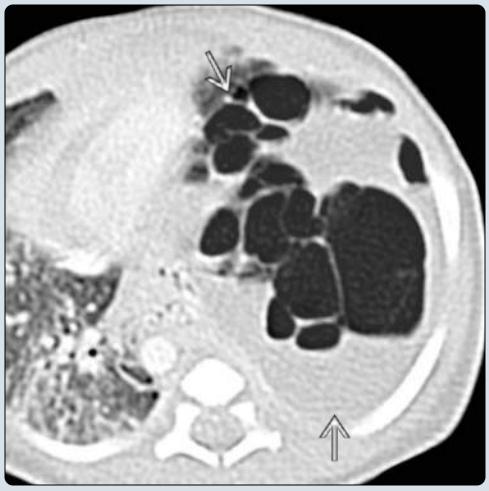
- Consider PPB when large chest mass seen in young child
- Imaging cannot distinguish type I PPB from benign cysts





(Left) *CT scanogram in an adolescent with a DICER1 mutation & pleuropulmonary blastoma (PPB) shows a cystic lesion ➡ occupying the lower 1/2 of the right hemithorax. PPB is the most common primary lung malignancy of childhood.* **(Right)** *Coronal CT of the chest in the same patient shows a large multiseptated cystic mass ➡ of the right hemithorax. Upon resection, a type I PPB was confirmed.*





(Left) *Coronal T2 SSFSE fetal MR shows a hyperintense solid-appearing left lung mass ➡ that is most typical of a type III congenital pulmonary airway malformation (CPAM) but was found after postnatal resection to be a PPB.* **(Right)** *Axial CECT in the same patient 3 days after birth shows a large multicystic left lower lobe lesion ➡ containing air & fluid, most typical of a type I CPAM. A type I PPB was actually confirmed upon resection. Limited fetal reports of PPB have been of type I lesions indistinguishable from CPAMs.*

TERMINOLOGY

Abbreviations

- Pleuropulmonary blastoma (PPB)

Definitions

- Rare malignant embryonal mesenchymal neoplasm of lung & pleura that arises during organ development

Associated Syndromes

- *DICER1* mutation
  - Associated with PPB, multilocular cystic nephroma (MLCN), ovarian sex cord-stromal tumors (especially Sertoli-Leydig cell tumor), multinodular goiter, & differentiated thyroid carcinoma

IMAGING

General Features

- Best diagnostic clue
  - Large pulmonary cystic, solid, or mixed mass in child < 6 years old
- Location
  - Intrathoracic, located in peripheral lung adjacent to or involving visceral pleura
  - No preference for right or left
- Size
  - 55% of type I lesions > 5 cm
  - 63% of type II or III lesions > 10 cm
- Morphology
  - Well-defined pulmonary mass
  - 3 types
    - Type I: Large, air-filled, multilocular cystic mass
    - Type II: Large, air-filled, cystic & solid mass
    - Type III: Solid pulmonary mass

Radiographic Findings

- Appearance depends on type of tumor
  - Type I: Well-circumscribed, multilocular cystic mass with thin septa
  - Type II: Cystic mass with variable soft tissue components
  - Type III: Soft tissue mass → opacified hemithorax with contralateral mediastinal shift
- Spontaneous pneumothorax at presentation in 30-65%
- Associated with pleural effusion in 77% of type II or III lesions

CT Findings

- Type I
  - Benign-appearing, air-filled lung cysts
  - Thin septa; no solid nodules or plaque-like thickening
  - Many different potential appearances: Unifocal (1 simple cyst), multilocular, cluster of contiguous cysts, or multifocal
- Type II
  - Air-filled cystic mass with variable soft tissue components
- Type III
  - Large, heterogeneous, solid mass with overall low attenuation
  - Originates from pulmonary pleura or parenchyma
  - Rarely invades chest wall
- Multiple lesions in 5-50% of patients
- Bilateral lesions in 11-26% of patients
- Metastases: Brain, bone, & rarely liver
- Imaging into upper abdomen may show associated cystic renal mass (MLCN)

Ultrasonographic Findings

- Solid heterogeneous mass (types II & III)
- Pleural fluid

Imaging Recommendations

- Best imaging tool
  - Radiograph often 1st imaging study due to respiratory symptoms
  - CT typically used to further characterize mass, evaluate initial extent of disease, & follow after treatment
    - Type I lesions often 1st imaged without contrast because of cystic appearance on radiograph
    - Type II & III lesions often imaged with contrast due to solid components

DIFFERENTIAL DIAGNOSIS

Congenital Pulmonary Airway Malformation

- Formerly known as congenital cystic adenomatoid malformation (CCAM)
- Types I & IV congenital pulmonary airway malformation (CPAM) indistinguishable from type I PPB
  - Factors favoring CPAM: Prenatal detection, systemic feeding vessel (in hybrid lesion), hyperinflated lung, or asymptomatic patient
- Type 1 CPAM: Macrocystic with large, air-filled cyst
  - ± air-fluid level on early neonatal imaging
- Type IV CPAM: Distal airway malformation
  - Current debate as to whether or not type IV CPAM is truly distinct from type I PPB

Congenital Lobar Overinflation

- Formerly known as congenital lobar emphysema
  - Hyperinflation of affected lobe/lung
  - On CT, not true cystic lesion: Lung has simplified appearance with small, widely spaced vessels

Mesenchymal Hamartoma of Chest Wall

- Intrathoracic mass of rib origin in neonate/infant
- Distortion/erosion of multiple adjacent ribs
- Ca^{2+} & fluid-fluid levels common

Undifferentiated Sarcoma

- May be indistinguishable from type III PPB
- More likely to invade chest wall

PATHOLOGY

General Features

- Genetics
  - Germline *DICER1* mutations in 65.5% of PPB patients
  - *DICER1* mutations present in ~ 100% with PPB & MLCN
- Associated abnormalities
  - Extrapulmonary lesions identified in ~ 25% of PPB patients
  - 10% associated with MLCN

Staging, Grading, & Classification

- Type I (33%): Purely cystic
- Type II (35%): Cystic & solid
- Type III (32%): Solid

Gross Pathologic & Surgical Features

- Type I: Cystic
- Types II & III: Soft, fleshy, friable, vascular tumor

Microscopic Features

- Histologically distinct from adult pulmonary blastoma, which has malignant epithelial & mesenchymal components
  - PPB has no malignant epithelial component
- Type I
  - Characteristic multilocular architecture with delicate septa
  - Thin cyst walls lined by alveolar-type epithelium
  - Small, primitive mesenchymal cells in stroma beneath epithelial lining
  - Focal areas of hypercellularity & hypervascularity
    - Hypercellular areas composed of hyperchromatic compact spindle cells & small round cells
  - ± foci of immature cartilage
- Types II & III
  - Solid components may have areas of necrosis & cystic degeneration
  - Higher grade cytologic features
    - Sheets of spindle, pleomorphic, or anaplastic cells
      - Anaplasia emerges as tumor progresses from type I to type III

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Respiratory distress ± spontaneous pneumothorax
    - Pneumothorax at presentation in 30% of patients with type I, 65% of patients with type II, & 20% of patients with type III
  - ± signs of upper respiratory tract infection
  - PPB may be incidental finding on prenatal US or postnatal chest radiograph

Demographics

- Age
  - 94% present in children < 6 years of age
  - Median age at diagnosis
    - Type I: 8 months
    - Type II: 35 months
    - Type III: 41 months
- Gender
  - Type I: 57% male
  - Types II & III: M = F
- Epidemiology
  - PPB: Most common primary pulmonary malignancy of childhood
    - 15-25 cases of PPB diagnosed in USA/year
    - Incidence of PPB estimated at 0.35-0.65 cases per 100,000 births

Natural History & Prognosis

- Clear progression from type I → type II → type III
- Type I
  - Presents at younger age
  - Better prognosis than types II & III
  - Complete surgical resection may be curative
  - Recurrent type I disease frequently progresses to more malignant type (II or III)
- Types II & III
  - Present at slightly older age
  - Multiple patients have history of pulmonary cysts preceding diagnosis of types II & III PPB
  - Worse prognosis
- 5-year survival
  - Type I: 91%
  - Type II: 71%
  - Type III: 53%
- Metastasis rate
  - Type I: No known metastases
  - Type II: 7%
  - Type III: 11%

Treatment

- Type I: Complete surgical resection ± chemotherapy
- Type II: Surgical resection + chemotherapy
- Type III: Surgical resection + chemotherapy; consider neoadjuvant chemotherapy
- Benefit of local radiation in types II & III PPB controversial

DIAGNOSTIC CHECKLIST

Consider

- PPB when large chest mass seen in young child
- Type I PPB radiologically indistinguishable from benign lung cysts
  - Argument for surgical excision of all cystic lung lesions found in young children

Image Interpretation Pearls

- PPB rarely invades chest wall
- Look for other *DICER1*-associated tumors

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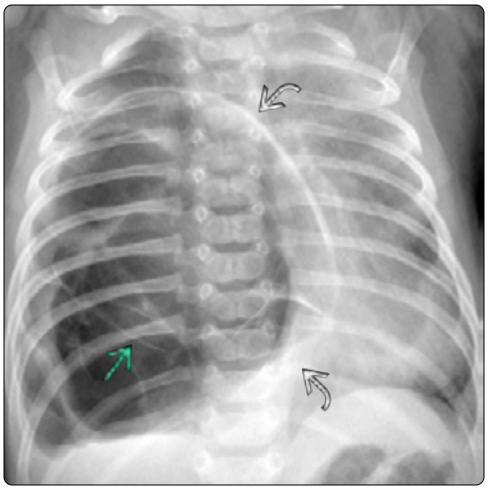
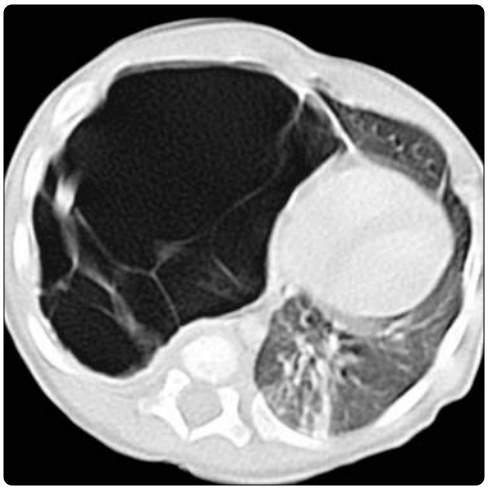
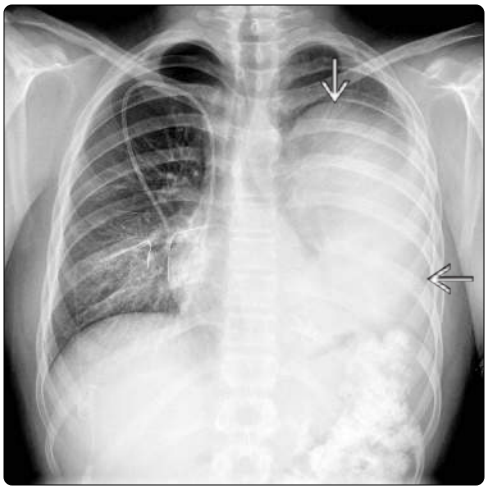
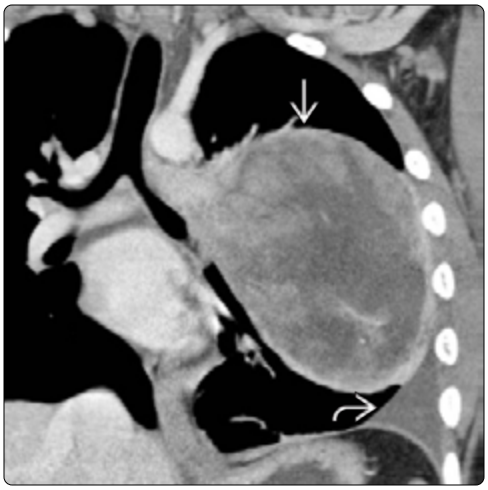


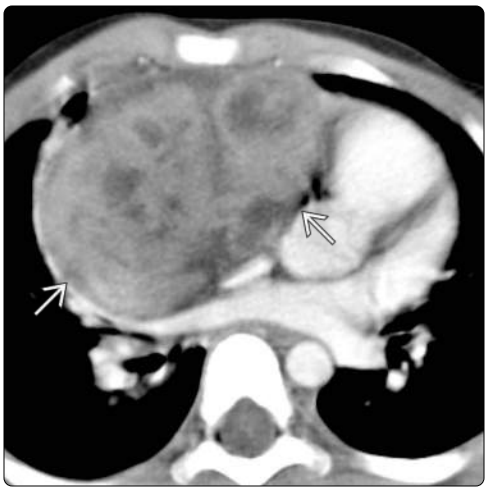


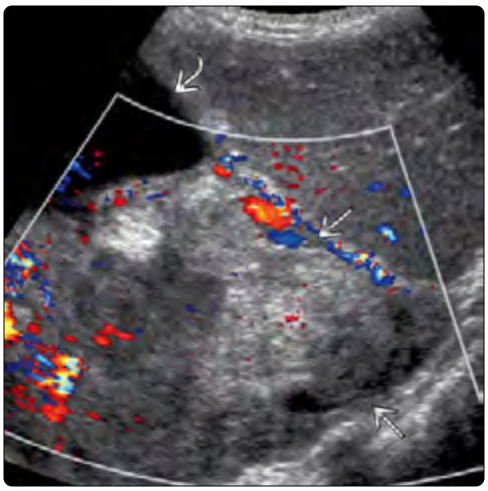
(Left) *AP radiograph of the chest shows a large, air-filled cystic mass of the right hemithorax. Numerous thin septations are seen within the mass. The mass crosses the midline & displaces the heart & mediastinum to the left.* **(Right)** *Axial CECT of the lungs in the same patient further details the multiseptated, cystic mass that occupies the entire right hemithorax. A multiloculated, air-filled, cystic mass without nodular or plaque-like solid tissue is typical of a type I PPB.*





(Left) *Chest radiograph in an adolescent boy with a PPB shows a large soft tissue mass occupying the left hemithorax. Type III PPB presents with a median age of 41 months.* **(Right)** *Coronal CECT of the chest in the same patient shows heterogeneous enhancement of the soft tissue mass in the left lung plus a small left pleural effusion. Pleural effusions are present in 77% of patients with type III PPB.*





(Left) *Axial CECT of the chest shows a solid mass of the anterior mediastinum. The mass is displacing the heart to the left. This is an unusual location for a PPB as they are typically located in the peripheral lung adjacent to or involving the visceral pleura.* **(Right)** *Longitudinal color Doppler ultrasound of a type III PPB shows a solid mass of the right hemithorax with mild internal vascularity. A moderate pleural effusion is present.*

Pulmonary Arteriovenous Malformation

KEY FACTS

TERMINOLOGY

- Pulmonary arteriovenous malformation (PAVM): Abnormal direct communication between pulmonary artery & vein
- Associated with hereditary hemorrhagic telangiectasia (HHT), a.k.a. Osler-Weber-Rendu syndrome

IMAGING

- Smoothly marginated, brightly enhancing nodule with feeding artery & draining vein; most in lower lobes
- Multidetector CTA allows best depiction of PAVMs & planning for transcatheter embolization

TOP DIFFERENTIAL DIAGNOSES

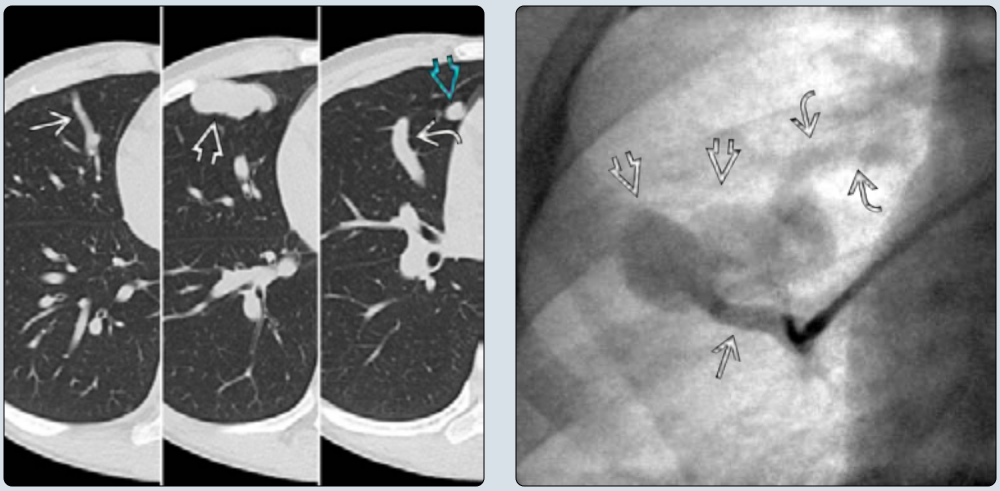
- Granuloma: No associated vessels; little/no enhancement; usually smoothly marginated; central CA^{2+} or densely calcified; ± associated calcified hilar & mediastinal lymph nodes
- Pulmonary varix: Enlarged pulmonary vein; no nidus or feeding artery; DSA useful to differentiate from PAVM
- Pulmonary metastasis: Frequently multiple; usually in setting of known primary malignant tumor

PATHOLOGY

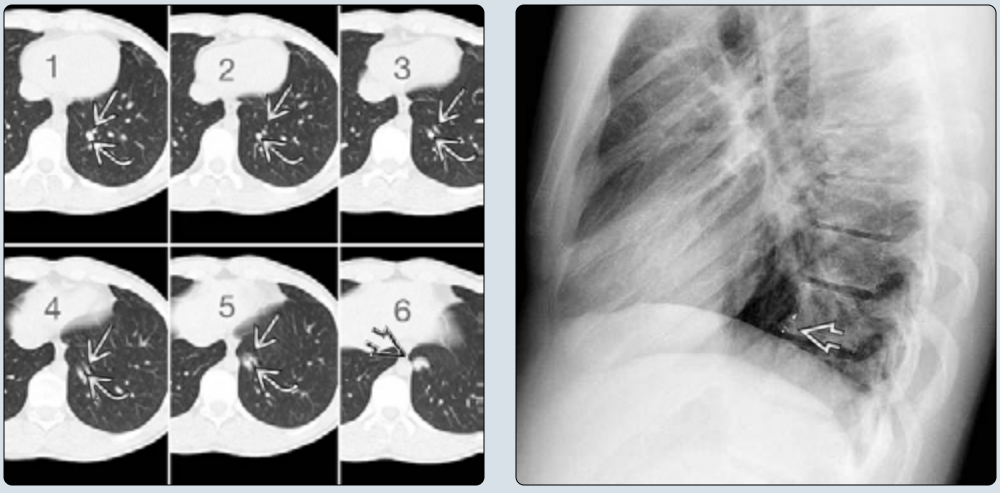
- HHT accounts for up to 90% of patients with PAVMs
- 30% of patients with HHT have PAVM
- Clinical triad: Epistaxis, telangiectasias, family history
- Should screen family members of patient with HHT & PAVM because 35% incidence of PAVM found

CLINICAL ISSUES

- Most HHT patients develop symptoms by age 20
- Consider referral to HHT Center of Excellence for endovascular treatment (coil, balloon, Amplatzer plug)
- Endovascular treatment allows embolization of multiple PAVMs at once; has very low morbidity & mortality with high success rate (permanent occlusion in > 98%)
- Untreated PAVMs can lead to neurologic complications (paradoxical emboli, abscess, stroke)



(Left) *Sequential (inferior to superior, left to right) lung CT images in a 12-year-old boy with hereditary hemorrhagic telangiectasia (HHT) show a large, lobulated pulmonary arteriovenous malformation (PAVM) in the RML. It is supplied by a single arterial feeder & has a larger caliber draining vein (which is typical). Note a second, smaller PAVM in the most superior image.* **(Right)** *Coned-down selective arteriogram in the same patient shows the lobulated PAVM, arterial feeder, & draining vein.*



(Left) *Sequential superior to inferior axial lung CT images (labeled 1-6) demonstrate a small PAVM in the left lower lobe with small feeding artery & adjacent draining vein.* **(Right)** *A follow-up lateral radiograph in the same patient shows a small Amplatzer occlusion device used to treat the PAVM shown in the preceding CT.*

TERMINOLOGY

Abbreviations

- Pulmonary arteriovenous malformation (PAVM)

Definitions

- Abnormal, capillary-free vascular communication between pulmonary artery & vein, allowing R → L shunt

IMAGING

General Features

- Best diagnostic clue
  - Smoothly marginated, avidly enhancing nodule with enlarged feeding artery & draining vein on CECT
- Location
  - 50-70% located in lower lobes
- Size: Most 1-5 cm; may enlarge over time to > 10 cm

Radiographic Findings

- Radiography
  - Abnormal in almost all symptomatic patients
  - Sharply defined pulmonary nodule with uniform density; may have lobulated borders

CT Findings

- CTA best to diagnose PAVMs (98% sensitivity)
  - Look for feeding artery & draining vein
  - Enhancement similar to that of other vascular structures

Angiographic Findings

- DSA: Selective pulmonary angiography
  - Less sensitive but may be more specific than CT
  - Large feeder artery + large early draining vein

Imaging Recommendations

- Multidetector CECT allows best detection of PAVMs & planning for transcatheter embolization
- Catheter angiography typically reserved for treatment

DIFFERENTIAL DIAGNOSIS

Granuloma

- No associated vessels; little to no enhancement
- Usually smoothly marginated but densely calcified or with central CA^{2+}; ± associated calcified hilar & mediastinal lymph nodes

Pulmonary Varix

- Enlarged pulmonary vein, no large feeding artery or nidus
- DSA may be needed to differentiate from PAVM

Pulmonary Metastasis

- Typically not associated with large vessels
- Frequently multiple & seen in setting of known primary malignant tumor

PATHOLOGY

General Features

- Etiology
  - Congenital (most common)
    - Thought to form from incomplete resorption of vascular septa during embryogenesis
  - Acquired
    - Surgery for congenital cyanotic heart disease: Late complication of Glenn & Fontan procedures
    - Hepatopulmonary syndrome: 50% with end-stage liver disease acquire abnormal arterial-venous communications
- Genetics
  - Autosomal dominant, variable penetrance
- Associated abnormalities
  - Hereditary hemorrhagic telangiectasia (HHT), a.k.a. Osler-Weber-Rendu syndrome, accounts for up to 90% of patients with PAVMs
    - 30% of patients with HHT have PAVM
    - Clinical triad: Epistaxis, telangiectasias, family history
    - Screen family members of HHT patients → 35% incidence of PAVM

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Incidental solitary pulmonary nodule on radiograph
  - Most HHT patients develop symptoms by age 20
    - Epistaxis is most common complaint
    - Findings or complications of R → L shunt
      - Neurologic (paradoxical emboli)
    - Mucocutaneous telangiectasias
- Other signs/symptoms
  - Right to left shunt: Dyspnea, cyanosis, clubbing
  - Hemoptysis

Demographics

- Age
  - Most are congenital; 10% detected in childhood

Natural History & Prognosis

- Patients with diffuse PAVMs are almost always symptomatic
- Lesions > 2 cm usually treated to avoid neurologic complications & heart failure

Treatment

- Options, risks, complications
  - Refer patient to HHT Center of Excellence
  - Endovascular coil, balloon, or Amplatzer vascular plugs
    - High success rate, permanent occlusion in > 98%
    - Low morbidity & mortality
    - Multiple PAVMs can be embolized at once

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Lymphoma

KEY FACTS

TERMINOLOGY

- Lymphoma: Malignant neoplasm arising from constituent cells of immune system or their precursors
- Hodgkin lymphoma (HL), non-Hodgkin lymphoma (NHL)

IMAGING

- Most common thoracic location: Anterior mediastinum
- US can help to distinguish normal thymus from abnormal thymus if nature of prominent anterior mediastinum unclear radiographically
- CECT: Typically used for initial diagnosis
  - Diffuse thymic infiltration: Enlarged & lobulated anterior mediastinal mass with fairly homogeneous soft tissue enhancement
    - Distorts, displaces, encases, & compresses adjacent structures
      - Airway compression: > 50% area reduction of trachea associated with respiratory failure during induction of anesthesia
    - Other locations in thorax: Hilum, axilla, supraclavicular region, lungs, pleura, pericardium
- PET: Very sensitive & specific (96.5%, 100%) for lymphoma
  - Changes initial stage in 10-23% of patients compared to conventional imaging
  - Can distinguish active disease from residual inactive mass
  - Negative PET performed after 2 cycles of chemotherapy: Good prognostic factor

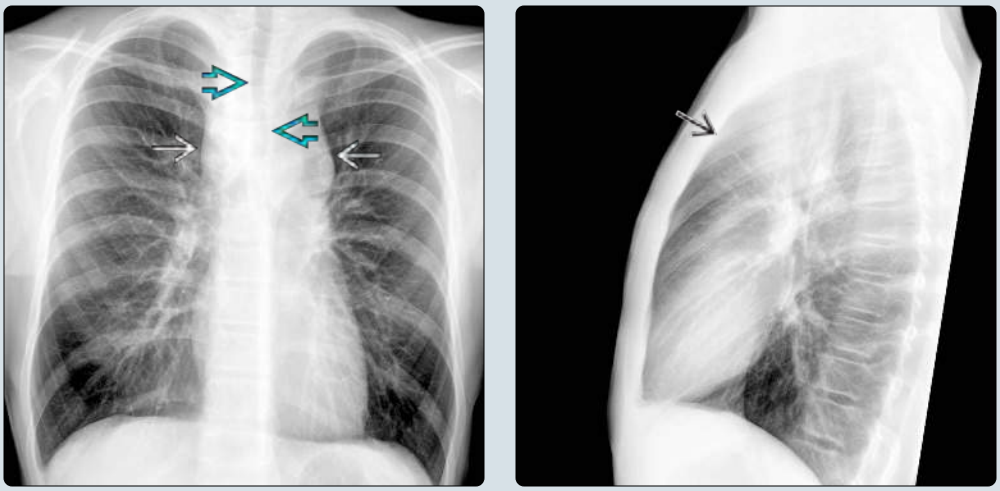
PATHOLOGY

- HL: Associated with Epstein-Barr virus in 50%
- NHL: Etiology depends on subtype, often translocation

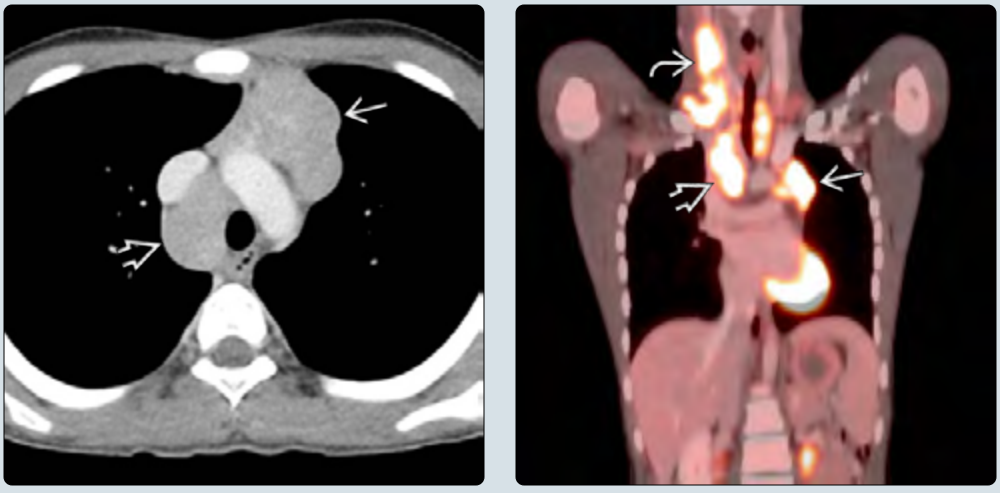
CLINICAL ISSUES

- Lymphoma 3rd most common pediatric malignancy
  - Accounts for 10-15% of all pediatric malignancies
- 60% of children with lymphoma have respiratory symptoms
- HL: 5-year survival = 91%
- NHL: 5-year survival = 70-76%

(Left) *Frontal chest radiograph in a 14-year-old boy with Hodgkin lymphoma shows a widened mediastinum with tracheal narrowing & deviation. Mediastinal involvement occurs in ~ 2/3 of pediatric patients with Hodgkin lymphoma.* **(Right)** *Lateral chest radiograph in the same patient shows partial obliteration of the anterior clear space. While this is a normal finding in infants due to the thymus, it is not normal in older children or adolescents & indicates the presence of an anterior mediastinal mass.*



(Left) *Axial CECT in the same patient shows a lobular anterior mediastinal mass & right paratracheal adenopathy. This mediastinal mass can be distinguished from the thymus due to its lobular contour & patient age.* **(Right)** *Coronal FDG PET/CT in the same patient shows abnormal FDG uptake in the aortopulmonary window, right paratracheal level, & right cervical chain. Approximately 80% of pediatric patients with Hodgkin lymphoma present with painless cervical adenopathy.*



TERMINOLOGY

Abbreviations

- Hodgkin lymphoma (HL), non-Hodgkin lymphoma (NHL)

Synonyms

- Hodgkin disease

Definitions

- Lymphoma: Malignant neoplasm arising from constituent cells of immune system or their precursors
  - Classic HL subtypes: Nodular sclerosis (NS), lymphocyte rich (LR), mixed cellularity (MC), lymphocyte depleted (LD)
  - NHL subtypes: Burkitt lymphoma (BL), diffuse large B-cell lymphoma (DLBCL), anaplastic large cell lymphoma (ALCL), lymphoblastic lymphoma (LBL)

IMAGING

General Features

- Best diagnostic clue
  - Anterior mediastinal mass or dominant nodal mass
- Location
  - Most common thoracic location: Anterior mediastinum
  - Other locations in thorax: Hilum, axilla, supraclavicular region, lungs, pleura, pericardium
- Size
  - Pathologic lymph nodes traditionally considered as > 1 cm in short axis
    - Negative & positive predictive values not 100%
  - Bulk disease in HL if ratio of maximum tumor:maximum intrathoracic diameter > 1:3
- Morphology
  - HL
    - Mediastinal disease in 2/3 of patients
      - Most often associated with NS subtype
    - Often associated with hilar adenopathy
  - NHL
    - Anterior mediastinum 2nd most common primary site
    - Frequency of intrathoracic involvement depends on subtype
      - 70% with LBL have mediastinal mass
      - ~ 25% with DLBCL have primary mediastinal disease
      - Mediastinum common location for ALCL
- Associated findings & complications
  - Superior vena cava compression/obstruction
  - Central airway compression/obstruction
  - Pleural effusion
  - Pericardial effusion
  - Pulmonary involvement

Radiographic Findings

- Anterior mediastinal mass
- Look for evidence of complications listed above
- Tracheal displacement or narrowing → airway compression

CT Findings

- CECT
  - Diffuse thymic infiltration: Enlarged & lobulated with fairly homogeneous soft tissue enhancement
    - Distorts, displaces, encases, & compresses adjacent structures
    - Ca^{2+} rare in untreated disease
  - Airway compression
    - > 50% reduction in area of trachea associated with respiratory failure during induction of anesthesia
      - Biopsy can be performed semierect with mild sedation in these patients
  - Lungs involved in 5-15% in HL & < 5% in NHL
    - Pulmonary nodules ± cavitation
    - Reticular interstitial pattern from venous or lymphatic obstruction
    - Lobar or segmental consolidation
  - ± pleural or pericardial effusions

MR Findings

- STIR
  - Sensitive for detection of bone marrow & soft tissue lesions
  - Cannot distinguish normal from abnormal lymph nodes
- Whole-body DWI with background body signal suppression (DWIBS)
  - Creates images similar to PET
  - Currently not as specific as FDG PET & requires correlation with lymph node size
    - All lymph nodes restrict diffusion; DWIBS ↑ conspicuity of nodes compared to other MR sequences
    - Must incorporate size criteria on anatomic imaging to help determine benign vs. malignant
    - ADC values currently not reliable indicator of benign vs. malignant
- Ultrashort TE
  - May be future of lung parenchymal assessment

Ultrasonographic Findings

- Grayscale ultrasound
  - Can help assess neck & axillary nodes, potentially guiding biopsy
  - Can look for pleural or pericardial effusions
  - Can help to distinguish normal thymus from abnormal thymus
    - Normal thymus: Uniform echogenic dot-dash pattern
    - HL/NHL: Heterogeneously hypoechoic lobular mass

Nuclear Medicine Findings

- PET
  - Very sensitive & specific (96.5%, 100%)
  - Changes initial stage in 10-23% of patients compared to conventional imaging
  - Can help to provide prognostic information
    - Negative PET performed after 2 cycles of chemotherapy: Good prognostic factor
  - Can distinguish active disease from residual inactive mass
  - PET/MR shows promise due to combination of functional imaging, anatomic imaging, DWI, & ↓ radiation

Imaging Recommendations

- Best imaging tool
  - CECT most commonly used for diagnosis
  - PET/CT used for staging & follow-up
- Protocol advice

- Beware placing patient in prone position or using sedation if there is compression of airway

DIFFERENTIAL DIAGNOSIS

Normal Thymus

- Large normal thymus occurs in children < 5 years of age
  - Lymphoma more common in teenagers
- Echogenic dot-dash pattern throughout on US
- Undulating margins; does not displace/compress airway or vessels

Germ Cell Tumor

- May be heterogeneous with cystic, fatty, or calcific foci

Lymphatic Malformation

- Multicystic mass of neck & mediastinum present since birth

Inflammatory Myofibroblastic Tumor

- Heterogeneously enhancing parenchymal or nodal mass with Ca^{2+} &/or necrosis

Thymoma

- Very uncommon in children

PATHOLOGY

General Features

- Etiology
  - HL: Associated with Epstein-Barr virus (EBV) in 50%
    - Other risks: Immunocompromised, ↓ socioeconomic status, small family size, early birth order
  - NHL: Etiology depends on subtype
    - BL: Translocation t(8;14) involving *MYC* (c-Myc) protooncogene
      - Also associated with EBV
    - DLBCL: Associated with immunocompromise
    - LBL: Associated with translocation of *TAL1* gene t(1;14)
- Associated abnormalities
  - Down syndrome has higher incidence of leukemia/lymphoma

Staging, Grading, & Classification

- HL subtypes (frequency): NS (70%), MC (20%), LR (5%), LD (5%)
- HL staging: Ann Arbor classification
  - Stage I: Single lymph node group
  - Stage II: 2 lymph node groups on same side of diaphragm
  - Stage III: Nodes on both sides of diaphragm
  - Stage IV: Extranodal sites
  - Designations applicable to any stage
    - A: Asymptomatic
    - B: Symptoms of fever, night sweats, weight loss > 10%
- NHL subtypes (frequency): BL (30%), DLBCL (10-20%), ALCL (10%), LBL (30%)
- NHL staging: St. Jude/Murphy classification
  - Stage I: Single nodal area outside abdomen & mediastinum
  - Stage II: Single mass with regional nodal involvement; 2 or more nodal areas on same side of diaphragm; or primary gastrointestinal tumor
  - Stage III: Nodal areas on both sides of diaphragm; primary intrathoracic or extensive abdominal disease; paraspinal or epidural tumor
  - Stage IV: Bone marrow or CNS disease

Microscopic Features

- Hodgkin: Reed-Sternberg cell
- Non-Hodgkin: Clonal proliferation of T- or B-cell origin

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 60% of children have respiratory symptoms
  - HL may present with painless adenopathy
- Other signs/symptoms
  - HL: B symptoms of fever, night sweats, weight loss
  - NHL: Can have life-threatening symptoms due to compression of trachea or superior vena cava or large pleural or pericardial effusion

Demographics

- Age
  - Much more common in 2nd decade of life
  - Prevalence of NHL ↑ with age
- Gender
  - HL: M > F under 15 years old; F > M over 15 years old
  - NHL: M > F
- Epidemiology
  - Lymphoma: 3rd most common pediatric malignancy
    - Accounts for 10-15% of all pediatric malignancies

Natural History & Prognosis

- HL: 5-year survival = 91%
- NHL: 5-year survival = 70-76%

Treatment

- HL: Chemotherapy ± radiation
- NHL: Chemotherapy ± bone marrow transplantation or radiation therapy

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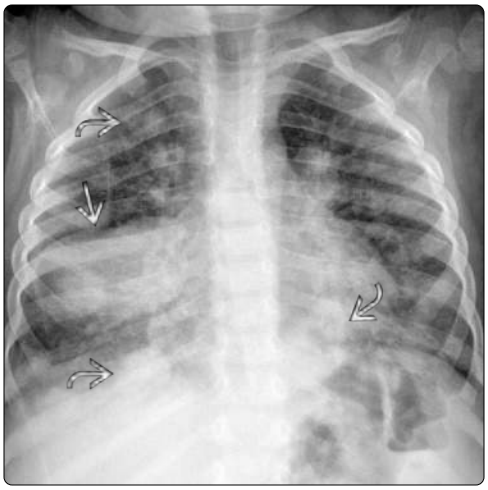
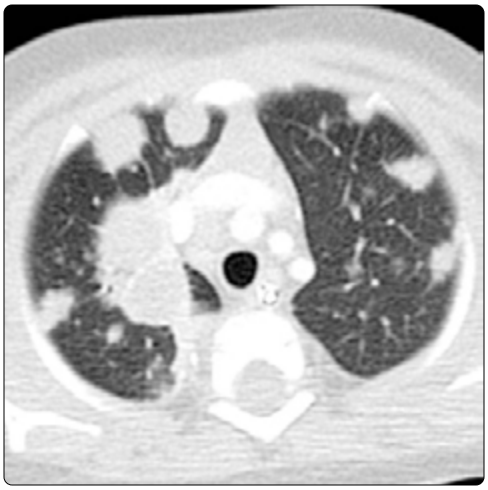
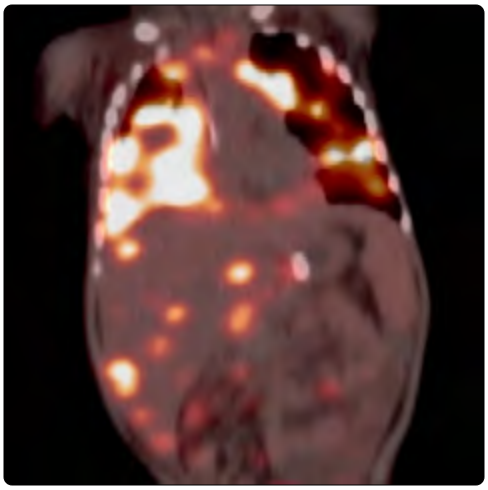
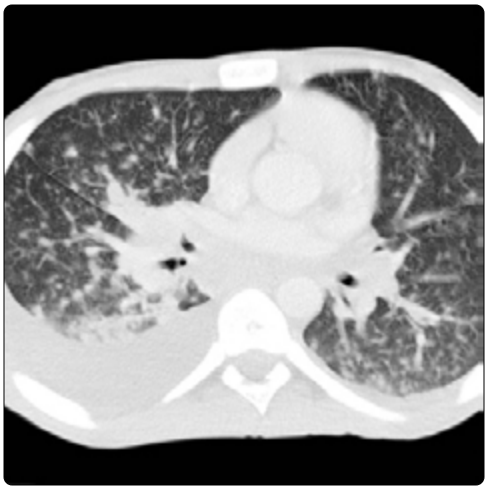


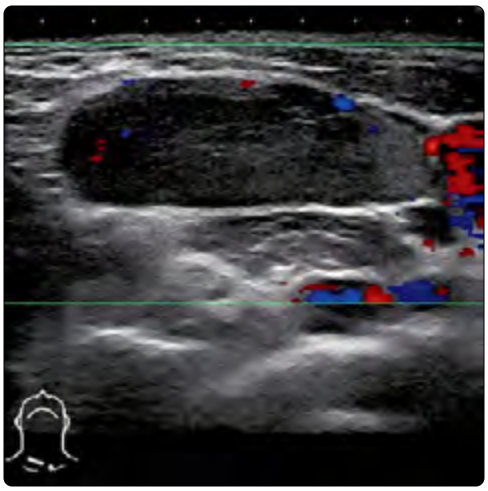


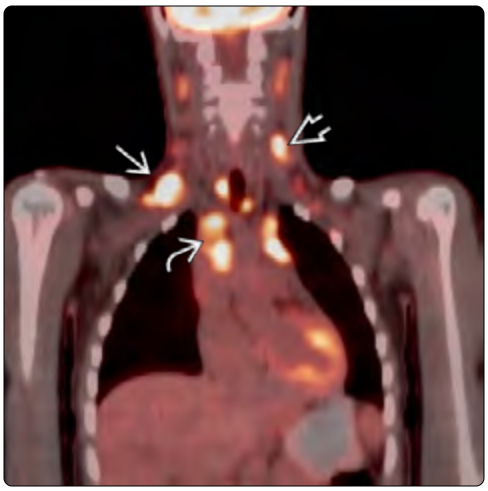
(Left) *Frontal chest radiograph in a 2-year-old patient with diffuse large B-cell lymphoma shows multiple pulmonary masses. The largest mass is in the right middle lobe. Smaller masses are present in both lungs.* **(Right)** *Axial chest CECT in the same patient shows multiple well-circumscribed pulmonary nodules. There are 3 potential patterns of lung involvement in lymphoma: Pulmonary nodules, reticular interstitial pattern, & airspace consolidation.*





(Left) *Coronal PET/CT in the same patient shows abnormal FDG uptake in the lungs & liver. Pulmonary involvement in non-Hodgkin lymphoma is uncommon, occurring in 5% of patients.* **(Right)** *Axial chest CECT in an adolescent with Hodgkin lymphoma shows a diffuse reticular interstitial pattern of pulmonary involvement. Moderate right & small left pleural effusions are also present. This pattern of disease occurs because of lymphatic or venous obstruction.*





(Left) *Ultrasound of the right supraclavicular region in a 15-year-old girl with Hodgkin lymphoma shows an enlarged lymph node with loss of the normal nodal architecture (i.e., fatty hilum & radiating vessels).* **(Right)** *Coronal PET/CT in the same patient shows abnormal FDG uptake in the right supraclavicular node in addition to mediastinal & left cervical chain nodes.*

Germ Cell Tumors

KEY FACTS

TERMINOLOGY

- Germ cell tumor (GCT) types: Teratoma, seminoma, nonseminomatous germ cell tumor (NSGCT)
  - Teratoma: Mature, immature, malignant
  - Seminoma: Germinoma & dysgerminoma
  - NSGCT: Embryonal cell, yolk sac tumor, choriocarcinoma, & mixed GCT
- Derived from primordial germ cells that differentiate into embryonic & extraembryonic structures

IMAGING

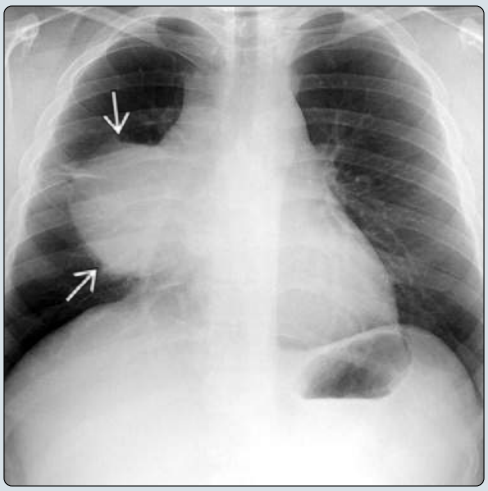
- Best clue: Heterogeneous anterior mediastinal mass arising within or adjacent to thymus
  - Less common locations: Posterior mediastinum, heart, pericardium
- Teratoma
  - Mostly cystic + soft tissue, fat, & calcium
    - 1 or more cysts; 20-40% calcify; 93% contain fat
    - CECT sensitive for these components
- Seminoma
  - Homogeneous, lobulated, bulky soft tissue mass
  - Often straddles midline & has mass effect
- NSGCT
  - Heterogeneous mass with hemorrhage & necrosis
  - Irregular margins: Obliterated fat planes & lung invasion
- Pericardial lesions often cause pericardial effusion
- Beware of potential airway collapse from tumor compression during sedation/general anesthesia

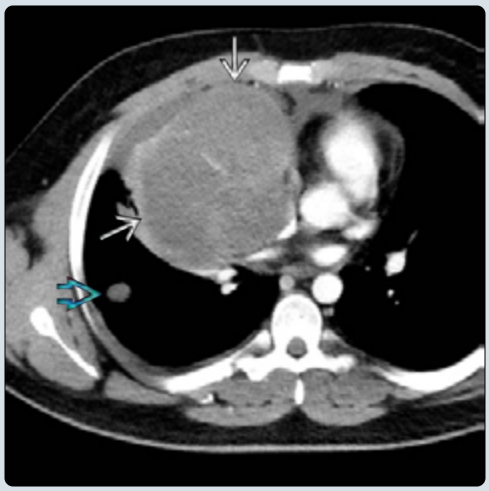
PATHOLOGY

- 50-300x ↑ risk of mediastinal GCT with Klinefelter syndrome

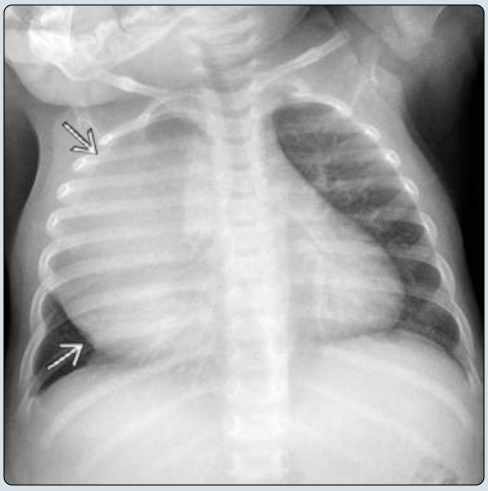
CLINICAL ISSUES

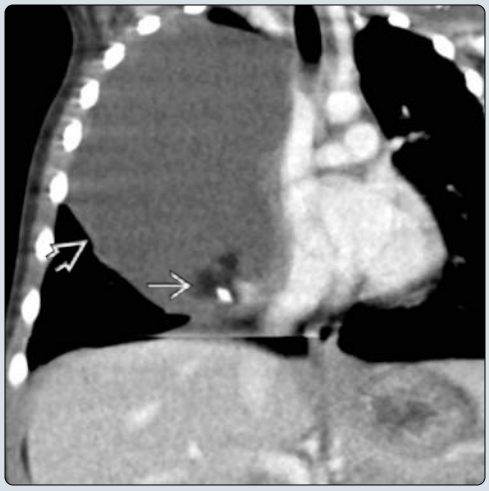
- Dyspnea, chest pain, cough, superior vena cava syndrome, hoarseness, fever, weight loss
- Asymptomatic in 50-60% of teratomas, 38% of seminomas, & 10% of NSGCTs





(Left) *Frontal chest radiograph in a young adult male with a yolk sac tumor shows a large mass in the right mid chest obscuring the right heart border. Yolk sac tumors are considered a nonseminomatous germ cell tumor.* **(Right)** *Axial CECT in the same patient shows a round mass of the anterior mediastinum that is hypodense compared to muscle. There is a solitary pulmonary nodule in the right lung base. Nonseminomatous germ cell tumors can metastasize to the lymph nodes, lung, & liver.*





(Left) *Chest radiograph in a 1-year-old patient with a mature teratoma shows a large mass abutting the right heart border & much of the right hemithorax. This is larger than typically seen for normal thymus, & the right hemidiaphragm is mildly depressed.* **(Right)** *Coronal CECT in the same patient shows a large mass in the right hemithorax. The mass is mostly fluid density but shows a nodule inferiorly that contains fat & calcium. The presence of cystic, fatty, & calcific foci is diagnostic of a teratoma.*

TERMINOLOGY

Abbreviations

- Germ cell tumor (GCT)

Synonyms

- Teratoma, seminoma, nonseminomatous germ cell tumor (NSGCT)

Definitions

- Tumor derived from primordial germ cells
- Teratoma: 3 major types
  - Mature teratoma: Characterized by mixture of adult-type tissues derived from 3 germinal layers
  - Immature teratoma: Contains embryonic or fetal tissue, often neuroepithelial structures
  - Malignant teratoma: Contains foci of malignant transformation
- Malignant GCT: Seminoma & NSGCT
  - Seminoma: Germinoma & dysgerminoma
  - NSGCT: Embryonal cell, yolk sac tumor (YST), choriocarcinoma, & mixed GCT

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous anterior mediastinal mass arising within or adjacent to thymus
- Location
  - Anterior mediastinum most common
  - Less common locations: Posterior mediastinum, heart, pericardium
- Size
  - Average size for NSGCT: 9 cm; seminoma: 5 cm
- Morphology
  - Teratoma: Mostly cystic with soft tissue, fat, & calcium elements
  - Seminoma: Homogeneous, lobulated, bulky mass
  - NSGCT: Heterogeneous soft tissue mass with hemorrhage & necrosis

Radiographic Findings

- Teratoma
  - Round, sharply marginated, anterior mediastinal mass
  - 20-40% calcify
- Seminoma
  - Bulky, lobulated, anterior mediastinal mass
- NSGCT
  - Large, anterior mediastinal mass
  - Pleural or pericardial effusions may be present

CT Findings

- NECT
  - Teratoma
    - Sharply marginated anterior mediastinal mass
    - Extends to 1 side of midline
    - Fatty, cystic/solid soft tissue, & calcified components often visible
      - 93% contain fat
  - Seminoma
    - Large solid mass with homogeneous density
    - Straddles midline; mass effect on adjacent structures
  - NSGCT
    - Large, heterogeneous mass
    - Central area of hemorrhage or necrosis
    - Irregular margins with obliterated fat planes
    - May invade lung, chest wall, & diaphragm
    - Metastasizes to lymph nodes, lung, & liver
- CECT
  - Better defines extent of lesion & invasion
    - Look for compression of vessels & airway
  - More clearly differentiates solid & cystic components
  - Teratoma: Enhancement of rim & septations
  - Seminoma: Mild enhancement
  - NSGCT: Enhancement of peripheral soft tissue around central necrotic region

MR Findings

- T1WI
  - Teratoma: Well-circumscribed lesion
    - Varied signal of fat, soft tissue, calcium, & fluid
      - Fluid varies in signal due to protein content
- T2WI
  - Teratoma: Signal dependent on tissue types in lesion
    - Fluid has high signal
    - Look for signal drop-out on fat saturated images

Imaging Recommendations

- Best imaging tool
  - CECT demonstrates differing tissue components, extent of disease, & complications
  - Beware of potential airway collapse from tumor compression during sedation/general anesthesia

DIFFERENTIAL DIAGNOSIS

Normal Thymus

- Large, quadrilateral-shaped, homogeneous tissue
- Uniform dot-dash pattern of echoes on US

Lymphoma

- Bulky, lobulated, homogeneous mass compressing & displacing airway & vessels
- Rarely calcifies before treatment

Lymphatic Malformation

- Congenital multicystic mass of neck & chest
- May have soft tissue attenuation following hemorrhage

Thymic Cyst

- Usually unilocular, may be bilobed with neck & mediastinal components
- Often asymptomatic but can compress adjacent structures

Thymoma

- Rare in children
- Associated with paraneoplastic syndromes

PATHOLOGY

General Features

- Etiology
  - Most common theory: Local transformation of primordial germ cells misplaced during embryogenesis

- Associated abnormalities
  - 50-300x ↑ risk of mediastinal GCT with Klinefelter syndrome
  - Possible relationship to Li-Fraumeni syndrome & Down syndrome
  - NSGCT associated with hematologic malignancies

Staging, Grading, & Classification

- Moran & Suster classification
  - Similar to WHO classification of gonadal GCT
  - Differs by further classifying malignant teratomas

Gross Pathologic & Surgical Features

- Teratoma: Well-encapsulated, predominantly cystic
  - Cystic component usually unilocular
  - May be adherent to mediastinal structures
- Seminoma: Unencapsulated, well circumscribed, large
  - Usually has solid, uniform appearance with focal areas of necrosis or hemorrhage
- NSGCT: Unencapsulated, irregularly marginated, heterogeneous large mass
  - Large areas of necrosis, hemorrhage, & cysts
  - Invades local structures

Microscopic Features

- Teratoma
  - Admixture of adult-type tissues
  - Derived from 3 germinal layers
  - Most common components: Skin & appendages, pancreatic, bronchial, neural, gastrointestinal, muscular, fatty, & cartilaginous elements
- Seminoma
  - Sheets of round or polygonal cells surrounded by septa of connective tissue infiltrated by lymphocytes
  - Prominent eosinophilic nucleoli with characteristic spiked appearance
- NSGCT
  - Embryonal: Large malignant cells arranged in sheets or tubular/acinar patterns
  - YST: Reticular pattern with cords of tumor cells embedded in myxoid stroma most common
  - Choriocarcinoma: Mix of syncytiotrophoblast, cytotrophoblast, & intermediate trophoblast

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic in 50-60% of teratomas, 38% of seminomas, & 10% of NSGCTs
  - Dyspnea (25-48%), chest pain (23-52%), cough (17-24%), superior vena cava syndrome (6-14%), hoarseness (1-14%), fever (13%), weight loss (11%)
  - α-fetoprotein elevated in 74% of patients with NSGCT; β-HCG elevated in 38%
- Other signs/symptoms
  - Gynecomastia, testicular atrophy, facial fullness, precocious puberty
  - If teratoma contains endocrine pancreas, can cause hyperinsulinism & hypoglycemia
  - Rare rupture of cystic component of teratoma → erosion into tracheobronchial tree followed by expectoration of sebaceous material & hair

Demographics

- Age
  - Separated into prepubertal & postpubertal due to differences in genetics & clinical behavior
- Gender
  - Mature teratoma: M = F
  - Malignant GCTs
    - Children: M = F [except in YST where F > M (4:1)]
    - Teenagers & adults: M >> F
- Epidemiology
  - GCT 3rd most common mediastinal tumor in children
  - Mature teratoma: 19-24% of anterior mediastinal tumors in children
    - Teratoma: 44% of mediastinal GCT
    - Seminoma: 16-37% of mediastinal GCT
    - NSGCT: 14% of mediastinal GCT
    - Mixed GCT: 13-25% of mediastinal GCT

Natural History & Prognosis

- Prognosis dependent on multiple factors
  - Age < 30 years associated with better prognosis
  - Histological type
    - Mature teratoma: Benign (80% of mediastinal GCT)
    - Immature teratoma: Good prognosis, no risk of recurrence or metastasis
    - Teratoma with component of malignant GCT: Prognosis depends on component
    - Seminoma: 5-year survival rate = 90%
    - NSGCT: 5-year survival rate = 48%
  - Presence of metastases
  - Stage: In children, prognosis related to stage
  - Status of resection: Complete resection strongest prognostic indicator
  - Tumor markers: α-fetoprotein > 10,000 ng/mL = worse prognosis; ↑ β-HCG = worse prognosis in adults

Treatment

- Mature teratoma: Surgery
- Seminoma: Chemotherapy followed by surgery for residual disease
- Nonseminomatous: Chemotherapy & surgery

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- If Ca^{2+} present, mass = teratoma until proven otherwise
- NSGCT invades surrounding structures
- Large mediastinal masses have risk of cardiopulmonary arrest at induction of anesthesia in patients with tracheal or vascular compression

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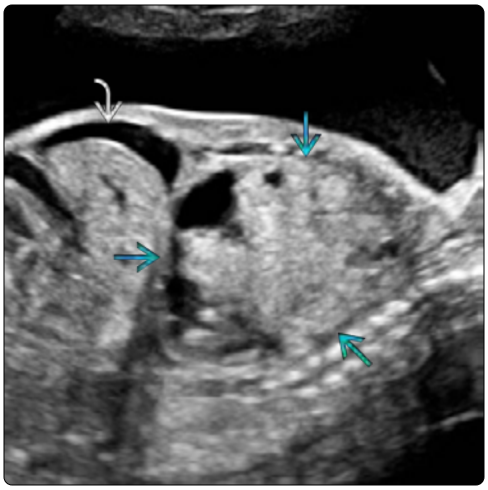
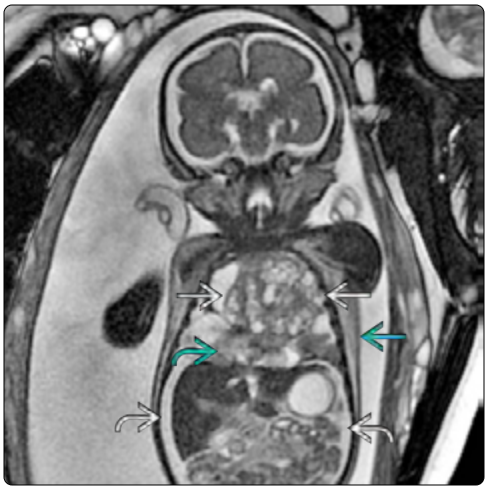
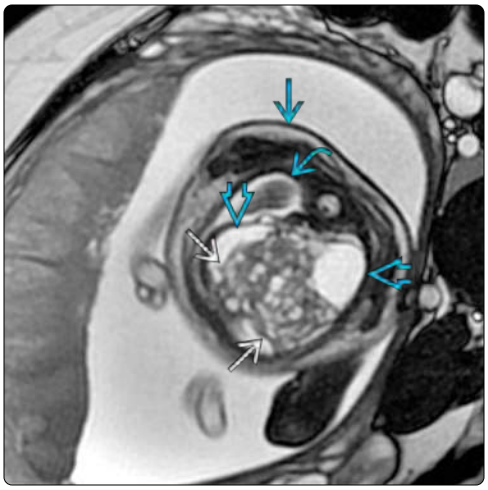
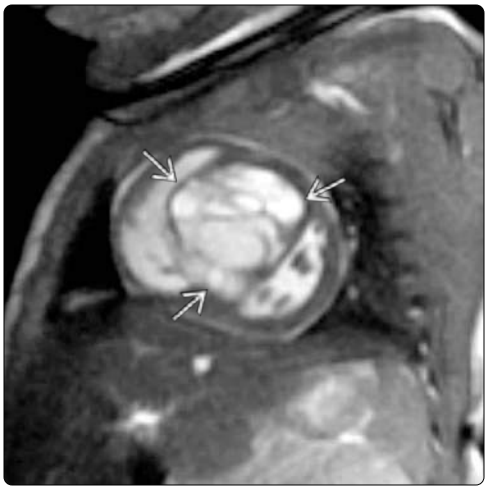


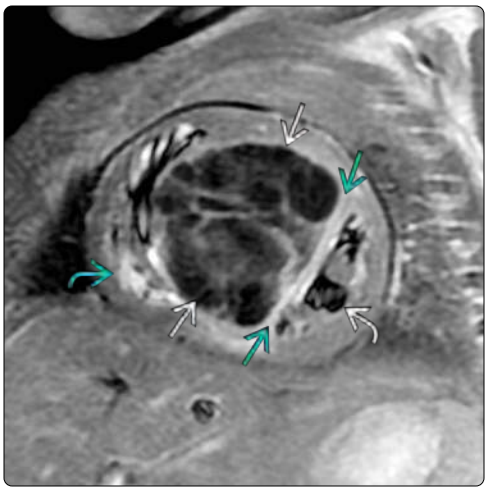


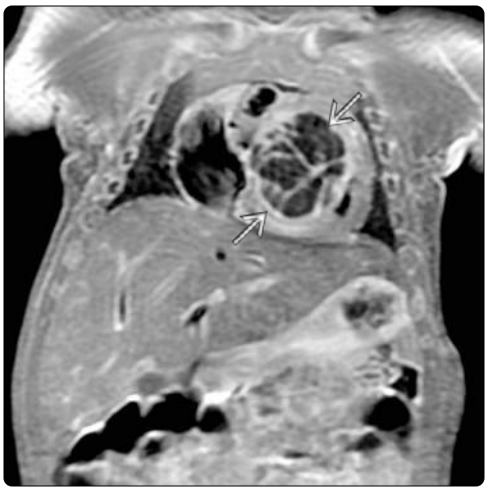
(Left) *Longitudinal oblique prenatal ultrasound of the chest & upper abdomen shows a heterogeneous chest mass with cystic & solid areas. There are signs of fetal distress with ascites.* **(Right)** *Coronal SSFP MR in the same fetus shows a mixed cystic & solid mass in the thorax. There are signs of fetal distress with ascites & skin edema. The heart is being compressed by the mass.*





(Left) *Axial SSFP MR in the same fetus shows the heterogeneous mass with a large pericardial effusion. These features strongly suggest a pericardial teratoma. Hydrops is again noted with body wall edema & a pleural effusion.* **(Right)** *Two-chamber view of the heart using a white blood MR technique shows a multiloculated mass in the interventricular septum of a young child. Germ cell tumors of the heart & pericardium occur less frequently than in the anterior mediastinum.*





(Left) *Two-chamber T1 C+ FS MR of the heart in the same patient shows a multicystic mass arising from the interventricular septum. The mass compresses both the right & left ventricles.* **(Right)** *Coronal T1 C+ FS MR of the chest in the same patient shows the multiloculated cystic mass of the interventricular septum. A teratoma was found upon resection.*

Neuroblastoma, Thoracic

KEY FACTS

TERMINOLOGY

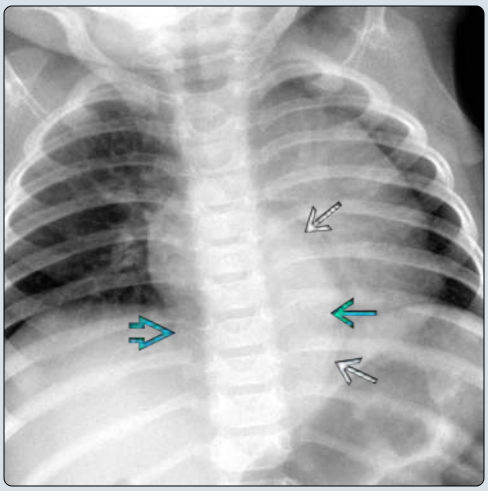
- Malignant tumor of primitive neural crest cells
- Continuous spectrum with more mature/benign counterparts: Ganglioneuroma & ganglioneuroblastoma

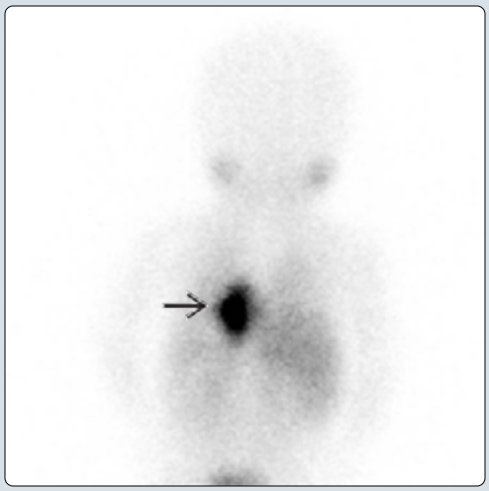
IMAGING

- Posterior mediastinal (paraspinal) mass with Ca^{2+}
  - Solid, elongated mass with paraspinal stripe widening ± rib splaying/erosion
- Tendency to invade spinal canal via neural foramen
- MR ± contrast best imaging modality to assess local tumor (due to its ability to evaluate neural foraminal extension)
- MIBG scan best for determining extent of disease

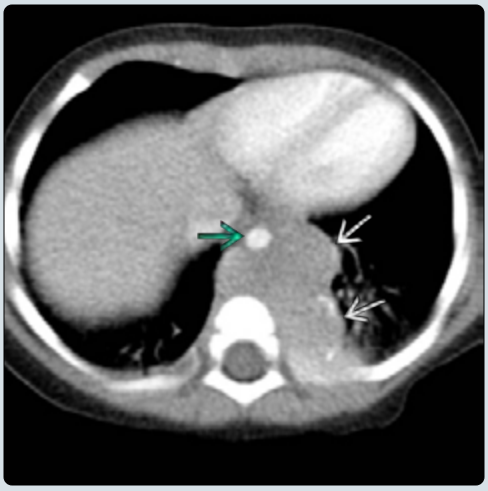
CLINICAL ISSUES

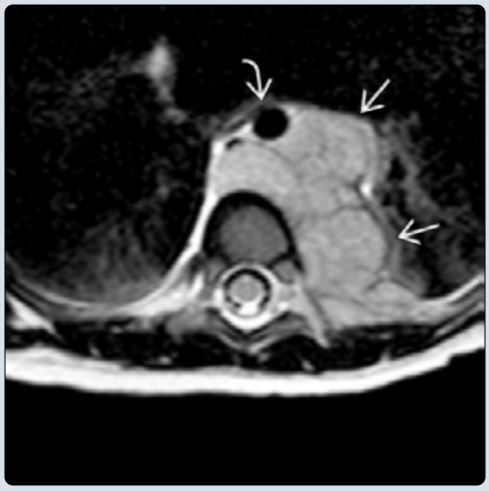
- 3rd most common pediatric malignancy after leukemia & central nervous system tumors
  - Most common extracranial solid tumor in children
- Posterior mediastinum: 3rd most common site for neuroblastoma (NBL) (15-20% of cases)
  - Many thoracic NBL asymptomatic & discovered incidentally
  - ± respiratory symptoms &/or neurologic symptoms (with cord compression)
- Mean age for thoracic presentation: 25 months
- 60-76% of patients with thoracic NBL have ↑ levels of urinary catecholamines (e.g., vanillylmandelic acid)
- Metastases most commonly to liver & bone (including marrow)
- Treatment options: Surgical resection, chemotherapy, radiation, stem cell transplant, I-131 MIBG therapy
  - Based on anatomic stage, age, & histologic features
  - Stage 4S/MS: High rates of spontaneous regression
- Overall 5-year survival: 78%
  - Thoracic NBL associated with higher survival rates than abdominal disease





(Left) *AP chest radiograph in a neonate shows a left paraspinal mass. There is mild splaying of the left 8th-9th ribs as compared to the right intercostal space at this level. When an elongated paraspinal mass is identified on a chest radiograph in a young child, it should be considered a neuroblastic tumor until proven otherwise.* **(Right)** *Posterior I-123 MIBG scan in the same patient shows intense uptake within the left paraspinal mass. MIBG is a sensitive & specific method of diagnosing neuroblastoma (NBL).*





(Left) *Axial CECT in the same patient shows an enhancing left paraspinal mass with faint peripheral CA^{2+}. Note how the mass lifts the aorta off of the spine, also typical of NBL. CA^{2+} are present in 85-90% of NBLs on CT.* **(Right)** *Axial T2 MR in the same patient shows a lobulated, intermediate signal intensity left paraspinal mass lifting the aorta off of the spine. The tumor surrounds 180° of the aorta but does not fully encase it. The tumor extends toward the neural foramen but does not invade the spinal canal.*

TERMINOLOGY

Abbreviations

- Neuroblastoma (NBL)

Definitions

- Malignant tumor of primitive neural crest cells

IMAGING

General Features

- Best diagnostic clue
  - Posterior mediastinal (paraspinal) mass with Ca^{2+}
  - Invasive mass that tends to encase & displace vessels
  - Often extends into spinal canal via neural foramina
- Location
  - Posterior mediastinum arising from sympathetic chain
    - Most commonly inferior; may be apical
- Morphology
  - Solid, elongated, or round paraspinal mass

Radiographic Findings

- Paraspinal soft tissue mass with widening of paraspinal stripe(s)
- Ca^{2+} within mass on up to 30% of radiographs
- Rib involvement common
  - Widening/splaying of intercostal spaces
  - Mild erosion/destruction of ribs
- Neural foraminal widening from intraspinal extension
- Pleural effusion may be present
- Bone metastases usually lucent, often subtle

CT Findings

- CECT
  - Posterior mediastinal mass, most commonly inferior
  - Mass often heterogeneous (from necrosis & hemorrhage)
  - Ca^{2+} seen on CT in 85-90%
  - ± local bone erosion or metastases

MR Findings

- T1WI
  - Intermediate to low-signal paraspinal mass
- T2WI FS/STIR
  - Intermediate to moderately high-signal paraspinal mass
  - Possible extension into neural foramen
    - Cord compression in up to 25%
  - Hyperintense bone marrow metastases
- DWI
  - May show restricted diffusion
- T1WI C+ FS
  - Variable enhancement, may be heterogeneous

Nuclear Medicine Findings

- Bone scan
  - Uptake in cortical bone metastasis &/or local bone invasion
  - Uptake in calcified primary mass (up to 74%)
- PET/CT
  - F-18 FDG useful in non-MIBG avid disease
- MIBG scintigraphy
  - Avid uptake related to catecholamine production
  - 88% sensitive & 98% specific in detection of NBL
    - ~ 10% of NBL not MIBG avid
  - Superior to PET/CT in depicting advanced disease & bone marrow metastases
  - Excellent for following therapy response

Imaging Recommendations

- Best imaging tool
  - MR ± contrast: Evaluates intraspinal extension & bone marrow metastases
  - MIBG for determining full extent of disease

DIFFERENTIAL DIAGNOSIS

Widening of Inferior Paravertebral Soft Tissues

- Normal paravertebral soft tissues
  - May be more prominent in obese & supine patients
  - Should not exceed adjacent pedicle in thickness or be inferolaterally oriented
- Thoracic discitis
  - Disc space narrowing, vertebral endplate irregularity
- Vertebral injury
  - Trauma & pain history usually clear
  - Vertebral body compression or malalignment

Posterior Mediastinal Mass in Child < 3 Years of Age

- Other neuroblastic tumors
  - Ganglioneuroma
    - Most common posterior mediastinal mass in adolescents & young adults
    - Benign; similar imaging features as NBL
    - May represent mature form of NBL
  - Ganglioneuroblastoma
    - Contains both mature & immature neuroblasts
    - May represent maturing form of NBL
- Round pneumonia
  - No evidence of rib erosion or intraspinal extension
  - ± air bronchograms, parapneumonic effusion
  - Short-term follow-up radiographs after antibiotics can help confirm round pneumonia (with expected interval clearance)
- Extralobar bronchopulmonary sequestration
  - May appear similar to NBL as paraspinal mass
  - Lung relationship & feeding vessel seen on CTA

PATHOLOGY

General Features

- Etiology
  - Malignant tumor of primitive neural crest cells
  - Continuous spectrum with more benign counterparts: Ganglioneuroma & ganglioneuroblastoma
  - Most commonly arises from adrenal gland but can arise anywhere along sympathetic chain
- Genetics
  - ↑ copies of *MYCN* (n-MYC) protooncogene associated with poor prognosis (*MYCN* amplification)
  - CD44: Glycoprotein on cell surface
    - ↑ levels: Better prognosis
- Associated abnormalities
  - Most neuroblastic tumors occur in isolation

Staging, Grading, & Classification

- International NBL Risk Group Staging System [pretherapy staging system using image-defined risk factors (IDRFs)]
  - L1: Localized tumor confined to 1 body compartment; not involving vital structures
  - L2: Localized tumor in 2 body compartments OR encasing/invading major structures
  - M: Distant metastases (except stage MS)
  - MS: Metastatic disease in children < 18 months of age with metastases confined to skin, liver, &/or bone marrow
  - IDRFs (thoracic) include
    - Encasement of aorta &/or major branches
    - Tumor compressing trachea &/or principal bronchi
    - Lower mediastinal tumor, infiltrating costovertebral junction between T9 & T12
    - Tumor extending into neck or abdomen
    - Intraspinal tumor extension so that > 1/3 of spinal canal in axial plane invaded
    - Infiltration of pericardium or diaphragm
- International NBL Staging System (older, surgically based staging system)
  - Stage 1: Localized tumor with gross total resection
  - Stage 2a: Localized tumor with incomplete gross total resection
  - Stage 2b: Localized tumor ± gross total resection & positive ipsilateral lymph nodes
  - Stage 3: Unresectable unilateral tumor crossing midline
    - Or localized unilateral tumor with positive contralateral lymph nodes
    - Or midline tumor with bilateral infiltration
  - Stage 4: Tumor with distant metastases
  - Stage 4S: Localized primary tumor; age < 1 year; metastatic disease confined to skin, liver, &/or bone marrow
- Bones (including marrow) in both staging systems must be clear by MIBG to qualify for stage 4S/MS (i.e., marrow disease is limited to < 10% involvement by biopsy)

Microscopic Features

- Immature, undifferentiated sympathetic cells: Small, round, blue cells
- Homer Wright rosettes: Circular groups of cells
- International NBL Pathology Classification (Shimada): Combines histiologic features & age of patient to define favorable & unfavorable histology for prognosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Many thoracic NBL asymptomatic, discovered incidentally
  - Respiratory symptoms, including cough
  - Neurologic symptoms with cord compression
  - 60-76% of patients with thoracic NBL have ↑ levels of urinary catecholamines (vanillylmandelic acid)
- Other signs/symptoms
  - Fever, chest pain, opsoclonus-myoclonus, Horner syndrome
  - Skin mets: Blueberry muffin syndrome

Demographics

- Age
  - Mean for thoracic NBL presentation: 25 months
  - Mean for all NBL: 15-17 months
- Gender
  - F:M = 1.4:1
    - Different from other sites of NBL where M > F
- Epidemiology
  - NBL: 3rd most common pediatric malignancy after leukemia & central nervous system tumors
    - Most common extracranial solid tumor in children
    - 7-10% of childhood cancers
    - 10-15% of childhood cancer deaths
  - Posterior mediastinum: 3rd most common site for NBL (15-20% of cases) after adrenal & extraadrenal retroperitoneum

Natural History & Prognosis

- Thoracic NBL associated with higher survival rates
  - Overall 5-year survival: 78%; 90% survival for stages 1, 2, 3; 29% for stage 4
- Features of NBL associated with better prognosis
  - Thoracic primary
  - Age at diagnosis < 18 months, histological grade (Shimada system), ↓ *MYCN* amplification (copies of gene), stage MS/4S
- Metastasizes most commonly to liver, bone, & bone marrow
- Some lesions may spontaneously regress/mature into less malignant tumors (ganglioneuroma)

Treatment

- Options: Surgical resection, chemotherapy, radiation, stem cell transplant, biologic agents, I-131 MIBG therapy
  - Based upon stage, age, & histologic features
  - Less aggressive lesions may be treated with surgical resection alone
  - For stage 4S/MS: Some institutions advocate no treatment due to rates of spontaneous regression

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Look for rib splaying/erosions & soft tissue Ca^{2+} when focal paraspinal mass is seen in child

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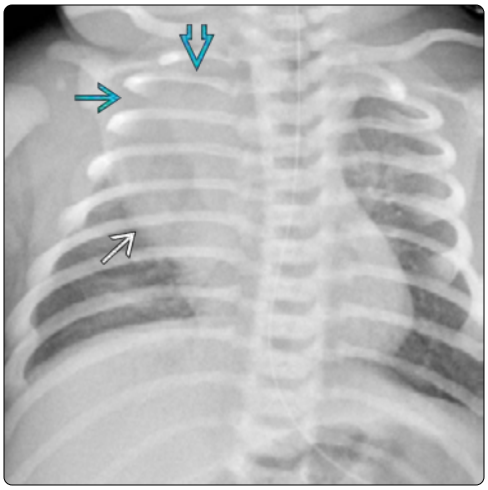
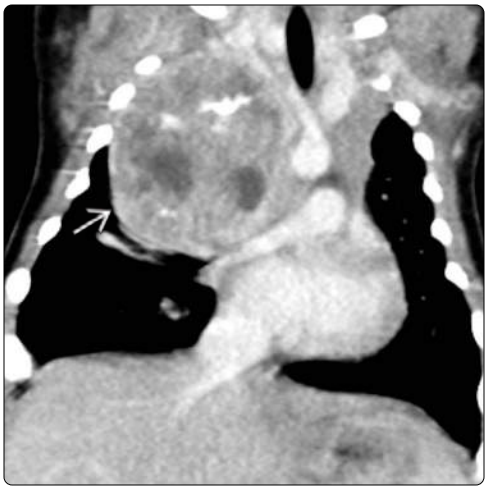
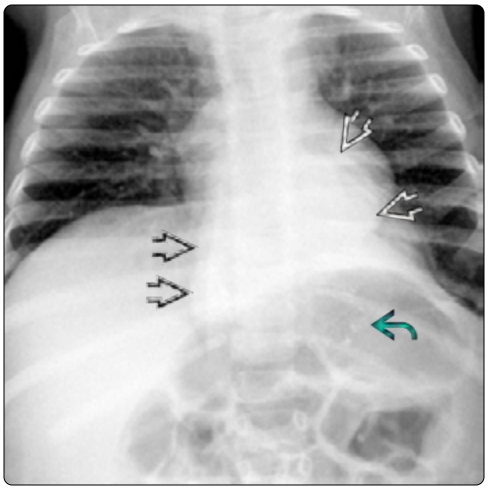
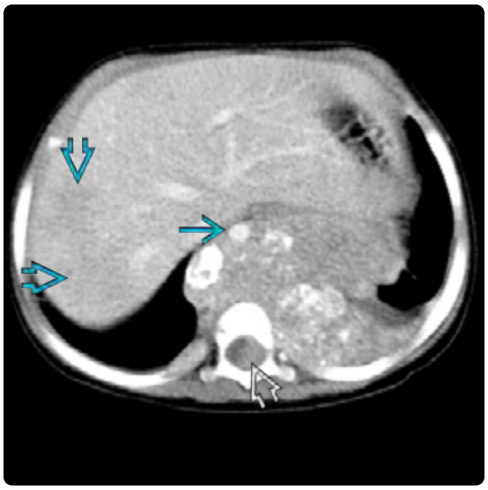


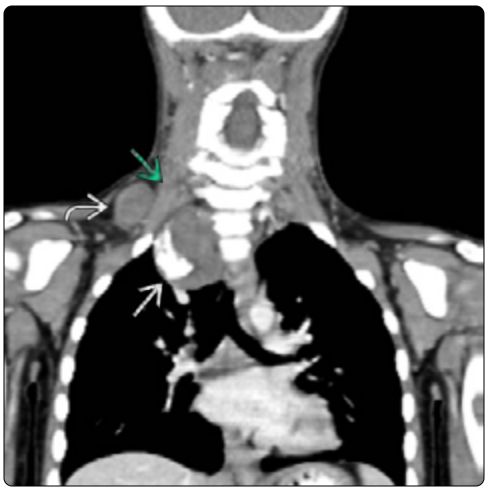


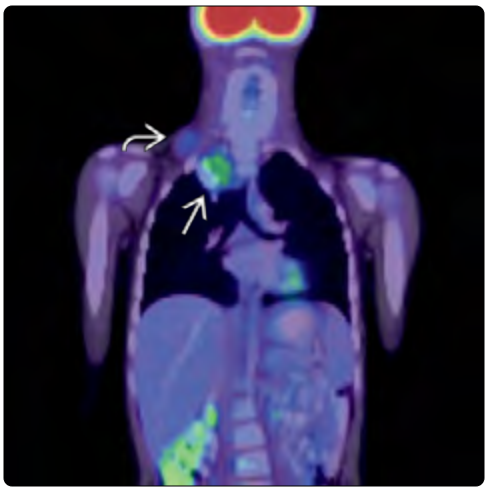
(Left) *AP chest radiograph in a neonate shows a large right apical mass. There is mild splaying & erosion of several right upper ribs.* **(Right)** *Coronal CECT in the same patient shows a right apical mass with internal heterogeneity & CA^{2+}, typical of NBL. In patients with NBL, it is important to look for any of the image-defined risk factors as their presence will change the stage of the tumor.*





(Left) *AP radiograph shows widening of the right paraspinal stripe & a retrocardiac mass. Speckled CA^{2+} are also seen in the abdominal left upper quadrant.* **(Right)** *Axial CECT of the same patient shows a large, heterogeneously calcified paravertebral NBL. The mass involves > 1/3 of the spinal canal diameter & lifts the aorta off of the spine, partially encasing it. Also note the poorly defined, hypoenhancing foci in the liver, consistent with distant metastases.*





(Left) *Coronal CECT of the chest in a 6 year old shows a partially calcified right apical/paraspinal NBL. The mass extends to the base of the neck, just below the brachial plexus. In addition, there is right supraclavicular adenopathy.* **(Right)** *Coronal PET/CT in the same patient shows FDG avidity in the right paraspinal mass & right supraclavicular lymph node. There is still controversy on when to best use PET/CT in NBL.*

Child Abuse, Rib Fractures

KEY FACTS

TERMINOLOGY

- Child abuse: Any act or failure to act by parent/caretaker that causes harm or imminent risk of harm to child

IMAGING

- Radiography
  - Linear lucency of acute fracture often not visible
  - Callus/subperiosteal new bone formation may become visible 7-10 days after injury
    - Ranges from indistinct margins & broadening of rib → sharply marginated nodular/bulbous callus
  - Rib head fracture may appear fragmented with mixed sclerosis & lucency; often no subperiosteal new bone
  - Arcuate density "ring shadow" comparable to "bucket handle" fracture at rib end/costochondral junction
- Tc-99m MDP bone scan or 18F-NaF PET complementary
  - Focal ↑ radiotracer activity within 24 hours
  - Costochondral junction fractures difficult to assess due to variable physiologic radiotracer uptake
- CT not advocated for identifying rib fractures but may be used to evaluate intrathoracic or intraabdominal injury
- Indications for initial radiographic skeletal survey
  - < 2 years old with suspicion of nonaccidental trauma
  - < 5 years old with suspicious fracture
  - Suspicion of NAT in any child unable to communicate
- Follow-up skeletal survey, typically after 2 weeks

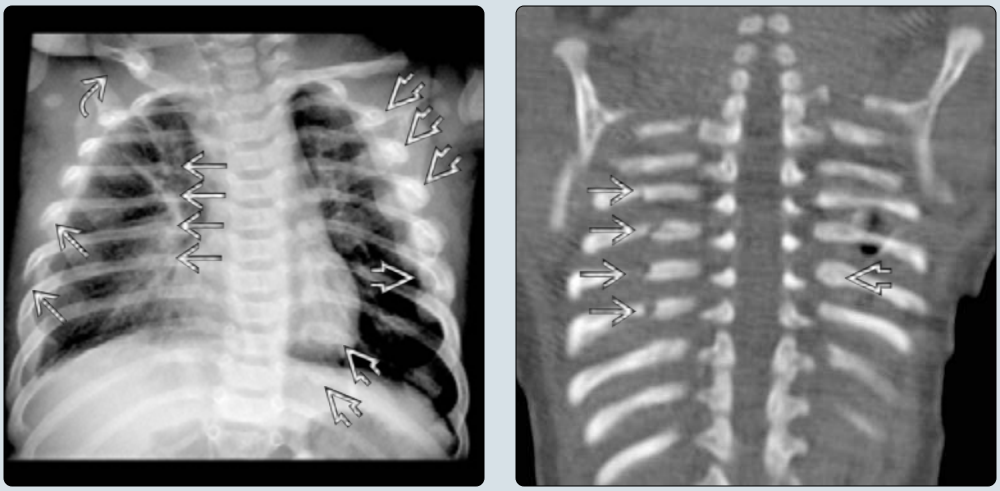
PATHOLOGY

- Posterior rib fractures most common & specific for NAT
  - Adult hands squeeze child's chest → leverage posterior ribs on transverse processes → fractures

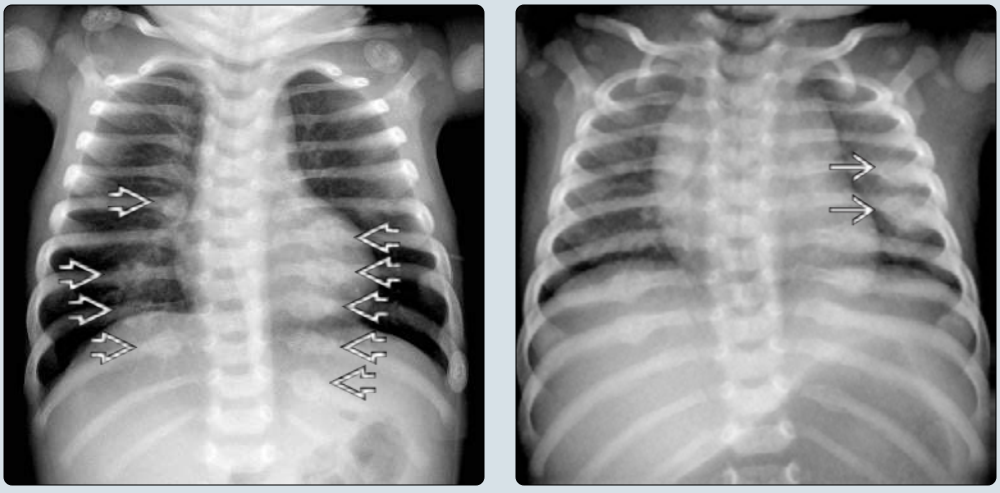
CLINICAL ISSUES

- Majority of rib fractures not suspected on clinical exam
  - Overlying bruising in ~ 9%
  - ± clicking or popping sounds with crepitus
- Likely most common skeletal injury in NAT
- Almost all rib fractures in children < 1 year old due to NAT

(Left) *AP radiograph in an infant with suspected abuse demonstrates multiple left lateral & posterior rib fractures with callus formation* ➡ *as well as more recent right-sided rib fractures without evidence of healing* ➡*. Also note the right clavicle fracture* ➡ *& right pleural effusion.* **(Right)** *Coronal CECT (in bone windows) in the same patient obtained shortly after the radiographs shows the acute right posterior rib fractures* ➡ *& one of the healing left posterior rib fractures* ➡*.*



(Left) *AP radiograph in this 1 month old shows multiple healing posterior rib fractures with bulbous callus formation* ➡*.* **(Right)** *AP radiograph obtained 2 weeks later in the same patient shows 2 additional healing rib fractures* ➡*, which, even in retrospect, are difficult to appreciate on the initial study.*



TERMINOLOGY

Abbreviations

- Nonaccidental trauma or injury (NAT, NAI), child abuse & neglect

Definitions

- Child abuse: Any act or failure to act by parent/caretaker that causes harm or imminent risk of harm to child

IMAGING

General Features

- Best diagnostic clue
  - Bulbous callus/subperiosteal new bone formation of multiple adjacent posteromedial ribs in infant
- Location
  - Can occur anywhere along rib
  - Posterior near costovertebral junction most common

Radiographic Findings

- Acute fractures often not visualized radiographically
  - Typically vertical/oblique linear lucency in rib short axis
  - Difficult to visualize if nondisplaced & fracture plane oriented coronal relative to x-ray beam
- Healing fractures may become visible 7-10 days after injury
  - Callus/subperiosteal new bone formation: Ranges from indistinct margins & broadening of rib → sharply marginated nodular/bulbous callus
  - "Hole-in-rib": Radiolucency (bone resorption) surrounded by sclerosis
  - Rib head: Often without subperiosteal new bone
    - May appear fragmented with mixed sclerosis & lucency
  - Rib end/costochondral junction: Arcuate density "ring shadow" comparable to "bucket handle," ± round callus
- Sensitivity 73% in 1 study vs. follow-up radiographs
- Sensitivity as low as 26% in 1 study vs. postmortem histology

CT Findings

- Performed for suspected thoracoabdominal injury
- Not advocated for identifying rib fractures alone due to radiation exposure
- Greater sensitivity than initial skeletal survey for detection of rib fractures
- Findings similar to radiography
  - Linear lucency, often in rib short axis
  - Callus & subperiosteal new bone formation

MR Findings

- Whole-body MR investigated for evaluating child abuse
- Low sensitivity (57%) for rib fractures vs. radiographs
- May see fluid signal changes in bone marrow & soft tissues ± elevation of periosteum before fracture evident on radiographs

Ultrasonographic Findings

- Adult studies show ↑ sensitivity of US over radiography for rib fractures, particularly of cartilaginous portion
- May show occult rib fracture in children when initial skeletal survey negative or equivocal, though patient cooperation may be issue
- Imaging findings include discontinuity of echogenic shadowing cortex, acoustic edge shadow at margin of fracture, local hematoma with elevated periosteum

Nuclear Medicine Findings

- Tc-99m MDP skeletal scintigraphy or 18F-NaF PET complementary to initial skeletal survey
  - Focal ↑ in radionuclide activity within 24 hours, normalizes within 6 months
  - Costochondral junction fractures (metaphyseal equivalent in anterior rib) difficult to assess due to variable physiologic uptake
- Compared to initial skeletal survey, 18F-NaF PET shows greater sensitivity for rib fractures but lower sensitivity for classic metaphyseal lesions
- Detection of bony injury by Tc-99m MDP bone scan & initial skeletal survey from 1 study demonstrated
  - 60 rib fractures: 63% identified on bone scan & 73% identified on skeletal survey
  - 6 cases with solitary rib fractures: 50% identified on both modalities, remaining 50% identified only on bone scan
  - Other fractures: 33% seen on both modalities, 44% seen on skeletal survey only, 25% seen on bone scan only

Imaging Recommendations

- Initial skeletal survey
  - Indications
    - < 2 years old with suspicion of NAT
    - < 5 years old with suspicious fracture
    - Concern for NAT in any child unable to communicate
  - Images include: AP & lateral skull, lateral cervical & lumbar spine, AP & lateral & both obliques of thorax, AP pelvis, AP humeri, AP forearms, PA hands, AP femurs, AP lower legs, AP feet; additional views based on clinical & imaging findings
- Follow-up skeletal survey
  - Typically 2 weeks (not < 10 days) from initial evaluation
  - Indications
    - Concerning fractures on initial study
    - Normal initial study with persistent suspicion based on clinical or imaging findings
  - Used to confirm suspected fractures & identify additional fractures
  - In 1 study, clarified questionable fractures or identified new fractures in 48% of cases
    - Most additional fractures were of ribs
- Tc-99m MDP bone scan or 18F-NaF PET used as complementary & problem-solving tools
- CECT for suspected intrathoracic or intraabdominal injury

DIFFERENTIAL DIAGNOSIS

Entities Associated With Multiple Fractures

- Osteogenesis imperfecta (OI)
  - Type IV OI most commonly mistaken for abuse
    - Sclera normal in color (not blue)
  - May see osteoporosis, multiple fractures, & wormian bones
- Menkes syndrome
  - Osteoporosis, metaphyseal spurs, brittle hair, tortuous intracranial vessels
  - Excessive wormian bone formation

- Rickets
  - Widening/lengthening of physes with loss of normal zones of provisional CA^{2+} + metaphyseal fraying, cupping, & splaying
- Leukemia
  - Osteoporosis ± lucent metaphyseal bands, permeative destruction, aggressive periosteal new bone formation

Birth Trauma

- Rare but can occur with large babies & difficult deliveries
- No rib fractures in one study of 34,946 births

Trauma From Cardiopulmonary Resuscitation

- Rib fractures very rare in pediatric CPR (< 1%)
- Most such fractures involve anterior ribs

Accidental Trauma

- Age & history must be consistent with injury
- Rib fractures more likely to be anterior & lateral
- Greater likelihood of intrathoracic injury & fewer fractures

PATHOLOGY

General Features

- Assailant holds infant, wrapping hands around chest with finger tips at posterior ribs & thumbs situated anteriorly extending to midline
- Anteroposterior compression while squeezing & shaking child results in fractures
  - Posterior rib fractures occur from leveraging posterior ribs on transverse processes
    - Most common location & most specific for abuse

CLINICAL ISSUES

Presentation

- Wide range of clinical presentations for NAT: Injury inconsistent with history, multiple injuries in various stages of healing, bruising in nonmobile infant, genitalia injury, cigarette burns, other injuries with high specificity for NAT (e.g., classic metaphyseal lesion/corner fracture)
- Rib fractures
  - Only radiographic finding of NAT in up to 29% of cases
  - Majority not suspected based on clinical exam
    - Associated bruising at fracture site is rare, 9%
  - "Clicking" or "popping" sound from back or chest on physical exam, ± crepitus
    - Consider follow-up radiographs or scintigraphy if initial radiographs negative
  - Associated intrathoracic injury less common than in accidental injury
    - 12.8% of NAT vs. 55.6% of accidental injuries
    - Difference relates to injury mechanism
  - Rib fractures in children < 3 years of age usually associated with abuse (61%)
  - Up to 82% of rib fractures in children < 1 year of age from NAT

Demographics

- Epidemiology
  - Estimated 702,000 child maltreatment victims in 2014 in USA
    - Infants ≤ 1 year old most at risk, account for 36.7%
    - Physical abuse in 41% of child maltreatment cases
  - Rib fractures most common skeletal injury in NAT
    - In 1 study of abused children, 79% of all fractures involved rib cage
    - Rib fractures present in 10-14% of skeletal surveys performed for suspected abuse

Natural History & Prognosis

- Estimated 1,580 fatalities from child maltreatment in 2014
  - Mortality highest in infants < 1 year old, 17.96/100,000 same-aged children in population
  - 71% of all deaths occurred in children < 3 years old
  - 79% of fatalities involved parent as perpetrator

Treatment

- Multidisciplinary investigation of maltreatment allegation involves physicians, social worker, Child Protective Services, legal authorities
- Ensure "at risk" child & siblings placed in safe environment
  - May involve removal of child from home with temporary/protective custody
    - 23% of maltreatment victims placed in foster care

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Carefully review visible ribs on any modality in young child, regardless of study indication
  - Fractures may be incidentally noted on unrelated exams

Reporting Tips

- Concern for child abuse based on imaging findings must be conveyed to referring clinician ASAP
- Final report represents legal document: Review carefully

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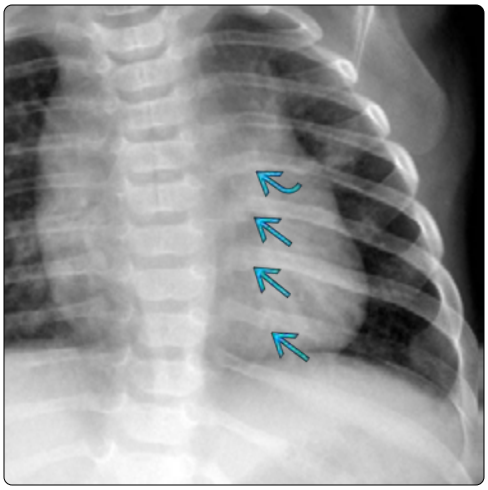
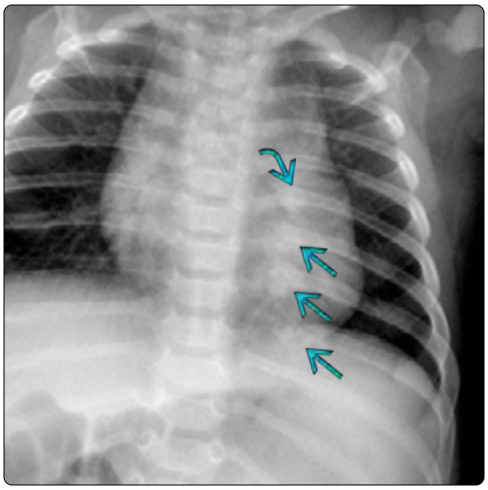
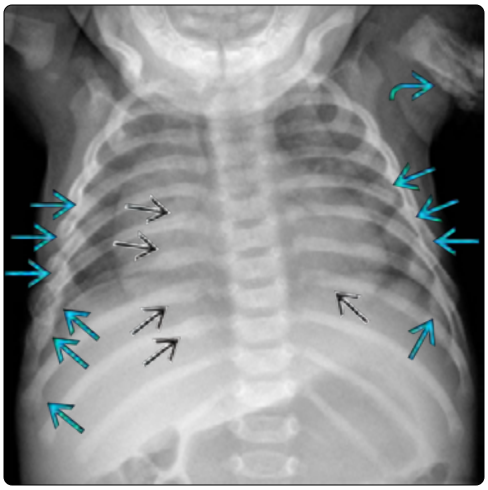
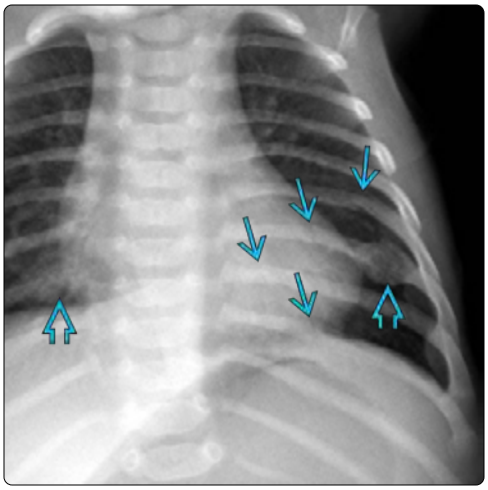


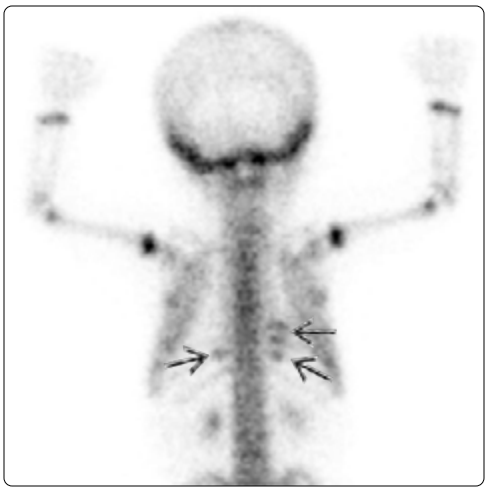


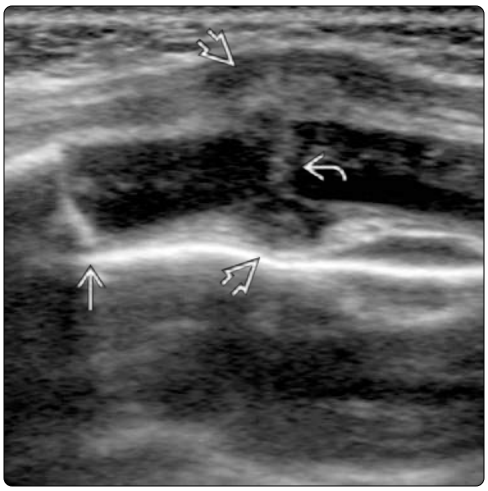
(Left) *AP chest radiograph from a 4 month old with "popping" sounds from his left back shows acute left 7th, 8th, & 9th posterior rib fractures. A 6th rib fracture is also suspected. (Right) AP chest radiograph from a follow-up skeletal survey of the same patient shows healing of the 7th, 8th, & 9th rib fractures. The suspected 6th rib fracture was also confirmed by visualization of callus formation. The provided history was incompatible with the injuries.*





(Left) *AP chest radiograph from an 8 week old with concern for nonaccidental trauma demonstrates multiple healing anterior & lateral rib fractures. Also present are healing posterior rib fractures with subtle rib broadening with callus. A healing fracture of the left humerus is partially visualized. (Right) AP chest radiograph in a vomiting 7 week old with palpable "clicking" from her back shows acute & healing posterior rib fractures. Multiple fractures of varying ages are highly concerning for nonaccidental trauma.*





(Left) *Anterior nuclear medicine bone scan in an abused infant shows multiple posterior rib fractures.* **(Right)** *Transverse ultrasound demonstrates the anterior edge of an ossified rib with a fracture extending through the anterior cartilaginous rib with adjacent hematoma.*

Lung Contusion and Laceration

KEY FACTS

TERMINOLOGY

- Lung contusion: Hemorrhage + edema in alveoli & interstitium due to traumatic alveolar capillary damage
- Lung laceration: Frank tear of lung parenchyma

IMAGING

- Radiograph: Often sufficient for blunt chest trauma evaluation
  - Contusion: Nonsegmental patchy or diffuse opacification
    - May not be radiographically evident < 4-6 hours
  - Laceration: Round lucency ± air-fluid level
    - May be obscured by contusion initially
- CECT: Performed if strong clinical or imaging findings suggest significant thoracic injury
  - ↑ sensitivity for detecting contusion & ↑ accuracy for assessing extent compared to radiographs
    - Significance of contusions seen only on CT doubtful → may not be associated with ↑ morbidity
  - Contusion: Confluent/nodular, crescentic/amorphous consolidative or ground-glass opacities
    - Commonly peripheral with rim of subpleural sparing
  - Laceration: Air-/fluid-filled cavity surrounded by opacity

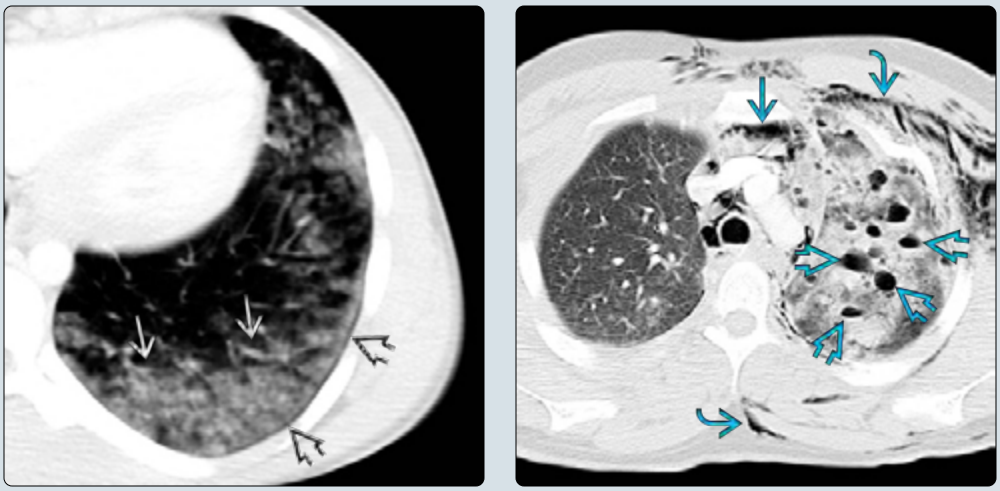
PATHOLOGY

- Associated chest injuries
  - Rib fractures less common than adults
  - Heart & great vessels (8%), diaphragm (< 5%), tracheobronchial tree (< 3%), esophagus (< 0.1%)

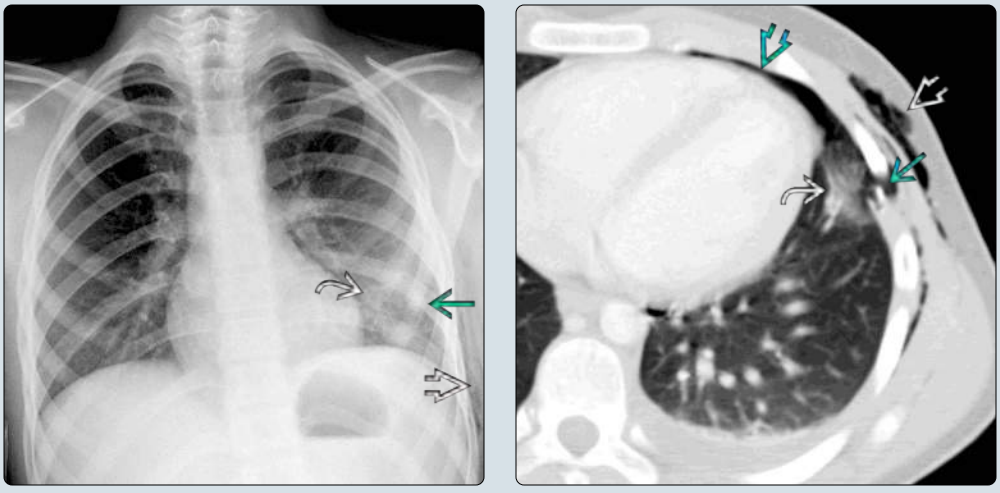
CLINICAL ISSUES

- Patients with lung injuries typically have ↑ overall injury severity with multisystem trauma
  - Other systems account for high morbidity & mortality
- Consider pneumonia or acute respiratory distress syndrome (ARDS) if worsening clinical & imaging findings > 48 hours
- Supportive treatment, including pulmonary toilet, pain control, noninvasive positive pressure ventilation, mechanical ventilation

(Left) *Axial CECT in a trauma patient shows amorphous density in the left lower lobe. Note the thin crescentic lucency adjacent to the chest wall. This finding of subpleural sparing is frequently seen in pulmonary contusions.* **(Right)** *Axial CECT of a 17 year old after a car accident shows multiple air- & fluid-filled cysts throughout the left upper lobe, consistent with lung lacerations. Also note the pneumomediastinum & extensive subcutaneous emphysema.*



(Left) *AP chest radiograph of a 13 year old after a bicycle collision shows an anterior left 5th rib fracture with adjacent subcutaneous emphysema & patchy airspace opacity consistent with contusion.* **(Right)** *Axial CECT in the same patient shows the left 5th rib fracture, subcutaneous emphysema, a pneumothorax, & adjacent ground-glass lung opacity consistent with contusion. Associated rib fractures are less frequent in pediatric patients (as compared to adults) due to the pliability of the rib cage.*



TERMINOLOGY

Definitions

- Lung contusion: Blunt chest trauma → alveolar capillary damage → hemorrhage + edema in alveoli & interstitium
- Lung laceration: Chest trauma (blunt or penetrating) → frank tear of lung parenchyma

IMAGING

General Features

- Best diagnostic clue
  - Crescentic or amorphous nonsegmental pulmonary opacities of mixed confluent & nodular quality
    - Subpleural sparing of overlying lung periphery
- Location
  - Posterior lung most common

Radiographic Findings

- Radiography
  - Contusions
    - Irregular patchy regions of poorly defined airspace consolidation (mild) vs. diffuse homogeneous consolidation (severe)
      - Due to hemorrhage & edema in alveoli & peribronchovascular interstitium
    - Located at impaction or contrecoup lung injury sites
      - Adjacent to ribs & vertebral bodies
    - Present in 30-50% of initial chest radiographs for blunt trauma
      - Radiographs underestimate volume of contusion
    - May not be evident in first 4-6 hours after trauma
      - Radiographs still negative > 6 hours in up to 33%
    - Complete clearing within 7-10 days unless other cause of opacification develops [pneumonia, acute respiratory distress syndrome (ARDS)]
  - Lacerations
    - Thin-walled, air-filled cysts of variable size (pneumatoceles) ± air-fluid levels
      - Single/multiple, oval/spherical, uni/multilocular
      - May fill with hematoma & subsequently expand (uncommon)
      - May be obscured by contusion initially, appearing hours or days after trauma
    - Occur at maximum impact or contrecoup injury sites
    - Persist up to 4 months; gradually ↓ in size by 1-2 cm/week
  - Bronchial injury
    - Very rare
    - Persistent pneumothorax despite chest tube
    - Fallen lung sign: Lung falls away from hilum

CT Findings

- Contusions
  - Mixed confluent & nodular lung opacities (70%)
    - Crescentic (50%) or amorphous (45%) in shape
    - Posterior location (75%); lower lobes most common
  - Subpleural sparing in up to 95% of cases
    - Thin 1-2 mm crescentic rim of uniformly nonopacified subpleural lung
      - Separates lung opacity from adjacent chest wall
    - Less likely in larger contusions
  - CT more sensitive than radiographs for contusion & more accurately determines contused volume
    - Up to 69% of contusions underestimated (24%) or not identified (45%) on initial radiograph compared to CT
- Lacerations
  - Air- or fluid-filled cavity/cavities surrounded by parenchymal contusion
    - Due to frank tear of parenchyma (worse injury)

Ultrasonographic Findings

- High sensitivity (> 90%) & specificity (> 90%) in adults with blunt chest trauma when 2 sonographic features present
  - Multiple B lines (comet-tail artifacts) arising from pleural line
    - Represents interstitial edema 1-2 hours after injury
    - Other processes with alveolar/interstitial involvement can appear similar (cardiogenic pulmonary edema, ARDS)
  - Peripheral parenchymal lesion with C lines, confluent consolidations (hepatization), or parenchymal disruption with effusion
    - Represents hemorrhage & debris in alveoli

Imaging Recommendations

- Chest radiographs usually sufficient for evaluation of blunt trauma
- CT performed in patients with strong clinical or imaging findings of significant thoracic injury
- Differentiation of contusions from other lung opacities in blunt chest trauma has relevance to child's prognosis

DIFFERENTIAL DIAGNOSIS

Aspiration

- Segmental distribution of opacities (as aspirated materials reach lungs via airways)
  - Posterior lower lobes most common
- Absence of subpleural sparing
- Findings develop in 24-48 hours

Pneumonia

- Focal or multifocal airspace opacification
- Absence of subpleural sparing
- If contusion worsens after 48 hours, consider superinfection

Atelectasis

- Triangular shape with segmental distribution
- Often dependent with lack of subpleural sparing
- Signs of volume loss

Acute Respiratory Distress Syndrome

- Extensive bilateral ground-glass opacities & consolidation
- Diffuse alveolar damage in setting of pulmonary or nonpulmonary injury

Neurogenic Pulmonary Edema

- Symmetric confluent airspace opacities; normal heart size

PATHOLOGY

General Features

- Etiology

- Blunt trauma: 85%-90% of thoracic injuries
  - Most commonly motor vehicle-related
  - Pedestrian vs. vehicle 36.5%, motor vehicle collision 31.7%, assault 11.5%, falls 9%
- Penetrating trauma: 10%-15% of thoracic injuries
  - Gunshot wound, stab wound, puncture from adjacent fractured rib
- Nonaccidental trauma (NAT)
  - Thoracic injury may be more common in nonaccidental than accidental trauma cases (17% vs. 6%)

- Associated abnormalities
  - With major chest trauma in child
    - Rib fractures less common as compared to adults
      - Due to elasticity of pediatric rib cage
      - Still present in 20-62% of cases
    - Uncommon injuries: Heart & great vessels (8%), diaphragm (1-5%), tracheobronchial tree (0.7-2.8%), esophagus (< 0.1%)
- Mechanism of injury
  - ↑ pliability of anterior pediatric chest wall + contrecoup forces of rapid deceleration → compression of relatively fixed posterior lung against ribs & vertebral column
  - Dispersion of forces along least mobile lung regions explains posterior location, crescentic shape, & nonsegmental distribution
  - Subpleural sparing may be due to
    - Terminal arterial branches ending prior to subpleural lung, possibly protecting this region from hemorrhage
      - Less common sign in larger contusions, possibly due to extension of hemorrhage into subpleural lung
    - Compression of subpleural lung against chest wall may "squeeze" extravasated blood & edema from periphery
  - Systemic inflammatory response: Secondary ↓ production of surfactant by injured alveolar tissue may lead to ARDS
  - Lung laceration
    - Frank tear of lung parenchyma results in cavity that fills with hemorrhage &/or air

Gross Pathologic & Surgical Features

- American Association for Surgery of Trauma Injury Scale
  - Grade I: Unilateral contusion < 1 lobe
  - Grade II: Unilateral contusion of single lobe or laceration resulting in simple pneumothorax
  - Grade III: Unilateral contusion > 1 lobe, laceration with persistent (> 72 hours) air leak from distal airway, or nonexpanding intraparenchymal hematoma
  - Grade IV: Major segmental/lobar laceration with air leak, expanding intraparenchymal hematoma, or primary branch intrapulmonary vessel disruption
  - Grade V: Hilar vessel disruption
  - Grade VI: Total uncontained transection of pulmonary hilar vessel

CLINICAL ISSUES

Presentation

- ↓ breath sounds, dullness to percussion, tachypnea, chest tenderness, hemoptysis (rarely)
- May present with respiratory failure in large contusions
  - Hypercarbia, hypoxia, acidosis

Natural History & Prognosis

- Children with thoracic trauma typically more severely injured overall than those without thoracic involvement
  - Morbidity & mortality in patients with lung contusion/laceration usually related to severity of multisystem trauma
    - Mortality rate of pediatric patients with lung contusion & laceration up to 34% & 43%, respectively
    - Presence of thoracic injury ↑ NAT mortality from 15% to 50%
  - 20-37% of children with pulmonary contusion require mechanical ventilation
    - Radiographic severity of lung contusion correlates with oxygenation impairment, carbon dioxide exchange, & duration of ventilatory support
      - Radiographically occult contusions seen by CT: No ↑ in patient morbidity
    - Contusion volume > 28% on CT predictive of need for mechanical ventilation
      - Study including adult & pediatric patients; not reproduced in solely pediatric population
    - Contusion volume > 20% on CT predictive of ARDS development in adults; not reproduced in children
- Complications: Pneumonia 20%, ARDS 5-20%

Treatment

- Supportive therapy: Pulmonary toilet, pain control, alveolar recruitment maneuvers, noninvasive positive pressure ventilation, mechanical ventilation, rarely ECMO
- Surveillance for other major organ injuries
- Observation for complications: Infection, ARDS, tension pneumothorax, hemopneumothorax, hemoptysis
- No evidence to support prophylactic broad spectrum antibiotics, but often used
- No proven role for corticosteroids

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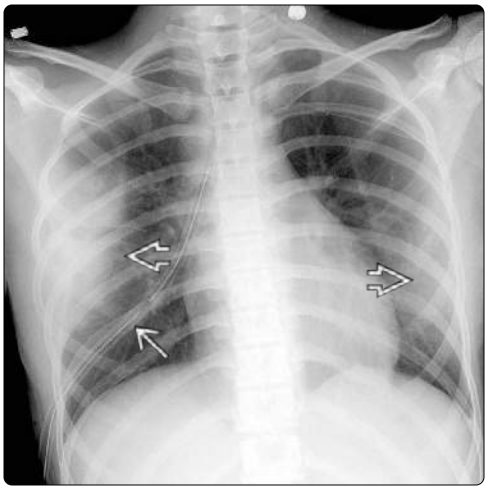
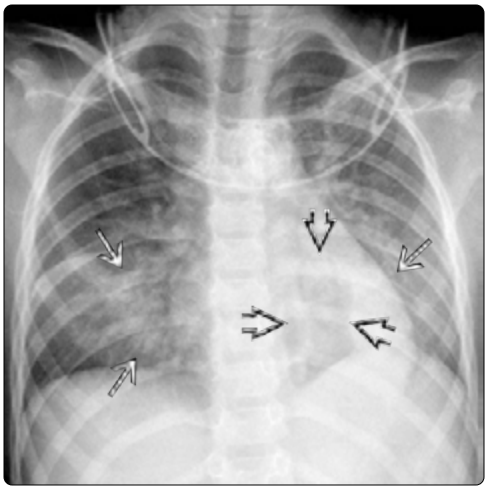
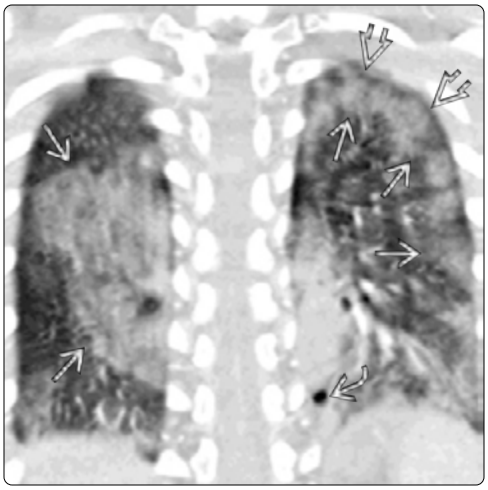
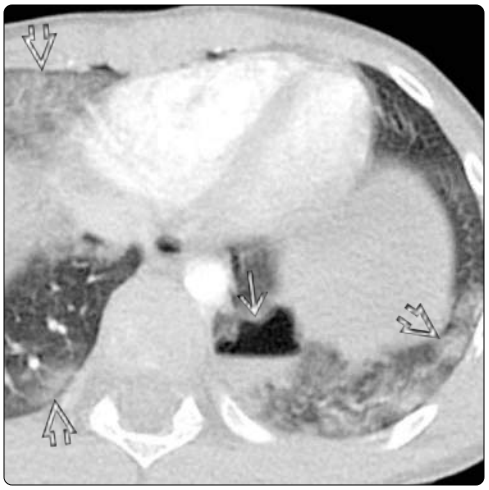


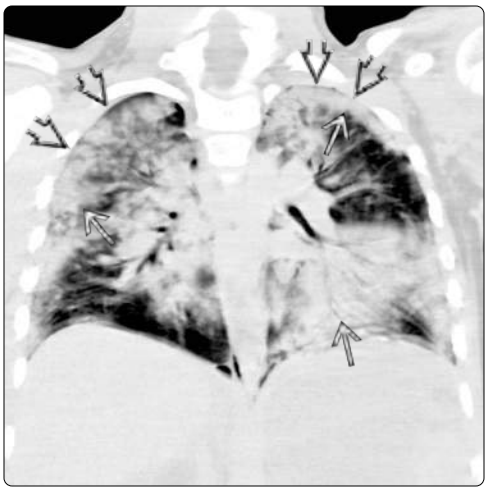


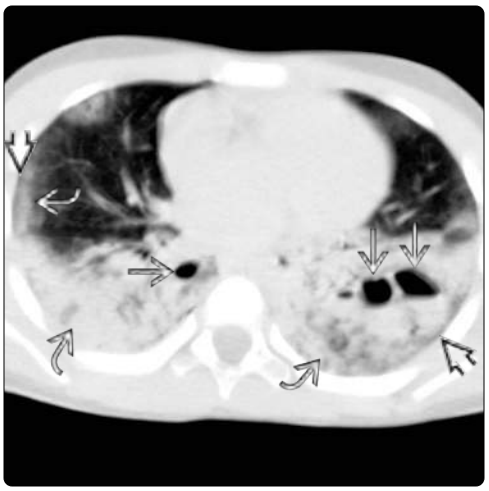
(Left) *AP radiograph in a trauma patient shows increased density in the lateral aspects of both the right & left lungs, a nonspecific finding that is consistent with contusions. There is a right-sided chest tube in place.* **(Right)** *Frontal radiograph of an 8 year old after an all-terrain vehicle accident shows confluent right middle & bilateral lower lobe opacities. The left lower lobe lucency is suspicious for a pneumatocele from a laceration.*





(Left) *Coronal CECT in the same patient shows nonsegmental opacities with subpleural sparing in the lungs bilaterally. A small round lucent focus in the left lower lobe is consistent with a laceration.* **(Right)** *Axial CECT in the same patient shows a posttraumatic pneumatocele with an air-fluid level in the left lower lobe. Scattered patchy & ground-glass opacities are also present in the right middle & bilateral lower lobes, consistent with contusions.*





(Left) *Coronal CECT in a trauma patient shows bilateral, primarily peripheral amorphous opacities that do not respect lobar anatomy, consistent with lung contusions. Note the subpleural parenchymal sparing.* **(Right)** *Axial CECT in the same patient shows bilateral, primarily peripheral amorphous densities that do not respect lobar anatomy, consistent with lung contusions. There is associated subpleural sparing. The multiple fluid-containing cysts are typical of lacerations.*

Aortic Injury

KEY FACTS

TERMINOLOGY

- Blunt aortic injury (BAI) may manifest as intramural hematoma, intimal tear, larger intimal flap, contained pseudoaneurysm, or frank aortic rupture

IMAGING

- Chest radiograph: Sensitive, not specific; high NPV (> 90%)
  - Most common findings: Indistinctness of aorta, prominent aortic knob, left apical cap
- CTA: Sensitivity & specificity 90-100%; NPV nearly 100%
  - Modality of choice: Fast, accessible, accurate; has largely replaced conventional aortography
  - Most common finding: Pseudoaneurysm, followed by intimal tear; usually near aortic isthmus

TOP DIFFERENTIAL DIAGNOSES

- Normal thymus
- CT artifacts
- Ductus diverticulum

PATHOLOGY

- Majority of pediatric BAIs due to rapid & forceful deceleration injuries (motor vehicle crashes > falls, ATV crashes, & bicyclist/pedestrian versus automobile collisions)

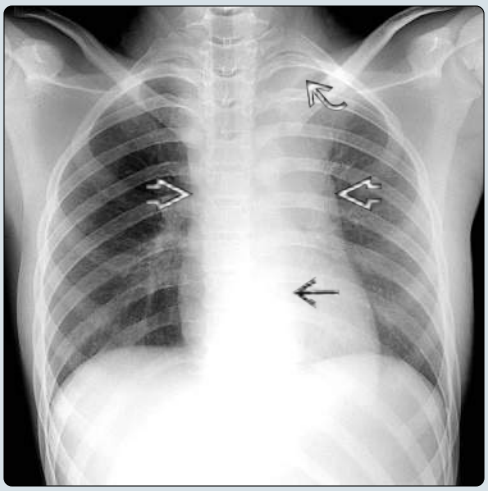
CLINICAL ISSUES

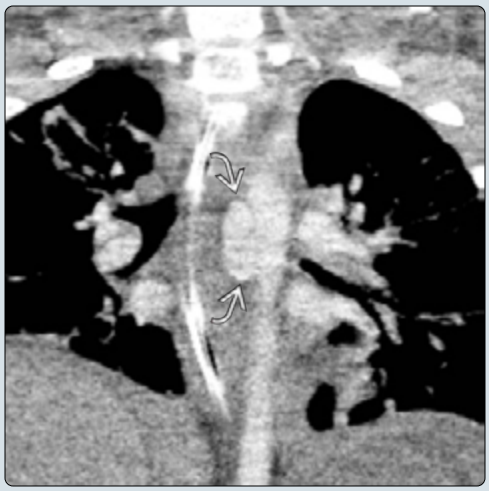
- Significant change in prognosis/treatment depending on presence or absence of aortic external contour alteration
  - Absent (intimal tears & flaps): Mortality 5-15% with treatment
  - Present: Mortality 25% for pseudoaneurysm & up to 90% for aortic rupture
- Delayed endovascular treatment becoming more common; blood pressure control essential in interval

DIAGNOSTIC CHECKLIST

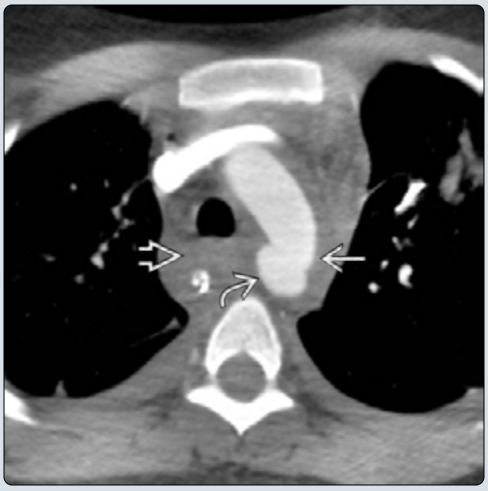
- Look for BAI in all cases of blunt, high-energy chest trauma
- Up to 50% of pediatric BAIs lack external signs of chest trauma & have obvious CNS, abdominal, or skeletal injuries

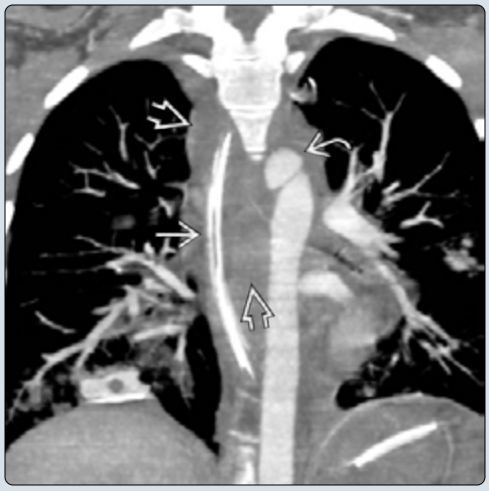
(Left) *AP chest radiograph in a 13-year-old girl involved in a motor vehicle crash (MVC) shows a left apical cap, a common radiographic finding in children with blunt aortic injury (BAI). Also note the widened mediastinum & prominent paraspinal soft tissue density behind the heart.* **(Right)** *Coronal CTA from a teenager ejected during a MVC shows a well-defined collection of extraluminal contrast-enhanced blood within a large aortic pseudoaneurysm just below the level of the ligamentum arteriosum.*





(Left) *Axial CTA in a teenager who was in a MVC shows an abnormal external contour of the aorta caused by a pseudoaneurysm near the aortic isthmus. Also note the extensive mediastinal hematoma.* **(Right)** *Coronal CTA in the same patient again shows the pseudoaneurysm as a focal, rounded contour alteration of the aorta. Note how the large mediastinal hematoma causes rightward deviation of the nasogastric tube.*





TERMINOLOGY

Abbreviations

- Traumatic aortic injury (TAI), blunt aortic injury (BAI)

Definitions

- Intramural hematoma: Isolated circumferential or crescentic thickening of aortic wall without intimal injury
- Intimal tear: Small, isolated injury to intimal layer of aorta without external aortic contour abnormality; linear intimal defect &/or associated thrombus < 10 mm
- Large intimal flap: Larger intimal injury than tear but still without external aortic contour abnormality; linear intimal defect &/or associated thrombus > 10 mm
- Pseudoaneurysm: Focally contained outpouching of aortic lumen causing external aortic contour abnormality
- Rupture: Uncontained dissection causing external aortic contour abnormality with extraluminal contrast (extravasation)

IMAGING

General Features

- Best diagnostic clue
  - Portable chest radiograph: Indistinctness of aorta or prominence of aortic knob + left apical cap
  - CTA: External aortic contour abnormality with mediastinal hematoma ± active extravasation
- Location
  - Most commonly (95%) near aortic isthmus between left subclavian & 1st intercostal arteries
  - Far less common at aortic root & diaphragmatic hiatus
  - Multiple tears seen in small number of patients
  - May occur in abdominal aorta in seat belt injury

Radiographic Findings

- Chest radiograph sensitive but not specific; low positive predictive value (< 20%), high negative predictive value (> 90%)
- Chest radiograph may be normal with only small mediastinal hematoma
- Radiographic signs
  - Left apical cap or left hemothorax
  - Obscured outline of descending aorta
  - Obliteration of aortopulmonary window
  - Widened mediastinum (which may be mimicked by normally prominent thymus in younger children)
  - Wide paraspinal stripe
  - Rightward shift of endotracheal &/or gastric tube(s)
  - Rightward shift of trachea
  - Depressed left main bronchus
  - Fractures of 1st &/or 2nd rib(s) &/or scapula

CT Findings

- NECT
  - Low sensitivity & specificity
  - High attenuation in aortic wall: Intramural hematoma
  - Obliteration/↑ attenuation of mediastinal fat suggests hemorrhage
  - Hematoma displacing trachea & esophagus to right
  - Hemopericardium
- CTA
  - Modality of choice: Rapid, accessible, high detail of acute thoracoabdominal injuries
    - Has largely replaced conventional aortography
  - Sensitivity 90-100%, specificity 90-100%, negative predictive value nearly 100%
  - Direct signs
    - Circumferential or crescentic thickening of aortic wall due to intramural hematoma
    - Linear filling defect (intimal flap) due to dissection
    - Intraluminal thrombus
    - Well-defined external contour abnormality of pseudoaneurysm, especially at aortic isthmus
    - Aortic wall irregularity
    - Abrupt change of aortic contour or caliber
    - Irregular uncontained extraluminal collection of extravasated contrast
  - Indirect signs
    - Periaortic hematoma (absent in up to 20% of patients)

MR Findings

- Impractical, generally no role in evaluation of acute BAI
  - May require sedation, especially young children
  - Patients usually have multisystem trauma, especially head injuries, requiring urgent treatment
- Can demonstrate delayed complications such as pseudoaneurysm, posttraumatic coarctation of aorta

Echocardiographic Findings

- May be performed in operating room; especially useful when patients go immediately to surgery
- Especially sensitive for intimal tears

Angiographic Findings

- Essentially replaced by CTA
- Used in equivocal cases or if endovascular repair planned

Imaging Recommendations

- Best imaging tool
  - Multidetector CT angiography (at least 64 slice) less likely to show pulsation artifact, allows thin multiplanar isotropic reconstructions
- Protocol advice
  - Quarter/half rotation volume data reconstructions or cardiac gating can reduce pulsation artifacts

DIFFERENTIAL DIAGNOSIS

Normal Thymus

- Prominent in young children, often accentuated by rotation or low lung volumes
- May obscure aortic contour

CT Artifacts

- Cardiac motion transmitted to aorta may mimic intramural hematoma or intimal tear
  - Rapid heart beat in children ↑ chances of pulsation artifact
- Streak artifact from contrast bolus may obscure aortic lumen or mimic intimal tear
- Atelectasis or parenchymal contusion in adjacent lung may be mistaken for aortic wall thickening

Ductus Diverticulum

- Broad-based outpouching of contrast with smooth contour & obtuse margins
- Like most traumatic aortic injuries, located at aortic isthmus
- Found in up to 10% of normal population
- No hematoma, contrast leak, or intimal flap

PATHOLOGY

General Features

- Etiology
  - Majority of pediatric BAIs due to rapid & forceful deceleration injuries
    - Mostly motor vehicle crashes (MVCs)
    - Falls, ATV crashes, & bicyclist/pedestrian versus automobile collisions also common
  - Sudden deceleration combined with fixation of aorta at ligamentum arteriosum → shearing, stretching, & torsional forces that injure aortic wall
  - Remainder of pediatric TAIs due to gunshot wounds, penetrating injuries, & crushing/compression forces
- Pediatric BAI rare compared to adults, as children have
  - Greater chest wall compliance
  - Relatively elastic vascular tissues (due to lack of atherosclerotic disease)
  - Lower body mass (thus less kinetic energy on impact)
  - Lower likelihood of MVCs

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Up to 50% of children with BAI lack external signs of chest trauma
    - Often have more obvious neurologic, abdominal, or skeletal injuries
  - May have no signs/symptoms, or only nonspecific chest pain, dyspnea
  - Hypotension, ↓ pulses in lower extremities, paraplegia
- Other signs/symptoms
  - Associated thoracic cage injuries: Diaphragm rupture, pulmonary contusion, rib fractures
  - High association with other injuries, especially traumatic brain injury & solid abdominal organ injury

Demographics

- Trauma most common cause of death in children, but vascular trauma accounts for < 1%; BAI even more rare
  - Retrospective review of ~ 11,000 children with severe blunt trauma showed < 0.1% sustained BAI
- Associated with high-risk recreational activities
  - Most pediatric BAI from MVCs in older children/teenagers
  - M:F ~ 3:1

Natural History & Prognosis

- Very low incidence, even in children involved in high-energy blunt chest trauma, but mortality rate very high
  - ~ 85% die at scene of injury
- Survival dependent on type of injury as well as time from injury to intervention
  - Injuries that do not alter external contour of aorta (intimal tears & intimal flaps) have relatively good prognosis (mortality 5-15% with treatment)
  - Injuries that do alter external contour of aorta have higher mortality (~ 25% for pseudoaneurysm & up to 90% for aortic rupture)
- Without intervention, mortality from BAI very high (80% mortality < 1 hour, 85% mortality < 24 hours, & 98% mortality < 10 weeks)
- Late complications include posttraumatic pseudoaneurysm & coarctation or spinal cord ischemia → paralysis

Treatment

- Rapid recognition of diagnosis critical
- Emergent operative treatment has been gold standard, but delayed repair becoming more common
  - β-blockade for all patients if hemodynamically stable
  - Intimal tears (< 10 mm) may require only observation
  - Intimal flaps (> 10 mm) often observed, treated with endovascular repair if progression of injury
  - Patients with external aortic contour abnormality treated emergently, urgently, or on semielective basis depending on hemodynamic stability & likelihood of surviving associated injuries
- Delayed endovascular treatment allows other injuries to be assessed & treated while ↓ operative complications
  - Requires strict blood pressure control
  - Provides time to appropriately size endograft & ensure that access sites can accommodate device
  - Short-term complications mainly at access site (bleeding, infection, pseudoaneurysm, etc.)
  - Long-term complications include device migration, fracture, endoleak, & arterial wall injury

DIAGNOSTIC CHECKLIST

Consider

- Look for signs of BAI in all cases of blunt or high-energy chest trauma
- Look for proxy signs of high-energy trauma, such as fractures of 1st &/or 2nd rib(s) &/or scapula
- Potential CTA pitfalls: Pulsation artifact, streak artifact, &/or periaortic focal atelectasis

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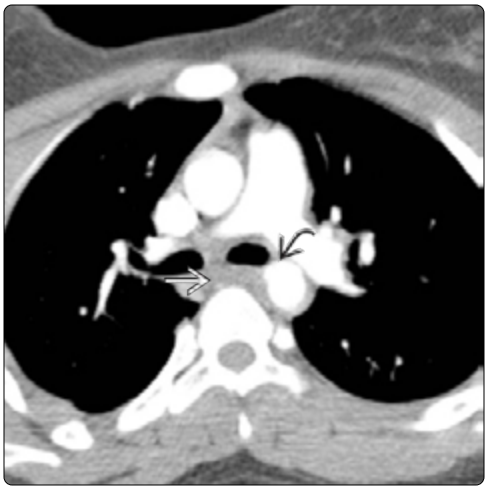
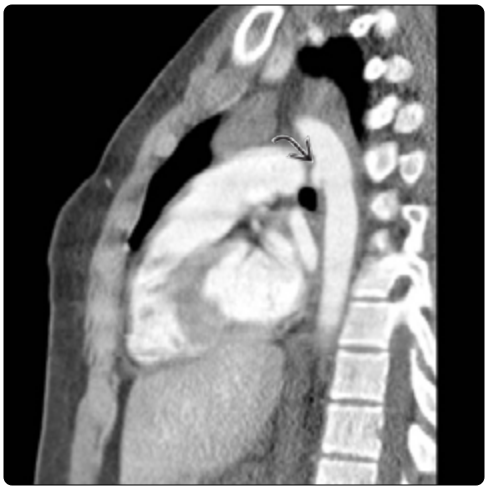
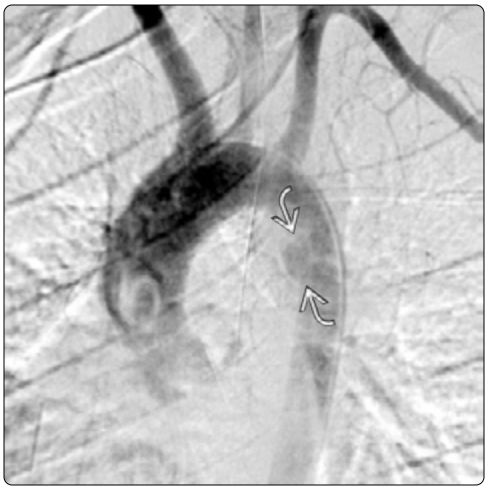
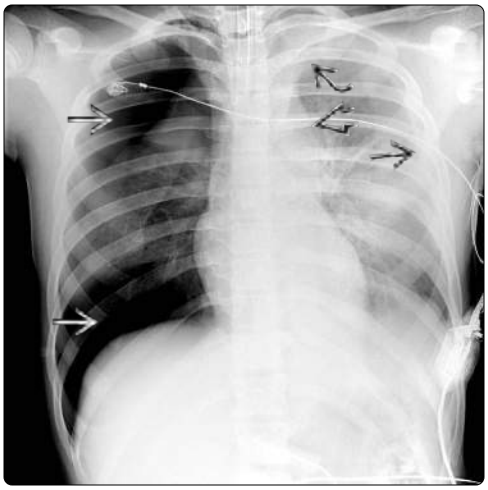


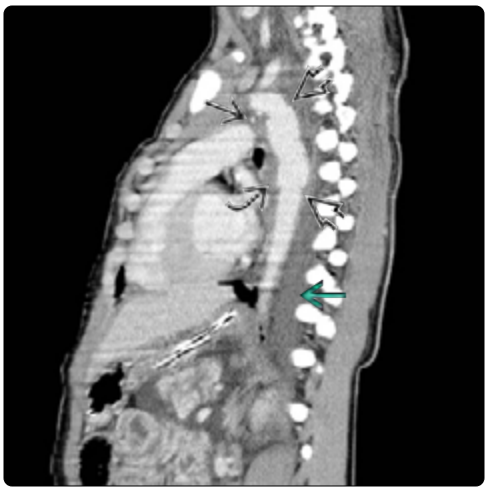


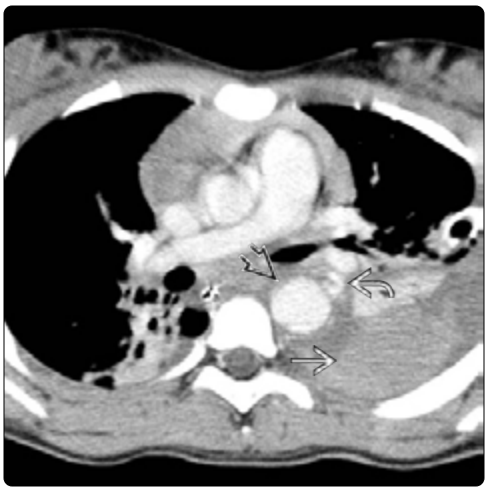
(Left) *Axial CTA in a 13-year-old girl shows a subtle, focal outpouching of contrast from the anteromedial wall of the descending aorta. The regional fat is slightly increased in attenuation from mediastinal hemorrhage.* **(Right)** *Sagittal CTA in the same patient again shows the subtle traumatic pseudoaneurysm.*





(Left) *Oblique DSA in the same child confirms the presence of a traumatic pseudoaneurysm just below the level of the aortic isthmus.* **(Right)** *Portable chest radiograph of an 11-year-old patient who sustained an aortic tear during a MVC demonstrates a left apical cap, obscuration of the aortic contour, & a hemothorax. There is also a large tension pneumothorax on the right.*





(Left) *Sagittal CTA in the same child shows a long segment of aortic caliber change & wall irregularity from the level of the ligamentum arteriosum (which is partially calcified) to the mid-descending aorta. Note the high-attenuation mediastinal fat & pleural effusion from hemorrhage.* **(Right)** *Axial CTA from the same study shows active extravasation from an aortic rupture as well as a large, high-attenuation hemothorax. The aorta is focally enlarged & irregular. This child died from these injuries.*

Pneumomediastinum

KEY FACTS

IMAGING

- Best imaging tool for pneumomediastinum (PM): Chest radiograph
  - Pleural line lateral to main pulmonary artery & aortic arch
  - Vertical lucencies tracking on either side of superior mediastinum into neck
  - Continuous diaphragm sign: Air between pericardium & diaphragm results in visualization of normally obscured superior surface of central diaphragm
  - Spinnaker sail sign: Thymic elevation by mediastinal air
  - Ovoid retrocardiac air collection: Infraazygos vs. inferior pulmonary ligament
  - Naclerio V sign: Air outlines lateral margin of descending aorta & extends laterally along left hemidiaphragm
- CT or esophagram only with radiographic evidence of trauma, risk factors for aerodigestive tract injury (foreign body, surgery, mediastinitis), or respiratory distress

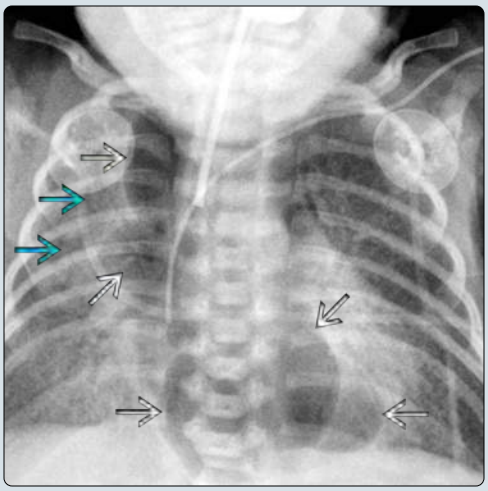
PATHOLOGY

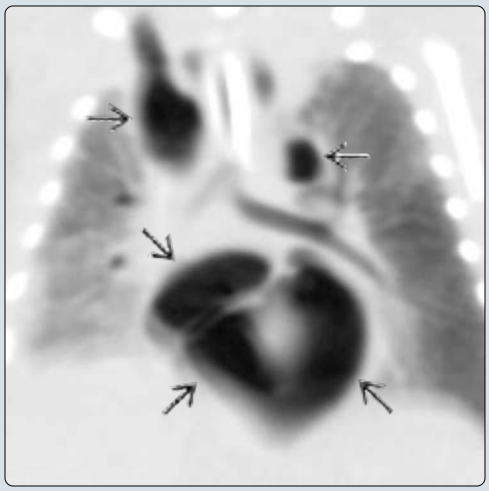
- Spontaneous PM: Extension of air from ruptured alveolus into interstitium & mediastinum
  - Asthma & respiratory infection most common
- Secondary PM: Disruption of aerodigestive tract (trauma, foreign body, Boerhaave syndrome), surgery, mediastinitis

CLINICAL ISSUES

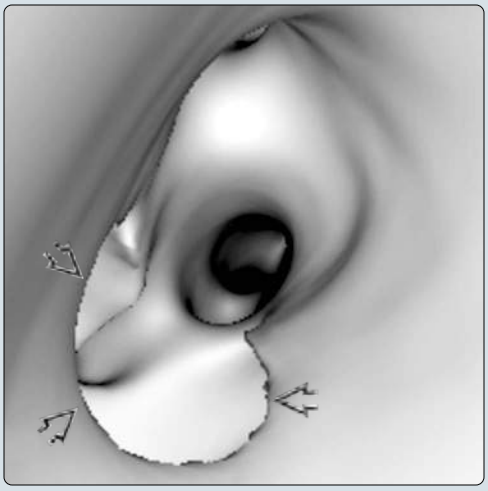
- Common symptoms: Chest pain, cough, dyspnea
- Secondary PM likely to present with rib fracture, pneumothorax, hemothorax, intracranial injury, respiratory distress, tachycardia
- Isolated spontaneous PM in stable patient: Self-limited
- Spontaneous PM with symptom progression: Further evaluate & treat underlying pulmonary cause
- Secondary PM: CT or esophagram if clinical or imaging evidence of aerodigestive tract injury
  - Widespread screening of low yield in blunt trauma patients with otherwise normal chest radiograph

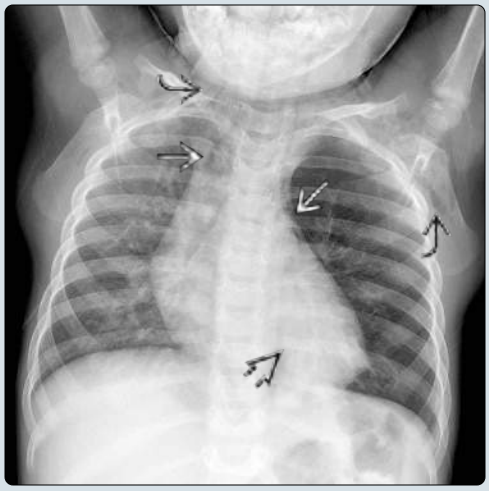
(Left) *AP chest radiograph in this intubated neonate demonstrates a large amount of abnormal lucency ➡ throughout the mediastinum, consistent with pneumomediastinum. The thymic tissue ⇨ is displaced to the right.* **(Right)** *Coronal NECT in the same patient again demonstrates the large amount of pneumomediastinum ⇨.*





(Left) *Virtual bronchoscopy CT reconstruction in the same patient demonstrates perforation ⇨ of the left main bronchus from an endoluminal perspective. The perforation was the result of iatrogenic trauma during intubation.* **(Right)** *AP chest radiograph in a 2-year-old patient shows hyperinflation of the left lung with retrocardiac atelectasis ⇨ & scattered foci of pneumomediastinum ➡ & subcutaneous emphysema ⇨. Subsequent bronchoscopy demonstrated an aspirated peanut in the left main bronchus.*





TERMINOLOGY

Definitions

- Pneumomediastinum (PM): Extraluminal mediastinal gas

IMAGING

Radiographic Findings

- Central lucencies with pleural line lateral to main pulmonary artery & aortic arch
- Vertical lucencies tracking on either side of superior mediastinum into neck
- Spinnaker sail sign: Elevation of thymus by mediastinal air
- Continuous diaphragm sign: Air between pericardium & diaphragm results in visualization of normally obscured superior surface of central diaphragm
- Air in inferior pulmonary ligament: Round/ovoid off-midline retrocardiac collection
- Infraazygos pneumomediastinum: Midline retrocardiac collection; more likely to displace esophagus than inferior pulmonary ligament air
- Naclerio V sign: Air outlines lateral margin of descending aorta & extends laterally along left hemidiaphragm

Fluoroscopic Findings

- ± esophageal leakage of contrast into mediastinum

CT Findings

- Extraluminal gas in mediastinal space
- May depict underlying disruption of tracheobronchial tree

Imaging Recommendations

- Chest radiograph as initial modality
- CT or esophagram in setting of risk factors for aerodigestive tract injury (foreign body, surgery, mediastinitis, definite thoracic injury) or respiratory distress

DIFFERENTIAL DIAGNOSIS

Pneumothorax

- Upright & decubitus views can help determine whether air is localized in mediastinum vs. freely mobile in pleural space

Pneumopericardium

- Round lucency with continuous outline of heart margins; displaces pericardium outward
- Air in pericardial space changes with patient position
- Air does not extend into superior mediastinum

Paramediastinal Pneumatocele

- ± internal fluid-fluid level or "claw" of lung parenchyma

PATHOLOGY

General Features

- Spontaneous PM: Pressure gradient between alveolus & surrounding tissue causes rupture with extension of air into interstices & mediastinum
  - Asthma, respiratory infection > straining against closed glottis, surfactant deficiency, meconium aspiration, inhalation drug use, forceful coughing
- Secondary PM: Disruption of aerodigestive tract (trauma, foreign body, Boerhaave syndrome), surgery, mediastinitis
  - Aerodigestive tract injury rare in pediatric population (4.1% of traumatic PM)
- Extension of air from peritoneum or retroperitoneum

CLINICAL ISSUES

Presentation

- Most common: Chest pain, cough, shortness of breath
- ± crepitus in neck or chest (from subcutaneous emphysema)
- Secondary PM likely to present with rib fracture, pneumothorax, hemothorax, intracranial injury, respiratory distress, tachycardia

Natural History & Prognosis

- Spontaneous PM: Self-limited, recurrence rare
  - Morbidity related to underlying pulmonary condition
- Secondary PM: Mortality related to other injuries
- Complications uncommon, can include pseudotamponade, mediastinitis, pneumothorax

Treatment

- Isolated spontaneous PM in stable patient
  - Supportive care, emergency department observation
  - No evidence to support further studies
- Spontaneous PM with symptom progression
  - Evaluate & treat underlying pulmonary condition
- Secondary PM
  - CT or esophagram if underlying aerodigestive tract injury specifically suspected
  - Treat underlying condition

DIAGNOSTIC CHECKLIST

Consider

- Widespread screening (especially with esophagram) of low yield in blunt trauma patients with otherwise normal chest radiograph
  - Selective investigation with high suspicion (e.g., rib fracture, effusion, pneumothorax)

Image Interpretation Pearls

- In toddler with no history of asthma or trauma, consider aspirated foreign body

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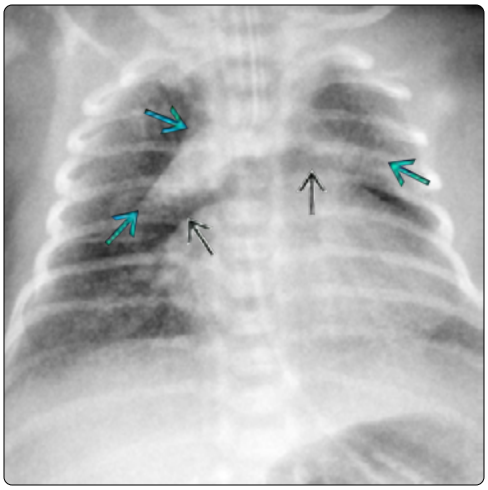
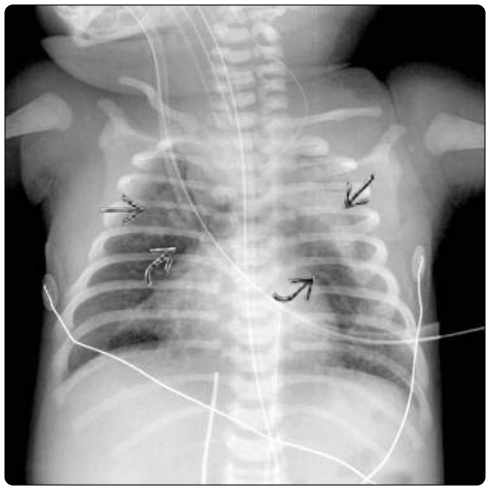
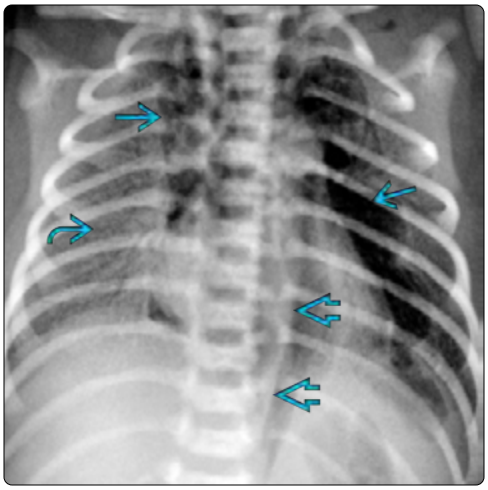
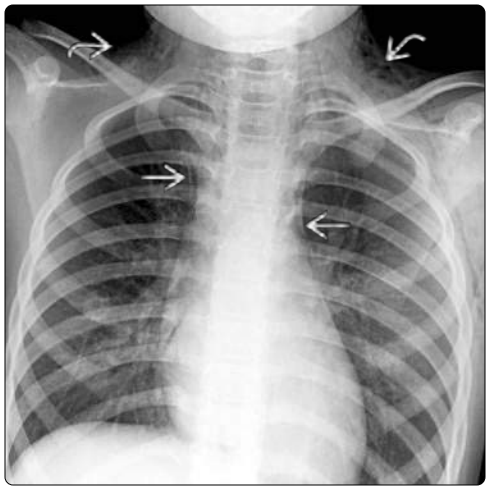
(Left) *Supine chest radiograph of a 2 day old demonstrates air uplifting the thymus, an appearance known as the spinnaker sail sign that is consistent with pneumomediastinum. The diffuse granular lung opacities relate to the patient's known surfactant deficiency.* **(Right)** *AP chest radiograph demonstrates uplifting of the thymus by pneumomediastinum in this newborn with a history of meconium aspiration. The pneumomediastinum resolved spontaneously after several days.*



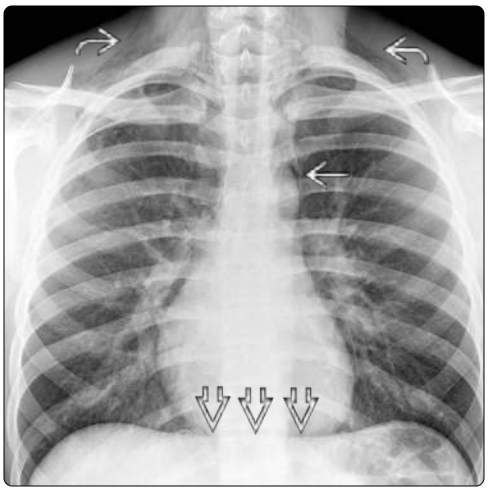


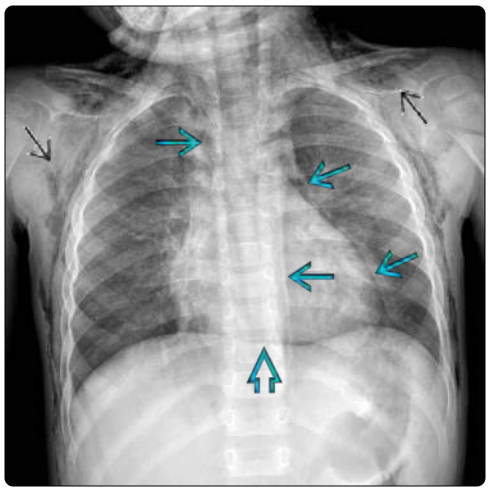
(Left) *Supine chest radiograph in a neonate shows lucency of pneumomediastinum outlining the descending thoracic & upper abdominal aorta. Diffuse granular opacities with air bronchograms relate to the patient's surfactant deficiency.* **(Right)** *PA chest radiograph demonstrates pneumomediastinum & subcutaneous emphysema in this child with chest pain & dyspnea after a bicycle accident with a direct handlebar injury to the thoracic inlet.*

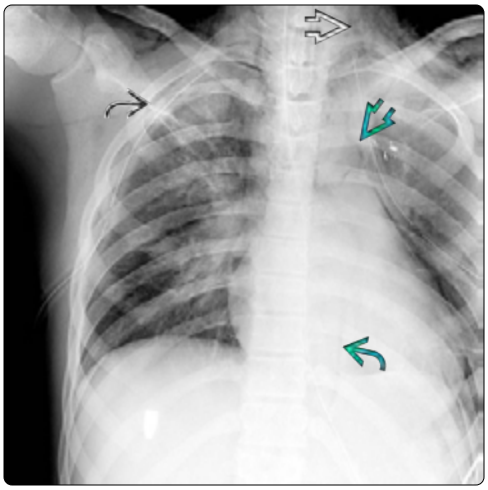


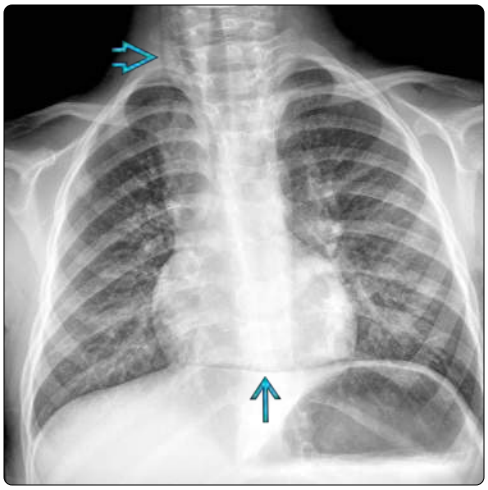


(Left) *PA chest radiograph of a teenager after a hockey injury demonstrates a continuous diaphragm sign & lucency along the left aspect of the superior mediastinum, consistent with pneumomediastinum. There is also subcutaneous emphysema.* **(Right)** *Supine chest radiograph of a 3 year old presenting with cough, fever, & facial swelling shows extensive subcutaneous emphysema & pneumomediastinum. There is a continuous diaphragm sign in this patient with RSV.*

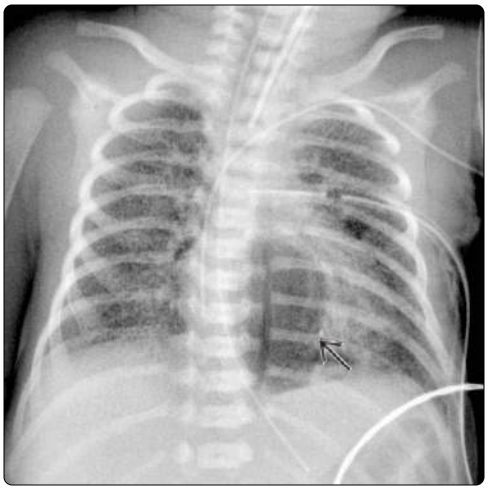


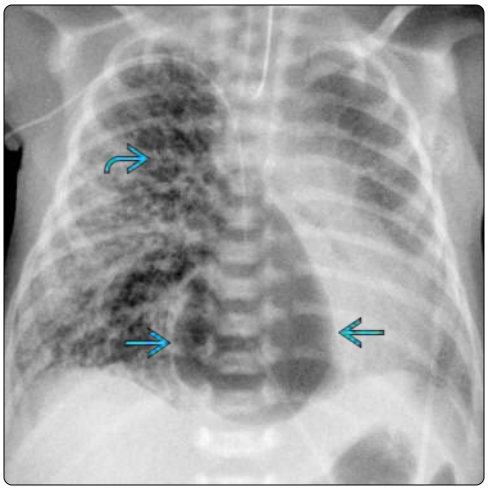




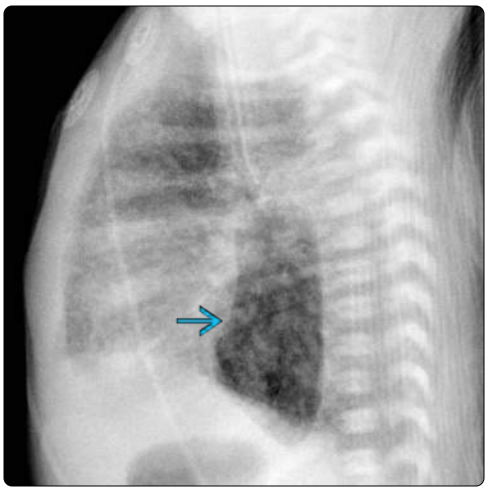


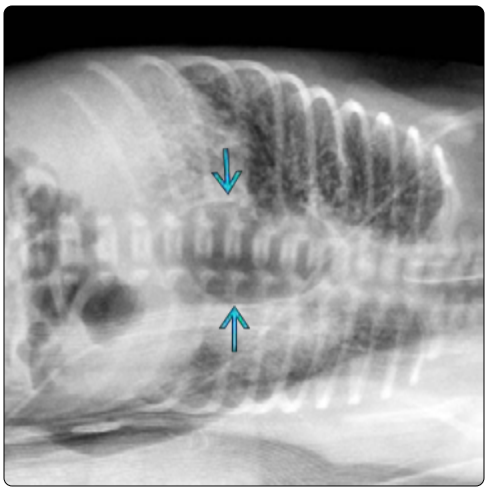
(Left) *Supine chest radiograph of a 17 year old with gun shot wounds demonstrates pneumomediastinum with air outlining the aortic arch & descending aorta. Other findings include left upper lobe contusions, a right pneumothorax, & subcutaneous emphysema. The trachea & esophagus were lacerated.* **(Right)** *Upright chest radiograph in a 4 year old with a respiratory infection shows subcutaneous emphysema & a continuous diaphragm sign, consistent with pneumomediastinum.*





(Left) *AP chest radiograph in a neonate with surfactant deficiency disease demonstrates a relatively large, well-circumscribed lucent collection, typical in appearance for retrocardiac pneumomediastinum.* **(Right)** *Supine chest radiograph in a 2-day-old premature infant with surfactant deficiency shows a round lucent collection projecting over the inferior midline, consistent with pneumomediastinum. Associated right pulmonary interstitial emphysema (PIE) is noted.*





(Left) *Lateral chest radiograph of the same patient localizes the pneumomediastinum to the retrocardiac region.* **(Right)** *Left decubitus view of the chest in the same patient was obtained to assess the mobility of the lucent collection (& ensure that this did not represent mobile air from a pneumothorax). The lucency did not move & was self-limited, resolving in 5 days. These features are consistent with retrocardiac pneumomediastinum.*

Neuroendocrine Cell Hyperplasia of Infancy

KEY FACTS

TERMINOLOGY

- NEHI: Form of childhood interstitial lung disease (chILD) associated with neuroendocrine cell hyperplasia

IMAGING

- Best test: HRCT (with inspiratory & expiratory images)
  - ~ 80% sensitive; ~ 100% specific with characteristic imaging pattern & clinical presentation
- Characteristic appearance
  - Geographic central-predominant ground-glass opacification (GGO), especially of right middle lobe & lingula
    - Affects ≥ 4 lobes (counting lingula) in > 90% of cases
  - Diffuse mosaic air-trapping
  - No other abnormalities in 57-65% of cases

TOP DIFFERENTIAL DIAGNOSES

- Bronchiolitis obliterans (constrictive bronchiolitis)
- Asthma
- Surfactant dysfunction disorder

PATHOLOGY

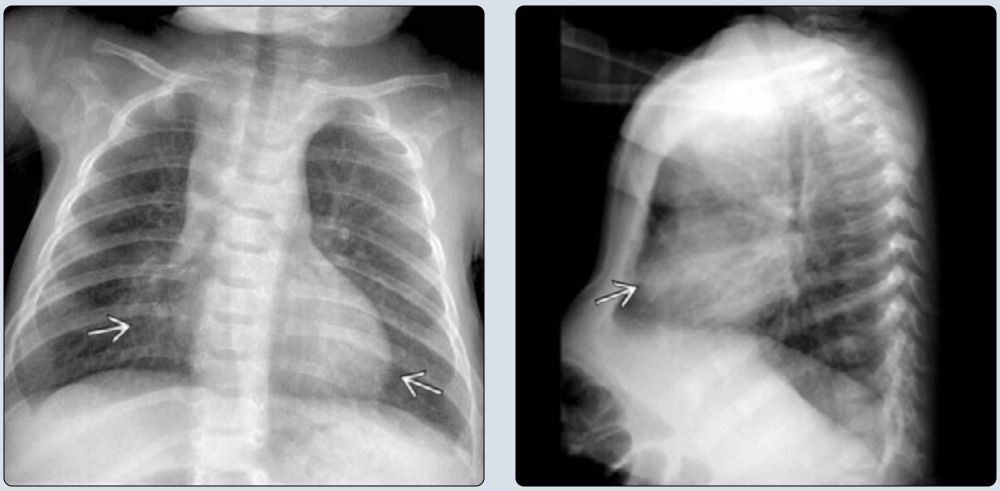
- ↑ number of bombesin-immunopositive pulmonary neuroendocrine cells (PNECs) in distal respiratory bronchioles & alveolar ducts
- Etiology unknown

CLINICAL ISSUES

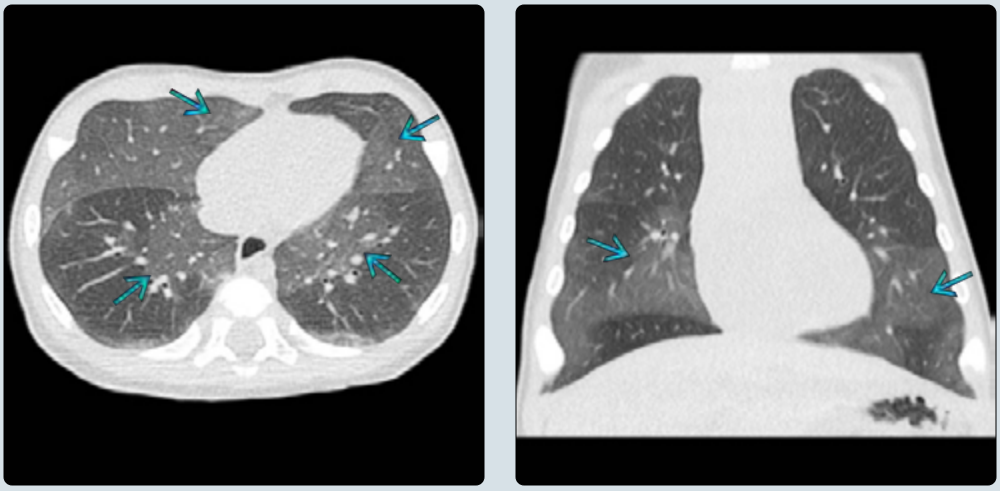
- Typically presents in first 6-12 months of life with persistent retractions, tachypnea, crackles, hypoxemia, &/or failure to thrive
- Not responsive to steroids (unlike other chILDs)
- Does not progress to respiratory failure; gradually improves

DIAGNOSTIC CHECKLIST

- Important to suggest NEHI as diagnostic possibility
  - Immunostaining for bombesin not routinely performed
  - If NEHI not suggested by radiologist, diagnosis likely to be missed by histopathology



(Left) *AP chest radiograph of a 4-month-old boy with an oxygen requirement & failure to thrive (FTT) shows subtle hazy or ground-glass opacification (GGO) in the right middle lobe & lingula plus mild overall hyperinflation.* **(Right)** *Lateral chest radiograph of the same 4-month-old boy shows the GGO in the middle lobe/lingula.*



(Left) *Axial inspiratory HRCT of the chest in the same 4-month-old boy with an oxygen requirement & failure to thrive (FTT) shows central-predominant ground-glass opacification (GGO) in the lower lobes, lingula, & middle lobe.* **(Right)** *Coronal inspiratory HRCT of the same 4-month-old boy shows the middle lobe & lingular distribution of GGO. The imaging findings are sufficient, along with the clinical history, to make the diagnosis of NEHI without biopsy.*

TERMINOLOGY

Abbreviations

- Neuroendocrine cell hyperplasia of infancy (NEHI)

Synonyms

- Familial neuroendocrine cell hyperplasia of infancy
- Persistent tachypnea of infancy (term no longer used)

Definitions

- Form of childhood interstitial lung disease (chILD) associated with neuroendocrine cell hyperplasia
  - First descriptions in 2001-2005

IMAGING

General Features

- Best diagnostic clue
  - Characteristic ground-glass opacification (GGO) pattern in central lungs, especially right middle lobe & lingula, with mosaic air-trapping

Radiographic Findings

- Nonspecific radiographic findings
- Perihilar opacities & hyperinflation may be similar to viral or reactive airways disease
  - Bronchial wall thickening typically absent

CT Findings

- HRCT
  - Geographic GGO
    - Central-predominant distribution
      - Does not change with patient position (supine vs. prone)
    - Most pronounced in right middle lobe & lingula
    - Often widely distributed
      - Affects ≥ 4 lobes (counting lingula) in > 90% of cases
  - Diffuse mosaic air-trapping
  - No other abnormalities in 57-65% of cases
  - Other findings are less common & nonspecific
    - Bronchial wall thickening
    - Bronchiectasis
    - Linear/reticular opacity
    - Nodules
  - Imaging findings may confirm but do not exclude diagnosis of NEHI
    - Misdiagnosis more likely when other abnormalities present & distribution of GGO not classic

Imaging Recommendations

- Best imaging tool
  - HRCT ~ 80% sensitive; specificity approaches 100% with characteristic CT pattern & clinical presentation
- Protocol advice
  - Routine HRCT including inspiratory & expiratory images
  - May use low-dose technique (e.g., 50 mAs) if evaluation of mediastinum not needed

DIFFERENTIAL DIAGNOSIS

Bronchiolitis Obliterans (Constrictive Bronchiolitis)

- Follows insulting event such as infection (especially adenovirus, influenza, & *Mycoplasma*), toxic inhalation, or lung/bone marrow transplantation
- Marked asymmetric radiolucency, often unilateral (Swyer-James-MacLeod syndrome)
- ↓ pulmonary vasculature in affected areas
- Inspiratory lung volumes may be ↓ with expiratory air-trapping
- Bronchiectasis & mild bronchial wall thickening

Asthma

- Air-trapping, but GGO much less prominent
- Does not preferentially affect middle lobe & lingula
- Associated with bronchial wall thickening

Surfactant Dysfunction Disorder

- GGO more diffuse than in NEHI
- Associated with septal thickening, unlike NEHI

PATHOLOGY

General Features

- Etiology
  - Unknown
- Genetics
  - Genetic mechanisms thought to play role as suggested by familial patterns in some cases
  - Reports of heterozygous mutations in thyroid transcription factor 1 (TTF1) gene (*NKX2-1*)

Staging, Grading, & Classification

- Form of childhood interstitial lung disease (chILD)
- Considered in class of disorders of unknown etiology
  - Pulmonary interstitial glycogenosis (PIG) also in this category

Microscopic Features

- ↑ number of bombesin-immunopositive pulmonary neuroendocrine cells (PNECs) in distal respiratory bronchioles & alveolar ducts
  - Enlarged & ↑ number of PNEC clusters in neuroepithelial bodies (NEBs)
    - ~ 10% in normal patients; > 70% in patients with NEHI
  - Minor patchy inflammation present in small percentage of airways
- Biopsy may not be necessary if clinical history, imaging, & pulmonary function tests typical for NEHI
  - Some authors advocate term "NEHI syndrome" if diagnosis based on clinical + CT findings without lung biopsy

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Persistent retractions, tachypnea, crackles, hypoxemia
- Other signs/symptoms
  - Failure to thrive (FTT)
  - Hypoxemia out of proportion to physical exam findings

- Pulmonary symptomatology out of proportion to minor, nonspecific lung biopsy findings
  - Despite symptoms, patients do not progress to respiratory failure
  - Usually hospitalized only for investigation or biopsy
    - Symptoms not severe enough to merit admission
- Patients usually have less complicated history than with other chILDs
- Clinical profile
  - Pulmonary function tests
    - ↓ forced expiratory volume (FEV), ↑ functional residual capacity (FRC), ↑ residual volume (RV)

Demographics

- Age
  - Typically diagnosed in first 12-18 months of life
    - Mean age at presentation to pulmonologist: 7 months
    - Mean age at diagnosis: 12 months
    - Age of presentation older than other chILDs, which present in early infancy
- Gender
  - Slight male predominance in original series of 15 cases
- Epidemiology
  - Incidence unknown but thought to be rare

Natural History & Prognosis

- Long-term prognosis unclear
  - Most children gradually improve over time
  - Preliminary research suggests symptoms may persist into adulthood
  - No reported deaths
- NEHI exacerbation
  - Episodes of ↑ air-trapping in children with NEHI who had previously experienced significant clinical improvement

Treatment

- Unlike other chILDs, NEHI not responsive to steroid therapy
  - Important to differentiate NEHI from other chILDs to avoid unnecessary side effects of steroids
- Treatment supportive
  - Oxygen supplementation
  - Nutrition optimization to prevent FTT

DIAGNOSTIC CHECKLIST

Consider

- NEHI in older infants with persistent tachypnea
- Expiratory images may be helpful for diagnosis (due to air-trapping)

Image Interpretation Pearls

- Characteristic CT appearance
  - Geographic GGO pattern in central lungs, especially right middle lobe & lingula
  - Mosaic air-trapping

Reporting Tips

- Important to specifically suggest NEHI as diagnostic possibility
  - Immunostaining for bombesin not routinely performed
  - If NEHI not suggested by radiologist, diagnosis likely to be missed

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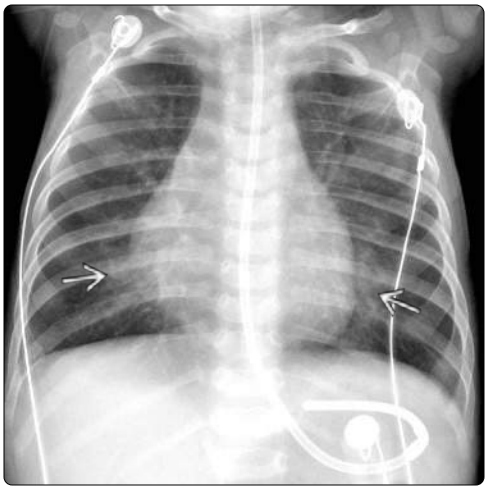
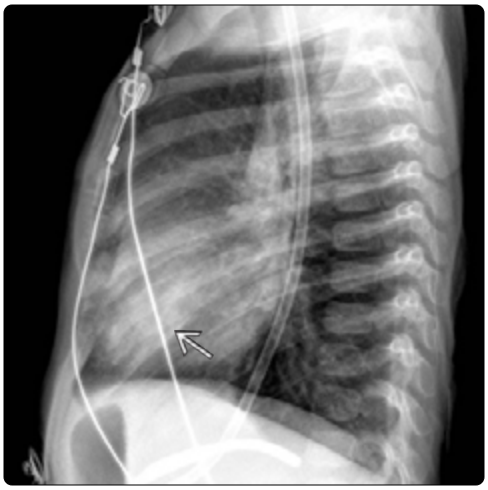
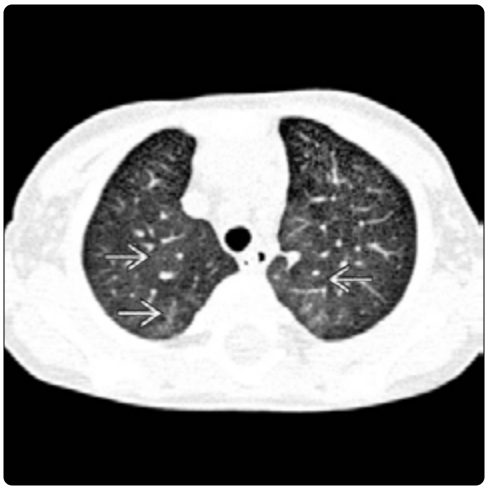
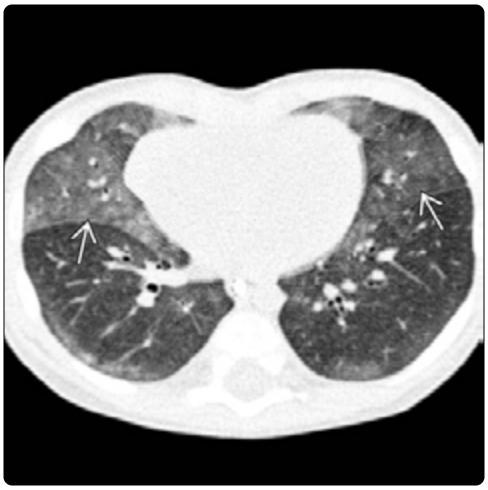


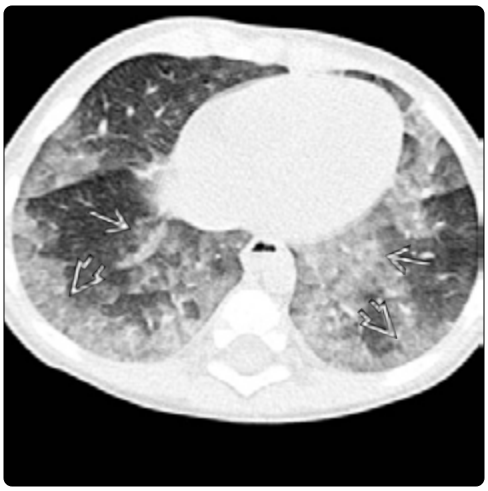


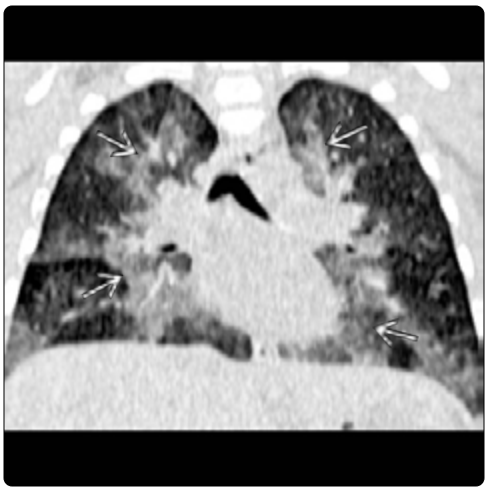
(Left) *AP chest radiograph of a 5-month-old boy with failure to thrive (FTT) shows hazy or ground-glass opacification (GGO) ⇨ in the right middle lobe & lingula.* **(Right)** *Lateral chest radiograph of the same 5-month-old boy shows the GGO ⇨ in the right middle lobe/lingula. Moderate hyperinflation is noted.*





(Left) *Axial inspiratory HRCT of the same 5-month-old boy shows subtle central-predominant upper lobe GGO ⇨.* **(Right)** *Axial inspiratory image from the same HRCT shows that the GGO ⇨ is most prominent in the right middle lobe & lingula. Biopsy confirmed NEHI.*





(Left) *Axial inspiratory HRCT of a 6-month-old boy with FTT, tachypnea, & hypoxia shows confluent GGO that is more severe than is often seen in NEHI, demonstrating both central ⇨ & peripheral ⇨ distributions.* **(Right)** *Coronal inspiratory image from the same HRCT again shows central & peripheral GGO ⇨. A subsequent lung biopsy showed numerous bombesin-immunopositive pulmonary neuroendocrine cells (PNECs) within bronchioles & alveolar ducts, indicating NEHI.*

Pulmonary Interstitial Glycogenosis

KEY FACTS

TERMINOLOGY

- Specific disorder of unknown etiology in childhood interstitial lung disease (chILD) classification system
- More accurate term: Neonatal pulmonary interstitial glycogen accumulation disorder

IMAGING

- Nonspecific imaging findings of diffuse or patchy hazy/ground-glass opacities, interstitial thickening, & hyperinflation
  - Appearances largely could be attributed to alveolar growth abnormalities, which pulmonary interstitial glycogenosis (PIG) often accompanies histopathologically
  - Other lung diseases, including infection & surfactant dysfunction disorders, could also show similar findings

TOP DIFFERENTIAL DIAGNOSES

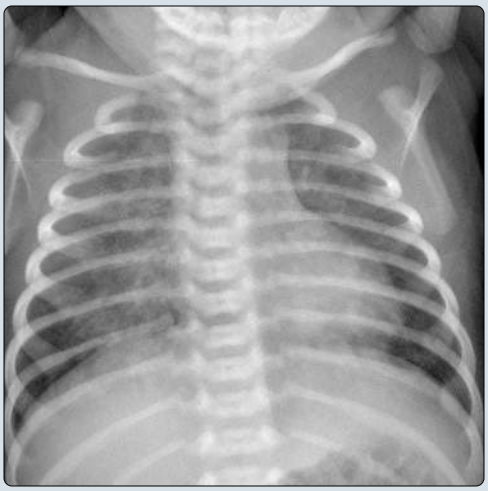
- Alveolar growth abnormality (AGA)
  - PIG commonly associated with AGA (which confounds assessment of imaging appearance of "pure" PIG)
- Diffuse lung developmental disorders
  - Profound impairment of gas exchange
  - Death usually < 1 month of age (unless ECMO bridges to lung transplantation)
- Disorders of surfactant
  - Surfactant deficiency in premature neonates & genetic disorders affecting surfactant metabolism
- Neonatal pneumonia
  - Pleural effusion frequent

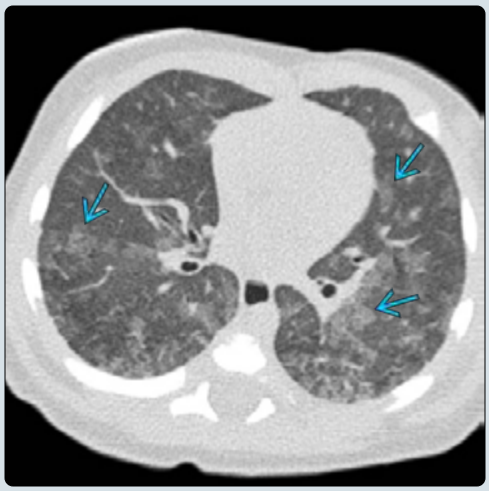
PATHOLOGY

- Infiltration & expansion of alveolar interstitium/septa by glycogen-laden immature mesenchymal cells

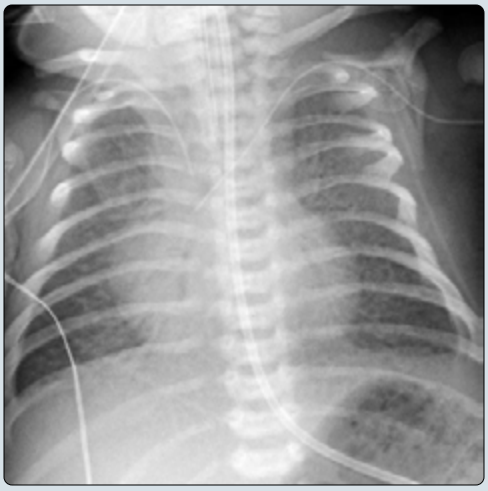
CLINICAL ISSUES

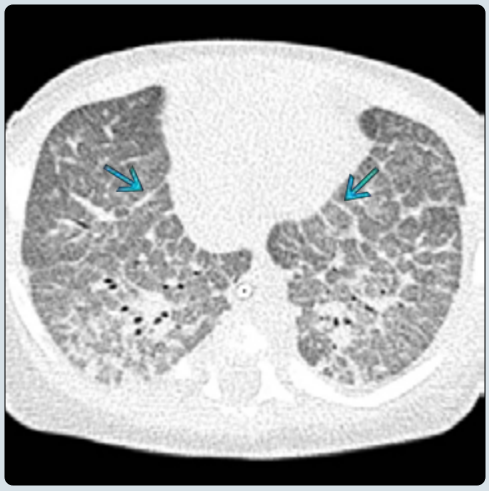
- PIG affects neonates & young infants
- Prognosis may be favorable without concurrent disease





(Left) *AP radiograph in a term neonate with respiratory difficulty shows slight hyperinflation with patchy perihilar & peripheral hazy opacities. No pleural effusion or pneumothorax is seen.* **(Right)** *Axial HRCT from the same 11-day-old neonate at the level of the middle & lingular bronchi confirms patchy areas of ground-glass opacity. Biopsy revealed a mild alveolar growth abnormality with patchy pulmonary interstitial glycogenosis (PIG).*





(Left) *AP radiograph in a term neonate with Turner syndrome & respiratory failure reveals hyperinflation with diffuse ground-glass opacities & septal thickening.* **(Right)** *Axial HRCT from the lower lobes of the same patient with Turner syndrome shows septal thickening & areas of patchy ground-glass opacity. Biopsy revealed diffuse PIG.*

TERMINOLOGY

Abbreviations

- Pulmonary interstitial glycogenosis (PIG)

Synonyms

- Infantile cellular interstitial pneumonitis (former name)
- Neonatal pulmonary interstitial glycogen accumulation disorder

Definitions

- Specific disorder of unknown etiology in childhood interstitial lung disease (chILD) classification system
- Results from infiltration & expansion of alveolar interstitium/septa by glycogen-laden mesenchymal cells

IMAGING

General Features

- Best diagnostic clue
  - Variable imaging features
  - Patchy or diffuse ground-glass opacity & interstitial thickening described
  - However, coexistent lung disease (e.g., alveolar growth abnormality) likely impacts imaging appearance
- Location
  - No specific lung zonal predilection; opacities may be diffuse or heterogeneous

Radiographic Findings

- Interstitial thickening
- Diffuse or patchy hazy/ground-glass opacities
- Hyperinflation

CT Findings

- Architectural distortion
- Ground-glass opacities
- Interstitial thickening
- Hyperlucent areas

Imaging Recommendations

- Best imaging tool: HRCT

DIFFERENTIAL DIAGNOSIS

Alveolar Growth Abnormality

- PIG commonly associated with alveolar growth abnormality (AGA), which confounds assessment of imaging appearance of "pure" PIG

Diffuse Lung Developmental Disorders

- Acinar dysplasia, congenital alveolar dysplasia, alveolar capillary dysplasia with misalignment of pulmonary veins

Surfactant Dysfunction Disorders

- Abnormal proteins (SP-B, SP-C, ABCA3, & TTF-1) important in surfactant production & function

Neonatal Pneumonia

- Pleural effusions frequent

PATHOLOGY

General Features

- Presence of immature interstitial cells containing abundant cytoplasmic glycogen

Microscopic Features

- Infiltration & expansion of alveolar interstitium/septa by glycogen-laden immature mesenchymal cells, which stain positive with vimentin
  - Accumulation of glycogenated mesenchymal cells often accompanies other conditions, particularly AGA
- Little to no inflammatory change
- Diffuse or patchy involvement of pulmonary parenchyma
  - Patchy form more common in patients with AGA

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Severity of presentation highly variable
    - Respiratory failure with pulmonary hypertension
    - Tachypnea
    - Hypoxemia

Demographics

- Age
  - Neonates or young infants
    - Typically < 6 months of age

Natural History & Prognosis

- Natural history of PIG unknown
- Case reports suggest favorable prognosis in absence of concurrent disease
- Mortality reported with presence of comorbidities such as AGA, congenital heart disease, & pulmonary hypertension

Treatment

- Possible benefits from high-dose pulsed steroids based on limited cases; no controlled studies yet performed

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- No specific imaging appearance
- Published reports of PIG imaging findings describe appearances that largely could be attributed to AGA (which PIG often accompanies histopathologically)

Reporting Tips

- Caution during HRCT interpretation as nonspecific findings of septal thickening & diffuse ground-glass opacities can overlap with other disorders, including infection or disorders of surfactant

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Alveolar Growth Abnormality

KEY FACTS

TERMINOLOGY

- Alveolar growth abnormality (AGA): Pathologic pattern of enlargement & simplification of alveoli with concomitant underdevelopment of pulmonary vasculature
- Synonyms: Alveolar growth disorder, alveolar simplification

IMAGING

- Radiologic findings range from near normal to
  - Variably sized but often large secondary pulmonary lobules with vascular rarefaction
  - Subpleural & perilobular reticular opacities
  - Ground-glass opacities
  - Discrete subpleural/subfissural cysts (often in trisomy 21)
  - Hyperlucent regions that can resemble cystic change

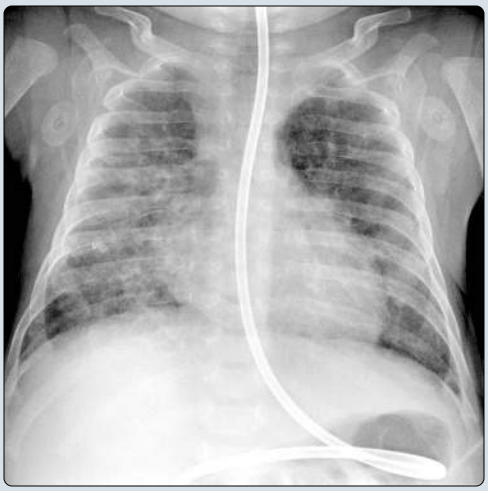
PATHOLOGY

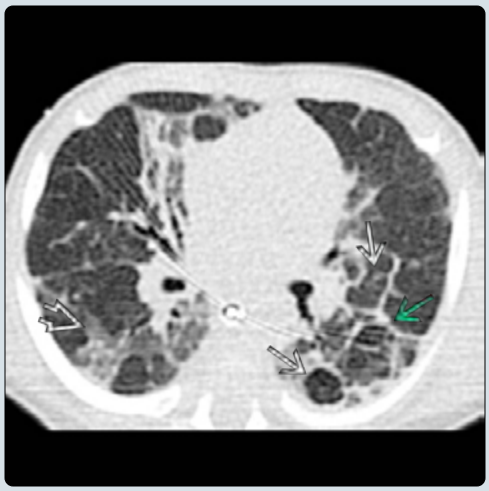
- Enlarged alveolar spaces with few alveoli + deficient vascular & alveolar septal development
  - ± concurrent pulmonary interstitial glycogenosis
- AGA often secondary; seen in wide variety of diseases
  - Prenatal conditions
    - Restriction of thoracic space, ↓ or absent breathing movements, cardiac anomalies limiting pulmonary blood supply, chromosomal abnormalities
  - Postnatal conditions
    - Prematurity (most common) with BPD
    - Term infants with early postnatal lung injury

CLINICAL ISSUES

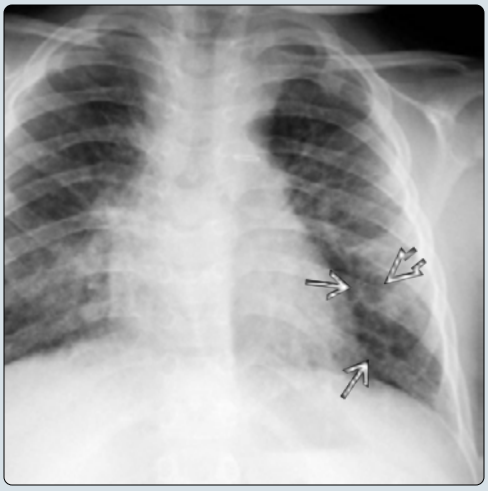
- Most common chronic diffuse lung disease in infancy
- Respiratory difficulties occur in neonatal period but natural history variable
  - Slow improvement or worsening with time depending on underlying disorder, AGA extent, ability to develop new alveoli, & presence of pulmonary hypertension
  - Filamin A (*FLNA*) mutations: Particularly severe AGA with periventricular gray matter heterotopia, cardiovascular anomalies, & connective tissue anomalies

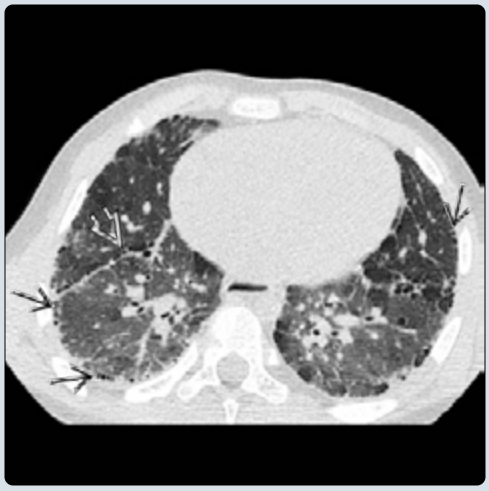
(Left) *AP chest radiograph of a former 23-week premature infant at 10 weeks of age shows bilateral hyperinflation with coarse parenchymal opacities.* **(Right)** *Axial HRCT from the same infant at 10 weeks of age shows disordered secondary pulmonary lobules of variable shape with ground-glass opacities & thick perilobular opacities, typical of chronic lung disease of prematurity with alveolar growth abnormality.*





(Left) *AP chest radiograph in a child with trisomy 21 shows perilobular opacities outlining large secondary pulmonary lobules.* **(Right)** *Axial HRCT in a child with trisomy 21 shows large secondary pulmonary lobules & subpleural/perilobular reticular opacities with subpleural & subfissural cysts.*





TERMINOLOGY

Definitions

- Alveolar growth abnormality (AGA): Pathologic pattern of enlargement & simplification of alveoli with concomitant underdevelopment of pulmonary vasculature

IMAGING

General Features

- Best diagnostic clue
  - Large secondary pulmonary lobules + vascular rarefaction
    - Imaging findings range from severe to near normal
- Location
  - Variable; cystic changes often most conspicuous in subpleural & subfissural lungs
  - Unilateral in certain congenital cases with in utero ipsilateral mass effect (e.g., lung lesion, diaphragmatic hernia, chylothorax)

Radiographic Findings

- Hyperinflation with pulmonary vascular rarefaction & generalized lucency of parenchyma
- Coarse reticular opacities

CT Findings

- Variably sized (but often large) secondary pulmonary lobules with vascular rarefaction
- Perilobular & subpleural linear opacities, parenchymal bands, ground-glass opacities
- Subpleural/subfissural cysts (often in trisomy 21)
- Hyperlucent regions that can appear cystic

Imaging Recommendations

- Best imaging tool: CT
  - Generally most conspicuous on inspiratory images
  - Expiratory images helpful if clinical &/or pulmonary function test (PFT) findings of obstructive airways disease/air trapping
- Protocol advice: Maximize lung recruitment if patients under sedation/anesthesia

DIFFERENTIAL DIAGNOSIS

Parenchymal Lung Diseases That May Result in Widespread Architectural Distortion

- Meconium aspiration
- Chronic aspiration
- Bronchopulmonary dysplasia (BPD)

PATHOLOGY

General Features

- Most common chronic diffuse lung disease in infancy
- AGA often secondary; seen in wide variety of diseases
  - Prenatal conditions
    - Restriction of thoracic space, ↓ or absent breathing movements, cardiac anomalies limiting pulmonary blood supply, chromosomal abnormalities
  - Postnatal conditions
    - Prematurity (most common) with BPD
    - Term infants with early postnatal lung injury

Microscopic Features

- Reduced radial alveolar count
- Enlarged alveolar spaces with fewer alveoli & deficient vascular & alveolar septal development
- ± concurrent pulmonary interstitial glycogenosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Respiratory difficulties in neonatal period
- Other signs/symptoms
  - Filamin A (*FLNA*) X-linked genetic mutations have particularly severe AGA
    - Pulmonary hypertension & respiratory decline

Natural History & Prognosis

- Slow improvement or worsening with time depends on underlying disorder, AGA extent, ability to develop new alveoli, & presence of pulmonary hypertension
- With prematurity
  - Advances in neonatal care (improved ventilation equipment/strategies, antenatal corticosteroids, surfactant, nutrition) → shift from classic BPD (airway obstruction & fibrosis) to "new BPD" (impaired alveolar development)
  - Unclear how "new BPD" patients remodel alveoli & pulmonary microvasculature with age & growth

Treatment

- Supportive measures: Supplemental oxygen, nutrition, & prevention of respiratory syncytial virus (RSV)
- Lung transplantation in most severely affected

DIAGNOSTIC CHECKLIST

Reporting Tips

- Clinical history important to prevent reporting all cases as chronic lung disease of prematurity/BPD
- Bilateral findings without prematurity could indicate genetic cause (trisomy 21, filamin A mutation) or peripartum condition resulting in impaired pulmonary parenchymal development

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Surfactant Dysfunction Disorders

KEY FACTS

TERMINOLOGY

- Group of rare lung diseases caused by mutations/deletions of genes affecting surfactant homeostasis
- Most frequent surfactant dysfunction disorders
  - Surfactant protein B (SP-B): *SFTPB* gene
  - Surfactant protein C (SP-C): *SFTPC* gene
  - ATP binding cassette transporter A3: *ABCA3* gene
  - Receptors for GM-CSF: *CSF2RA*, *CSF2RB* genes
    - Congenital alveolar proteinosis
  - Thyroid transcription factor: *TTF1/NKX2-1* genes
    - Brain-lung-thyroid syndrome

IMAGING

- Radiographs: Bilateral granular opacities in term neonate
- CT: Diffuse ground-glass opacification & thickened interlobular septa: "Crazy paving" pattern
  - Can progress to fibrotic changes with persistent interlobular septal thickening, cysts

TOP DIFFERENTIAL DIAGNOSES

- Surfactant deficiency related to prematurity
- Neonatal pneumonia
- Transient tachypnea of newborn
- Alveolar growth abnormalities

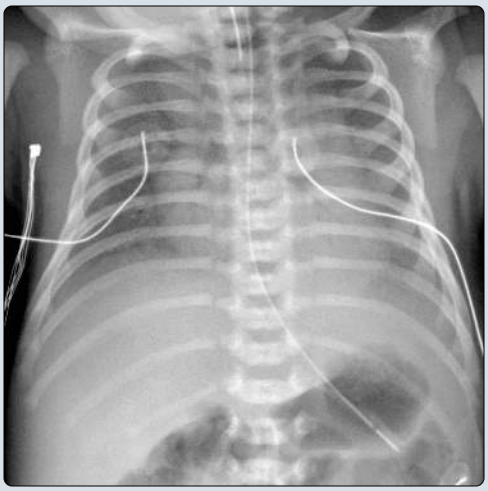
PATHOLOGY

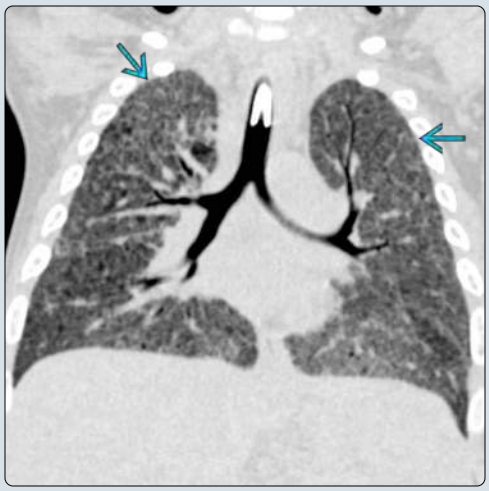
- Pulmonary surfactant: Complex mixture of lipid (90% by weight) & protein lining alveolar surface
  - Prevents end-expiratory atelectasis

CLINICAL ISSUES

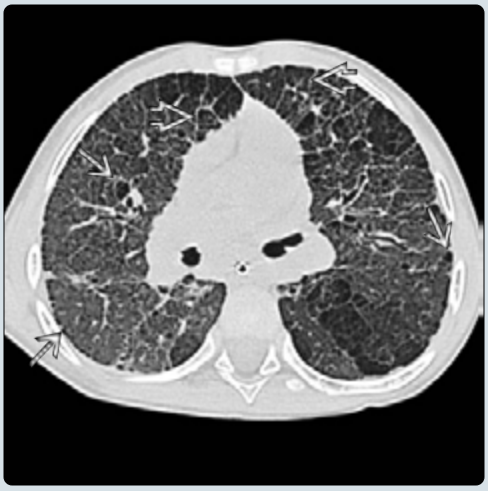
- Acute respiratory distress in full-term infants at birth
- SP-B: Progressive & usually fatal by 3-6 months of age with lung transplantation as only effective treatment
- SP-C, ABCA3: More commonly associated with diffuse lung disease in older infants, children, & adults
  - Onset of symptoms highly variable

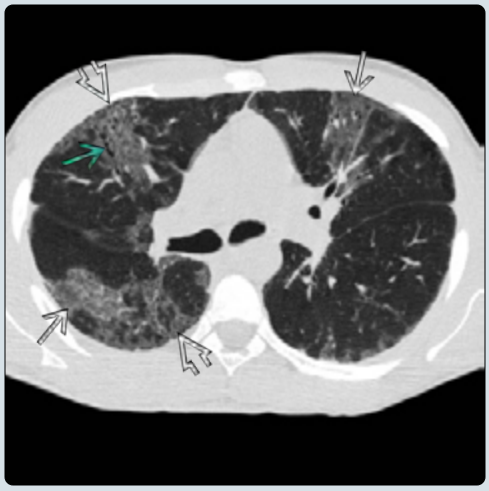
(Left) *AP radiograph in a term neonate presenting with respiratory distress secondary to an ABCA3 gene mutation shows low lung volumes with widespread granular opacities resembling surfactant deficiency of prematurity.* **(Right)** *Coronal HRCT in the same neonate with an ABCA3 mutation shows diffuse ground-glass opacification with areas of interlobular septal thickening.*





(Left) *Axial HRCT from an older infant with a surfactant protein C mutation shows areas of mild ground-glass opacity, numerous parenchymal cysts, & foci of interlobular septal thickening.* **(Right)** *Axial HRCT from a 17-year-old with a known ABCA3 mutation shows areas of ground-glass opacity with concomitant reticulation & traction bronchiectasis, indicating fibrosis.*





TERMINOLOGY

Definitions

- Group of rare lung diseases caused by mutations/deletions of genes affecting surfactant homeostasis
- Most frequent surfactant dysfunction disorders
  - Surfactant protein B (SP-B): *SFTPB* gene
  - Surfactant protein C (SP-C): *SFTPC* gene
  - ATP binding cassette transporter A3 (ABCA3): *ABCA3* gene
  - Receptors for GM-CSF: *CSF2RA*, *CSF2RB* genes
    - Congenital alveolar proteinosis
  - Thyroid transcription factor: *TTF1/NKX2-1* genes
    - Brain-lung-thyroid syndrome
- Pulmonary alveolar proteinosis of adults & older children
  - GM-CSF autoantibodies
  - Alveolar macrophage dysfunction

IMAGING

General Features

- Best diagnostic clue
  - Term infant with radiographic presentation similar to surfactant deficiency of prematurity

Radiographic Findings

- Diffuse or patchy granular opacities

CT Findings

- SP-B
  - Diffuse ground-glass opacification & thickened interlobular septa: Crazy paving pattern
  - Can progress to fibrotic changes with persistent interlobular septal thickening
  - Rarely imaged with CT during severe neonatal presentation due to fragile clinical state
- SP-C or ABCA3
  - Infants
    - Diffuse ground-glass opacification or consolidation with interlobular septal thickening: Crazy paving
  - Older infants & children
    - Ground-glass opacities that ↓ in extent with age
    - Parenchymal cysts that ↑ in number & size with age
    - Interlobular septal thickening
    - Pectus excavatum

DIFFERENTIAL DIAGNOSIS

Surfactant Deficiency Related to Prematurity

- Differentiated clinically based on gestational age
  - Affects ~ 60% born < 28-weeks gestation
  - Affects ~ 5% born at term

Neonatal Pneumonia

- Can be acquired antenatally, perinatally, or postnatally
- Etiologies vary depending on timing of disease acquisition
- Pleural effusion common

Transient Tachypnea of Newborn

- Self-limited illness due to delay in clearance of fetal lung fluid

Alveolar Growth Abnormalities

- Commonly due to prematurity but also observed in congenital heart disease & genetic disorders (e.g., trisomy 21)

PATHOLOGY

General Features

- Etiology
  - Pulmonary surfactant: Complex mixture of lipid (90% by weight) & protein lining alveolar surface
    - Prevents end-expiratory atelectasis

CLINICAL ISSUES

Natural History & Prognosis

- SP-B
  - Acute respiratory distress in full-term infants at birth
  - Transient or modest improvement with surfactant replacement &/or corticosteroid therapy
  - Progressive & usually fatal by 3 -6 months of age with lung transplantation as only effective treatment
- SP-C
  - More commonly associated with diffuse lung disease in older infants, children, & adults
  - Onset of symptoms highly variable: Influenced by mutation as well as environmental factors (e.g. viral infections)
    - Average onset of 2-3 months of age
      - 10-15% present in 1st month of life
      - Presentation in full-term neonates: Similar to surfactant deficiency of prematurity; may be fatal
      - Presentations in older infants: Tachypnea, retractions, hypoxemia, digital clubbing, & failure to thrive
  - Wide ranging long term clinical status: From no supplemental oxygen requirement to death while awaiting transplant
  - Older adults often present with pulmonary fibrosis
- ABCA3
  - Variable: Associated with above phenotypes

Treatment

- SP-B
  - Almost always fatal without lung transplant
  - Rare cases of surviving children with partial defects in SP-B production
- SP-C & ABCA3
  - Treatment varies based on phenotypic manifestations
    - Lung transplant in infancy in severe cases

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KEY FACTS

TERMINOLOGY

- Acquired unilateral pulmonary hypoplasia thought to result from childhood viral infection (postinfectious bronchiolitis obliterans)
- Results in small, hyperexpanded lung with relative decrease in vascularity

IMAGING

- Radiograph
  - Asymmetric hyperlucent lung with diminished number & caliber of vessels
  - May simulate pneumothorax
- HRCT
  - Air-trapping in affected lung
  - Small central & peripheral pulmonary arteries
  - Occasional bronchiectasis with minor subpleural scarring
  - Bronchial wall thickening, usually mild

TOP DIFFERENTIAL DIAGNOSES

- Foreign body aspiration
- Obstructive endobronchial lesion
- Unilateral absence of pulmonary artery
- Bullous disease

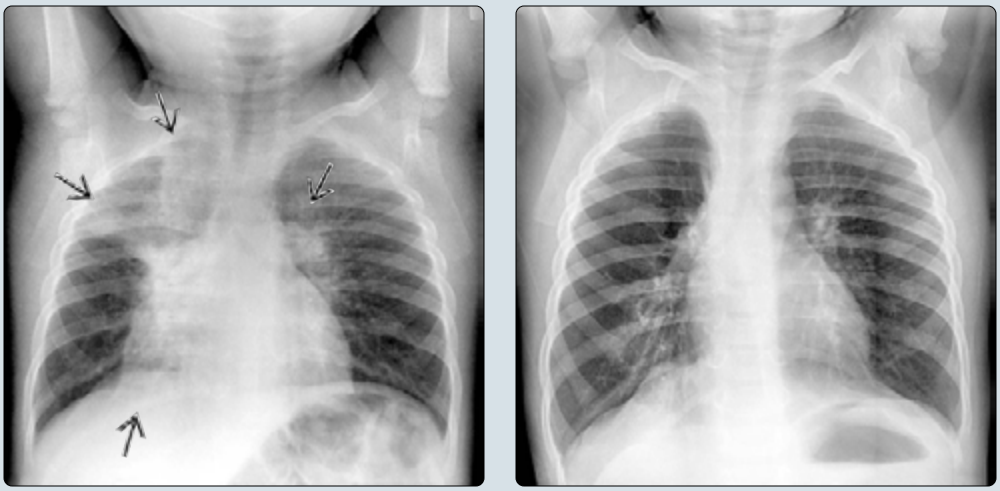
PATHOLOGY

- Inflammation & fibrosis of walls & contiguous tissues of membranous & respiratory bronchioles
- Do not confuse bronchiolitis obliterans (BO) with bronchiolitis obliterans organizing pneumonia (BOOP)

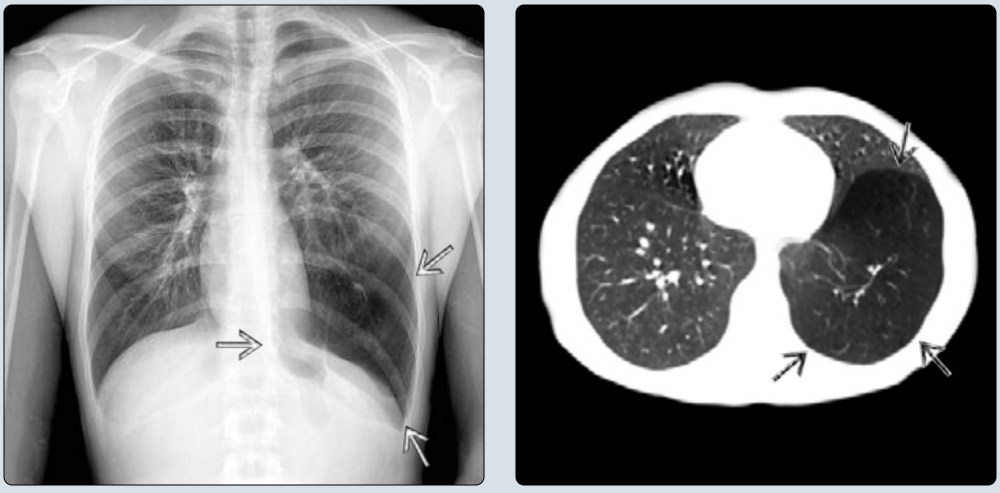
CLINICAL ISSUES

- Frequent respiratory infections in infancy or childhood
- Chronic cough, wheezing, recurrent pneumonia
- Variable presentation depending on severity of disease
- Prognosis usually good
- Treatment supportive

(Left) *Frontal upright radiograph shows a 13 month old with typical symptoms & radiographic findings of viral airway disease, a risk factor for later developing Swyer-James syndrome (SJS). Note the asymmetric geographic areas of air-space opacity & volume loss that mostly involve the right lung.* **(Right)** *8-month follow-up radiograph in the same patient demonstrates asymmetric lucency & oligemia throughout the right lung, consistent with SJS.*



(Left) *Frontal chest radiograph in an 18-year-old patient performed in order to obtain a visa demonstrates focal hyperexpansion, lucency, & oligemia in the left lung base.* **(Right)** *Axial NECT through the lung bases confirms the findings of localized hyperexpansion & oligemia with diminishment of normal lung architecture, consistent with SJS.*



TERMINOLOGY

Abbreviations

- Swyer-James syndrome (SJS)

Synonyms

- Swyer-James-MacLeod syndrome, Brett syndrome, postinfectious bronchiolitis obliterans (BO)

Definitions

- Acquired unilateral pulmonary hypoplasia thought to result from childhood viral infection
- Results in small, hyperexpanded lung with relative decreased vascularity & (rarely) bronchiectasis
- Imaging findings may appear as early as 9 months after initial infectious insult

IMAGING

General Features

- Best diagnostic clue
  - Asymmetric hyperlucent lung with diminished number & caliber of vessels
  - Classically unilateral, but usually multifocal

Radiographic Findings

- Radiography
  - Pronounced patchy unilateral or unilobar hyperlucency (due to oligemia of involved segments)
    - May simulate pneumothorax on radiographs
  - Possible reduced lung volume on inspiration, with air-trapping on expiration

CT Findings

- HRCT
  - Mosaic pattern of attenuation
  - Inspiratory: Affected lung volume small or normal-sized
  - Expiratory: Air-trapping in affected lung
  - Small central & peripheral pulmonary arteries
  - Bronchiectasis with minor subpleural parenchymal scarring
  - Bronchial wall thickening, usually relatively mild

Imaging Recommendations

- Protocol advice
  - Important to do HRCT with expiratory images

DIFFERENTIAL DIAGNOSIS

Foreign Body Aspiration

- Acute presentation, often with supportive history (e.g., recent choking on peanuts)
- Endobronchial foreign body may be visible on CT

Obstructive Endobronchial Lesion

- More in common in older teenagers & adults
- Endobronchial filling defect on CT

Bullous Disease

- Absent pulmonary architecture in bullae

Unilateral Absence of Pulmonary Artery

- Also presents as unilateral small, hyperlucent, oligemic lung
- No air-trapping on expiration (as in SJS)
- VQ scan demonstrates ventilation with no perfusion
- All prior chest radiographs will be abnormal (as opposed to previously normal in SJS)

PATHOLOGY

General Features

- Inflammation & fibrosis of walls & contiguous tissues of membranous & respiratory bronchioles
  - Postobstructive hyperexpansion of terminal air sacs
  - Compensatory decreased perfusion
- Result: Underdevelopment of alveoli & pulmonary arteries
- Main histopathological finding: Constrictive bronchiolitis
- Do not confuse BO with bronchiolitis obliterans organizing pneumonia (BOOP)
  - BOOP is separate entity, a.k.a. cryptogenic organizing pneumonia (COP)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Frequent respiratory infections in infancy or childhood
    - Most common infections: Adenovirus, RSV, influenza A, *Mycoplasma* pneumonia
    - Childhood BO usually postinfectious, but also may be related to other insults such as aspiration, toxic fumes, organ transplantation
  - Chronic cough, wheezing, recurrent pneumonia
- Other signs/symptoms
  - Variable presentations: Range from severe to asymptomatic depending on severity of disease
  - Restrictive pattern on pulmonary function tests

Demographics

- Age
  - Usually presents in infancy or childhood
  - Less severe cases may be missed until adulthood (often found incidentally on radiograph)

Natural History & Prognosis

- ~ 1% of adenovirus-induced bronchiolitis progress to BO
- If insult occurs before alveolar maturation (8 years), may affect number of alveoli & pulmonary vessels
- Prognosis usually good

Treatment

- Treatment supportive
- Pneumonectomy reserved for intractable recurrent infections

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Lymphangioleiomyomatosis

KEY FACTS

TERMINOLOGY

- Cystic disease of lungs & thoracic/retroperitoneal lymphatics due to proliferation of lymphangioleiomyomatosis (LAM) cells
  - LAM cells: Low-grade neoplastic smooth muscle-like cells metastatic from unknown primary tumor
- 2 subtypes nearly identical by histology & imaging
  - Tuberous sclerosis complex-associated (TSC-LAM)
  - Sporadic (S-LAM)

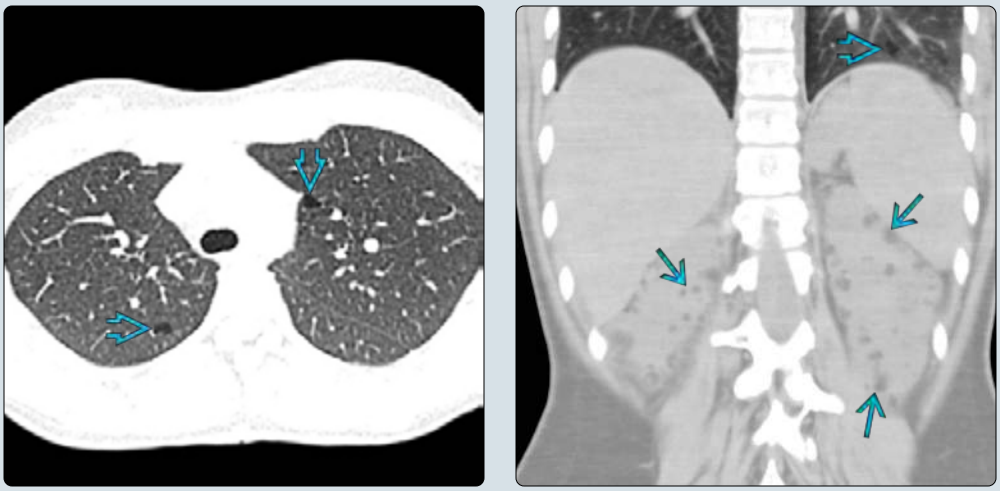
IMAGING

- Chest radiograph
  - Normal to ↑ lung volumes
  - Diffuse bilateral fine reticular opacities
  - ± pneumothorax, pleural effusion
- HRCT/lung CT
  - Numerous randomly distributed thin-walled cysts
    - Range from few scattered cysts to complete lung parenchymal replacement
    - Variable size: Usually 2 mm to 2 cm (may be larger)
      - Expiratory images more sensitive for small cysts
  - Normal intervening parenchyma
  - Small nodules in minority of patients
  - CT underestimates functional disease severity

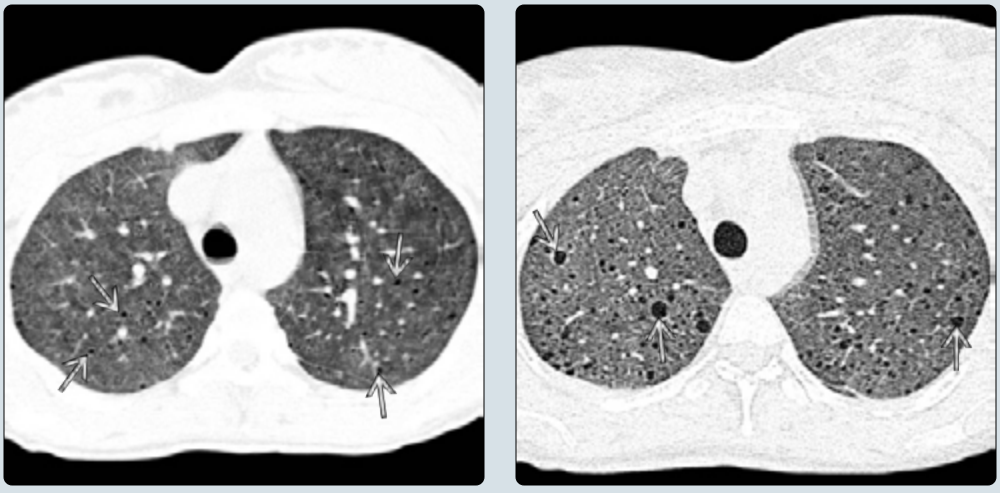
CLINICAL ISSUES

- S-LAM
  - Occurs nearly exclusively in females
  - Mean age of diagnosis: 35 years
- TSC-LAM higher in women than men (42% vs. 13% of TSC cases, respectively)
  - TSC-LAM cysts may be seen in childhood
  - 22% affected by age 20; 80% by age 40
- Most common presentations: Dyspnea on exertion, recurrent pneumothorax
- Gradual pulmonary function deterioration from slowly advancing disease
- Treatments include mTOR inhibitors, lung transplant

(Left) *Axial HRCT of the lungs in a 17-year-old girl with tuberous sclerosis complex (TSC) shows small scattered cysts bilaterally amidst otherwise normal-appearing lung parenchyma. These cysts represent early manifestations of lymphangioleiomyomatosis (LAM).* **(Right)** *Coronal NECT in a 14-year-old patient with TSC shows a small lung cyst in the left lower lobe. Despite the lung window settings, numerous fat-containing lesions can be seen throughout the kidneys, typical of renal angiomyolipomas.*



(Left) *Axial HRCT expiratory image of a 22-year-old patient with TSC-LAM shows numerous scattered 1- to 2-mm round thin-walled cysts with normal intervening lung parenchyma.* **(Right)** *Axial expiratory HRCT image obtained 3 years later in the same patient shows that the cysts have increased in both size & number. The intervening parenchyma remains normal.*



TERMINOLOGY

Definitions

- Lymphangioleiomyomatosis (LAM): Disease characterized by abnormal proliferation of LAM cells in lungs & thoracic/retroperitoneal lymphatics
  - Occurs almost exclusively in women of childbearing age
- 2 main forms
  - Tuberous sclerosis complex-associated LAM (TSC-LAM)
  - Sporadic LAM (S-LAM)

IMAGING

General Features

- Best diagnostic clue
  - Bilateral, randomly distributed thin-walled pulmonary cysts surrounded by normal intervening parenchyma

Radiographic Findings

- Chest radiograph normal in early stages of LAM
- Eventual development of diffuse fine reticular opacities
- Normal to ↑ lung volumes
- ± pneumothorax (39-53%), pleural effusion (10-44%)

CT Findings

- Numerous thin-walled cysts with uniform distribution
  - Variable size: Usually 2 mm to 2 cm (may be larger)
    - Cyst sizes correlate with extent of disease
  - Variable number: From few scattered cysts to complete replacement of lung parenchyma
- Normal intervening lung parenchyma in majority
- Solid or ground-glass 1- to 10-mm nodules in 3-20%, predominantly upper lobe & peripheral
  - Multifocal micronodular pneumocyte hyperplasia
- ± pleural effusion, pneumothorax, thoracic duct enlargement

DIFFERENTIAL DIAGNOSIS

Langerhans Cell Histiocytosis

- Small pulmonary nodules ± cysts

Cystic Fibrosis

- Central & peripheral bronchiectasis, greatest in upper lobes

Bronchopulmonary Dysplasia

- History of prematurity + prolonged oxygen requirement

Papillomatosis

- Due to vertical transmission of human papillomavirus

PATHOLOGY

General Features

- Etiology
  - Abnormal neoplastic smooth muscle-like cell (LAM cell)
  - Evidence supports LAM cell metastases as cause for pulmonary manifestations
    - Clonal origin consistent across affected organs
    - Recurrence in transplanted lungs shows original host TSC gene mutations
    - Primary lesion unknown: Renal angiomyolipomas (AML) vs. uterine LAM cells
- Genetics
  - TSC-LAM: 2-hit Knudson hypothesis of (sporadic superimposed on inherited germline) mutations causing complete functional loss of *TSC1* or *TSC2* genes
    - *TSC1* gene product: Hamartin protein
    - *TSC2* gene product: Tuberin protein
    - Hamartin & tuberin necessary for functioning **m**ammalian **t**arget **o**f **r**apamycin (mTOR) signaling pathway
  - S-LAM: 2 sporadic somatic hits to *TSC2*

Extrapulmonary Findings

- S-LAM
  - Renal angiomyolipomas (AMLs) in 30% (usually small)
  - Chylous ascites, uterine leiomyomas, liver & pancreas cysts, abdominal/pelvic cystic lymphangioleiomyomas
- TSC-LAM
  - Renal AMLs in 80% (usually larger than in S-LAM) + renal cysts
  - Hepatic & pancreatic lesions, PEComas
  - Cardiac rhabdomyomas (majority involute over time)
  - Cerebral > cerebellar cortical/subcortical tubers, white matter lesions, subependymal nodules, subependymal giant cell astrocytomas
  - Other: Retinal astrocytomas/hamartomas, facial angiofibromas, ash leaf spots, shagreen patches, subungual fibromas, cystic & sclerotic bone lesions

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Progressively worsening dyspnea on exertion in women of childbearing age
  - Spontaneous pneumothorax, often recurrent

Demographics

- Age
  - S-LAM mean age of diagnosis: 35 years
  - TSC-LAM lung disease can be seen on CT in childhood
    - 22% affected by age 20; 80% by age 40

Natural History & Prognosis

- Progressive pulmonary function deterioration from slowly advancing lung disease
- Most recent 10-year survival reported to be 79-91%

Treatment

- Primarily symptomatic (e.g., evacuating pleural effusion, pneumothorax; bronchodilators)
- Conflicting results on antiestrogen therapies
- Sirolimus/rapamycin (mTOR inhibitor)
- Lung transplant

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Langerhans Cell Histiocytosis, Pulmonary

KEY FACTS

TERMINOLOGY

- Neoplastic proliferation of monoclonal Langerhans cells leading to formation of destructive granulomas

IMAGING

- Diffuse lung involvement typical in children
  - Classic sparing of lower lungs/costophrenic sulci applies to adult pulmonary Langerhans cell histiocytosis (LCH)
    - Should not be used as diagnostic criterion in pediatrics
- CXR: Reticulonodular pattern of opacities
- CT: Combination of small nodules & cysts
  - Irregularly shaped nodules in centrilobular, peribronchial, or peribronchiolar locations
  - Cysts with variable wall thickness
- Chronic LCH: Fibrosis & traction emphysema
- May also see osseous, thymic, hepatic, & splenic involvement on chest imaging
  - High likelihood of multisystem LCH if lung disease present in child

TOP DIFFERENTIAL DIAGNOSES

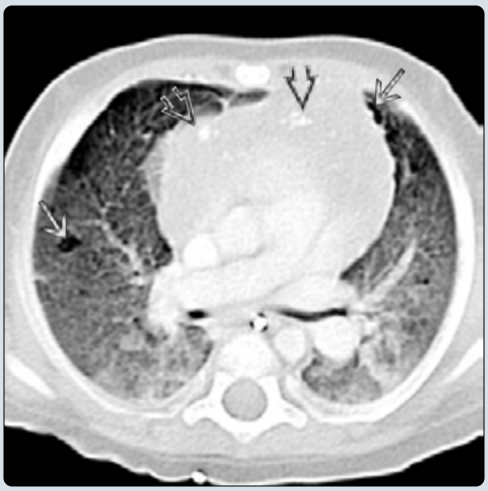
- Cystic fibrosis
- Papillomatosis
- Lymphangioleiomyomatosis
- Sarcoidosis

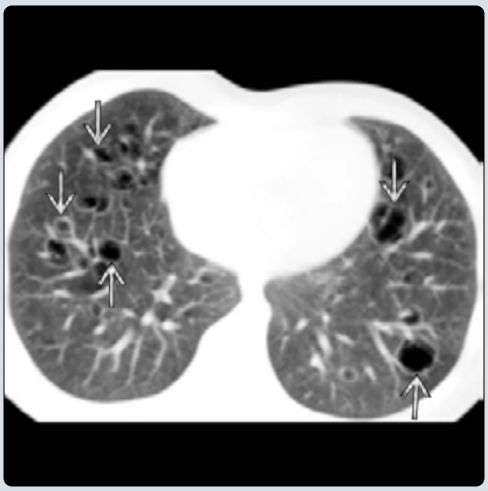
CLINICAL ISSUES

- May present at any age in childhood
- Typically occurs with multisystem disease in children
  - Lung no longer considered risk organ for worse prognosis in LCH
- Leads to end-stage lung disease in 27% of cases
- HRCT helpful to follow disease progression in young children as pulmonary function tests less reliable

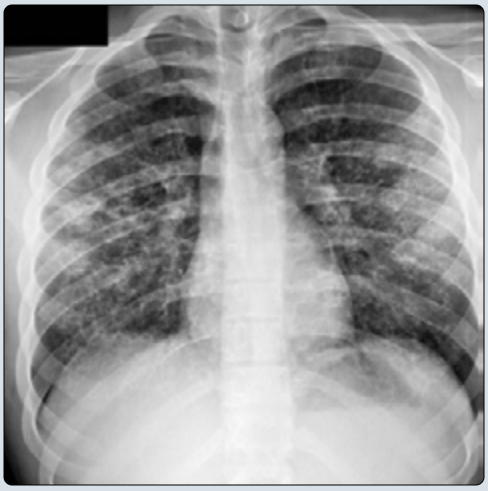
DIAGNOSTIC CHECKLIST

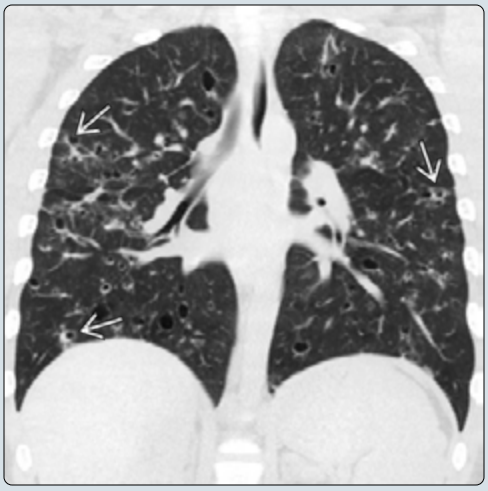
- Pediatric LCH not to be confused with adult pulmonary LCH (typically single system disease associated with smoking)





(Left) *Axial HRCT of a 7-month-old girl shows bilateral pulmonary cysts ➡ within diffusely opacified lungs. The thymus is also involved by Langerhans cell histiocytosis (LCH) in this patient, resulting in scattered thymic Ca^{2+} ⇨. Follow-up CT imaging (not shown) demonstrated clear lungs & resolved cysts after systemic therapy.* **(Right)** *Axial NECT of a 14-year-old boy with pulmonary LCH shows numerous cysts of varying size & wall thickness ➡ scattered throughout the lungs.*





(Left) *PA chest radiograph in a 14-year-old boy with pulmonary LCH shows nodular opacities intermixed with reticulation secondary to peribronchovascular cysts.* **(Right)** *Coronal CT in a 14-year-old boy with pulmonary LCH shows both nodules & cysts. Note the peribronchovascular distribution of cysts ➡ reflecting the bronchiolocentric nature of the disease.*

TERMINOLOGY

Abbreviations

- Langerhans cell histiocytosis (LCH)

Definitions

- Neoplastic proliferation of monoclonal Langerhans cells in 1 or more organ systems
  - Leads to formation of destructive granulomas

IMAGING

General Features

- Best diagnostic clue
  - Combination of small pulmonary nodules & cysts

Radiographic Findings

- Early findings
  - Characteristic reticulonodular pattern (due to summation of nodules & thin-walled cysts)
  - Lung volumes normal or ↑ (unlike many other interstitial diseases)
- Late findings
  - Honeycomb-like pattern with architectural distortion (due to summation of air-filled cysts)
- ± pneumothorax, pneumomediastinum, pleural effusion

CT Findings

- HRCT
  - Progression: Nodules → cavitary nodules → thick-walled cysts → thin-walled cysts → confluent cysts
    - Irregularly shaped nodules in centrilobular, peribronchial, or peribronchiolar locations
    - Cysts show variable wall thickness
  - Diffuse disease common in children
    - Classic sparing of lower lungs/costophrenic sulci should not be used as diagnostic criterion in pediatrics
  - Chronic LCH: Fibrosis & traction emphysema
  - May also see osseous, thymic, hepatic, & splenic involvement on chest CT

DIFFERENTIAL DIAGNOSIS

Cystic Fibrosis

- Patchy, upper lobe predominant disease
- Cylindrical to cystic bronchiectasis
- Bronchial wall thickening, mucous plugging

Papillomatosis

- Nodules intermixed with thick-walled cysts
- Nodules of larynx & trachea more common than lung involvement

Lymphangioleiomyomatosis

- Small, round, thin-walled cysts with normal surrounding parenchyma
- No nodules (unless multifocal micronodular pneumocyte hyperplasia present)

Sarcoidosis

- Apical predominant disease with ground-glass opacities, fibrotic change, subpleural honeycombing
- Hilar adenopathy typical

PATHOLOGY

General Features

- Pulmonary LCH: Proliferation of Langerhans cells in bronchial/bronchiolar epithelium
  - Cavitating granulomas adjacent to small airways
  - Lead to airway obstruction, air trapping, cystic change
- Diagnosed by bronchoalveolar lavage, possible biopsy
- Electron microscopy: Birbeck granule in cytoplasm of 40% of Langerhans cells
  - Classic "tennis racquet" organelle
- Immunostaining: CD1a, CD207, S100 protein positive

Staging, Grading, & Classification

- Multisystem LCH
  - Pulmonary involvement in 23-50%
  - Most pediatric pulmonary LCH occurs in multisystem disease
  - Lung disease no longer denotes risk organ involvement with worse prognosis
- Single system LCH
  - Pulmonary involvement in < 10% of pediatric patients

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Tachypnea, dyspnea, wheezing
- Clinical profile
  - Not associated with smoking (unlike adult LCH)

Natural History & Prognosis

- Leads to end-stage lung disease in 27% of cases

Treatment

- Corticosteroids & other chemotherapeutic agents

DIAGNOSTIC CHECKLIST

Consider

- Pediatric LCH not to be confused with adult pulmonary LCH
- Recommend chest radiography when LCH discovered in any body system
- Degree of pulmonary involvement on CT correlates with functional impairment
  - HRCT helpful to follow disease progression in young children as pulmonary function tests less reliable

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Asthma

KEY FACTS

TERMINOLOGY

- Chronic, reversible, paroxysmal airway hyperresponsiveness leading to airflow obstruction
- Not single disease; rather, umbrella term for numerous phenotypes in which normally harmless environmental allergens cause airway hyperresponsiveness due to pathologic immune-mediated host response

IMAGING

- Usually normal; may have symmetric hyperexpansion with flattened hemidiaphragms & ↑ retrosternal airspace
- Usually not necessary but helps exclude complications & mimics
- Consider chest radiograph if poor response to therapy

TOP DIFFERENTIAL DIAGNOSES

- Viral bronchiolitis: May be impossible to differentiate radiographically & clinically
- Foreign body aspiration: Static asymmetric lung volumes on bilateral decubitus or inspiratory/expiratory imaging
- Cystic fibrosis: Focal disease with bronchiectasis & mucous plugging, most common in upper lobes

PATHOLOGY

- Risk factors include frequent symptoms in 1st year of life, maternal smoking or history of asthma, & signs of atopy

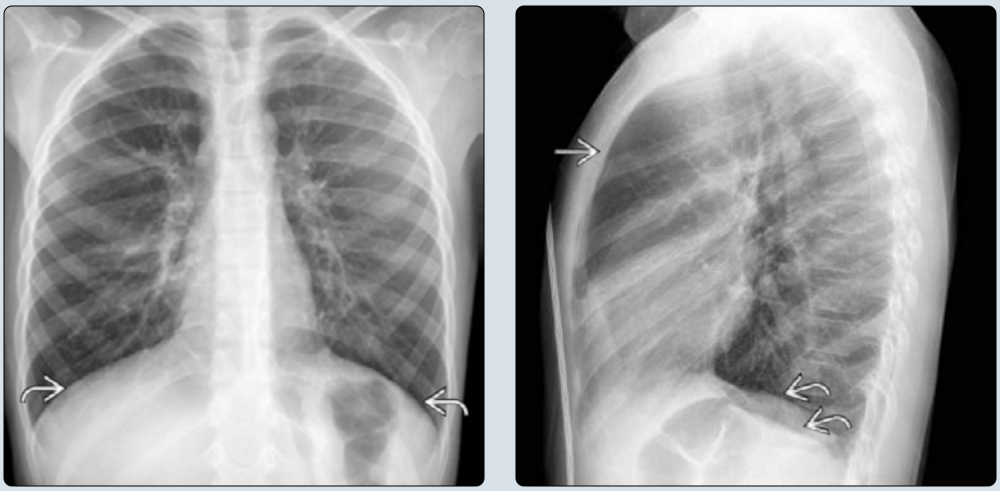
CLINICAL ISSUES

- Intermittent wheezing before age 6 is usually benign & typically resolves within few years
- Severity of asthma symptoms between ages of 7-10 years predicts persistence into adulthood

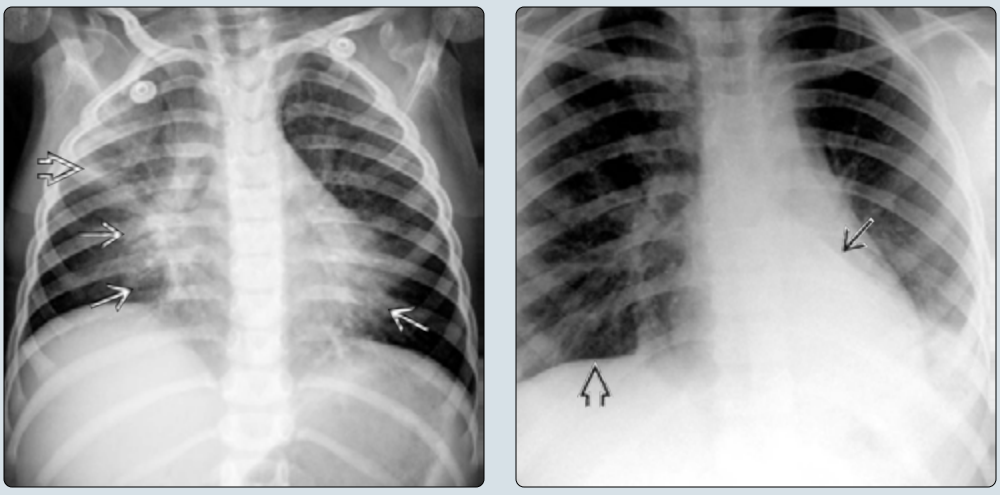
DIAGNOSTIC CHECKLIST

- Radiographs usually normal; indicated to exclude suspected alternate diagnosis, complication, or poor response to therapy

(Left) *AP chest radiograph in a 9-year-old boy with severe asthma who presented with respiratory distress & decreased breath sounds shows overall hyperexpansion & flattening of the hemidiaphragms.* **(Right)** *Lateral radiograph from the same patient shows an enlarged retrosternal airspace & flattening of the hemidiaphragms.*



(Left) *Frontal chest radiograph in a younger child with an acute asthma exacerbation shows an irregular cardiac contour due to multifocal subsegmental atelectasis & peribronchial thickening.* **(Right)** *Frontal chest radiograph in an older child shows a triangular-shaped homogeneous opacity due to left lower lobe collapse with loss of visualization of the left hemidiaphragm (as compared to the sharply defined right hemidiaphragm).*



TERMINOLOGY

Synonyms

- Bronchial asthma
- Airway hyperreactivity
- Reactive airway disease

Definitions

- Not single disease; rather, umbrella term for numerous phenotypes in which normally harmless environmental allergens cause airway hyperresponsiveness due to pathologic immune-mediated host response
- Airway hyperresponsiveness → contraction of bronchial wall smooth muscle & cascade of inflammation → acute, reversible, airway narrowing & airflow obstruction

IMAGING

General Features

- Best imaging clue
  - Symmetric lung hyperexpansion: Flattening of hemidiaphragms, ↑ retrosternal airspace

Radiographic Findings

- Radiography
  - Not part of most acute childhood asthma algorithms; reserved for those with fever, suspected foreign body (FB) aspiration, failure to improve with treatment, or focal findings on physical exam
  - Most common finding: Normal chest radiograph
  - Next most common: Subtle & nonspecific signs of hyperinflation, usually symmetric
    - Flattening of hemidiaphragms
    - ↑ AP diameter of chest & retrosternal airspace
    - ± bulging intercostal spaces
  - Less common findings
    - Peribronchial thickening/cuffing
    - Atelectasis & lobar/partial lobar collapse
    - Irregular cardiac contour = "shaggy heart"
    - Peripheral oligemia
  - Chronic findings
    - Bronchiectasis, mucous plugging
  - Complications: More frequent in younger children (as smaller bronchi are more easily narrowed or occluded) & more likely with concurrent viral bronchiolitis
    - Barotrauma (pneumomediastinum, subcutaneous emphysema, & rarely pneumothorax)
    - Lobar collapse, segmental & subsegmental atelectasis
    - Secondary allergic bronchopulmonary aspergillosis
    - Pneumonia

CT Findings

- HRCT
  - Generally not indicated, particularly in acute asthma
  - Signs of airway narrowing
    - Bronchial wall thickening
    - Narrowing or dilation of bronchial lumen
    - Mucous plugging
  - Signs of abnormal aeration
    - Mosaic attenuation due to combination of regional air-trapping & oligemia
    - Focal peripheral air-trapping on expiratory images
  - Signs of airspace disease
    - Segmental & subsegmental atelectasis
    - Centrilobular opacities
  - Bronchiectasis suggests asthma mimics (cystic fibrosis, ciliary dyskinesias, or immune deficiencies)

MR Findings

- Hyperpolarized noble gas MR shows promise in measuring ventilation volumes; findings also correlate with disease severity, lung function, symptom control, & level of inflammatory marker elevation

Imaging Recommendations

- Imaging generally not recommended except in
  - Febrile children (suspected complicating pneumonia)
  - Suspected FB aspiration
  - Those who fail to improve with treatment
  - Suspected barotrauma or lung collapse

DIFFERENTIAL DIAGNOSIS

Viral Bronchiolitis

- Often acts as precipitating trigger for asthma & may be impossible to differentiate radiographically or clinically
- Look for preceding or concurrent symptoms of viral upper respiratory infection (e.g., nasal congestion, low-grade fever)

Foreign Body Inhalation or Ingestion

- Vast majority of aspirated FBs not radiopaque
- Stable air-trapping or collapse on serial radiographs, usually unilateral
- Static lung volumes on bilateral decubitus or inspiratory/expiratory imaging
- Persistent symptoms that do not respond to bronchodilator therapy

Croup

- Stridor rather than wheezing
- Barking cough
- Narrowed subglottic trachea

Cystic Fibrosis

- Early bronchial wall thickening that progresses to bronchiectasis
- Persistent hyperinflation or recurrent consolidation
- Mucous plugging of dilated bronchi
- Focal disease most common in upper lobes

Ciliary Dyskinesias

- Situs inversus or dextrocardia (50%), paranasal sinusitis, & bronchiectasis
- Recurrent pneumonias

Immune Deficiencies

- Recurrent pulmonary infection → bronchiectasis

Vascular Sling

- Symptoms often present from birth
- No response to bronchodilator therapy
- Abnormal impression of left pulmonary artery coursing between trachea & esophagus
- Often has associated complete cartilage rings of trachea (appearing round & narrow in cross section)

PATHOLOGY

General Features

- Etiology
  - Due to complex interplay of environmental triggers & host factors
    - Environmental triggers
      - Cockroach & pet dander; dust mites in bedding, carpets, furniture
      - Inhaled allergens & irritants (tobacco smoke, household chemicals, mold, pollen, etc.)
      - Cold, dry weather
    - Host factors
      - Genetic predisposition
      - Upper respiratory tract viral illnesses
      - Exercise & strong emotional states (anger, anxiety, fear)
      - Hormonal fluctuations
  - Complex response to these triggers causes release of inflammatory mediators from mast cells, macrophages, eosinophils, epithelial cells, & activated T lymphocytes
    - Bronchial wall smooth muscle contraction
    - ↑ production of excessive mucus
- Risk factors
  - Frequent symptoms in 1st year of life
  - Maternal smoking & history of asthma
  - Eczema, atopy, elevated IgE levels
  - Low birthweight
  - Male sex

Gross Pathologic & Surgical Features

- Bronchial wall thickening
- Mucous plugging of airway lumen

Microscopic Features

- Inflammatory cell infiltration & edema of airway wall
- Mucous gland hyperplasia

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Chronic &/or recurrent cough, wheezing
  - Shortness of breath
  - Chest tightness
- Other signs/symptoms
  - ↓ peak expiratory flow rates & forced expiratory volume in 1-second (FEV_1) values
  - Symptomatic improvement with bronchodilation therapy
  - Use of accessory muscles to breathe at rest
  - Exercise limitation

Demographics

- Age
  - Prevalence peak between 6-11 years
- Gender
  - M > F = 1.5:1.0 before puberty
  - M < F = 1.0:1.5 after puberty
- Epidemiology
  - Most common chronic disease of childhood
  - Prevalence 1-30% worldwide
  - Positive correlation with urbanization
  - Increasing incidence worldwide with increasing mortality
  - In USA, asthma more common in African American & Hispanic children

Natural History & Prognosis

- Prognosis usually excellent with appropriate treatment
- Wheezing before age 6 usually benign, usually resolves within few years
- Severity of asthma symptoms between ages of 7-10 years predictive of persistence into adulthood
  - Small subgroup (30%) of children characterized by signs of atopy, severe & persistent symptoms at young age, & maternal history of smoking or asthma

Treatment

- Avoidance of exposure to known precipitating environments
- Inhaled β-agonists for bronchospasm
- Inhaled & oral corticosteroids that dampen inflammatory response
  - Used to prevent acute exacerbations & for treatment of chronic asthma
- Inhaled mast cell stabilizers that prevent release of mediators from mast cells that cause airway inflammation & bronchospasm

DIAGNOSTIC CHECKLIST

Consider

- Radiographs usually normal but indicated with poor response to therapy, suspected complication, or entertaining alternate diagnosis
- Hyperinflation with varying degrees of atelectasis
- Difficult to distinguish from viral bronchiolitis

Image Interpretation Pearls

- Look for asthma mimics & signs of complication

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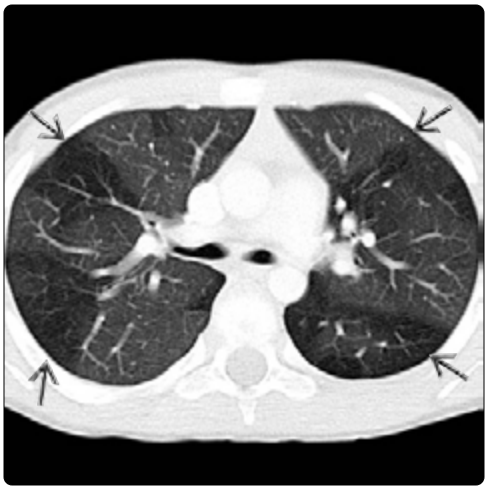
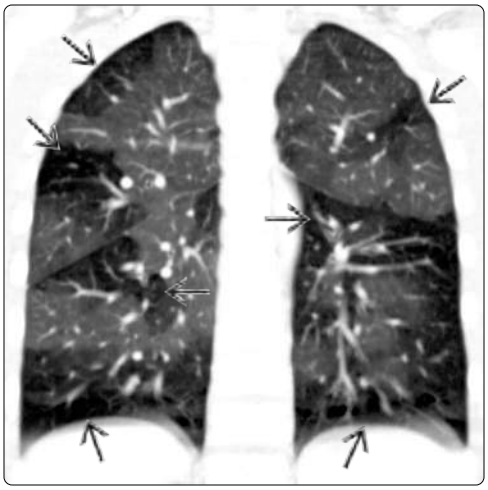
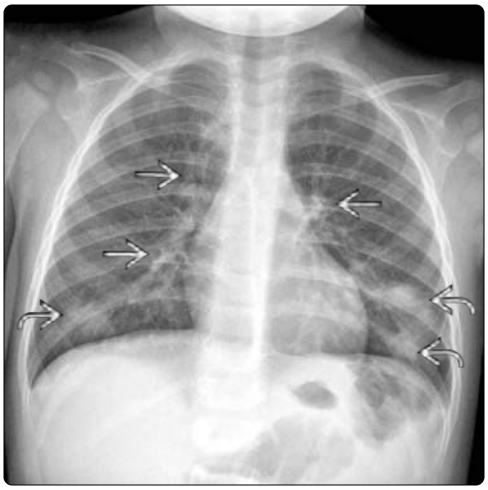
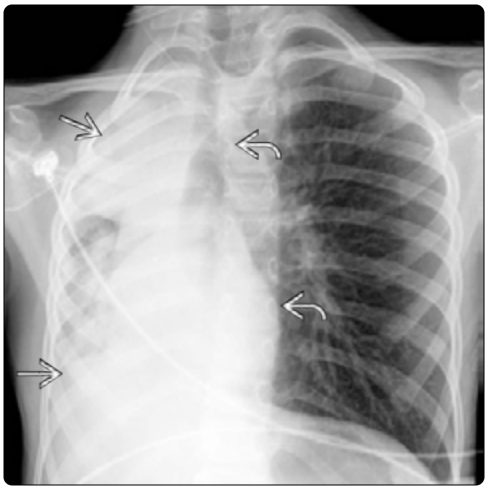


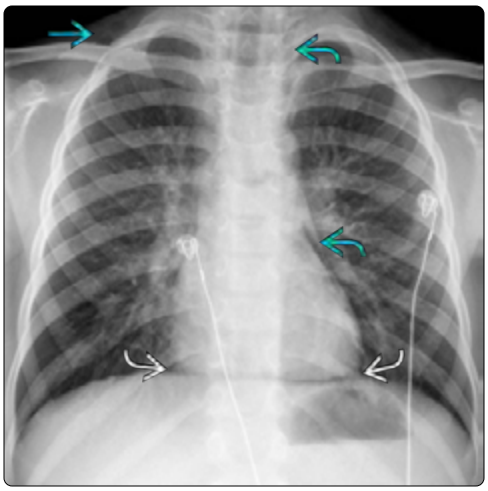


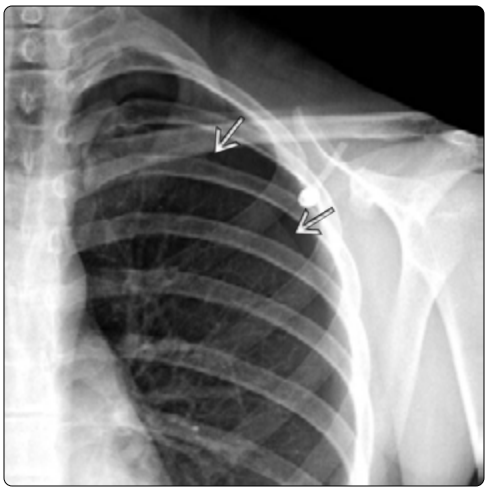
(Left) *Axial CECT in a 9-year-old boy with chronic asthma shows mosaic attenuation with subsegmental areas of relative lucency ⇨ secondary to air trapping & oligemia.* **(Right)** *Coronal CECT reconstruction in the same patient shows the widespread nature of the mosaic attenuation ⇨.*





(Left) *PA chest radiograph from a young child with wheezing who was later diagnosed with asthma shows peribronchial airway thickening ➡ & multifocal atelectasis ➡ bilaterally.* **(Right)** *Frontal radiograph from a child with chronic asthma complicated by plastic bronchitis (which is the development of luminal casts, in this case due to inflammation) that caused bronchial occlusion & near total right lung collapse shows widespread opacity ➡ & shift of the mediastinum to the right ➡.*





(Left) *PA chest radiograph in a child with an acute asthma exacerbation complicated by pneumomediastinum ⇨ shows the continuous diaphragm sign caused by gas in the inferior mediastinum ➡. Subcutaneous emphysema is also seen in the right supraclavicular region ⇨.* **(Right)** *Coned-down image from a frontal chest radiograph in another patient shows another sign of asthma complicated by barotrauma: A small pneumothorax ➡ overlies the left apex.*

Bronchial Obstruction

KEY FACTS

TERMINOLOGY

- Foreign body aspiration, foreign body (FB)
- Complete or partial bronchial occlusion by aspirated FB

IMAGING

- Vast majority of aspirated FBs are **not** radiopaque
- Look for unilateral static lung volume on chest radiographs
  - In uncooperative patients (most common), obtain frontal view + bilateral decubitus images: Look for lack of passive deflation of dependent lung, suggesting air-trapping
  - In cooperative patients, radiographs can be obtained at maximum inspiration & expiration: Look for lack of ↓ expiratory volume on affected side
- Volume of affected lung segments can be normal, ↑, or ↓
- Consider CT with multiplanar reconstructions & 3D virtual bronchoscopy in cases with persistent clinical suspicion & negative chest radiographs

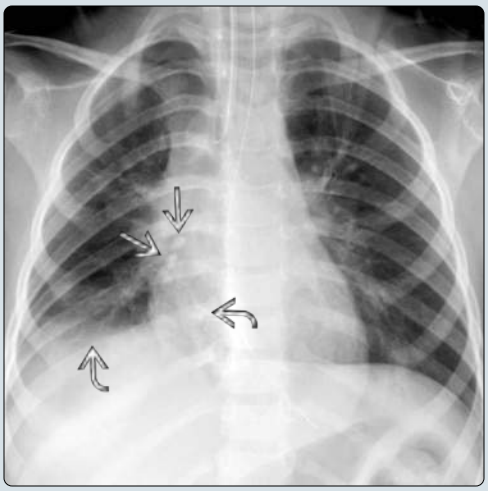
TOP DIFFERENTIAL DIAGNOSES

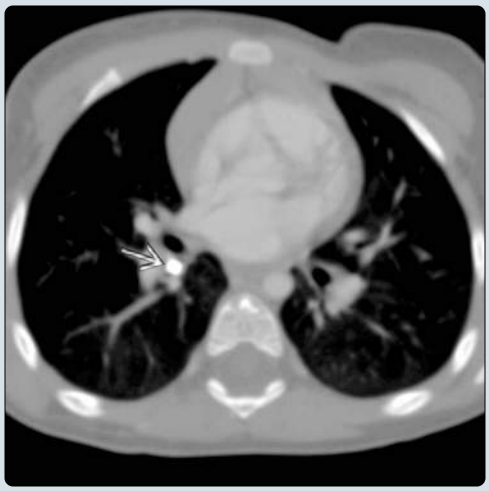
- Refractory asthma
  - Very common; ↑ peribronchial markings & symmetrically hyperexpanded lungs ± foci of atelectasis
- Viral lower respiratory tract infection
  - Very common; ↑ peribronchial markings & symmetrically hyperexpanded lungs ± foci of atelectasis
- Pulmonary sling
  - Much less common than FB; typically presents with respiratory distress at birth & asymmetric lung volumes
- Congenital lobar emphysema
  - Lobar hyperexpansion, generally since newborn period or early infancy

CLINICAL ISSUES

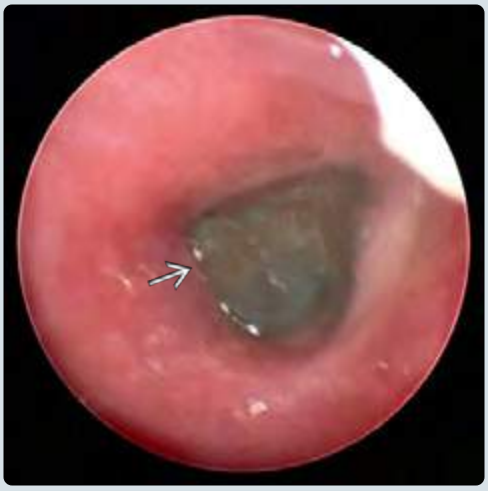
- Typically presents with wheezing, cough, sometimes fever
- More indolent symptoms if presentation delayed (10-25%)
- Delay in diagnosis associated with ↑ complication rate
- Treatment: Bronchoscopic removal of FB

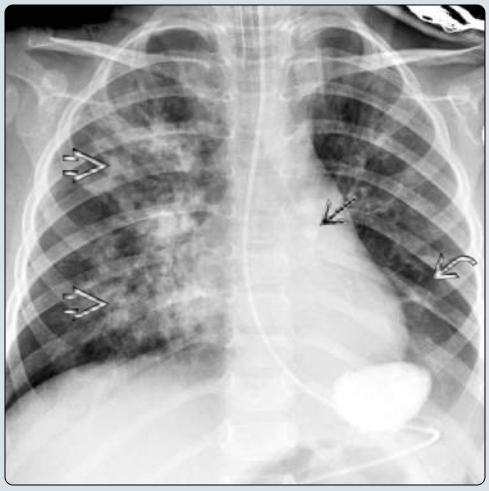
(Left) *Portable chest radiograph of a 3-year-old patient who aspirated glass fragments during a car crash shows 3 small radiopaque foreign bodies in the right lower lobe bronchus. There is resulting airspace opacity as well as volume loss causing elevation of the right hemidiaphragm & slight mediastinal shift.* **(Right)** *Axial CECT in the same child shows a square-shaped radiopaque glass foreign body in a segmental bronchus of the right lower lobe.*





(Left) *Bronchoscopic image from the same patient shows complete occlusion of the segmental bronchus by glass fragments.* **(Right)** *Portable chest radiograph of a toddler found unconscious in a debris pile after a flood shows a small, well-defined opacity in the left main bronchus. The left lower lobe is relatively hyperlucent due to air-trapping, & there are widespread patchy airspace opacities due to aspiration. The bronchial obstruction was due to a small aspirated stone.*





TERMINOLOGY

Abbreviations

- Foreign body aspiration (FBA)
- Foreign body (FB)

Definitions

- Complete or partial bronchial occlusion by aspirated FB

IMAGING

General Features

- Best diagnostic clue
  - Visualization of radiopaque FB in airway (rare)
  - Unilateral static lung volume on inspiratory/expiratory or bilateral decubitus chest radiographs
- Location
  - Most FBs lodge in main bronchi
    - Bronchial 76%, laryngeal 6%, tracheal 4%
    - Right bronchi (58%) > left bronchi (42%)
      - Due to straighter course & larger caliber of right vs. left main bronchi
- Size
  - Most FB: 5-12 mm

Radiographic Findings

- Radiography
  - Best direct evidence: Radiopaque FB
    - However, most FBs are **not** radiopaque
  - Best indirect evidence is static unilateral lung volume on inspiration/expiration or bilateral decubitus imaging
  - Volume of affected lung segments can be normal, ↑, or ↓ (i.e., larger lung not always abnormal)
    - Hyperinflation & oligemia from air-trapping
      - ± depressed hemidiaphragm, widening of intercostal spaces, mediastinal shift towards opposite side
    - Atelectasis from total bronchial obstruction, pneumonia from superinfection
      - ± elevation of hemidiaphragm, narrowing of intercostal spaces, ipsilateral mediastinal shift with atelectasis
  - Pneumothorax & pneumomediastinum from tracheobronchial laceration
  - Reported incidence of chest radiograph findings in FBA
    - Normal: 14-35%
    - Hyperinflation: 21-43%
    - Opacification/atelectasis: 18-29%
    - Mediastinal shift: 10-37%
    - Radiopaque FB: 3-23%
- Bilateral decubitus radiographs
  - Obtain frontal view + bilateral decubitus images
  - Normal, nonobstructed lung will become smaller & more opaque when dependent
  - Abnormal, obstructed lung will remain inflated & relatively lucent when dependent
  - Most appropriate maneuver for infants & toddlers who cannot cooperate with inspiratory & expiratory imaging
- Inspiratory/expiratory radiographs
  - In patients who are cooperative, radiographs can be obtained at maximum inspiration & expiration
    - Useful in minority of patients as this clinical scenario most often occurs from 1-2 years of age
  - Volume of nonobstructed lung diminishes significantly with expiration

Fluoroscopic Findings

- Child fluoroscoped in supine frontal view
  - Normally, both hemidiaphragms will move superiorly & inferiorly in synchronous manner
  - With obstruction, lung volume in affected lung static with decreased or no motion of ipsilateral hemidiaphragm
- Fluoroscopy with forced expiration
  - Technique has been described where evaluator places gloved hand on child's abdomen & gently applies pressure immediately prior to & while obtaining image
  - Pressure drives diaphragms superiorly
  - Abnormal side with bronchial obstruction will remain static in volume
  - Normal side will show elevation of diaphragm as compared to initial neutral radiograph
  - Generally not recommended

CT Findings

- CT not typically advocated in imaging algorithm for suspected FB; may be obtained for
  - Persistent lung collapse or pneumonia
  - Work-up of suspected extrinsic airway compression
- Some advocate CT with multiplanar reconstructions & 3D virtual bronchoscopy in cases with persistent clinical suspicion & negative chest radiograph
  - May see FB as filling defect in bronchus ± focal hyperinflation or atelectasis

Imaging Recommendations

- Best imaging tool
  - Chest radiographs
- Protocol advice
  - Normal inspiratory chest radiograph alone does not exclude aspirated FB: Look for unilateral static lung volume with bilateral decubitus or inspiratory/expiratory imaging

DIFFERENTIAL DIAGNOSIS

Refractory Asthma

- Much more common than bronchial FB
- ↑ peribronchial markings
- Hyperinflation typically symmetric

Viral Lower Respiratory Tract Infection

- Much more common than FB
- ↑ peribronchial markings
- Hyperinflation typically symmetric

Pulmonary Sling

- Much less common than FB
- Chronic; often presents with respiratory distress at birth
- Often associated with other congenital heart disease & complete tracheal rings
- Only vascular ring that results in asymmetric aeration
  - Either side may be larger

Extrinsic Tracheal Compression by Mass

- Bronchogenic cysts, lymphadenopathy, & other masses may compress bronchi & present with asymmetric aeration

Swyer-James Syndrome

- Bronchiolitis obliterans
- Asymmetric lung hyperlucency
  - Affected lucent lung may be smaller than contralateral lung

Congenital Lobar Overinflation

- Gradually increasing symmetric lung hyperlucency confined to single lobe
- Generally presents in newborn period or early infancy

PATHOLOGY

General Features

- Etiology
  - Young children explore environment with their mouth, often putting discovered items in mouth
  - Young infants may be given FBs or inappropriate foods by older siblings
  - Lack of molars & poor swallowing coordination predispose infants to FBA
  - Older children have tendency to talk, laugh, & be active while chewing
- Mechanism
  - Aspirated FB lodges in bronchus & leads to partial or intermittent obstruction
  - May have "ball valve" effect leading to
    - Air-trapping & hyperinflation
    - Complete obstruction leading to atelectasis & collapse
- Typology of FBs
  - Vast majority organic or plastic & therefore radiolucent
    - Peanuts & seeds most common
    - Fruit, meat, plastic, soil, other radiolucent items less common
    - Teeth, metallic items rarely

Gross Pathologic & Surgical Features

- FB lodged in tracheobronchial tree
  - Dried foods absorb water & may swell
  - Peanuts & tree nuts elicit most airway irritation
  - Leukocyte infiltration & edema in surrounding bronchial wall
  - Chronic FB leads to granuloma formation/granulation tissue

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - FBA may present acutely or in delayed fashion; event often unwitnessed or not remembered until later
    - Same day (25%): Wheezing, cough, ± fever
    - Day 2-7 (45%): Indolent cough, medically refractory wheezing, dyspnea
    - Delayed by > 1 week (30%): Same as above

Demographics

- Age
  - Most common age: 1-3 years; peak: 18 months
- Gender
  - FBA more common in boys (2:1)

Natural History & Prognosis

- High degree of suspicion important
- Delay in diagnosis associated with ↑ risk of major complications
  - Incidence of major complications low if rapidly diagnosed
  - Complications: 4% at > 4 days, 91% > 30 days
- Complications of chronic FBs
  - Bronchopulmonary fistula, bronchial rupture, damage to distal lung
- Death rare, ~ 100 per year in USA
  - In 1 series of 2,165 pediatric autopsies, only 10 deaths due to FBA

Treatment

- Endobronchial removal of FB

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Unilateral static lung volume at different phases of respiratory cycle (need more than inspiratory chest radiograph alone)

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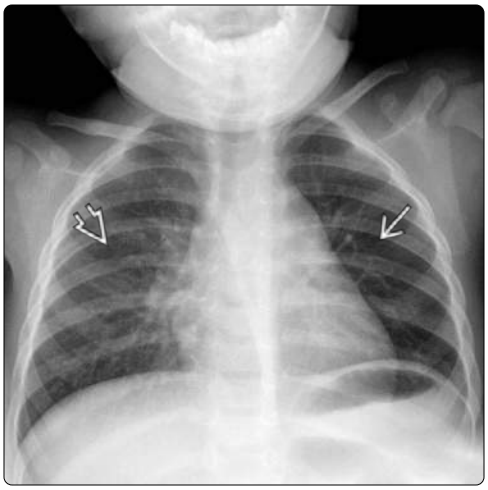
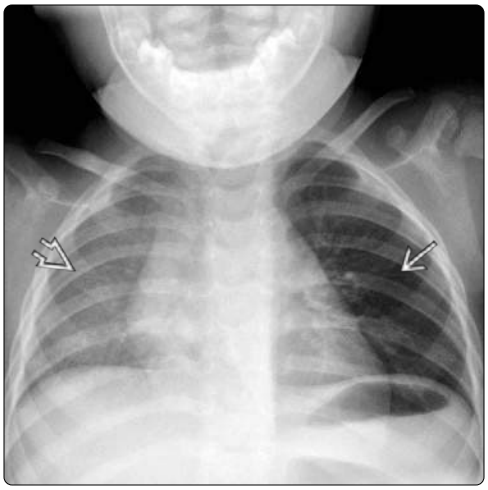
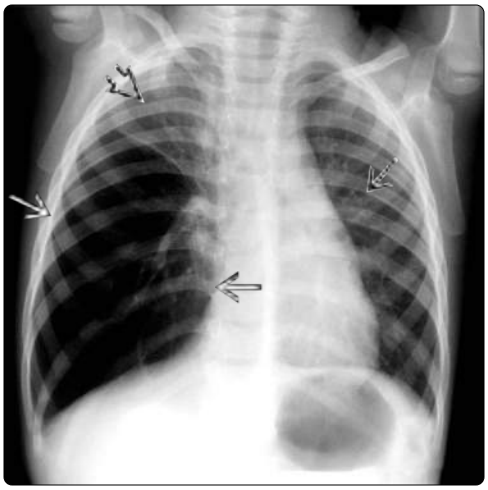
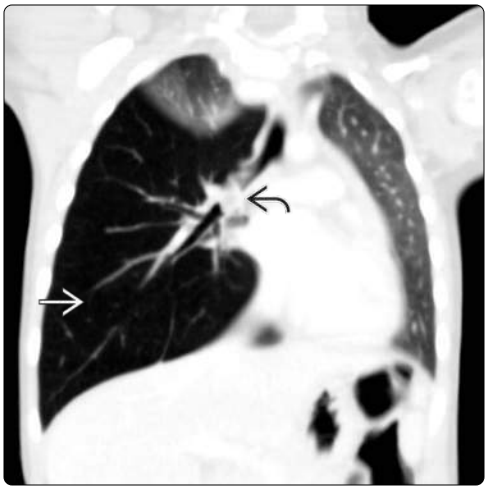


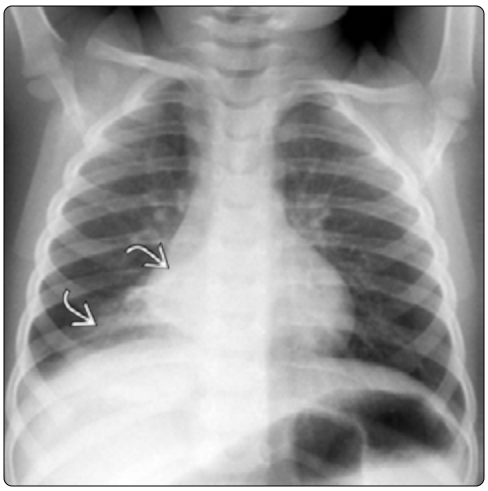


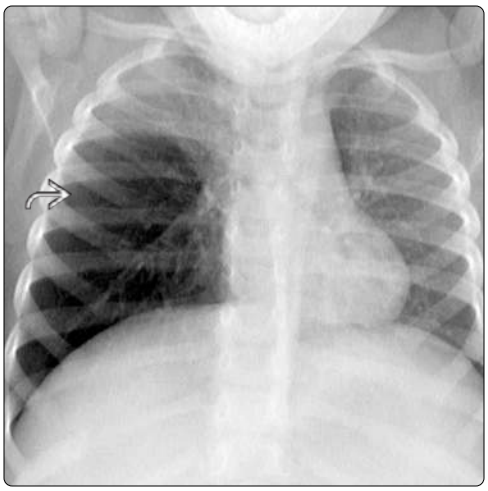
(Left) *Inspiratory PA chest radiograph in a toddler with acute respiratory distress demonstrates subtle relative lucency of the left lung compared to the right.* **(Right)** *Expiratory PA chest radiograph in the same patient demonstrates a static volume & persistent hyperlucency of the left lung compared to the right. In the correct clinical setting, static lung volumes on inspiratory & expiratory or bilateral decubitus chest radiographs are very suggestive of air-trapping due to foreign body aspiration.*





(Left) *PA chest radiograph shows asymmetric hyperexpansion & lucency in the right middle & lower lobes with relative increased opacity in the right upper lobe & left lung.* **(Right)** *Coronal oblique CECT reconstruction in the same patient shows that the right middle lobe (RML) & right lower lobe air-trapping was due to a foreign body occluding the bronchus intermedius.*





(Left) *Frontal chest radiograph of a coughing 2-year-old patient who aspirated a peanut (unrecognized at the time) shows opacity & volume loss in the RML presumed to be atelectasis in the setting of viral airway disease.* **(Right)** *AP radiograph in the same child 3 months later shows RML hyperlucency & oligemia due to air-trapping. During bronchoscopy, an old peanut was found in the RML bronchus. Depending on the degree of bronchial occlusion, a foreign body may cause atelectasis, hyperexpansion, or a changing picture over time.*

Cystic Fibrosis, Pulmonary

KEY FACTS

TERMINOLOGY

- Autosomal recessive multisystem disorder caused by dysfunctional chloride ion transport across epithelial surfaces → thickening of secretions (e.g., mucus, digestive fluids, sweat)
- In lungs, abnormal mucus & degraded WBCs → chronic airway impaction → recurrent inflammation & infections → chronic airway damage (in progressively worsening cycle)

IMAGING

- Most common in upper lobes, superior lower lobes
  - Peribronchial thickening (early finding)
  - Mosaic attenuation due to air-trapping, best seen on expiratory scan
  - Bronchiectasis with signet ring sign (bronchus larger than adjacent artery)
  - Mucus plugging within dilated bronchi (finger-in-glove appearance)
  - "Tree-in-bud" centrilobular nodular opacities

PATHOLOGY

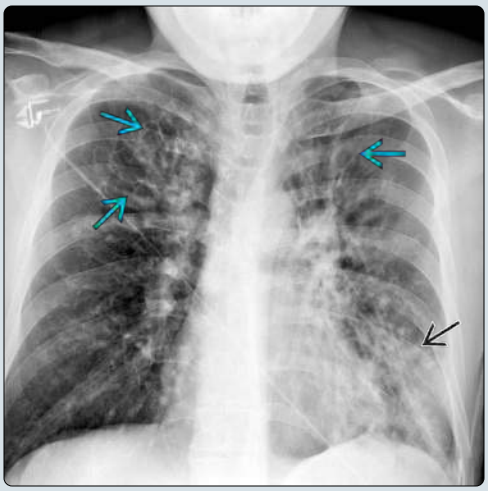
- Most common lethal genetic disorder in Caucasians
  - ~ 1 in 2,500 affected
- Mutation in both copies of CF transmembrane conductance regulator (*CFTR*) gene at chromosome 7q31.2 → defective chloride transport → abnormal water regulation
- > 1,000 genetic defects can result in CF; ΔF508 mutation of *CFTR* most common (~ 90%)

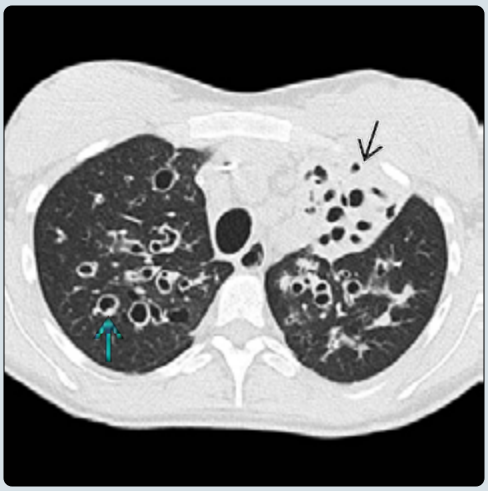
CLINICAL ISSUES

- 70% present < 1 year of age: GI symptoms more common
- 90% by 12 years of age: Respiratory more typical
  - Often have asthma type symptoms
- Median survival: 41.1 years

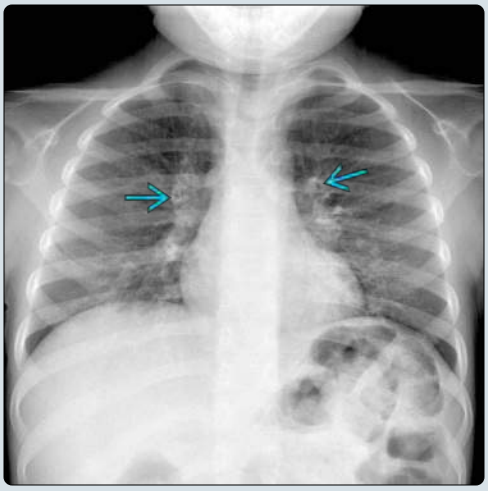
DIAGNOSTIC CHECKLIST

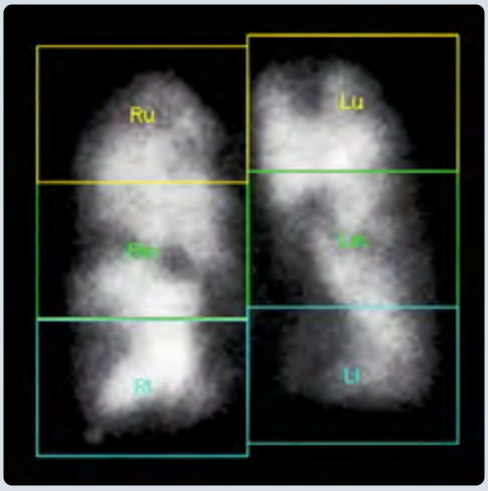
- Annual chest radiograph or low-dose surveillance CT
  - CT best assesses progressive disease & predicts future exacerbations vs. pulmonary function tests, radiographs





(Left) *Single frontal radiograph in a patient with cystic fibrosis (CF) shows prominent bronchiectasis in the upper lobes + left lower lobe consolidation with nodularity.* **(Right)** *Axial NECT in a CF patient shows bilateral upper lobe bronchiectasis with consolidation of the anterior segment of the left upper lobe. Note the signet ring sign (or pearl ring sign) in the right upper lobe with the dilated bronchus forming the ring & the adjacent artery forming the attached jewel.*





(Left) *Frontal radiograph of the chest in a 7 year old with CF shows mild peribronchial thickening in the perihilar regions, an early finding of CF that is indistinguishable from common viral bronchiolitis.* **(Right)** *Anterior image from a Tc-99m MAA arterial perfusion scan in a patient with CF shows multifocal decreased uptake in the lungs bilaterally. Perfusion abnormalities may be the earliest findings in patients with CF.*

TERMINOLOGY

Abbreviations

- Cystic fibrosis (CF)

Definitions

- Autosomal recessive multisystem disorder caused by dysfunctional chloride ion transport across epithelial surfaces → thickening of secretions (e.g., mucus, digestive fluids, sweat)

IMAGING

General Features

- Best diagnostic clue
  - Upper lobe predominant bronchiectasis & bronchial wall thickening in child with respiratory (± gastrointestinal) symptoms
- Location
  - In lungs, more common in upper lobes & superior segments of lower lobes
  - Also affects sinuses, pancreas, hepatobiliary system, GI tract, & sex organs

Radiographic Findings

- Radiographs insensitive to early changes of CF
- Late changes on radiographs include bronchiectasis, bronchial wall thickening, mucoid impaction, hyperinflation, lobar collapse, & pulmonary arterial enlargement due to pulmonary artery hypertension
- ± microcardia with chronic pulmonary hyperinflation
- ± abdominal manifestations visible on chest radiographs
  - Dilated bowel in newborn with meconium ileus
  - Ca^{2+} in newborn peritoneum if complicated meconium ileus → perforation → meconium peritonitis
  - Ca^{2+} over upper abdomen in older children & adolescents with chronic pancreatitis

CT Findings

- HRCT
  - Peribronchial thickening (early finding)
  - Bronchiectasis with signet ring sign
    - Ectatic bronchus > adjacent pulmonary artery
  - Mosaic attenuation due to air-trapping, best seen on expiratory scan
  - "Tree-in-bud" centrilobular nodular opacities
    - Bronchiolar impaction of mucus &/or infectious/inflammatory debris
  - Finger-in-glove appearance of mucus plugging within dilated bronchi
  - Other findings include: Emphysema, multifocal atelectasis, sacculations, & bullae
  - ± lymphadenopathy
- CTA
  - May show bronchial artery hypertrophy &/or aberrant/collateral arterial supply in setting of hemoptysis

MR Findings

- Pulmonary MR ± use of inhaled hyperpolarized gas
  - May yield imaging results comparable to CT
  - May be able to detect early mucus plug & differentiate mucoid impaction from atelectasis
  - Phase contrast imaging
    - Volume of flow to each lung may be calculated
    - ↑ aortopulmonary collateral flow may precede lung function decline
  - Functional cardiac information & ventricular volumes may be obtained

Ultrasonographic Findings

- Useful in evaluating pleural effusions
- Not helpful in lung evaluation in CF

Angiographic Findings

- Bronchial arteriography to detect (± embolize) source of pulmonary hemorrhage/hemoptysis

Nuclear Medicine Findings

- Normal chest radiograph with normal Tc-99m perfusion scan essentially excludes bronchiectasis
  - Nuclear medicine perfusion more sensitive for detection of early disease in patients with CF
    - Multifocal perfusion defects typically seen

Imaging Recommendations

- Best imaging tool
  - High-resolution CT with full inspiration & expiration

DIFFERENTIAL DIAGNOSIS

Asthma

- Peribronchial thickening & hyperinflation common but without bronchiectasis

Allergic Bronchopulmonary Aspergillosis

- Findings overlap CF; allergic bronchopulmonary aspergillosis (ABPA) often complicates CF
- Other causes of ABPA include refractory asthma, tuberculosis, sarcoidosis, infectious pneumonia, Churg-Strauss syndrome

Ciliary Dyskinesia

- Abnormal ciliary movement (not abnormally thick secretions) responsible for mucus accumulation
- Basilar predominant bronchial wall thickening, bronchiectasis
- Situs inversus & sinus disease in Kartagener syndrome

Tracheobronchomegaly (Mounier-Kuhn Syndrome)

- Rare congenital disorder with abnormal widening of upper airways

Williams-Campbell Syndrome

- Cystic bronchiectasis with bronchomalacia due to defective bronchial cartilage

PATHOLOGY

General Features

- Mutation in both copies of CF transmembrane conductance regulator (*CFTR*) gene at chromosome 7q31.2
  - > 1,000 genetic defects can result in CF; ΔF508 mutation most common (~ 90%)
- CFTR protein malfunction → lack of chloride ion secretion → ↑ sodium retention & fluid resorption → ↑ viscosity of luminal secretions → obstruction of ducts of solid organs & hollow viscera

- In lungs, abnormal mucus & WBC degradation products (including DNA) → stasis in airways → recurrent infection & inflammation → chronic airway damage with worsening susceptibility to infection & inflammation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 1st symptoms may be delayed passage of meconium with bowel obstruction ± perforation in newborn with meconium Ileus
  - In childhood, patients often look similar to asthma patients, presenting with chronic cough & wheezing
- Other signs/symptoms
  - Chronic constipation
  - Recurrent pancreatitis
  - Hepatobiliary disease
  - Clubbing of fingers & toes
- Clinical profile
  - Most now detected on neonatal screening
  - After symptom development, testing by sweat chloride (> 60 mEq/mL = + for CF)

Demographics

- Age
  - 70% present < 1 year: GI symptoms more common
  - 90% by 12 years: Respiratory more typical
- Epidemiology
  - Most common lethal gene defect in Caucasians (1 in 2,500 affected)
    - Much less common with African or Asian ancestry

Natural History & Prognosis

- Infancy: Often only mild or absent pulmonary symptoms
- Early childhood: Present similar to asthma patients with cough, wheezing, & bronchitis
- Late childhood & adolescence: Recurrent pneumonias, bronchitis, & bronchiectasis with mucus plugging
- End-stage lung disease occurs at variable ages but often in late adolescence to early adulthood
- Exact timing of progression in CF unpredictable
- Complications of CF lung disease
  - Recurrent infections most common complication: > 1/2 develop infection with *Pseudomonas aeruginosa*
  - Allergic bronchopulmonary aspergillosis
  - Pneumothorax & pneumomediastinum
  - Hemoptysis
  - Bullous emphysema
  - Cor pulmonale
- Median survival: 41.1 years

Treatment

- Goal: ↓ lung damage from mucus plugging & infection
- Various internal & external airway clearance techniques
- Prophylactic antibiotics to ↓ chance of infection
- ± long-term IV (Port-a-Cath) for recurrent lung infections
- New medicines targeted at specific *CFTR* mutations
- ± lobectomy for patients with single lobe complications
- End-stage lung disease may require lung transplantation
  - Both lungs typically transplanted to avoid infection of new lung from native lung
  - Clinical parameters typically guide transplantation

DIAGNOSTIC CHECKLIST

Consider

- Annual chest radiograph or low-dose CT surveillance of bronchiectasis complications
- Pulmonary MR showing promise for future utility

Image Interpretation Pearls

- CT more accurate in assessing progressive lung disease than pulmonary function tests or radiographs
- CT improves with treatment of acute exacerbations
- Brody CT score predicts future pulmonary exacerbations better than initial FEV_1; scoring based on
  - Extent of bronchiectasis
  - Size of abnormally dilated bronchi
  - Peribronchial thickening
  - Parenchymal involvement including consolidation, ground-glass opacity, cysts & bullae
  - Hyperinflation

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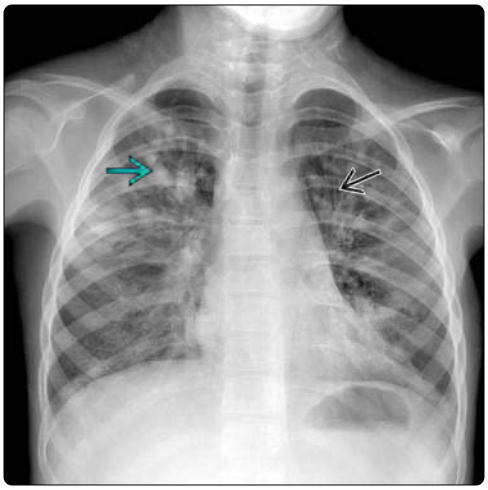
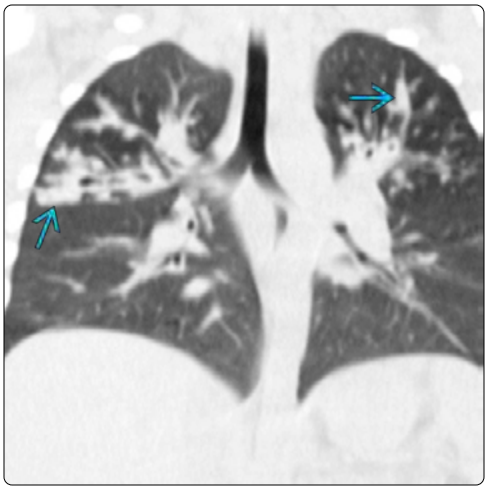
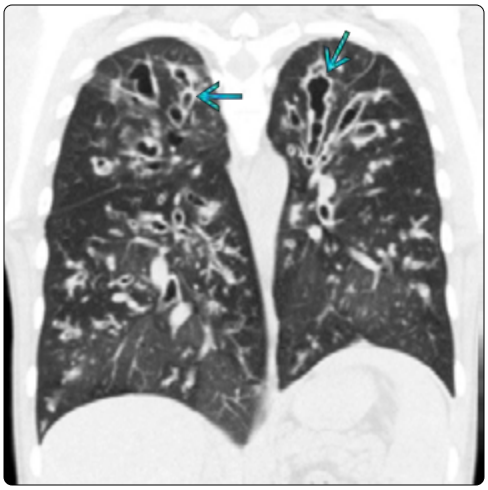
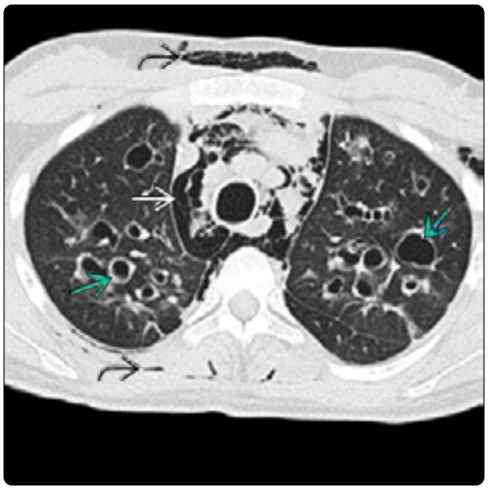


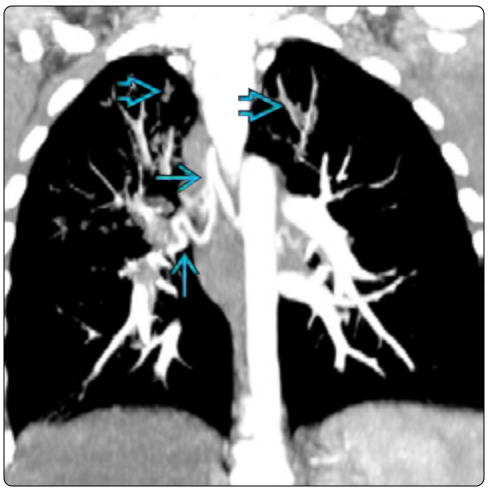


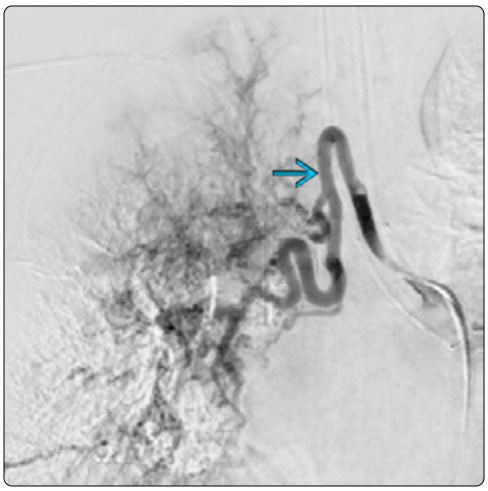
(Left) *Frontal view of the chest in a 7-year-old girl with CF shows thick tubular & nodular densities in the right upper lobe. Note the bronchiectasis in the left upper lobe. Mucus impacted in the dilated bronchi is referred to as a finger-in-glove appearance.* **(Right)** *Coronal NECT image in a 7-year-old CF patient shows increased nodular & tubular densities in the right > left upper lobes, consistent with mucus impaction within dilated bronchi.*





(Left) *Coronal HRCT in a patient with CF shows bronchiectasis & peribronchial thickening that are more prominent in the upper lobes.* **(Right)** *Axial HRCT from a CF patient shows pneumomediastinum & subcutaneous emphysema. Note the prominent bronchiectasis in the upper lobes bilaterally.*





(Left) *Coronal CTA in a 12-year-old CF patient with hemoptysis shows an enlarged right bronchial artery. Evaluation of the more distal branches & adjacent parenchyma (for alveolar hemorrhage & extravasation) may help localize the source of bleeding. There is also upper lobe bronchiectasis & mucus plugging bilaterally.* **(Right)** *Selective DSA of the enlarged right bronchial artery in the same patient shows no active extravasation, though there is heterogeneous lung perfusion & tortuosity of more distal arteries.*

KEY FACTS

TERMINOLOGY

- Foreign body in esophagus for prolonged period of time

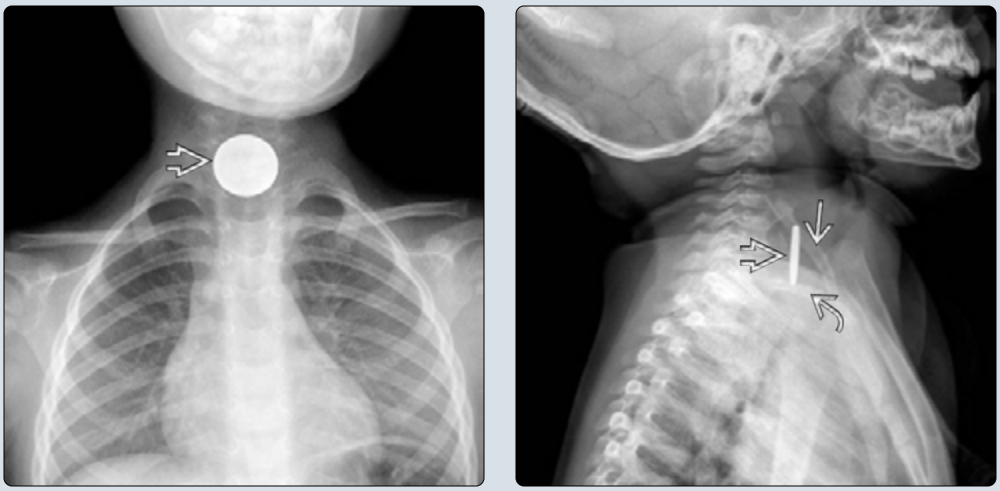
IMAGING

- Foreign body may or may not be radiopaque
- Airway narrowing suggests chronicity
  - Airway displaced anteriorly
  - Proximal esophagus may be dilated
- May present with complications: Abscess, pneumomediastinum, or pneumothorax
- Most common site: Upper esophagus at thoracic inlet
  - 2nd most common site: Level of carina & aortic arch
- Coins are most commonly swallowed foreign body
  - Button batteries show characteristic double-density (2-layer) periphery
- Imaging recommendations
  - AP & lateral chest radiograph best initial study; include nasal cavity on lateral airway view if respiratory symptoms present
  - Esophagram for nonradiopaque foreign bodies
  - CECT to diagnose complications, such as abscess, mediastinitis, aortic wall injury

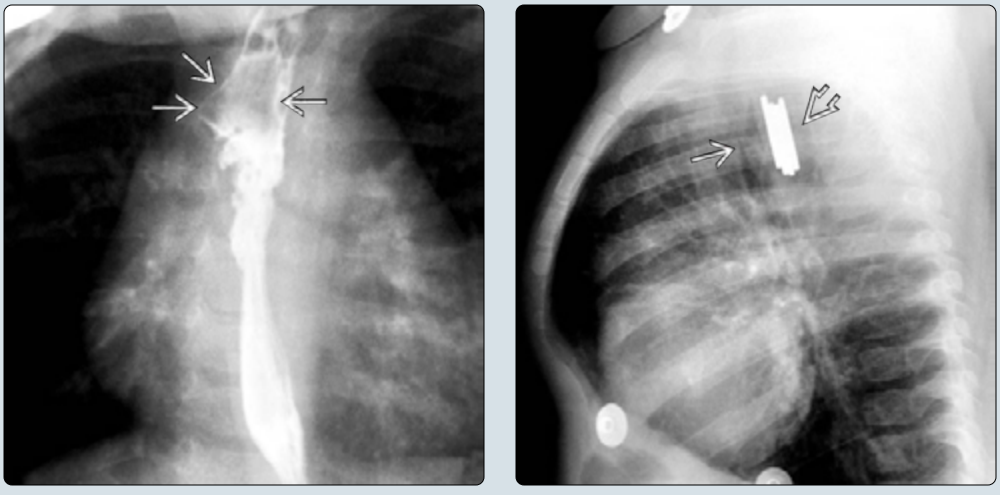
CLINICAL ISSUES

- Respiratory/airway or feeding symptoms most common
  - Cough, stridor, fever, wheezing
- Removal success rate 95-100% regardless of technique
- Use endoscopy immediately for batteries in esophagus
  - Batteries quickly cause damage by pressure against wall of esophagus, leakage of caustic alkali, & generation of electrical current
  - Aortic wall inflammation can lead to delayed rupture & exsanguination; NASPGHAN guidelines recommend serial cross-sectional imaging
- Foreign bodies present for > 24 hours have ↑ risk of esophageal perforation

(Left) *AP radiograph of the chest shows a round metallic foreign body at the level of the thoracic inlet ➡, consistent with the history of a swallowed coin. This is the most common location for an esophageal foreign body.* **(Right)** *Lateral radiograph of the upper chest & neck shows an esophageal foreign body ➡. Notice the narrowing of the trachea ➡ with soft tissue swelling around the metallic foreign body ➡.*



(Left) *Frontal esophagram shows a large, nonradiopaque foreign body ➡ proximal to the anastomotic stricture in a patient who is status post esophageal atresia-tracheoesophageal fistula repair.* **(Right)** *Lateral radiograph shows 4 coins in the esophagus ➡ with anterior displacement of the airway, which is significantly narrowed ➡, indicating inflammation.*



IMAGING

General Features

- Best diagnostic clue
  - Radiopaque object in region of esophagus
  - High suspicion for chronicity when airway narrowed &/or anteriorly displaced
  - May present with complications: Abscess, pneumomediastinum, pneumothorax
- Location
  - Most common site: Upper esophagus at thoracic inlet
  - 2nd most common site: Level of carina & aortic arch from physiologic narrowing
  - 3rd most common site: Distal esophagus slightly above gastroesophageal junction
- Morphology
  - Coins are most commonly swallowed foreign body
    - Coins in esophagus will usually appear in coronal plane (en face) on frontal view
    - Coins in trachea typically appear in sagittal plane (on end) on frontal view
  - Button batteries show characteristic double-density (2-layer) shadow at periphery
    - Laterally, edges are rounded with step-off at junction of positive & negative terminals
    - Important to identify since batteries can cause caustic burn injury to esophagus in little time
  - Nonradiopaque foreign body
    - Hot dog & plastic toys common

Radiographic Findings

- Radiography
  - Radiopaque foreign body in esophagus
    - Entirety or portion of foreign body may be radiolucent
  - Lateral view shows soft tissue thickening anterior to esophagus with displacement, bowing, & narrowing of trachea
    - Indicative of chronic inflammation & predictive of more difficult removal of foreign body

Fluoroscopic Findings

- Esophagram
  - Useful for evaluating nonradiopaque foreign bodies
  - May diagnose stricture, fistula, or perforation

CT Findings

- CECT
  - Postremoval CT: Esophageal leak, diverticulum, mediastinitis/abscess; inflammation or injury of aorta if battery or sharp object removed

Imaging Recommendations

- Best imaging tool
  - AP & lateral chest radiograph best initial study
    - Include nasal cavity with respiratory symptoms
  - Esophagram for nonradiopaque foreign bodies
  - CECT to diagnose complications

DIFFERENTIAL DIAGNOSIS

Airway Obstruction/Inflammation

- May simulate foreign bodies in esophagus

Achalasia

- Failure of normal relaxation of lower esophageal sphincter

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Respiratory or airway problems most common
    - Cough, stridor, fever, wheezing
    - Chronic upper respiratory infection or pneumonia
    - Hemoptysis, choking, cyanosis
  - Gastrointestinal symptoms
    - Dysphagia, drooling, vomiting, gagging
  - Chest pain when swallowing
  - Fever of unknown origin

Demographics

- Age
  - Most < 5 years; typically, 8 months to 2 years

Natural History & Prognosis

- Foreign bodies present for > 24 hours have ↑ risk of esophageal perforation
- Vast majority of ingested objects pass through gastrointestinal tract without problems

Treatment

- Removal success rate 95-100% regardless of technique
- Strategy depends on type, location, & duration
- General complications prior to or without treatment
  - Most common complication: Perforation & subsequent mediastinitis
  - Rare complications include tracheoesophageal fistula & aortoesophageal fistula
- Endoscopy or surgery
  - Use immediately for sharp or irregular objects & unknown foreign bodies
  - Use immediately for batteries in esophagus
    - Cause damage by pressure against wall of esophagus, leakage of caustic alkali, & electrical current
    - Reports of exsanguination (even delayed) due to inflammation of aortic wall by battery have led to NASPGHAN guidelines for serial cross-sectional imaging after battery removal

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Sickle Cell Disease, Acute Chest Syndrome

KEY FACTS

TERMINOLOGY

- New pulmonary opacity on chest radiograph + ≥ 1 additional symptom (such as fever, cough, sputum production, tachypnea, dyspnea, or hypoxia) in setting of sickle cell disease (SCD)

IMAGING

- Upper & middle lobe opacities more common in children
- Lower lobe disease more common in adults
- Initial chest radiograph may be normal (46%)
  - Opacity may not appear until 2-3 days after symptoms develop
- Opacities on CT may be more extensive than on radiograph

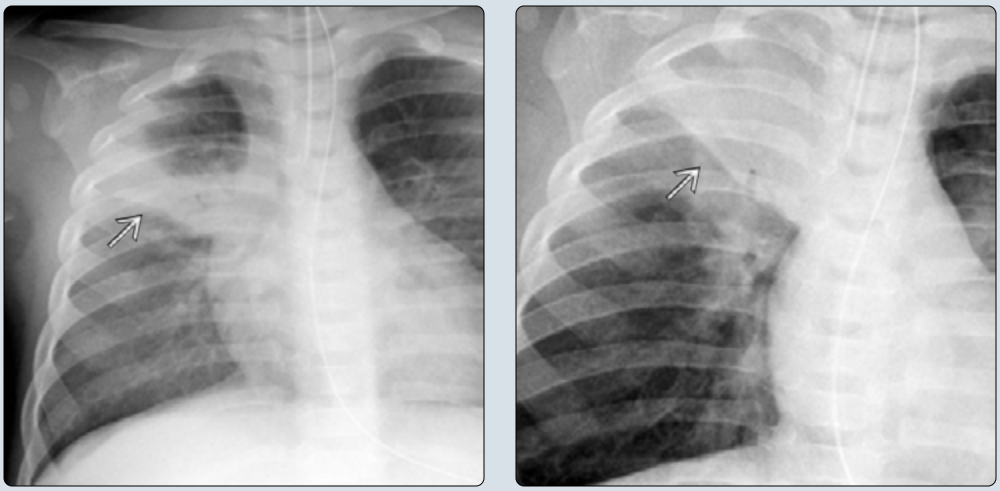
PATHOLOGY

- Potential causes: Infection (30%), pulmonary fat embolism (9%), pulmonary infarction (18%), & rib infarction

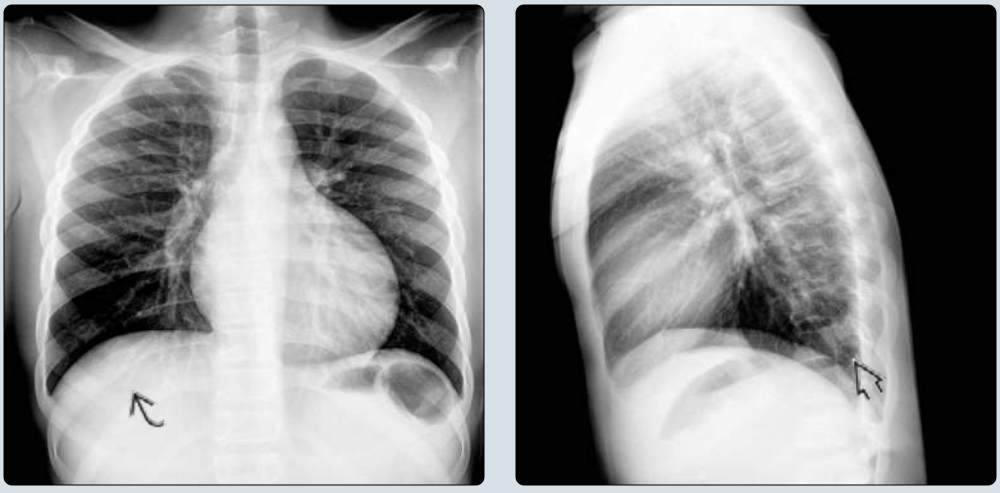
CLINICAL ISSUES

- Acute chest syndrome (ACS) most common in patients aged 2-4 years; incidence ↓ with age
  - Fever, cough, & tachypnea most common symptoms in patients < 10 years of age
  - Pain (chest, extremity, abdominal) more common in adolescents & adults
- ACS 2nd most common cause of hospitalization in patients with SCD
- ACS most common cause of premature death in patients with SCD
  - Mortality 4-9x higher in adults than in children
- Treatment
  - Supportive: Oxygen, antibiotics, pain control, IV fluids, incentive spirometry, & blood transfusions
  - Prevention: Pneumococcal vaccine, *Haemophilus influenzae* vaccine, & hydroxyurea

(Left) *AP radiograph of the chest shows an opacity in the right upper lobe ➡ of a 1 year old with sickle cell disease presenting with fever & dyspnea.* **(Right)** *AP radiograph of the chest in the same patient 12 hours later shows worsening opacification & volume loss of the right upper lobe ➡. Acute chest syndrome is defined as a new pulmonary opacity in a symptomatic patient with sickle cell disease. In children, it occurs more commonly in the upper & middle lobes.*



(Left) *Frontal radiograph of the chest in a patient with sickle cell disease shows a subtle opacity ⇨ in the right lower lobe. Lower lobe opacities are more common in adults.* **(Right)** *Lateral radiograph in the same patient shows the subtle retrocardiac opacity ⇨ overlying the spine. Acute chest syndrome can be caused by infection, infarction, & fat embolism.*



TERMINOLOGY

Abbreviations

- Acute chest syndrome (ACS)
- Sickle cell disease (SCD)

Definitions

- New pulmonary opacity on chest radiograph + ≥ 1 additional symptom (such as fever, cough, sputum production, tachypnea, dyspnea, or hypoxia) in setting of SCD

IMAGING

General Features

- Best diagnostic clue
  - Pulmonary opacity in patient with SCD who has fever & respiratory symptoms
- Location
  - Upper & middle lobe involvement more common in children
  - Lower lobe disease more common in adults
- Size
  - Variable: May be segmental, lobar, or multilobar consolidation

Radiographic Findings

- Radiography
  - ACS
    - Initial chest radiograph may be normal (46%)
      - Radiograph lags behind physiological changes
      - Opacity may not appear until 2-3 days after symptoms develop
    - Opacity mimics pneumonia ± volume loss
    - Pleural effusions present in > 50% of patients
  - Other findings of SCD
    - Cardiomegaly due to chronic anemia
    - Avascular necrosis/sclerosis of humeral heads
    - H-shaped or biconcave vertebrae
    - Enlarged ribs due to marrow expansion
    - Small spleen (autosplenectomy) identified by lateralization of stomach bubble
    - Cholecystectomy clips in right upper quadrant

CT Findings

- NECT
  - Limited clinical use for ACS
  - Can have consolidation or ground-glass opacity
    - Opacities may be more extensive than on radiograph
  - Simplification of lung with ↓ vascularity
  - Pleural effusion common
  - May see healed bone infarcts
- HRCT
  - Mosaic perfusion due to microvascular occlusion
  - Sequelae of ACS
    - Parenchymal bands & interlobular septal thickening
    - Peripheral wedge-shaped opacities
    - Architectural distortion
    - Traction bronchiectasis
- CTA
  - Pulmonary embolism 3rd most common cause of ACS
    - Accounts for 16-17% of ACS episodes
    - Occurs in segmental or subsegmental pulmonary arteries
    - No associated lower extremity deep vein thrombosis

Nuclear Medicine Findings

- Bone scan
  - Foci of abnormal radiotracer uptake in ribs
    - ↓ or ↑ uptake: Acute or subacute bone infarcts
  - May have other bone infarcts vs. osteomyelitis
- V/Q scan
  - Limited clinical use in ACS
  - Can see perfusion defect
    - May mimic pulmonary embolism
  - Defects often resolve quickly with supportive therapy

Imaging Recommendations

- Best imaging tool
  - Chest radiograph for evaluation of pulmonary opacity
- Protocol advice
  - Routine frontal & lateral chest radiographs
  - Repeat chest radiograph in 48-72 hours if clinical concern high as initial chest radiograph often normal

DIFFERENTIAL DIAGNOSIS

Bacterial Pneumonia

- Clinically similar to ACS with fever, leukocytosis, pleuritic chest pain, pleural effusion, & productive cough
- Multiple lobe involvement & recurrent opacities more common in SCD
- Clinical symptoms & radiographic abnormalities prolonged, lasting 10-12 days

Viral Chest Infection

- Viral infection causes 8% of ACS
  - Respiratory syncytial virus (RSV) common

Pulmonary Infarction

- ~ 50% of cases in patients with SCD caused by fat embolism
- Vascular occlusion by sickle cells also important cause of ACS
- Usually diagnosis of exclusion

Asthma

- Significant comorbidity in patients with SCD; may be underdiagnosed in SCD
  - 40% of patients with SCD have positive response to cold air or exercise challenge
  - Patients diagnosed with asthma have 4-6x greater risk of developing ACS

PATHOLOGY

General Features

- Etiology
  - ACS may be multifactorial
    - Infection most common cause of ACS in children
    - 50% of adult patients initially admitted with vasoocclusive crisis causing pain
  - Infection
    - Documented in 38-54% of cases

  - Most common pathogens: *Chlamydia pneumoniae*, *Mycoplasma pneumoniae*, RSV
  - Pulmonary opacity persists longer than cases where infection not documented
- Pulmonary fat embolism
  - Cause of ACS in 9-16%
  - Etiology: Vasoocclusive crisis → edema & infarction of marrow compartment → marrow necrosis → fat cells enter venous bloodstream → embolize in branches of pulmonary artery
  - Frequently have bone pain
  - Lab findings: Thrombocytopenia, anemia; ↑ LDH, lipase, phospholipase A2, & uric acid; ↓ serum calcium
  - Diagnosis supported by lipid-laden macrophages in bronchoalveolar lavage fluid
- Pulmonary infarction
  - Cause of ACS in 16-17%
  - Occurs in segmental & subsegmental pulmonary arteries
  - No associated lower extremity deep vein thrombosis suggests in situ thrombosis
- Rib infarction with hypoventilation from pain &/or analgesics
  - High correlation between rib infarction & pulmonary opacity
  - Pain may lead to splinting & atelectasis
  - Incentive spirometry helps prevent pulmonary complications of ACS
  - Analgesics may ↓ splinting but may cause hypoventilation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Fever, cough, & tachypnea most common in patients < 10 years of age
  - Pain (chest, extremity, abdominal) more common in adolescents & adults

Demographics

- Age
  - Most common in patients aged 2-4 years
  - Lower incidence in patients < 2 years due to higher fetal hemoglobin concentrations
  - Incidence gradually declines with age
    - Excess mortality in group with ACS
    - Fewer viral illnesses due to acquired immunity
- Gender
  - Slightly more common in males
- Ethnicity
  - In USA, almost exclusively seen in African Americans
- Epidemiology
  - Overall incidence of 12.8 episodes per 100 patient-years
  - ACS most common cause of premature death in patients with SCD
  - ACS 2nd most common cause of hospitalization in patients with SCD after pain crisis
    - ~ 50% of patients admitted with diagnosis other than ACS
    - Diagnosed with ACS 2-3 days after admission

Natural History & Prognosis

- More severe in patients > 20 years
  - Mortality 4-9x higher in adults than in children
- Features associated with poor prognosis
  - Physical exam: Altered mental status, tachycardia > 125 beats/minute, tachypnea > 30 breaths/minute, temperature > 40°C, hypotension
  - Lab findings: Arterial pH < 7.35, O_2 saturation < 88%, hemoglobin concentration ↓ by ≥ 2 g/dL, platelet count < 200,000, multiorgan failure
- Risk factors: Asthma, smoking, abdominal surgery, trauma

Treatment

- Supportive
  - Oxygen, antibiotics, pain control, IV fluids, incentive spirometry, & blood transfusions
  - Corticosteroids controversial; may be associated with ↑ readmission rate
- Prevention
  - At higher risk for pneumonia from encapsulated organisms due to autosplenectomy
    - Pneumococcal vaccination
    - *Haemophilus influenzae* vaccination
  - Hydroxyurea
    - Reduces sickling by ↑ fetal hemoglobin level
    - Reduces incidence in patients with recurrent ACS
  - Transfusion therapy

DIAGNOSTIC CHECKLIST

Consider

- ACS in patient with SCD, chest syptoms, & pulmonary opacity

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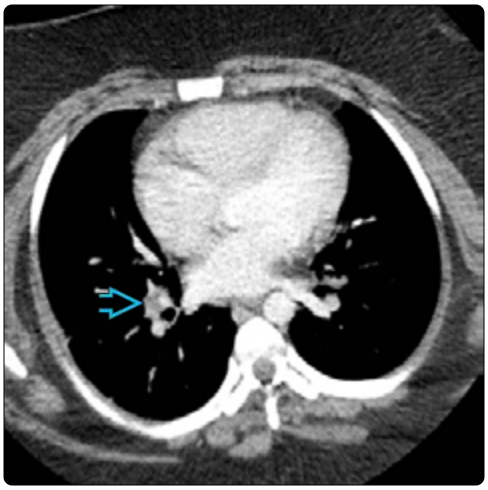
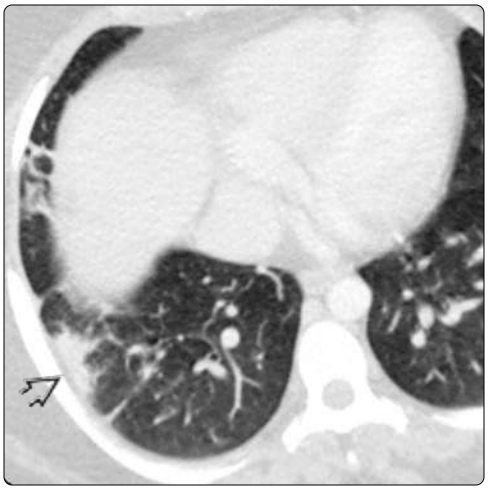
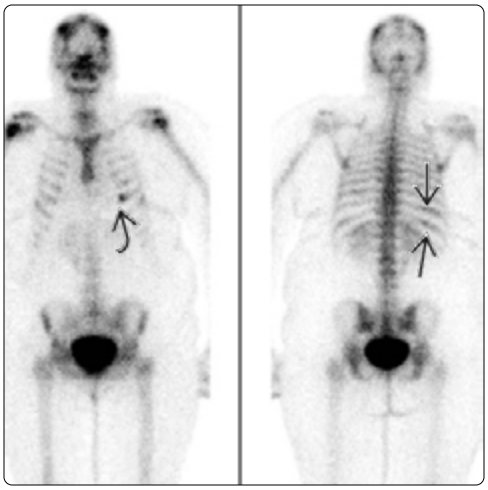
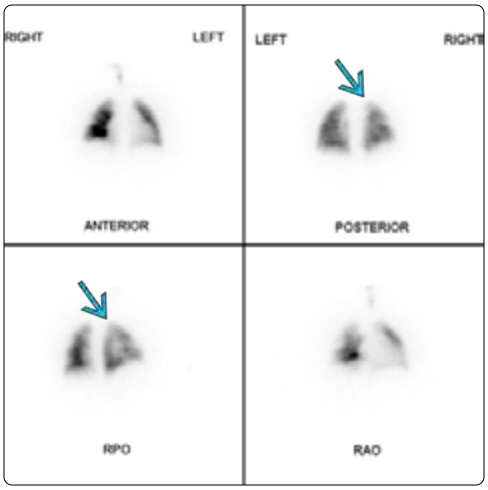


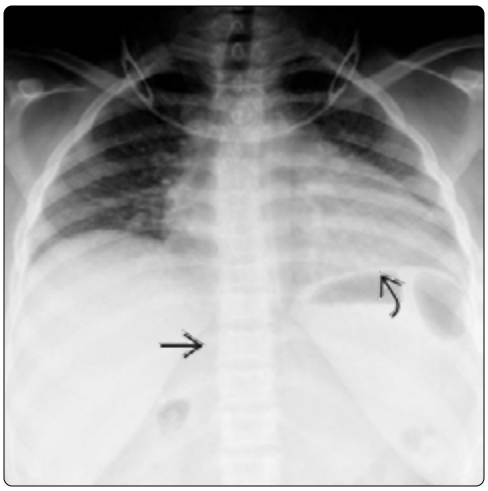


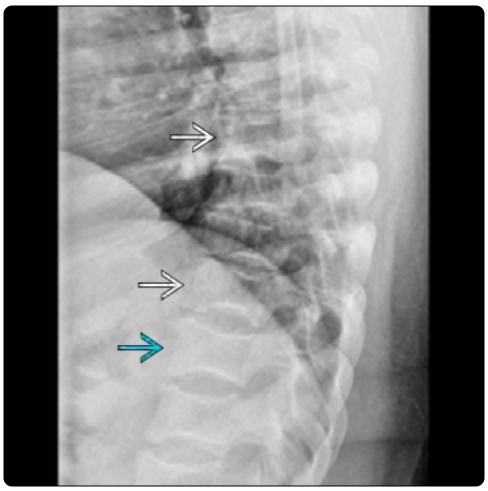
(Left) *Axial CECT in an adolescent with sickle cell disease shows a filling defect in the right lower lobe pulmonary artery. Pulmonary embolism is a common cause of acute chest syndrome.* **(Right)** *Axial CECT in lung windows in the same patient shows a wedge-shaped area of opacity in the right lower lobe. Pulmonary embolism can cause pulmonary infarction with hemorrhage.*





(Left) *Anterior & posterior projections from a bone scan in a patient with sickle cell disease show focally increased uptake in the left anterior 7th rib & right posterior 10th & 11th ribs, concerning for bone infarct.* **(Right)** *Perfusion study with Tc-99m MAA in a patient with sickle cell disease shows a moderate-sized defect in the right upper lobe. Perfusion defects in sickle cell disease may represent acute or chronic infarction.*





(Left) *AP radiograph of the chest in an adolescent with sickle cell disease shows a new streaky opacity in the left lower lung. The vertebral bodies have an abnormal H-shaped configuration typical of sickle cell disease.* **(Right)** *Lateral radiograph in the same patient shows abnormal H-shaped (due to central infarctions) or biconcave (due to bone softening) vertebral bodies at every imaged level. Other bone findings in sickle cell disease include sclerotic bone infarcts & enlargement of the ribs due to marrow expansion.*

Pectus Excavatum

KEY FACTS

TERMINOLOGY

- Depression of sternum posteriorly → sunken appearance of midline anterior inferior chest wall

IMAGING

- Radiographs: Right heart border blurring with vertical anterior ribs; degree of depression best seen on lateral view
- Pre-Nuss procedure evaluation with CT/MR
  - Haller index
    - Ratio of transverse (left-right) diameter divided by sagittal (anterior-posterior) diameter of chest
    - Haller index > 3.25 considered enough deformity for surgical candidacy (by most insurance agencies)
    - Consider reporting correction & depression indices to capture patients with clinically significant deformities but possibly "normal" Haller indices
  - CT: Noncontrast, low mA images
  - MR: Single cardiac MR can replace echocardiogram & CT
    - ↓ right ventricular ejection fraction & hemodynamically insignificant pericardial effusion common
- Post-Nuss procedure
  - Assess for complications → bar displacement/rotation, pneumothorax, pleural effusion, sternal infection

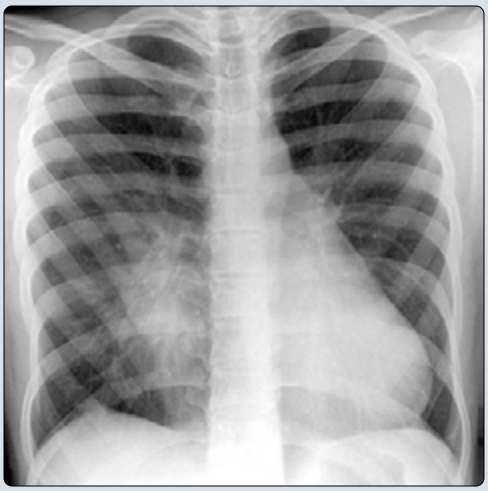
TOP DIFFERENTIAL DIAGNOSES

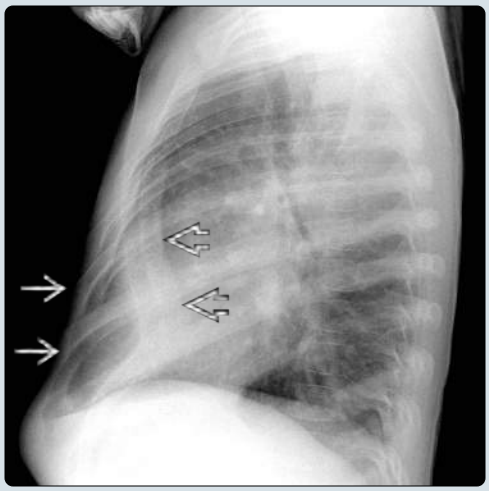
- Pectus carinatum
- Chest wall aggressive lesions
- Chest wall vascular malformations
- Palpable normal variants of chest wall

CLINICAL ISSUES

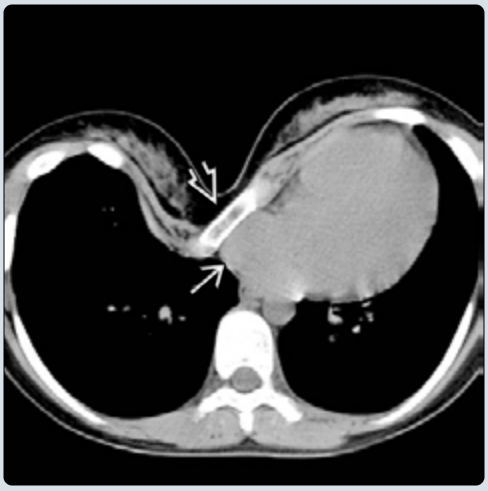
- Minimally invasive pectus repair (Nuss procedure)
  - Transverse curved metal bar surgically inserted internal to sternum & ribs
  - Excellent results in > 85% of patients

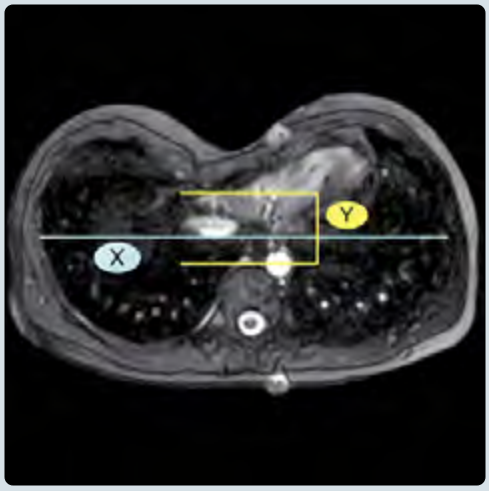
(Left) *AP radiograph shows a silhouette sign with an apparent opacity obscuring the right heart border (mimicking middle lobe disease). Note the vertically oriented anterior ribs.* **(Right)** *Lateral radiograph in the same patient shows posterior positioning of the sternum ⇨ as compared to the anterior ribs ⇨, consistent with pectus excavatum. There is no middle lobe pathology.*





(Left) *Axial NECT of the chest shows marked pectus excavatum with posterior displacement & rotation of the sternum ⇨ as compared with the anterior chest wall. Note the position of the right atrium ⇨ immediately behind the sternum.* **(Right)** *Axial SSFP MR shows the landmarks used to calculate the Haller index. The left-to-right (blue line) & anterior-to-posterior (sagittal yellow line) diameters are measured, & the ratio is calculated as X/Y.*





TERMINOLOGY

Definitions

- Deformity of chest wall characterized by sternal depression

IMAGING

General Features

- Imaging metrics
  - Haller index (HI)
    - Most frequently used metric to determine severity of pectus deformity; serves as indicator for Nuss procedure candidacy
    - Ratio of transverse (left-right) diameter divided by sagittal (anterior-posterior) diameter of inner aspects of osseous chest wall
    - HI > 3.25 considered requirement of many insurance companies to qualify for payment of surgical correction
  - Correction index (CI)
    - % chest depth missing from patient (i.e., % of chest depth to be corrected by bar placement)
  - Depression index (DI)
    - Measurement of severity independent of thoracic diameters

Radiographic Findings

- Right heart border blurring with vertical anterior ribs ± obliteration of descending thoracic aortic interface
- Degree of depression best seen on lateral radiograph
- Post-Nuss procedure
  - Nuss bar: Transverse metal bar with T-shaped stabilizer at 1 end
  - Potential complications: Bar displacement/rotation, pneumothorax, pleural effusion, sternal infection, cardiac injury
  - Lateral radiograph: Best view for migration of bar

CT Findings

- Pre-Nuss procedure evaluation with CT
  - Imaging metrics: HI, CI, DI, sternal tilt
  - Heart position (for surgical planning)
  - Central airway compression

MR Findings

- Expiratory axial slices with cardiac MR: Chest wall metrics + functional data (e.g., right ventricular ejection fraction, valve function)
- Right ventricle dysfunction due to extrinsic compression
- Hemodynamically insignificant pericardial effusion common

DIFFERENTIAL DIAGNOSIS

Pectus Carinatum

- Anterior chest convex outward: "Pigeon chest"

Chest Wall Aggressive Lesions

- Firm, painful mass ± rib destruction, pleural effusion

Chest Wall Vascular Malformations

- Soft, compressible mass; may fluctuate in size

Palpable Normal Variants of Chest Wall

- Rib anomalies may mimic sternal tilting/depression

PATHOLOGY

General Features

- Associated abnormalities
  - Connective tissue abnormalities
  - Neuromuscular disease
  - Genetic conditions
  - Pulmonary conditions

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Concerns about physical appearance common
  - Exercise intolerance (82%), chest pain (68%), poor endurance (67%), shortness of breath (42%)
- Other signs/symptoms
  - Cardiac (pulmonic murmur, mitral valve prolapse, syncope, Wolff-Parkinson-White syndrome), restrictive lung disease, central airway compression
  - Palpable bony asymmetry associated with mild pectus deformity may be mistaken for soft tissue mass

Demographics

- Epidemiology
  - Accounts for up to 90% of anterior chest wall disorders

Treatment

- Conservative management for mild cases
- Minimally invasive repair (Nuss procedure): Transverse curved metal bar inserted deep to sternum & rib cage
  - Excellent results in > 85% of patients

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Minimally Invasive Pectus Repair Appearance

KEY FACTS

TERMINOLOGY

- Minimally invasive Nuss procedure has replaced open surgeries to repair pectus excavatum
  - Introducer tunneled posterior to sternum
  - Convex stainless steel bar passed (with convexity down) through 2 incisions, then flipped & fixed in place
    - Immediate cosmetic improvement
  - Bar remains in place 2-4 years

IMAGING

- Postoperative radiographs
  - AP & lateral views to evaluate Nuss bar position & improvement in pectus deformity
- Early postoperative complications
  - Pneumothorax, pneumomediastinum
  - Atelectasis, pneumonia
  - Hemothorax, pleural effusion
    - If lasts > 4 days, should be cultured
    - May also be due to nickel allergy
- Late postoperative complications
  - Bar displacement
    - May rotate, requiring surgical revision
    - Apex of bar may slip from deepest portion of sternal depression, causing unsatisfactory contour change
      - 2 bars may be placed in these patients
      - Typically no intervention if slippage < 20%
    - Displacement more likely in postpubertal patients

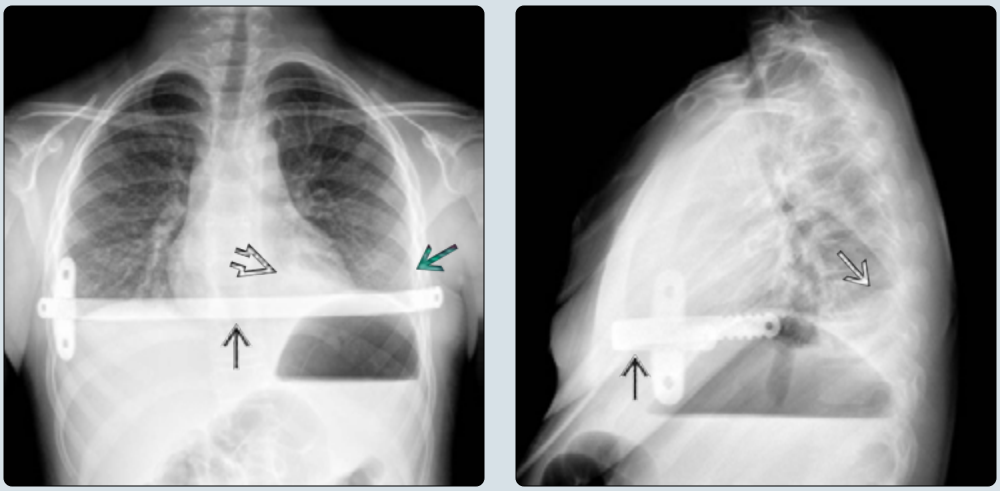
CLINICAL ISSUES

- Ideal age for repair: Just before puberty
  - May be performed earlier in cases of cardiac/pulmonary compression
  - Can also be performed after puberty with good outcomes

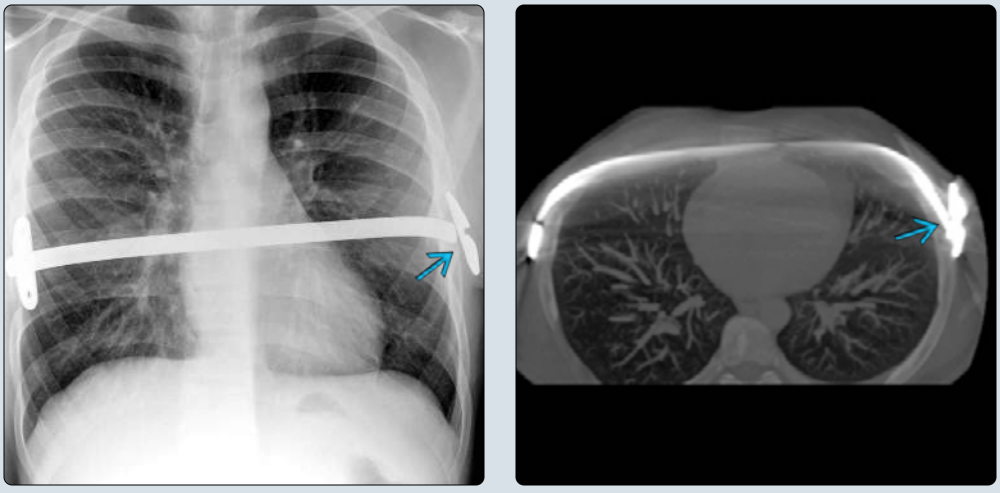
DIAGNOSTIC CHECKLIST

- Small postoperative pneumothoraces common; may be managed conservatively

(Left) *PA chest radiograph in a 10-year-old girl following a minimally invasive pectus excavatum repair shows the stainless steel bar ⇨ is in satisfactory position. Note the small left postoperative pleural effusion ⇨ & passive left lower lobe atelectasis ⇨. No pneumothorax is seen.* **(Right)** *Lateral chest radiograph of the same patient shows the stainless steel bar ⇨ passing posterior to the sternum at the point of maximum sternal concavity. Note the posterior pleural fluid & atelectasis ⇨.*



(Left) *PA chest radiograph after the Nuss procedure shows disruption at the junction of the pectus bar & left stabilizer ⇨.* **(Right)** *Volume-rendered image from a CT in the same patient shows disruption at the junction of the pectus bar & left stabilizer ⇨.*



KEY FACTS

TERMINOLOGY

- Askin tumor: Extraskeletal Ewing sarcoma of chest
  - Ewing sarcoma & Askin tumor closely related: Ewing sarcoma family of "tumors" (ESFT)

IMAGING

- Unilateral thoracic opacification
  - Large, lobular mass may occupy most of or entire hemithorax, especially with pleural effusion
  - Rib destruction common (> 50%)
  - Ca^{2+} uncommon
- Mildly heterogeneous enhancement on CECT, MR
- Mass effect on mediastinal structures
  - Vessels & airway typically compressed & shifted rather than encased or invaded
- Mediastinal lymphadenopathy: 25% at presentation
- Pulmonary metastasis: 38% at presentation
- CT & MR can be complimentary
  - CT better detects small pulmonary metastases
  - MR better evaluates involvement of chest wall

PATHOLOGY

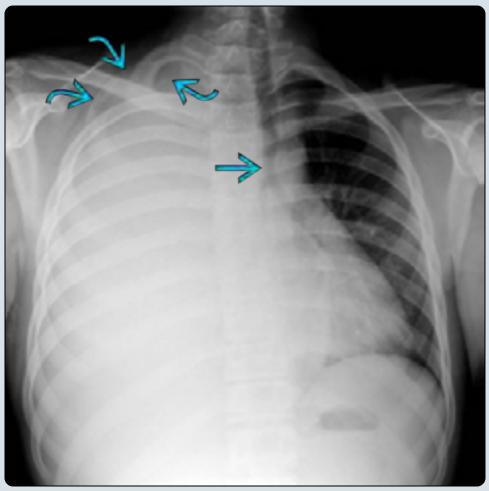
- Identical to other Ewing sarcomas
  - Small round blue cells
  - Positive immunohistochemical stain for *MIC2* gene product (CD99): 90%
  - Classic chromosomal translocation [t(11;22)(q24;q12)]: 85-95%

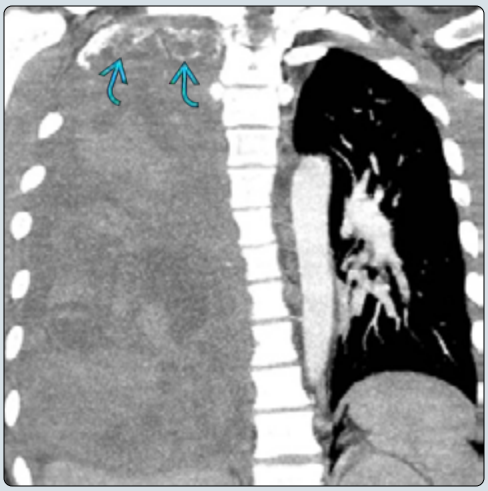
CLINICAL ISSUES

- Median age: 14.5 years
- Poor prognosis: Recurrence > 50%; 6-year survival of 14%

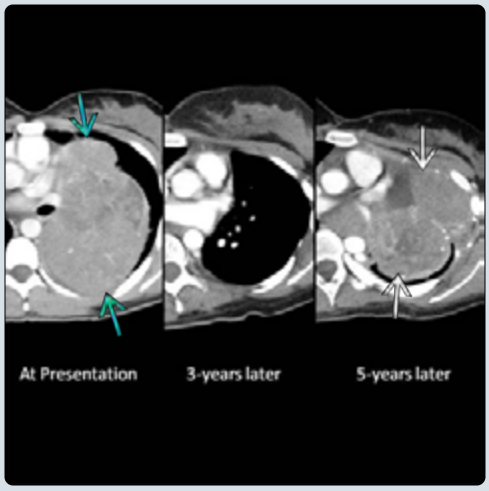
DIAGNOSTIC CHECKLIST

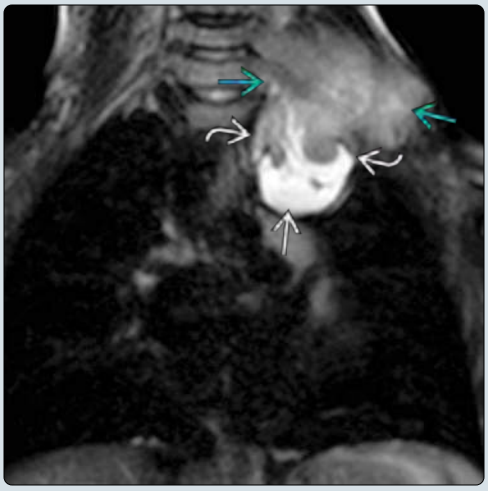
- On posttherapy surveillance CT, look closely for
  - Local recurrence in chest wall
  - Mediastinal lymphadenopathy
  - Pulmonary metastases





(Left) *PA radiograph shows a 15-year-old girl with a chest wall Ewing sarcoma. There is complete opacification of the right hemithorax with mass effect on the mediastinum. Note the lucency, expansion, permeation, & erosion of the right 2nd rib.* **(Right)** *Coronal CECT in the same patient shows a heterogeneous mass occupying the right hemithorax. There is expansion & destruction of the right 2nd rib.*





(Left) *Initial axial CECT of a girl with an Askin tumor at presentation (L) shows a large heterogeneous mass in the left upper hemithorax. After chemotherapy, resection, & radiation, no tumor was detectable on the 3-year follow-up CT (M). Tumor recurrence is present at 5 years (R).* **(Right)** *Coronal T2 FS MR shows a 3-year-old patient with an Askin tumor. A heterogeneous mass in the left upper hemithorax is partly cystic & extends into the soft tissues of the left lower neck.*

Chest Wall Mesenchymal Hamartoma

KEY FACTS

TERMINOLOGY

- Mesenchymal chest wall hamartoma = benign infantile lesion of proliferating skeletal elements arising from rib(s)

IMAGING

- Partially calcified soft tissue mass of chest wall
  - Chondroid > > osseous mineralization (100% by CT)
- Multiple internal compartments containing fluid-fluid levels
  - Secondary aneurysmal bone cysts (60-80%, CT vs. MR)
- Bizarre remodeling/distortion & erosion of multiple adjacent ribs
- Prenatal US may show chest wall mass with echogenic rim ("capsule")
  - ± polyhydramnios, pleural effusion
- Typical locations
  - Extrapleural posterior > anterior chest wall
  - Multifocal (unilateral or bilateral) in < 20%

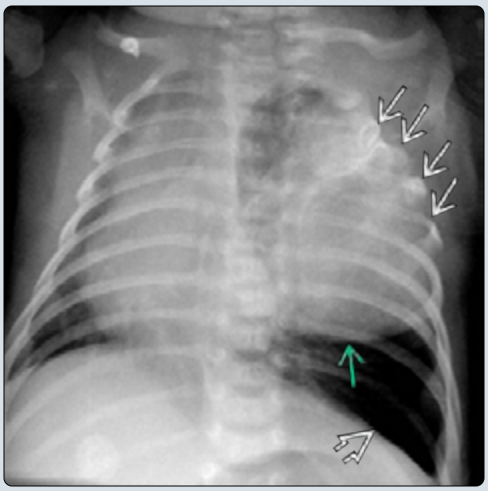
PATHOLOGY

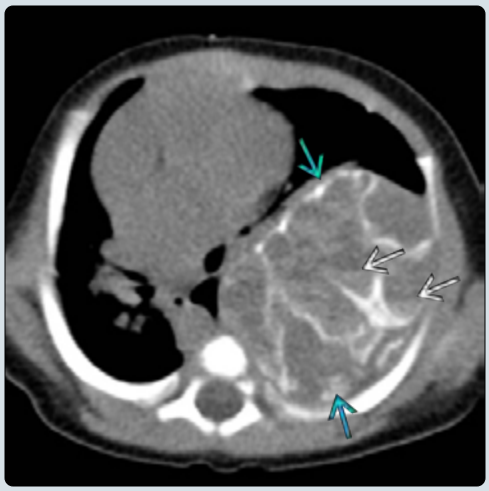
- Hemorrhagic cavities intermixed with cartilage & bone

CLINICAL ISSUES

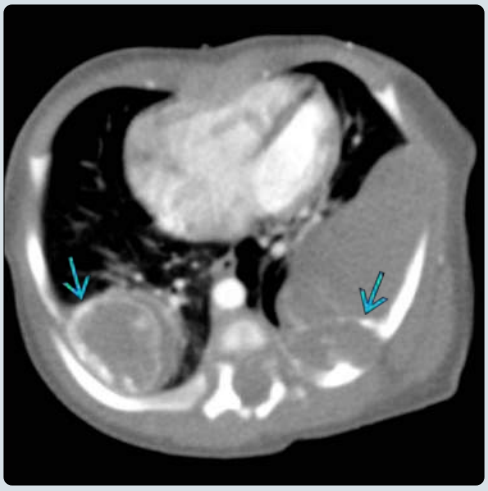
- 1 in 3,000 primary bone tumors
- Typically detected prenatally or within first 6 months of life
- Classic presentations: Congenital chest wall mass/deformity, respiratory distress
- No known risk of malignant degeneration, local invasion, or metastasis
- Many reports of spontaneous regression ≥ 2 years of age
- Observation if asymptomatic with classic age, presentation, & imaging findings
  - Potential for significant hemorrhage with disruption of blood-filled cavities during biopsy or excision
- Resection in setting of respiratory, cardiovascular, or neurologic compromise
  - Scoliosis occurs with resection of multiple posterior ribs

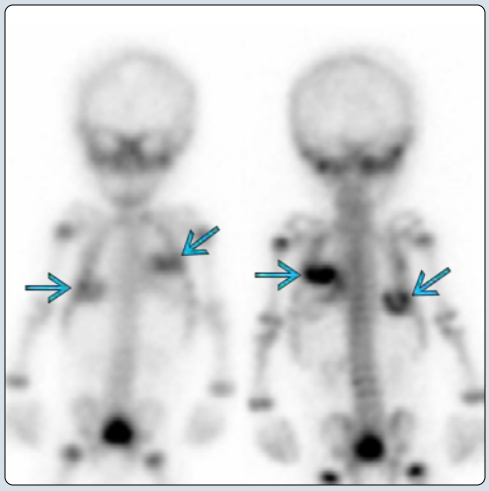
(Left) *AP radiograph in a 1 month old with respiratory distress shows a large soft tissue mass in the left hemithorax with bizarre remodeling & erosion of multiple adjacent ribs. There is mediastinal shift & lower lobe air-trapping due to mass effect.* **(Right)** *Axial NECT in the same patient shows rim & septal Ca^{2+} throughout the mass with numerous fluid-fluid levels due to layering blood products. Biopsy confirmed a chest wall mesenchymal hamartoma.*





(Left) *Axial CECT in a newborn with respiratory distress shows bilateral posterior chest wall masses with rim & septal Ca^{2+}. Additional CT images demonstrated distortion of multiple adjacent ribs bilaterally.* **(Right)** *Anterior (L) & posterior (R) Tc-99m bone scan images in the same patient show increased uptake within the bilateral chest wall mesenchymal hamartomas secondary to Ca^{2+}/ossification.*





KEY FACTS

TERMINOLOGY

- Kaposiform lymphangiomatosis (KLA): Uncommon but agressive lymphatic disease with poor prognosis
  - Features of both neoplasia & malformation
- Predominantly affects thorax with progressive respiratory symptoms & hemorrhages

IMAGING

- Poorly defined, infiltrative, fluid signal intensity/attenuation soft tissue lesions of mediastinum & chest wall + peribronchial & interlobular septal thickening of lung parenchyma, ± pleural & pericardial effusions
  - Enhancement of infiltrating abnormal lymphatic tissue on T1WI C+ FS MR
- Location
  - Thorax: Mediastinum (100%) > lung, pleura (80%)
  - Bone, spleen (50% each, typically as discrete lesions)
  - Subcutaneous tissues, retroperitoneum

TOP DIFFERENTIAL DIAGNOSES

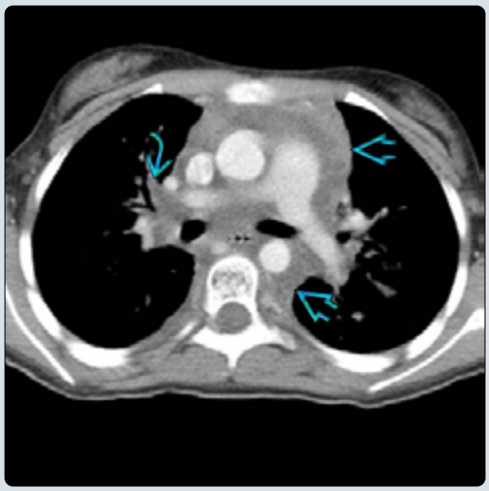
- GLA
- Gorham-Stout disease
- Lymphoma
- Langerhans cell histiocytosis
- Cat scratch disease

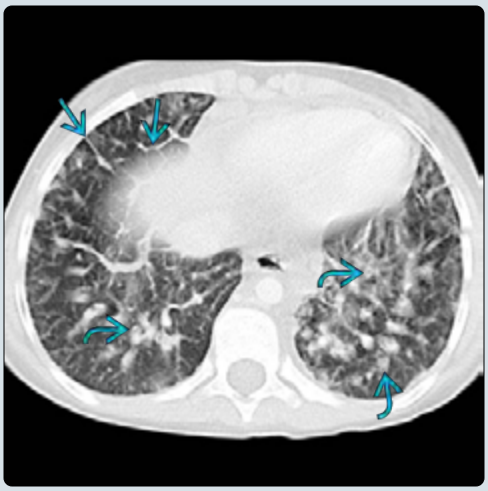
PATHOLOGY

- Abnormal lymphatic channels with spindled endothelial cells; stain positive for lymphatic markers PROX1, D2-40
- Intermixed extravasated red blood cells, hemosiderin

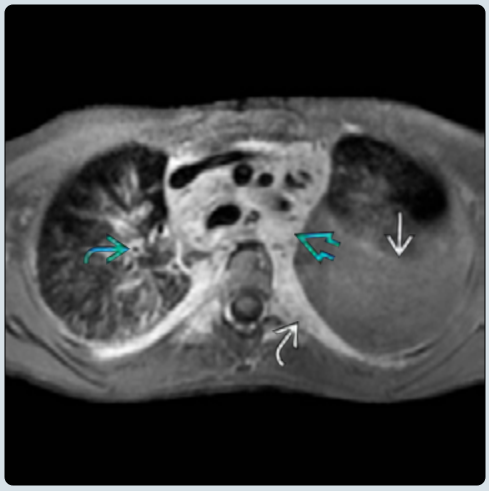
CLINICAL ISSUES

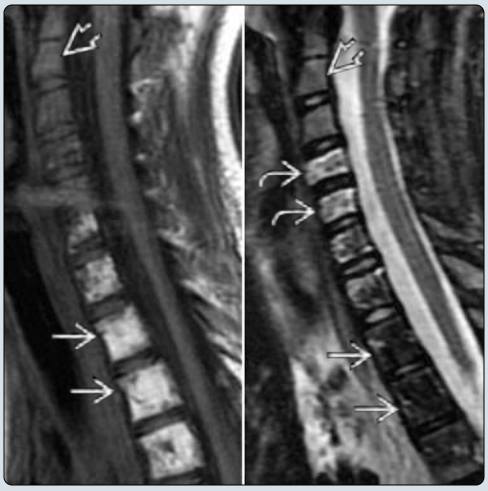
- Most common presentations
  - Respiratory (50%): Cough, dyspnea
  - Hemorrhage from underlying coagulopathy (50%)
  - Mass (35%)
- Median age of presentation: 6.5 years
- 5-year survival of ~ 50%





(Left) *Axial CECT in an adolescent with shortness of breath shows infiltration & expansion of the peribronchial ⇨ & mediastinal ⇨ soft tissues by poorly defined fluid-attenuation lesions.* **(Right)** *Axial CECT (in lung windows) of the same patient with kaposiform lymphangiomatosis (KLA) shows extensive peribronchial ⇨ & interlobular septal ⇨ thickening.*





(Left) *Axial T1 C+ FS MR in the same patient several months later shows peribronchial thickening ⇨ with diffuse enhancement of the infiltrating mediastinal ⇨ & pleural ➡ lesions. A large left pleural effusion ➡ has developed.* **(Right)** *Sagittal T1 (left) & STIR (right) MR images show multifocal vertebral lesions in the same patient. Some lesions shows abnormal fatty deposition ➡ compared to normal (for this age) vertebral marrow ➡, while other lesions show increased fluid signal intensity ➡.*

SECTION 3

Cardiac

Cardiomyopathies

Miscellaneous

Imaging Anatomy

The days in which congenital heart disease (CHD) was characterized by chest radiography & defined by angiography are gone. Both still play some role in the diagnosis & management of CHD. However, with echocardiography, cardiac MR, & CT, we now have robust imaging modalities that can provide detailed anatomic & functional evaluation in patients with complex heart issues. A patient who undergoes a CT or MR for CHD will often have multiple findings. For example, an asplenia heterotaxy patient with right-sided isomerism may have a long list of diagnoses, such as total anomalous pulmonary venous return (TAPVR), transposition of great arteries, patent ductus arteriosus (PDA), atrial septal defect (ASD), ventricular septal defect (VSD), single coronary artery, situs inversus, coarctation, & tracheal stenosis. This is not out of the ordinary in a complex CHD patient referred for advanced imaging (CT or MR). A systematic approach is therefore crucial in any cross-sectional imaging modality of a patient with CHD. This introduction attempts to provide a systematic approach to cardiac CT or MR in a patient with CHD.

Situs

Evaluation of situs involves the designation of an individual as normal (situs solitus), reversed (situs inversus), or some other combination (situs ambiguous). In situs solitus, the stomach, left atrium, apex of the heart, & spleen are on the left, while the right atrium, liver, superior vena cava (SVC), & inferior vena cava (IVC) are on the right. The pulmonary veins drain to the left atrium. The tracheobronchial branching in situs solitus includes a trilobed lung & main bronchus that sits posteriorly &/or superiorly to the pulmonary artery (eparterial bronchus) on the right & a bilobed lung with a main bronchus sitting below the pulmonary artery on the left (hyparterial bronchus). Situs inversus is the exact opposite of situs solitus. There are multiple variations to the above basic designation of situs. These fall into the category of situs ambiguous. Some additional observations will often be necessary. In the abdomen, besides looking at the side of the stomach, one must look for the position of the liver, absence of the spleen or presence of multiple spleens, & presence of an intrahepatic IVC (to exclude azygous continuation of the IVC). In the heart, be aware of symmetric atria & appendages in patients with heterotaxy as these may represent 2 right atria or 2 left atria in right or left isomerism. It is usually not difficult to differentiate the ventricles based on the prominence of the trabeculations (i.e., the more trabeculated ventricle is usually the right ventricle). The right ventricle may more confidently be identified by the presence of a moderator band (muscular fibers connecting the free wall of the right ventricle to the interventricular septum). Also, pay attention to the tracheobronchial branching relative to the pulmonary arteries: It may be abnormal bilateral left-sided branching (hyparterial bronchi) or bilateral right-sided branching (eparterial bronchi).

Lungs & Airways

There is an increased incidence of airway anomalies in children with CHD. These include pig bronchus, congenital stenosis, & bilateral right- or left-sided tracheobronchial branching seen with heterotaxy syndromes. More common than congenital airway anomalies is airway compression from cardiomegaly & enlarged or anomalous vasculature. The child with CHD is also susceptible to chronic lung disease from extended mechanical ventilation as well as postoperative effusions, atelectasis, tracheobronchomalacia, & infections.

Atria & Veins

It is important to make sure the venous drainage empties into the appropriate chamber of the heart. Anomalies like TAPVR & partial anomalous pulmonary venous return can easily be missed unless specifically investigated. Always look for a left SVC & a crossing innominate vein. The unsuspected presence of a left SVC may complicate cardiac surgery & cardiopulmonary bypass unnecessarily. Also, scan the atrial septum for defects. Artifact can sometimes hide or mimic atrial septal defects. As technology advances, the ability to evaluate valvular & septal structures will also improve. The artrioventricular valves should be evaluated for position, size, & thickness. They can also be difficult to evaluate if there is motion, beam hardening artifact, or a delayed contrast bolus.

Ventricles

A ventricular septal defect is any communication of the right & left ventricles. These defects are most commonly in the membranous portion of the interventricular septum. In complex congenital heart patients, defects are also frequently seen in the muscular portion of the ventricular septum. Rarely, defects can also occur along the outflow tracts. Included in the evaluation of the ventricles are the right & left ventricular outflow tracts. Outflow tract evaluation includes assuring the patency (no atresias), separation (no truncus anomaly), & connection to the appropriate great vessel (no transposition).

Great Vessels

The size, course, & position of the great vessels should be evaluated by CT & MR. Basic measurements of the size of the aorta are made at the sinuses of Valsalva, aortic valve annulus, sinotubular junction of the ascending aorta, transverse aortic arch, & descending aorta. Basic measurements are made for the main pulmonary artery at the pulmonary valve annulus, mid main pulmonary artery, & proximal & distal right & left pulmonary arteries. Any other obvious stenosis or dilation should also be measured. Vascular rings, slings, & aberrant vessels should be identified. A patent ductus arteriosus is usually present in complex congenital heart patients. Expect to find a patent ductus arteriosus & measure its length & width.

Coronary Arteries

The origin, course, & termination of the coronary arteries should be evaluated when possible. Visualization of the coronaries is variable depending on the type of exam performed. Generally, the coronaries are best seen with an ECG-gated cardiac CTA. It is important to note that certain coronary anomalies that will alter the surgical approach. Rarely, the left (less commonly the right) coronary artery may arise from the pulmonary artery (ALCAPA). This can lead to a steal phenomenon & cause the patient to go into heart failure. Sudden death can occur in patients (typically athletes) with an interarterial (malignant) course of the coronary arteries. This is most commonly seen when the left coronary arises from the right coronary sinus or when the right coronary artery arises from the left coronary sinus. The aberrant artery then courses between the right & left ventricular outflow tracts. Some coronary anomalies are associated with certain types of CHD. This helps focus the search for the coronary anomalies. For example, patients with transposition of the great arteries (TGA) typically have a right coronary arising from the

noncoronary sinus. If the coronary anatomy is something different from the typical pattern in TGA patients, the surgeon should be alerted as he or she will have to translocate the coronaries during the arterial switch procedure. Coronary artery fistulas are frequently seen in patients with a hypoplastic right ventricle. It is common to see a partial interarterial course of the right coronary artery in a patient with tetralogy of Fallot since clockwise rotation of the aortic root is seen in a large percentage of these patients. Finally, for any anomaly where a coronary artery courses in front of an outflow tract, alert the surgeon so he or she can avoid severing the vessel during surgery.

Functional

Basic functional evaluation of the heart can be performed by both CT & MR cardiac exams. Routine functional evaluation includes calculating the volumes & ejection fractions of both ventricles. Indexed volumes are obtained by dividing the gross volume by the body surface area. CT & MR can also be used to evaluate the left ventricular muscle mass. Standardized data is available to compare volume & muscle mass results with other size & age matched data sets. MR has the added capabilities to evaluate & quantify flow volumes & velocities. A pulmonary to systemic flow ratio (Qp/Qs) is routinely provided by interrogating the flow from the right & left ventricular outflow tracts. In patients with pulmonic & aortic regurgitation, velocity-encoded phase-contrast imaging can be used to calculate regurgitant fractions. The regurgitant fraction is calculated by dividing the regurgitant volume by the stroke volume. Similar applications can be used to evaluate other veins, arteries, & valves as needed. Regurgitant fractions are typically followed yearly in patients with a pulmonary homograft to assess the severity of pulmonic regurgitation. When MR thresholds of end-systolic, end-diastolic, & pulmonic regurgitant fractions are met, a pulmonic valve replacement is recommended.

Anatomy-based Imaging Issues

It is crucial to know as much as possible about the patient's anatomy & postsurgical history before protocoling a cardiac CT or MR. For example, IVC (Fontan) & SVC (Glenn) shunts may introduce unopacified blood directly into the pulmonary arterial system. This can simulate pulmonary embolism when unsuspected. Thoughtful protocoling of patients with CHD can help avoid artifacts that may simulate pathology & provide additional information to the cardiologist & cardiothoracic surgeon to better care for these patients.

Indications for Cardiac MR & CT

The routine congenital heart patient with a simple lesion (ASD, PDA, VSD) would likely never undergo an advanced imaging study, such as a cardiac CT or MR. Echocardiography provides excellent delineation of the intracardiac anatomy, great artery relationships, & function of the heart. Cardiac CT & MR are reserved for problem solving in difficult echocardiography cases.

Extracardiac anatomy can be difficult to completely evaluate by echocardiography in complex cases. CT & MR are frequently done to evaluate the pulmonary veins, pulmonary arteries, systemic veins, & aorta & its branches. Heterotaxy patients often have abnormalities involving the extracardiac anatomy & airways & may now routinely undergo cardiac CT or MR.

There are certain circumstances where the evaluation of the intracardiac anatomy may be helpful by CT & MR. These may include cardiac tumors, cardiomyopathies, ischemic heart disease, myocarditis, & coronary artery abnormalities. A more accurate volumetric analysis with CT or MR may be desirable in patients to evaluate the possibility of a 2-ventricle repair. MR has proven to be an effective tool to follow tetralogy of Fallot patients with pulmonic regurgitation to determine the timing of pulmonary valve replacement.

Cardiac MR vs. Cardiac CT

With higher temporal resolution, cardiac MR is superior to cardiac CT for functional imaging of the heart & evaluation of intracardiac anatomy. With higher spatial resolution, cardiac CT is superior to MR for extracardiac anatomy, airway, & coronary artery evaluation. There is significant overlap between the 2 modalities, as cardiac CT can provide volumetric functional evaluation & MR gives adequate delineation of the extracardiac anatomy.

Cardiac MR requires longer sedation times than CT.

Cardiac CT exposes the patient to ionizing radiation. Adjusting for size & using prospective gating, as well as other techniques, can significantly reduce the radiation dose.

Cardiac 3D Models

Segmentation of the anatomy in a patient with complex disease is often performed, & 3D images are used in presurgical planning. Once the images are segmented, they can be exported to 3D printers to create physical models of the heart. 3D printers are now widely available & can print the models using a variety of flexible & nonflexible materials. Simple resin models of the heart can be printed & used in educating parents, patients, & other health professionals. 3D congenital heart models are now used in the teaching of physicians in training at all levels. These patient-specific models can be printed in flexible material & used to test devices on the patient's anatomy before performing the actual procedure. This method has proven effective in decreasing procedure times. Examples include the coiling of aneurysms, sizing of devices for closure of intramuscular ventricular septal defects, & Melody valve placement along the pulmonary outflow tract.

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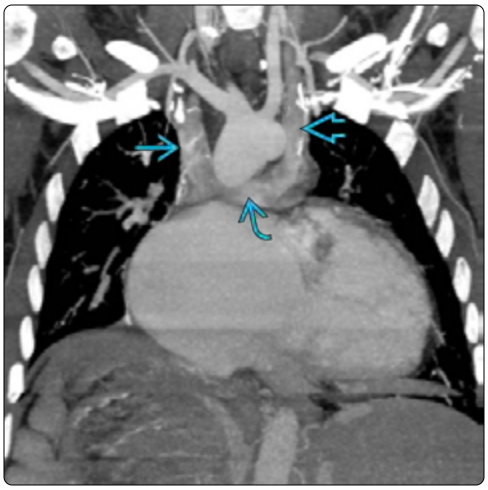
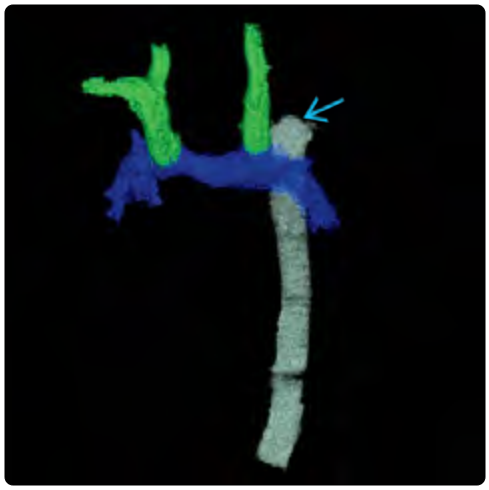
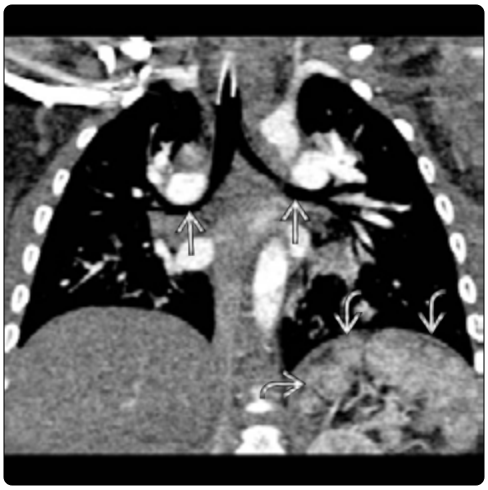
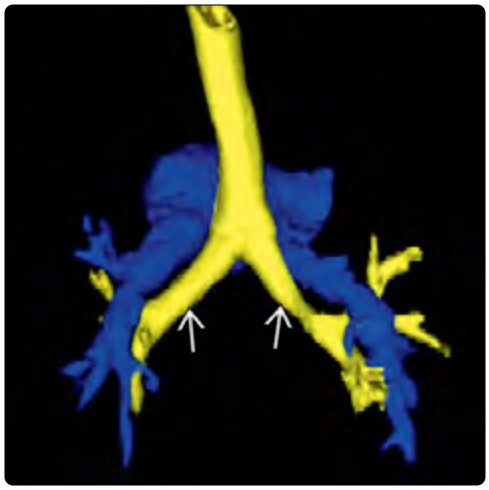
(Left) *Coronal image from a cardiac CTA shows right & left SVCs connected to the pulmonary arteries, consistent with bilateral Glenn procedures. Bilateral upper extremity injections were performed to get even contrast enhancement of the pulmonary arteries & exclude embolism.* **(Right)** *Frontal 3D image from a cardiac CTA shows bilateral SVCs (green) connected to the pulmonary arteries (blue), consistent with a Glenn procedure. Notice the azygous vein also connecting to the left pulmonary artery.*



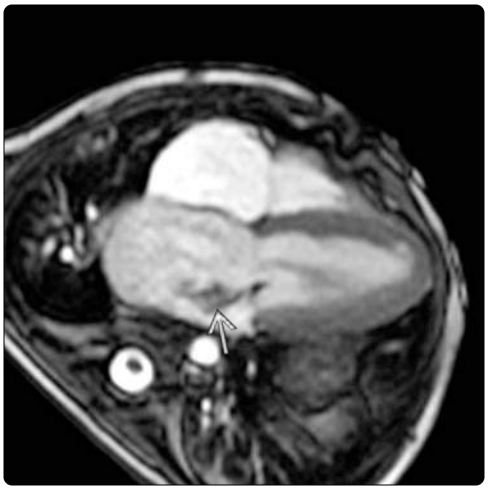


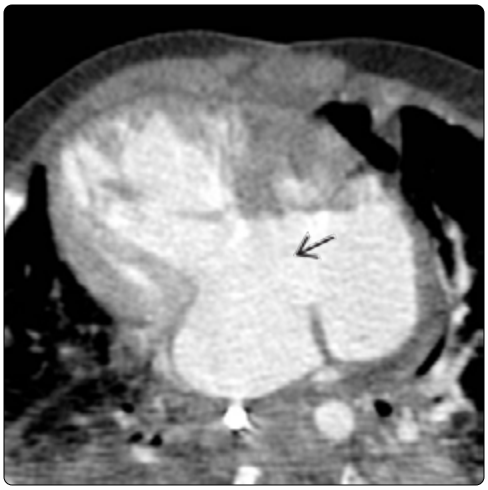
(Left) *A single coronal image from a cardiac CTA shows bilateral hyparterial bronchi with multiple spleens in this patient with polysplenia syndrome.* **(Right)** *Posterior image from a 3D color-coded cardiac CTA shows bilateral hyparterial bronchi with the left & right pulmonary arteries (blue) passing superior to the main bronchi (yellow).*



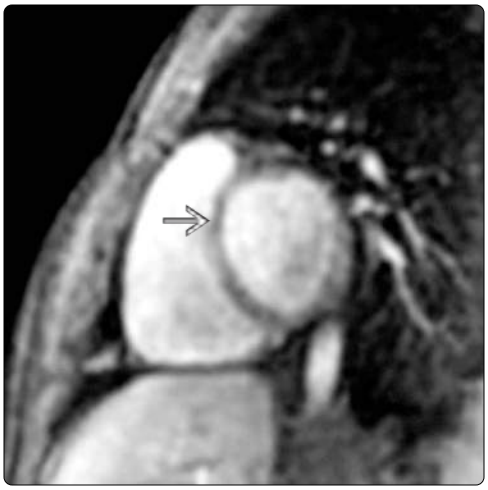


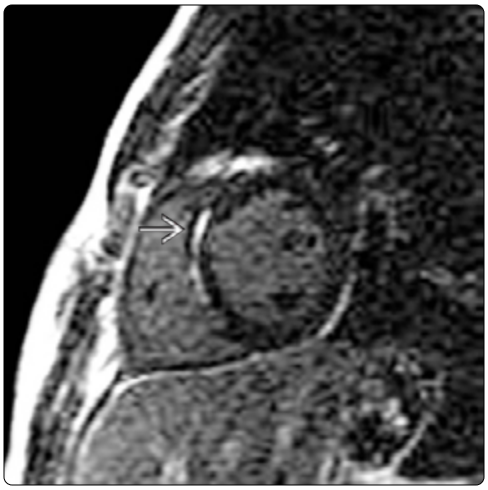
(Left) *Anterior 3D image of a CTA shows an anomalous vertical vein draining blood from the pulmonary venous system (purple) to the innominate vein. Notice that a right single pulmonary vein loops around a narrowed left bronchus.* **(Right)** *A single frontal image from a 3D color-coded chest CTA shows a right arch (red) with an aberrant left subclavian artery. Notice the tortuous path of a patent ductus arteriosus (green) arising from the left subclavian & inserting into the left pulmonary artery, completing the vascular ring.*



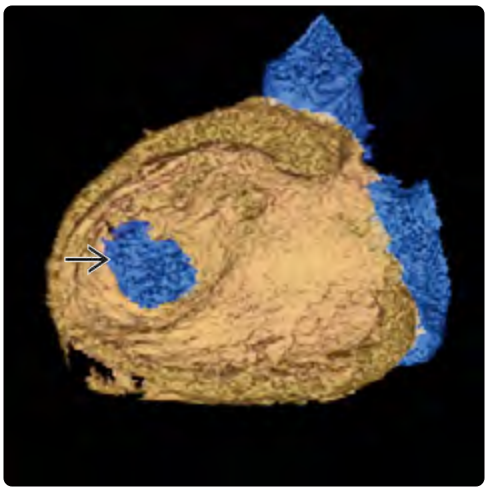


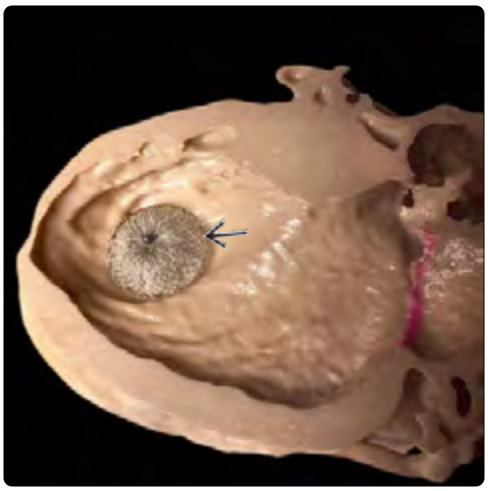
(Left) *A single image from a 4-chamber view SSFP cine cardiac MR shows dephasing artifact ⇒ directed back into the left atrium from the mitral valve in this patient with mitral regurgitation.* **(Right)** *A single axial image from a cardiac CTA shows a large defect in the atrial septum ⇨ adjacent to the endocardial cushion in this patient with dextrocardia & a septum primum atrial septal defect.*





(Left) *An immediate postcontrast short-axis image from an MR cardiac perfusion study shows decreased uptake in the interventricular septum ⇒.* **(Right)** *Late gadolinium enhanced short-axis image from an MR cardiac perfusion study in the same patient shows increased uptake in the mid muscular portion of the interventricular septum ⇒. This was consistent with myocarditis in this 10 year old with acute onset heart failure.*





(Left) *Lateral 3D view from a segmented digital model looking from within the left ventricle shows a large intramuscular ventricular septal defect (VSD) ⇨ in a patient that had suffered infarction of the interventricular septum. Images were used in presurgical planning for closure device placement.* **(Right)** *Resin 3D-printed model looking from within the left ventricle at the interventricular septum shows placement of a closure device ⇨ within an intramuscular VSD.*

Atrial Septal Defect

KEY FACTS

TERMINOLOGY

- ASD: Defect(s) in cardiac atrial septum; may be isolated anomaly or associated with other congenital heart lesions
- Left-to-right (LTR) shunt: Blood from left heart bypasses systemic circulation to enter right heart
  - Most ASD sequelae related to long-term LTR shunting
- Types of ASD
  - LTR shunts: Ostium secundum (70-90%), ostium primum, sinus venosus, unroofed coronary sinus defects
  - Patent foramen ovale only allows right-to-left shunting, usually transient given normal atrial pressures
    - Increased stroke risk; unclear migraine association

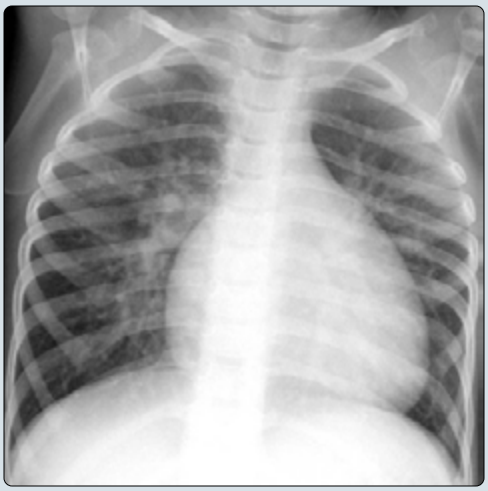
IMAGING

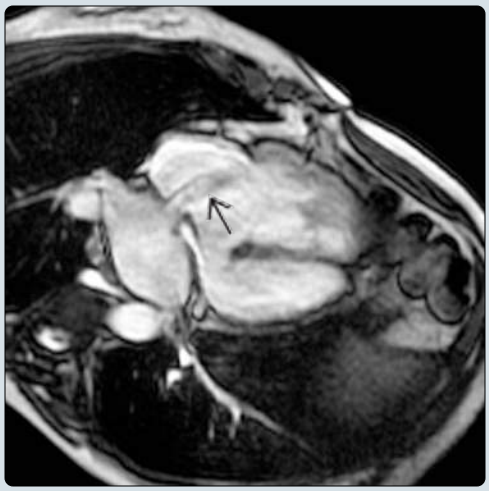
- LTR shunting leads to chronic volume overload of right heart, eventual enlargement of RA, RV, & PA
  - Secondary findings & symptoms uncommon in children
- Diagnosis of actual defect primarily made by echocardiography
- Cardiac MR accurate alternative for depiction of function, flow, & anatomy

CLINICAL ISSUES

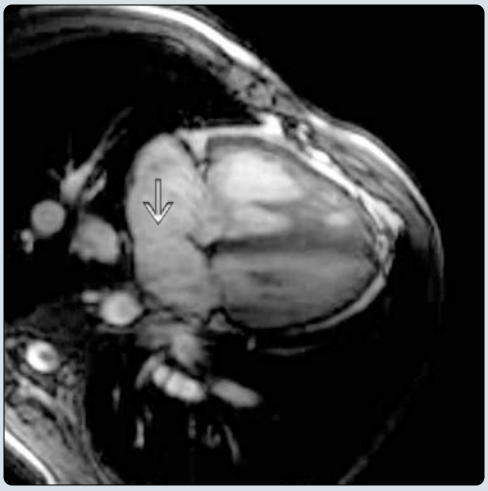
- ASD: 10% of CHD in children, yet 30% of CHD in adults
- Secundum ASD: Majority of patients asymptomatic
  - Spontaneous closure occurs in many children
  - Subtle symptoms more likely in 2nd decade, though large defects frequently do not present until adulthood
    - Fatigue, exercise intolerance, syncope, shortness of breath, palpitations
    - ASD leading to severe pulmonary hypertension: Median age of detection is 51 years
  - Repair indicated if shunt ratio > 1.5:1 or defect > 10 mm
  - Percutaneous closure with occlusion device
- Primum/atrioventricular septal defect more severe; requires early surgical repair
- Sinus venosus ASDs also require surgery due to complex anatomy

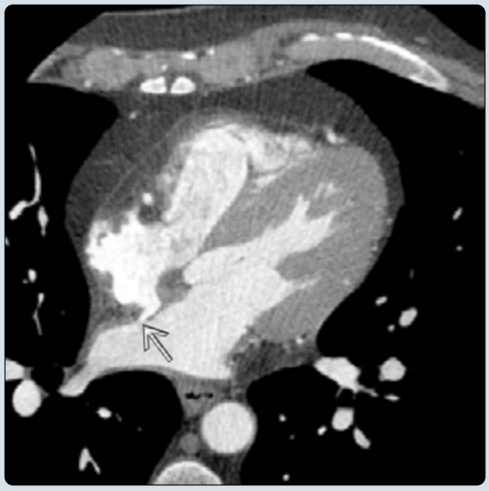
(Left) *A single frontal chest radiograph in a 4-year-old child demonstrates cardiomegaly, increased pulmonary vascularity, & mild pulmonary edema. The patient had a large, untreated atrial septal defect (ASD).* **(Right)** *4-chamber view from an SSFP (bright blood) cardiac MR shows a secundum-type defect of the atrial septum. Note the left-to-right (LTR) flow across the ASD with dephasing artifact ⇨ in the right atrium (RA).*





(Left) *4-chamber view from an SSFP (bright blood) cardiac MR shows a sinus venosus ASD with the defect in the superolateral aspect of the atrial septum ➡. This defect is nearly always associated with right upper lobe partial anomalous pulmonary venous return.* **(Right)** *Axial image from a coronary CTA shows a patent foramen ovale ➡ with a typical oblique defect & flap in the atrial septum. This is seen in 25% of the population. Note that the size of the RA is normal as LTR shunting does not occur.*





TERMINOLOGY

Abbreviations

- Atrial septal defect (ASD)
- Congenital heart disease (CHD)

Definitions

- ASD: Defect(s) in cardiac atrial septum; may be isolated anomaly or associated with other congenital heart lesions
- Left-to-right (LTR) shunt: Blood from left heart bypasses systemic circulation to enter right heart
  - Most ASD sequelae related to long-term LTR shunting

IMAGING

General Features

- Best diagnostic clue
  - Defect in atrial septum seen on any imaging modality
  - Sequelae of isolated ASD uncommon in children
- Location
  - Patent foramen ovale: Persistence of normal fetal interatrial communication that allows flow from inferior vena cava (IVC) to largely bypass right atrium (RA) & freely enter left atrium (LA) in utero
    - Most common (25-30% of adults), but not generally considered with other ASDs as flap only allows flow from right to left (not LTR shunting)
      - Only occurs with elevations in right heart pressures, typically transient phenomenon
      - Main concern in this population is stroke; questionable association with migraine headaches
  - Ostium secundum ASD (70-90% of LTR shunt ASDs): Defect of fossa ovalis due to defect of septum primum
  - Ostium primum ASD (2-3%): Simplest form of atrioventricular septal (endocardial cushion) defect (AVSD); this ASD lies between anterior/inferior portion of septum & AV valves
  - Sinus venosus ASD (4-11%): Posterior to fossa ovalis; superior-type (communication of cardiac SVC & at least 1 right pulmonary vein) > inferior-type (posterior inferior right atrial wall) defects
  - Unroofed coronary sinus (least common): Defect between LA & coronary sinus roof
- Size
  - Variable
- Morphology
  - Many ASDs irregular or complex, not circular

Radiographic Findings

- Chest radiograph findings
  - Ostium secundum ASD
    - Small to moderate defects: Normal chest radiographs
    - Large defects: Mild cardiomegaly, normal to increased main pulmonary artery (PA) size, shunt vascularity
  - Ostium primum ASD or AVSD
    - Young child with cardiomegaly & increased pulmonary vascularity
    - Volume overload of RA, right ventricle (RV), & PA
  - Sinus venosus defect, superior type
    - Horizontal position of right upper pulmonary vein as it enters SVC
  - Adults with pulmonary hypertension
    - Classic enlarged, convex main PA with peripheral decrease in size of vessels
    - Right ventricular enlargement
      - Upturned cardiac apex on frontal view
      - Diminished retrosternal clear space on lateral view

CT Findings

- CTA
  - Defect in atrial septum
    - Large shunt suggested with unexpected equalization of contrast between atria during cardiac CTA with saline chaser
  - Enlargement of RA, RV, PA
  - Useful to look for associated congenital heart lesions
    - Partial anomalous pulmonary venous return (PAPVR) associated with sinus venous ASD
  - Volumetric functional information obtained with retrospective gating

MR Findings

- SSFP cine
  - Shunt quantification using biventricular stroke volume ratio; net shunt volume = difference in stroke volumes
  - Interrupted septum visible on 4-chamber views & short-axis stacks
    - Shunting blood may show signal loss (dephasing) on these bright blood sequences
- Gradient-recalled echo cine with saturation band
  - Saturation band applied across one atrial chamber
  - Shunting from chamber of saturated blood (dark) into chamber of unsaturated blood (bright) reveals defect
- First-pass gadolinium perfusion
  - Rapid dynamic imaging can show ASD by
    - Dark (unenhanced) LA blood shunting into bright (enhanced) RA, followed by bright (enhanced) LA blood shunting into less enhanced RA
- Phase contrast velocity
  - Shunt ratio (Qp/Qs) calculated by determining velocity & flow in ascending aorta & main PA
    - Qp/Qs of 1.5:1 typically symptomatic
  - Also used to measure ASD flow directly
  - En face evaluation accurately depicts ASD size & shape as well as rim assessment
    - Critical parameters for assessing possibility of percutaneous closure
- MR angiography
  - Depiction of anomalous pulmonary veins

Angiographic Findings

- Utilized for transcatheter percutaneous treatment with closure device

Imaging Recommendations

- Best imaging tool
  - Primary diagnosis made by echocardiography
  - Cardiac MR accurate alternative for depiction of function, flow, & anatomy
  - Catheterization for percutaneous closure

DIFFERENTIAL DIAGNOSIS

Normal Chest Radiograph

- Main PA can be prominent normally, particularly between ages 8-12 years

Ventricular Septal Defect

- Small shunts have normal radiographs
- Moderate or large shunts have cardiomegaly & increased PA flow

Pulmonary Hypertension

- Variety of causes, mostly secondary to chronic lung disease
- Enlarged main & central PA with pruning distally, mosaic perfusion on CT

PATHOLOGY

General Features

- Genetics
  - Down syndrome (trisomy 21): Up to 65% with CHD
    - Up to 45% due to AVSD; up to 42% due to ostium secundum ASD
  - Holt-Oram syndrome (*TBX5* gene mutation on 12q24): ASD plus upper extremity anomalies
  - Various other syndromes
  - Most ASDs sporadic
    - Mutations have been found in several genes associated with cardiac septation
- Associated abnormalities
  - Sinus venosus
    - Superior: PAPVR of RUL pulmonary vein to SVC
    - Inferior: PAPVR of RLL pulmonary vein to IVC or RA
  - Ostium secundum
    - Mitral valve degeneration with regurgitation in 25%
  - Unroofed coronary sinus: Persistent left-sided SVC
- Pathophysiology: Volume overload primary issue
  - ASD: Low-pressure LTR shunt
  - VSD, AVSD: High-pressure LTR shunts
  - All eventually lead to pulmonary hypertension if untreated

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Most children with ASDs have no symptoms
    - Detected due to murmur or other medical work-up
  - Rarely present in childhood with failure to thrive, respiratory infection, tachypnea
  - Subtle symptoms more likely in 2nd decade, though large defects frequently do not present until adulthood
    - Fatigue, exercise intolerance, syncope, shortness of breath, palpitations
  - ASD leading to severe pulmonary hypertension: Median age of detection is 51 years
  - Ostium secundum, sinus venosus: Majority of patients asymptomatic
  - AVSD or ostium primum defects: More likely to have early symptoms
- Other signs/symptoms
  - Abnormal heart sounds
    - Crescendo-decrescendo systolic ejection murmur of 2nd left intercostal space
      - Increased volume through pulmonic valve
    - Widely split 2nd heart sound
      - Delayed closure of pulmonic valve

Demographics

- Age
  - Congenital defect; most ASDs asymptomatic in infancy with symptoms developing in adulthood
  - AVSD detected prenatally or in 1st week of life
- Gender
  - Ostium secundum defects: F:M = 2:1
- Epidemiology
  - 3rd most common CHD
  - ASD: 10% of CHD in children, yet 30% of CHD in adults
  - 56-100/100,000 live births
    - True incidence may be higher as many close spontaneously (especially ostium secundum)

Treatment

- Repair indicated: Shunt ratio > 1.5:1 or defect > 10 mm
  - Repair unless child under 2 years of age (due to rates of spontaneous closure)
- Patients with small shunts: Monitor for right heart dysfunction
- Repair contraindicated if pulmonary hypertension has already developed
- Ostium secundum ASD
  - Spontaneous closure occurs in many children
  - If persistent, close percutaneously with catheter-placed occlusion device
  - Large defects may need patch or direct suture closure
- Ostium primum defects not amenable to percutaneous device closure due to relationship to AV valves
  - Surgery by 3-5 years of age
- Sinus venosus defect: More complex surgical repair required due to pulmonary vein anatomy

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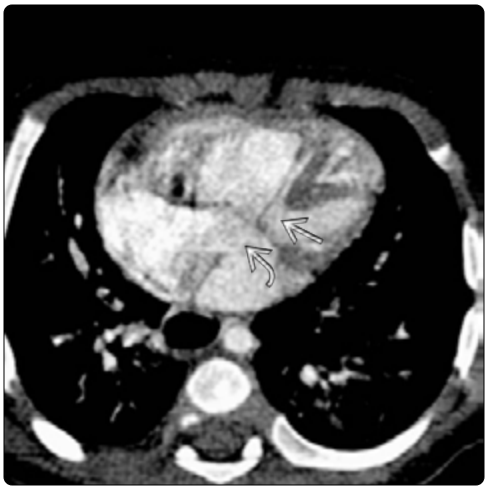
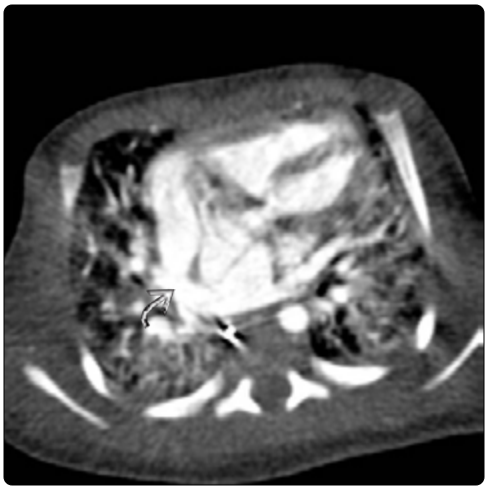
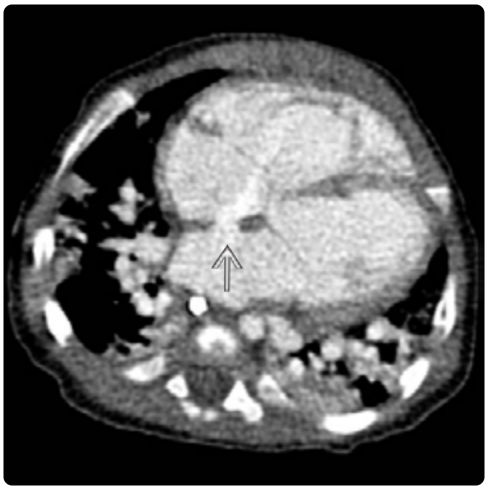
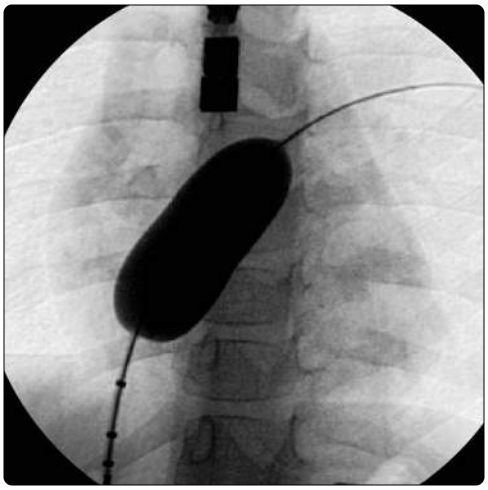


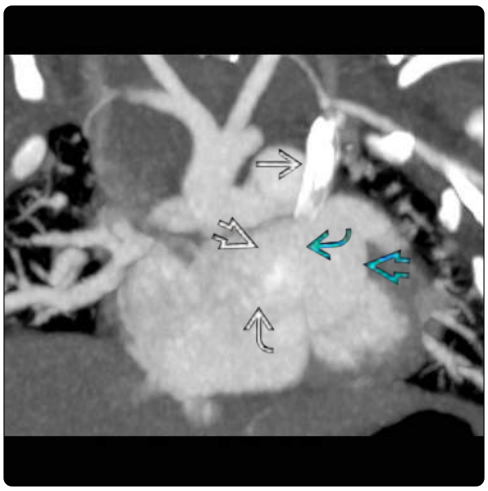


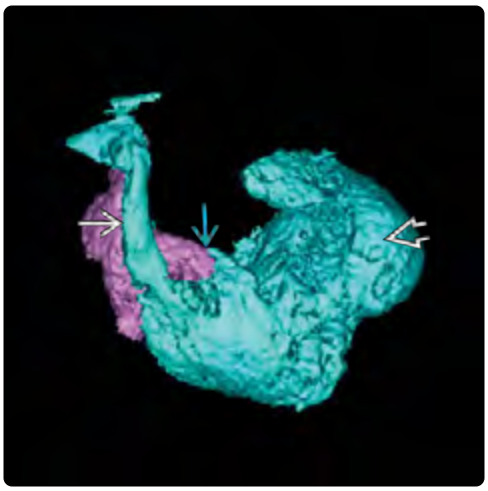
(Left) *Axial image from a cardiac CTA in a newborn shows a septum primum defect. This defect is seen adjacent to a common atrioventricular (AV) valve. Note the VSD in this patient with a complete AV canal-type defect.* **(Right)** *Axial image from a cardiac CTA in a newborn shows a sinus venosus ASD with the defect seen in the superolateral aspect of the atrial septum.*





(Left) *Axial image from a cardiac CTA of a newborn shows a typical septum secundum ASD with the defect seen in the mid portion of the atrial septum.* **(Right)** *A single frontal fluoroscopic image of the chest demonstrates a balloon in an ASD of a patient undergoing a Rashkind balloon septostomy. A larger ASD was needed in this patient with transposition of the great arteries to improve mixing of the oxygenated & deoxygenated blood.*





(Left) *Coronal MIP CTA shows a left SVC with drainage to a markedly enlarged coronary sinus. A defect is seen superiorly between the coronary sinus & left atrium, consistent with an unroofed coronary sinus defect.* **(Right)** *Superior view from a color-coded 3D CTA shows a left SVC draining to a dilated coronary sinus, which then empties into right atrium. Note communication between superior aspect of coronary sinus & left atrium; this was confirmed surgically as unroofed coronary sinus.*

Ventricular Septal Defect

KEY FACTS

TERMINOLOGY

- Cardiac anomaly with communication(s) between left & right ventricles through septum
  - Perimembranous septal defect (80%)
  - Posterior or inlet defect associated with atrioventricular septal defect (AVSD) (8-10%)
  - Muscular or trabecular septal defect (5-10%)
  - Outlet septal defect or supracristal ventricular septal defect (VSD) (5%)
- Complex cardiac anomalies with VSD: TOF, truncus, DORV

IMAGING

- Cardiomegaly with ↑ size of main pulmonary artery, ↑ pulmonary artery flow, left atrial enlargement, & usually small aorta
- Hyperinflation in large shunts from abnormal lung compliance & bronchial compression by dilated pulmonary arteries
- CT & MR delineate anatomy
  - Multiple muscular VSDs: "Swiss cheese" septum
  - Shunt volume estimated by velocity encoded cine MR imaging

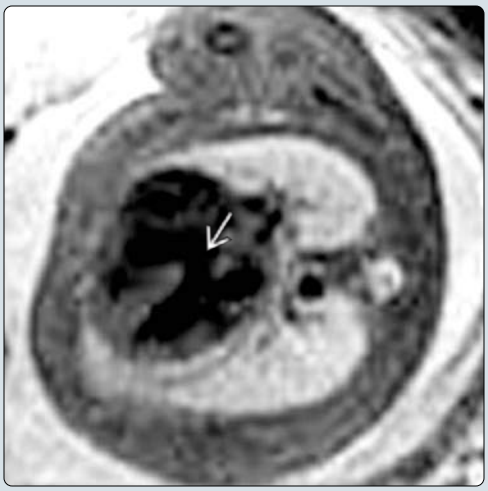
TOP DIFFERENTIAL DIAGNOSES

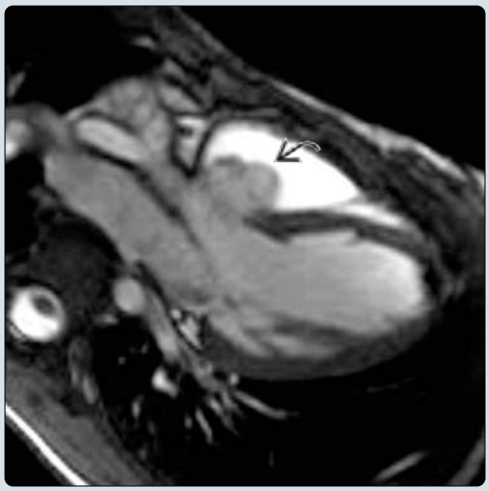
- Atrioventricular canal defects
- Patent ductus arteriosus
- Double outlet right ventricle

CLINICAL ISSUES

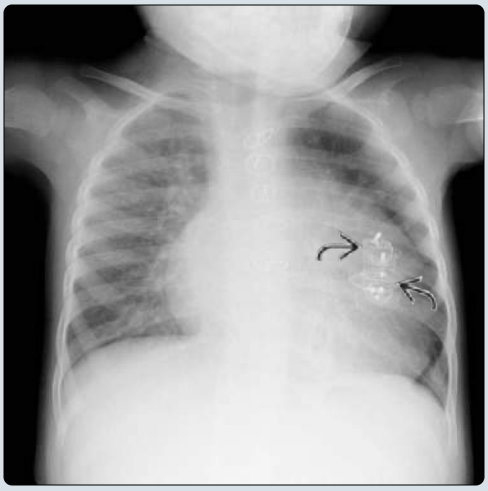
- Small VSD: Children asymptomatic but have heart murmur
  - May close spontaneously
- Moderate or large shunts often asymptomatic early until pulmonary vascular resistance drops
  - Children develop tachypnea, tachycardia, diaphoresis, & failure to thrive
  - Treated medically with subsequent surgical approach
    - Muscular lesions require more difficult surgical approach; VSD catheter closure devices often used

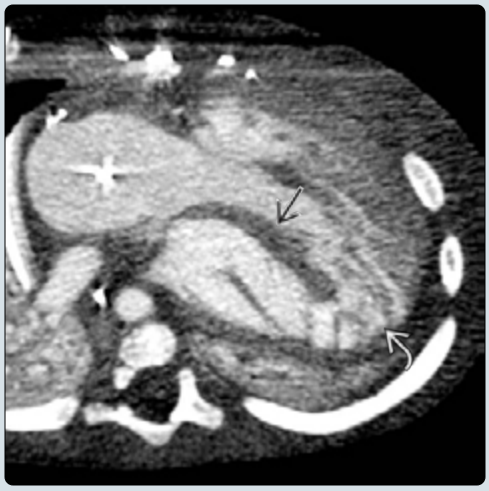
(Left) *Axial SSFSE T2 fetal MR shows a perimembranous-type ventricular septal defect (VSD) in this fetus with tetralogy of Fallot.* **(Right)** *Three-chamber view from a cine sequence cardiac MR shows an aneurysm of the membranous ventricular septum protruding into the right ventricle. This likely represents the spontaneous membranous closure of a previously patent VSD. When large, the aneurysm may cause right ventricular outflow tract obstruction.*





(Left) *Frontal radiograph of the chest shows cardiomegaly with increased pulmonary vascularity in a patient with multiple intramuscular-type VSDs. Multiple VSD closure devices have been placed in an attempt to decrease shunting.* **(Right)** *Oblique axial cardiac CTA shows a Swiss cheese appearance of the interventricular septum near the apex in this patient with multiple muscular-type VSDs. Notice the normal positions of the septum.*





TERMINOLOGY

Abbreviations

- Ventricular septal defect (VSD)

Definitions

- Cardiac anomalies characterized by defect(s) in ventricular septum
  - Perimembranous septal defect
  - Muscular or trabecular septal defect
  - Posterior or inlet defect associated with atrioventricular septal defect (AVSD)
  - Outlet septal defect or supracristal VSD
- Complex cardiac anomalies with VSD
  - Tetralogy of Fallot, truncus arteriosus, double outlet right ventricle
  - Associated with other congenital lesions: Coarctation, tricuspid atresia

IMAGING

General Features

- Best diagnostic clue
  - Chest radiograph with cardiomegaly (particularly left atrial enlargement) + ↑ pulmonary artery flow in small child
  - Defect in ventricular septum on any cross-sectional imaging modality
- Location
  - Membranous or perimembranous defects occur in 80%
    - Defects lie in outflow tract of left ventricle immediately beneath aortic valve
  - Inlet VSD occurs in 8-10%
    - Posterior & inferior defects, beneath septal leaflet of tricuspid valve
    - Associated AVSD with usual involvement of atrioventricular valves
  - Outlet VSD occurs in 5%
    - Conal, subpulmonic, subaortic, supracristal, or infundibular
    - Malalignment defects associated with truncus arteriosus, tetralogy of Fallot, & double outlet right ventricle
    - Supracristal defect located about crista muscle high in ventricular outlet portion
      - May cause prolapse of aortic coronary cusp with development of aortic insufficiency & injury to aortic valve
  - Muscular or trabecular VSD occurs in 5-10%
    - Confined to muscular portion of interventricular septum
    - Central muscular, apical muscular, or marginal; may have multiple defects described as "Swiss cheese" muscular septum
- Size
  - Defects can be small, moderate, or large & involve adjacent structures

Radiographic Findings

- Small VSD
  - Normal chest radiograph does not exclude small shunt
- Moderate to large VSD
  - Cardiomegaly with ↑ size of main pulmonary artery, ↑ pulmonary artery flow, left atrial enlargement, & (usually) small aorta
    - Main pulmonary artery high in position in infants, frequently confused with aortic knob
  - Heart failure may occur with venous edema
  - Hyperinflation seen in large shunts due to abnormal lung compliance & possibly bronchial compression by dilated pulmonary arteries
- Supracristal VSD
  - Left-to-right shunt usually small as anterior leaflet of aortic valve prolapses & may partially cover defect
  - May have evidence of dilated ascending aorta if aortic insufficiency present
  - Difficult to diagnosis on chest radiographs

CT Findings

- Cardiac gated CTA
  - Delineates cardiac anatomy ± functional & volumetric evaluation
  - 3D images for planning percutaneous closure device
  - 3D printed models (made from various materials) now occasionally used to test closure device placement before procedure

MR Findings

- Delineates cardiac anatomy & quantification of physiologic function
- Morphologic information provided by ECG-gated spin-echo & cine MR imaging
- Shunt volume can be estimated by using velocity-encoded cine MR imaging
- Qp:Qs ratio (amount of blood flowing from right ventricle compared to left ventricle) may be calculated using phase-contrast imaging of right & left ventricular outflow tracts
  - Qp:Qs ratio > 1.5:1 is 1 indication for closure of VSD
- High-resolution 3D examination of vessels

Echocardiographic Findings

- Characterizes type, location, & number of septal defect(s) as well as function & hemodynamic assessment
- Echocardiography utilized as main diagnostic modality in infants & young children

Angiographic Findings

- Cardiac catheterization & angiography findings
  - Catheterization utilized in complex lesions to obtain hemodynamic information & delineate anatomy

DIFFERENTIAL DIAGNOSIS

Atrioventricular Canal Defects

- Congenital defect involving atrial & ventricular septum & associated atrioventricular valves
- Chest radiograph demonstrates cardiomegaly & ↑ flow
- Presents early with clinical symptoms of large shunt
- High association with trisomy 21

Patent Ductus Arteriosus

- Persistent flow through ductus from high-pressure aorta to main pulmonary artery
- When shunt is large, chest radiograph demonstrates cardiomegaly & ↑ flow

- Presents early & has loud, continuous murmur during both systole & diastole

Double Outlet Right Ventricle

- Both great vessels have their origins from right ventricle
- Aortic-mitral discontinuity present with valves at similar level
- Pulmonary artery pressure lower than systemic with significant flow into pulmonary arteries
  - Clinically & radiographically simulates large left-to-right shunt
- Complex lesion with many variants & classifications

PATHOLOGY

General Features

- Genetics
  - No specific genetic defect in majority
- Embryology
  - Complex, dependent on location of defect & associated anomalies
- Pathophysiology
  - Determinants of left-to-right shunt: Defect size, relative resistance or pressure in ventricular chambers (which may reflect systemic or pulmonary artery pressures)
  - Small defects have high resistance to flow across defect, resulting in small shunts
  - Large-sized VSDs are defined as defects that approximate size of aorta (which may have large flow)
  - ↑ flow across shunt → ↑ work of right ventricle & ↑ volume of venous return to left atrium & ventricle
  - Marked volume overload occurs, & child develops tachycardia + congestive heart failure
  - Long-term ↑ in flow to pulmonary arteries associated with vessel injury → pulmonary hypertension
    - Complex interaction between vascular endothelium & smooth muscle reaction is incompletely understood
    - May be reversible, & those with early pulmonary hypertension may need early surgical closure

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Dependent on size of shunt, associated lesions, & pulmonary vascular pressure
    - Small VSD: Children asymptomatic but have heart murmur
    - Moderate or large VSD: Children have tachypnea, tachycardia, diaphoresis, & failure to thrive
      - Congestive heart failure may occur
- Other signs/symptoms
  - Loud systolic murmur near left heart border

Demographics

- Age
  - Although defect present at birth, children not symptomatic immediately due to high pulmonary vascular resistance of newborn
  - Moderate or large shunts usually symptomatic in 1st few months of life
- Gender
  - M = F
- Epidemiology
  - Accounts for 20% of all congenital heart lesions
  - Most common congenital lesion associated with other heart lesions

Natural History & Prognosis

- Most small muscular VSDs close spontaneously
- Untreated large shunt will develop pulmonary vascular disease
  - Reversal of shunt from right to left with late onset cyanosis
- Associated cardiac anomalies determine final outcome
- Lifetime risk of bacterial endocarditis

Treatment

- Small defects may close spontaneously
  - Includes many small muscular defects
  - Aneurysm of ventricular septum may be part of spontaneous closure
- Moderate & large defects treated medically & followed by surgical approach
  - Medical therapy with diuretics & afterload reduction
  - Many infants improve & will grow
  - Poor growth &/or congestive heart failure may be indication for early surgery
- Surgical treatment depends on site of VSD
  - Perimembranous VSD: Surgical closure of shunt lesion usually performed with right atrial approach on bypass during 1st or 2nd year if shunt moderate or large
  - Outlet defects, such as supracristal VSD, closed earlier to prevent aortic sinus prolapse, injury to valve leaflet, & subsequent aortic regurgitation
  - Muscular lesions require more difficult surgical approach; VSD catheter closure devices often used

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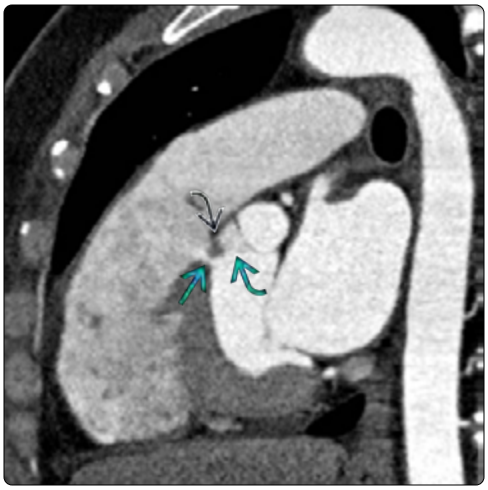
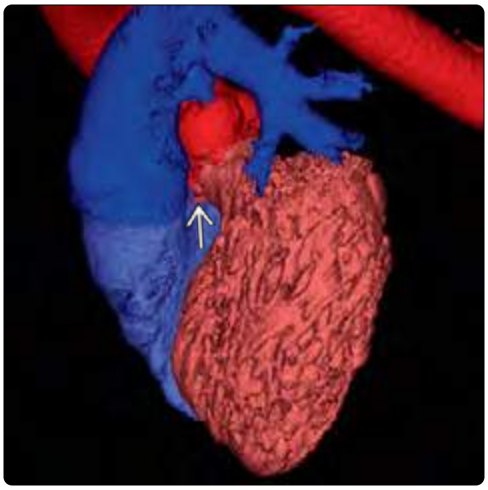
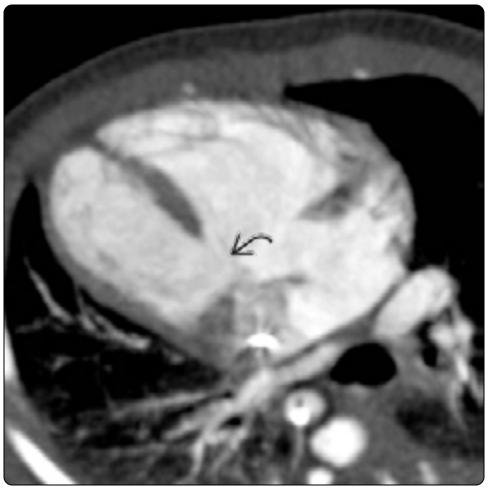
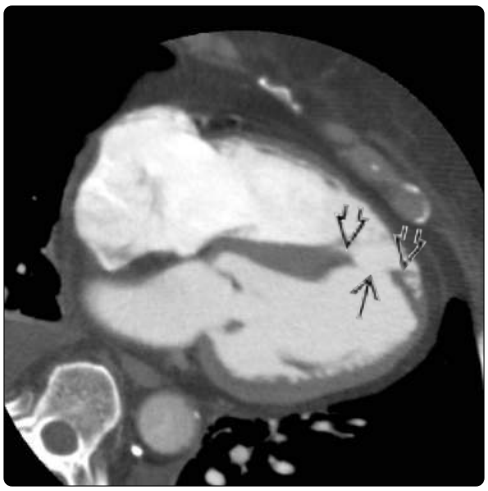


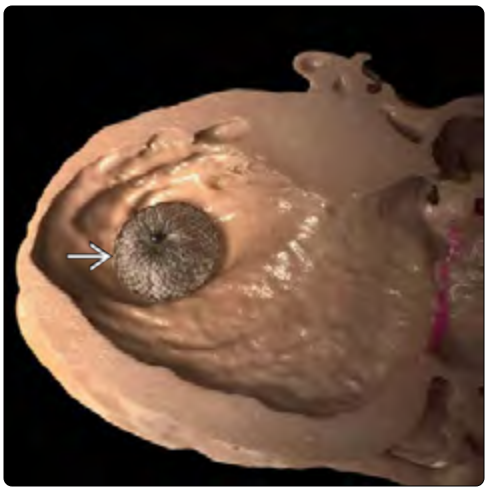


(Left) *Sagittal view from a cardiac CTA shows a small subaortic VSD. Notice the thickened valvular tissue adjacent to the VSD as the patient had a ruptured sinus of Valsalva.* **(Right)** *Volume-rendered 3D oblique CTA shows a small VSD between the left (salmon) & right (purple) ventricles.*



(Left) *Axial image from a cardiac CTA in a Down syndrome patient shows dextrocardia & a posterior-type VSD. Notice that the apex of the heart is to the right in this patient with situs inversus.* **(Right)** *Left lateral view from a color-coded 3D CTA in a patient with D-TGA shows a posterior VSD. Transposition of the great arteries is present with the aorta (red) arising off of the RV (purple). Notice that there is pulmonic atresia with the PDA (green) giving rise to the PAs (blue). Patients with CHD often have multiple findings.*





(Left) *Gated cardiac CTA shows a large intramuscular-type VSD. Notice that there is adjacent thinning of the interventricular septum as this VSD was the result of a myocardial infarction.* **(Right)** *Endoluminal view of the left ventricle from a printed 3D resin model of the heart shows placement of a closure device through a defect in the interventricular septum. This allowed the interventional cardiologist to test several closure devices for the best possible fit.*

Atrioventricular Septal Defect

KEY FACTS

TERMINOLOGY

- Atrioventricular septal defect: Complete atrioventricular canal (AVC) defect, endocardial cushion defect
- Broad spectrum of defects characterized by involvement of atrial septum, ventricular septum, & 1 or both atrioventricular valves
- Ostium primum defect: Partial AVC defect or partial atrioventricular septal defect

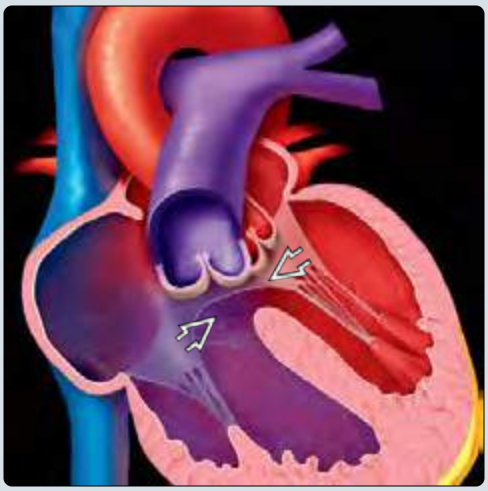
IMAGING

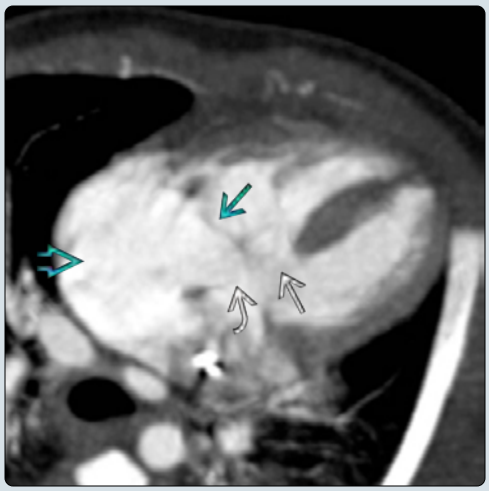
- Large right atrium, right ventricle, pulmonary artery with increased pulmonary artery flow
- Large defect in anterior inferior portion of atrial septum (ostium primum defect)
- Large defect in ventricular septum (posterior type most common)
- Anterior & superior aortic position with elongation + dysplastic common 5-leaflet AV valve narrowing subvalvular LVOT → "gooseneck" deformity on angiography
- When AV valve opens toward 1 ventricle → unbalanced canal defect (right ventricular or left ventricular dominance can occur with single ventricle physiology)
- Pulmonary hypertension patients have abnormal lung compliance: Lungs often hyperinflated with eversion of hemidiaphragms
- Mitral insufficiency may occur both pre- & postoperatively

CLINICAL ISSUES

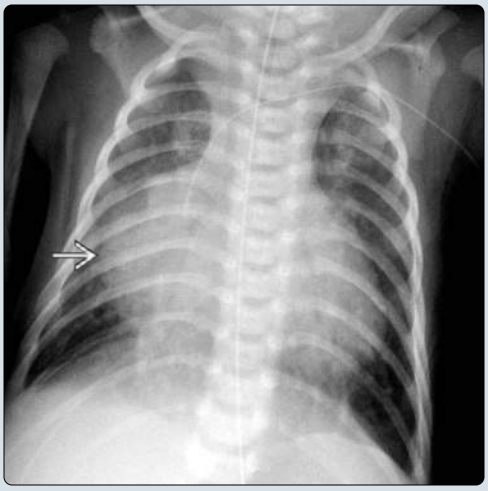
- Associated with trisomy 21 in 44-48%
- Large shunts present early with tachypnea, tachycardia, & failure to thrive
- Small shunts may be well tolerated through 1st decade; children may be asymptomatic
- Single ventricle physiology may necessitate staged procedure such as Glenn & then Fontan for unbalanced AVC
- Partial AVC closed by pericardial patch via right atrial approach

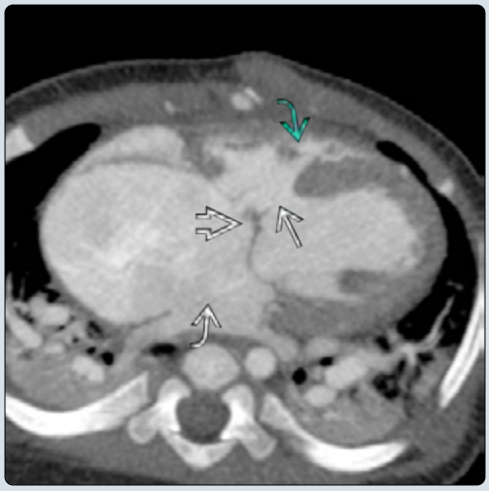
(Left) *Graphic shows a defect in the atrioventricular (AV) septum connecting the right atrium & right ventricle to the left atrium & left ventricle.* **(Right)** *Axial image from a cardiac CTA in a newborn with Down syndrome shows a large ventricular septal defect (VSD), septum primum atrial septal defect (ASD), & a common AV valve, consistent with an atrioventricular septal defect (AVSD). Notice the large right atrium.*





(Left) *Frontal chest radiograph in a 2 month old with Down syndrome shows cardiomegaly, increased pulmonary vascularity, & venous congestion. Notice the massive enlargement of the right atrium.* **(Right)** *Axial image from a cardiac CTA shows an AVSD. There is a common dysplastic AV valve with an inlet-type VSD & a septum primum ASD. Notice the enlargement of the right atrium & the small right ventricle in this unbalanced AV canal defect.*





TERMINOLOGY

Abbreviations

- Atrioventricular septal defect (AVSD)

Synonyms

- AVSD
  - Atrioventricular canal (AVC) defect
  - Endocardial cushion defect
  - Complete AVC defect
- Ostium primum defect
  - Partial AVC defect
  - Partial AVSD
  - Incomplete endocardial cushion defect

Definitions

- Broad spectrum of defects characterized by involvement of atrial septum, ventricular septum, & 1 or both atrioventricular (AV) valves
  - Complete AVSD indicates presence of both atrial & ventricular septal defects with common AV valve
  - Partial AVSD indicates atrial septal involvement with separate mitral & tricuspid valve orifices
  - Unbalanced AVSD indicates 1 ventricular chamber is hypoplastic compared with other, depending on direction of AV valve flow

IMAGING

General Features

- Best diagnostic clue
  - Chest radiograph
    - Large heart with large main pulmonary artery & increased pulmonary artery flow
    - Serial radiographs to evaluate for pulmonary hypertension
    - Mitral insufficiency may occur both pre- & postoperatively
      - When mitral insufficiency severe, left atrium can be large & cause left lower lobe collapse
    - Children with large shunts have increased incidence of upper respiratory infections & pneumonia
    - Pulmonary hypertension patients have abnormal lung compliance: Lungs are often hyperinflated with eversion of hemidiaphragms
- Location
  - Complete AVC
    - Large defect in anterior inferior portion of atrial septum (ostium primum defect)
    - Large defect in ventricular septum (posterior type most common)
    - Common AV valve with variable chordal attachments to ventricle
    - When AV valve opens toward 1 ventricle → unbalanced canal defect
    - Right ventricular or left ventricular dominance can occur with single ventricle physiology (or unbalanced defect)
    - Hypoplasia of inlet & outlet septum → hypoplasia of chamber with malalignment of ventricular septum
  - Ostium primum defect
    - Defect in anterior inferior aspect of atrial septum
    - ± coexistent cleft in anterior leaflet of mitral valve
    - 5-leaflet AV valve present with separate valve orifices to right & left ventricles
- Size
  - Broad spectrum of size of defects in AV septum & respective sizes of ventricles

Echocardiographic Findings

- Echocardiogram
  - Defines lesion in infants & young children
    - Primum defects have echo dropout in lower portion of septum, cleft in mitral valve
    - Anterior & superior displacement of aorta with elongation & narrowing of left ventricular outflow tract (LVOT)
    - 3D echocardiography can better define anatomy of valve
- Color Doppler
  - Demonstration of left-to-right shunt, severity of mitral regurgitation, & tricuspid regurgitation
    - LVOT obstruction can be quantified

CT Findings

- CECT
  - Noninvasive alternative to further depict anatomy, chamber volumes, & function
  - Large right atrium, right ventricle, & pulmonary artery

MR Findings

- Excellent noninvasive alternative for depiction of function & anatomy
- Best imaging modality for calculating complex regurgitation across common AV valve
- Qp:Qs ratio calculated from velocity-encoded cine MR technique

Angiographic Findings

- Conventional
  - Cardiac catheterization not usually done to evaluate anatomy but to measure pulmonary vascular resistance
    - Left ventriculogram shows cleft in mitral valve, shunts, respective sizes of ventricles, & LVOT obstruction
    - Classic "gooseneck" deformity seen on frontal projection of left ventricular angiocardiogram
      - Dysplastic common AV valve narrows subvalvular LVOT

Imaging Recommendations

- Best imaging tool
  - Echocardiography in infants & young children: Defines lesion
    - Primum defects have echo dropout in lower portion of septum, mitral valve cleft
    - Complete AVSD demonstrates varying degrees of absence of septum, size of defect, & relative size of ventricles
- 3D CT & MR reconstructions: Useful for demonstrating complex global, coronary, & extracardiac anatomy for presurgical planning

DIFFERENTIAL DIAGNOSIS

Ventricular Septal Defect

- Most common congenital heart disease (CHD) with left-to-right shunt
- Most common CHD associated with other lesions
- Cardiac enlargement with increased pulmonary flow

Atrial Septal Defect

- Defect in superior portion of atrial septum
- Presents in older children who are usually asymptomatic from shunt
- Left-to-right shunt usually not large but can cause Eisenmenger physiology in adult if unrecognized

Patent Ductus Arteriosus

- Communication between high-pressure aorta & lower pressure pulmonary artery
- Left-to-right shunt usually presents in infancy
- Closed by percutaneous occlusion devices

PATHOLOGY

General Features

- Etiology
  - Malformation occurring during 5th week of gestation
  - Abnormal or inadequate fusion of superior & inferior endocardial cushion
  - Abnormal fusion of ventricular (trabecular) portion of septum
  - Iatrogenic AVSD has been described
- Genetics
  - Associated with trisomy 21 in 44-48%
- Associated abnormalities
  - Trisomy 21 children have constellation of clinical & radiographic findings
    - Chest radiograph may show 11 ribs, double manubrial ossification center in 80%
  - Heterotaxy
  - Tetralogy of Fallot

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Complete AVSD
    - Large shunts present early with tachypnea, tachycardia, & failure to thrive
    - Mitral insufficiency adds complexity & earlier symptoms
  - Partial AVSD
    - Small shunts may be well tolerated through 1st decade, children may be asymptomatic
    - Mitral insufficiency adds complexity & earlier symptoms
- Other signs/symptoms
  - Pathophysiology of lesions
    - Degree of left-to-right shunting determined by size of defect & relative compliance of atria & ventricles
    - Right ventricular compliance reflects pulmonary vascular resistance
    - Infants have high pulmonary vascular resistance & therefore rarely have shunts
    - As pulmonary vascular resistance decreases, left-to-right shunting increases with age
    - Subsequent enlargement of right atrium & right ventricle & increase in pulmonary vascularity
    - Cleft directs regurgitant blood through atrial defect

Demographics

- Epidemiology
  - 4-8:1,000 live births have congenital heart defects
  - 5-8% have AVSD
  - 44-48% of patients with Down syndrome or trisomy 21 have AVC defect

Natural History & Prognosis

- Complete AVC defect presents in infancy with symptoms
- Children assessed for surgical repair
  - Postoperative course may be complicated by mitral insufficiency
  - Pulmonary hypertension occurs in children without surgical intervention

Treatment

- Medical management until surgery (depending on lesion & severity)
- Surgical management: Partial AVSD
  - Closed by pericardial patch via right atrial approach
  - Percutaneous closure devices not typically deployed, as inferior attachment may injure AV valves
- Surgical management: Complete AVSD (mortality rate 3%)
  - Elective repair in children 2-5 years of age unless mitral regurgitation present
  - Complications include mitral insufficiency (which may require reoperation, valvuloplasty, or replacement)
  - Arrhythmias such as sinus node dysfunction or heart block
- Surgical management: Complete unbalanced AVSD
  - Single ventricle physiology may necessitate staged procedure such as Glenn & then Fontan

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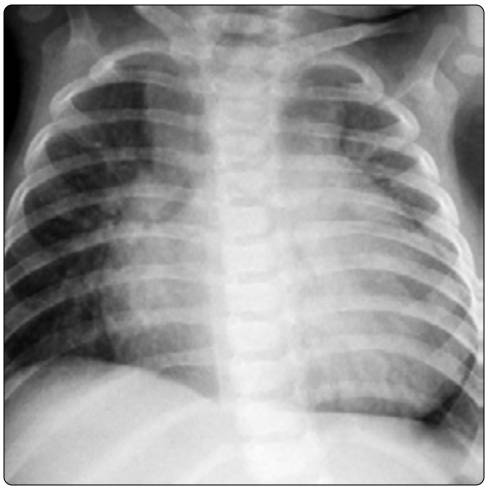
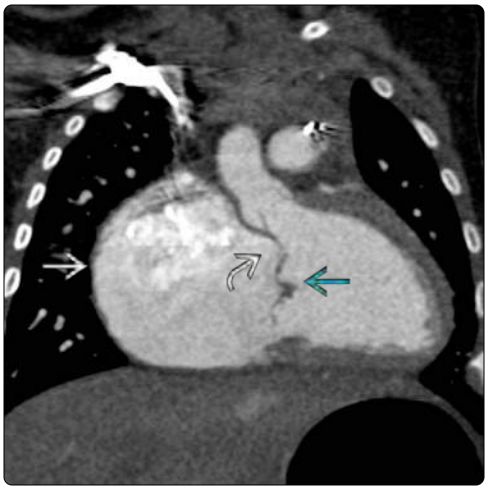
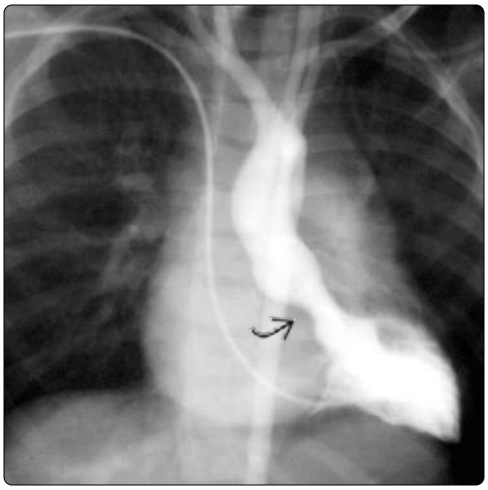
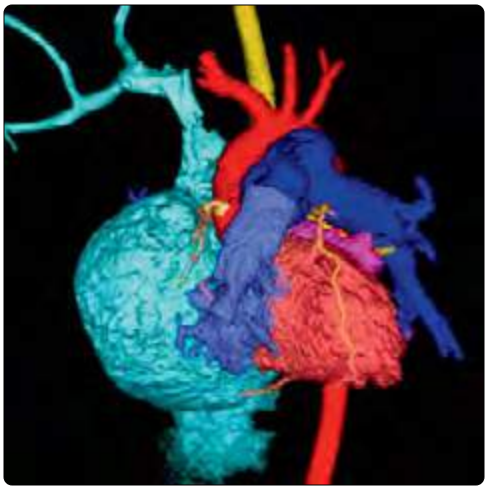


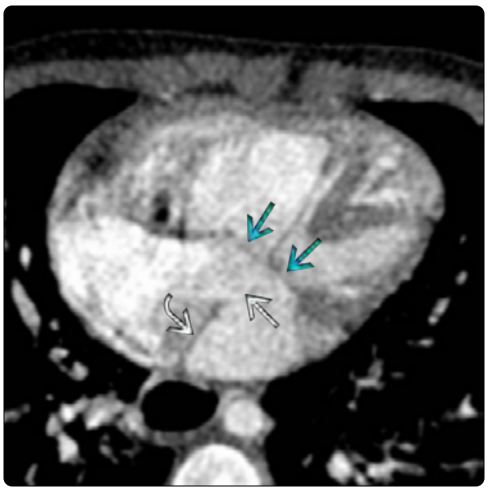


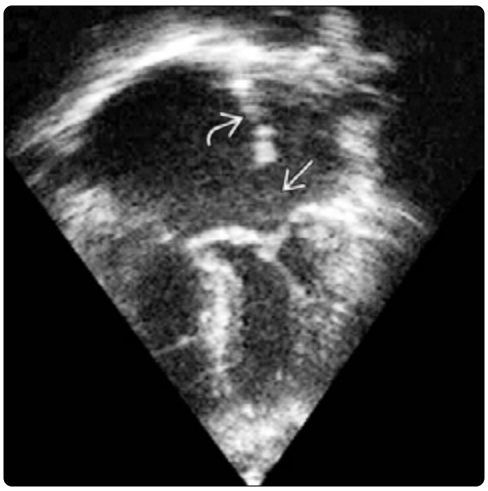
(Left) *AP radiograph shows cardiomegaly in a 1-month-old infant. There is enlargement of all chambers & an increase in pulmonary artery flow secondary to left-to-right shunting.* **(Right)** *Coronal image from a cardiac CTA shows the dysplastic common AV valve ⇨ narrowing the subvalvular left ventricular outflow tract (LVOT) ⇨. This is the cause of the "gooseneck" deformity seen on conventional angiography in patients with AVSD. Notice the marked enlargement of the right atrium ⇨.*





(Left) *Left ventricular angiogram shows a typical "gooseneck" deformity ⇨ of the LVOT in a patient with AVSD. Subvalvular narrowing of the left ventricular outflow tract is due to the enlarged common AV valve.* **(Right)** *Single frontal view from a color-coded cardiac CTA shows massive right atrium enlargement (light blue) in a patient with AVSD. Note the hypoplastic right ventricle (purple) compared with the left ventricle (pink), consistent with an unbalanced AV defect.*





(Left) *Four-chamber view from a cardiac CTA in an infant demonstrates an ostium primum defect ⇨ in the atrial septum ⇨ with a common AV valve ⇨ in a patient with an AVSD.* **(Right)** *Four-chamber view echocardiogram shows echo dropout in the inferior portion of the atrial septum ⇨, which is characteristic of a primum ASD. Notice the intact portion of the atrial septum ⇨.*

Patent Ductus Arteriosus

KEY FACTS

TERMINOLOGY

- Persistent postnatal patency of normal prenatal connection from PA to proximal descending aorta
- Hemodynamics: L → R shunt between aorta & PA
- PDA frequently essential in complex congenital heart disease: L → R or R → L flow, depending on other anomalies
- PDA in persistent fetal circulation syndrome: R → L flow

IMAGING

- Well demonstrated by CTA & MRA: Usually linear, directed anterior to posterior, of variable size
- PDA may be tortuous vessel connecting aorta &/or innominate artery with PA
- CTA modality of choice for showing airway compression from tortuous PDA or vascular ring

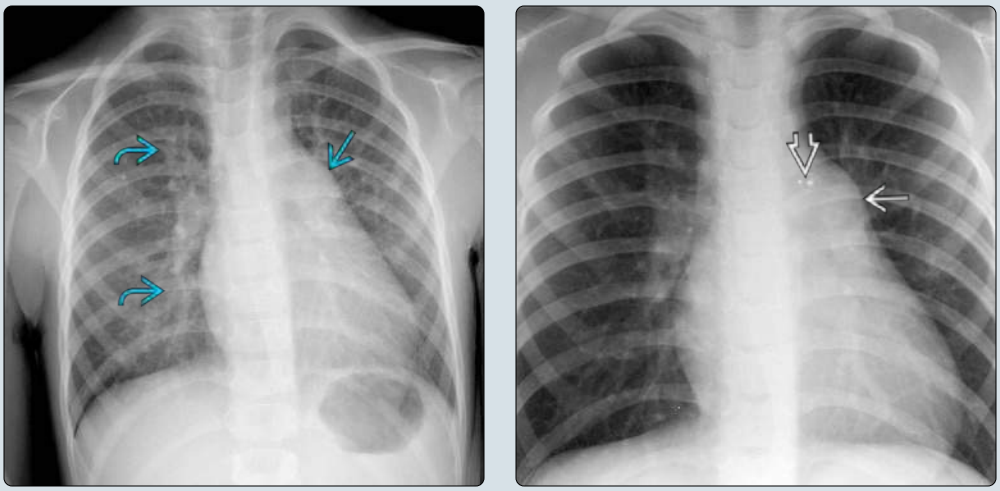
PATHOLOGY

- With normal drop of pulmonary vascular resistance, L → R shunt to PA through PDA
  - Volume overload of left-sided cardiac chambers
  - Diastolic flow reversal in aorta can lead to renal & intestinal hypoperfusion → renal dysfunction & necrotizing enterocolitis
  - Pressure overload of right ventricle eventually causes reversal of shunt (R → L) → cyanosis (Eisenmenger physiology)
- When closed: Forms ligamentum arteriosum, ± Ca^{2+}
- In right arch, aberrant left subclavian artery ductus completes vascular ring

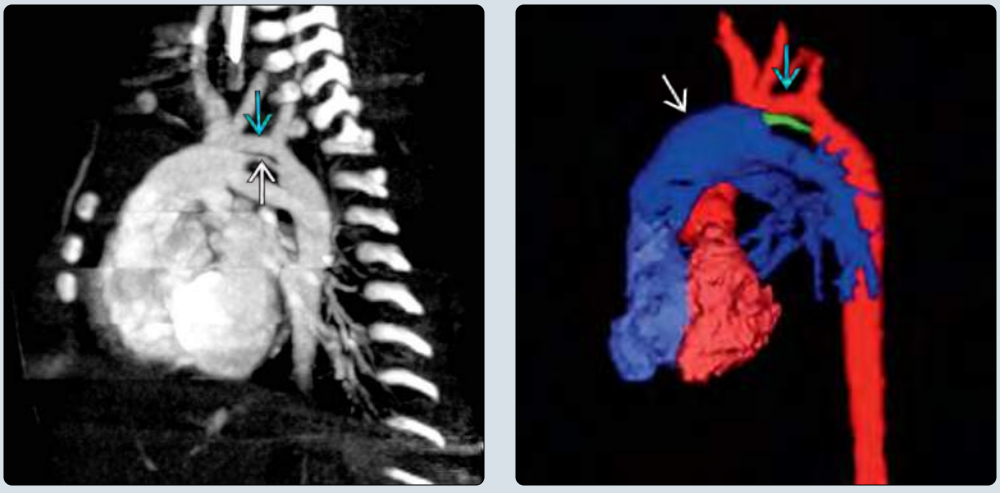
CLINICAL ISSUES

- To close ductus in premature infants: Indomethacin
- To keep ductus open in cyanotic heart disease: Prostaglandin E1
- Term infants, older children: Surgical clipping or ligation versus endovascular closure with duct occluder devices &/or coils

(Left) *Frontal view of the chest in an 8-year-old patient with a persistent patent ductus arteriosus (PDA) shows increased (shunt) vascularity with prominence of the pulmonary artery (PA). Chronic PDA can cause Eisenmenger syndrome.* **(Right)** *Frontal chest radiograph shows prominence of the main PA & mild cardiomegaly in a patient with a recent PDA closure device placement. Notice that the pulmonary vascularity appears normal.*



(Left) *Oblique MIP image from a cardiac CTA shows a small PDA connecting the aorta to the main PA. Notice that the transverse aortic arch is mildly hypoplastic. A PDA is commonly seen in association with a left ventricular outflow tract obstruction.* **(Right)** *Oblique 3D color-coded cardiac CTA in the same patient shows a PDA (green) with a mildly hypoplastic aorta. Compare this with the previous MIP image. Notice the enlarged size of the MPA caused from increased left → right shunting.*



TERMINOLOGY

Abbreviations

- Patent ductus arteriosus (PDA)

Synonyms

- Persistent arterial duct, patent ductus Botalli

Definitions

- Persistent postnatal patency of normal prenatal connection from pulmonary artery (PA) to proximal descending aorta
- Category: Acyanotic, ↑ pulmonary blood flow
- Hemodynamics: L → R shunt between aorta & PA
- PDA is frequently essential part of complex congenital heart disease
  - Typical ductal-dependent lesions, including hypoplastic left heart syndrome, severe hypoplastic-type coarctation, & interruption of aortic arch: Conduit for systemic perfusion (R → L flow)
  - D-transposition of great arteries: Necessary for admixture between systemic & pulmonary circuits (L → R flow)
  - Pulmonary atresia & other severe cyanotic heart diseases with right-sided obstruction: Conduit for pulmonary perfusion (L → R flow)
- PDA is part of persistent fetal circulation syndrome: R → L flow
  - Severe lung disease (meconium aspiration, surfactant deficiency disease, neonatal pneumonia)
  - Primary pulmonary hypertension of neonate

IMAGING

General Features

- Best diagnostic clue
  - Cardiomegaly & heart failure once pulmonary vascular resistance drops in premature infant recovering from surfactant deficiency disease

Radiographic Findings

- Cardiomegaly (left atrium & left ventricle)
- ↑ pulmonary vascularity
- Wide vascular pedicle (large aortic arch with ductus bump)

CT Findings

- CTA
  - Volume renditions of aortic arch depict PDA with associated complex anatomy
  - Excellent modality for sizing of ductus prior to cardiac catheterization for placement of occluder device or prior to stenting in hybrid procedure
  - Modality of choice for showing airway compression from tortuous PDA or vascular ring

MR Findings

- SSFP bright blood cine
  - Functional right ventricular assessment in cases with Eisenmenger pulmonary hypertension
  - Dephasing artifact depicted in direction of PDA flow
- Double inversion recovery ("black blood") sequence
  - Sagittal oblique plane through aortic arch depicts ductus
- 3D gadolinium MRA with volume rendition to depict anatomy
- Velocity-encoded cine sequences used to calculate Qp:Qs ratio
  - Closure usually indicated for Qp:Qs ratio > 1.7

Echocardiographic Findings

- Echocardiogram
  - Suprasternal notch view: Direct visualization of ductus
  - Size & flow across ductus in early infancy have been shown to correlate with prognosis of chronic lung disease in premature infants
- M-mode
  - ↑ left atrium:aorta ratio (> 1.2:1)
- Pulsed Doppler
  - Diastolic flow reversal in descending & abdominal aorta (ductus steal)
  - Flow acceleration across constricting ductus: Transductal velocity ratio
- Color Doppler
  - For flow direction through ductus

Angiographic Findings

- Conventional
  - Cardiac catheterization only needed for associated complex cyanotic heart disease & to determine reversibility of pulmonary hypertension
  - Placement of PDA closure device

Imaging Recommendations

- Protocol advice
  - Treatment decisions based only on echocardiographic findings in majority of cases
  - CTA recommended for complex anatomy with airway compression

DIFFERENTIAL DIAGNOSIS

Other Causes of Left-to-Right Shunting

- Atrial & ventricular septal defects
- Atrioventricular canal

Persistent Fetal Circulation Syndrome

- Pulmonary hypertension (primary or secondary to severe lung disease)
- Patent foramen ovale
- PDA secondary to profound irreversible hypoxia

PATHOLOGY

General Features

- Etiology
  - Prematurity: Persistent postnatal hypoxia → failure of contraction of ductus
  - Term infant: Associated with maternal rubella
- Genetics
  - No specific genetic defect identified in most cases of isolated PDA
- Embryology
  - Ductus originates from primitive 6th aortic arch
- Pathophysiology (for simple PDA)
  - PDA is persistence of normal prenatal structure after birth
  - In normal neonate, ductus arteriosus closes functionally 18-24 hours after birth, anatomically at 1 month of age

- With normal drop of pulmonary vascular resistance, L → R shunt occurs to PA through PDA
- Volume overload of left-sided cardiac chambers
- With pulmonary hypertension, pressure overload of right ventricle causes reversal of shunt (R → L) → cyanosis (Eisenmenger physiology)
- Diastolic flow reversal in aorta can lead to renal & intestinal hypoperfusion → renal dysfunction & necrotizing enterocolitis

Gross Pathologic & Surgical Features

- PDA usually wider on aortic side
  - Length: 2-8 mm; diameter: 4-12 mm
  - Makes acute angle with aorta in simple PDA, blunt angle with associated congenital heart disease
- Contractile tissue mainly on pulmonary side: Spirally arranged muscle bundles in media
  - Prostaglandin E1 present in fetal life maintains relaxation
  - ↑ oxygen pressure causes constriction
- Thickening of intima with mucoid degeneration
- When closed, arterial duct forms ligamentum arteriosum
- Calcified ligamentum arteriosum often incidentally seen on radiograph or CT
- Can be right-sided & can originate from base of brachiocephalic artery or from aberrant right subclavian artery
- In right arch with aberrant left subclavian artery anatomy, ductus completes vascular ring
- Rarely ducti may be bilateral with right ductus typically originating from right brachiocephalic or subclavian & left ductus originating from undersurface of aorta
  - Double ducti anatomy may be reversed with situs anomalies
  - Spontaneous closure of ductus may help differentiate from aortopulmonary collaterals

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Characteristic machinery-like murmur
  - Bounding peripheral pulses
  - Congestive heart failure
  - Special situation: Premature infant recovering from surfactant deficiency disease
    - ↓ in hypoxia → ↓ in pulmonary vascular resistance → ↑ shunt flow through ductus arteriosus → clinical & radiographic signs of congestive heart failure (cardiomegaly, pulmonary edema)
- Other signs/symptoms
  - Subacute bacterial endocarditis
    - Need for treatment of clinically "silent" PDA (incidentally detected with echocardiography) controversial
  - Ductal aneurysm
    - Can result from premature narrowing of ductus on pulmonary side

Demographics

- Epidemiology
  - 10-12% of congenital heart disease
  - 1/2,500-5,000 live births
  - Slightly more common in female patients
  - Associated with prematurity (21-35%)

Natural History & Prognosis

- Isolated PDA: Excellent prognosis with early closure
- When associated with complex heart disease, prognosis determined by underlying disorder
- Irreversible pulmonary hypertension (Eisenmenger physiology) → shunt reversal & development of cyanosis
- Persistent fetal circulation, pulmonary hypertension: Treatment with extracorporeal membrane oxygenation often necessary to disrupt vicious cycle
  - Hypoxia → pulmonary vasoconstriction → ↓ pulmonary flow → more severe hypoxia

Treatment

- To close ductus in premature infants: Indomethacin
  - Side effects: Renal failure, intestinal perforation, intracranial hemorrhage
- To keep ductus open (cyanotic heart disease): Prostaglandin E1
- Term infants, older children: Surgical clipping, ligation, or device closure of PDA
  - Can be performed at bedside under video-assisted thoracoscopic &/or robotic guidance
  - Complications: Inadvertent ligation of aortic isthmus or PA, recurrent laryngeal nerve injury
- Endovascular closure with duct occluder devices &/or coils
  - Small ductus (< 4 mm): Gianturco coils
  - Large ductus (> 4 mm): Ivalon plug, Rashkind & Amplatz duct occluders
  - Complications: Protrusion of occluder device into left PA orifice → ↓ left lung perfusion, peripheral embolization
  - Incomplete closure in 10-20%

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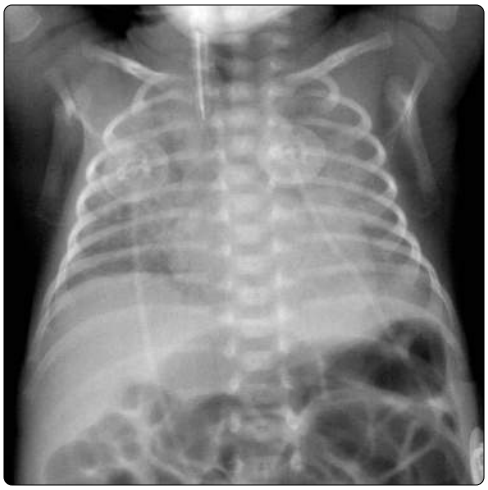
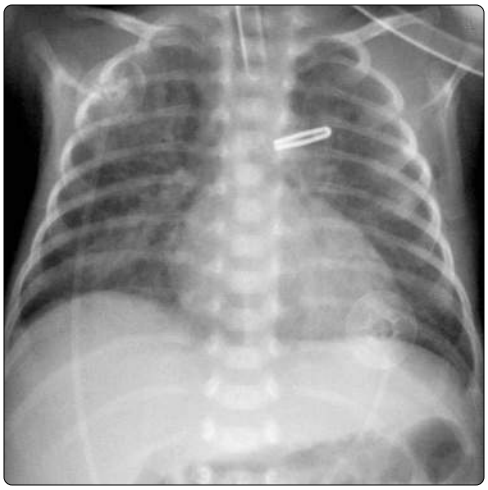
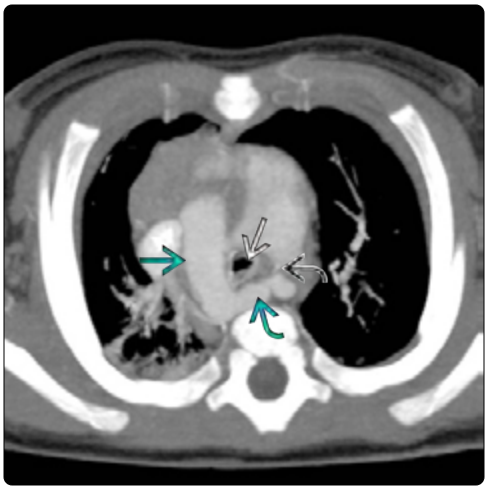
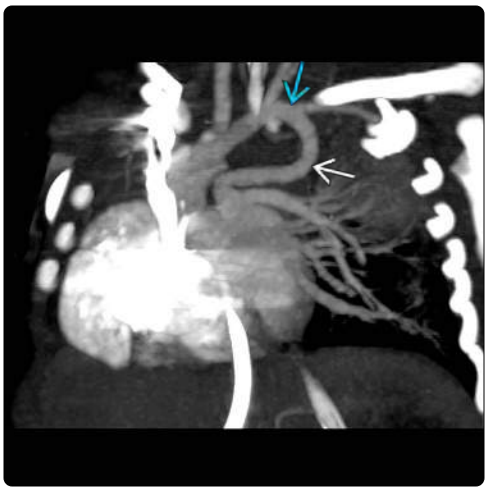


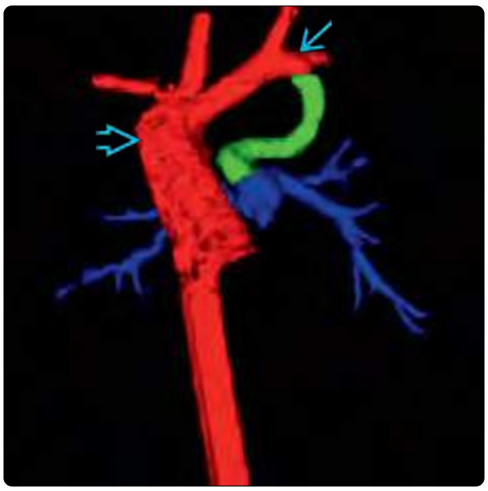


(Left) *AP chest radiograph in a 7-day-old premature infant shows diffuse bilateral hazy opacification of the lungs. An echocardiogram showed a PDA with left → right flow.* **(Right)** *AP view of the chest in the same patient shows a surgical clip from interval ligation of a PDA with increased aeration of the lungs & decreased pulmonary edema.*



(Left) *Oblique 3D color-coded CTA shows a large PDA (green) connecting the aorta (red) to the PA (blue) in a patient with D-transposition of the great arteries. Notice that the right ventricle (purple) is connected to the aorta (red), & the left ventricle (pink) is connected to the PA.* **(Right)** *Axial MIP CTA image shows a right aortic arch ⇨ with an aberrant left subclavian artery ⇨ & small PDA ⇨. Notice that the small PDA completes the vascular ring around the trachea ➡.*





(Left) *Coronal oblique MIP CTA shows a tortuous PDA ➡ arising from the left subclavian artery ⇨ in a patient with a right aortic arch.* **(Right)** *Frontal projection from a 3D color-coded cardiac CTA shows the tortuous PDA (green) arising from the left subclavian artery ⇨ in a patient with a right aortic arch ⇨ & pulmonic atresia. Notice that there is no aberrant left subclavian artery, so this does not represent a vascular ring.*

Tetralogy of Fallot

KEY FACTS

TERMINOLOGY

- Most common cyanotic congenital heart lesion
- Tetralogy: 4 heart defects from embryological anterocephalad deviation of conoventricular septum
  - Infundibular or subpulmonary narrowing
  - Anterior malalignment ventricular septal defect (VSD)
  - Aorta overriding VSD
  - Secondary right ventricular hypertrophy (RVH)
- Spectrum of tetralogy of Fallot disease
  - "Blue Tet": More subpulmonary obstruction → VSD shunts right-to-left → cyanotic appearance
  - "Pink Tet": Less subpulmonary obstruction → VSD shunts left-to-right (normal) → acyanotic appearance
  - Tetralogy with pulmonary atresia & major aortopulmonary collaterals: Severe congenital heart disease
  - Tetralogy with absent pulmonary valve: "To-fro" flow in pulmonary artery (PA) leading to massively dilated branch PAs, tracheobronchial compression

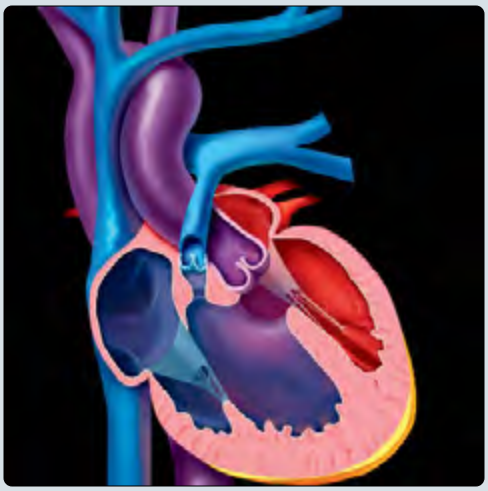
IMAGING

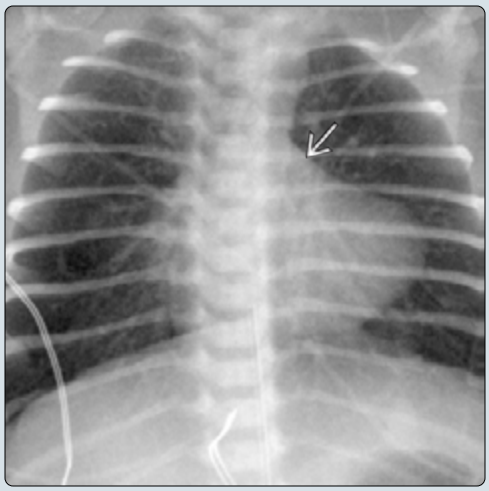
- Radiography: Normal heart size, concave PA segment, ↓ pulmonary vascularity (oligemia)
  - RVH → upturned cardiac apex → boot-shaped heart
  - Right-sided aortic arch in 25%
- Echocardiography: Initial diagnosis, often prenatal
  - Coronary artery anomalies important for surgical planning: Left anterior descending artery arising from right coronary & crossing RVOT (4%)
- Cardiac MR: Becoming gold standard for postoperative assessment; critical biomarker data for timing of pulmonary valve replacement (PVR)

CLINICAL ISSUES

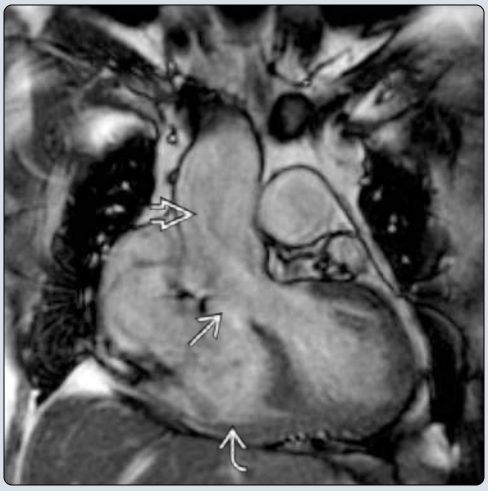
- Typical repair within 1st year to life: VSD closure, relieve RVOT obstruction (transannular patch)
- Significant PR leads to RV chamber dilation over time: ↓ exercise tolerance, RV dysfunction, ventricular arrhythmias
- PVR increasingly utilized to prevent progressive RV dilation

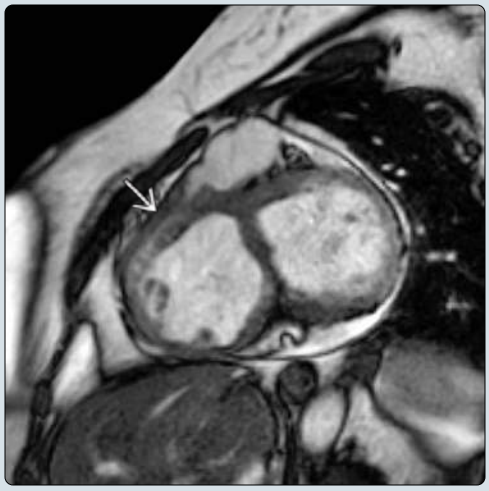
(Left) *Graphic shows subvalvular (infundibular) pulmonary stenosis, a small pulmonic valve, the aorta overriding a high ventricular septal defect (VSD), right ventricular hypertrophy (RVH), & a right-sided aortic arch.* **(Right)** *AP radiograph shows the classic appearance of tetralogy of Fallot (TOF) with a concave pulmonary artery segment ➔ & an upturned cardiac apex creating the "coeur en sabot" (boot-shaped heart) appearance. Note the pulmonary oligemia.*





(Left) *Coronal bright blood cine MR in an adult TOF patient palliated with a Potts shunt illustrates the VSD ➔, overriding aorta ➔, & RVH ➔.* **(Right)** *Two-chamber bright blood cine MR in the same patient shows marked infundibular narrowing ➔ due to muscle bundle hypertrophy. Additionally, the right ventricle (RV) has become more round to handle the elevated systemic pressures due to the unrepaired VSD & outflow tract obstruction.*





TERMINOLOGY

Definitions

- Tetralogy of Fallot (TOF): 4 heart defects in combination
  - Obstruction to pulmonary outflow via infundibular right ventricular outflow tract (RVOT) narrowing or pulmonary valve stenosis
  - Secondary right ventricular hypertrophy from ↑ right ventricle (RV) pressures/fixed subpulmonary obstruction
  - Anterior malalignment ventricular septal defect (VSD)
  - Aorta overriding VSD
- Category: Cyanotic, normal heart size, ↓ pulmonary vascularity
- Hemodynamics: Obstruction to pulmonary blood flow via infundibular narrowing by anterior deviation of subpulmonary conal septum
  - Spectrum: Classic "blue tet," "pink tet," TOF with pulmonary atresia & major aortopulmonary collaterals (MAPCAs), TOF with absent pulmonary valve

IMAGING

General Features

- Best diagnostic clue
  - Anterocephalad deviation of subpulmonary conoventricular septum → 4 characteristic heart defects
  - Aorta overriding large anterior malalignment VSD

Radiographic Findings

- RV hypertrophy + concave pulmonary artery (PA) segment: Boot-shaped heart ("coeur en sabot")
  - RV hypertrophy rotates heart, creating rounded, upturned apex
- ↓ pulmonary vascularity (oligemia) due to obstruction of pulmonary blood flow
- Normal heart size at birth
- Right-sided aortic arch (25%)

CT Findings

- CTA
  - Preoperative: Multidetector-row CT can diagnose coronary anomalies, obviating need for angiography
  - Postoperative: Cardiac-gated cine CT can perform
    - Assessment of RV volumes & function in patients with contraindication for MR
    - CTA also less affected by metal artifact than MR (to assess results of interventions, e.g., stents, coils)

MR Findings

- T1WI
  - Cardiac-gated axial images for preoperative definition of PA anatomy, PA stenosis
  - Postoperative PA anatomy, patency of Blalock-Taussig (BT) shunts assessed by presence of flow void
- T2* GRE
  - Short-axis steady-state free precession cine bright blood MR for RV & left ventricular (LV) volumes, ejection fraction (EF), pulmonary regurgitant fraction, branch PA differential flow
    - Functional MR: Biventricular response to exercise & recovery
    - Presence of RVOT akinesia/aneurysm & wide annulus correlates with need for pulmonary valve replacement (PVR)
- MRA
  - Gadolinium-enhanced MRA: Depiction of PA anatomy & aortopulmonary collaterals
  - Phase contrast assists with estimating right ventricular ejection fraction & pulmonary regurgitation

Echocardiographic Findings

- Location of VSD, additional muscular VSDs
- Degree of aortic override, position of arch
- RVOT obstruction, pulmonary valve morphology
- Branch PA anatomy
- Coronary artery origin & proximal courses

Imaging Recommendations

- Best imaging tool
  - Initial diagnosis with pre- & postnatal echocardiography
- Protocol advice
  - MRA/3D whole-heart sequence or CTA for detailed PA & coronary anatomy
  - Cardiac catheterization for coronary anatomy & percutaneous interventions (RVOT or ductal stent as initial palliative procedure)
  - MR in older child/young adult with poor acoustic echocardiography windows; becoming gold standard for functional assessment of postoperative regurgitation & ventricular dysfunction prior to PVR

DIFFERENTIAL DIAGNOSIS

Double Outlet Right Ventricle With Normally Related Great Vessels, Subaortic Ventricular Septal Defect, & Pulmonary Stenosis

- Defined as both outlets (aorta & PA) arising from RV
- Can be difficult to differentiate from TOF; therefore, secondary characteristics often employed
  - > 50% aortic override of VSD
  - Bilateral subarterial conal septum leading to mitral-aortic fibrous discontinuity

Pulmonary Atresia With Intact Ventricular Septum

- Massive cardiomegaly with right atrial enlargement at birth

Pulmonary Valve Stenosis & Perimembranous Ventricular Septal Defect

- Significant pulmonary valve stenosis with VSD should be evaluated for conal septal deviation

Tricuspid Atresia With Normally Related Great Vessels, Subpulmonary Obstruction

- Large right atrium with obligate right-to-left atrial level shunt; subpulmonary obstruction through restricted VSD

PATHOLOGY

General Features

- Genetics
  - Chromosomal anomalies in 11% (chromosome 22)
  - Other congenital anomalies in 16%; 8% syndromic
- Associated abnormalities
  - PA branch stenosis or hypoplasia

- Absence of pulmonary valve: Severe pulmonary regurgitation → aneurysmal dilation of PAs → tracheobronchomalacia & compression
- Pulmonary atresia & MAPCAs
  - Extreme end of TOF spectrum; severe congenital heart disease with often poor prognosis
- Patent foramen ovale vs. secundum atrial septal defect
- Right aortic arch, mirror image branching (25%)
- Coronary anomalies: Left anterior descending arising from right coronary & crossing RVOT (4%) with implications for surgical repair

- Pathophysiology: Balance between RVOT obstruction & VSD determines shunt direction
  - Classic TOF: Right-to-left shunting, ↓ pulmonary flow, cyanosis
  - "Pink" TOF: Left-to-right shunting, normal to ↑ pulmonary flow, congestive heart failure

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Varying degrees of cyanosis at birth (most often apparent by 3 months)
  - Older child: Cyanotic spells, relieved by squatting
  - Congestive heart failure (large VSD)
- Other signs/symptoms
  - After repair: Decreased exercise tolerance, RV dysfunction
  - Severe arrhythmias, which may be fatal
    - RV chamber dilation from postoperative pulmonary regurgitation
    - Damage to conduction system during VSD closure
    - Scar from right ventriculotomy → ectopy focus
  - Bacterial endocarditis
  - Stroke due to paradoxical embolus to brain
  - Hyperviscosity syndrome due to polycythemia

Demographics

- Epidemiology
  - Incidence: 3-5 per 10,000 live births
  - 4th most common congenital heart anomaly
  - Most common cyanotic heart lesion

Natural History & Prognosis

- 10% of untreated patients live > 20 years
- Short term: Excellent results after early "complete" repair
- Long term: Determined by RV diastolic & systolic dysfunction; chronic regurgitation leading to dilation, arrhythmias, & risk of sudden death
  - Timing of PVR determined by results of functional MR

Treatment

- Palliative shunt (needed primarily with hypercyanotic episodes or extreme desaturation)
  - Classic BT shunt: End-to-side subclavian artery to PA (opposite from aortic arch)
  - Modified BT shunt: Interposition of Gore-Tex graft
  - Central shunt: Ductus-like connection between aorta & PA
- Complete repair: Removal of RVOT obstruction, VSD closure
  - Limited transannular patch with RVOT enlargement: Postoperative pulmonary regurgitation
    - RV dysfunction, arrhythmias
  - Rastelli shunt: Valved conduit between RV & PAs in case of severe RVOT stenosis or pulmonary valve atresia
- Pulmonary valve or conduit replacement after early complete repair
  - Conduit stenosis: RV pressure overload, systolic dysfunction
  - Pulmonary regurgitation: RV volume overload, diastolic & systolic dysfunction, reciprocal left ventricle systolic dysfunction, arrhythmias
  - Timely surgery allows RV remodeling with ↓ RV size, ↑ EF, ↑ exercise capacity
- Percutaneous balloon dilation of residual pulmonary valve stenosis &/or peripheral PA stenosis (with stent placement)
- Transcatheter PVR
  - Less invasive, new option for PVR
  - Risk:benefit ratio of earlier reintervention is being evaluated compared with surgical replacement
  - Risk of bacterial endocarditis higher compared to surgical PVR

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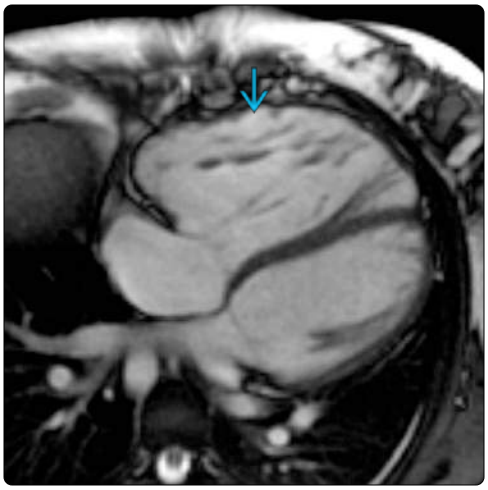
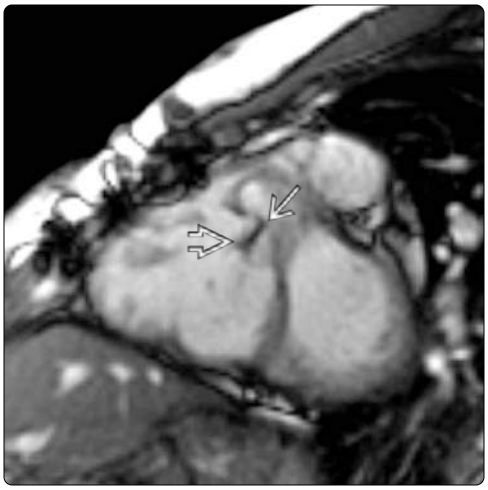
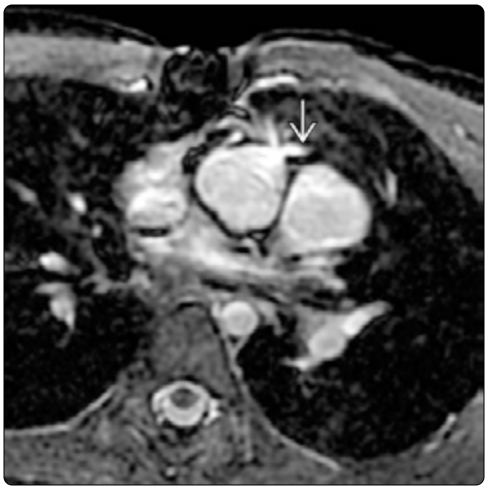
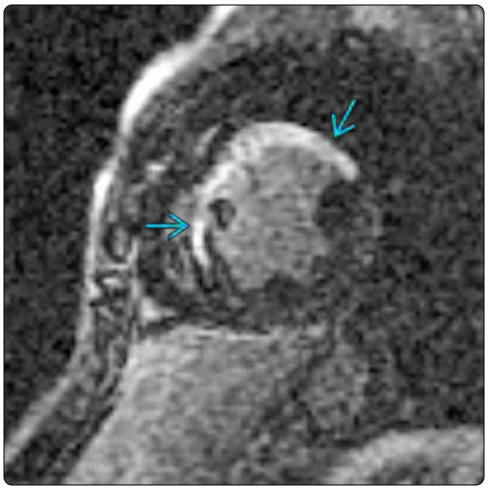


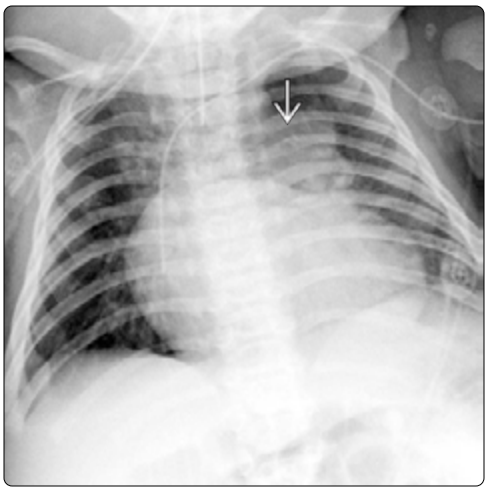


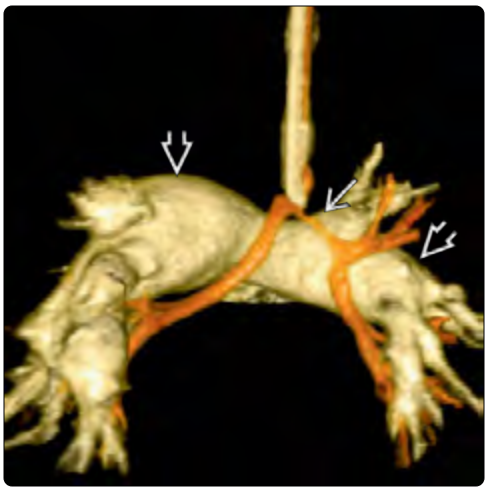
(Left) *Four-chamber bright blood cine MR shows a typical appearance of the RV after "complete" repair. The RV often dilates from volume overload secondary to a large regurgitant fraction across the pulmonic valve.* **(Right)** *Two-chamber bright blood cine MR in a repaired TOF patient shows a patch leak located where the VSD patch is sewn to the aortic annulus. The leak is indicated by the dark, dephased blood shooting into the RV. This patient was revised with a conduit & closure of the patch leak.*





(Left) *Axial bright blood cine MR in a 6 year old (who had recurrent RV outflow tract obstruction requiring repeat subpulmonary muscle bundle resection at age 4) shows the left anterior descending (LAD) coronary artery arising from right coronary artery & coursing over the RV outflow tract.* **(Right)** *Two-chamber late gadolinium enhancement (LGE) MR in a TOF patient shows myocardial fibrosis due to prior infarction in the distribution of the LAD, which was presumably injured during the subpulmonary muscle bundle resection.*





(Left) *AP radiograph in a 1 month old with TOF & pulmonic valve absence shows enlargement of the pulmonary arteries, most pronounced on the left. The patient was difficult to manage because of severe airway narrowing.* **(Right)** *Coronal posterior 3D reformation of a CTA in the same patient shows a markedly narrowed right bronchus & dilated branch pulmonary arteries. Severe tracheobronchomalacia is often seen in TOF with pulmonary valve absence.*

Pulmonary Atresia

KEY FACTS

TERMINOLOGY

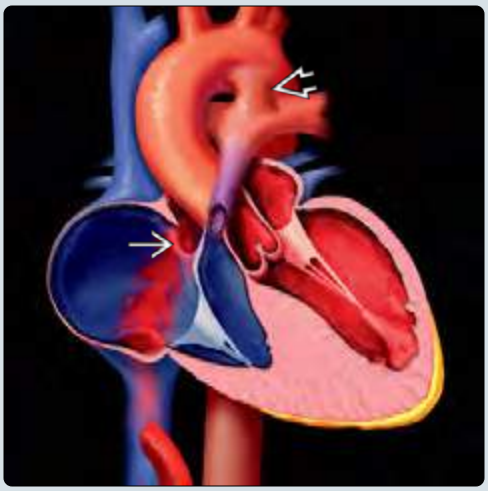
- 2 distinct entities, differentiated by presence or absence of ventricular septal defect (VSD)
  - Pulmonary atresia (PAt), intact VS: Normal-sized pulmonary arteries (PAs) supplied by patent ductus arteriosus (PDA), patent foramen ovale (PFO)
  - PAt, VSD, multiple aortopulmonary collateral arteries (MAPCAs): Hypoplastic/absent PAs; MAPCAs supply 1 or both lungs
    - At extreme end of spectrum of RVOT-obstructive (Fallot-type) heart lesions, with complex & highly variable PA anatomy

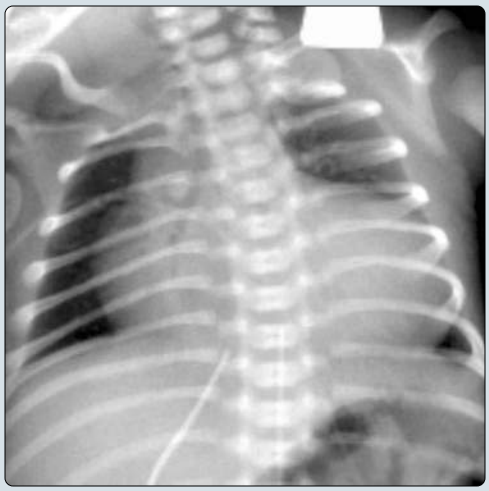
IMAGING

- Extreme boot-shaped appearance of heart
- Right-sided aortic arch common
- PAt, intact VS: Severe cardiomegaly from massive right atrial dilation
- Initial diagnosis with echocardiography
- Cardiac CTA delineates PAs, MAPCAs, & coronary artery fistulas & sinusoids
  - Provides roadmap for subsequent catheterization
- CT or MR postoperatively for shunt/conduit patency
- Cardiac catheterization for hemodynamic assessment, selective injection studies, & catheter-based interventions

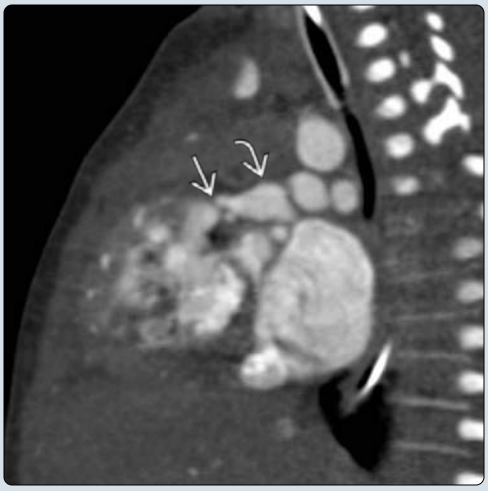
CLINICAL ISSUES

- Progressive cyanosis after birth at closure of PDA
  - Prostaglandin E1 to maintain PDA
- Congestive heart failure with large unobstructed high-flow MAPCAs
- Treatment
  - PAt, VSD, MAPCAs: Unifocalization of MAPCAs & true PAs (if existent, to allow for PA growth)
  - PAt, intact VS: Type of repair dependent on RV size & RV-dependency on coronary circulation





(Left) *Graphic shows pulmonary atresia with an intact ventricular septum. Note the patent foramen ovale, dilation of the right atrium, & right ventricular hypertrophy. The pulmonary arteries are perfused by flow from the aorta through a patent ductus arteriosus (PDA).* **(Right)** *Frontal view of the chest in a neonate with pulmonic atresia shows cardiomegaly with decreased pulmonary vascularity. The differential diagnosis for this appearance includes pulmonic atresia, Ebstein anomaly, & tricuspid atresia.*



(Left) *Sagittal MIP from a cardiac CTA shows an atretic thickened pulmonary valve with normal caliber. A PDA feeds the main pulmonary artery. This patient is considered ductal dependent as survival is not possible without the PDA.* **(Right)** *Left posterior oblique view of a color-coded 3D CTA shows complete pulmonic atresia with a gap seen between the right ventricle (RV) (purple) & the pulmonary artery (blue). Notice the large PDA (green) that provides flow to the lungs.*

TERMINOLOGY

Abbreviations

- Pulmonary atresia (PAt) with ventricular septal defect (VSD) & multiple aortopulmonary collateral arteries (MAPCAs)
- PAt with intact ventricular septum (PAt, intact VS)

Synonyms

- Sometimes referred to as truncus arteriosus type 4 or pseudotruncus (misnomer)

Definitions

- 2 distinct entities, differentiated by presence or absence of VSD
  - PAt, VSD, MAPCAs: Hypoplastic/absent pulmonary arteries (PAs); MAPCAs supply 1 or both lungs
    - Type A: Normal-sized PAs with small aortopulmonary (AP) collaterals
    - Type B: Mild to moderate PA hypoplasia with multiple AP collaterals
    - Type C: Markedly hypoplastic PAs with pulmonary flow from numerous AP collaterals
  - PAt, intact VS: Normal-sized PAs supplied by patent ductus arteriosus (PDA), patent foramen ovale (PFO)
    - Coronary sinusoids or fistulas are often present
- Both are characterized by underdevelopment of right ventricular outflow tract (RVOT) & pulmonary valve
  - PAt, VSD, MAPCAs: At extreme end of spectrum of RVOT-obstructive (Fallot-type) heart lesions, with complex & highly variable PA anatomy
- Category: Cyanotic, cardiomegaly, decreased &/or irregular pulmonary vasculature
- Hemodynamics: Extreme outflow obstruction of right ventricle (RV); (almost) entire cardiac output goes into dilated overriding ascending aorta

IMAGING

General Features

- Best diagnostic clue
  - Atresia of RVOT &/or pulmonary valve

Radiographic Findings

- Extreme boot-shaped appearance of heart
- Right-sided aortic arch common
- Diminutive hilar shadows
- Irregular branching patterns of MAPCAs
- PAt, intact VS: Severe cardiomegaly from massive right atrial dilatation

CT Findings

- CTA
  - Cardiac CTA provides detailed evaluation of PAs & MAPCAs
    - 3D images have proven helpful in planning for unifocalization
  - CTA best used to provide anatomic roadmap for subsequent catheterization
    - Saves overall radiation, contrast, procedure time
    - Cardiac CTA provides delineation of coronary artery fistulas & sinusoids
  - CTA is excellent modality for unstable postoperative patients

MR Findings

- T1WI
  - PAt, VSD, MAPCAs: Cardiac-gated axial images for preoperative definition of PA anatomy
- T2* GRE
  - Short- & long-axis SSFP cine MR for functional assessment, tricuspid regurgitation
- MRA
  - Coronal gadolinium-enhanced MRA for detailed analysis of PA anatomy & MAPCAs
- Phase-contrast imaging
  - Helpful in following RV volumes & regurgitant fractions in postsurgical patients

Echocardiographic Findings

- Echocardiogram
  - PAt, VSD, MAPCAs
    - Characterizes intracardiac anatomy, position, & size of VSD, aortic root override
    - Development of branch PAs & their confluence
  - PAt, intact VS
    - Morphology of interatrial septum: Identifies restriction to flow across PFO
    - Size of RV & tricuspid annulus (expressed as "z-score"), degree of tricuspid regurgitation: Important for planning of surgical repair

Angiographic Findings

- Conventional
  - PAt, VSD, MAPCAs
    - Selective injection with pressure recordings of all MAPCAs + imaging of true PAs
    - Pulmonary venous wedge injections for retrograde filling of diminutive PAs
  - PAt, intact VS
    - Suprasystemic pressure recordings in RV
    - Detailed imaging of coronary anatomy through RV & aortic root injections: RV to coronary communications, stenoses, interruptions

Imaging Recommendations

- Protocol advice
  - PAt, VSD, MAPCAs
    - Initial diagnosis with echocardiography
    - CT or MR for preoperative assessment of PA anatomy, postoperative assessment for shunt/conduit patency
    - Cardiac catheterization for hemodynamic assessment, selective injection studies, & catheter-based interventions

DIFFERENTIAL DIAGNOSIS

Tetralogy of Fallot

- At least partial patency of RVOT

Complex Cyanotic Heart Lesions With Component of (Sub)Pulmonary Stenosis

- Double outlet RV
- Transposition of great arteries with VSD
- Single ventricle
- Tricuspid atresia

Ebstein Anomaly

- May mimic PAt, intact VS with large tricuspid annulus & massive tricuspid regurgitation

PATHOLOGY

General Features

- Embryology (PAt, VSD, MAPCAs)
  - RVOT obstruction → hypoplasia of PAs
  - Persistence or hypertrophy of primitive arterial connections to lungs
  - Hypertrophy of bronchial arteries
- Pathophysiology of PAt, VSD, MAPCAs: Balance between flow though PAs & MAPCAs determines pulmonary perfusion
  - PA flow at subsystemic pressures, restricted by narrow caliber & eventual closure of ductus arteriosus
  - MAPCA flow leads to increased lung perfusion at systemic pressures (unless restricted by stenosis)
  - Degree of cyanosis determined by intracardiac admixture & amount of pulmonary flow
  - Large amount of pulmonary blood flow through unrestricted MAPCAs → congestive heart failure
- Pathophysiology of PAt, intact VS: Obligatory right → left shunt through PFO
  - PAs supplied by PDA
  - Small heavily trabeculated RV with suprasystemic pressures
  - Depending on size of tricuspid valve annulus: Severe tricuspid regurgitation, leading to massive right atrial dilation (comparable to Ebstein anomaly)
  - Transmyocardial sinusoids connecting right ventricular cavity with coronary artery system cause coronary flow reversal during diastole, leading to myocardial ischemia & infarction

Staging, Grading, & Classification

- PAt with VSD & MAPCAs
  - Type A: Majority of pulmonary flow from PAs with small MAPCAs
  - Type B: Equal pulmonary flow from PAs & MAPCAs
  - Type C: Majority of pulmonary flow from MAPCAs with markedly hypoplastic native PAs

Gross Pathologic & Surgical Features

- Hilar arteries = true PAs
- Presence & confluence of central portions of true PAs important for surgical repair
- MAPCAs originating from
  - Ascending aorta
  - Brachiocephalic or intercostal arteries
  - Ductus arteriosus
  - Descending aorta (most common)

Microscopic Features

- Pulmonary vascular disease develops in vascular bed of high-flow MAPCAs → increase in cyanosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Progressive cyanosis after birth with closure of ductus arteriosus
  - Congestive heart failure with large, unobstructed, high-flow MAPCAs

Natural History & Prognosis

- Progressive cyanosis due to development of pulmonary vascular disease → irreversible pulmonary hypertension
- Life expectancy when untreated < 10 years
- Survival into adulthood now possible: "Adult congenital heart disease"
  - Need for lifelong follow-up with multiple imaging tests
- Prognosis is guarded, depends on feasibility of surgery

Treatment

- Prostaglandin E1 to keep ductus arteriosus open
- Palliative: Systemic-to-PA shunt (Blalock-Taussig, central), initial banding of high-flow MAPCAs
- PAt, VSD, MAPCAs: Staged complete repair
  - Unifocalization of MAPCAs & true PAs (if existent, to allow for PA growth)
  - Early 1-stage repair in infancy, with incorporation of all MAPCAs in PA conduit, may be feasible
  - Complete repair with incorporation of MAPCAs & PAs in conduit, connected to reconstructed RVOT, & closure of VSD (may not be possible due to high pressure in pulmonary system from residual stenosis/hypoplasia & pulmonary vascular disease)
  - Catheter-based interventions (balloon angioplasty with stenting of stenoses, coil embolization of small superfluous &/or bleeding MAPCAs)
- PAt, intact VS: Type of repair dependent on RV size & RV-dependency on coronary circulation
  - Restriction in flow across PFO: Balloon atrial septostomy
  - Catheter-based or surgical pulmonary valvotomy
  - Sudden decompression of RV through valvotomy, RVOT repair, or transannular patch may lead to myocardial ischemia/infarction
  - When RV is too hypoplastic for biventricular repair: Cavopulmonary (Glenn) shunt, staged completion of univentricular repair (Fontan)

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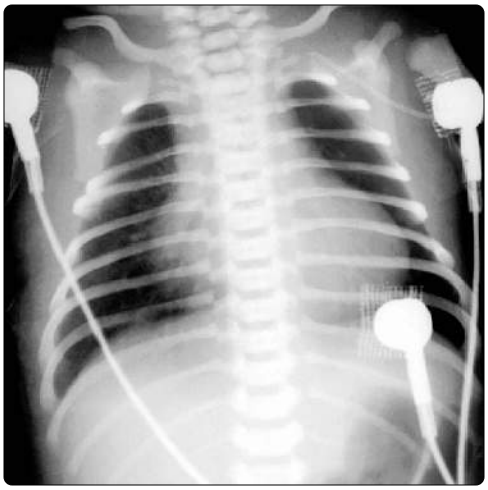
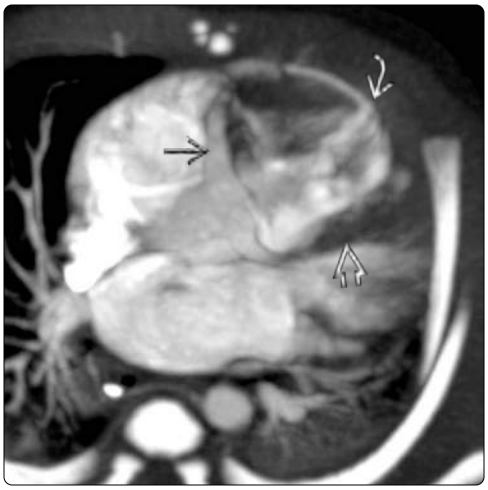
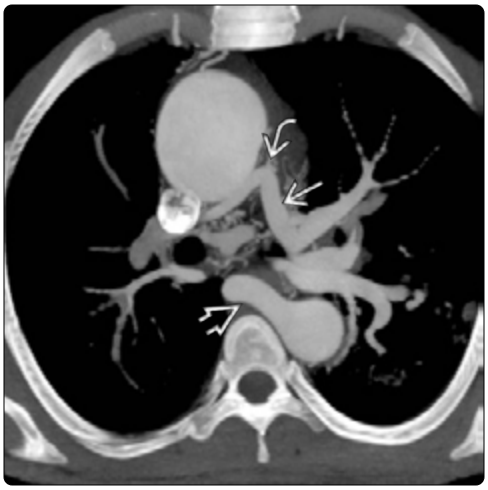
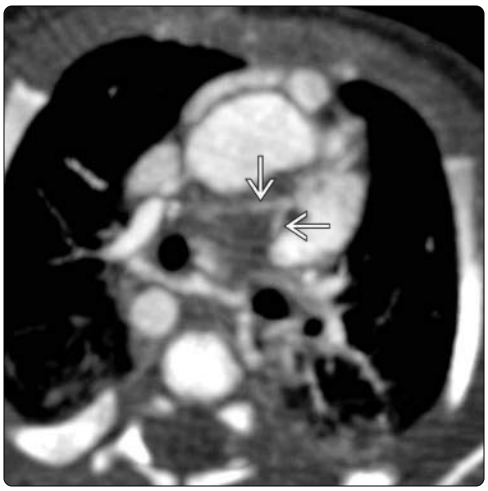


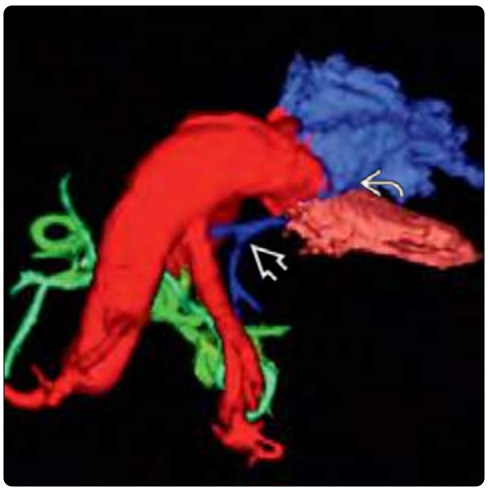


(Left) *AP radiograph in a patient with pulmonary atresia & intact ventricular septum after a right Blalock-Taussig shunt (note rib splaying) reveals right cardiomegaly & pulmonary oligemia.* **(Right)** *Four-chamber cardiac CTA MIP in a patient with pulmonic atresia & intact ventricular septum shows a prominent right coronary artery with a coronary sinusoid communicating with the RV . In patients with an intact ventricular septum, a PDA or PFO is required for blood mixing & oxygenation.*



(Left) *Axial MIP from a cardiac CTA shows pulmonic atresia with moderately hypoplastic pulmonary arteries . Notice the large aortopulmonary collateral arising from the descending aorta. This is a type B pulmonic atresia because the pulmonary arteries are only moderately hypoplastic.* **(Right)** *Lateral view of a color-coded 3D CTA shows moderately hypoplastic pulmonary arteries (blue) with multiple aortopulmonary collaterals (green) in a patient with pulmonic atresia.*





(Left) *Axial MIP from a cardiac CTA shows markedly hypoplastic pulmonary arteries in a patient with pulmonic atresia.* **(Right)** *Superior view of a color-coded 3D CTA shows the markedly hypoplastic native pulmonary arteries (blue). A ventricular septal defect is present . Multiple aortopulmonary collaterals (green) are providing the majority of the pulmonary flow (type C pulmonic atresia).*

Ebstein Anomaly

KEY FACTS

TERMINOLOGY

- Downward/apical displacement of septal & posterior leaflets of tricuspid valve with tricuspid regurgitation
- Category: Cyanotic congenital heart disease with cardiomegaly & ↓ pulmonary vascularity

IMAGING

- Classic radiographic appearance: Massive right-sided cardiomegaly (box-shaped heart)
- All cross-sectional modalities can show right atrial enlargement, apical displacement of tricuspid septal leaflet, & "atrialized" portion of right ventricle
  - Apical displacement of septal tricuspid leaflet (> 15 mm in children < 14 years; > 20 mm in adults)
- MR excellent to evaluate ventricular volumes, ejection fraction, tricuspid regurgitant fraction

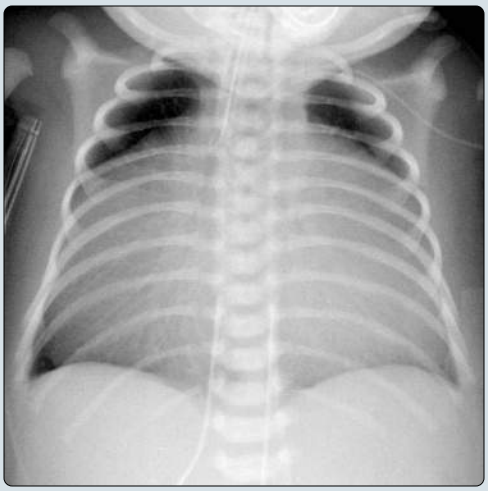
PATHOLOGY

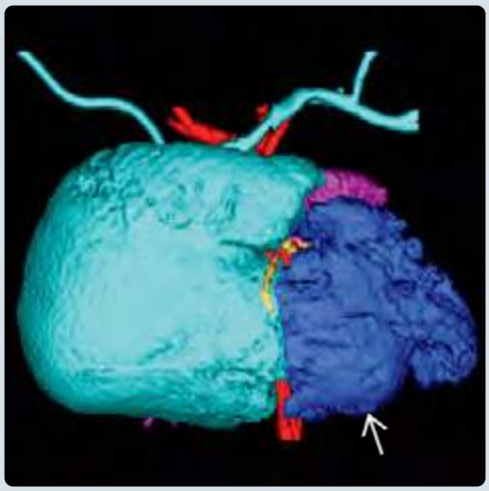
- Patent foramen ovale (PFO), secundum atrial septal defect (ASD) in 90%

CLINICAL ISSUES

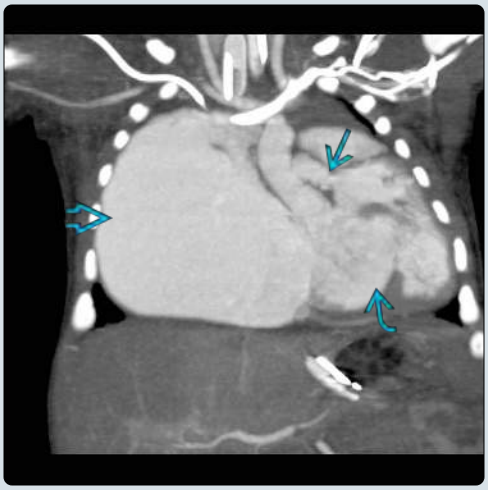
- Wide spectrum of findings & ages at 1st presentation
  - Some patients asymptomatic
- Presence of cyanosis depends on balance between right & left atrial pressures
- Prognosis dependent on hemodynamic significance of tricuspid regurgitation & presence of cyanosis, arrhythmias
- Supportive treatment in cyanotic neonate
  - Oxygen, nitric oxide ventilation to lower pulmonary vascular resistance
- Tricuspid valve replacement &/or reconstruction (valvuloplasty)
  - Definitive repair procedure

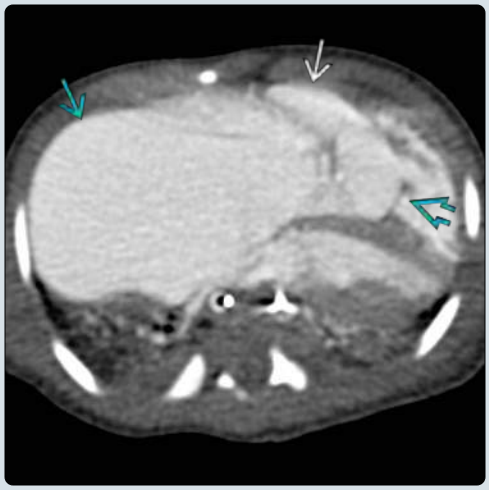
(Left) *Single frontal view of the chest shows massive cardiomegaly (the box-shaped heart) with decreased pulmonary vascularity, typical of patients with Ebstein anomaly.* **(Right)** *Frontal view from a 3D color-coded cardiac CTA shows massive dilation of the right atrium (light blue). Note the smooth portion of the right ventricular wall that has become "atrialized."*





(Left) *Coronal MIP image from a cardiac CTA shows massive dilation of the right atrium with enlargement of the right ventricle in a patient with Ebstein anomaly. Note the narrowed pulmonary outflow tract.* **(Right)** *Axial cardiac CTA shows a massively dilated right atrium with the septal leaflet of the tricuspid valve deviated toward the apex of the heart. Notice the atrialized (smooth) right ventricular wall.*





TERMINOLOGY

Definitions

- Downward/apical displacement of septal & posterior leaflets of tricuspid valve → tricuspid regurgitation
- Classic radiographic appearance: Massive right-sided cardiomegaly (box-shaped heart)
- Category: Cyanotic, (severe) cardiomegaly, normal or ↓ pulmonary vascularity
- Hemodynamics: Determined by severity of tricuspid valve regurgitation
  - Volume overload to right heart
  - Right-to-left shunting through patent foramen ovale (PFO) → cyanosis

IMAGING

General Features

- Best diagnostic clue
  - Downward/apical displacement of septal tricuspid leaflet (≥ 8 mm/m² body surface area)
- Location
  - Tricuspid valve

Radiographic Findings

- Severe right-sided cardiomegaly: May mimic large pericardial effusion
  - Heart size ranges from near normal in newborn period to massively enlarged
  - Heart ↑ gradually in size over time, reaching massive proportions in untreated cases during adulthood
  - Cardiothoracic ratio used as parameter for follow-up
- Small vascular pedicle

CT Findings

- Cardiac CT can be used to obtain volumes & functional analysis of ventricular contraction
- Cardiac CT useful in defining associated complex extracardiac anomalies

MR Findings

- MR cine
  - Cardiac-gated steady-state free precession bright blood MR
    - Apical displacement of septal tricuspid valve leaflet
    - Smooth "atrialized" component of right ventricle
    - Dephasing signal void from tricuspid regurgitation
    - Can calculate ventricular volumes, ejection fraction, tricuspid regurgitation fraction
      - Left ventricular function affected by right ventricular dilation, bowing of septum, mitral valve prolapse
    - Indexed volumes & function of right heart correlate with overall prognosis

Echocardiographic Findings

- Echocardiogram
  - Right chamber enlargement with "atrialized" portion of right ventricle
  - Enlarged tricuspid annulus (expressed in z score)
  - Apical displacement of septal tricuspid leaflet (> 15 mm in children < 14 years; > 20 mm in adults)
- Color Doppler
  - Tricuspid regurgitation
  - PFO with right-to-left shunting

Angiographic Findings

- Characteristic notch at inferior right ventricular border at insertion of displaced anterior tricuspid leaflet
- Seldom required for primary diagnosis

Imaging Recommendations

- Protocol advice
  - Anatomic & functional assessment with echocardiography in infants
  - Cardiac MR in (young) adults

DIFFERENTIAL DIAGNOSIS

Large Atrial Septal Defect

- Acyanotic
- ↑ pulmonary vascularity
- Left-to-right flow through large atrial septal defect (ASD)

Pericardial Effusion

- Acyanotic
- Easy differentiation with echocardiography

Tricuspid Insufficiency

- Primary, due to dysplastic valve
- Often secondary to pulmonary atresia with intact ventricular septum

Uhl Anomaly

- Congenital absence of right ventricular myocardium

Arrhythmogenic Right Ventricular Dysplasia

- Fatty infiltration of right ventricle; fat often not visible by imaging

Right-Sided Obstructive Cyanotic Heart Lesions With Decreased Pulmonary Vascularity

- Tetralogy of Fallot
- Pulmonary atresia
  - With ventricular septal defect & aortopulmonary collaterals
  - With intact ventricular septum
    - Causes severe cardiomegaly
- Tricuspid atresia
- Transposition of great arteries (TGA) with pulmonary stenosis
- Double outlet right ventricle with pulmonary stenosis

PATHOLOGY

General Features

- Genetics
  - Most often sporadic
- Associated abnormalities
  - PFO, secundum ASD in 90%
  - Ebstein anomaly frequently involves left-sided tricuspid valve in congenitally corrected (L) TGA
- Embryology
  - Insufficient separation of tricuspid valve leaflets & chordae tendineae from right ventricular endocardium

- Pathophysiology
  - 3 compartments: Right atrium, atrialized or noncontracting inlet portion of right ventricle, & functional outlet portion of right ventricle
  - Massive tricuspid regurgitation
  - Volume overload to right side of heart
  - Right-to-left shunt through PFO → cyanosis
  - Left ventricular diastolic dysfunction may result from massive right-sided cardiac enlargement
  - Arrhythmias due to conduction abnormalities

Gross Pathologic & Surgical Features

- Thickened valve leaflets, adherent to underlying myocardium
- Downward/apical displacement of septal & posterior tricuspid leaflets
- Normally placed, redundant sail-like anterior tricuspid leaflet
- May occur on left side of heart with congenitally corrected (L) transposition

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Wide spectrum of findings & ages at 1st presentation; some patients asymptomatic
  - Chronic right heart failure
    - ↓ exercise tolerance [classified as New York Heart Association (NYHA) classes I-IV]
  - Presence of cyanosis depends on balance between right & left atrial pressures
    - Physiological ↓ in pulmonary vascular resistance in neonatal period → ↓ in right-to-left shunting through PFO → gradual improvement in cyanosis in 1st weeks of life
- Other signs/symptoms
  - Hydrops fetalis in neonatal cases
  - Severe cardiomegaly in fetal life → pulmonary hypoplasia
  - Thrombosis, paradoxical embolus
  - Arrhythmias
    - Atrial fibrillation, atrial flutter → irregular heartbeat
    - Accessory atrioventricular conduction pathways (preexcitation) → tachyarrhythmias, which can be unexpected & fatal

Demographics

- Age
  - 1st presentation can range from newborn period through old age (average: 14 years)
- Epidemiology
  - < 1% of congenital cardiac anomalies, incidence 1/210,000 live births
  - M:F = 1:1

Natural History & Prognosis

- Sudden death due to fatal atrial arrhythmias
- Uncomplicated pregnancies possible in women with hemodynamically well-balanced lesions
- Prognosis highly variable, dependent on hemodynamic significance of tricuspid regurgitation, presence of cyanosis

Treatment

- Supportive treatment in cyanotic neonate: Oxygen, nitric oxide ventilation to lower pulmonary vascular resistance
- Systemic to pulmonary (Blalock-Taussig & central) shunts ineffective
- Some patients benefit from total right-sided heart bypass procedures (Glenn → Fontan surgical treatment pathway)
- Tricuspid valve replacement &/or reconstruction (valvuloplasty): Definitive repair procedure
  - Valvuloplasty & bioprosthesis placement preferable to mechanical valve (allow growth, no need for lifelong anticoagulation)
  - Valvuloplasty uses tissues from existing valve (redundant anterior tricuspid leaflet)
  - Bioprosthesis: Homograft or xenograft (porcine valve)
- Indications for valve repair
  - NYHA classes III & IV
  - NYHA classes I & II with cardiothoracic ratio > 0.65
  - Significant cyanosis (arterial saturation < 80%) &/or polycythemia (Hb > 16 g/dL)
  - History of paradoxical embolus
  - Arrhythmia due to accessory atrioventricular pathway
- Conal reconstruction may improve long-term right ventricle performance & prognosis
- Arrhythmia treatments
  - Antiarrhythmic drugs
  - Permanent pacemaker implantation
  - Radiofrequency ablation

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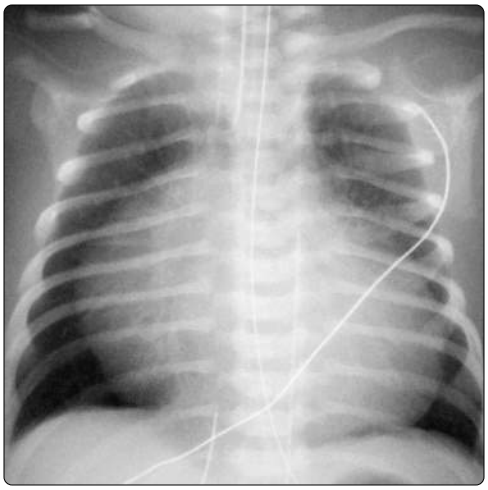
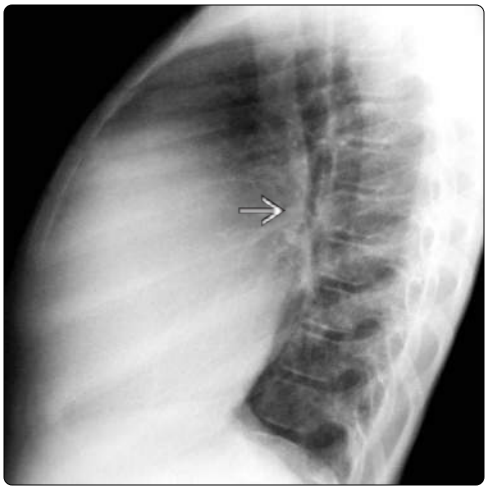
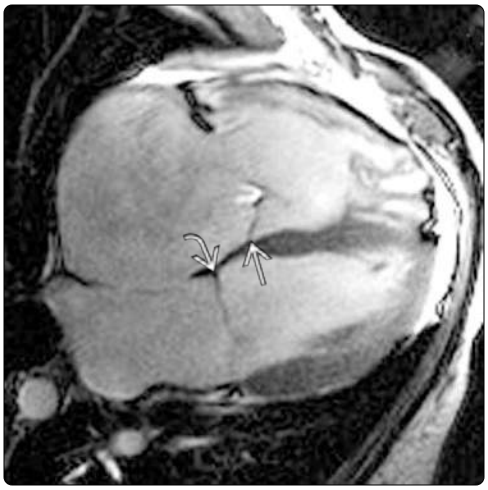
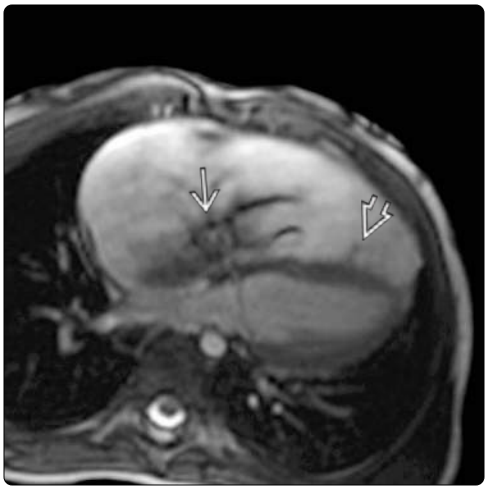


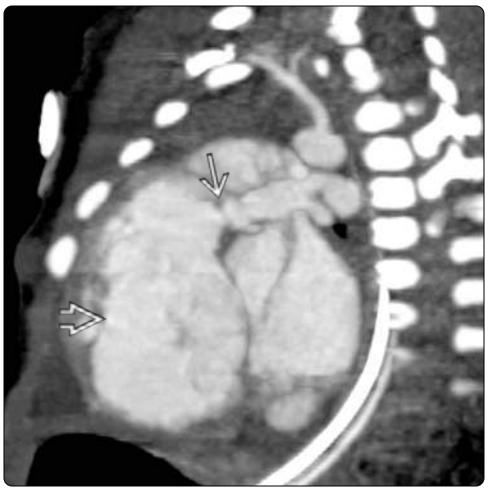


(Left) *AP radiograph of the chest demonstrates cardiomegaly with decreased pulmonary vascularity in this patient with Ebstein anomaly. The classic differential considerations in patients with cardiomegaly & decreased pulmonary vascularity are Ebstein anomaly, pulmonic atresia, & tricuspid atresia.* **(Right)** *Lateral radiograph of the chest shows cardiomegaly with very small pulmonary vessels around the hilum ⇨ in this patient with Ebstein anomaly.*





(Left) *Four-chamber bright blood cine MR shows a 16-mm distance between the septal leaflets of the mitral ⇨ & tricuspid ⇨ valves, which is diagnostic of Ebstein anomaly.* **(Right)** *Four-chamber bright blood cine MR of the heart shows dephasing artifact of turbulent flow from tricuspid regurgitation ⇨. The septal leaflet of the tricuspid valve is displaced apically ⇨.*



(Left) *Oblique cardiac CTA in an infant with Ebstein anomaly shows valvular pulmonic stenosis ⇨. Notice the dilated right ventricle ⇨.* **(Right)** *Oblique 3D reformation from a cardiac CTA in an infant with Ebstein anomaly shows valvular pulmonic stenosis ⇨. Note the massively dilated right atrium (light blue) & the moderately dilated right ventricle (purple).*

D-Transposition of Great Arteries

KEY FACTS

TERMINOLOGY

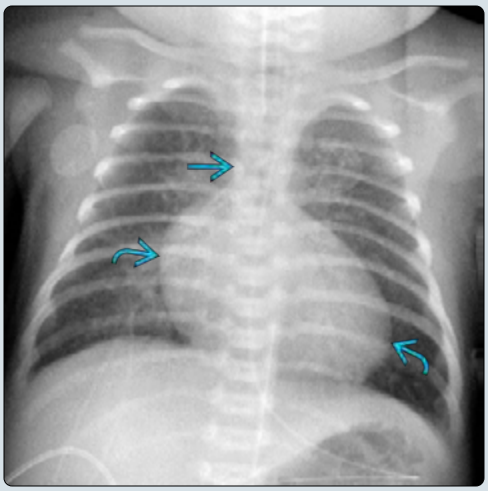
- Ventriculoarterial discordance with atrioventricular concordance: Aorta arises from right ventricle (RV) & pulmonary artery (PA) arises from left ventricle (LV)
- Complete separation of pulmonary & systemic circulations, lethal without flow admixture: Patent foramen ovale, ventricular septal defect (VSD), patent ductus arteriosus (PDA)

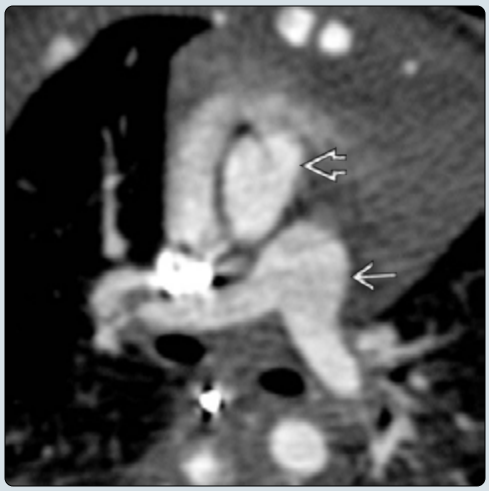
IMAGING

- Preoperative
  - Great vessels lie parallel & almost in same sagittal plane, with aortic valve in anterior position & slightly to right (D-loop) of pulmonary valve; coronary anomalies common
  - Classic radiographic appearance: Narrow mediastinum with cardiomegaly ("egg on string/egg on its side" heart) + ↑ pulmonary vascularity
- Postoperative (after arterial switch/Jatene procedure)
  - Classic alterations of great vessel anatomy
    - PA now anteriorly positioned with posterior aorta in same sagittal plane; right & left PAs now drape over ascending aorta
  - Transposed coronary arteries
  - Traction on both branch PAs may lead to stenosis

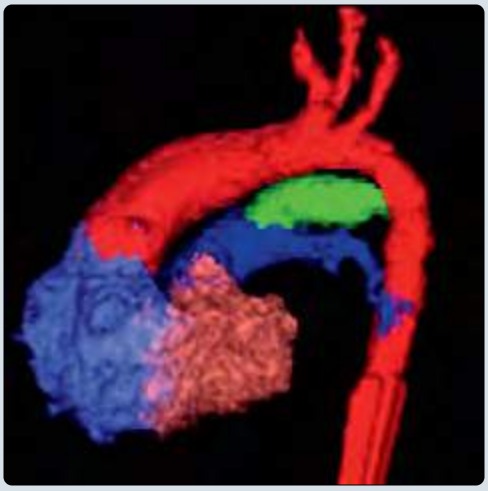
CLINICAL ISSUES

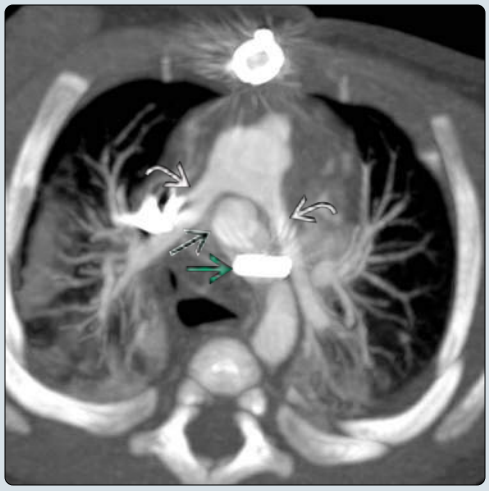
- Simple transposition: Good prognosis with early switch
- Large VSD: Congestive heart failure as neonate
- Patients with large VSD & (sub-) pulmonic stenosis have mild symptoms & may survive for years without treatment
- Long-term prognosis determined by potential coronary abnormalities
- Potential treatments: Prostaglandin E1 to maintain PDA preoperatively; emergency balloon atrial septostomy (Rashkind); early surgery (preferred): Arterial switch with transposition of coronaries (Jatene); late surgery: Rerouting of venous flow in atria with pericardial baffle (Mustard) or reorientation of atrial septum (Senning)





(Left) *Frontal radiograph of the chest shows cardiomegaly & increased pulmonary vascularity in a patient with D-transposition of great arteries (D-TGA). The superior mediastinum is narrow, & the heart is globular in shape. This combination is referred to as the egg-on-a-string appearance.* **(Right)** *Axial cardiac CTA shows the pulmonary artery (PA) posterior & slightly to the left of the ascending aorta. This is the classic relationship of the great vessels seen with D-TGA.*





(Left) *Lateral color-coded 3D CTA reformation in a D-TGA patient shows that the aorta (red) arises from the right ventricle (purple), & the PA (blue) arises from the left ventricle (pink). A large patent ductus arteriosus (PDA) (green) connects the aortic arch to the PA (blue).* **(Right)** *Axial MIP from a cardiac CTA status post arterial switch procedure shows the typical postop anatomy with the PAs draped around the ascending aorta. PDA clip is noted. Most TGA patients have a persistent large PDA that needs to be ligated.*

TERMINOLOGY

Definitions

- Aorta arises from right ventricle (RV) & pulmonary artery (PA) arises from left ventricle (LV)
- Ventriculoarterial discordance with atrioventricular concordance
- Category: Cyanotic, cardiomegaly, ↑ pulmonary vascularity
- Hemodynamics
  - RV connected with systemic circulation: Pressure overload
  - LV connected with pulmonary circulation: Volume overload
  - Lethal without flow admixture: Patent foramen ovale (PFO), ventricular septal defect (VSD), patent ductus arteriosus

IMAGING

General Features

- Best diagnostic clue
  - Great vessels lie parallel in almost same sagittal plane: Aortic valve in anterior position & slightly to right (D-loop) of pulmonary valve

Radiographic Findings

- Radiography
  - Classic appearance: Narrow mediastinum with cardiomegaly ("egg on a string or egg on its side" heart) + ↑ pulmonary vascularity

CT Findings

- CTA
  - Shows classic alterations of great vessel anatomy status post arterial switch procedure: PA now anteriorly positioned with posterior aorta in same sagittal plane; right & left PAs now drape over ascending aorta
    - Traction on both branch PAs may lead to stenosis
    - Anterior tracheal or left main bronchus compression may be seen
    - Variable coronary anomalies in D-TGA; transposition performed during surgery

MR Findings

- T1WI
  - Cardiac-gated axial images for segmental cardiac analysis: Atrioventricular concordance & ventriculoarterial discordance
    - Presence of PFO, VSD, (sub-) pulmonary stenosis
    - Postoperative assessment of PA stenosis
- T2* GRE
  - Multiplanar bright blood SSFP cine gold standard for cardiac function evaluation, ventricular volume measurements
    - RV dysfunction following atrial switch procedures (as RV not able to sustain systemic circulation)
- MRA
  - Gadolinium-enhanced MRA for postoperative PA stenosis
- Phase-contrast MR
  - Flow velocity measurements to calculate gradients across stenoses: Mustard/Senning or PA stenoses

CLINICAL ISSUES

Presentation

- Severe cyanosis not improving with oxygen, with little respiratory distress

Demographics

- Epidemiology
  - Incidence: 1 in 3,000 live births
  - 5% of congenital heart disease

Natural History & Prognosis

- Early death without communicating shunt
- Large VSD: Congestive heart failure in neonatal period
- Patients with large VSD & (sub-) pulmonic stenosis have mild symptoms & may survive for years without treatment
- Simple transposition: Good prognosis with early switch
  - Long-term prognosis determined by potential coronary abnormalities
- Complication of arterial switch: Traction on branch PAs by anteriorly transposed main PA → branch origin stenosis

Treatment

- Prostaglandin E1 to keep ductus arteriosus open preoperatively
- Emergency balloon atrial septostomy (Rashkind)
- Early surgery: Arterial switch with transposition of coronaries (Jatene)
- Late surgery (if Jatene not performed): Rerouting of venous flow in atria with pericardial baffle (Mustard) or reorientation of atrial septum (Senning)

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L-Transposition of Great Arteries

KEY FACTS

TERMINOLOGY

- "Congenitally corrected transposition" (misnomer)
- Inversion of ventricles & great arteries: Atrioventricular (AV) discordance & ventriculoarterial discordance
  - Right atrium connects via mitral valve to right-sided morphologic left ventricle (LV), which connects to pulmonary circulation
  - Left atrium connects via tricuspid valve to left-sided morphologic right ventricle (RV), which connects to systemic circulation
- Category: Dependent on associated anomalies
  - Ventricular septal defect (VSD) (60-70%): Acyanotic, ↑ pulmonary vascularity
  - Left ventricular outflow tract (subpulmonary) obstruction (30-50%): Cyanotic
  - Only 1% have no associated anomalies: True congenitally corrected transposition

IMAGING

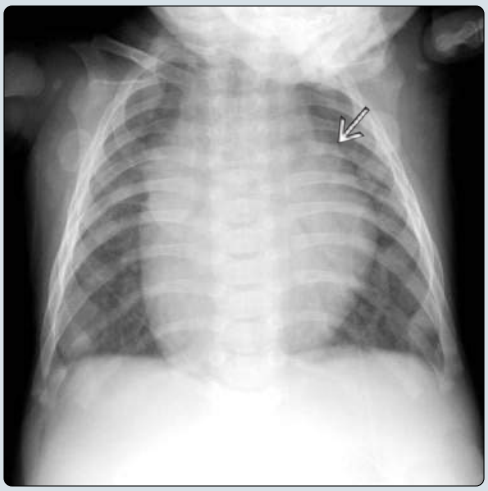
- Classic radiograph: Straight upper left heart border
- CT & MR demonstrate complex anatomy

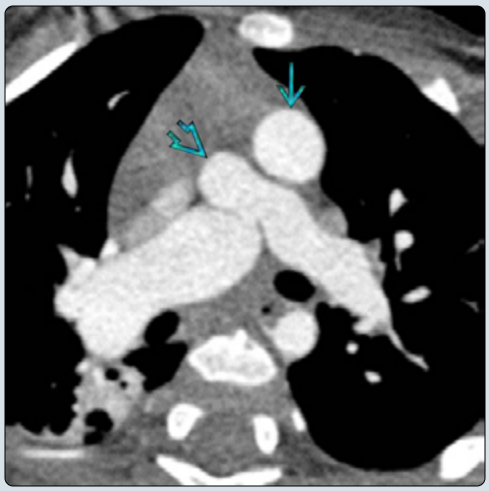
PATHOLOGY

- VSD: 80%
- LV outflow tract (subpulmonary) obstruction: 30-50%
- Left-sided tricuspid valve dysplasia, Ebstein anomaly, regurgitation: 30%

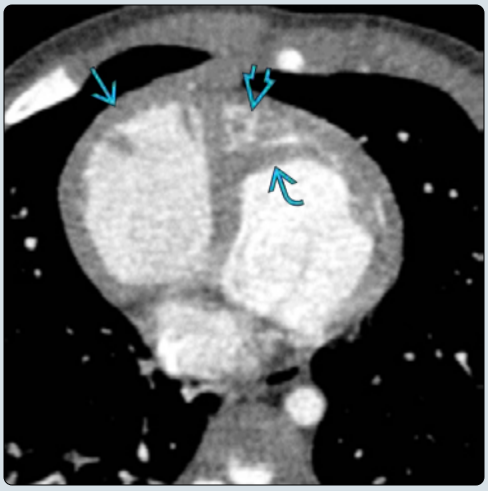
CLINICAL ISSUES

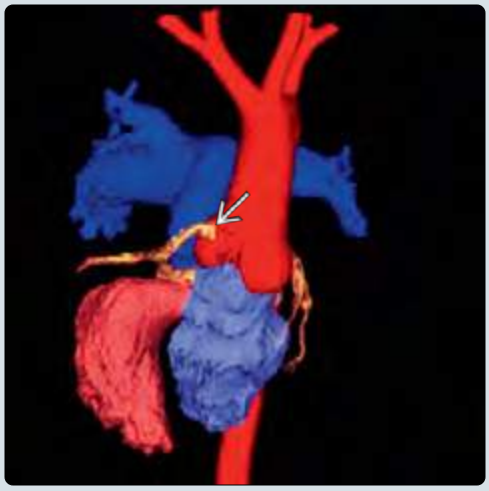
- Guarded prognosis due to progressive systemic AV valve & RV dysfunction after corrective surgery: 50% mortality after 15 years
- Patients with true congenitally corrected transposition may have normal life expectancy





(Left) *Frontal radiograph of the chest shows a straightened left upper heart border in a patient with levo-transposition of the great arteries (L-TGA).* **(Right)** *Axial image from a cardiac CTA shows the typical position of the great vessels in L-TGA. The aorta (Ao) is anterior & to the left of the pulmonary artery (PA).*





(Left) *Axial image from a cardiac CTA in a patient with L-TGA shows a smooth-walled morphologic left ventricle (LV) on the right. The morphologic right ventricle (RV) is on the left, demonstrating a characteristic moderator band.* **(Right)** *Frontal view from a 3D color-coded cardiac CTA shows the morphologic LV (pink) on right giving rise to the PAs (blue) & the morphologic RV (purple) on the left giving rise to the Ao (red). The right coronary artery arises from the right coronary sinus & branches into the LAD & circumflex.*

TERMINOLOGY

Synonyms

- "Congenitally corrected transposition" (misnomer)
- Discordant transposition

Definitions

- Inversion of ventricles & great arteries: Atrioventricular (AV) discordance & ventriculoarterial discordance
- Category: Dependent on associated anomalies
  - Ventricular septal defect (VSD) (60-80%): Acyanotic, ↑ pulmonary vascularity
  - Left ventricular outflow tract (subpulmonary) obstruction (30-50%): Cyanotic
  - Only 1% have no associated anomalies: True congenitally corrected transposition

IMAGING

General Features

- Best diagnostic clue
  - Great vessels lie parallel & almost in same coronal plane with aortic valve in anterior position & slightly to left (L-loop) of pulmonary valve
- Morphology
  - S, L, L heart: Atrial situs solitus, L-loop, L-transposed great arteries
    - Right-sided morphologic left ventricle (LV) characterized by associated mitral valve, smooth wall, & absent outflow chamber to pulmonary valve
    - Left-sided morphologic right ventricle (RV) characterized by associated tricuspid valve, trabeculated wall with moderator band, & infundibulum below aortic valve

Radiographic Findings

- Classic plain film appearance: Straight upper left heart border

CT Findings

- 3D CT angiography can depict abnormal AV & ventriculoarterial relationships
- Cardiac CTA to establish coronary artery anatomy

MR Findings

- Multiplanar cardiac-gated T1WI & 3D gadolinium MRA for segmental cardiac analysis & anatomic evaluation
- SSFP cine & phase contrast to evaluate function & outflow tract obstruction
  - Phase contrast imaging used to calculate shunt fraction (Qp:Qs)

DIFFERENTIAL DIAGNOSIS

Congestive Heart Failure, Increased Pulmonary Blood Flow

- Isolated VSD
- Double inlet ventricle
- Tricuspid atresia with ↑ pulmonary blood flow
- Double outlet right ventricle + subaortic VSD

PATHOLOGY

General Features

- Associated abnormalities
  - VSD: 60-80%
  - LV outflow tract (subpulmonary) obstruction: 30-50%
  - Left-sided tricuspid valve dysplasia, Ebstein anomaly, regurgitation: 30%
  - Coronary distribution is mirror image of normal (right-sided coronary bifurcates into circumflex & anterior descending arteries)
- Embryology: Primitive cardiac tube loops to left (L-loop) → ventricular inversion & left-sided position of ascending aorta

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Congestive heart failure (VSD, systemic AV valve dysfunction)
  - Cyanosis (subpulmonary stenosis)
  - Rarely completely asymptomatic, presenting as incidental finding on chest radiograph (straight upper left heart border)

Natural History & Prognosis

- Determined by presence of AV valve dysfunction
- Guarded prognosis due to progressive systemic AV valve & RV dysfunction after corrective surgery: 50% mortality after 15 years

Treatment

- Surgical treatment focused on associated abnormalities
  - Congestive heart failure from VSD shunt: PA banding or VSD closure
  - Cyanosis from subpulmonary stenosis: Systemic to PA shunt (Blalock-Taussig) or LV to PA conduit (Rastelli)
- Double switch operation to prevent late systemic ventricular (RV) failure
  - Venous switch (Senning) reroutes atrial blood into appropriate ventricles
  - Ventricular (Rastelli) or arterial switch: Morphologic LV becomes systemic ventricle

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Tricuspid Atresia

KEY FACTS

TERMINOLOGY

- Congenital absence or agenesis of tricuspid valve & inlet portion of right ventricle

IMAGING

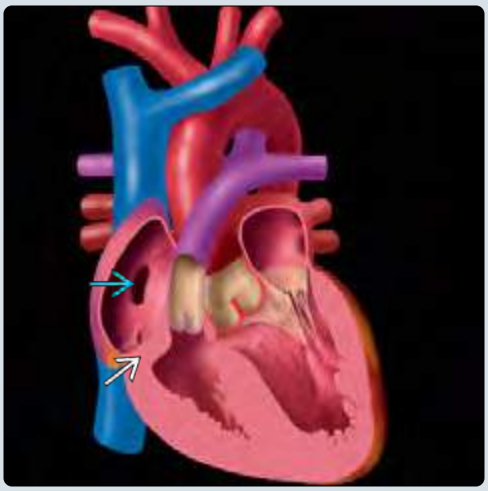
- Type I: Normally related great arteries (70-80%)
- Type II: D-transposition of great arteries (12-25%)
- Type III: L-transposition of great arteries/malposition (3-6%)
- Small ventricular septal defect (VSD) → heart usually normal in size, right ventricle hypoplastic, pulmonary flow diminished
- Large VSD → heart usually large with ↑ flow or transposition of great arteries
- MR: Excellent for postoperative left ventricular functional assessment & anatomy of caval-pulmonary artery anastomosis
- CTA: Useful to assess for pulmonary artery embolus & collateral vessels in children who have increasing cyanosis
  - Knowledge of previous surgical procedure is critical to correctly prescribing protocol & interpreting imaging, particularly in Glenn anastomosis & Fontan procedures (as unopacified blood can mimic thrombus)

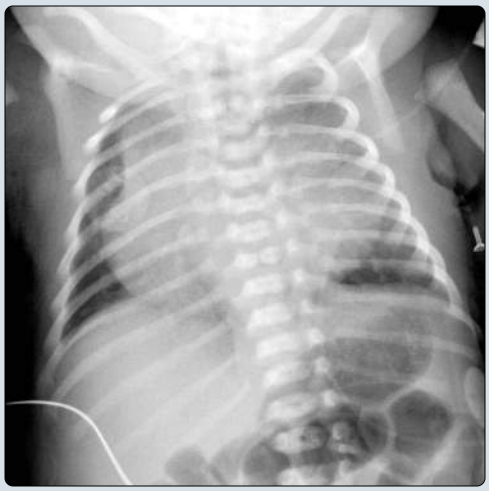
TOP DIFFERENTIAL DIAGNOSES

- Ebstein anomaly
- Tetralogy of Fallot

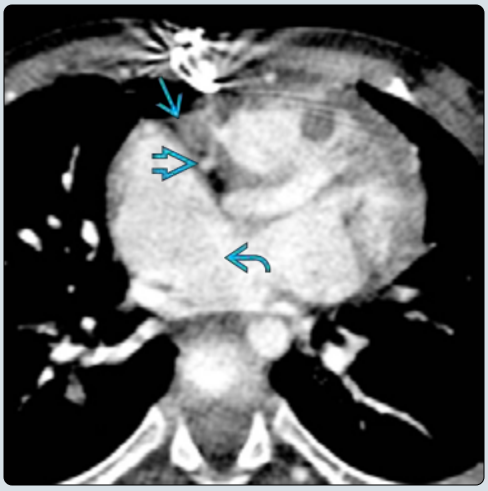
CLINICAL ISSUES

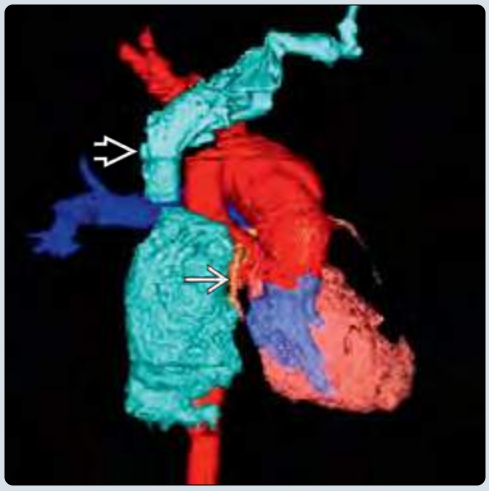
- 50% of neonates present with cyanosis in first 24 hours
- 30% present with signs of congestive heart failure
- Left axis deviation on newborn ECG usually diagnostic
- Staged surgical approach, similar to single ventricle
  - Modified Blalock-Taussig shunt with systemic artery to pulmonary flow
  - Bidirectional Glenn anastomosis with superior vena cava to pulmonary artery
  - Modified Fontan procedure with inferior vena cava conduit to pulmonary artery





(Left) *Coronal graphic shows tricuspid atresia where there is no forward flow into the right ventricle due to obstruction at the level of the right atrioventricular (AV) groove. There is an atrial septal defect (ASD), a ventricular septal defect (VSD), & a hypoplastic right ventricle (RV).* **(Right)** *Single frontal radiograph of the chest shows cardiomegaly with decreased pulmonary vascularity in an infant with tricuspid atresia.*





(Left) *Image from a cardiac CTA shows fat & soft tissue in the right AV groove where the tricuspid valve is normally located. Notice the right coronary artery deep within the groove. A large ASD is also seen.* **(Right)** *3D reformat of a cardiac CTA shows a Glenn shunt with SVC directly connected to the PAs (dark blue). Note the appearance of tricuspid atresia with wide separation of the right atrium (light blue) & hypoplastic right ventricle (purple). Right coronary artery lies in the AV groove where the tricuspid valve should be.*

TERMINOLOGY

Definitions

- Congenital absence or agenesis of tricuspid valve & inlet portion of right ventricle

IMAGING

General Features

- Best diagnostic clue
  - Absence of inflow portion of right ventricle with atretic tricuspid valve
  - Outlet portion of right ventricle depends on size of ventricular septal defect (VSD)
- Location
  - Tricuspid valve absent, fused, or stenotic; size of VSD & right ventricle variable
- Morphology
  - Type I: Normally related great arteries (70-80%)
  - Type II: D-transposition of great arteries (12-25%)
  - Type III: L-transposition of great arteries or malposition (3-6%)
  - VSD can occur with atresia, stenosis, or normal pulmonary valve

Radiographic Findings

- Neonatal chest variable depending on size of VSD

CT Findings

- CECT
  - Demonstrates postoperative anastomosis, relative size of pulmonary arteries & systemic veins, & presence of collateral venous anatomy
  - Can be useful to assess for pulmonary artery embolus & collateral vessels in children with increasing cyanosis
    - Must know underlying anatomy & type of surgical repair to interpret contrast studies as unopacified blood can mimic thrombus

MR Findings

- MR cine
  - Excellent for postoperative left ventricular functional assessment & anatomy of caval-pulmonary artery anastomosis
  - 3D contrast-enhanced MRA or spin-echo imaging can be used to assess connections & relations as well as size of proximal pulmonary arteries

Echocardiographic Findings

- Defines size & position of VSD, right ventricle, & associated abnormalities

Angiographic Findings

- Cardiac catheterization prior to staged cardiac surgery repairs or for complications
  - Coiling of collateral arteries & veins to reduce workload of left ventricle

Other Modality Findings

- Newborn pattern of left axis deviation on electrocardiography (ECG) is usually diagnostic

DIFFERENTIAL DIAGNOSIS

Tetralogy of Fallot

- Large aorta (right sided in 25%), concave left hilum, & ↓ peripheral flow

Ebstein Anomaly

- Newborn chest may show massive cardiomegaly
- Spectrum of disease, which involves downward displacement of septal & posterior leaflets of tricuspid valve

PATHOLOGY

General Features

- Etiology
  - Early embryologic insult with fusion of valve leaflet → stenosis (partial fusion) or atresia (complete fusion) of valves
- Genetics
  - Associated with asplenia syndromes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 50% of neonates present with cyanosis in first 24 hours
  - 30% present with signs of congestive heart failure
- Other signs/symptoms
  - Extracardiac anomalies may occur in 20% of patients

Demographics

- Epidemiology
  - 3% of congenital heart lesions

DIAGNOSTIC CHECKLIST

Consider

- Knowledge of previous surgical procedure critical to correctly prescribing protocol & interpreting imaging, particularly in Glenn anastomosis & Fontan procedures

Image Interpretation Pearls

- Hallmark is lack of direct anatomic continuity between right atrium & ventricle
- Right coronary artery & fat seen within atrioventricular groove where tricuspid valve should be normally positioned
- Ventricular anatomy, type & size of VSD & relationship of great vessels, ventriculoarterial connections, & sources of pulmonary artery flow all need to be assessed

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KEY FACTS

TERMINOLOGY

- Common arterial vessel (trunk) arising from heart; gives rise to aorta, pulmonary arteries (PAs), & coronaries
- Congenital heart lesion most commonly associated with right aortic arch (30-40%)

IMAGING

- High (outlet) ventricular septal defect (VSD) immediately below truncal valve
- Classic radiograph: Cardiomegaly, ↑ pulmonary vascularity, narrow mediastinum, right aortic arch

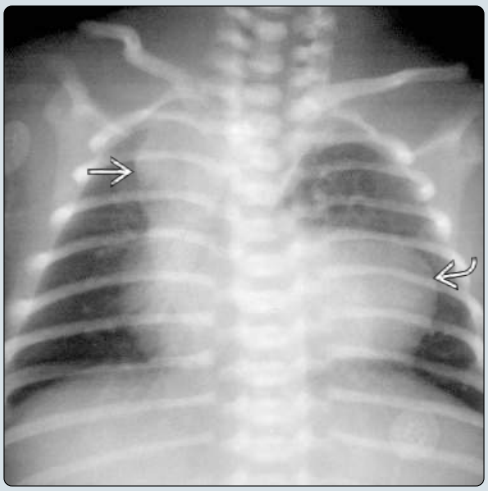
PATHOLOGY

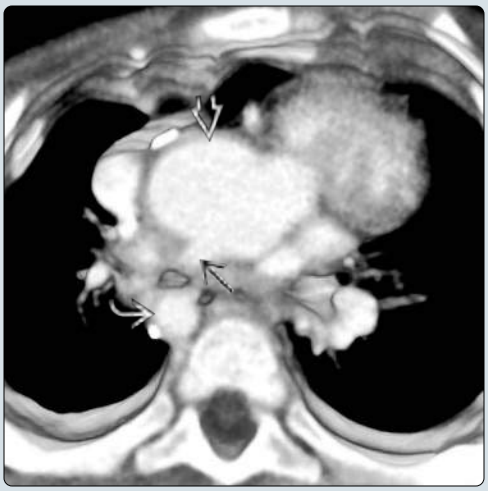
- Classification of Collett & Edwards
  - Type 1: Separation of common trunk into ascending aorta & main PA
  - Type 2: Common take-off of branch PAs from trunk with no main PA
  - Type 3: Both branch PAs originate separately from posterolateral aspect of ascending aorta
  - Type 4: "Pseudotruncus": Pulmonary arterial supply from major aortopulmonary collateral arteries (MAPCAs) arising from descending aorta; controversial entity, misnomer for pulmonary atresia with VSD & MAPCAs

CLINICAL ISSUES

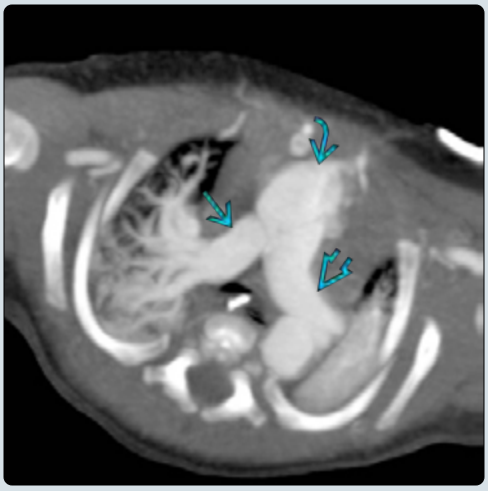
- Untreated: 65% 6-month & 75% 1-year mortality
- Intractable congestive heart failure → pulmonary hypertension → shunt reversal → increasing cyanosis
- Associated thymic agenesis in DiGeorge syndrome → T-cell immunodeficiency
- Early complete repair (at 2-6 weeks of life) favored by most surgeons: Placement of conduit between right ventricle & PA with closure of VSD
- Truncal valve dysfunction (regurgitation) common → need for valvuloplasty, prosthesis

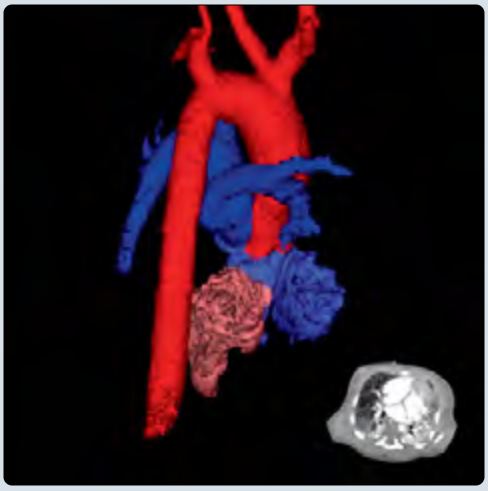
(Left) *AP radiograph shows cardiomegaly with a right aortic arch & increased pulmonary vascularity in an infant with truncus arteriosus. The apex of the heart is upturned. This appearance is often mistaken for tetralogy of Fallot.* **(Right)** *Axial CTA MIP shows a small right pulmonary artery (PA) originating from the posterior aspect of the large common trunk. Notice the descending thoracic aorta on the right.*





(Left) *Axial CTA MIP of an infant with truncus arteriosus shows both right & left PAs coming off of a central common trunk.* **(Right)** *Posterior oblique image from a 3D color-coded cardiac CTA reconstruction shows the PAs (blue) arising from the posterior aspect of the ascending aorta (red), consistent with truncus arteriosus.*





TERMINOLOGY

Abbreviations

- Common arterial trunk (CAT)

Definitions

- Common arterial vessel arising from heart, giving rise to aorta, pulmonary arteries (PAs), & coronaries
- Classic radiographic appearance: Cardiomegaly, ↑ pulmonary vascularity, narrow mediastinum, right aortic arch
  - Truncus arteriosus is heart lesion most commonly associated with right aortic arch (30-40%)
- Category: Cyanotic, cardiomegaly, ↑ pulmonary vascularity
- Hemodynamics
  - Both ventricles connected to pulmonary & systemic circulation
  - Flow admixture across ventricular septal defect (VSD) & within truncus → cyanosis
  - Postnatal drop in pulmonary vascular resistance → relative ↑ in pulmonary blood flow → volume overload of pulmonary circulation
- Frequently associated with absent thymus & parathyroid glands: DiGeorge syndrome

IMAGING

General Features

- Best diagnostic clue
  - CAT arising from both ventricles
  - Large common valve with 2-5 leaflets

Radiographic Findings

- Radiography
  - Cardiomegaly
  - Active pulmonary vascular congestion (shunt vascularity)
  - Right aortic arch common
  - Narrow mediastinum due to thymic agenesis
  - Dilated PAs may compress neighboring bronchi → atelectasis

CT Findings

- CTA
  - Shows relationship of branch PAs with truncus
  - Coronary anatomy well demonstrated
  - Postoperative: Patency & size of conduit, Ca^{2+}, & stenosis
  - CTA is best technique to evaluate stent placement & airway compression

MR Findings

- T1WI
  - Cardiac-gated axial images for preoperative definition of PA anatomy
  - Postoperative: Conduit stenosis, anastomotic pseudoaneurysm
- T2* GRE
  - Steady state free precession cine MR: Truncal valve regurgitation, ventricular function & volumes
- MRA
  - Gadolinium-enhanced MRA for global anatomy, patency of PA conduit
- Phase contrast
  - Velocity-encoded phase-contrast imaging to evaluate regurgitant fraction of common truncal valve or postoperative regurgitation/stenosis
  - Qp:Qs calculated with phase-contrast imaging

Echocardiographic Findings

- Echocardiogram
  - CAT originating from both ventricles
  - High (outlet) VSD immediately below truncal valve
  - Common truncal valve with 2 (5%), 3 (60%), or 4 (25%) cusps, rarely 5
- Color Doppler
  - Bidirectional flow across VSD
  - Truncal valve regurgitation

Angiographic Findings

- Conventional
  - Cardiac catheterization with angiography
    - To define exact type of truncal anatomy
    - Truncal valve insufficiency
    - Hemodynamic study is gold standard for calculation of pulmonary vascular resistance

Imaging Recommendations

- Protocol advice
  - Primary diagnosis made with echocardiography
  - MR/CTA for preoperative delineation of PA anatomy
  - MR/CTA for postoperative assessment of conduit regurgitation/stenosis, stent placement

DIFFERENTIAL DIAGNOSIS

Transposition of Great Arteries

- Presents earlier in life with more severe cyanosis; ductus dependent

Aortopulmonary Window

- Congenital fenestration between separate ascending aorta & PA, with separate aortic & pulmonary valves

Common Atrioventricular Canal

- When unbalanced (right or left dominant), cyanosis frequently occurs due to admixture

PATHOLOGY

General Features

- Genetics
  - Strong association with deletion on long arm of chromosome 22 (22q11 syndrome)
  - Associated findings: Cardiac anomalies (truncus arteriosus), abnormal facies, thymic hypoplasia, cleft palate, & hypocalcemia with deletion of chromosome 22 (CATCH-22)
    - Includes DiGeorge syndrome, velocardiofacial syndrome, & conotruncal anomaly face syndrome
  - Theory: Abnormal migration of neural crest tissue that interferes with development of cardiac tube
- Associated abnormalities
  - Right-sided aortic arch with mirror-image branching (30-40%)
  - Persistence of primitive aortic arches
  - Absent thymus & parathyroid glands

- Embryology
  - Lack of separation of primitive bulbus cordis into aorta & main PA
  - Associated persistence of primitive aortic arches
- Pathophysiology: Congestive heart failure vs. cyanosis (degree of cyanosis determined by balance between pulmonary & systemic vascular resistances)
  - Marked ↑ in pulmonary blood flow in early neonatal period due to drop in pulmonary vascular resistance → slight improvement in cyanosis but worsening congestive heart failure
  - Development of pulmonary vascular obstructive disease → improvement in congestive heart failure but worsening cyanosis

Staging, Grading, & Classification

- Classification of Collett & Edwards
  - Type 1: Separation of common trunk into ascending aorta & main PA
  - Type 2: Common take-off of branch PAs from trunk with no main PA
  - Type 3: Both branch PAs originate separately from posterolateral aspect of ascending aorta
  - Type 4: "Pseudotruncus": Pulmonary arterial supply from major aortopulmonary collateral arteries (MAPCAs) arising from descending aorta; controversial entity, misnomer for pulmonary atresia with VSD & MAPCAs
- Classification of Van Praagh
  - Type A1: Same as Collett & Edwards type 1
  - Type A2: Collet & Edwards types 2 & 3 combined
  - Type A3: Unilateral PA with collateral supply to contralateral lung
  - Type A4: Truncus with interrupted aortic arch

Gross Pathologic & Surgical Features

- Common outflow tract of both ventricles over nonrestrictive VSD
- Position of common trunk with respect to VSD
  - Predominantly positioned over right ventricle (42%)
  - Predominantly positioned over left ventricle (16%)
  - Equally shared (42%)
- No separate outflow portion (infundibulum) of right ventricle
- Many variations exist involving interruption of aortic arch (11-14%), absence of branch PA (hemitruncus), & patent ductus arteriosus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Progressive congestive heart failure with drop in pulmonary vascular resistance in young infant
  - Increasing cyanosis due to shunt reversal with development of pulmonary hypertension
- Other signs/symptoms
  - T-cell immunodeficiency (thymic agenesis in DiGeorge syndrome)
  - Neonatal tetany (absent parathyroid glands)

Demographics

- Epidemiology
  - 2% of congenital cardiac anomalies

Natural History & Prognosis

- Untreated: 65% 6-month & 75% 1-year mortality
- Intractable congestive heart failure
  - Marked ↑ in pulmonary flow after drop in pulmonary vascular resistance
  - Aggravated by presence of truncal valve regurgitation (in 50% of cases)
- Eventual shunt reversal with progressive cyanosis & sudden death
  - Pulmonary vascular obstructive disease with Eisenmenger physiology can develop as early as 6 months of age
- Postoperative course determined by function of PA conduit & morbidity of conduit replacement

Treatment

- Palliative: Banding of main PA
  - Initial palliation with PA banding is often unsatisfactory with early development of pulmonary vascular disease → pulmonary hypertension
  - Early complete repair (at 2-6 weeks of life) favored by most surgeons
- Surgical repair with placement of conduit between right ventricle & PA or creation of connection with atrial appendage & monoleaflet valve with closure of VSD
  - Conduit revisions frequently necessary throughout patient's lifetime
    - Patient outgrows fixed conduit size
    - Ca^{2+}, stenosis, neointimal hyperplasia
    - Anastomotic pseudoaneurysm
    - Conduit valve dysfunction (regurgitation)
  - Truncal valve dysfunction (regurgitation) is common → need for valvuloplasty, prosthesis

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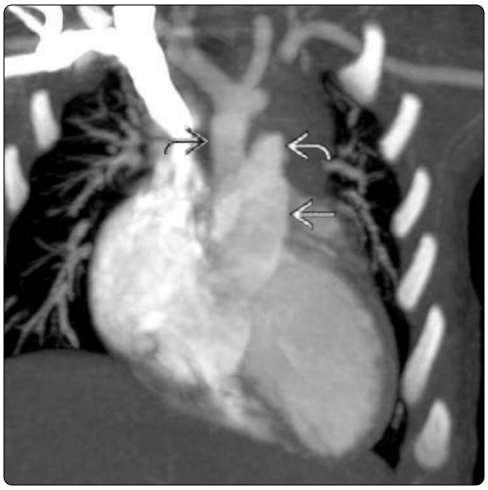
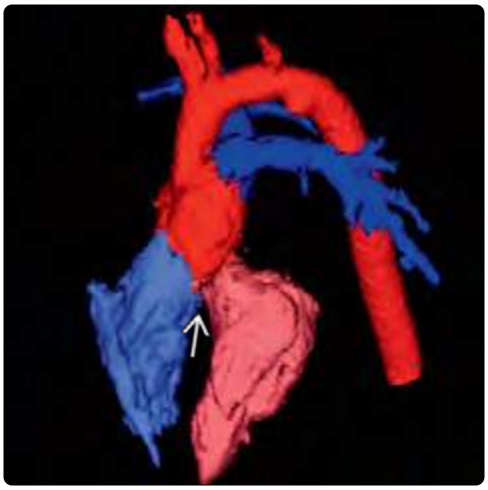
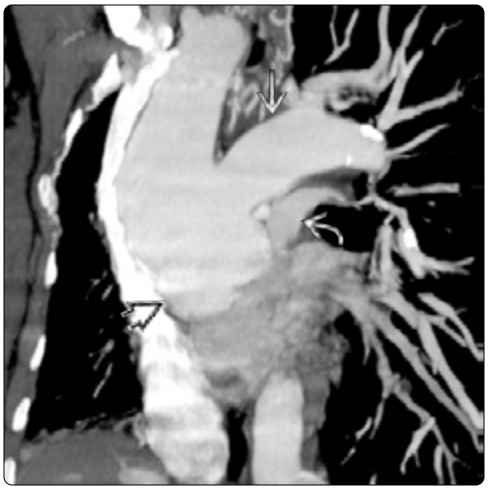
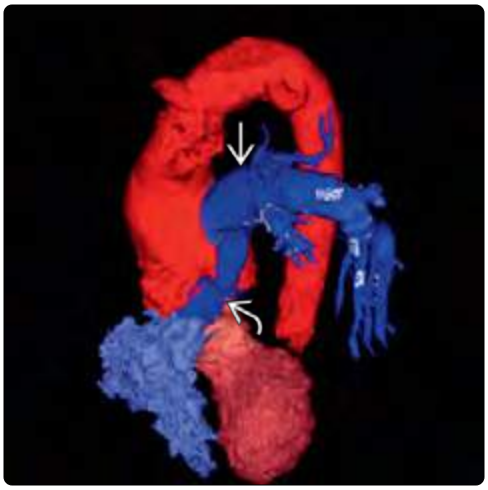


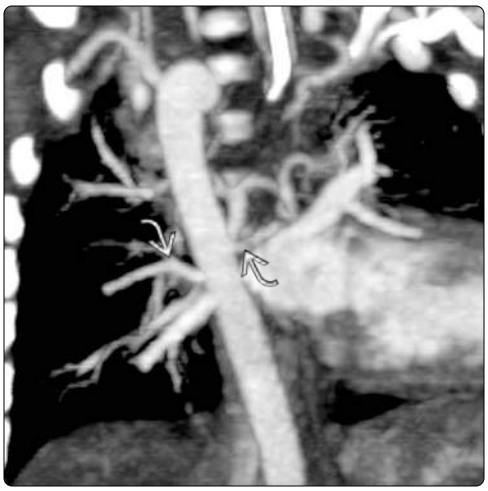


(Left) *Oblique MIP from a cardiac CTA in an infant with truncus arteriosus shows a common trunk ➡ giving rise to both the aorta ⇨ & the PA ➡. (Right) Lateral view from a 3D color-coded cardiac CTA shows a common trunk giving rise to a main PA (blue) that divides into right & left PAs (type 1 truncus arteriosus). Notice the ventricular septal defect where the right (purple) & left (pink) ventricles communicate ➡ with the overriding common trunk.*





(Left) *Oblique MIP from a cardiac CTA shows the left PA ➡ arising from a common aortic trunk ⇨. There is a small right PA ➡. Notice the PA Ca^{2+} that developed on the left from longstanding pulmonary hypertension.* **(Right)** *Oblique view from a 3D color-coded cardiac CTA shows the left PA ➡ arising from a common aortic trunk (red) & the right PA ➡ arising from the RV (purple). This is a hemitruncus, which is a unilateral variation of truncus arteriosus.*



(Left) *Coronal MIP from a cardiac CTA in an infant shows multiple collateral vessels ➡ arising from the descending aorta (pseudotruncus) in a patient with severe pulmonary artery hypoplasia & tetralogy of Fallot.* **(Right)** *Frontal view from a 3D color-coded cardiac CTA in a patient with tetralogy of Fallot & pseudotruncus shows multiple aortopulmonary collaterals (green) arising from the descending aorta (red).*

Total Anomalous Pulmonary Venous Return

KEY FACTS

TERMINOLOGY

- Total anomalous pulmonary venous return (TAPVR) or "drainage": Failure of connection between pulmonary veins (PVs) & left atrium
- Category: Cyanotic; heart size & pulmonary vascularity depend on type
- All pulmonary venous return goes to right heart (extracardiac left-to-right shunt)
- All types are admixture lesions
- Supracardiac TAPVR (type I, 40-50%): "Vertical" common PV joins left innominate vein
- Cardiac TAPVR (type II, 20-30%): Common PV joins coronary sinus or right atrium
- Infracardiac TAPVR (type III, 10-30%): Common PV joins portal vein, ductus venosus, or inferior vena cava

IMAGING

- Cardiomegaly (types I, II); small heart (type III)
- Shunt vascularity (types I, II); pulmonary edema (type III)
- Type I: "Snowman" heart
- Type II: Indistinguishable from atrial septal defect (ASD)
- Type III: Small heart, reticular pattern in lungs (edema)

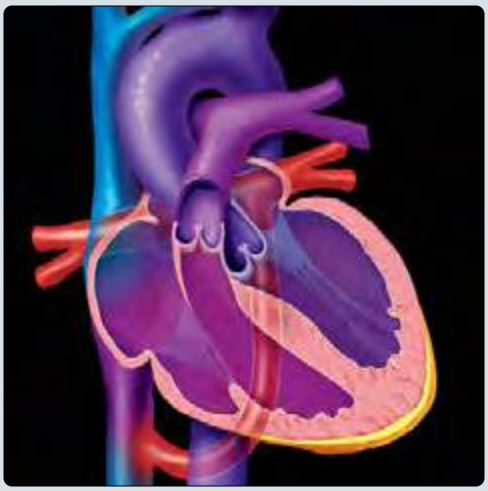
PATHOLOGY

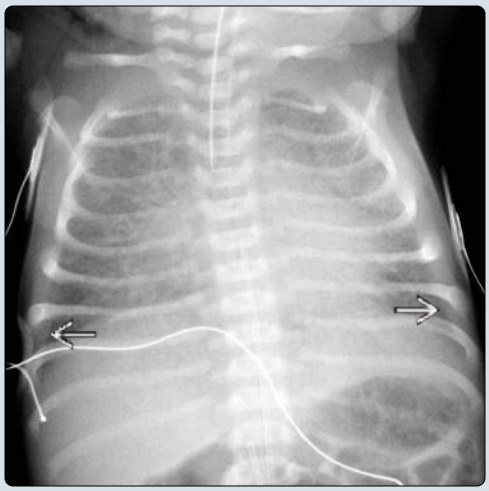
- All types have patent foramen ovale (PFO) to allow for obligatory right-to-left flow → varying degrees of cyanosis
- TAPVR (type III): Common PV obstructed by diaphragmatic hiatus → pulmonary venous congestion & edema
- Left-sided cardiac chambers may be underdeveloped, especially in TAPVR type III (due to ↓ in systemic flow)

CLINICAL ISSUES

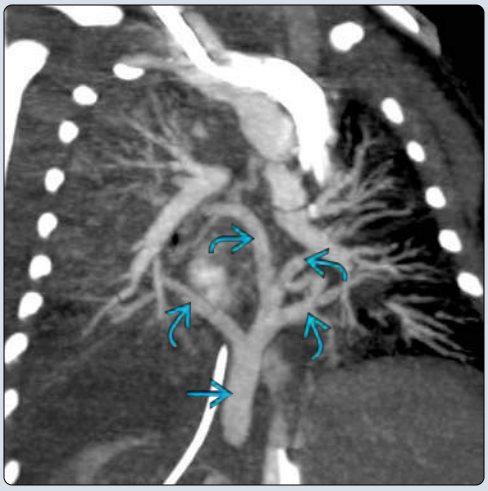
- Treatment: Early surgical anastomosis of pulmonary venous confluence to left atrium
- Anastomotic PV stenosis in up to 18% of TAPVR repairs
- Longstanding PV stenosis → irreversible pulmonary hypertension (arterial pulmonary vascular disease)
- Angioplasty of distal PVs often required after surgery

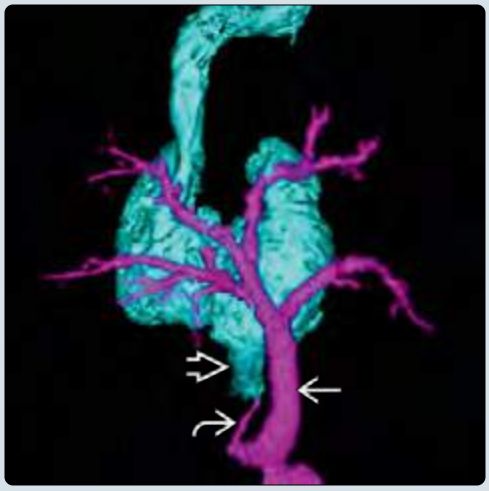
(Left) *Graphic shows an infradiaphragmatic total anomalous pulmonary venous return (TAPVR) (type III) to the inferior vena cava constituting an obligatory extracardiac left-to-right shunt. Mixed blood flows to the left atrium through a patent foramen ovale.* **(Right)** *Frontal radiograph in a newborn shows diffuse pulmonary edema with small bilateral pleural effusions. Notice that the heart size is normal in this patient with type III TAPVR.*





(Left) *Coronal MIP CTA of an infant with an obstructed type III TAPVR shows that the pulmonary veins connect to a large vertical vein that then drains below the diaphragm.* **(Right)** *Posterior view of a color-coded 3D CTA shows the pulmonary veins (purple) emptying into a large inferior vertical vein in a patient with obstructed infracardiac TAPVR. Only a small venous connection was seen to the inferior vena cava. Notice the symmetric atria (light blue) in this patient with heterotaxy.*





TERMINOLOGY

Abbreviations

- Total anomalous pulmonary venous return (TAPVR) or "drainage"
  - Refers to hemodynamics: To where does pulmonary venous flow return (drain)

Synonyms

- Total anomalous pulmonary venous connection
  - Refers to anatomy: To where do pulmonary veins (PVs) connect

Definitions

- Failure of connection between PVs & left atrium
- Category: Cyanotic; heart size & pulmonary vascularity depend on type
- Hemodynamics
  - All pulmonary venous return goes to right heart (extracardiac left-to-right shunt)
  - Intracardiac right-to-left shunt through patent foramen ovale (PFO)
  - All types are admixture lesions
- 3 types
  - Supracardiac TAPVR (type I, 50%): "Vertical" common PV joins left innominate vein
  - Cardiac TAPVR (type II, 20%): Common PV joins coronary sinus or right atrium
  - Infracardiac TAPVR (type III, 20%): Common PV joins portal vein, ductus venosus, hepatic veins, or inferior vena cava
  - Mixed or complex TAPVR when there is mixture of above types (10%)

IMAGING

General Features

- Best diagnostic clue
  - No PVs connecting to left atrium
  - Supra or infracardiac vertical vein

Radiographic Findings

- Radiography
  - Cardiomegaly (types I, II); small or normal heart size (type III)
  - Shunt vascularity (types I, II); pulmonary edema (type III)
    - Types I & II rarely may be obstructive with pulmonary edema & effusions
  - Wide mediastinum (type I, "snowman heart"), narrow mediastinum (types II, III, thymic atrophy)
  - Classic plain film appearance
    - Type I: "Snowman" heart (due to left vertical vein)
    - Type II: Indistinguishable from atrial septal defect (ASD)
    - Type III: Small heart, reticular pattern in lungs: Edema

CT Findings

- CTA useful to define anatomy in complex types of TAPVR & to define additional congenital heart lesions
  - Thickened interlobular septa, peribronchial cuffing, & ground-glass opacities suggest anastomotic PV stenosis
  - Airway compression well visualized by CT
- Can be used postoperatively for evaluation of PV caliber & anastomoses

MR Findings

- T1WI
  - Cardiac-gated black blood imaging: Anomalous connections best seen in coronal plane
- MRA
  - Velocity-encoded phase-contrast MRA: For detection of PV anastomotic stenosis (flow velocities > 100 cm/sec diagnostic)
    - Phase contrast
      - Qp:Qs calculation
      - Elevated velocities in distal vertical vein
  - Dynamic time-resolved gadolinium-enhanced 3D MRA: For detailed depiction of PV anatomy
- MR cine
  - Steady-state free precession cine MR for functional assessment, flow jets, regurgitation

Echocardiographic Findings

- Echocardiogram
  - Lack of connection of PVs to left atrium
  - Right-sided chamber enlargement in types I, II
  - PFO
  - Associated cardiac & abdominal situs abnormalities
  - Evaluation of complex types limited by ultrasound
  - Limited assessment for postoperative anastomotic PV obstruction

Angiographic Findings

- Conventional
  - Seldom required for primary diagnosis
  - Balloon atrial septostomy when flow across ASD restricted
  - After repair: For diagnosis & treatment of anastomotic PV stenosis

Imaging Recommendations

- Protocol advice
  - Primary diagnosis with echocardiography
  - CT, MR for postoperative PV anastomotic stenosis
  - Conventional angiography to treat postoperative PV stenosis

DIFFERENTIAL DIAGNOSIS

Cor Triatriatum

- Pulmonary venous connection occurred but remains stenotic with lack of incorporation of common PV into left atrial wall

Hypoplastic Left Heart Syndrome

- PVs insert normally into left atrium with left-to-right shunting through PFO

Persistent Fetal Circulation Syndrome/Primary Pulmonary Hypertension

- Associated with
  - Severe surfactant deficiency disease
  - Meconium aspiration

PATHOLOGY

General Features

- Genetics
  - No specific genetic defect found
  - Associated with heterotaxy syndromes
- Associated abnormalities
  - Single ventricle, atrioventricular septal defect, truncus arteriosus, tetralogy of Fallot, anomalous systemic venous connection
  - Asplenia syndrome with symmetric bilateral morphologic right atria
  - Polysplenia syndrome less common
  - Biliary atresia
- All anomalous pulmonary venous drainage eventually flows into right atrium
- Embryology
  - Lack of normal incorporation of primitive common PV into posterior wall of left atrium
  - Persistence & enlargement of embryological pathways for pulmonary venous return via umbilicovitteline & cardinal veins
- Pathophysiology
  - All types have PFO to allow for obligatory right-to-left flow → varying degrees of cyanosis (less severe in types I, II: Pulmonary hypercirculation)
  - Nonobstructive TAPVR (types I, II): ASD physiology, pulmonary plethora, congestive heart failure
  - Obstructive TAPVR (type III): Common PV obstructed by diaphragmatic hiatus → pulmonary venous congestion & edema
  - Left-sided cardiac chambers may be underdeveloped, especially in TAPVR type III, due to prenatal ↓ in systemic blood flow

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Types I, II: Congestive heart failure
  - Type III: Severe cyanosis at birth
  - Patent ductus arteriosus: Persistent fetal circulation

Demographics

- Epidemiology
  - 1-3% of congenital heart disease
  - More frequent in neonatal period

Natural History & Prognosis

- No patients survive without surgical treatment
- Natural history is highly variable
  - Types I, II: Initially asymptomatic, with gradual development of congestive heart failure when pulmonary vascular resistance drops (ASD physiology)
  - Type III, obstructive forms: Death within month
- After surgical repair: Prognosis determined by associated cardiac anomalies & development of PV anastomotic stenosis
  - Progressive distal PV stenosis often seen → significant morbidity & mortality

Treatment

- Prostaglandin E1 to improve systemic perfusion in pulmonary hypertension
- Preoperative extracorporeal membrane oxygenation occasionally necessary to improve oxygenation & systemic perfusion
- Early surgical anastomosis of pulmonary venous confluence to left atrium
  - Anastomotic PV stenosis may occur in up to 18% of TAPVR repairs
  - Longstanding PV stenosis → irreversible pulmonary hypertension (arterial pulmonary vascular disease)
  - Reoperation performed using sutureless technique, with pericardial patch augmentation of anastomotic stenoses
- Angioplasty of distal PVs often required after surgery

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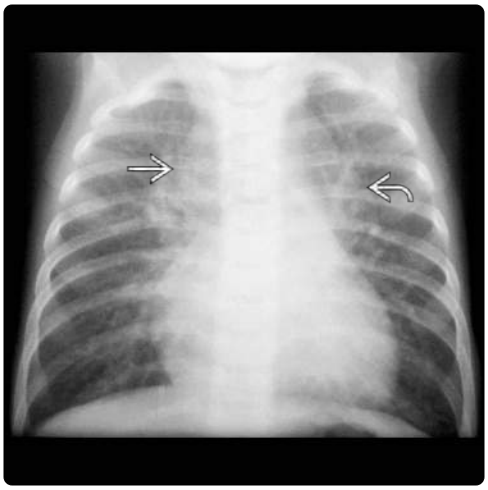
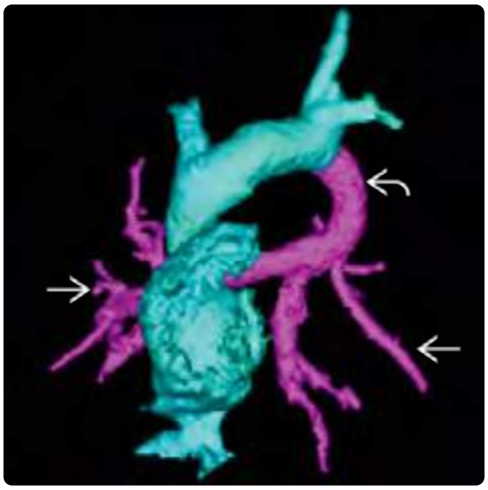
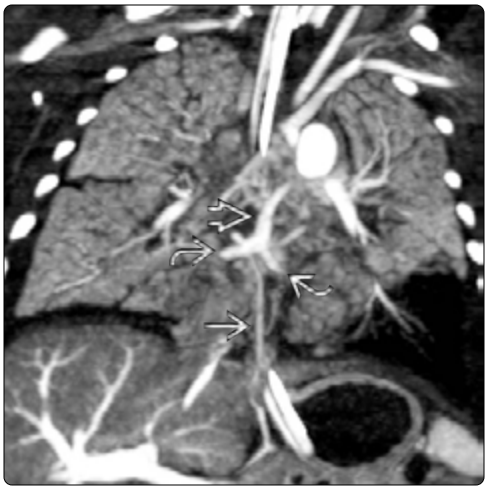
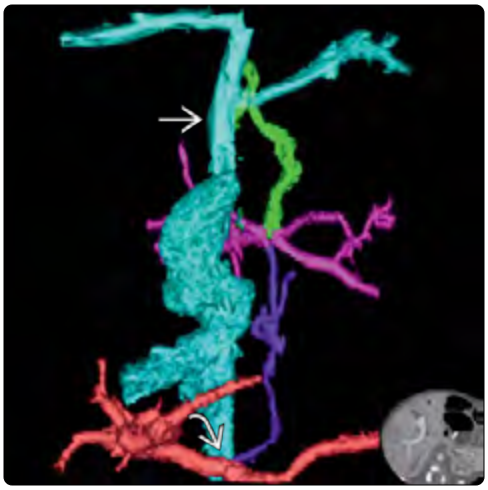


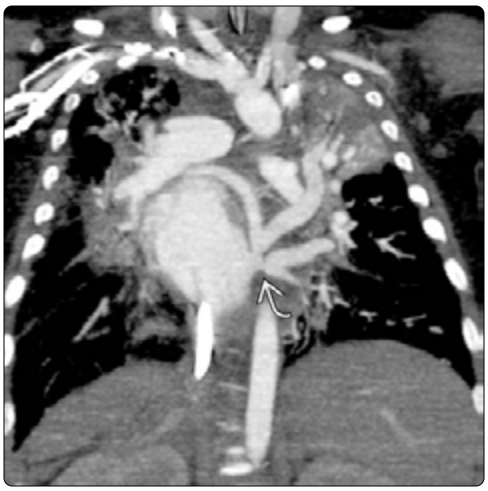


(Left) *Frontal radiograph shows a snowman appearance of the superior mediastinum in a patient with a supracardiac TAPVR. The left-sided vertical vein forms the border of the superior left mediastinum. The dilated innominate vein & superior vena cava form the superior & right borders of the superior mediastinum, respectively.* **(Right)** *Frontal projection from a 3D color-coded cardiac CTA shows the pulmonary veins emptying into a large left-sided vertical vein in this patient with a supracardiac TAPVR.*





(Left) *Coronal MIP from a cardiac CTA shows 2 pulmonary veins emptying into superior & inferior vertical veins. Notice the diffuse pulmonary edema from insufficient vertical vein drainage of the lungs in this obstructed complex TAPVR.* **(Right)** *Frontal view from a color-coded 3D CTA in a complex TAPVR patient shows pulmonary veins (pink) draining into superior (green) & inferior (purple) vertical veins. The superior vertical vein drains to the SVC, & the inferior vertical vein drains to the portal vein.*



(Left) *Coronal cardiac CTA in a patient status post TAPVR repair shows stenosis of the distal pulmonary veins. This is a common complication seen in postoperative TAPVR patients.* **(Right)** *Posterolateral view from a cardiac CTA in a patient after TAPVR repair shows stenosis of the distal pulmonary veins (pink) at the anastomosis with the atrium (blue). 3D images can be very helpful for surgical planning.*

Hypoplastic Left Heart Syndrome

KEY FACTS

TERMINOLOGY

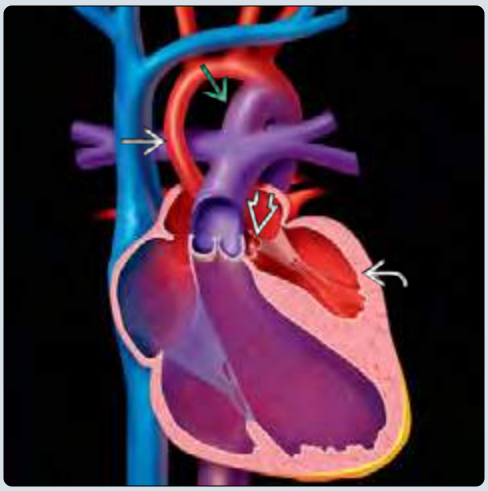
- Most severe congenital heart lesion
  - Presents in neonatal period with congestive heart failure, cardiogenic shock, & cyanosis
- Category: Cyanotic, cardiomegaly, ↑ pulmonary vascularity
- Hypoplasia/atresia of ascending aorta, aortic valve, left ventricle (LV), & mitral valve
- Secondary findings: Patent ductus arteriosus, juxtaductal coarctation

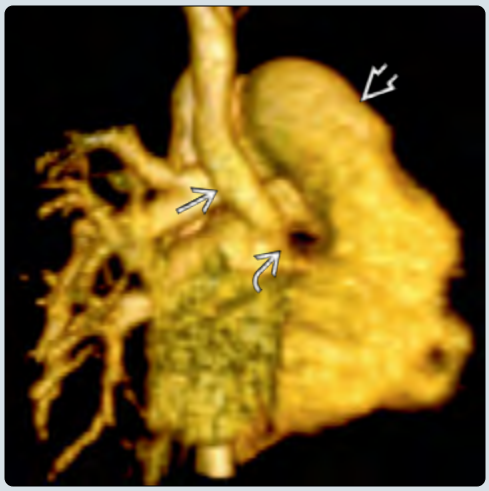
IMAGING

- Chest radiograph shows cardiomegaly, large right atrium, pulmonary venous congestion with interstitial fluid
- CT can evaluate patency of aortopulmonary (Blalock-Taussig) & cavopulmonary (Glenn) shunts
- MR for functional assessment of univentricular heart to determine suitability for Fontan operation
- Postnatal echocardiogram sufficient for treatment planning
  - Diminutive ascending aorta < 5 mm
  - Small, thick-walled LV; presence of endocardial fibroelastosis (EFE)
  - Mitral valve size important to decide whether biventricular repair possible in marginally hypoplastic LVs
  - Dilation of right cardiac chambers & pulmonary artery
  - Size & location of ductus arteriosus
  - Unrestricted atrial level shunt (patent foramen ovale vs. atrial septal defect) critical for immediate postnatal care; may require emergent balloon atrial septostomy; saturations dependent on degree of atrial level shunting
  - Abnormal ventricular wall motion (ischemic damage, EFE)

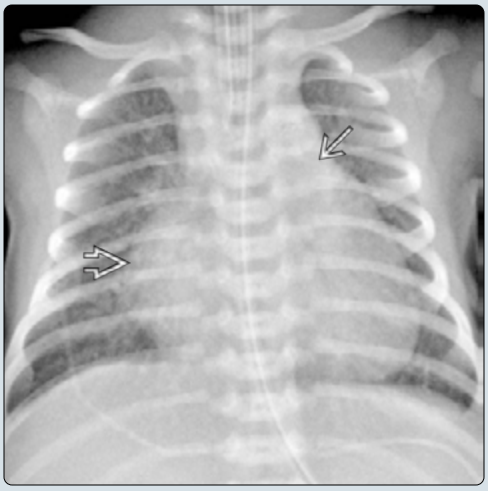
CLINICAL ISSUES

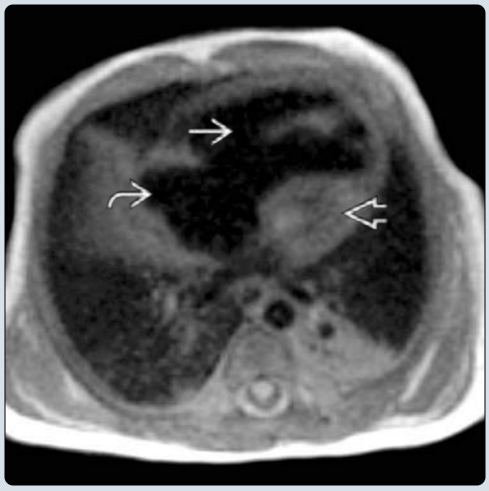
- Mortality high if not treated in neonatal period
- Prognosis improves substantially with surgical intervention





(Left) *Graphic shows hypoplasia of the LA, MV, LV, aortic valve, & ascending aorta. Systemic blood flow depends on the patency of the ductus arteriosus.* **(Right)** *Coronal oblique 3D reformation of an MRA in a 3 day old shows a small ascending aorta & a large main pulmonary artery (MPA). The proximal portion of the aortic sinuses & left ventricular outflow tract (LVOT) are not seen due to signal dropout caused by rapid blood flow across a stenotic LVOT. This area could be seen on source images.*





(Left) *AP radiograph in the same 3 day old shows the typically large MPA along the left border of the heart plus an enlarged right atrium due to left-to-right shunting of blood across the atrial septal defect. Mild pulmonary edema & increased pulmonary blood flow characterize the vasculature.* **(Right)** *Axial T1 MR at the level of the ventricles in the same patient shows the large right ventricle & the diminutive LV. Note the large right atrium.*

TERMINOLOGY

Abbreviations

- Hypoplastic left heart syndrome (HLHS)

Synonyms

- 4 classic types: Mitral atresia/aortic atresia, mitral atresia/aortic stenosis, mitral stenosis/aortic atresia, mitral stenosis/aortic stenosis

Definitions

- Hypoplasia/atresia of ascending aorta, aortic valve, left ventricle (LV), & mitral valve (MV)
  - Secondary findings: Patent ductus arteriosus (PDA), juxtaductal coarctation
- Most severe congenital heart lesion
  - Presents in neonatal period with congestive heart failure, cardiogenic shock, & cyanosis
- Category: Cyanotic, cardiomegaly, ↑ pulmonary vascularity
- Hemodynamics
  - Severe obstruction of flow to systemic circulation (ductal dependent)
  - Retrograde flow in hypoplastic aortic arch & ascending aorta for cranial & coronary perfusion
  - Volume overload in pulmonary circulation
  - Obligate left-to-right shunt at atrial level via patent foramen ovale (PFO) or secundum atrial septal defect (ASD)
    - Restrictive atrial septum in neonate is highly lethal & requires emergent catheter-based balloon atrial septostomy
  - Flow admixture in right atrium → severe cyanosis

IMAGING

General Features

- Best diagnostic clue
  - Hypoplasia of ascending aorta, LV

Radiographic Findings

- Cardiomegaly, pulmonary venous congestion with interstitial fluid, hyperinflation, narrow mediastinum due to thymic atrophy

Echocardiographic Findings

- Echocardiogram
  - HLHS often diagnosed prenatally
    - Retrograde flow in diminutive ascending aorta
    - LV growth arrest only becomes manifest between 18-22 weeks of gestation
  - Postnatal diagnosis with echo sufficient for treatment planning
    - Diminutive ascending aorta < 5 mm
    - Small, thick-walled LV
    - Mitral valve annulus z-score: Important parameter to decide whether biventricular repair is possible in marginally hypoplastic LVs
    - Dilation of right-sided cardiac chambers & main pulmonary artery (MPA)
    - Size & location of ductus arteriosus
    - Atrial level shunt: PFO or secundum ASD (often superior secundum defect); any degree of atrial level restriction can play significant role in decision making pre- & postnatally
    - Abnormal ventricular wall motion (ischemic damage); endocardial fibroelastosis of diminutive LV
    - Tricuspid valve regurgitation: Best marker of clinical outcomes
- Color Doppler
  - Diagnosis of HLHS subtype (mitral stenosis or atresia; aortic stenosis or atresia)
  - Left to right shunting through PFO/ASD: Mean diastolic gradient estimates degree of atrial-level restriction
  - Tricuspid valve regurgitation: Clinically important indicator

CT Findings

- CTA
  - Presurgical: Small left atrium with atretic/stenotic MV, hypoplastic LV with ascending & transverse aortic arch hypoplasia, single dilated right ventricle & MPA with large PDA continuation to descending aorta
  - Postsurgical: Patency of aortopulmonary (Blalock-Taussig) & cavopulmonary (bidirectional Glenn) shunts
  - Seroma associated with Blalock-Taussig shunt
  - Airway compression (left bronchus) &/or left pulmonary artery compression by dilated neoaortic arch following Norwood repair

MR Findings

- T2* GRE
  - SSFP cine MR for ventricular volume measurements in marginally hypoplastic left heart (to determine feasibility of biventricular repair)
  - Short-axis SSFP cine MR for functional assessment of univentricular heart (to determine suitability for Fontan operation)
- MRA
  - Velocity-encoded phase-contrast MRA for measurements of flow through aortic isthmus, PDA, & PFO
    - Can predict response to intraoperative test closure of ASD & PDA to determine feasibility of biventricular repair

Angiographic Findings

- Cardiac catheterization with angiography
  - Primarily interventional (as opposed to diagnostic): Obtained in case of atrial level restriction & need for early balloon atrial septostomy (BAS)
    - Delivery at pediatric-care center where immediate BAS can be performed
    - Cath & BAS can be performed via umbilical venous & arterial catheters
    - Retrograde flow in hypoplastic ascending aorta
    - Catheter can be placed across ductus arteriosus to image pulmonary arteries retrograde

Other Modality Findings

- CTA, MR: Occasionally performed after staged Norwood procedure

- Presurgical: Unusual anatomic variants or borderline LV volumes (mitral stenosis/aortic stenosis variants); severe single ventricle dysfunction
- Postsurgical: Functional assessment of marginally hypoplastic left heart with cine MR & velocity-encoded phase-contrast MRA, prior to Fontan operation
- Residual stenosis of neoaortic arch, coarctation

DIFFERENTIAL DIAGNOSIS

Interrupted Aortic Arch

- Pressure overload of normally developed LV

Hepatic Congenital Hemangioma

- Structurally normal heart with volume overload of all chambers

Endocardial Fibroelastosis

- Globally enlarged, structurally normal heart with myocardial dysfunction

Anomalous Left Coronary Artery From Pulmonary Artery

- Left coronary originates from pulmonary artery (PA) → myocardial infarction

Severe Arrhythmias: Paroxysmal Supraventricular Tachycardia

- Characteristic electrocardiogram

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - No circulatory symptoms immediately at birth but rapid deterioration
    - Congestive heart failure (volume overload pulmonary circulation)
    - Cardiogenic shock after closure of PDA
    - Cyanosis (flow admixture in right heart)
    - Hypoxia → pulmonary hypertension, persistent fetal circulation
- Other signs/symptoms
  - Poor systemic perfusion, metabolic acidosis
    - Acute tubular necrosis, renal failure
    - Necrotizing enterocolitis

Demographics

- Epidemiology
  - 1-3 per 10,000 live births, M:F = 2:1
  - 4th most common congenital heart lesion presenting under 1 year (7-9%)

Natural History & Prognosis

- Death within days/weeks when untreated
- Prognosis improves substantially with treatment
- Prognosis determined by complications, residua, & sequelae of staged Norwood repair & Fontan operation (right ventricular dysfunction, venous hypertension)
- Significant tricuspid regurgitation after surgical palliation correlates with poor outcome

Treatment

- Medical: Prostaglandins initiated at birth/time of diagnosis to keep PDA open (provides right-to-left flow to descending aorta)
- Prenatal: US-guided balloon dilation of aortic valve in mid/late fetal period is being studied
  - Change in fetal hemodynamics may enhance prenatal growth of left-sided cardiac structures
- Rashkind BAS (in case of flow restriction across PFO)
- Palliative repair: 3-stage approach
  - Norwood: Neoaorta creation from pulmonary artery (with incorporation of native hypoplastic ascending aorta), atrial septectomy, & Blalock-Taussig shunt (Gore-Tex shunt from base of innominate artery to right pulmonary artery) OR Sano shunt (Gore-Tex shunt from RV to midpulmonary artery) for controlled pulmonary blood flow (1-3 weeks of age)
  - Damus-Kaye-Stansel anastomosis: Essentially occurs with every Norwood as hypoplastic ascending aorta is filleted & incorporated into neoaorta/pulmonary artery (classic description is side-to-side anastomosis between PA & aorta)
  - Conversion to hemi-Fontan: Glenn shunt between superior vena cava & right PA (4-6 months)
  - Bidirectional Glenn anastomosis: Off loads ventricle, not shunt dependent (more stable), prepares lungs for passive lung perfusion via cavopulmonary anastomosis
  - Fontan: Fenestrated venous conduit through right atrium for inferior cava flow to right PA (1.5-2 years)
- Marginally hypoplastic LV: Biventricular repair may be feasible
  - LV volume commonly underestimated with echocardiography
  - Functional MR (SSFP cine): Ventricular volumes & mass determination more reliable
- In some centers: Cardiac transplantation

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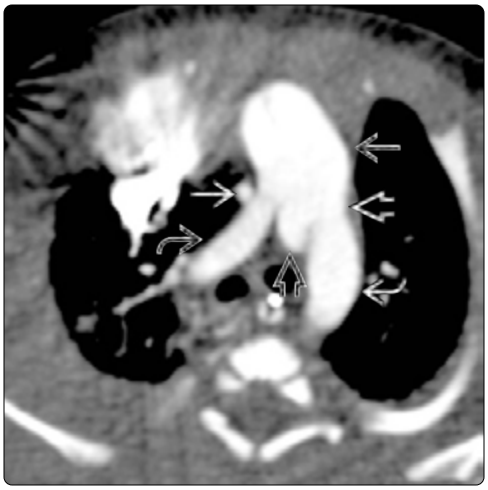
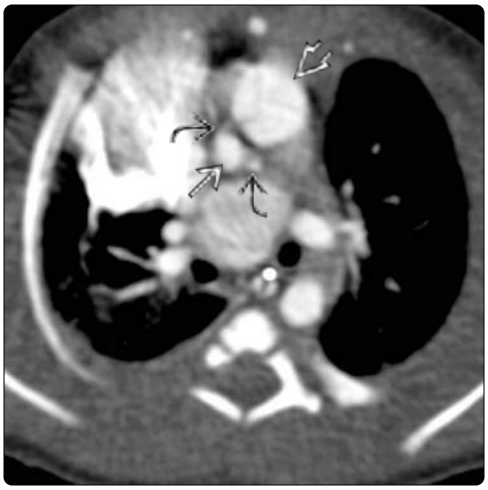
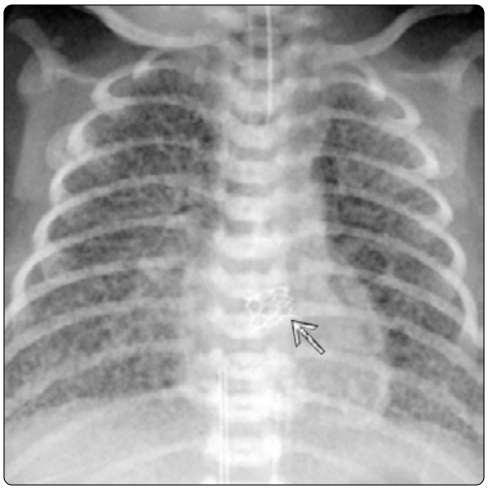
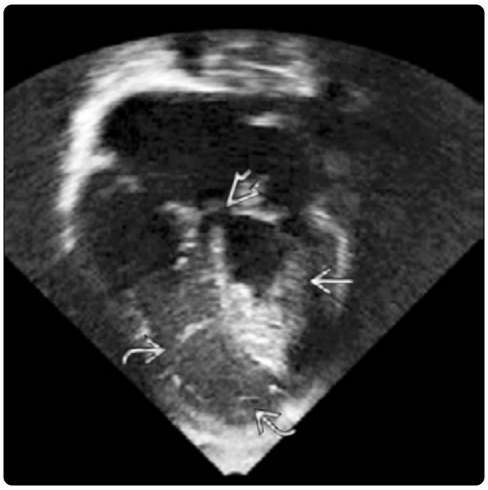


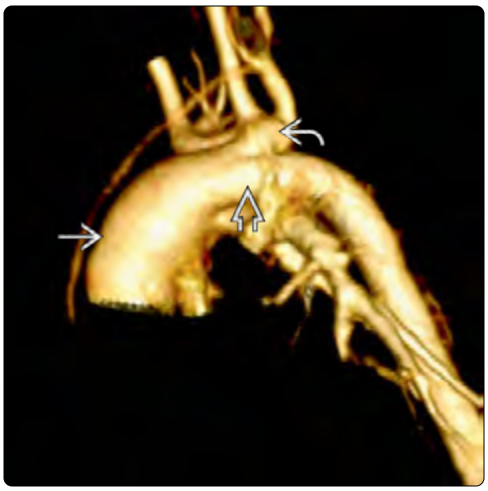


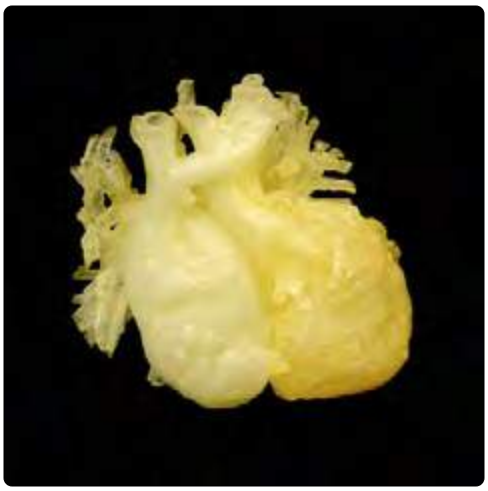
(Left) *Axial CTA shows a hypoplastic ascending aorta & a large MPA with a large patent ductus arteriosus supplying the descending aorta. Notice the right & left pulmonary arteries.* **(Right)** *Axial CTA in the same patient shows the coronary arteries arising from the coronary sinuses. Notice the normal relationship of the MPA & ascending aorta. Hypoplastic left heart syndrome (HLHS) patients usually have atrial-ventricular & ventricular-arterial concordance.*





(Left) *AP radiograph of the chest in a newborn with HLHS shows severe interstitial edema. The stent was placed after perforation of the interatrial septum to provide blood flow from the left atrium to the right atrium. Restricted blood flow to the right atrium explains the absence of right atrium enlargement.* **(Right)** *Four-chamber view echocardiogram shows a small muscle-bound LV & small LVOT. The right ventricle is large relative to the hypoplastic LV & wraps around the apex of the LV.*





(Left) *Oblique surface rendering of a cardiac CTA shows the very large MPA feeding a large patent ductus arteriosus, which provides blood flow to the descending aorta & the native aortic arch.* **(Right)** *3D printed heart model of an adult with HLHS status post classic atriopulmonary Fontan is shown. The 3D model was created for Fontan conversion surgery.*

Left Coronary Artery Anomalous Origin

KEY FACTS

TERMINOLOGY

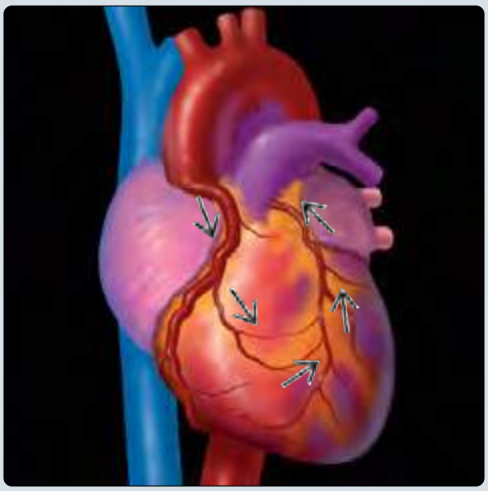
- Anomalous origin of left coronary artery from pulmonary artery: Most common congenital coronary artery anomaly presenting in children
- Causes cardiac ischemia & infarction, poor left ventricular (LV) systolic function, & significant mitral regurgitation

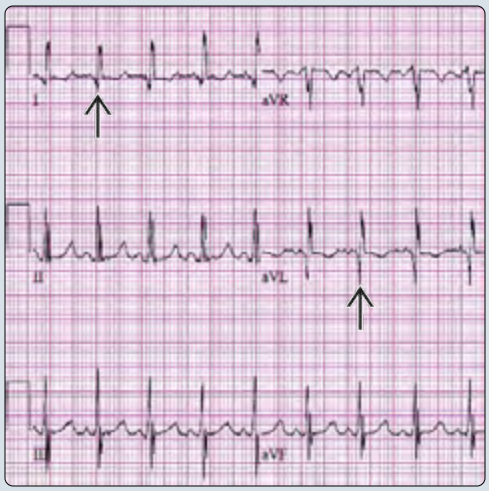
IMAGING

- Chest radiograph demonstrates cardiomegaly
- Echocardiogram (~ 90% diagnostic accuracy) demonstrates
  - Abnormal left coronary artery (LCA) ostium arising from pulmonary trunk
  - Retrograde flow in LCA toward pulmonary artery
  - Right coronary artery dilation & abundant intercoronary septal collaterals
  - Depressed LV systolic function & dilated LV chamber
  - Significant mitral regurgitation from ischemic papillary muscle dysfunction & mitral annular dilation
- CTA/MRA to identify coronary origins when echo limited (~ 100% diagnostic accuracy)

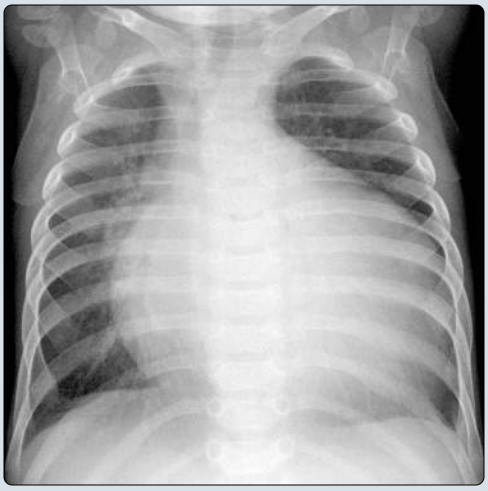
CLINICAL ISSUES

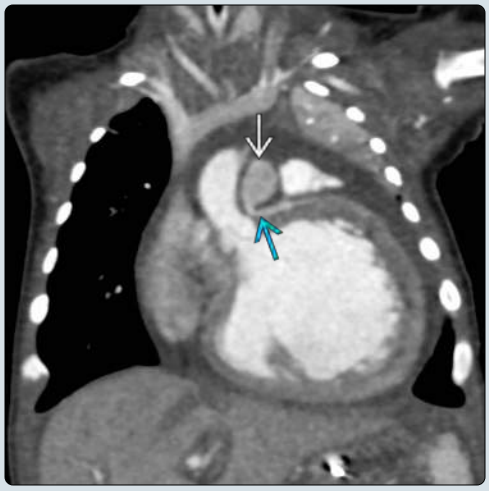
- Rare congenital anomaly; up to 90% mortality if not identified & surgically corrected
- Up to 90% present in infancy with nonspecific symptoms of irritability, failure to thrive, & wheezing (from mitral regurgitation)
  - ECG shows anterior lateral wall infarct pattern
- Older children often asymptomatic until sudden event with syncope, dysrhythmia, & occasional sudden cardiac death
  - Surgical options include coronary reimplantation to aorta, bypass grafting with anomalous origin ligation, Takeuchi baffle
  - Simultaneous repair of mitral valve controversial as severe regurgitation may improve with revascularization





(Left) *Graphic shows an anomalous origin of the left coronary artery (LCA) from the main pulmonary artery (PA). Collateral flow develops from the normal right coronary artery (RCA) to the LCA, allowing retrograde flow through the LCA to the low-resistance PA (with flow direction denoted ⇨). This flow bypasses the high-resistance myocardial bed of the left ventricle (LV).* **(Right)** *ECG of a neonate shows pathologic deep Q-waves in leads I & aVL ⇨, consistent with a diagnosis of ALCAPA & myocardial ischemia.*





(Left) *AP radiograph from an 8 month old with wheezing, grunting, & weight loss (& a normal radiograph 4 months prior) shows a markedly enlarged cardiac silhouette. Echocardiography showed the LCA arising from the PA.* **(Right)** *Coronal projection of a CT angiogram demonstrates an anomalous LCA ⇨ originating from the underside of the PA trunk ➡, consistent with ALCAPA. The LV is markedly dilated.*

TERMINOLOGY

Synonyms

- Anomalous origin of left coronary artery from pulmonary artery (ALCAPA) = Bland-White-Garland (BWG) syndrome

Definitions

- ALCAPA: Most common congenital coronary artery anomaly presenting in children
  - Causes cardiac ischemia & infarction, depressed left ventricular (LV) systolic function, & significant mitral valve regurgitation

IMAGING

General Features

- Best diagnostic clue
  - Chest radiograph: Cardiomegaly in infant
  - Electrocardiogram (ECG): Deep Q waves in leads I & aVL, consistent with anterior lateral wall infarct
  - Echocardiogram: Abnormal left coronary artery (LCA) ostium arising from pulmonary trunk, retrograde LCA flow, right coronary artery (RCA) dilation, abundant intercoronary septal collaterals, significant mitral regurgitation, & depressed LV systolic function; ~ 90% diagnostic accuracy
  - CTA/MRA: Anomalous origin of LCA ostium; large tortuous coronary vessels with dilated RCA & significant collaterals; ~ 100% diagnostic accuracy

Radiographic Findings

- Radiography
  - Cardiomegaly, left atrial & LV enlargement, ↑ pulmonary vascularity or pulmonary edema (from depressed LV function & mitral regurgitation)

CT Findings

- Multidetector CTA demonstrates coronary artery anatomy & LV enlargement
  - Volume or surface 3D rendering from contrast-enhanced imaging assists with visualization

MR Findings

- T2WI
  - Myocardial edema in LV
- MRA
  - Anomalous LCA from pulmonary trunk, dilated RCA, significant intercoronary septal collaterals
- MR cine
  - Ventricular function quantified by summation of short-axis stack
  - Wall motion abnormalities assessed on 4-chamber & short-axis stacks, particularly LCA distribution
  - Ventricular chamber size quantified by end-diastolic volume
- Double IR FSE
  - Thin slice (4 mm) demonstrates excellent vessel anatomy: ALCAPA & dilated RCA
- Delayed enhancement
  - Myocardial late gadolinium enhancement (LGE) in LCA distribution consistent with infarcted or fibrotic segments: Focal with transmural or near transmural LGE

Echocardiographic Findings

- Abnormal origin of coronary artery recognized by retrograde flow from LCA into pulmonary artery, dilated RCA, & multiple collateral vessels
- Echocardiography demonstrates ↓ LV systolic function, segmental wall abnormalities in LCA distribution (typically representing anterolateral infarct), & significant mitral regurgitation

Angiographic Findings

- Invasive coronary angiography by cardiac catheterization: 100% diagnostic accuracy; performed only when echo & cross-sectional imaging not diagnostic
  - Pulmonary arteriogram demonstrates anomalous origin of LCA
  - Selective injection of RCA demonstrates dilated vessel & tortuous collateral vessels

Imaging Recommendations

- Best imaging tool
  - ECG in infancy: Infarct pattern with abnormal deep Q waves (particularly in leads I & aVL), ST segment depression, & T wave inversion
  - Chest radiograph: Cardiomegaly
  - Echocardiogram: ↓ LV systolic function, anomalous origin of coronary artery, collateral vessels, significant mitral regurgitation
  - CTA/MRA when echocardiogram not definitive
- Protocol advice
  - CTA performance best with ECG-synchronized gating (prospective), suspended respiration (if possible), high contrast injection rate (4-5 mL/sec), & contrast bolus timed or tracked to fill entire PA & aorta

DIFFERENTIAL DIAGNOSIS

Dilated Cardiomyopathy

- Most common cause of depressed LV function in infancy
  - Genetic/metabolic vs. viral origin
- Similar clinical presentation with irritability, failure to thrive, wheezing

Kawasaki Disease

- Vasculitis of medium-sized vessels leading to coronary artery ectasia or aneurysms; etiology unknown
- Myocarditis common; aneurysms occur in < 2%
- May develop stenosis associated with aneurysm → clot → infarction

Coronary Artery Fistula

- May produce cardiac ischemia if coronary steal leads to reduced blood flow to coronary circulation

Single Coronary Artery Origin

- 40% associated with other anomalies, including tetralogy of Fallot, transposition of great arteries, etc.
- Usually asymptomatic but occasional sudden cardiac death

PATHOLOGY

General Features

- Associated abnormalities

- Coronary artery anomalies can be isolated or associated with other defects; important to identify for surgical planning
  - Tetralogy of Fallot: Coronary anomalies include small conal branch (88%), large conal branch (6%), left anterior descending (LAD) from RCA (3%), dual LAD (2%), single RCA (0.3%), & single LCA (0.2%)
  - Transposition of great arteries: Coronary anomalies from aortic valve malposition include left circumflex from RCA (14%), single RCA (9%), LAD from RCA (6%), interarterial course of LCA or RCA (4%)
- Embryology
  - Abnormalities in signaling pathways or alterations in local factors that direct coronary development

Microscopic Features

- Signs of LV ischemia & varying degrees of reparative changes evident on autopsy or transplant
  - Anterolateral papillary muscle atrophic & scarred
  - Thinning & scarring of anterolateral LV wall & apex due to infarction

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - ~ 90% present in infancy with nonspecific symptoms of irritability, fussiness, wheezing, &/or failure to thrive
- Other signs/symptoms
  - Diaphoresis or ↓ oxygenation & perfusion (severe cyanosis or grayish appearance)
- Older child or adolescent
  - Usually asymptomatic until sudden catastrophic event occurs with syncope, dysrhythmic event, occasional death
    - May occur with exercise (suggesting coronary artery or other heart disease)

Demographics

- Gender
  - No sex predilection

Natural History & Prognosis

- Fetal life: Lungs collapsed; normal myocardial perfusion
- Stage 1: Newborn with high pulmonary vascular resistance (PVR)
  - Preferential flow into anomalous LCA (antegrade) from pulmonary artery → normal myocardial perfusion without symptoms or ischemia
- Stage 2: PVR ↓ ~ 2-4 months of life
  - ↓ PA diastolic pressure inadequate to provide forward flow to anomalous LCA to keep LV myocardium perfused
  - Collateral vessels develop from RCA → RCA dilation
  - Flow from RCA & collaterals meets high resistance of LV myocardial bed → preferential retrograde LCA flow into low-resistance PA
    - Retrograde flow of fully oxygenated blood into PA creates left-to-right shunt
- Later stages: Cardiac ischemia & infarct
  - Myocardial steal occurs with ↑ in collateral vessels
  - Ischemia & infarct develop in anterolateral distribution → global ventricular dilation & dysfunction plus significant mitral regurgitation (secondary to papillary muscle infarction)

Treatment

- Medical therapy to stabilize patient initially
  - Mechanical ventilation with oxygen to prevent hypoxia & treat cardiogenic shock
  - Sedation to ↓ myocardial oxygen demand
    - Cardiac anesthesia team involved due to ↑ potential for cardiac arrest or arrhythmia
- Surgical therapy: Up to 90% mortality if left unrepaired; prognosis related to degree of preoperative LV systolic dysfunction
  - LV assist device may be utilized preoperatively
  - Surgical techniques include
    - Direct transfer of LCA to aortic root: Button of tissue around LCA ostium transferred posteriorly to aorta
    - Bypass grafting: Proximal anomalous LCA ligated + bypass grafting to reestablish normal flow direction & tissue perfusion
    - Takeuchi repair: Intrapulmonary baffle from aorta to anomalous LCA origin
    - Simultaneous repair of mitral valve controversial as severe regurgitation may improve with revascularization
  - Surgical mortality ~ 5-10%, usually related to poor LV function
    - Complications include bleeding, cardiac arrest, heart failure, & stroke
    - Heart transplant warranted in cases where LV function does not improve after coronary artery surgical intervention

DIAGNOSTIC CHECKLIST

Consider

- ALCAPA in infant with abnormal ECG & unknown cause of LV systolic dysfunction
  - Readily diagnosed by imaging

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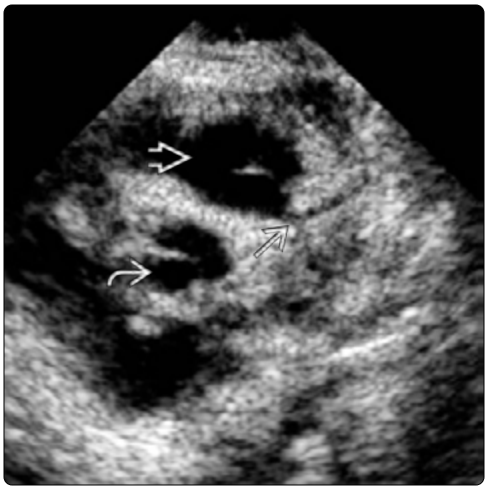
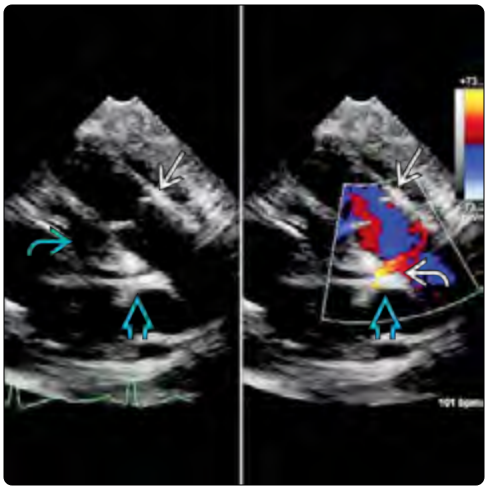
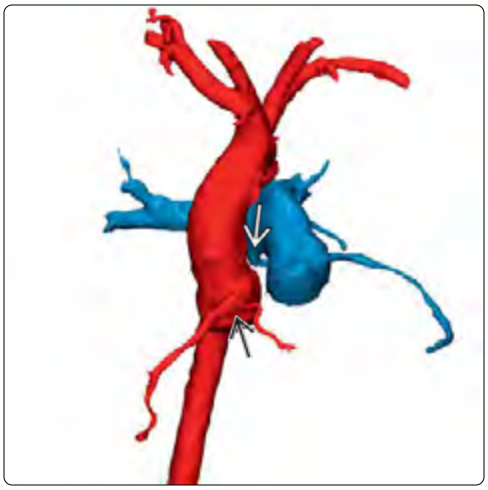
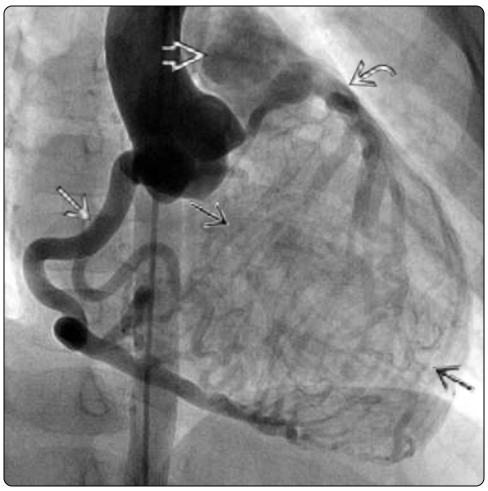


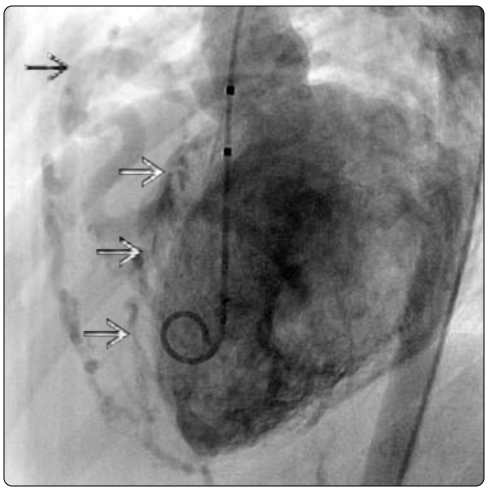


(Left) *Echocardiogram in an infant at the level of the aorta & main PA shows the LCA arising from the main PA. (Courtesy Michael Taylor, MD.)* **(Right)** *Echocardiogram demonstrates an ALCAPA with retrograde blood flow into the PA. While the LCA appears to arise from the aorta on grayscale imaging, the color Doppler image shows flow reversal of the LCA into the PA (demonstrated by a red jet toward the transducer). The ALCAPA was causing myocardial ischemia.*



(Left) *Anterior oblique projection of a 3D rendered CTA performed for 3D printing demonstrates a normal origin of the RCA (with acute marginal branch) from the aorta (red). An ALCAPA arises from the underside of the pulmonary trunk (blue).* **(Right)** *Sagittal view of the same 3D rendered CTA shows the ALCAPA arising from the underside of the pulmonary trunk (blue).*





(Left) *Anterior oblique conventional angiogram shows antegrade flow through a large RCA during an injection of the ascending aorta. Retrograde flow in the LCA (which is filled by multiple collateral vessels) opacifies the PA. The coronary arteries outline the enlarged LV.* **(Right)** *Lateral angiography after a left ventricular injection shows an enlarged right coronary supplying an arcade of collateral vessels in a teenager with ALCAPA.*

Double-Outlet Right Ventricle

KEY FACTS

TERMINOLOGY

- Double outlet right ventricle (DORV): Form of abnormal ventriculoarterial connection in which both great arteries arise completely or predominately from morphologic right ventricle
  - 16 variants based on relationship of great arteries & position/location of ventricular septal defect (VSD)
- May be part of complex congenital heart disease coexisting with ventricular anomalies, valve stenosis or atresia, abnormal atrioventricular valve, aortic valve anomalies, coarctation, coronary anomalies, & anomalies of systemic or pulmonary venous return

IMAGING

- Pulmonary flow dependent on site of VSD & degree of outflow or pulmonary valve stenosis
- Side-by-side relationship of aorta & pulmonary artery with aorta on right in 50-64%
- 3D imaging can help with complex intracardiac relationships for presurgical planning

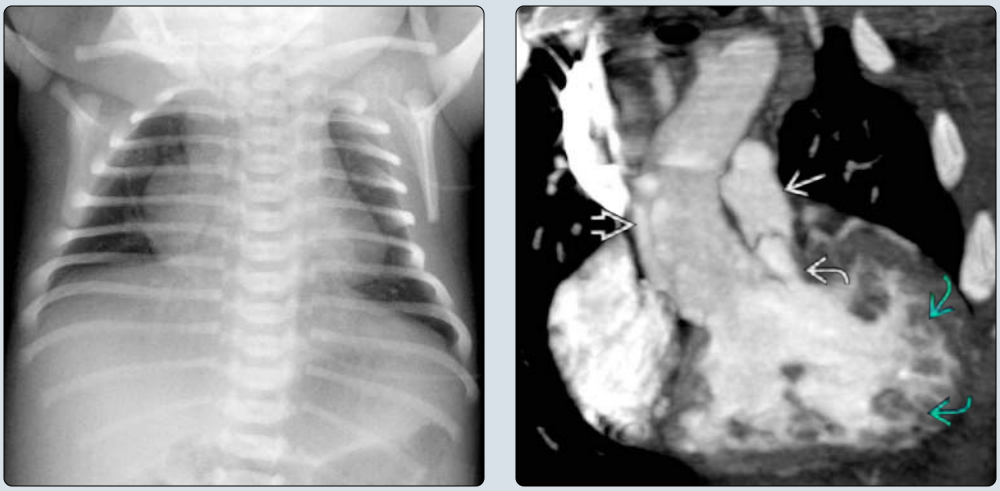
PATHOLOGY

- Conotruncal heart defect that may be of neural crest origin

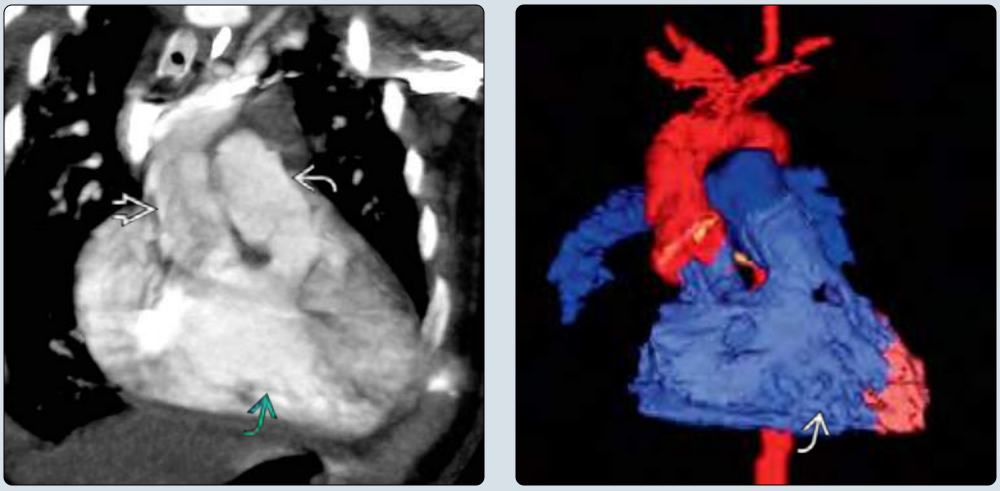
CLINICAL ISSUES

- May be diagnosed in utero; usually has clinical symptoms at birth or in 1st month
  - DORV with pulmonic stenosis: Cyanosis, failure to thrive, tachypnea
  - DORV with subaortic VSD without pulmonic stenosis: Symptoms of large left-to-right shunt & early evidence of pulmonary hypertension
- Surgery depends on anatomy
  - Closure of VSD & placement of right ventricle to pulmonary artery conduit
- Mortality rate after operation higher for complex lesions
- 15-year survival rate for noncomplex lesions: 85-90%

(Left) *AP chest radiograph in a patient with double outlet right ventricle (DORV) shows decreased pulmonary vascularity & mild cardiomegaly. Pulmonic stenosis in this patient makes the radiographic appearance similar to that of a patient with tetralogy of Fallot.* **(Right)** *Coronal MIP image from a cardiac CTA in a newborn with DORV shows both the pulmonary artery & aorta arising from the trabeculated right ventricle. Notice the subpulmonic stenosis of the right ventricular outflow tract.*



(Left) *Coronal MIP image from a cardiac CTA in a patient with DORV shows the aorta & pulmonary artery arising from the trabeculated right ventricle.* **(Right)** *Frontal view of a 3D cardiac CTA shows both the aorta (red) & pulmonary artery (blue) arising from the right ventricle (purple) in a patient with DORV. Note the prominent trabeculations of the right ventricle.*



TERMINOLOGY

Abbreviations

- Double outlet right ventricle (DORV)

Definitions

- Form of abnormal ventriculoarterial connection in which both great arteries arise completely or predominately from morphologic right ventricle
- 16 variants based on relationship of great arteries & position/location of ventricular septal defect (VSD)

IMAGING

General Features

- Best diagnostic clue
  - Radiologic appearance, regardless of modality, depends on physiology of lesion, which reflects variable anatomy of DORV
    - Careful assessment of segmental anatomy & relationships
    - Pulmonary flow dependent on site of VSD & degree of outflow or pulmonary valve stenosis

Radiographic Findings

- Radiography
  - DORV with subaortic VSD + severe pulmonic stenosis + normally related great vessels
    - ↓ pulmonary flow
    - Similar appearance to tetralogy of Fallot (TOF)
  - DORV without pulmonic stenosis but with normally related great vessels
    - ↑ pulmonary flow
    - Appears similar to large VSD

CTA or MRA

- Shows relationships of great vessels, position of VSD, relative size of ventricles, & associated anomalies
  - Altered ventricular-great vessel relationship
  - Aortic valve & pulmonic valve may be at same level
  - Demonstrates situs abnormalities & associated extracardiac anomalies such as coarctation, anomalies of systemic venous connections, & anomalies of pulmonary venous connections
- 3D imaging can help with complex intracardiac relationships for presurgical planning
- Postoperative assessment in older patients useful for anatomy & functional assessment

Angiographic Findings

- Opacification of both great arteries during right ventricular injection
- Filling defect dividing 2 outflow tracts or double conus
- Aortic & pulmonary valves may be on same horizontal plane
- ± malposition of aorta

DIFFERENTIAL DIAGNOSIS

Ventricular Septal Defect

- Distinguishing feature is normal fibrous continuity between posterior leaflet of aortic valve & anterior leaflet of mitral valve

Tetralogy of Fallot

- Physiology may be similar to DORV, but TOF has aortic-mitral continuity despite anterior position of aorta & overriding of right ventricle

Transposition of Great Arteries

- D-transposition of great arteries shows atrioventricular concordance & ventriculoarterial discordance (e.g., aorta arises from right ventricle & pulmonary artery arises from left ventricle)

PATHOLOGY

General Features

- Etiology
  - Conotruncal heart defect that may be of neural crest origin
    - Neural crest involved in development of cardiac septum
  - Occurs during looping of bulboventricular tube

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - May be diagnosed in utero; usually have clinical symptoms at birth or in 1st month
    - DORV with pulmonic stenosis: Cyanosis, failure to thrive, tachypnea
    - DORV with subaortic VSD without pulmonic stenosis: Symptoms of large left-to-right shunt & early evidence of pulmonary hypertension

Demographics

- DORV accounts for 1.0-1.5% of all congenital heart disease with incidence of 1 per 10,000 live births
- No race or sex predilection

Treatment

- Surgery depends on anatomy
  - Closure of VSD & placement of right ventricle to pulmonary artery conduit if 2 developed ventricles
  - Norwood/Fontan procedure if there is hypoplasia of ventricle
- Survival statistics depend on specific type of DORV
  - Mortality rate after operation higher for complex lesions
  - 15-year survival rate for noncomplex lesions: 85-90%

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Aortic Coarctation

KEY FACTS

TERMINOLOGY

- Narrowing of aortic lumen with obstruction to blood flow

IMAGING

- Locations: Preductal, typically hypoplastic (infantile); juxtaductal or postductal, typically focal (adult); abdominal, middle aortic syndrome (rare)
  - May have diffuse hypoplasia of aortic isthmus + focal coarctation (important for surgical planning)
- Can be simple (isolated coarctation in adult) or complex (additional cardiac anomalies, presenting in infancy)
- Classic radiographic findings
  - Poststenotic dilatation of proximal descending aorta (figure 3 sign)
  - Rib notching (age > 5 years)
  - Left ventricular hypertrophy: Rounded cardiac apex
- Echocardiography for primary diagnosis in infancy

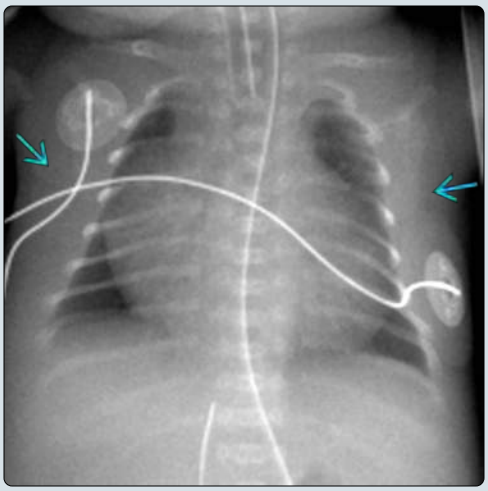
PATHOLOGY

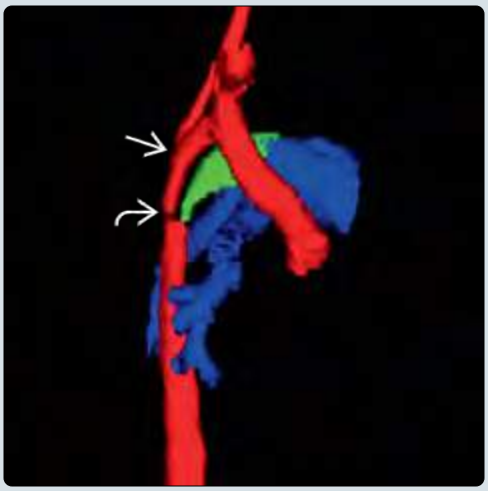
- Most common additional cardiac anomalies: Ventricular septal defect (33%), PDA (66%), bicuspid aortic valve (50%)
- Turner syndrome: 20-36% have coarctation

CLINICAL ISSUES

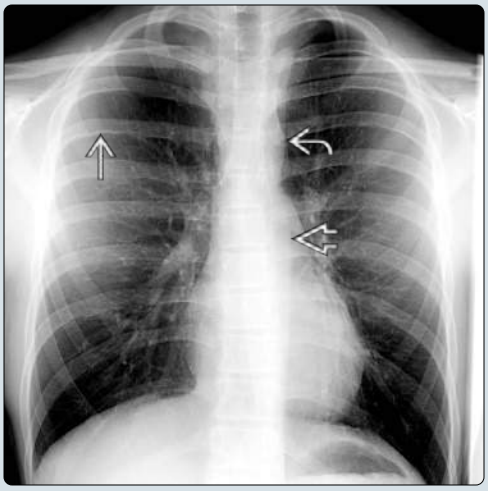
- Presentations include
  - Infancy: Congestive heart failure (due to aortic arch interruption, associated anomalies)
  - Older child, adult: Hypertension, diminished femoral pulses, differential blood pressure between upper & lower extremities (arm-leg gradient)
- Treatment: Resection + end-to-end anastomosis, interposition graft, patch + aortoplasty, balloon angioplasty
- Complications: Recoarctation (< 3%; ↑ if operation in infancy), postoperative aneurysms (24% after patch aortoplasty)
- ↓ long-term survival (late hypertension, coronary artery disease)

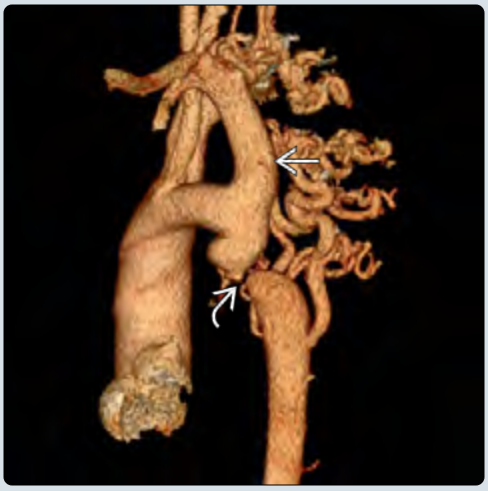
(Left) *Frontal chest radiograph in a 2-week-old infant with congestive heart failure & severe coarctation of the aorta shows cardiomegaly & anasarca.* **(Right)** *Lateral oblique view of a 3D color-coded cardiac CTA shows a preductal hypoplastic-type coarctation of the aorta with a periductal focal coarctation. Notice that the patent ductus arteriosus (PDA) (green) is beginning to close with severe narrowing near the aortic connection.*





(Left) *Frontal chest radiograph in a teenage patient with coarctation of the aorta shows prominence of the aortic knob & descending aorta, creating a figure 3 sign. Note the sclerosis & undulation of the undersurface of the ribs from collateral vessels causing rib notching.* **(Right)** *Lateral oblique 3D image from a cardiac CTA shows a focal coarctation of the aorta with the formation of multiple large arterial collaterals. Note the massive enlargement of the left subclavian artery.*





TERMINOLOGY

Definitions

- Narrowing of aortic lumen with obstruction to blood flow
- Category of congenital heart disease: Acyanotic, normal heart size (most commonly), normal pulmonary vascularity
- Hemodynamics: Left ventricular (LV) pressure overload

IMAGING

General Features

- Best diagnostic clue
  - Focal or diffuse aortic narrowing with presence of collaterals in older patients
- Location
  - Preductal, typically hypoplastic (infantile)
  - Juxtaductal or postductal, typically focal (adult)
  - Abdominal, middle aortic syndrome (rare)
    - Should be specifically looked for in patients with differential blood pressures + normal thoracic aorta
- Morphology
  - Simple (isolated coarctation in adults) or complex (associated with other cardiac anomalies, presenting in infancy)

Radiographic Findings

- Poststenotic dilation of proximal descending aorta (figure 3 sign)
- Rib notching (age > 5 years) or undulation & sclerosis of undersurface of ribs due to collaterals
- LV hypertrophy: Rounded cardiac apex

Fluoroscopic Findings

- Esophagram: Impression by dilated descending aorta (reverse figure 3 sign)

CT Findings

- CTA
  - Cardiac CTA with prospective ECG gating increasingly used in diagnosis
    - Advantages: Speed of exam, anatomic detail (coronaries, collaterals)
    - Disadvantages: Radiation dose, no hemodynamic data
  - Depicts coarctation site, percentage of stenosis, & presence/location of collaterals

MR Findings

- T1WI
  - "Black blood" imaging (cardiac-gated spin-echo or double-inversion recovery)
    - Sagittal oblique ("candy cane") plane through aortic arch shows location of coarctation
    - Perpendicular views for cross-sectional diameter measurements
- T2* GRE
  - "Bright blood" imaging (cardiac-gated steady-state free-precession cine MR)
    - More reliable for diameters than "black blood" imaging
    - Sagittal oblique plane for anatomy
    - Length of systolic (dark) flow jet correlates with hemodynamic significance
    - ± aortic regurgitation (bicuspid aortic valve)
- MRA
  - Velocity-encoded phase-contrast MRA: For estimate of gradient, collateral flow, & aortic valve regurgitation fraction
  - 3D gadolinium-enhanced MRA: For anatomy & depiction of collaterals
- Phase-contrast MR technique: Alternative method to obtain velocities to calculate gradient across coarctation
  - Multiple MR techniques now available to calculate pressure gradients

Ultrasonographic Findings

- Pulsed Doppler
  - Aortic & visceral arterial waveforms distal to coarctation may show parvus et tardus morphology
    - Slowly rising & diminished systolic upstroke

Echocardiographic Findings

- Echocardiogram
  - Imaging of aortic arch & branches in suprasternal long-axis view
  - Relationship of coarctation with patent ductus arteriosus (PDA)
  - Notch in descending aorta at level of coarctation (shelf sign)
- Pulsed Doppler
  - Estimate of gradient across coarctation

Angiographic Findings

- Cardiac catheterization: Direct measurement of gradient
- Intervention: Balloon angioplasty

Imaging Recommendations

- Protocol advice
  - Echocardiography for primary diagnosis in infancy
  - Cardiac CTA with prospective ECG gating in infancy for complex anatomy
  - MR in older children for preoperative work-up & postoperative surveillance for recoarctation & aneurysms
  - Cardiac catheterization reserved for gradient measurement & intervention

DIFFERENTIAL DIAGNOSIS

Hypoplastic Left Heart Syndrome

- Congestive heart failure in newborn
- Hypoplastic LV
- Ductus-dependent systemic perfusion
- Retrograde flow in hypoplastic ascending aorta

Interrupted Aortic Arch

- Flow to descending aorta via PDA

Pseudocoarctation

- Elongation & kinking of aorta without obstruction/gradient

Takayasu Arteritis

- Acquired inflammatory condition
- Acute phase: Aortic wall enhancement
- Chronic phase: Narrowing/occlusion of aorta & branch vessels

PATHOLOGY

General Features

- Etiology
  - 2 developmental theories
    - Abnormal fetal hemodynamics [when associated with cardiac lesions that ↓ LV output & flow through aortic isthmus, such as hypoplastic left heart syndrome or large ventricular septal defect (VSD)] can lead to preductal coarctation & diffuse hypoplasia of isthmus
    - Postnatal contraction of fibrous ductal tissue in aortic wall at time of PDA closure
- Genetics
  - Usually sporadic
- Associated abnormalities
  - Cardiac: VSD (33%), PDA (66%), bicuspid aortic valve (50%), transposition, subaortic & mitral stenosis ("parachute" deformity: Shone syndrome), Taussig-Bing anomaly, endocardial fibroelastosis
  - Associated with Turner syndrome (20-36% have coarctation)
  - Berry aneurysms of circle of Willis
  - Scoliosis (in boys)
  - Abdominal coarctation associated with neurofibromatosis, Williams syndrome, Alagille syndrome, fibromuscular dysplasia, mucopolysaccharidosis, fetal alcohol syndrome, & arteritis
- Pathophysiology
  - ↑ in systemic vascular resistance → ↑ in LV afterload → LV hypertrophy
  - Hypertension due to renal hypoperfusion
  - Congestive heart failure (newborn, associated complex heart disease)
  - Arterial collaterals develop to bypass stenosis
    - Internal mammary, intercostal, paravertebral, epigastric

Gross Pathologic & Surgical Features

- Focal shelf or waist lesion
- Diffuse narrowing of aortic isthmus
- Poststenotic dilatation of descending aorta

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Frequently asymptomatic, incidentally found
  - Infancy: Congestive heart failure (due to aortic arch interruption, associated anomalies)
  - Older child, adult: Hypertension, ↓ femoral pulses, differential blood pressure between upper & lower extremities (arm-leg gradient)
- Other signs/symptoms
  - Bacterial endocarditis

Demographics

- Epidemiology
  - Incidence: 2-6 per 10,000 live births
  - More common in male patients (2:1), Caucasians

Natural History & Prognosis

- ↓ long-term survival (late hypertension, coronary artery disease)

Treatment

- Resection & end-to-end anastomosis
- Interposition graft
- Prosthetic patch, subclavian flap aortoplasty
- Balloon angioplasty
- Complications
  - Recoarctation (< 3% but higher when operation occurs in infancy)
  - Postoperative aneurysms (24% after patch aortoplasty)

DIAGNOSTIC CHECKLIST

Consider

- Diffuse hypoplasia of aortic isthmus in addition to focal coarctation (important for surgical planning)

Image Interpretation Pearls

- Rib notching in asymptomatic individuals can be 1st clue to hemodynamically significant coarctation

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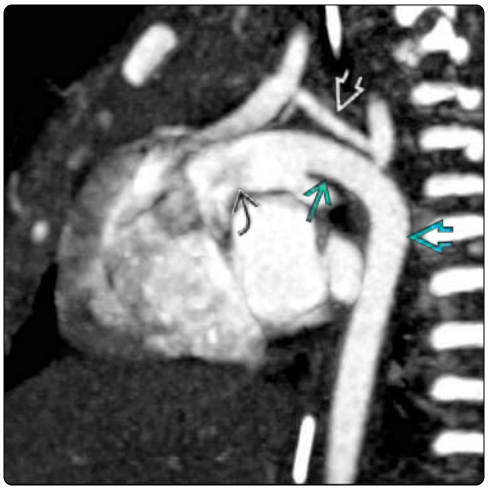
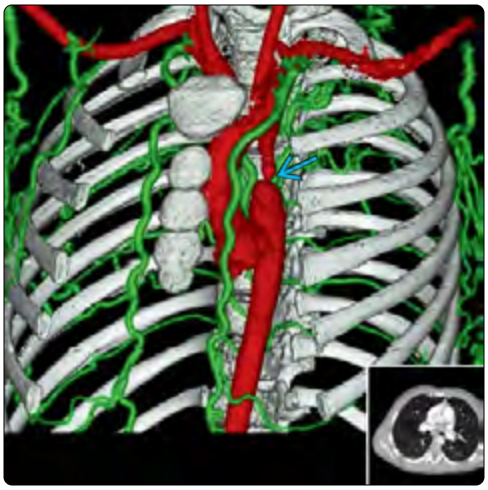
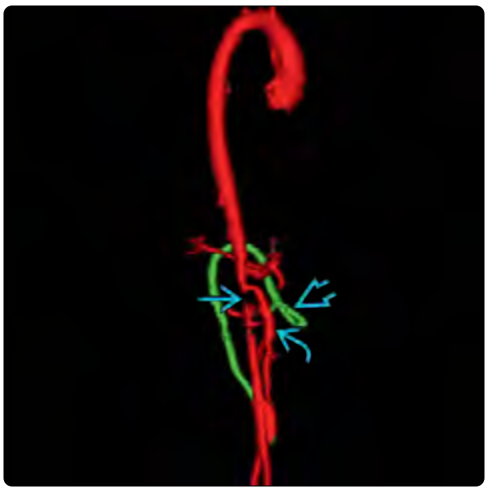
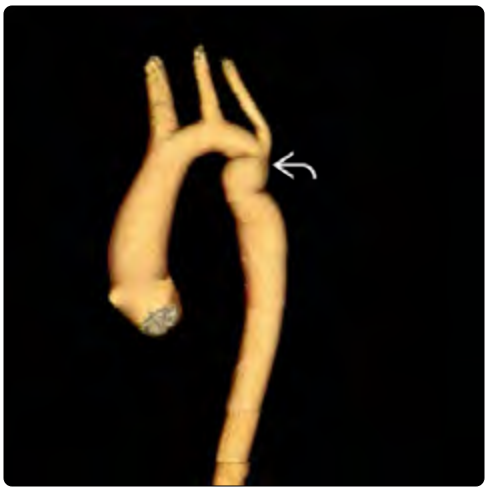


(Left) *Sagittal oblique cardiac CTA in a newborn shows a hypoplastic-type coarctation of the aorta with a large PDA connecting the main pulmonary artery to the descending aorta.* **(Right)** *Lateral oblique view of a 3D color-coded model from a cardiac CTA shows a hypoplastic-type coarctation of the aorta (red) with a large PDA (green) connecting the pulmonary arteries (blue) to the descending aorta.*



(Left) *Frontal oblique view from a 3D color-coded CTA of the chest shows a focal coarctation of the aorta with prominent arterial collaterals (green) circumventing the obstruction.* **(Right)** *A lateral image from a chest CTA 3D color-coded model shows an aortic vascular stent (white) at the origin of the left subclavian artery. A large graft (green) has been placed between the ascending & descending aorta to improve flow.*





(Left) *Oblique 3D aortogram from an MRA shows focal obstruction of the midabdominal aorta with a dilated superior mesenteric artery & a large collateral (green) circumventing the obstruction.* **(Right)** *Lateral oblique 3D image from a CTA shows a tortuous but normal-caliber portion of the descending aortic arch. No collaterals were seen, & no significant gradient was detected in this patient with pseudocoarctation of the aorta.*

Aortic Stenosis

KEY FACTS

TERMINOLOGY

- Spectrum of aortic valve abnormalities that ranges from asymptomatic bicuspid aortic valve to thickened & obstructed aortic valve stenosis to severe neonatal aortic atresia & hypoplastic left heart syndrome
- Aortic stenosis (AS) may be valvar, supravalvar, or subvalvar
  - Valvar stenosis most common at 80%

IMAGING

- Varies by location, etiology, & severity of stenosis
- Chest radiographs range from cardiomegaly & edema in severely affected infants to normal in adolescents
- Poststenotic dilation of ascending aorta in valvar stenosis
  - Due to flow jet through stenotic valve
- Supravalvar shows hourglass shape of ascending aorta
- Subaortic stenosis may have hypertrophic cardiomyopathy
- Cardiac enlargement may not be seen in childhood
- MR & echo allow quantitative assessments
- Cardiac catheterization for interventional treatment with balloon valvotomy, leads to aortic regurgitation

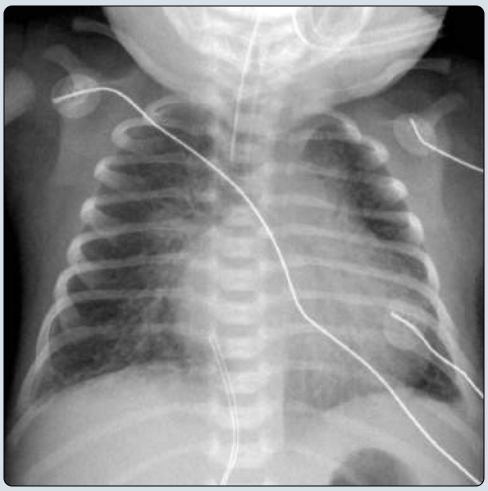
PATHOLOGY

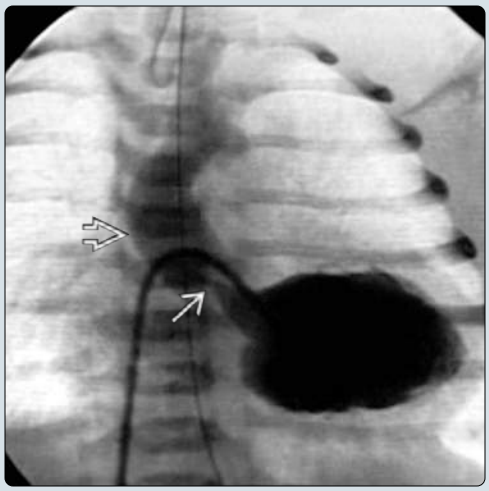
- Grading of AS: Jet velocity, gradient across valve, valve area
  - Mild has gradient < 20 mmHg
  - Moderate has gradient from 20-40 mmHg
  - Severe has gradient > 40 mmHg
- Associations include Williams syndrome, hypoplastic left heart syndrome, bicuspid aortic valve, hypertrophic cardiomyopathy, endocarditis

CLINICAL ISSUES

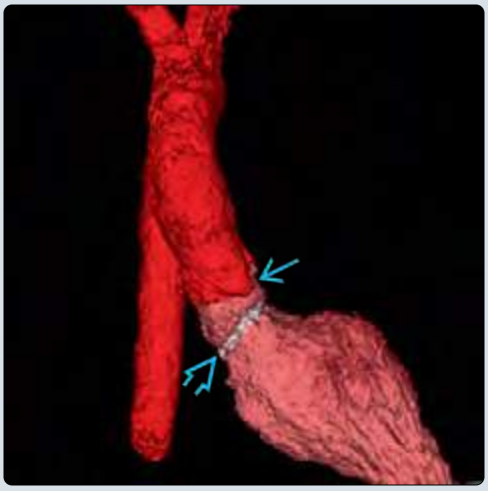
- 10-20% of AS presents in 1st year of life
- Neonatal: Signs of poor or low cardiac output with tachypnea & feeding problems
- Childhood: Usually asymptomatic but may have systolic murmur or suprasternal thrill
- 1% of sudden deaths may be related to undetected AS

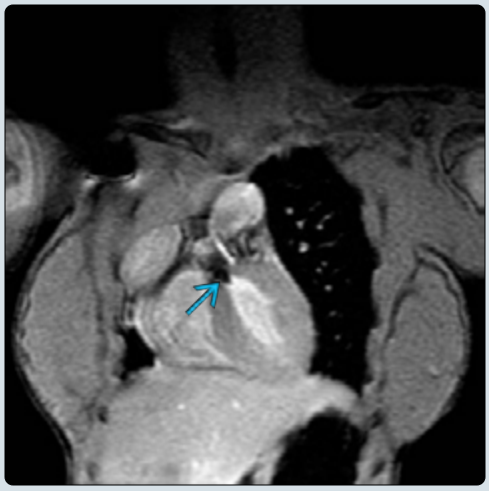
(Left) *Frontal chest radiograph shows a 2-week-old infant in heart failure with perihilar pulmonary edema & cardiomegaly. This patient had critical aortic stenosis.* **(Right)** *Single frontal image from a left ventricular angiogram shows a jet across the aortic valve ⇨ from severe aortic stenosis in a 2-week-old infant. Notice the dilation of the ascending aorta ⇨.*





(Left) *Oblique 3D view from a cardiac CTA shows a subvalvular membrane (white) causing narrowing of the left ventricular outflow tract (LVOT). Notice that the membrane ⇨ is well below the aortic valve ⇨.* **(Right)** *Oblique coronal bright blood GRE cine cardiac MR along the LVOT in a patient with a subvalvular membrane shows dephasing signal void artifact ⇨ from stenosis below the aortic valve.*





TERMINOLOGY

Abbreviations

- Aortic stenosis (AS)

Synonyms

- Aortic valvar stenosis, aortic valvular stenosis

Definitions

- Spectrum of aortic valve abnormalities that ranges from asymptomatic bicuspid aortic valve to thickened, obstructed aortic valve stenosis to severe neonatal aortic atresia & hypoplastic left heart syndrome
- AS may be valvar, supravalvar, or subvalvar (subaortic)
  - Valvar stenosis most common at 80%

IMAGING

General Features

- Best diagnostic clue
  - Thickened valve leaflets with fusion
    - Bicuspid aortic valve in 80% of valvar AS
    - Unicuspid aortic valve seen in hypoplastic left heart with endocardial fibroelastosis
  - High-velocity jet of blood ejected from left ventricle during systole
  - Poststenotic dilation of ascending aorta
  - Concentric left ventricular hypertrophy
- Location
  - Stenosis may be subvalvar, valvar (most common), or supravalvar
- Size
  - Valve annulus may be small for age; valve leaflets usually thickened; commissures may be fused
- Morphology
  - Normal valve tricuspid & mobile; stenotic valve usually thickened with restricted systolic motion

Radiographic Findings

- Neonates & infants may have normal chest radiograph vs. mild cardiomegaly & edema
- Children & adolescents may have normal heart size even in severe AS
  - Dilation of ascending aorta
  - Left ventricle may enlarge secondary to aortic regurgitation (AR) following balloon valvotomy for treatment
  - Ca^{2+} of valve rare in childhood

CT Findings

- CTA
  - Used to evaluate complex anatomy
  - Can provide volumetric functional data in hypoplastic left heart patients with AS
  - Supravalvar stenosis (as seen in Williams syndrome) shows concentric narrowing of ascending aorta (hourglass shape)
    - ± coarctation or pulmonary artery stenoses

MR Findings

- MR cine
  - Ventricular function can be qualitatively assessed on 4-chamber view & quantitatively assessed on stack of short-axis views
  - Stenosis & regurgitation evaluation
    - Flow jet: Signal loss caused by high-velocity flow & turbulence
    - Valve motion: Abnormal motion of stenotic valve evaluated in plane parallel to annulus
    - Secondary changes in chamber size & degree of wall thickening can be measured
- Velocity-encoded (VENC) or phase contrast MR
  - Allows quantification of transvalvular pressure gradient & valve area
  - Can quantify regurgitant fraction
  - Flow velocity maps across valve can be generated
- Delayed enhancement
  - Look for infarction or fibrosis both pre- & postablation in subaortic stenosis obstructive cardiomyopathy patients

Echocardiographic Findings

- Gold standard for making diagnosis in infants; used to assess
  - Aortic jet velocity, gradient across valve, valve area
  - Left ventricular function, aortic valve thickness & motion, & associated anomalies such as mitral insufficiency
- Doppler echocardiography
  - Systolic high-velocity flow jet in left ventricle outflow tract

Angiographic Findings

- Cardiac catheterization for interventional treatment with balloon valvotomy
- Findings include thickened aortic valve, doming of aortic valve, systolic flow jet into ascending aorta, enlarged ascending aorta, & thickened left ventricle

DIFFERENTIAL DIAGNOSIS

Rheumatic Heart Disease

- Multisystem disease with fever, rash, carditis, & valvular disease

Marfan Disease

- Connective tissue disorder associated with aneurysmal dilation of ascending aorta

Coarctation of Aorta

- May have dilation of aorta proximal to narrowing
- 75% may have associated bicuspid valve

Systemic Hypertension

- Left ventricular hypertrophy & prominent ascending aorta, typically in older individuals

PATHOLOGY

General Features

- Genetics
  - Bicuspid aortic valve one of most common congenital malformations
  - Williams syndrome due to chromosomal abnormality of 7q11.2; autosomal dominant
- Associated abnormalities

- Endocarditis occurs in 4%
- Williams syndrome: Associated with supravalvar AS & pulmonary artery stenosis
- Coarctation of aorta: Associated with bicuspid aortic valve & AS
- Hypoplastic left heart syndrome

Staging, Grading, & Classification

- Grading of AS incorporates aortic jet velocity, valve gradient, valve area
  - Mild has gradient ≤ 20 mmHg
  - Moderate has gradient from 20-40 mmHg
  - Severe has gradient > 40 mmHg
- Classification of AS
  - Subvalvar (left ventricular outflow tract) AS (or subaortic stenosis) due to
    - Membrane that partially obstructs left ventricular outflow
    - Thickening of interventricular septum in patients with obstructive cardiomyopathy
  - Valvar in 80% of cases, most frequently in association with bicuspid aortic valve
  - Supravalvar shows concentric narrowing in ascending aorta
    - May be isolated or associated with Williams syndrome

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Neonatal: Signs of poor or low cardiac output with tachypnea & feeding problems
  - Childhood: Usually asymptomatic but may have systolic murmur or suprasternal thrill
    - If prior valvotomy, there will be dilation of ascending aorta & large left ventricle secondary to AR
    - Sudden death, which usually occurs during exercise
    - Children with Williams syndrome recognized due to multiple manifestations of disease

Demographics

- Age
  - 10-20% in 1st year of life
- Gender
  - M:F = 4:1
- Epidemiology
  - Occurs in 3-5% of children with congenital cardiac defects

Natural History & Prognosis

- Infants with critical AS may be diagnosed in utero
  - Mortality relates to degree of stenosis, presence of neonatal symptoms, & lower birth weights
    - Hypoplastic left heart syndrome has highest mortality
    - Endocardial fibroelastosis with decreased left ventricular outflow
    - Coronary blood flow to subendocardium reduced, ischemia may occur
- Children with AS constitute 3-5% of all congenital heart defects
  - Usually stenosis progresses; 20% have associated lesions
  - 1% of sudden deaths thought to be related to undetected AS
  - Once balloon valvotomy occurs, children will have both residual AS & AR

Treatment

- In infants, AS may be part of spectrum of hypoplastic left heart syndrome
  - Treated with prostaglandin to maintain ductal patency
  - May need balloon atrial septostomy or Rashkind procedure with staged Norwood procedure
  - Occasionally treated with heart transplant
- In infants with "critical" AS
  - Percutaneous balloon valvotomy urgently (but may result in AR)
  - Ross surgical procedure performed when valvotomy inadequate or regurgitation moderate
    - Native pulmonary valve placed in aortic position; homograph placed for pulmonary valve
- In children, depends on degree of obstruction & progression
  - Mild stenosis: Monitored with echocardiogram & EKG
  - Moderate stenosis: May require valvotomy
  - Severe stenosis: Valvotomy required
- Surgical aortic valvotomy
  - Performed for supravalvar AS, resection of subaortic membrane, & enlargement of aortic annulus
  - Mechanical valves usually need anticoagulation (undesirable for normal children)
- Replacement of aortic valve with pulmonary valve (Ross procedure)
  - Pulmonary valve replaced with homograft or reconstruction with conduit
  - Indicated in peak-to-peak gradients > 70 mm & where valvotomy has failed
- All children need prophylaxis to prevent bacterial endocarditis

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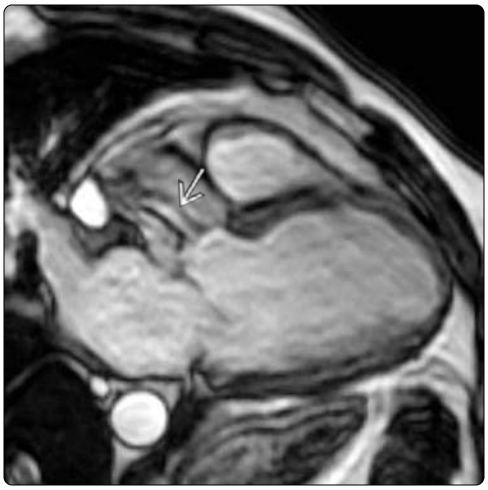
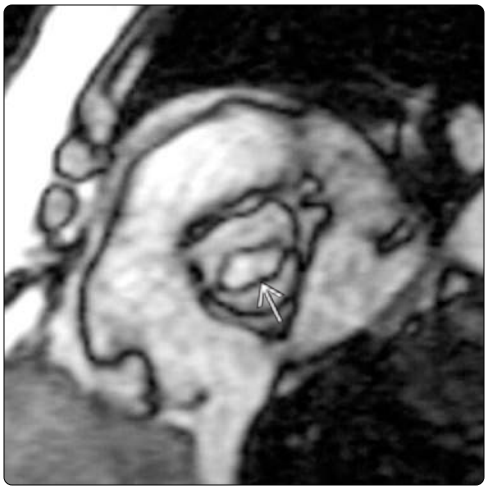
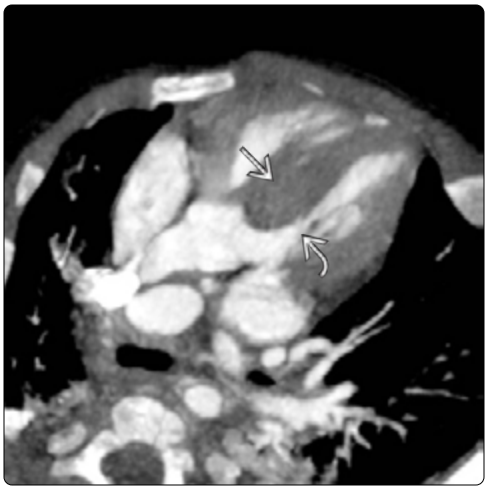
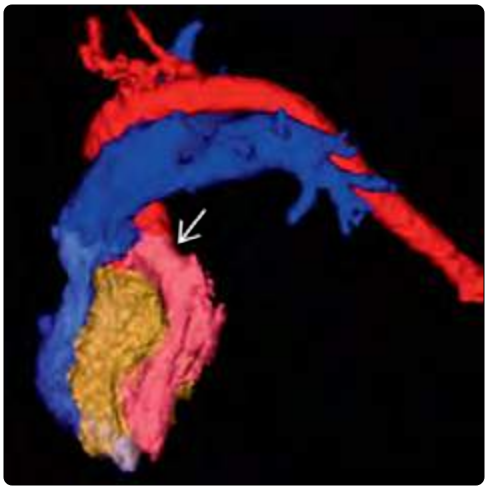


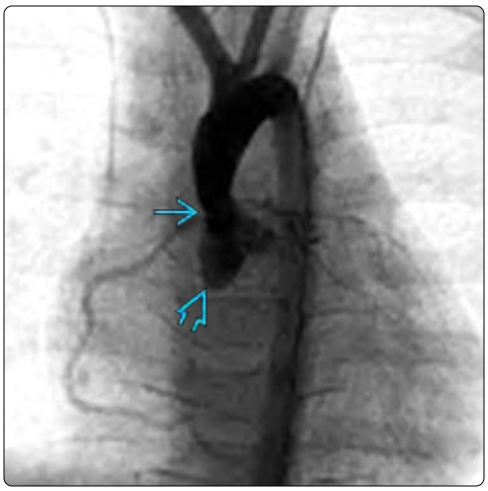


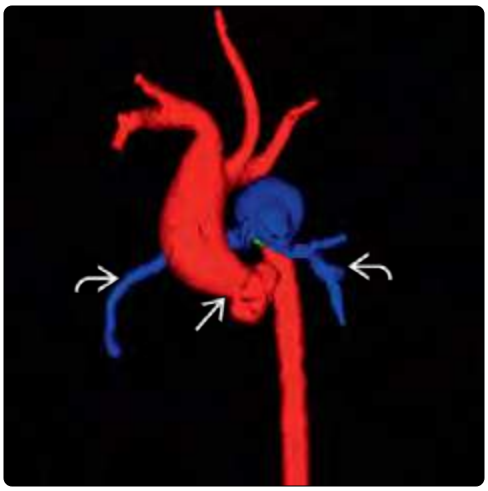
(Left) *Three-chamber view from a bright blood GRE cine cardiac MR shows a linear signal void ➡ from elevated velocity flow across a stenotic aortic valve.* **(Right)** *Cross-sectional view through the aortic valve from a bright blood GRE cine cardiac MR shows a bicuspid aortic valve ➡.*





(Left) *Axial MIP from a cardiac CTA in an infant shows marked thickening of the interventricular septum ➡ & narrowing of the LVOT ➡ in this patient with idiopathic hypertrophic subaortic stenosis (IHSS).* **(Right)** *Oblique view from a 3D color-coded cardiac CTA shows narrowing of the LVOT ➡ with severe contouring of the left ventricle (pink) from the thickened interventricular septum (gold) in this patient with IHSS.*





(Left) *Frontal view from an aortogram shows a supravalvular aortic stenosis ➡ with a typical hourglass shape of the ascending aorta in a patient with Williams syndrome. Note the dilated sinuses of Valsalva ➡.* **(Right)** *Frontal image from a 3D color-coded cardiac CTA shows supravalvular aortic stenosis ➡ in a patient with Williams syndrome. Notice the hypoplastic pulmonary arteries ➡, which are often seen in this syndrome.*

Pulmonary Artery Stenosis

KEY FACTS

TERMINOLOGY

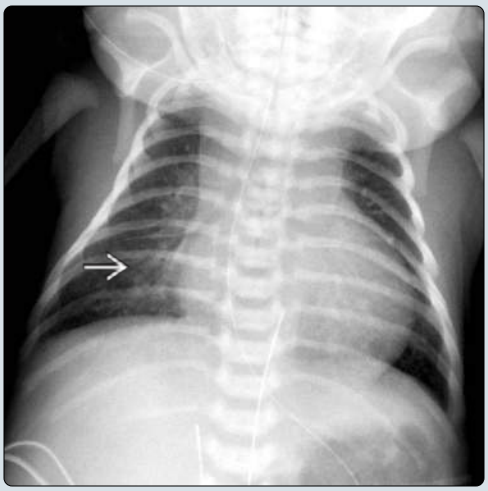
- Stenosis at level of infundibulum, pulmonary valve, supravalvar main pulmonary artery (PA), or branches of PA
- Pulmonary valvar stenosis most common (> 90%)

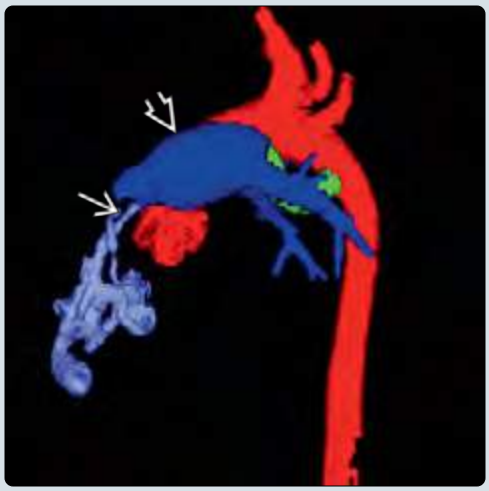
IMAGING

- Valvar stenosis: Normal heart size with dilated main pulmonary artery segment in ~ 80%
  - Thickened valve leaflets, doming of valve, systolic high-velocity flow jet in pulmonary outflow tract
- Supravalvar PA stenosis (PS) in Williams syndrome
- Alagille syndrome has valvar PS & peripheral PA stenosis
- Infundibular narrowing in tetralogy of Fallot & complex malformations
- Right ventricular hypertrophy occurs secondary to ↑ work

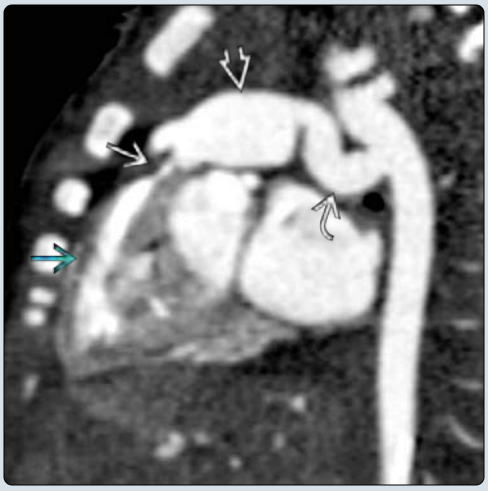
CLINICAL ISSUES

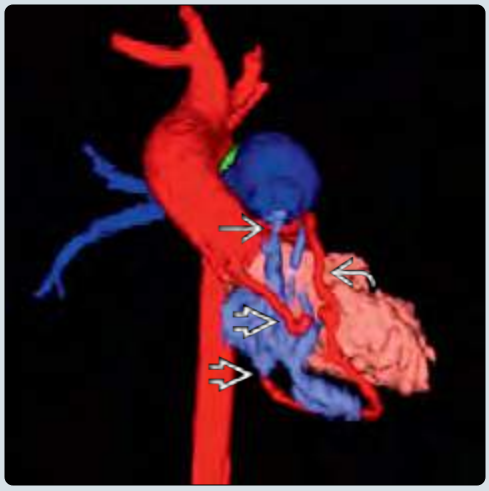
- PS often diagnosed between 2-6 years of age during routine physical exam
- Mild stenosis: Usually asymptomatic with systolic ejection murmur
- Moderate stenosis: Exertional dyspnea, easy fatigability
- Severe stenosis: Infants may present with severe cyanosis
- Balloon valvuloplasty is treatment of choice for moderate to severe gradients > 50 mmHg
  - Newborns with critical PS may need immediate valvotomy
    - Prostaglandin to maintain ductus arteriosus flow
    - May need surgical valvotomy or palliative Blalock-Taussig shunt
  - Not as effective in dysplastic valves (e.g., Noonan syndrome), which may require surgery





(Left) *Frontal radiograph shows mild cardiomegaly & perihilar pulmonary edema in a patient with severe pulmonic stenosis. Notice how interstitial edema makes the pulmonary vessels indistinct. (Right) Lateral image from a 3D color-coded cardiac CTA shows severe pulmonic stenosis with poststenotic dilation of the main pulmonary artery. Notice the large PDA (green) extending from the aorta (red) to the pulmonary artery (blue).*





(Left) *Sagittal MIP from a cardiac CTA shows severe pulmonic stenosis with poststenotic dilation of the main pulmonary artery & right ventricular (RV) hypertrophy. Notice the tortuous PDA extending from the descending aorta to the main pulmonary artery. (Right) Frontal view from a 3D color-coded cardiac CTA shows severe pulmonic stenosis with a hypoplastic RV (purple). Notice that the coronary arteries communicate with the RV, consistent with coronary artery fistulas.*

TERMINOLOGY

Abbreviations

- Pulmonary stenosis (PS)

Definitions

- Stenosis at level of infundibulum, pulmonary valve, supravalvar main pulmonary artery, or branches of pulmonary artery
- Pulmonary valvar stenosis most common (> 90%)

IMAGING

General Features

- Best diagnostic clue
  - Normal heart size & prominent main pulmonary artery segment
  - Left pulmonary artery occasionally larger than right
  - Normal pulmonary flow
- Location
  - Infundibulum, pulmonary valve, supravalvar main pulmonary artery, &/or pulmonary artery branches
- Size
  - Orifice of valve reduced, which creates turbulence & ↑ work of right ventricle
- Morphology
  - Thickened & stenotic pulmonary valve most common
    - Valvar stenosis in > 90%
    - Poststenotic dilation of pulmonary artery frequently seen
  - Dysplastic pulmonary valve seen in Noonan syndrome
  - Supravalvar PS in Williams syndrome
    - Also have supravalvar aortic stenosis
  - Infundibular narrowing seen in tetralogy of Fallot & complex malformations
  - Pulmonary artery branch stenosis seen in tetralogy of Fallot
  - Alagille syndrome has PS & peripheral pulmonary artery stenosis
  - Congenital rubella may have pulmonary branch stenosis
  - Right ventricular hypertrophy occurs secondary to ↑ work

Radiographic Findings

- Normal heart size with dilated main pulmonary artery segment in ~ 80%

CT Findings

- CTA
  - Most useful for supravalvar PS or peripheral PS

MR Findings

- Assessment of right ventricular function
- Can quantitate degree of pulmonary insufficiency
- MRA useful for supravalvar & peripheral PS

Echocardiographic Findings

- Echocardiogram
  - Pulmonary valvar stenosis
    - Fusion of valve commissures
    - Thickened valve with systolic restricted motion & doming
    - Poststenotic dilation of pulmonary artery
    - Right ventricular hypertrophy
- Pulsed Doppler
  - Systolic high-velocity flow jet in pulmonary outflow tract
  - Can accurately determine velocity of flow, which can help predict pressure gradients

Angiographic Findings

- Conventional
  - Cardiac catheterization utilized to measure pressures
    - Demonstrates degree of outflow tract obstruction
    - Degree of trabeculation of right ventricle relates to degree of obstruction
  - Cardiac catheterization utilized for treatment with balloon valvotomy

Imaging Recommendations

- Best imaging tool
  - Diagnosis in infancy by echocardiography
  - Cardiac catheterization done as part of therapeutic intervention
    - Shows thickened valve leaflets, doming of valve, & poststenotic dilation

DIFFERENTIAL DIAGNOSIS

Normal Chest Radiograph in Adolescent

- Main pulmonary artery is prominent normally
- Occasionally confused with dilation of pulmonary artery
- Patients have no murmur or symptoms

Pulmonary Hypertension

- Heart may have normal size with right ventricular enlargement
- Central pulmonary arteries is large

Congenital Heart Disease With Large Left-to-Right Shunt

- Main pulmonary artery is large with ↑ pulmonary flow

PATHOLOGY

General Features

- Etiology
  - Pulmonary valvar stenosis is congenital anomaly
    - Maldevelopment of pulmonary valve tissue & distal portion of bulbus cordis
- Genetics
  - No genetic predisposition in valvar stenosis
  - Williams syndrome related to contiguous gene deletion of locus 7q4
    - Distinct facial features & personality, mild retardation, cardiovascular abnormalities, elastin arteriopathy
  - Noonan syndrome is autosomal dominant
    - > 50% of individuals have changes in *PTPN11* gene
    - Multiple anomalies with dysplastic pulmonary valve, growth failure, mild intellectual impairment
  - Alagille syndrome is autosomal dominant related to mutations on chromosome 20p12
    - Pulmonic valve & peripheral stenoses associated with cholestatic jaundice in infancy
      - Jaundice due to bile duct hypoplasia/paucity

  - Butterfly vertebrae, abnormal facies, intellectual impairment, eye & renal anomalies
- Associated abnormalities
  - Most children normal without any other lesions
  - Can be associated with other common congenital lesions in 10%
    - Infundibular stenosis & ventricular septal defect in tetralogy of Fallot
    - Atrial septal defect, patent ductus arteriosus
    - Complex cardiac lesions
  - Williams syndrome
    - Supravalvar aortic stenosis, coarctation, supravalvar PS, peripheral pulmonary stenoses
  - Noonan syndrome
    - Dysplastic pulmonary valve
  - Alagille syndrome
    - PS with multiple peripheral artery stenoses

Staging, Grading, & Classification

- Pulmonary valvar stenosis: Mild, moderate, severe

Gross Pathologic & Surgical Features

- Valve thickened with fused commissure
- Dysplastic valves have redundant tissue & thickened leaflets

Microscopic Features

- Valve thickened with fibrous, myxomatous, & collagenous tissue

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Mild stenosis: Usually asymptomatic with systolic ejection murmur
  - Moderate stenosis: Exertional dyspnea, easy fatigability
  - Severe stenosis: Infants may present with severe cyanosis
    - ↓ pulmonary flow, ↑ right ventricular pressure, tricuspid regurgitation, & shunting from right atrium to left atrium
    - Severe stenosis may appear similar to pulmonic atresia, right ventricular hypoplasia, or coronary artery fistulas
- Other signs/symptoms
  - Loud systolic ejection murmur & click at left upper heart border

Demographics

- Age
  - Critical pulmonic stenosis occurs in newborns
  - PS often otherwise diagnosed between 2-6 years of age during routine physical exam
- Gender
  - M = F

Natural History & Prognosis

- Critical pulmonic stenosis in infancy will progress; can be fatal if not treated
- Pulmonary valvar stenosis with mild gradient does not usually progress
  - Children have normal life expectancy
- Pulmonary valvar stenosis, which is moderate, will progress
  - Clinically well tolerated
  - After valvotomy, may have pulmonary insufficiency, which is usually tolerated
- Mortality depends on severity of lesions, but mild to moderate have normal life expectancy

Treatment

- Observation, medical management for mild valvar stenosis gradients < 25 mmHg
- Balloon valvuloplasty is treatment of choice for moderate to severe valvar gradients > 50 mmHg
  - Long-term ↓ in right ventricular pressure & estimated gradient
  - Hemodynamically insignificant pulmonary insufficiency may occur in 80%
  - Recurrence of PS happens in 15% at 10 years
  - Excellent outcome with survival similar to general population
  - Balloon valvotomy not as effective in dysplastic valves, which may need surgery
- Newborns with severe or critical pulmonic stenosis
  - May need immediate valvotomy
  - Prostaglandin to maintain ductus arteriosus flow
  - Surgical valvotomy or palliative Blalock-Taussig shunt
  - Lesion may be associated with hypoplasia of right ventricle requiring univentricular repair

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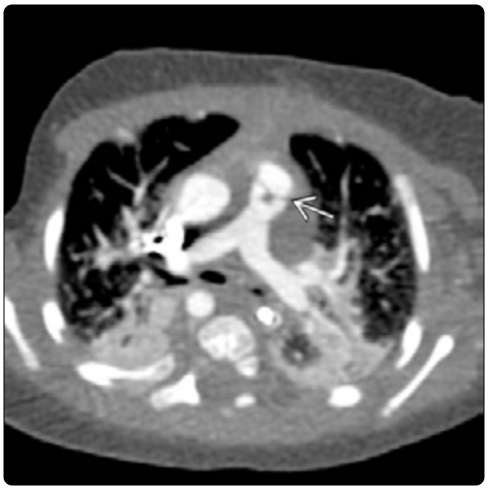
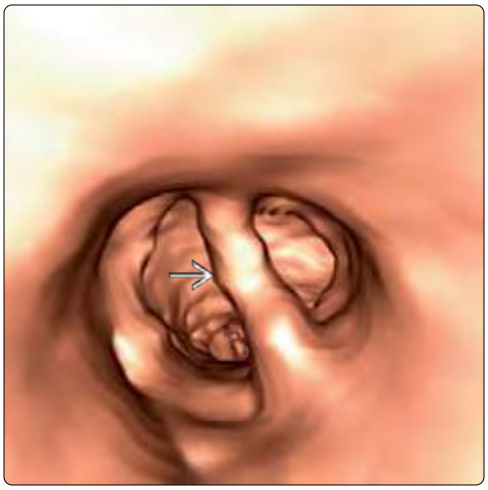
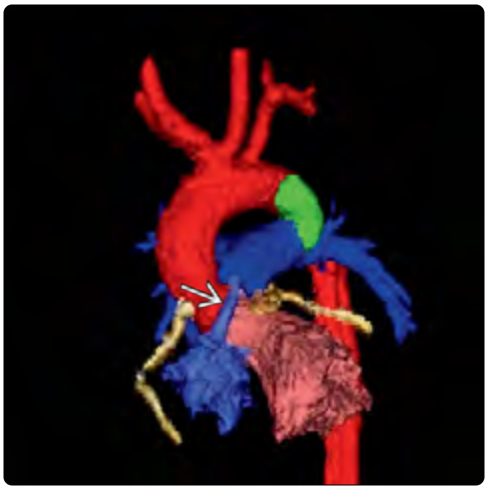
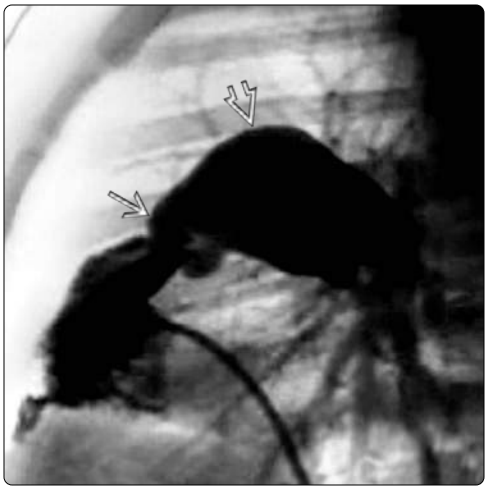


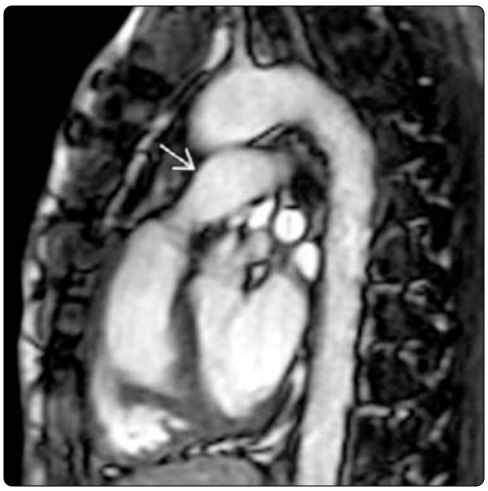


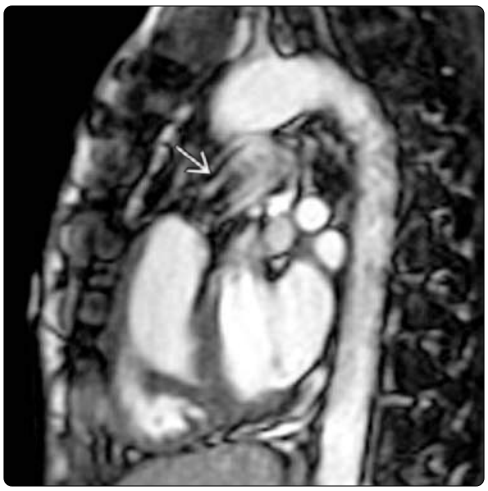
(Left) *Axial cardiac CTA shows valvular pulmonic stenosis with a markedly thickened pulmonic valve ➡. (Right) An intraluminal view from a 3D cardiac CTA shows a thickened, dysplastic pulmonary valve with a thick septation centrally ➡.*





(Left) *Anterior oblique color-coded 3D cardiac CTA in an infant shows focal pulmonic stenosis with marked narrowing of the pulmonary outflow tract ➡. A large PDA (green) is seen connecting the main pulmonary artery (blue) to the descending aortic arch (red). (Right) Lateral RV angiogram shows a focal valvular stenosis ➡ with poststenotic dilation of the main pulmonary artery ➡.*





(Left) *Sagittal GRE cine image during diastole from a cardiac MR shows poststenotic dilation ➡ of the main pulmonary artery in a patient with pulmonic stenosis. (Right) Sagittal GRE cine image during systole from a cardiac MR shows a signal void ➡ in the main pulmonary artery from the turbulent flow jet in a patient with severe pulmonic stenosis. The velocity can be measured on phase-contrast images.*

Scimitar Syndrome

KEY FACTS

TERMINOLOGY

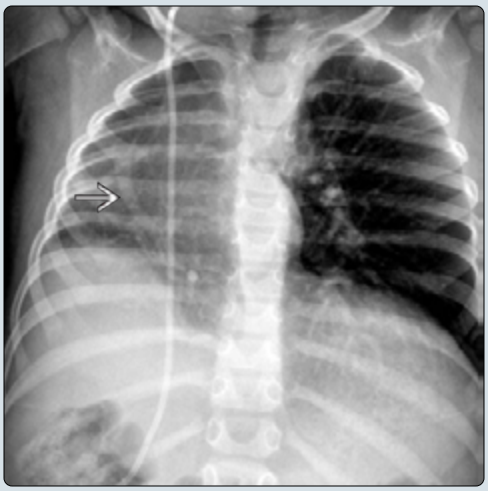
- Synonyms: Hypogenetic lung syndrome, congenital pulmonary venolobar syndrome
- Scimitar syndrome triad: Right lung hypoplasia, anomalous right pulmonary venous connection to inferior vena cava (IVC), & anomalous systemic arterial supply to right lower lobe
  - Extracardiac left-to-right shunt
  - Form of partial anomalous pulmonary venous return (PAPVR)
- Category: Acyanotic, right-sided cardiac chamber enlargement, ↑ pulmonary vascularity

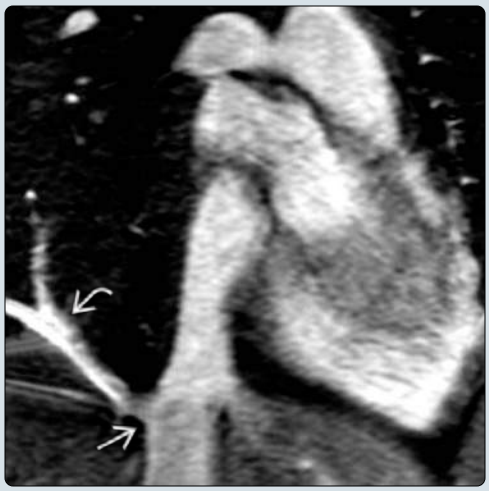
IMAGING

- Scimitar sign: Curved anomalous venous trunk, resembling Turkish sword, in right medial costophrenic sulcus near right heart border; typically ↑ in caliber in caudad direction
- Right lung hypoplasia with mediastinal shift
- Dextroversion of heart (not dextrocardia as apex still directed toward left)
- Prominent right atrium, active pulmonary vascular congestion: Shunt vascularity

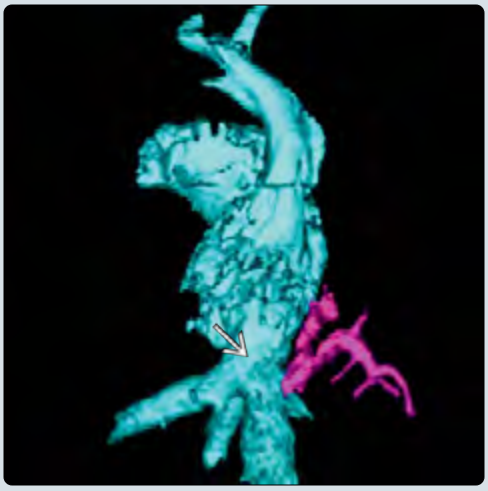
CLINICAL ISSUES

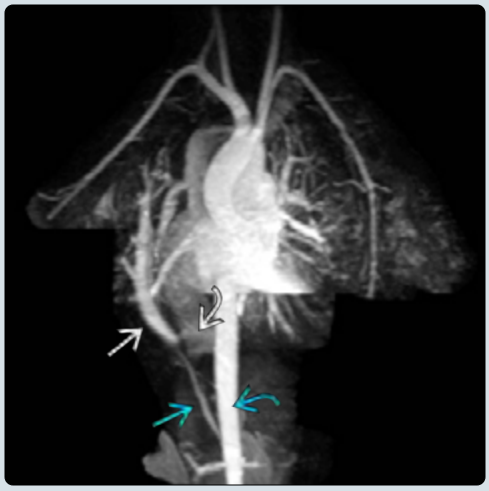
- Newborn: Congestive heart failure, right heart volume overload, pulmonary hypertension
- Young child: Recurrent infections in right lung base
- Older child & adult: Often asymptomatic (incidental finding on chest radiograph)
- Treatment
  - Embolization of systemic arterial supply
  - Baffling of common right pulmonary vein onto left atrium
    - Surgical repair indicated when left-to-right shunt > 2:1





(Left) *Frontal radiograph shows mediastinal shift to the right from a hypoplastic right lung. There is an enlarged curvilinear vein ➡ in the right lower lobe from partial anomalous pulmonary venous return (PAPVR) in this patient with scimitar syndrome.* **(Right)** *Coronal MIP image from an MRA of the chest shows PAPVR with the right lower lobe (RLL) pulmonary vein ➡ draining into the inferior vena cava (IVC) ➡.*





(Left) *Posterior view from a 3D color-coded CTA shows PAPVR of the RLL with the anomalous vein (pink) draining to the IVC ➡.* **(Right)** *Frontal image from an MR angiogram shows a large scimitar (pulmonary) vein ➡ draining to the IVC ➡ with systemic arterial supply ➡ to the RLL arising from the proximal abdominal aorta ➡.*

TERMINOLOGY

Synonyms

- Hypogenetic lung syndrome, congenital pulmonary venolobar syndrome

Definitions

- Scimitar syndrome triad: Right lung hypoplasia, anomalous right pulmonary venous connection to inferior vena cava (IVC), & anomalous systemic arterial supply to right lower lobe
  - Scimitar vein represents form of partial anomalous pulmonary venous return (PAPVR)
- Hemodynamics: Venous flow from right lung returns to right atrium → volume overload of right heart [atrial septal defect (ASD) type physiology]

IMAGING

General Features

- Best diagnostic clue
  - Scimitar sign: Curved anomalous pulmonary vein, resembling Turkish sword, typically draining right lower & middle lobes
    - ↑ in caliber in caudad direction

Radiographic Findings

- Right lung hypoplasia with mediastinal shift
- Prominent right atrium & shunt vascularity when multiple lobes drain to IVC
- Scimitar vein may be seen in right medial costophrenic sulcus

CT Findings

- CTA with 3D reconstruction most helpful to demonstrate venous drainage, anomalous systemic arterial supply, & right pulmonary & main bronchus hypoplasia

MR Findings

- Phase-contrast MRA for shunt flow calculation (Qp/Qs)
- Gadolinium-enhanced MRA, coronal acquisition with 3D reconstruction, for anomalous right pulmonary venous & arterial development

Echocardiographic Findings

- Echocardiogram
  - Scimitar vein connecting to IVC
  - Enlarged right atrium & ventricle with significant shunt

Angiographic Findings

- Conventional
  - Scimitar vein opacifies during venous phase of pulmonary artery injection
  - Injection of abdominal aorta: Anomalous systemic arterial supply to right lung base originating from abdominal or thoracic aorta

Imaging Recommendations

- CTA or MRA better than echocardiography for complete assessment & can replace diagnostic angiocardiography
- Angiography reserved for coil embolization

DIFFERENTIAL DIAGNOSIS

Other Forms of PAPVR

- Right pulmonary vein(s) to azygous vein, superior vena cava (SVC), right atrium (with sinus venosus ASD)

True Dextrocardia With Abdominal Situs Solitus

- Other complex cardiac anomalies

Isolated Right Pulmonary Hypoplasia

- Normal right pulmonary venous connection to left atrium

Bronchopulmonary Sequestration

- Mass in right lung base not connected to bronchial tree, receives systemic arterial supply

PATHOLOGY

General Features

- Embryology
  - Primary abnormality in development of right lung, with secondary anomalous pulmonary venous connection
- Pathophysiology
  - Extracardiac left-to-right shunt: ASD physiology

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depends on age at presentation & size of left-to-right shunt
    - Newborn: Congestive heart failure, right heart volume overload, pulmonary hypertension
    - Young child: Recurrent infections in right lung base
    - Older child & adult: Often asymptomatic (incidental finding on chest radiograph)

Natural History & Prognosis

- Large shunt → irreversible pulmonary hypertension
- Moderate to poor prognosis with neonatal presentation
- May be asymptomatic for many years with small shunt

Treatment

- Surgical repair indicated when left-to-right shunt > 2:1
- Baffling of common right pulmonary vein into left atrium
- Embolization of systemic arterial supply

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Blalock-Taussig Shunt

KEY FACTS

TERMINOLOGY

- Original/classic Blalock-Taussig (BT) shunt
  - Ligation & division of subclavian artery with end-to-side anastomosis of proximal subclavian artery to pulmonary artery
  - Rarely performed today due to complications
    - Overshunting, nerve injury, & potential growth disturbance of ipsilateral upper extremity
- Modified BT shunt
  - Synthetic prosthetic graft (Gore-Tex) between subclavian artery & ipsilateral pulmonary artery
    - Contralateral to side of aortic arch
  - Up to 90% patency rate at 2 years of age
- Used as palliative procedure to ↑ pulmonary blood flow prior to definite repair
  - Tetralogy of Fallot
  - Tricuspid atresia
  - Pulmonary atresia
  - Hypoplastic left heart syndrome (part of stage 1 Norwood procedure)

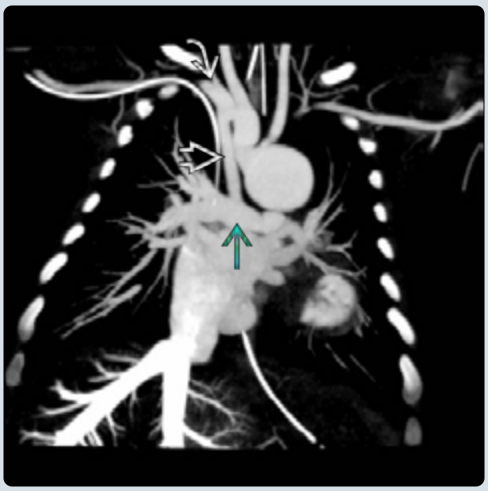
IMAGING

- CTA/MRA: Linear, tubular contrast-opacified structure connecting subclavian artery to ipsilateral pulmonary artery (usually right-sided)
  - Multiplanar & 3D reformations very helpful
- Dark signal intensity within patent shunt on black blood MR sequences
- Complications
  - Stenosis/thrombosis/occlusion
  - Perigraft seroma
  - Pseudoaneurysm

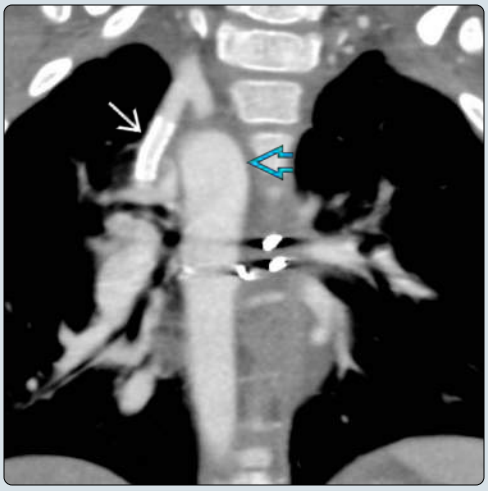
TOP DIFFERENTIAL DIAGNOSES

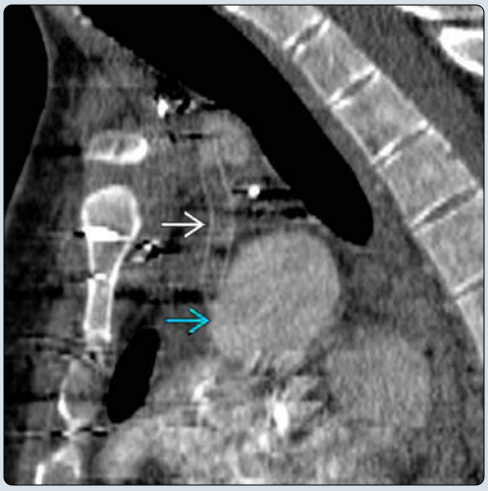
- Other palliative shunts (Glenn, Waterston, Potts, Sano)

(Left) *Coronal graphic shows a modified Blalock-Taussig (BT) shunt extending from the right subclavian artery to the right pulmonary artery. (Note that the superior vena cava has been excluded from the graphic in order to visualize the BT shunt.)* **(Right)** *Coronal MIP CTA in a 3 month old status post Norwood stage 1 procedure shows a widely patent modified BT shunt extending between the proximal right subclavian artery & the proximal right pulmonary artery.*



(Left) *Coronal CTA reconstruction in this 5 year old with a history of tetralogy of Fallot demonstrates opacification of a vascular structure with a stent connecting the right subclavian artery to the right pulmonary artery, consistent with a modified BT shunt. Note the right-sided aortic arch.* **(Right)** *Sagittal oblique CTA reconstruction demonstrates a thrombosed BT shunt connecting the right subclavian artery to a dilated pulmonary artery.*





TERMINOLOGY

Abbreviations

- Blalock-Taussig (BT) shunt

Definitions

- Palliative procedure to augment pulmonary blood flow in various uni- & biventricular anatomies: Tetralogy of Fallot, tricuspid atresia, pulmonary atresia, & hypoplastic left heart syndrome (part of Norwood stage 1)
  - Original/classic BT shunt
    - Developed in 1945 by Dr. Alfred Blalock, Dr. Helen Taussig, & Vivien Theodore Thomas
    - Ligation & division of right subclavian artery with end-to-side anastomosis of proximal right subclavian artery to right pulmonary artery
    - Complications included shunt thrombosis, overshunting, nerve injury, & potential growth disturbance of ipsilateral upper extremity
    - Rarely performed today
  - Modified BT shunt
    - Synthetic graft prosthesis (Gore-Tex) between subclavian artery & ipsilateral pulmonary artery, contralateral to side of aortic arch
    - End-to-side anastomosis at each connection performed through median sternotomy or lateral thoracotomy (with former preferred)
    - Up to 90% patency rate at 2 years of age

IMAGING

General Features

- Complications
  - Thrombosis
    - Can be early or late complication
  - Complete occlusion
    - ~ 10% of cases
    - More common in grafts < 3 mm in size
  - Stenosis, typically near proximal or distal anastomosis
  - Perigraft seroma
    - Complication secondary to leakage of fluid across Gore-Tex graft
  - Pseudoaneurysm

Radiographic Findings

- Typically ↑ pulmonary vascularity compared to pre-BT shunt radiograph
- Postthoracotomy rib changes
- Features of underlying congenital heart disease

CT Findings

- Tubular contrast-opacified structure (synthetic graft) connecting subclavian artery to ipsilateral pulmonary artery, usually right sided
- Abrupt cut-off consistent with occlusion or prior takedown in older child
- Filling defect within shunt compatible with thrombosis
- Evaluate for proximal or distal anastomotic stenosis
- Look for associated perigraft seroma or, rarely, pseudoaneurysm (especially if recently stented)
- In patients who have undergone classic BT shunt in past, ipsilateral subclavian artery may be absent

MR Findings

- Tubular structure connecting subclavian artery to ipsilateral pulmonary artery seen as dark signal intensity structure on black blood sequences
- Contrast enhanced MRA best to confirm & characterize thrombus, occlusion, stenosis, pseudoaneurysm, or associated perigraft seroma
- Can quantify flow on phase-contrast sequences

Echocardiographic Findings

- Can evaluate BT shunt for patency & measure gradient across stenosis
- Can evaluate perigraft seroma

Angiographic Findings

- Typically performed for angioplasty with stenting of shunt stenosis or occlusion
- Pseudoaneurysm can develop around stent

Imaging Recommendations

- Best imaging tool
  - CTA/MRA with multiplanar & 3D reformations

DIFFERENTIAL DIAGNOSIS

Other Palliative Shunts

- Glenn shunt
  - Superior vena cava to pulmonary artery
  - Requires low pulmonary vascular resistance
    - Not performed under 3-6 months of age due to high pulmonary vascular resistance
- Waterston shunt
  - Ascending aorta to pulmonary artery
  - No longer performed
    - Congestive heart failure & pulmonary hypertension due to excessive pulmonary blood flow
- Potts shunt
  - Descending aorta to left pulmonary artery
  - No longer performed
    - Pulmonary hypertension & pulmonary artery kinking
- Sano shunt
  - Direct shunt between right ventricle & pulmonary artery
  - Performed as part of Norwood stage 1 in lieu of BT shunt in some cases of hypoplastic left heart

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Sano Shunt

KEY FACTS

TERMINOLOGY

- Sano shunt provides alternate source of pulmonary blood flow to modified Blalock-Taussig (BT) shunt in hypoplastic left heart patients undergoing stage 1 Norwood procedure
- Extracardiac conduit between right ventricle & pulmonary artery
  - Small ventriculotomy made in right ventricular outflow tract
  - Conduit is nonvalved, thus allowing free regurgitation
- Typically performed during first few days of life
- Benefits
  - Elimination of "coronary steal" phenomenon secondary to reversal of diastolic flow with modified BT shunt
  - Forward flow through shunt only occurs during systole
  - Improved hemodynamic stability postoperatively compared to modified BT shunt
  - Higher rate of transplantation-free survival at 12 months compared with modified BT shunt

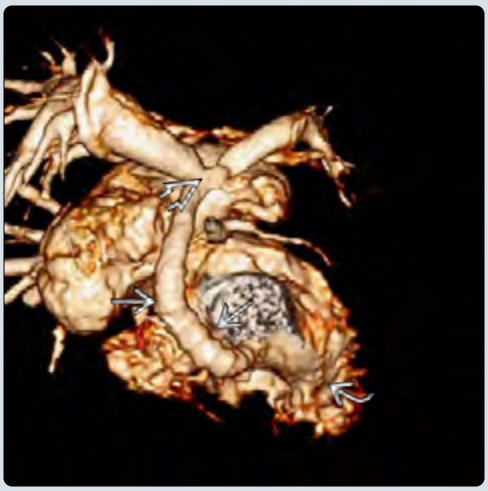
IMAGING

- CT useful in immediate postoperative period to assess for complications
- CT can also be used to evaluate status of Sano shunt itself & branch pulmonary arteries
  - Evaluate shunt thrombosis or occlusion
  - Branch pulmonary artery stenoses
  - Aneurysm/pseudoaneurysm at ventriculotomy site
- MR provides comprehensive, ionizing radiation-free evaluation of postoperative anatomy & function
- 3D reconstructions of CTA/MRA extremely helpful

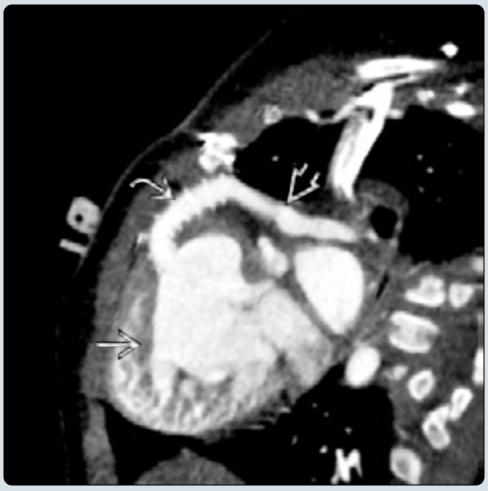
TOP DIFFERENTIAL DIAGNOSES

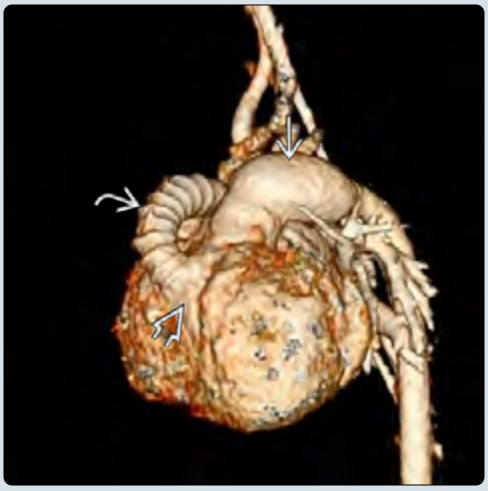
- Modified BT shunt

(Left) *Coronal graphic demonstrates a stage 1 Norwood with a Sano shunt. The neoaorta has been constructed using the patient's native main pulmonary artery ➪. The Sano shunt ➪ connects the right ventricle to the left pulmonary artery ➪.* **(Right)** *3D volume-rendered image demonstrates the Sano conduit ➪ extending between the systemic right ventricular chamber ➪ & the pulmonary arterial confluence ➪ in a 3-month-old patient with a history of hypoplastic left heart, status post stage 1 Norwood operation.*



(Left) *Oblique cardiac CTA shows a Sano shunt ➪ extending between the right ventricular chamber ➪ & the pulmonary artery ➪ in a 4 month old with a history of a Norwood operation for hypoplastic left heart syndrome.* **(Right)** *Lateral 3D cardiac CTA shows a Sano shunt ➪ is shown at the right ventricular end ➪ in an infant status post stage 1 of a Norwood procedure. Note the neoaorta ➪ adjacent to the Sano shunt.*





TERMINOLOGY

Synonyms

- Right ventricle-pulmonary artery (RV-PA) conduit

Definitions

- Norwood stage 1 involves using main pulmonary artery to augment ascending aorta in patients with hypoplastic left heart syndrome (HLHS)
  - New source of pulmonary blood flow is required
    - Blalock-Taussig (BT) shunt provides source of pulmonary blood flow from systemic arterial system
    - BT flow is continuous in both systole & diastole
    - Because ~ 75% of coronary blood flow occurs during diastole, "coronary steal" phenomenon develops
      - Diastolic retrograde flow occurs in coronaries & descending thoracic aorta
      - Resultant coronary insufficiency theorized to play major role in operative & postoperative morbidity/mortality of stage 1 Norwood with BT shunt
  - Sano conduit is alternative to modified BT shunt
  - Extracardiac conduit between right ventricle & pulmonary artery
    - Small ventriculotomy made in right ventricular outflow tract
    - 4- to 6-mm synthetic graft anastomosed to right ventricle & pulmonary artery to left of neoaorta
    - Conduit is nonvalved, thus allowing free regurgitation
- Typically performed during first few days of life
- Stage 2 Norwood (Glenn anastomosis) typically needs to be performed slightly earlier after Sano shunt than with modified BT shunt in 3- to 6-month timeframe
- Benefits
  - Elimination of "coronary steal" phenomenon secondary to reversal of diastolic flow with modified BT shunt
    - Forward flow through shunt only occurs during systole
  - Improved hemodynamic stability postoperatively compared to modified BT shunt
  - Higher rate of transplantation-free survival at 12 months compared with modified BT shunt
  - Nonrandomized & retrospective studies have shown potential benefit in outcomes associated with RV-PA conduit when compared to modified BT shunt
- One limitation cited in available literature is poor ventricular performance in view of ventriculotomy
  - Current available evidence, although weak, does not show any adverse effects of ventriculotomy on ventricular performance in patients with Sano shunt in short- & medium-terms

IMAGING

General Features

- Complications
  - Arrhythmias
  - Shunt stenosis/obstruction
  - Shunt thrombosis
  - Branch pulmonary artery stenoses
  - Right ventricular dysfunction
  - Aneurysm/pseudoaneurysm formation at ventriculotomy site
  - Shunt infection & pulmonary embolism
  - May require more extensive pulmonary artery reconstruction at time of Glenn anastomosis (stage 2 Norwood)

CT Findings

- Useful in immediate postoperative period to assess for complications
  - Mediastinitis
  - Hemorrhage
  - Aneurysm
- Can also be used to evaluate status of Sano shunt itself & branch pulmonary arteries
- Radiation exposure is concern; however, newer generation scanners can provide requisite information in sub mSv radiation doses
- 3D reconstructions extremely helpful

MR Findings

- Comprehensive evaluation of postoperative anatomy & function, without ionizing radiation
- Functional analysis for systemic ventricle
- Phase-contrast imaging can be used to quantify flow in
  - Sano shunt
  - Branch pulmonary arteries
  - Neoaorta
- Pulmonary:systemic arterial flow ratio (Qp/Qs) can be calculated
- 3D reconstructions extremely helpful

Echocardiographic Findings

- Duplex Doppler imaging of mid to distal portion of Sano shunt can provide flow information & evaluation for stenosis
- Proximal anastomosis of shunt & branch pulmonary arteries may not be adequately visualized in view of poor acoustic windows

DIFFERENTIAL DIAGNOSIS

Modified Blalock-Taussig Shunt

- Synthetic graft connection between subclavian artery & ipsilateral pulmonary artery as part of stage 1 of Norwood

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Glenn Shunt

KEY FACTS

TERMINOLOGY

- Superior cavopulmonary shunt
- Goal is to direct systemic venous return from upper 1/2 of body to pulmonary circulation, directly bypassing right heart
  - Superior vena cava (SVC) divided from right atrium at superior cavoatrial junction
  - End-to-side anastomosis between divided SVC & right pulmonary artery (PA)
  - SVC flow directed to confluent branch PAs
- Used in patients with single ventricle physiology as staged palliative procedure prior to Fontan, ultimately resulting in total cavopulmonary connection
- Comprises stage 2 of Norwood procedure for hypoplastic left heart syndrome
- Typically performed between 3-9 months of age when pulmonary vascular resistance has ↓ sufficiently
- If 2 SVCs are present, each can be anastomosed to its respective PA, creating bilateral Glenn shunts

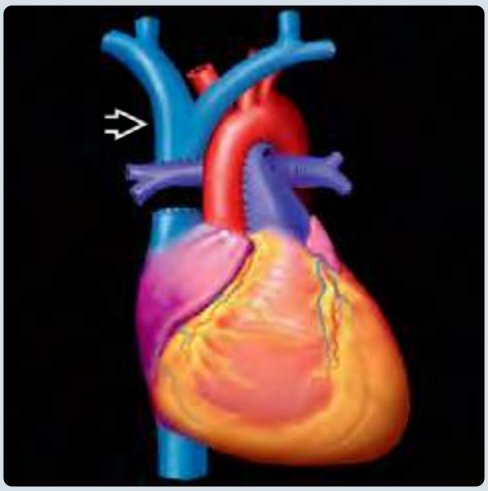
IMAGING

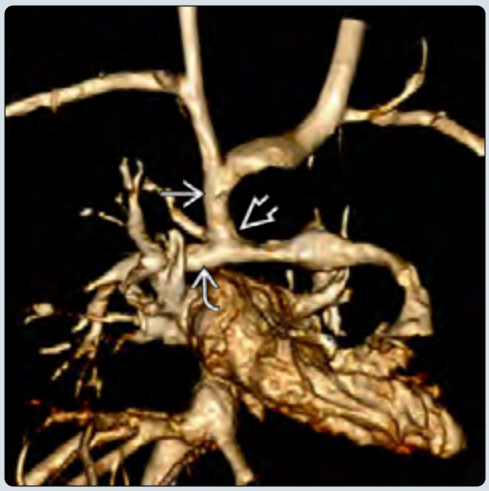
- CECT can be used to assess for shunt patency, collaterals, & branch pulmonary artery stenoses
  - Delayed equilibrium-phase CTA should be performed to avoid contrast mixing artifact within branch PAs
- MR can be used in pre-Glenn & post-Glenn evaluation
  - Evaluate cardiac anatomy with ventricular & atrioventricular valve function
  - Evaluate branch pulmonary arteries for stenoses along with flow differential calculation by phase-contrast imaging (PCI)
    - Velocities picked for PCI across branch PAs typically < 100 cm/s since venous flow via Glenn shunt
  - Evaluate neoaorta
  - Quantify pulmonary-to-systemic blood flow & collaterals

TOP DIFFERENTIAL DIAGNOSES

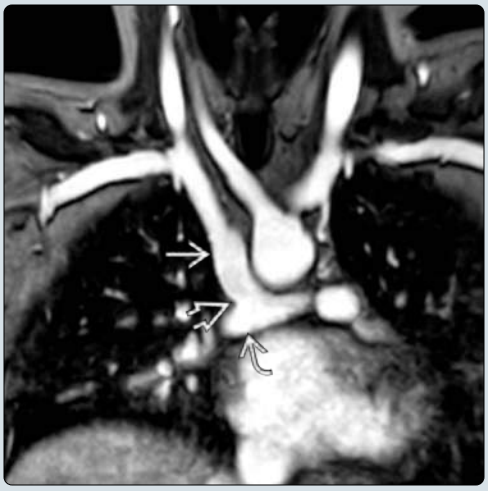
- Hemi-Fontan

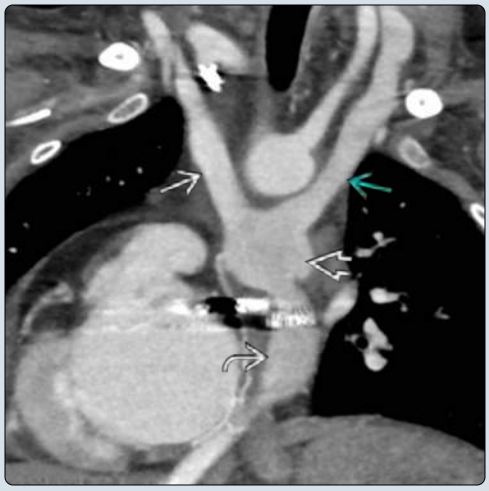
(Left) *Graphic shows a Glenn shunt as part of a stage 2 Norwood procedure. A direct connection between the superior vena cava (SVC) & the right pulmonary artery (RPA) is created with deoxygenated systemic venous flow directed to both lungs.* **(Right)** *3D volume-rendered CTA in the coronal plane in a 2-year-old patient with a univentricular heart shows the SVC to RPA anastomosis, compatible with a bidirectional Glenn shunt. Note the confluent branch pulmonary arteries (PAs).*





(Left) *Coronal MRA in a patient status post stage 2 Norwood demonstrates a bidirectional Glenn shunt with anastomosis of the right SVC to the RPA.* **(Right)** *Coronal reformat from a delayed-phase CTA study in a 6-year-old patient with underlying heterotaxy shows that the right & left SVC are anastomosed to the PAs, comprising a superior cavopulmonary communication (Glenn). Also note the left-sided Fontan.*





TERMINOLOGY

Synonyms

- Superior cavopulmonary shunt

Definitions

- Goal is to direct systemic venous return from upper 1/2 of body to pulmonary circulation, directly bypassing right heart
- Originally described by Dr. William Glenn in 1958
  - End-to-end anastomosis of divided superior vena cava (SVC) to divided right pulmonary artery (PA)
  - SVC flow directed to right lung only
- Bidirectional Glenn shunt more commonly used now
  - SVC divided from right atrium at superior cavoatrial junction followed by end-to-side anastomosis between divided SVC & right PA
  - SVC flow directed to both right & left PAs
- Performed in patients with single ventricle physiology as staged palliative procedure prior to Fontan; final result called total cavopulmonary connection
- Glenn shunt forms stage 2 of Norwood procedure for hypoplastic left heart syndrome
- By reducing volume load, bidirectional Glenn shunt reduces single ventricle wall stress & atrioventricular valve insufficiency
- Since no synthetic graft material used, shunt grows with child
- Typically performed between 3-9 months of age
  - By this age, pulmonary vascular resistance has ↓ to level where systemic venous return enters pulmonary circulation without assistance of right heart pump
- If 2 SVCs are present, each can be anastomosed to its respective PA, creating bilateral Glenn shunts
- Azygous & hemiazygos veins are ligated as part of procedure

IMAGING

General Features

- Complications
  - Superior-to-inferior systemic venous collaterals
    - Can be coil embolized
  - Progressive cyanosis
  - Formation of pulmonary arteriovenous malformations (AVM)
    - Hypothesized to be secondary to exclusion of hepatic venous flow through lungs
    - Resultant lack of hepatic factor delivery to pulmonary circulation
    - Subsequent lack of inhibition of endothelial proliferation in lungs
  - Branch pulmonary artery stenosis
  - Arrhythmias

CT Findings

- Can be used postoperatively to assess for shunt patency
- Useful in evaluating thoracic vasculature, including neoaorta, branch pulmonary arteries, pulmonary veins, & collaterals
- Can be used for detection of pulmonary AVM
- Pre-Glenn evaluation of cardiac anatomy, including branch PAs instead of cardiac MR
- 3D reconstructions helpful

MR Findings

- Traditionally used in pre-Glenn evaluation instead of conventional angiography
  - Evaluate cardiac anatomy & function
  - Evaluate valvular function
  - Evaluate central pulmonary arteries & veins
  - Evaluate neoaorta
  - Quantify pulmonary-to-systemic blood flow
- Useful in post-Glenn evaluation
  - Evaluate ventricular function
  - Evaluate shunt patency & flow
  - Evaluate for pulmonary artery narrowing
  - Evaluate collaterals & pulmonary AVMs

Ultrasonographic Findings

- Echocardiography used for pre- & postoperative anatomic & hemodynamic assessment
- Limited evaluation of branch pulmonary arteries & pulmonary veins

Angiographic Findings

- Considered gold standard in preoperative assessment of anatomic & hemodynamic suitability for Glenn shunt
- Catheter-based interventions, such as collateral embolization & aortic balloon dilation, can be performed if required

DIFFERENTIAL DIAGNOSIS

Hemi-Fontan

- Similar to Glenn, except continuity of SVC & right atrium maintained
- SVC anastomosed to right PA
- Patch of homograft tissue sewn across superior cavoatrial junction preventing systemic venous return from upper body into right atrium
- Simplifies subsequent lateral tunnel Fontan completion as continuity of right atrium & SVC maintained

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Fontan Operation

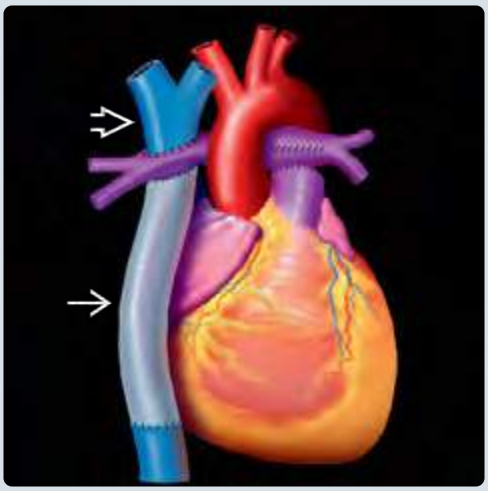
KEY FACTS

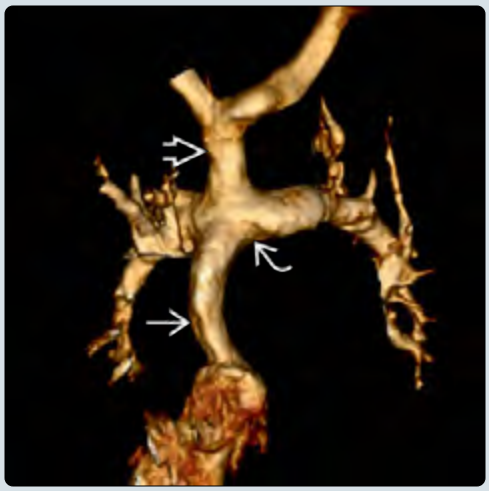
TERMINOLOGY

- Total cavopulmonary connection
- Used in single ventricle physiology as stage 3 of Norwood procedure
  - Tricuspid atresia
  - Hypoplastic left heart syndrome
  - Double-inlet ventricle
  - Some heterotaxies
- Typically performed between 18-36 months of age
- Lateral tunnel Fontan
  - Intraatrial tunnel created in right atrium connecting inferior vena cava (IVC) to pulmonary artery
- Extracardiac conduit Fontan (typically favored)
  - Synthetic graft tube connected to IVC & right pulmonary artery
- Excellent early, mid, & late-term outcomes, with mortality rates < 5-10%

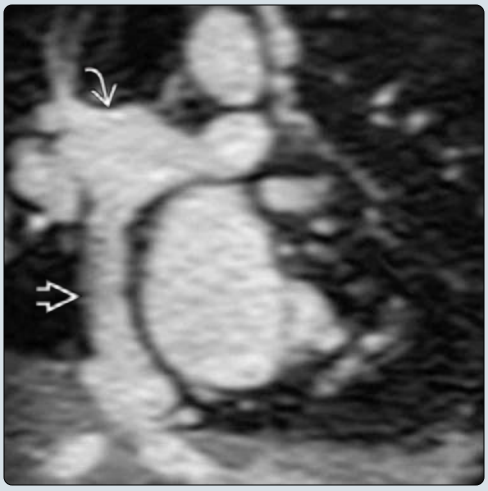
IMAGING

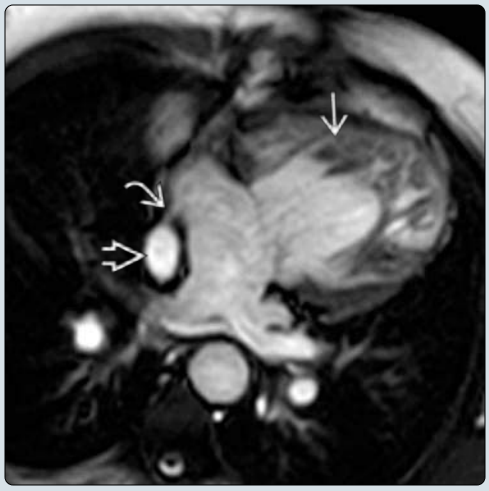
- CECT can be used for detection of Fontan complications such as thrombus, hepatomegaly, effusions, & pulmonary arteriovenous malformation
  - Delayed-phase of image acquisition during CTA eliminates contrast mixing artifacts within branch pulmonary arteries & Fontan conduit
- MR can be used in pre-Fontan & post-Fontan assessment
  - Evaluation of branch pulmonary arteries, including determination of flow differential using phase-contrast sequences
  - Evaluation of superior cavopulmonary connection (Glenn shunt)
  - Identification of collaterals
  - Evaluation of ventricular function
  - Determine patency of Fontan conduit & exclude thrombus
  - Assessment of hepatic fibrosis





(Left) *Graphic shows an extracardiac Fontan conduit. The conduit creates a direct connection between the inferior vena cava (IVC) & the right pulmonary artery (RPA). A bidirectional Glenn shunt is also depicted.* **(Right)** *Coronal 3D volume-rendered MRV image in a 15-year-old patient shows a right extracardiac Fontan anastomosed to the RPA. Also note the right bidirectional Glenn connection.*





(Left) *Coronal oblique view from a gadolinium-enhanced 3D gradient-echo sequence performed after administration of a blood-pool contrast agent in this 14-year-old patient with a univentricular heart shows the right extracardiac Fontan conduit connecting to the RPA.* **(Right)** *Axial SSFP sequence in a 14-year-old patient with a univentricular heart demonstrates a right-sided Fontan conduit. Note the fenestration along the medial wall of the conduit.*

TERMINOLOGY

Synonyms

- Total cavopulmonary connection

Definitions

- Goal is to direct systemic venous return to pulmonary circulation directly, bypassing right heart
- Initially described by Dr. Francis Fontan in 1968
- Used in single ventricle physiology
  - Tricuspid atresia
  - Hypoplastic left heart syndrome
  - Double-inlet ventricle
  - Some heterotaxies
- Stage 3 of Norwood procedure
- Typically performed between 18-36 months of age
- Originally described as extracardiac valved conduit between right atrium & left pulmonary artery
  - In combination with Glenn shunt, creates physiologic correction of blood flow
  - Superior vena cava (SVC) blood directed to right pulmonary artery & inferior vena cava (IVC) blood directed to left pulmonary artery
  - Complications of classic Fontan
    - Right atrial enlargement & hypertension
    - Impaired ventricular function
    - ↓ pulmonary vascular blood flow
    - Tricuspid valve insufficiency
- In order to preserve ventricular & pulmonary vascular function, modified Fontan now favored
  - Lateral tunnel Fontan
    - Intraatrial tunnel created in right atrium using prosthetic material
    - IVC anastomosed to caudal aspect of tunnel
    - Pulmonary artery anastomosed to cephalad aspect of tunnel
  - Extracardiac conduit Fontan (typically favored)
    - IVC divided from right atrium
    - Synthetic graft tube connected to IVC inferiorly & right pulmonary artery superiorly; travels along side of right atrium
  - In either form, IVC blood directed to pulmonary circulation, bypassing right heart structures
- Small fenestration often created between Fontan circuit & right atrium
  - Prevents volume overload to lungs
  - Buffers any ↑ in systemic venous pressure
- Excellent early, mid, & late-term outcomes, with mortality rates < 5-10%

IMAGING

General Features

- Complications
  - Arrhythmias
  - Liver dysfunction due to fibrosis
  - Protein-losing enteropathy
  - Heart failure
  - Thrombus & emboli
  - Formation of pulmonary arteriovenous malformations (AVM)
    - Hypothesized to be secondary to exclusion of hepatic veins from Fontan pathway with lack of protective hepatic factor delivery to lungs
    - Potentially reversible if hepatic venous flow can be redirected to lungs
  - Chylous pleural effusions
  - Plastic bronchitis

CT Findings

- Can be used for detection of Fontan complications such as thrombus, hepatomegaly, effusions, & pulmonary AVM
- 3D reconstructions very helpful

MR Findings

- Pre-Fontan evaluation
  - Can potentially obviate need for preoperative catheter angiography
  - Evaluation of superior cavopulmonary connection formed by Glenn procedure
  - Evaluation of branch pulmonary arteries
  - Identification of collaterals
  - Evaluation of ventricular function
- Post-Fontan evaluation
  - Evaluate patency of Fontan pathway
  - Evaluate ventricular function
  - Evaluate pulmonary artery stenoses
  - Identify collaterals
  - MR elastography for evaluation of hepatic fibrosis
  - MR lymphangiography for chylous effusion, plastic bronchitis

DIFFERENTIAL DIAGNOSIS

Kawashima Procedure

- Used in patients with single ventricle physiology & interrupted IVC
  - Fontan not feasible
- SVC, including azygous continuation, connected to right pulmonary artery
  - All systemic venous return directed to pulmonary artery
    - Except hepatic veins & coronary sinus

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Norwood Procedure

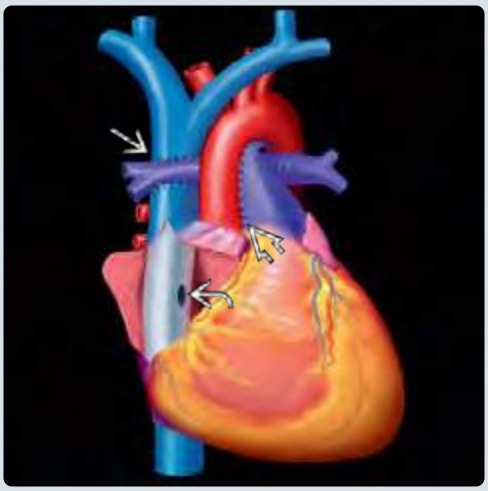
KEY FACTS

TERMINOLOGY

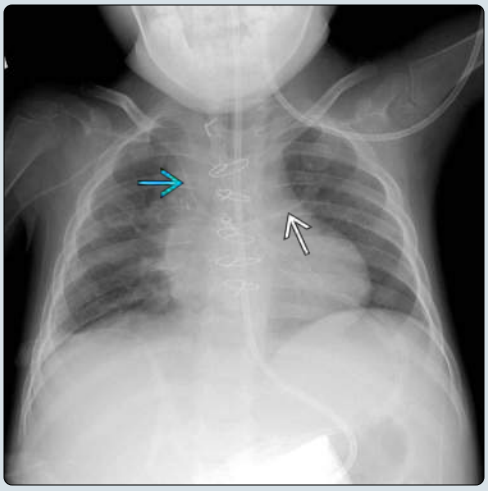
- 3-stage procedure to palliate single ventricle physiology (most commonly hypoplastic left heart syndrome)
- Goals of Norwood procedure
  - Utilize single right ventricle as systemic pump & reconstruct systemic arterial outflow
  - Ensure pulmonary venous return to right heart
  - Reroute systemic venous return directly to lungs
- Stage 1 Norwood
  - Neoaorta constructed using divided main pulmonary artery (PA) anastomosed to aortic root
  - Blalock-Taussig (BT) or Sano shunt created to provide blood flow to high-resistance pulmonary arterial circulation
  - Ductus arteriosus ligated
  - Atrial septum resected
- Stage 2 Norwood
  - BT or Sano shunt excised
  - Bidirectional Glenn shunt created
- Stage 3 Norwood
  - Lateral tunnel or extracardiac Fontan procedure performed
- ~ 70% 5-year survival of Norwood procedure
- Norwood procedure alternatives for hypoplastic left heart syndrome
  - Hybrid procedure (ductus arteriosus stenting, PA banding, & atrial septal defect ballooning in lieu of stage 1 Norwood)
  - Primary cardiac transplantation

IMAGING

- CECT can be used pre- & postoperatively to evaluate complex anatomy
- CECT useful in evaluation of complications such as mediastinitis, shunt thrombus, collaterals, & effusions
- MR most commonly used in between stages or after completion



(Left) *Graphic shows a completed 3-stage Norwood procedure. There is a neoaorta ➡ (constructed by using the patient's native main PA), a bidirectional Glenn shunt ➡ (having replaced the eliminated BT shunt), & a fenestrated lateral tunnel Fontan ➡.* **(Right)** *Color 3D CECT from a CTA shows a Norwood stage 1 with a hypoplastic native ascending aorta ⇨ combined with the main PA ➡ to form a neoaorta from a single ventricle (purple). A BT shunt (green) carries blood to the PAs (dark blue).*



(Left) *Single frontal view of the chest in a 4 month old shows a right upper extremity PICC that courses through the SVC ⇨ & then goes straight to the left PA ➡ from a Glenn shunt (SVC to PAs). A Glenn shunt is created in stage 2 of the Norwood procedure.* **(Right)** *Color 3D CECT from a CTA shows a Fontan shunt (green) carrying blood from the IVC ⇨ to the PAs (dark blue). A Glenn shunt is also present taking blood from the SVC ➡ to the PAs. Creation of a Fontan is stage 3 of the Norwood procedure. Notice the hypoplastic RV (purple).*

TERMINOLOGY

Definitions

- 3-stage procedure to palliate single ventricle physiology (most commonly hypoplastic left heart syndrome)
  - Staged approach necessary due to high pulmonary vascular resistance in neonatal period
  - Developed in early 1980s, initially as 2 stages
- Goals of Norwood procedure
  - Utilize single right ventricle (RV) [or less frequently single left ventricle (LV)] as systemic pump & reconstruct systemic arterial outflow
  - Ensure unobstructed pulmonary venous return to right heart
  - Reroute systemic venous return directly to lungs
- Stage 1 Norwood
  - Performed within 1st few days after birth
  - Neoaorta construction: Divide main pulmonary artery (PA) & anastomose to aortic root
    - Single, unobstructed arterial trunk from RV to systemic circulation now in place
  - Blalock-Taussig (BT) or Sano shunt created to provide blood flow to high-resistance PA circulation
  - Ductus arteriosus ligated
  - Atrial septum resected
    - Common atrium receives blood from superior & inferior vena cavas (SVC & IVC) & pulmonary veins
- Stage 2 Norwood
  - Performed between 3-6 months of age
    - Pulmonary vascular resistance has ↓ by this time to normal levels
  - BT or Sano shunt excised
  - Bidirectional Glenn shunt created
    - End-to-side anastomosis of SVC to PAs
- Stage 3 Norwood
  - Typically performed between 18-36 months of age
  - Lateral tunnel or extracardiac Fontan procedure performed
    - IVC blood flow directed to pulmonary arteries

IMAGING

General Features

- Complications
  - Heart failure, pleural & pericardial effusions, arrhythmias
  - Valvular dysfunction
  - Branch PA stenoses
  - Shunt thrombosis/stenosis/obstruction
  - Liver fibrosis
  - Protein-losing enteropathy
  - Pulmonary arteriovenous malformation
  - Neurologic complications
  - Unexplained sudden death

Radiographic Findings

- After stage 1: Signs of RV hypertrophy, ↓ pulmonary venous congestion, & occasional new paratracheal shadow from BT shunt
- After stage 2: Normalized pulmonary vascularity & elimination of BT shunt shadow, ↑ size of head & neck (initially) from ↑ pressure in SVC
- After stage 3: ↑ pulmonary vascularity & pleural effusions

CT Findings

- CECT
  - Can be used pre- & postoperatively to evaluate patients with complex anatomy &/or for whom long anesthesia times may be difficult
  - Useful in evaluation of complications such as mediastinitis, shunt thrombus, venous & arterial collaterals, effusions

MR Findings

- Occasionally used preoperatively in complex cases when biventricular vs. univentricular repair being contemplated
- Used frequently after stage 1, prior to bidirectional Glenn shunt
  - Assess anatomy & ventricular/valvular function
  - Can replace catheter angiography in prestage 2 evaluation
- Can be helpful after stage 2, prior to Fontan procedure
  - Glenn pathway can be evaluated in addition to ventricular & valvular function & other anatomy
  - PA sizes & anatomy can be ascertained, which affects surgical decision making for Fontan
- Very helpful after stage 3
  - Evaluate Fontan pathway patency & for presence of thrombus
  - Evaluate for PA stenoses
  - Evaluate for venous & arterial collateral formation
- MR elastography helpful in evaluating degree of liver fibrosis

DIFFERENTIAL DIAGNOSIS

Norwood Procedure Alternatives for Hypoplastic Left Heart Syndrome

- Hybrid procedure
  - In lieu of Norwood stage 1
  - Ductus arteriosus stenting, PA banding, & atrial septal defect ballooning performed
  - No cardiopulmonary bypass or thoracotomy required
  - Comparable outcomes to full Norwood
- Primary cardiac transplantation
  - ~ 70% 7-year survival
  - Lifelong immunosuppression needed

CLINICAL ISSUES

Natural History & Prognosis

- ~ 70% 5-year survival of Norwood procedure

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Arterial Switch Procedure

KEY FACTS

TERMINOLOGY

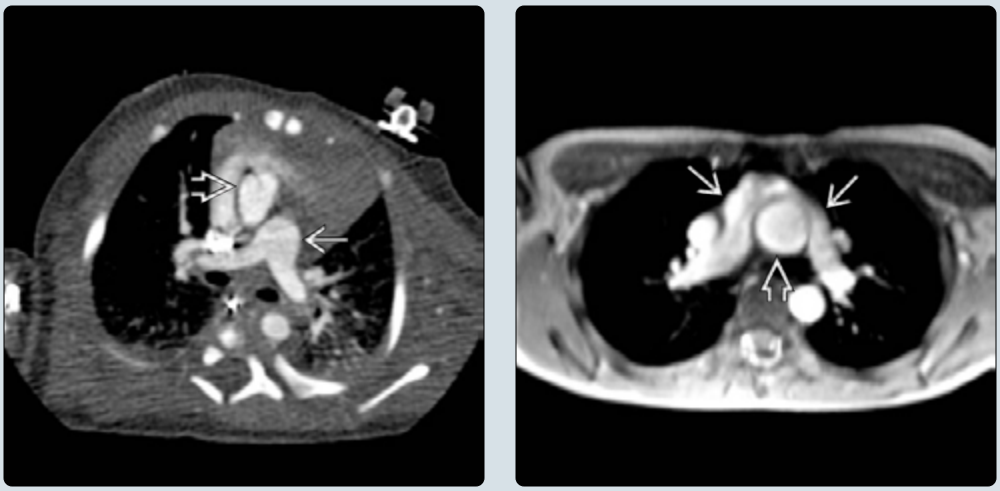
- Surgical procedure to correct D-transposition of great arteries (D-TGA)
  - Coronary arteries translocated to base of neoaorta
  - Ascending aorta & main pulmonary artery transected & transposed
    - Ascending aorta connected to left ventricular outflow tract
    - Main pulmonary artery relocated anterior to aorta & connected to right ventricular outflow tract as part of Lecompte maneuver
- Typically performed in first 2 weeks of life
- Survival rate & freedom of reoperation at 5 years: 90% & 97%, respectively

IMAGING

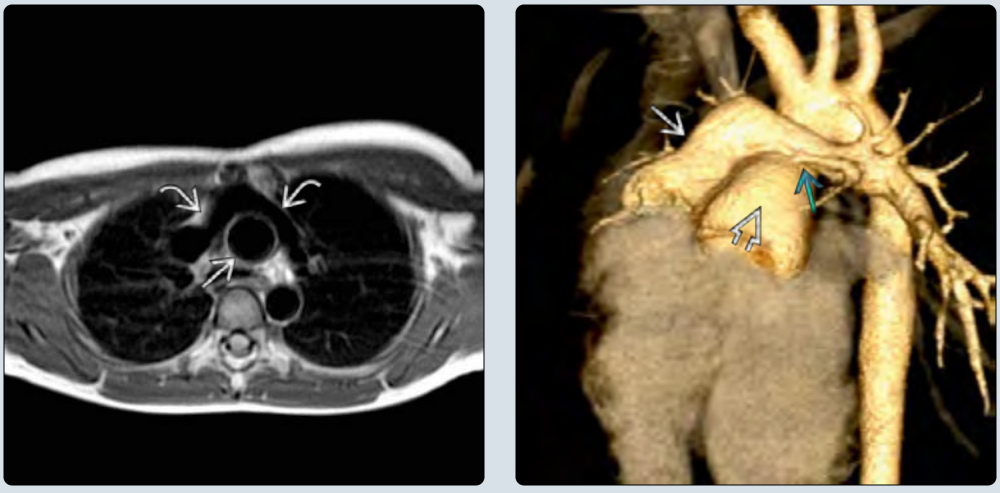
- Typical postoperative appearance: Pulmonary artery arises directly anterior to ascending aorta; branch pulmonary arteries drape over either side of ascending aorta
- Complications
  - Supravalvular pulmonic stenosis & branch pulmonary artery stenoses
  - Aortic root dilation & regurgitation
  - Ischemia secondary to coronary artery stenosis/occlusion
- CTA useful in immediate postoperative period to assess for complications & to follow status of coronary arteries & branch pulmonary arteries
- MR provides comprehensive evaluation of postoperative anatomy & function without ionizing radiation

TOP DIFFERENTIAL DIAGNOSES

- Mustard/Senning: Late atrial level repair of D-TGA with baffle
- Rastelli: Performed on subset of patients with D-TGA, ventricular septal defect, & left ventricular outflow tract stenosis/obstruction



(Left) *Preoperative axial CTA in a patient with D-transposition of great arteries (D-TGA) shows abnormal positions of the great vessel origins with the ascending aorta ➡ very anterior & mildly right of the pulmonary artery ➡.* **(Right)** *Axial magnitude MR from a cine-phase contrast acquisition in a patient with a history of D-TGA repair shows the pulmonary arteries ➡ draped over the ascending aorta ➡. This is the classic appearance of a Lecompte maneuver as part of the Jatene arterial switch procedure.*



(Left) *Axial double-inversion recovery black-blood MR demonstrates the pulmonary arteries ➡ draped over the ascending aorta ➡ in a classic Lecompte maneuver as part of a Jatene arterial switch procedure for surgical correction of D-TGA.* **(Right)** *Lateral 3D reformation status post Jatene arterial switch procedure shows the main pulmonary artery ➡ directly anterior to the ascending aorta ➡. The branch pulmonary arteries ➡ drape to either side of the ascending aorta.*

TERMINOLOGY

Synonyms

- Jatene arterial switch

Definitions

- Surgical procedure to correct D-transposition of great arteries (D-TGA)
  - TGA involves ventriculoarterial discordance & atrioventricular concordance
    - Pulmonary trunk arises from left ventricle (LV)
    - Aorta arises from right ventricle (RV)
    - Frequently associated with ventricular septal defect (VSD) & outflow tract obstruction
- Arterial switch first successfully used in 1975 by Dr. Adib Jatene
  - Coronary arteries transposed to base of neoaorta
  - Aorta & pulmonary trunks then sectioned, transposed, & anastomosed
    - Ascending aorta ends up being connected to left ventricular outflow tract
    - Main pulmonary artery (PA) relocated anterior to aorta & connected to right ventricular outflow tract
      - This relocation of pulmonary trunk referred to as Lecompte maneuver
      - Reduces risk of coronary artery kinking
- VSD corrected if present
  - If no VSD present, typically undergo PA banding prior to correction to prepare LV for systemic pressures
- Arterial switch performed in first 2 weeks of life
- If not performed early in neonatal period, PA banding ± Blalock-Taussig shunt used to acclimate LV to systemic pressures in preparation for connection to aorta
- Arterial switch contraindicated in presence of coronary anomalies such as intramural course
- Arterial switch may not be feasible in presence of significant LV outflow obstruction
  - Rastelli procedure often used instead in this situation
- Benefits of arterial switch
  - LV used as systemic pump & mitral valve as systemic atrioventricular valve
  - Lower incidence of arrhythmias compared to atrial switch (Senning/Mustard)
  - No baffle obstructions/leaks as with atrial switch (Senning/Mustard)
  - Can be performed earlier in neonatal period than atrial switch (Senning/Mustard)
  - Survival rate & freedom of reoperation at 5 years of 90% & 97%, respectively

IMAGING

General Features

- Complications
  - Supravalvular pulmonic stenosis
    - May require angioplasty or surgical correction in small percentage of patients
  - Branch PA stenoses
  - Supravalvular aortic stenosis
  - Aortic root dilation & regurgitation
  - Left ventricular dysfunction
  - Persistent pulmonary hypertension
  - Ischemia secondary to coronary artery stenosis/occlusion, typically at ostium

CT Findings

- Useful in immediate postoperative period to assess for airway compression, branch PA stenosis, & mediastinitis
- Can be utilized to evaluate coronary artery lesions such as ostial stenosis/kink

MR Findings

- Comprehensive evaluation of postoperative anatomy & function; excellent follow-up tool
  - Quantify RV & LV chamber sizes & function
  - Evaluate valvular function by flow quantification across vessels using phase-contrast imaging
  - Measure aortic root dilation & degree of aortic regurgitation
  - Delayed enhancement techniques useful for evaluation of myocardial ischemia or infarction
  - 3D SSFP sequence for coronary anatomy

DIFFERENTIAL DIAGNOSIS

Mustard/Senning

- Atrial level repair of D-TGA
- Intraatrial baffle created
  - Superior & inferior vena cava form superior & inferior limbs of systemic venous baffle, routed to morphologic LV & hence PA
  - Pulmonary veins routed to morphologic RV & hence aorta
- Mustard procedure uses autologous or synthetic pericardium to form baffle
- Senning uses native atrial tissue to form baffle
- Complications include arrhythmias, RV dysfunction, tricuspid regurgitation, pulmonary & systemic pathway stenoses, & baffle leaks/stenoses

Rastelli

- Performed on subset of patients with D-TGA, VSD, & left ventricular outflow tract stenosis/obstruction
- PA divided just above valve plane, & cardiac end closed, followed by placement of extracardiac RV to PA conduit
- Intraventricular tunnel created directing blood flow from LV through VSD into aorta

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Amplatzer Occluder Device

KEY FACTS

TERMINOLOGY

- Percutaneous transcatheter occlusion of septal defect or vessel
- 4 varieties
  - Septal occluder (for secundum atrial septal defect)
  - Muscular ventricular septal defect occluder
  - Duct occluder (for patent ductus arteriosus)
  - Vascular plug (for embolization of peripheral arteries & veins)
- Device made from nitinol mesh & polyester fabric, which provide rapid occlusion & substrate for tissue ingrowth
- Can be repositioned & recaptured during placement

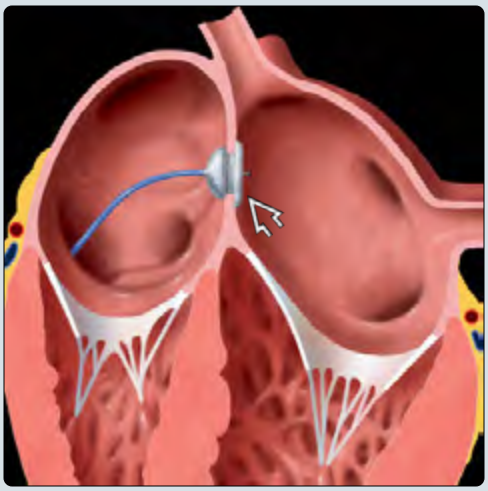
IMAGING

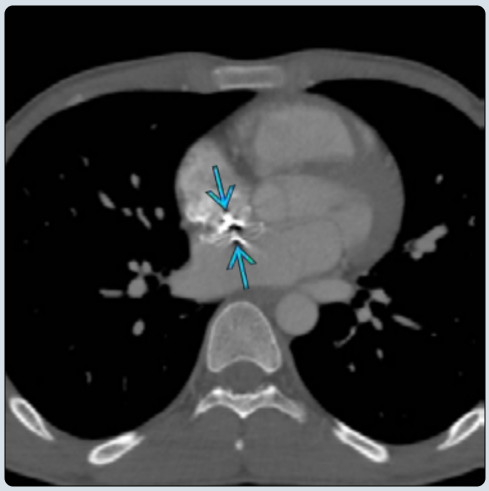
- Requires transesophageal or intracardiac echocardiography as well as fluoroscopic imaging during implantation
- Septal occluder device
  - 2 radiodense dots define ends of 3-4 mm long waist with 2 radiodense discs on either side of waist
- Ductal occluder device
  - 2 radiodense dots define ends of 5-8 mm long waist of device with single radiodense disc on aortic side of waist
- Best visualized with bone windows on CT
- Subsequent MR scanning safe up to 3.0 Tesla with maximum spatial gradient magnetic field of up to 720 Gauss/cm & maximum specific absorption rate of 3 W/kg for up to 15 minutes

CLINICAL ISSUES

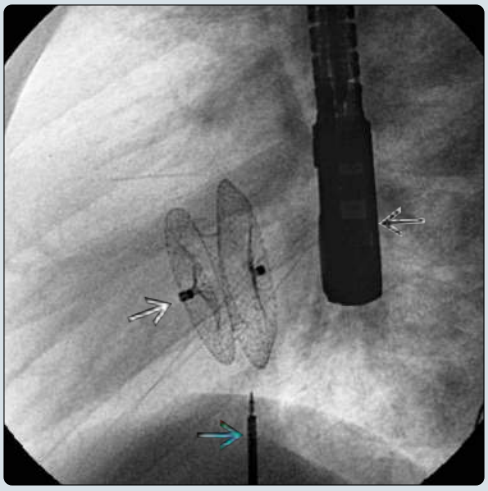
- Very high closure rates (> 95%)
- Requires endocarditis prophylaxis & antiplatelet/anticoagulation therapy for 6 months post implantation
- Early complications: Perforation of vessel or myocardium, thrombus formation, dislodgement/embolization, & percutaneous access site complications
- Late complications: Arrhythmia (most common), thrombosis, cardiac erosion, & infection or endocarditis

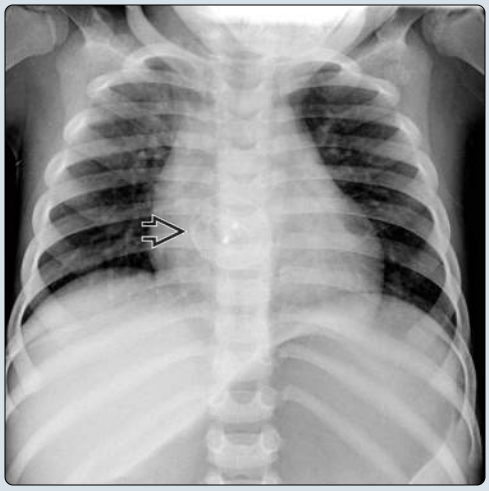
(Left) *Four-chamber view illustration shows an Amplatzer septal occluder device being placed from an inferior vena cava/right atrial approach across an atrial septal defect (ASD).* **(Right)** *Axial CECT shows a septal occluder device across an ASD. Note the 2 central metallic dots (indicating the ends of the device waist) & the flat discs on either side of the waist.*





(Left) *Lateral intraprocedural angiography image shows the recent placement of a septal occluder device across a septum secundum-type ASD. Note the probe from the transesophageal echocardiogram & the catheter in the inferior vena cava from the recent release of the device.* **(Right)** *AP chest radiograph demonstrates an Amplatzer septal occluder device projecting over the expected location of an ASD.*





TERMINOLOGY

Abbreviations

- Atrial septal defect (ASD), ventricular septal defect (VSD), patent ductus arteriosus (PDA)

Synonyms

- Septal occluder device
- Duct occluder device

Definitions

- Percutaneous, transcatheter occlusion of septal defect or vessel
- Invented by Dr. Kurt Amplatz, radiologist
- 4 varieties
  - Septal (ASD) occluder
    - FDA approved in 2001
    - Designed to close atrial septal wall on each side of secundum defect
    - Available in 4-38 mm sizes in USA
    - Multifenestrated ASDs require cribriform occluder subtype
  - Muscular VSD occluder
    - Designed to close VSD on each side of defect
    - 2 discs connected together by waist that corresponds to size of VSD
    - Available in 4-18 mm waist sizes
  - Duct occluder
    - Designed to seal PDA
    - Conical shape to conform to PDA
    - Available in 5-16 mm diameters
  - Vascular plug
    - Used for embolization of peripheral arteries & veins
    - Alternative to coils
- Device made from nitinol mesh & polyester fabric, which provide rapid occlusion & substrate for tissue ingrowth
- Sizing balloon used to determine correct size device
- Requires transesophageal or intracardiac echocardiography as well as fluoroscopic imaging during procedure
- Can be repositioned & recaptured during placement
- Very high closure rates (> 95%)
- Indications for ASD closure device
  - Ostium secundum ASD
  - Clinical evidence of right ventricular (RV) volume overload
    - Left-to-right shunt of 1.5:1 or greater, or RV chamber enlargement
- Contraindications
  - Sepsis within 1 month prior to implantation
  - Bleeding disorder or other contraindication to aspirin therapy after device placement
  - Intracardiac thrombi
  - Septal defect location < 5 mm from coronary sinus, atrioventricular valves, or pulmonary vein orifice
  - Nickel allergy (as nitinol contains nickel)
- Follow-up
  - Requires endocarditis prophylaxis & antiplatelet/anticoagulation therapy for 6 months post implantation
  - Devices "conditional 6" to 3.0 Tesla magnet

IMAGING

General Features

- Complications
  - Early complications include: Perforation of vessel or myocardium, thrombus formation, dislodgement/embolization, & percutaneous access site complications
  - Late complications include: Arrhythmia (most common), thrombosis, cardiac erosion, & infection or endocarditis

Radiographic Findings

- Septal occluder device
  - 2 radiodense dots define ends of 3-4 mm long waist of device
  - 2 radiodense flat discs on either side of waist
  - 2 dots & 2 discs may not be visualized on each view depending on projection & overlapping structures
- Ductal occluder device
  - 2 radiodense dots define ends of 5-8 mm long waist of device
  - 1 radiodense flat disc on aortic side of waist

CT Findings

- Best visualized on bone windows
- Radiodense dots (at either end of waist) & radiodense flat discs easily identified

MR Findings

- Device not evaluated/followed by MR but can be scanned immediately after placement
  - Scanned safely up to 3.0 Tesla, with maximum spatial gradient magnetic field of up to 720 Gauss/cm & with maximum specific absorption rate of 3 W/kg for up to 15 minutes
- Will cause susceptibility artifact

Ultrasonographic Findings

- Transesophageal or intracardiac echocardiography used to monitor implantation & to assess success of ASD closure

Angiographic Findings

- Catheter angiography & fluoroscopy used to define anatomy & monitor implantation

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Myocarditis

KEY FACTS

IMAGING

- Chest radiograph: Many cases normal
  - Cardiomegaly &/or pericardial effusion in setting of cardiac dysfunction
  - Pulmonary edema in more severe cases
- Echocardiography: Often 1st imaging, identifies cardiac dysfunction &/or pericardial effusion
- Cardiac catheterization & endomyocardial biopsy: Historical gold standard (but invasive & can miss patchy inflammation)
- Cardiac MR: Increasingly utilized for diagnostic capabilities
  - Cardiac volumes & functional assessment: ↓ ejection fraction with wall motion abnormality, ± pericardial effusion
  - Myocardial tissue characterization (edema, hyperemia, & fibrosis): ↑ T2 signal, early & late gadolinium enhancement, native T1 values > 990 ms
  - ≥ 2 positive criteria in acute phase: 80% sensitivity, 90% specificity

TOP DIFFERENTIAL DIAGNOSES

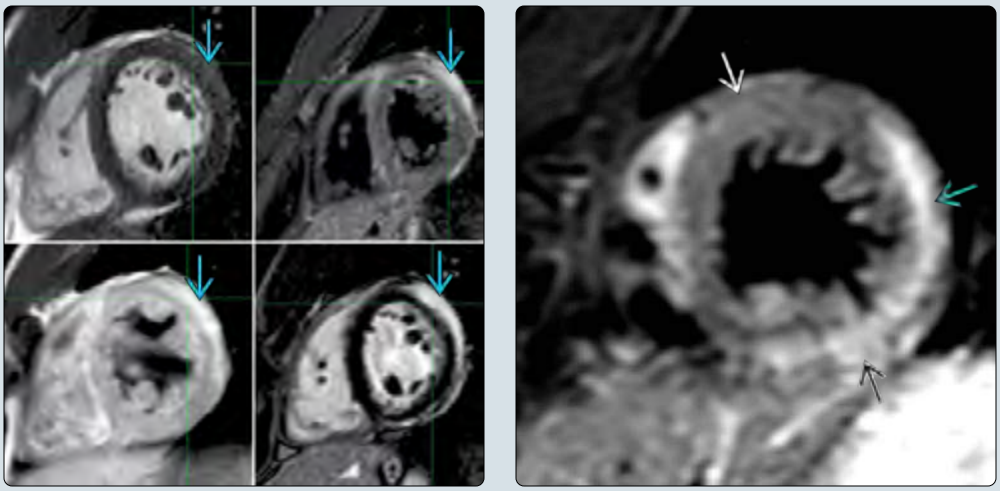
- Ischemic heart disease, inheritable cardiomyopathy, Kawasaki disease, septic shock, & sarcoidosis

PATHOLOGY

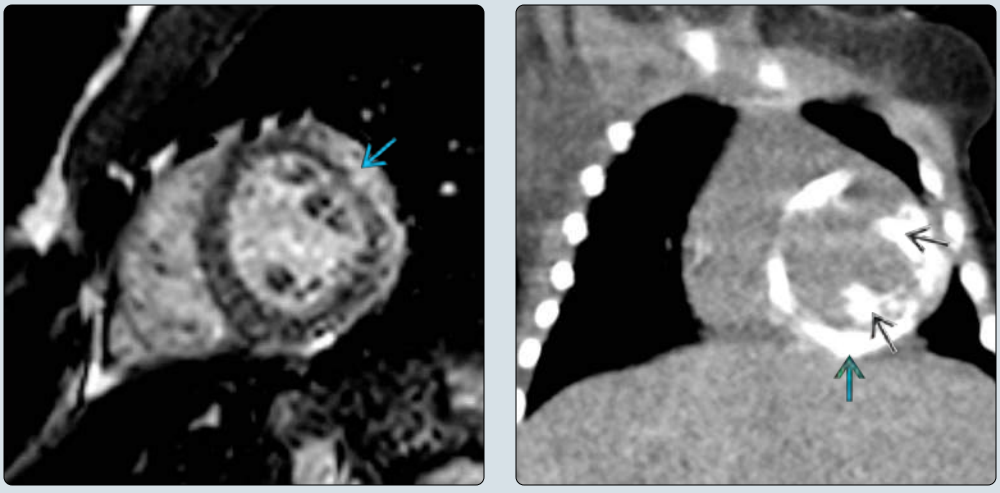
- Infectious etiology (typically viral) most common in otherwise healthy individuals
- Toxins/drug reactions: Antibiotics, antiepileptics, carbon monoxide, illegal drugs (cocaine)
- Autoimmune: Lupus, sarcoidosis, Takayasu arteritis
- Ischemic: Acute coronary syndrome/myocardial infarction

CLINICAL ISSUES

- Nonspecific symptoms: Fever, fatigue, malaise, dyspnea, muscle aches, & unexplained sinus tachycardia
- May imitate acute myocardial infarction
- Rarely, fulminant with cardiovascular collapse & shock
- May account for 10% of sudden death in young
- Most cases: Mild symptoms, need only supportive care



(Left) *Short-axis MR tissue characterization in myocarditis: A region of interest with increased signal is seen in the anterolateral segment ⇨ on SSFP (upper left), T2 (upper right), early gadolinium (lower left), & late gadolinium (lower right) enhancement images.* **(Right)** *Short-axis T2 MR demonstrates hyperintense signal in the lateral ⇨ & inferior ⇨ left ventricular segments compared to the relatively isointense signal of the remaining segments ➡ in a patient with myocarditis. The subendocardium is spared.*



(Left) *Short-axis T1 modified Look-Locker inversion recovery (MOLLI) demonstrates increased signal in the anterolateral segment of the midventricle ⇨. The post analysis native T1 value was 1047 ms, which is > the 990 ms threshold outlined for sensitivity in detecting edema in myocarditis patients.* **(Right)** *Coronal NECT demonstrates significant dystrophic calcification of the left ventricular myocardial wall ⇨ & papillary muscles ⇨ in a neonate with suspected enterovirus myocarditis.*

TERMINOLOGY

Definitions

- Inflammation of myocardium with edema, hyperemia, & fibrosis, often leading to cardiac dysfunction & pericardial effusion
  - Clinical symptoms often nonspecific

IMAGING

Radiographic Findings

- Chest radiograph findings vary based on severity
  - Many cases normal
  - Cardiomegaly &/or pericardial effusion in setting of cardiac dysfunction
  - Severe cases show interstitial &/or alveolar pulmonary edema

MR Findings

- T2WI
  - Triple inversion recovery (with inversion pulses for fat & blood) show ↑ T2 signal
    - Quantitative analysis of signal intensity more sensitive
      - Identification of edema throughout myocardium
    - Limited sensitivity in mild cases
- T1WI C+
  - Early gadolinium enhancement (EGE)
    - Hyperemia & capillary leak show enhancement 1 minute post-gadolinium
  - Late gadolinium enhancement (LGE)
    - Fibrosis & necrosis
      - May be irreversible
    - Differentiates ischemic injury (always subendocardial) from nonischemic injury
- Native T1 mapping demonstrates location, extent, & pattern of myocarditis
  - Threshold of T1 > 990 ms (sensitivity & specificity ~ 90%) detects significantly larger areas of involvement than T2WI & LGE

Echocardiographic Findings

- Often 1st imaging performed; neither sensitive nor specific
  - Mild cases: ↓ function ± wall motion abnormalities, ± pericardial effusion
  - Moderate to severe cases: ↓ function with associated wall motion abnormalities, left ventricular chamber dilation, ↑ wall thickness, ± pericardial effusion

Imaging Recommendations

- Best imaging tool
  - Cardiac MR: Functional assessment with myocardial tissue characterization
- Protocol advice
  - SSFP 2-chamber, 3-chamber, 4-chamber, & short-axis stack
  - Tissue characterization with precontrast T2 3-slice short axis (edema) & postcontrast 3-slice short-axis EGE (hyperemia) & LGE (fibrosis)
  - Native T1 values: Most accurate detection of myocarditis

DIFFERENTIAL DIAGNOSIS

Cardiac MR Findings

- Ischemic heart disease, inheritable cardiomyopathy, Kawasaki disease, septic shock, & sarcoidosis

PATHOLOGY

General Features

- Etiology
  - Infectious etiology most common if otherwise healthy
    - Direct viral infection of myocardium: Coxsackie B virus, adenovirus, parvovirus B19, echoviruses, Ebstein-Barr
      - May see dystrophic Ca^{2+} in neonates with enteroviral myocarditis
    - Immune-mediated postinfectious reaction
    - Chagas disease: *Trypanosoma cruzi* (Central & South America)
  - Other causes
    - Toxins/drug reactions: Antibiotics, antiepileptics, carbon monoxide, illegal drugs (cocaine)
    - Autoimmune: Lupus, Takayasu arteritis, Wegener granulomatosis, giant cell arteritis

Microscopic Features

- Endomyocardial biopsy in severe cases when definitive diagnosis required, otherwise not recommended
  - Histopathology shows infiltration of inflammatory cells
  - Immunohistochemistry: Best sensitivity but less available
  - Viral genome analysis: PCR for most common viruses

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often nonspecific with fatigue, dyspnea, muscle aches, unexplained tachycardia, or ventricular ectopy
- Other signs/symptoms
  - Many cases asymptomatic, no medical care sought
  - May imitate acute myocardial infarction
  - Rarely fulminant with cardiovascular collapse & shock
  - Infants & small children: Dyspnea, poor feeding, & fever
    - Mistaken for reactive airway disease or pneumonia
  - ~ 10% of sudden cardiac deaths in young

Treatment

- Most cases have mild symptoms, need only supportive care
- With cardiac dysfunction, treatment similar to other causes of heart failure
  - Drugs that stimulate heart should be avoided
  - Complete heart block requires temporary pacing
- Intravenous immunoglobulin: Variable results

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Left Ventricular Noncompaction

KEY FACTS

TERMINOLOGY

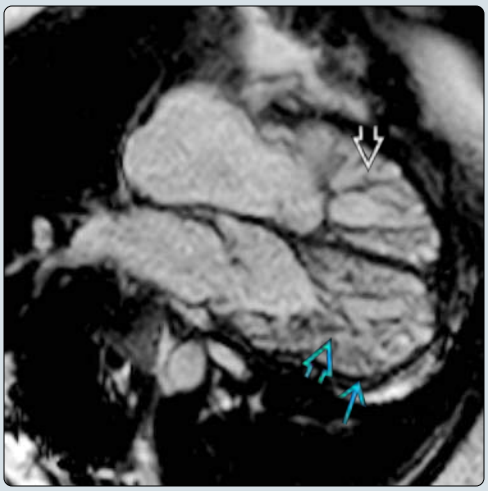
- Left ventricular noncompaction cardiomyopathy (LVNC): Cardiomyopathy characterized by sponge-like appearance of left ventricular (LV) myocardium
  - 2-layer appearance: Heavily trabeculated noncompacted (NC) layer & thin, dense compacted (C) layer
  - NC:C ratio > 2.3:1 strongly suggestive
- Controversial diagnosis due to lack of definitive criteria & variable prognostic data
  - ↑ in diagnosis has occurred with improved echocardiogram & MR imaging quality
- Primitive appearance of myocardium suggests potential arrest in fetal LV myocardial development
  - LVNC also seen in association with other cardiomyopathies & congenital heart disease

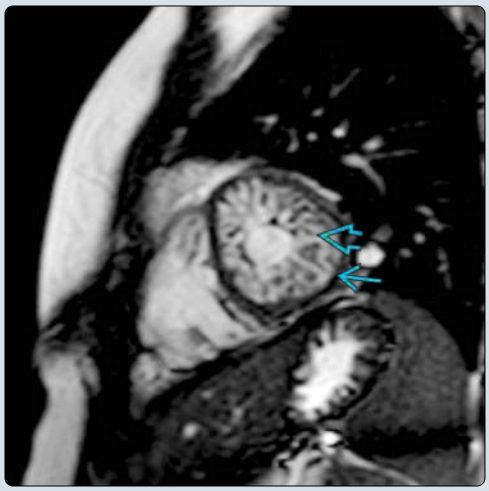
IMAGING

- Echocardiography widely used; however, cardiac MR (CMR) better at characterizing location & extent of trabeculations
- CMR defines location & extent of trabeculations, ventricular volumes & ejection fraction, fibrosis (with late gadolinium enhancement), & apical thrombus
- Multitude of imaging "criteria" proposed; however, varying results of imaging findings compared to prognostic data

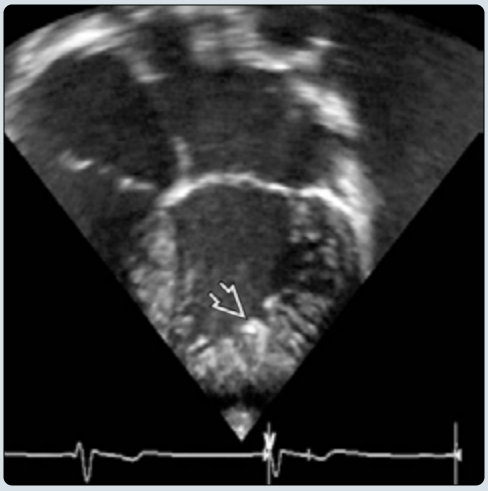
CLINICAL ISSUES

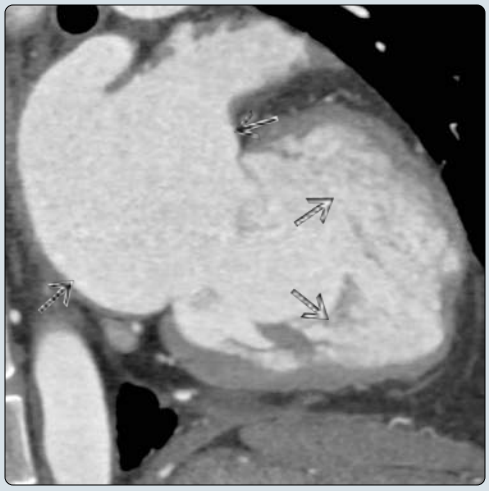
- Highly variable clinical presentations described, ranging from completely asymptomatic to symptomatic with heart failure, cardioembolic events, & ventricular arrhythmias
  - Mural thrombus develops within deep trabeculations of LV → significant risk factor for cardioembolic events
- Predictors of prognosis may include number of affected segments, heart failure at presentation, & ventricular arrhythmias
- Treatment aimed at treating or preventing heart failure, ventricular arrhythmias, & embolic stroke





(Left) *Four-chamber SSFP MR shows marked left ventricular hypertrabeculation of the noncompacted layer & a thin, dense compacted layer in a patient with left ventricular noncompaction cardiomyopathy (LVNC). Also note the hypertrabeculated right ventricle (RV), suggesting biventricular NC.* **(Right)** *Short-axis SSFP MR in end-diastole demonstrates marked LV hypertrabeculation of the noncompacted layer & a thin, dense appearance of the compacted layer in a patient with LVNC.*





(Left) *Four-chamber echocardiogram in a child with a ventricular septal defect shows a thick noncompacted layer that includes the apex.* **(Right)** *Vertical long axis cardiac CTA during end-diastole show prominent trabeculation in the mid & apical aspects of the left ventricle. The ratio of noncompacted to compacted myocardium is greater than 2.3:1, which is highly suggestive of LVNC. The left atrium is dilated.*

TERMINOLOGY

Abbreviations

- Left ventricular noncompaction cardiomyopathy (LVNC)

Synonyms

- Noncompacted left ventricle (NCLV), left ventricular hypertrabeculation, spongy myocardium

Definitions

- Inherited cardiomyopathy characterized by extensive hypertrabeculation of left ventricular (LV) myocardium
- 2-layer appearance of LV with sponge-like noncompacted (NC) inner layer & thinner, dense compacted (C) outer layer
- Primitive appearance of myocardium suggests potential arrest in fetal LV myocardial development

IMAGING

General Features

- Best diagnostic clue
  - LV myocardium in 2 layers: Thick hypertrabeculated inner layer (NC) & thin, dense outer layer (C)
  - NC:C thickness ratio > 2.3:1 in patients with LVNC
- Location
  - Typically LV with coalescent pattern in inferior & lateral segments of apical & midregions; rare in basal region
  - Normally RV somewhat trabeculated; can be heavily trabeculated in severe LVNC

Radiographic Findings

- Cardiomegaly in patients with heart failure

MR Findings

- SSFP cine
  - 2-, 3-, & 4-chamber views & short-axis stack best sequences to locate segment distribution & extent of trabeculations
  - NC:C thickness ratio > 2.3:1 strongly suggestive; measured in end-systole or end-diastole
  - ↑ LV end-diastolic volume (EDV): Contours performed on compacted layer in end-diastole
  - ↓ LV ejection fraction: Challenging to contour in end-systole due to heavy trabeculations coalescing
  - Can determine trabecular mass-to-total mass ratio
- Delayed enhancement
  - Myocardial fibrosis assessment by late gadolinium enhancement (LGE)
    - Difficult to accurately depict given deep trabeculations
      - Requires multiple slice planes
    - ± wall motion abnormalities if LGE positive

Echocardiographic Findings

- Primary diagnostic modality; 3 descriptions
  - Minor trabeculation: Prominent trabeculation in posteroapical LV in noncoalescent pattern; NC:C ratio < 2.3
  - Major trabeculation: Prominent trabeculation in posteroapical LV in coalescent pattern; NC:C ratio < 2.3
  - Noncompaction: Prominent trabeculation in posteroapical LV in coalescent pattern; NC:C ratio > 2.3
- Color Doppler demonstrates flow within trabeculations & communication with ventricular cavity
- Evaluate for septal defects, apical thrombus, LV systolic &/or diastolic dysfunction

Imaging Recommendations

- Best imaging tool
  - Cardiac MR with SSFP 2-, 3-, & 4-chamber views & short-axis stack; LGE for fibrosis
- Protocol advice
  - Artifact from parallel imaging may obscure trabeculations; best images without parallel imaging

DIFFERENTIAL DIAGNOSIS

Normal With Prominent Trabeculations

- Likely normal if NC:C ratio < 2.3, noncoalescent trabecular pattern, normal ejection fraction & LV EDV, absence of symptoms

Dilated Cardiomyopathy

- Hypertrophy of cardiac trabeculations can imitate noncompaction; cardiac apex not involved in dilated cardiomyopathy

Hypertrophic Cardiomyopathy

- Most commonly seen in apical hypertrophic cardiomyopathy (HCM); ↑ myocardial mass in HCM compared to LVNC

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Tachypnea due to low cardiac output or heart failure
    - Infants & small children with tachypnea, cyanosis, acute life-threatening event, or failure to thrive
    - Older patients with exertional dyspnea
  - Tachyarrhythmias; most have ECG abnormalities
  - Cardioembolic event

Demographics

- Familial in ~ 50%
- M > F, 3:2 ratio

Natural History & Prognosis

- Variable natural history & prognostic data → controversy in diagnosis
- Predictors of prognosis may include heart failure at presentation, systolic/diastolic dysfunction, thromboembolic events, & ventricular arrhythmias

Treatment

- Aimed at preventing/treating heart failure, ventricular arrhythmias, & stroke; new therapies target LV remodeling

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3. Towbin JA et al: Left ventricular non-compaction cardiomyopathy. Lancet. 386(9995):813-25, 2015

Hypertrophic Cardiomyopathy

KEY FACTS

TERMINOLOGY

- Familial cardiomyopathy characterized by thickened but nondilated LV & no identifiable systemic or cardiac cause
- Most common genetic cardiomyopathy (1 in 500)

IMAGING

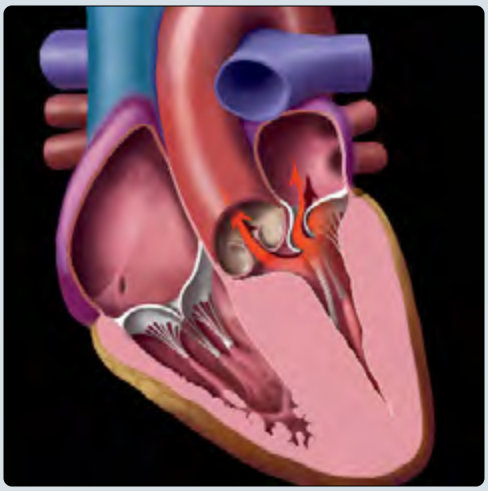
- Echocardiography primary diagnostic/screening tool
- Cardiac MR increasingly utilized for ventricular dimensions, LVOT obstruction, & late gadolinium enhancement (LGE)
  - Asymmetric septal hypertrophy, typically of basal septum
    - Septal wall thickness > 15 mm (or > 13 mm with positive family history)
    - > 30 mm risk factor for sudden cardiac death (SCD)
  - Dynamic LVOT obstruction
    - Severe thickening of septum during systole causing midcavitary obstruction
    - Systolic anterior motion of mitral valve
  - Systolic & diastolic dysfunction variable
  - Patchy or focal LGE (from fibrosis) of basal & midventricle septal segments
    - Risk factor for ventricular arrhythmia & SCD

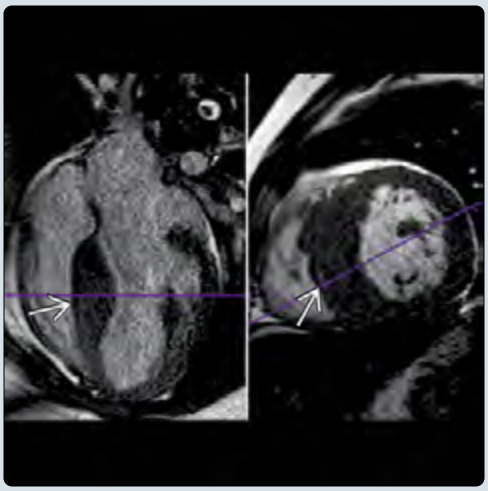
PATHOLOGY

- Primary: Typically autosomal dominant inheritance
- Secondary: Associated with syndromic, neuromuscular, & metabolic disorders
- Idiopathic: ~ 50% of affected children < 1 year old

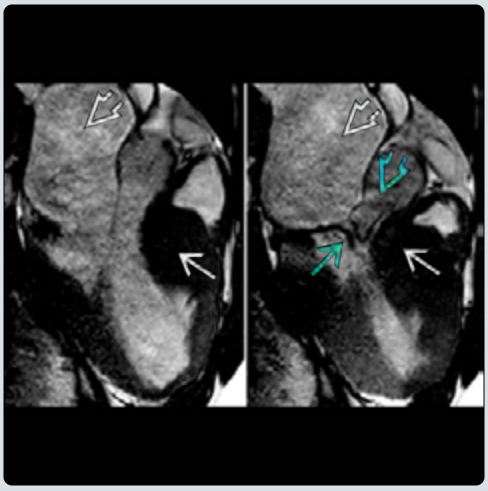
CLINICAL ISSUES

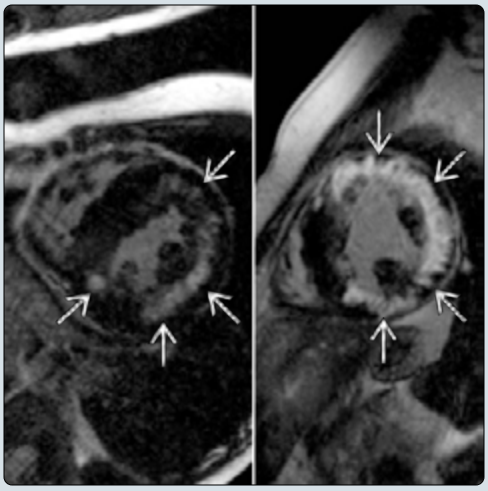
- Typical symptoms include dyspnea, chest pain, or syncope, typically with exertion
  - Ventricular arrhythmias common
  - Increasing intensity in systolic heart murmur with Valsalva, positional change (standing/squatting), or exercise
  - Leading cause of SCD in youth
- Treatments include β-blockers, antiarrhythmics, pacemaker, septal ablation, myomectomy





(Left) *Five-chamber graphic demonstrates concentric hypertrophic obstructive cardiomyopathy. Septal hypertrophy & systolic anterior motion (SAM) of the mitral valve leaflet combine to cause left ventricular outflow tract (LVOT) obstruction & mitral regurgitation.* **(Right)** *Four-chamber & orthogonal short-axis SSFP MRs demonstrate severe asymmetric septal hypertrophy. Both views are utilized to obtain a correlative septal measurement (along the purple line).*





(Left) *Three-chamber SSFP MRs during diastole (left) & systole (right) demonstrate LVOT obstruction from severe septal hypertrophy & SAM of the mitral valve apparatus. Also note the left atrial enlargement.* **(Right)** *Short-axis late gadolinium enhanced MRs of the same patient show progression of fibrosis on a follow-up study. The delayed enhancement is severe (though patchy) on the 1st image (left), becoming more extensive & nearly full thickness on the follow-up exam (right).*

TERMINOLOGY

Abbreviations

- Hypertrophic cardiomyopathy (HCM)

Synonyms

- Idiopathic hypertrophic subaortic stenosis, hypertrophic obstructive cardiomyopathy

Definitions

- Familial cardiomyopathy with thickening of left ventricular (LV) myocardium without other cardiac or systemic disease; typically involves ventricular septum

IMAGING

General Features

- Best diagnostic clue
  - Significant thickening of LV myocardium, typically septal
    - Diastolic wall thickness ratio: Thickest/thinnest > 2.0
- Location
  - Anteroseptal portion of basal LV most commonly involved; concentric/symmetric or apical secondarily involved
- Morphology
  - 3 types: Asymmetric septal, concentric, or apical

Radiographic Findings

- Typically normal chest radiograph
- Left atrial dilation with mitral regurgitation, diastolic issues
- Cardiomegaly evident in later stages with chamber dilation

MR Findings

- SSFP cine
  - 3-chamber, 4-chamber, & short-axis stack best sequences for ventricular dimensions & ejection fraction
  - Asymmetric septal hypertrophy, typically basal septum
    - Septal wall thickness > 15 mm (or > 13 mm with positive family history)
    - LV thickness > 30 mm: Risk factor for sudden cardiac death (SCD)
  - Mechanism of left ventricular outflow tract (LVOT) obstruction
    - Severe septal thickening → midcavitary obstruction
    - Systolic anterior motion of mitral valve apparatus → anterior leaflet & chordae pulled into LVOT via Venturi effect
- Myocardial late gadolinium enhancement (LGE)
  - Patchy LGE (from fibrosis) with occasional focal areas (anteroseptal > inferoseptal)
    - Positive LGE = poor prognostic indicator

Imaging Recommendations

- Best imaging tool
  - Echocardiography: Primary diagnostic/screening tool
    - Better for diastolic dysfunction & LVOT gradient
  - Cardiac MR: Secondary diagnostic/prognostic tool
    - Better for ventricular wall dimensions & fibrosis

DIFFERENTIAL DIAGNOSIS

Secondary Left Ventricular Hypertrophy

- Typically concentric
- Due to hypertension, aortic stenosis, coarctation

Athlete's Heart (Physiologic Hypertrophy)

- LV cavity size near normal or mildly ↑ (> 55 mm)

Left Ventricular Noncompaction

- Inner thick trabecular (noncompacted) myocardial layer + thin outer compacted layer
- Similar genetic abnormalities to HCM

PATHOLOGY

General Features

- Genetics
  - Familial form typically autosomal dominant (AD) with de novo mutations making up small % of cases
  - 70% of genes involved in HCM encode β-myosin heavy chain & myosin-binding protein C; remaining in troponin T or I, α-actin, or multiple other genes

Staging, Grading, & Classification

- Primary: Familial HCM (AD inheritance)
- Secondary: Syndromic, neuromuscular, & metabolic
- Idiopathic: ~ 50% of affected children < 1 year of age

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic (often discovered via family history)
  - Physical exam: Systolic heart murmur that ↑ in intensity with position changes (squatting), Valsalva, or exercise; prominent LV apical impulse
  - Red flags: Exertional symptoms including dyspnea, chest pain, or syncope; arrhythmias (typically ventricular)
  - SCD: Most common cause in children is HCM

Demographics

- Ethnicity
  - African American > Caucasian or Latino
- Epidemiology
  - Most common genetic heart disease (1:500)
  - Most common childhood cardiomyopathy (42%)

Natural History & Prognosis

- Variable clinical course with 1% annual mortality

Treatment

- Medical management
  - β-adrenergic blocking agents limit heart rate at baseline & with exertion
  - Antiarrhythmics reduce potential for arrhythmic events
- Invasive: Pacemaker, septal ablation, myomectomy

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Duchenne Muscular Dystrophy-Related Cardiomyopathy

KEY FACTS

TERMINOLOGY

- Dystrophin-associated inherited disorders with progressive muscle wasting & weakness

IMAGING

- Cardiac MR (CMR) important for early detection of Duchenne muscular dystrophy (DMD)-associated cardiomyopathy
  - Initially, normal function with normal ejection fraction (EF)
  - Late gadolinium enhancement (LGE) from fibrosis in left ventricle (LV), typically in inferolateral segments
  - ↓ EF over time with progressive LV dilation
  - CMR allows much earlier detection of cardiac involvement than echocardiography

PATHOLOGY

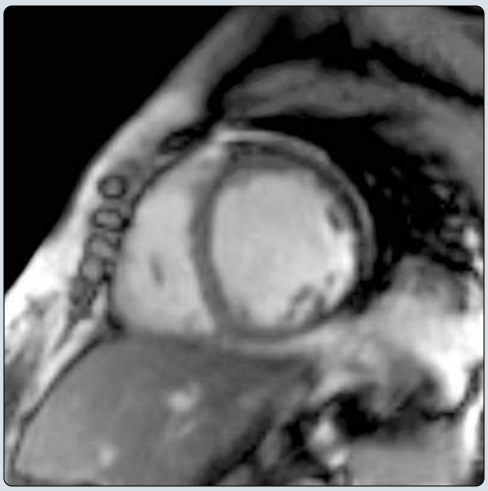
- Dystrophinopathies
  - DMD: Absent dystrophin
  - Becker MD: Abnormal/reduced dystrophin
- X-linked disorders occurring in males
- Dystrophin is large, sarcolemmal glycoprotein connecting actin filaments & extracellular matrix
- Dystrophin plays role in stabilizing plasma membranes
  - Absence, reduction, or dysfunction → sarcolemmal fragility → myocyte degeneration & death
  - End result is fibrosis & myocardial dysfunction

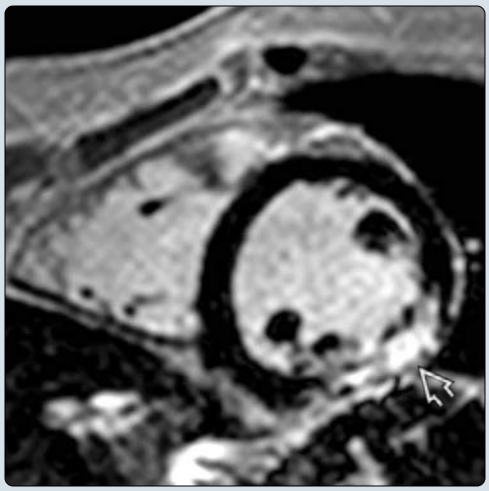
CLINICAL ISSUES

- In past, DMD patients often died of respiratory complications; improved respiratory therapy → more cardiac complications
- Patients often asymptomatic until late in heart failure (as immobility from skeletal muscle weakness obscures typical symptoms of heart failure)

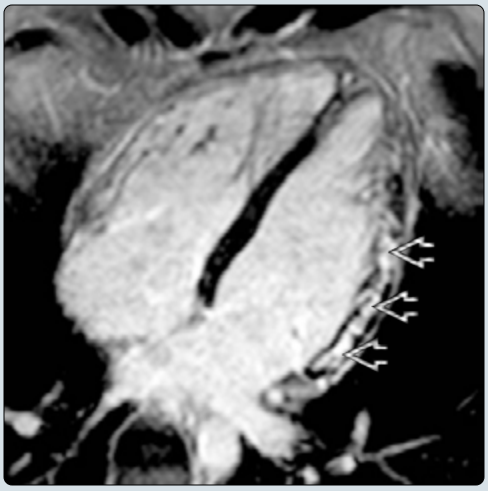
DIAGNOSTIC CHECKLIST

- LGE is usually subepicardial in inferolateral segments
- Myocardial dysfunction is progressive





(Left) *Short-axis SSFP cine MR at end diastole shows a dilated left ventricle in this patient with end-stage dilated cardiomyopathy due to DMD. The end-systolic phase in the same plane had a similar appearance, estimating a severely depressed ejection fraction (EF).* **(Right)** *Short-axis mid ventricle late gadolinium enhancement (LGE) MR imaging, performed using 3D phase-sensitive inversion recovery, shows LGE ⇨ of fibrosis located in the inferolateral wall, which is a typical pattern seen in DMD.*



(Left) *Four-chamber view of LGE in the myocardium of a 19-year-old man with DMD shows intense, well-defined enhancement ⇨ of fibrosis in the inferolateral wall. The enhancement is usually less intense & patchy.* **(Right)** *Four-chamber tagged cine MR is useful for detecting subclinical & regional myocardial dysfunction but should only be employed if able to post process for strain values. Currently, this technique is mainly used to investigate subtle changes in response to therapeutics.*

TERMINOLOGY

Abbreviations

- Duchenne muscular dystrophy (DMD)
- Becker muscular dystrophy (BMD)

Definitions

- Dystrophin-associated inherited disorders with progressive muscle wasting & weakness

IMAGING

General Features

- Best diagnostic clue
  - Muscle biopsy or genetics to confirm DMD prior to presenting for cardiac imaging
  - Subepicardial late gadolinium enhancement (LGE) in left ventricle (LV) inferolateral wall
  - ↓ ejection fraction (EF)

Echocardiographic Findings

- Used to follow ventricular size & function
  - Limited due to lung artifact, pectus deformity, scoliosis, ↑ fat due to steroids

MR Findings

- No heart failure symptoms due to nonambulatory state
- Progressive LV dysfunction develops over time
  - Initially, normal function with normal EF
  - LGE in LV, typically in inferolateral segments
    - Can occur with normal EF or ↓ EF
  - ↓ EF over time with progressive LV dilation

Imaging Recommendations

- Best imaging tool
  - Cardiac MR (CMR) with LGE assessment
    - Much more reproducible than echocardiography
    - Typical short-axis cine bright blood stack for LV function & volume
    - LGE can detect areas of damaged myocardium
      - Often precursor to more rapid decline in function
    - Specialized strain analysis utilizing tracking or tagging techniques can detect LV dysfunction before EF ↓
    - CMR provides prognostic information
- Protocol advice
  - Perform standard functional analysis, which provides left ventricular end-diastolic volume, end-systolic volume, EF, & myocardial mass
  - LGE performed in short-axis plane at minimum
    - 2-chamber, 4-chamber, & 3-chamber LGE images very helpful

DIFFERENTIAL DIAGNOSIS

Cardiomyopathy of Other Causes

- Acute viral myocarditis
- Hypertrophic cardiomyopathy
- Noncompaction cardiomyopathy

PATHOLOGY

General Features

- Etiology
  - Dystrophinopathies: DMD with absent dystrophin; BMD with abnormal/reduced dystrophin
    - X-linked disorders occurring in males
    - Dystrophin: Large, sarcolemmal glycoprotein connecting actin filaments & extracellular matrix
    - Dystrophin plays role in stabilizing plasma membranes
      - Anecdotally described as shock absorber
      - Dystrophin absence or dysfunction → sarcolemmal fragility → myocyte degeneration & death
      - End result is fibrosis & myocardial dysfunction

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - In past, DMD patients often died of respiratory complications; improved respiratory therapy → more cardiac complications
  - Often no heart failure symptoms until later due to immobility from skeletal muscle weakness
  - Nearly all dystrophinopathy patients develop cardiomyopathy by 3rd decade
  - Death typically related to cardiopulmonary complications in 3rd decade
  - CMR important for early detection of DMD-associated cardiomyopathy
    - Provides prognostic information
    - CMR allows much earlier detection of cardiac involvement than echocardiography
    - Cardioprotective medical therapies slow progression

Demographics

- Age
  - Progressive muscle weakness manifests in childhood with ambulation (Gower sign)
  - Loss of ambulation by teenage years
    - 50% by 8-10 years of age
  - By 20 years of age, nearly all have dilated cardiomyopathy
- Gender
  - X-linked disorder afflicting males
- Epidemiology
  - Incidence of 1 in 3,500 males

Natural History & Prognosis

- DMD has more rapid skeletal myopathy than BMD
- BMD experience worse cardiomyopathy than DMD
- Cardiomyopathy is progressive but can be modified by medical therapies that improve quality of life & survival

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KEY FACTS

TERMINOLOGY

- Disturbance of normal left-right asymmetry in position of thoracic & abdominal organs; typically described in terms of right atrial isomerism vs. left atrial isomerism

IMAGING

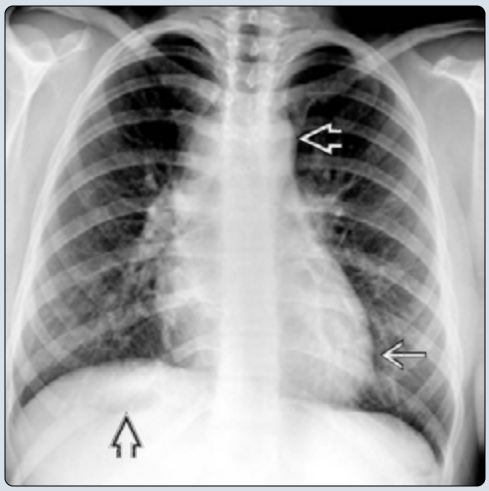
- Best diagnostic clue: Abnormal symmetry in chest & abdomen
- Classic plain film appearance: Transverse midline liver, discrepancy between position of cardiac apex & stomach, bilateral left- or right-sidedness in chest, cardiomegaly or other findings of congenital heart disease
- CTA: Rapid examination of chest & abdomen for abnormalities of situs, systemic & pulmonary venous connections, tracheobronchial anatomy
- Multiplanar MR for segmental analysis of intracardiac connections & defects
  - Gadolinium-enhanced 3D MRA: Comparable to CTA
- Upper GI study: Malrotation is frequently associated

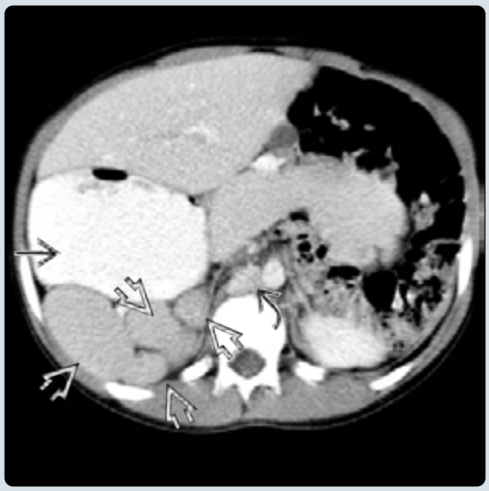
PATHOLOGY

- Any arrangement other than situs solitus or inversus is termed situs ambiguous
- Heterotaxy syndrome represents spectrum with overlap between classic asplenia & polysplenia manifestations & other anomalies

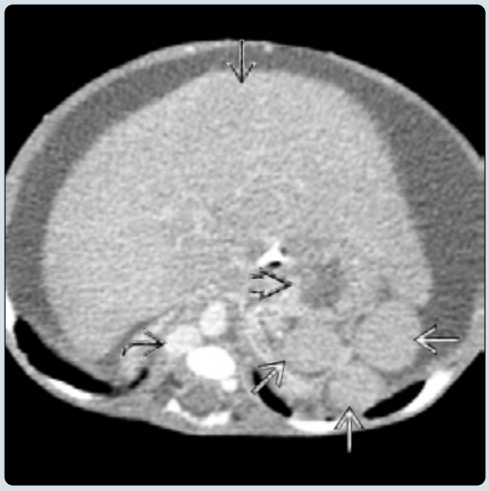
CLINICAL ISSUES

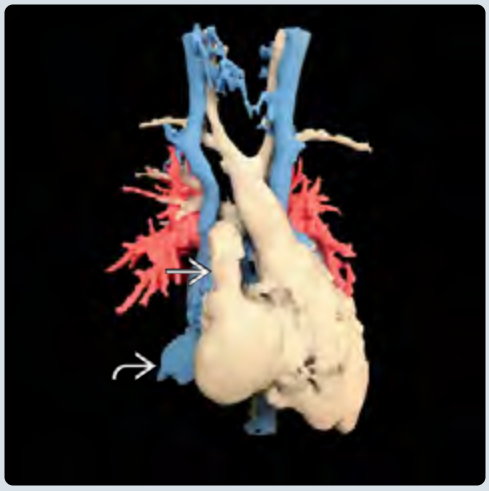
- Asplenia
  - Male neonate with severe cyanosis, susceptibility for infections, severe congenital heart disease
- Polysplenia
  - Less severe cardiac disease (i.e., systemic venous malformations, atrial septal defect), often presents later





(Left) *PA chest radiograph demonstrates a left-sided cardiac apex, a left-sided aortic arch, & a right-sided stomach bubble in this patient with situs ambiguous.* **(Right)** *Axial CECT through the upper abdomen in a child with heterotaxy shows a right-sided stomach & multiple spleens. An enlarged azygous vein is noted, but no intrahepatic inferior vena cava (IVC) is visualized, consistent with azygous continuation of the IVC.*





(Left) *Axial CECT through the upper abdomen in a child with heterotaxy demonstrates a midline liver, a left-sided stomach, multiple spleens, & an enlarged azygous vein without an intrahepatic IVC, consistent with azygous continuation of the IVC.* **(Right)** *3D-printed model of a patient with heterotaxy shows bilateral SVCs (blue), interrupted IVC with azygous continuation, hepatic veins draining directly to the RA, & complex pulmonary venous return (red). The 3D model was created for surgical planning.*

TERMINOLOGY

Synonyms

- Situs ambiguous, right/left isomerism, cardiosplenic syndromes, Ivemark syndrome

Definitions

- Disturbance of normal left-right asymmetry in position of thoracic & abdominal organs

IMAGING

General Features

- Best diagnostic clue
  - Abnormal symmetry in chest & abdomen

Radiographic Findings

- Radiography
  - Classic appearance: Transverse midline liver, discrepancy between position of cardiac apex & stomach, bilateral left- or right-sidedness in chest, cardiomegaly or other findings of congenital heart disease (CHD)
  - Asplenia syndrome or right atrial isomerism
    - Bilateral minor fissures
    - Symmetrical short main bronchi with right-sided morphology (narrow carinal angle, early take-off of upper lobe bronchus)
    - Bilateral eparterial bronchi: Main bronchus is superior to branch pulmonary artery (eparterial bronchus)
    - Cardiomegaly, pulmonary edema
  - Polysplenia syndrome or left atrial isomerism
    - No minor fissure on either side
    - Symmetrical long main bronchi with left-sided morphology (wide carinal angle)
    - Bilateral hyparterial bronchi: Main bronchus is inferior to branch pulmonary artery (hyparterial bronchus)
    - Absent inferior vena cava (IVC) shadow on lateral film, prominent azygous shadow on AP
  - Both syndromes
    - Cardiac malposition (40%: Mesocardia, dextrocardia)
    - Transverse liver
    - Right-sided stomach bubble with levocardia, left-sided stomach bubble with dextrocardia, or midline stomach

CT Findings

- CTA
  - Rapid examination of chest & abdomen: Situs abnormalities, systemic & pulmonary venous connections, tracheobronchial anatomy
  - Best for postoperative patients with metallic coils, stents, & clips
  - Preferred modality over conventional angiography due to more detailed anatomic information

MR Findings

- T1WI
  - Multiplanar imaging for segmental analysis of intracardiac connections & defects
- T2* GRE
  - Cine MR for ventricular volumes & function to determine suitability for biventricular vs. univentricular repair
- MRA
  - Ultrafast time-resolved MRA C+ with repeated acquisitions allows for dynamic circulation study
  - Velocity-encoded phase contrast MRA for flow quantification

Echocardiographic Findings

- Initial diagnostic test for characterization of intracardiac anomalies, abnormal systemic &/or pulmonary venous connections

Other Modality Findings

- Upper GI study: Malrotation frequently associated

Imaging Recommendations

- Protocol advice
  - Echocardiography, followed by MR
  - CTA for anatomic study in postoperative patients

DIFFERENTIAL DIAGNOSIS

Situs Inversus Totalis (I, L, L)

- Mirror image of normal
- Low association with CHD (3-5%)
- May be associated with immotile cilia syndrome (Kartagener): Sinusitis, bronchiectasis, infertility

True Dextrocardia + Abdominal Situs Solitus or Levocardia + Abdominal Situs Inversus

- Both have high association with CHD (95-100%)

Dextroversion of Heart

- Heart positioned in right chest with apex & stomach still directed toward left
  - Right lung hypoplasia (scimitar syndrome)
  - Left-sided mass lesions
    - Diaphragmatic hernia
    - Congenital pulmonary airway malformation

PATHOLOGY

General Features

- Heterotaxy syndrome represents spectrum with overlap between classic asplenia & polysplenia manifestations & other anomalies
- Embryology: Early embryological disturbance (5th week of gestation) leading to complex anomalies
- Pathophysiology: Determined by complexity of associated CHD
- Genetics: No specific genetic defect in majority (usually sporadic)

Staging, Grading, & Classification

- Segmental approach to analysis of complex cardiac anomalies with cardiac malposition (reported by Anderson or Van Praagh systematic descriptions)
  - Visceroatrial situs designated by S (solitus = normal) or I (inversus = mirror image of normal)
  - Always associated on same side
    - Major lobe of liver, IVC, anatomic right atrium, trilobed lung, eparterial bronchus
    - Spleen, stomach, descending aorta, anatomic left atrium, bilobed lung, hyparterial bronchus
  - Any arrangement other than situs solitus or inversus is termed situs ambiguous (heterotaxia)

- Ventricular loop: D loop (normal) or L loop (inverted)
- Orientation of great arteries (presence of transposition) also designated by D or L
- Van Praagh segmental analysis summarized by 3-letter code describing relationships of atria, ventricles, & great arteries: S,D,D; I,L,L; & S,D,L
- Anderson connections described as concordant or discordant between atria, ventricles, & great arteries
- Associated abnormalities: Transposition of great arteries (TGA), double-outlet right ventricle (DORV), total anomalous pulmonary venous return (TAPVR), unbalanced atrioventricular septal defects

- 2 major subtypes
  - Asplenia syndrome = right atrial isomerism or double right-sidedness
    - Absence of spleen
    - IVC & aorta on same side
    - Bilateral superior vena cavae (~ 36%); absent coronary sinus
    - Right isomerism of atrial appendages
    - Common atrium with band-like remnant of septum crossing atria in anteroposterior direction
    - Bilateral trilobed lungs
    - Bilateral eparterial bronchi
    - Associated with severe cyanotic CHD (atrioventricular septal defect, common atrioventricular valve, DORV, TGA, pulmonary stenosis/atresia)
    - Abnormalities of pulmonary venous connections
      - TAPVR, > 80%; often obstructed, below diaphragm (type III)
      - Findings of pulmonary venous outflow obstruction may be masked when there is restriction to pulmonary arterial inflow at same time (pulmonary atresia)
  - Polysplenia syndrome = left atrial isomerism or double left-sidedness
    - Multiple spleens, anisosplenia, multilobed spleen (functional asplenia)
    - Abnormalities of systemic venous connections: Interrupted IVC with azygous continuation (> 70%), hepatic veins drain separately into common atrium
    - Bilateral superior vena cavae (~ 41%); 1 or both may connect to coronary sinus
    - Left isomerism of atrial appendages
    - Common atrium or large ostium primum atrial septal defect
    - Bilateral bilobed lungs
    - Bilateral hyparterial bronchi
    - Associated with less severe CHD (common atrium, ventricular septal defect)
    - Malrotation, volvulus, preduodenal portal vein, absent gallbladder, extrahepatic biliary atresia, short pancreas (dorsal head agenesis)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asplenia: Male neonate with severe cyanosis, susceptibility for infections
  - Polysplenia: More variable, often presents later
- Other signs/symptoms

Demographics

- Epidemiology
  - Prevalence 1 per 22,000 to 24,000; 1-3% of CHD
  - Asplenia: M > F; polysplenia: M = F

Natural History & Prognosis

- 1st year mortality: 85% asplenia, 65% polysplenia

Treatment

- Supportive, prostaglandins (if CHD lesion has inadequate pulmonary blood flow or aortic arch interruption/obstruction), antibiotic prophylaxis (functional asplenia)
- Asplenia/polysplenia with pulmonary overcirculation: Pulmonary artery banding
- Asplenia with obstructed pulmonary flow & TAPVR: Delicate balance between pulmonary arterial inflow & venous outflow
  - Placement of palliative systemic to pulmonary artery (Blalock-Taussig or central) shunt increases inflow
  - TAPVR repair needs to be done at same time to reduce outflow obstruction
- Early biventricular repair, if possible
- Univentricular repair, step 1: Bidirectional Glenn (superior cavopulmonary anastomosis) or hemi-Fontan
- Polysplenia: Incorporation of azygous vein to cavopulmonary anastomosis (Kawashima operation) to reduce occurrence of microscopic pulmonary arteriovenous malformation
  - Postoperative: Development of pulmonary to systemic venous collaterals, arteriovenous malformations, pulmonary vein stenosis
- Completion of total cavopulmonary anastomosis (i.e., Fontan procedure), if possible
  - 1 or more hepatic veins may have to be excluded from Fontan shunt → venovenous collaterals
  - CTA or MRA prior to catheterization as road map for coil embolization of collaterals

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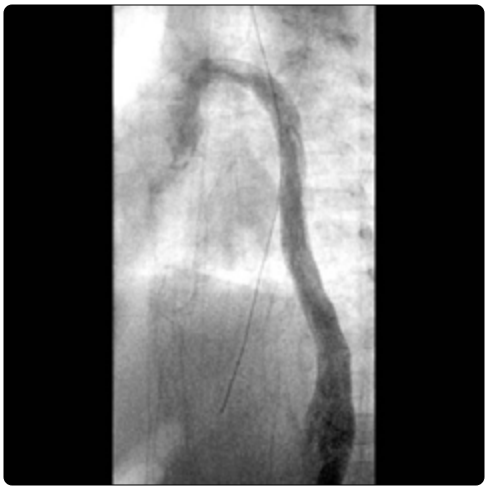
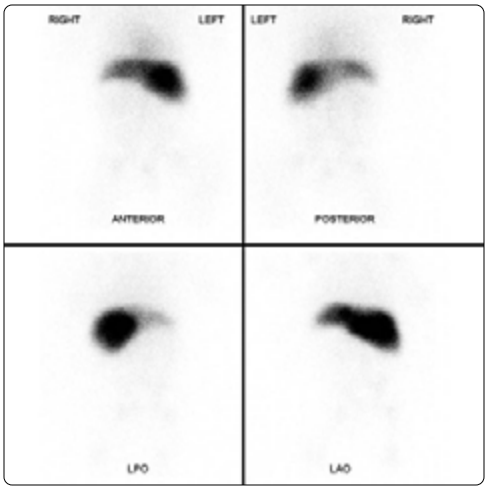
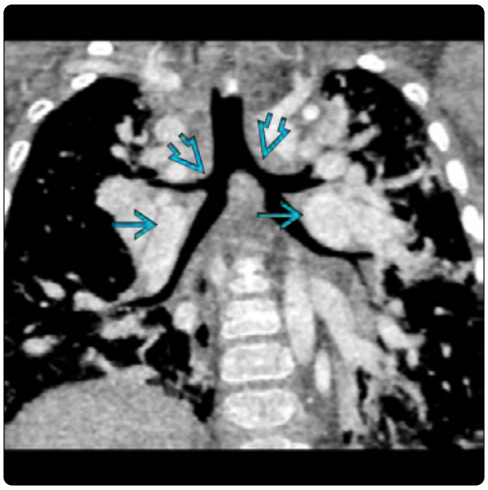
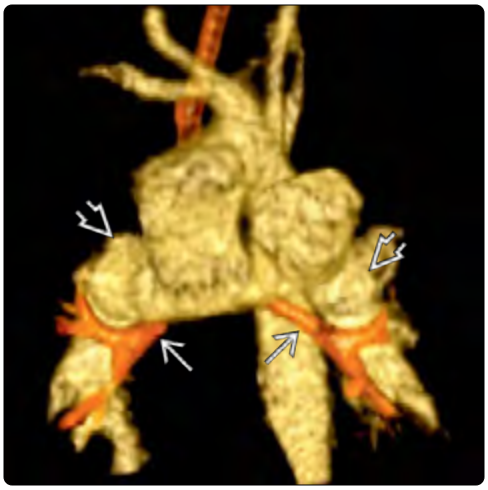


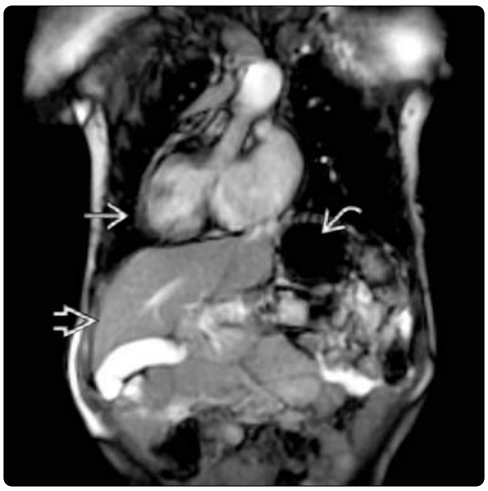


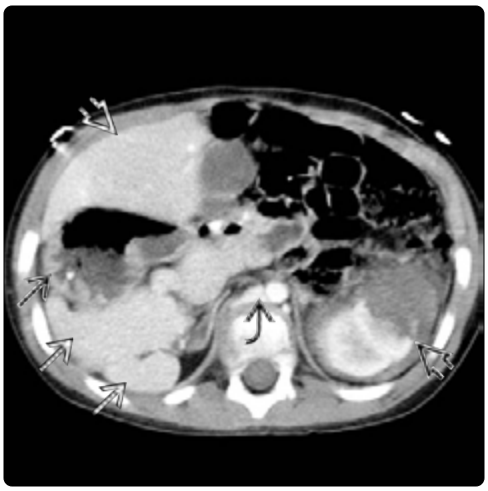
(Left) *Lateral projection catheter angiography demonstrates contrast opacifying a posteriorly located, vertically oriented vessel that drains to the right atrium via a superior approach, consistent with azygous continuation of the IVC in a patient with heterotaxy. Note the lack of any branches, indicating that this is not the aorta.* **(Right)** *Multiple images from a Tc-99m sulfur colloid scan in a patient with heterotaxy demonstrate normal liver uptake but no splenic tissue.*





(Left) *Coronal CTA shows symmetric bilateral eparterial bronchi above the visualized pulmonary arteries, consistent with right isomerism.* **(Right)** *Anterior view of a 3D reformation CTA in a heterotaxy patient illustrates the pulmonary arteries coursing over top of their respective main bronchi, indicating bilateral hyparterial bronchi (i.e., left isomerism).*





(Left) *Coronal T2* GRE MR in this patient with asplenia demonstrates a right-sided cardiac apex, right-sided liver, left-sided stomach, & no splenic tissue.* **(Right)** *Axial CECT in a 2-year-old patient demonstrates a right-sided liver, right-sided stomach, multiple spleens, azygous continuation of the IVC, & a left renal laceration. These findings were all unknown until the patient was involved in a motor vehicle accident, which led to this CT.*

Rhabdomyoma

KEY FACTS

TERMINOLOGY

- Congenital cardiac hamartoma composed of abnormal myocytes

IMAGING

- Initial diagnosis often by fetal &/or postnatal echocardiogram
  - Homogeneous, hyperechoic mass(es) of myocardium
    - Intramyocardial: May appear as wall thickening
    - Intracavitary: Mass attached to myocardium protrudes into lumen
- MR leading diagnostic test to delineate location, extent, & tissue characteristics of cardiac masses in children
  - T1WI: Iso- or mildly hyperintense to myocardium
  - T2WI: Hyperintense to myocardium
  - 1st-pass perfusion: Hypointense to myocardium
  - Late gadolinium enhancement: Isointense to myocardium
  - Homogeneous appearance on all sequences
- Image brain (MR) & kidneys (US) for findings of tuberous sclerosis complex (TSC)
  - ~ 100% of patients with multiple rhabdomyomas & 50% with single rhabdomyoma have TSC

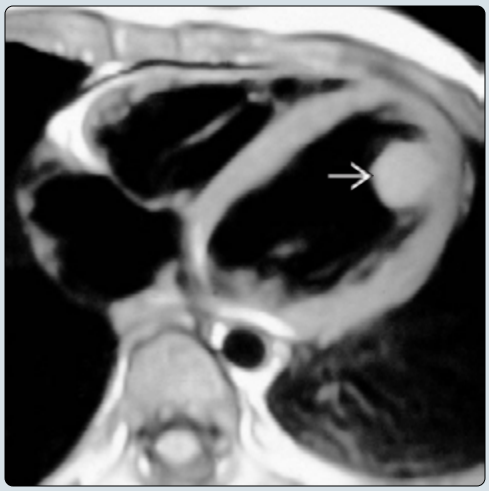
TOP DIFFERENTIAL DIAGNOSES

- Fibroma
- Pericardial teratoma

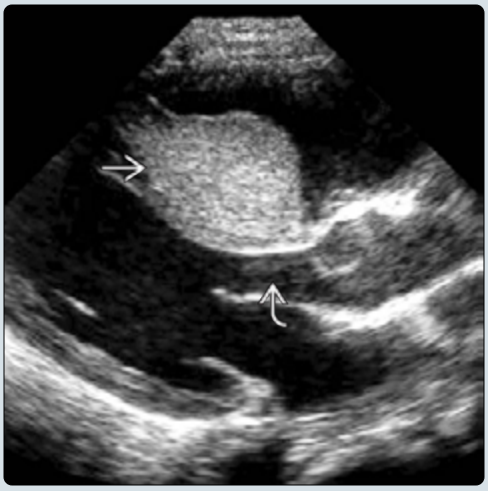
CLINICAL ISSUES

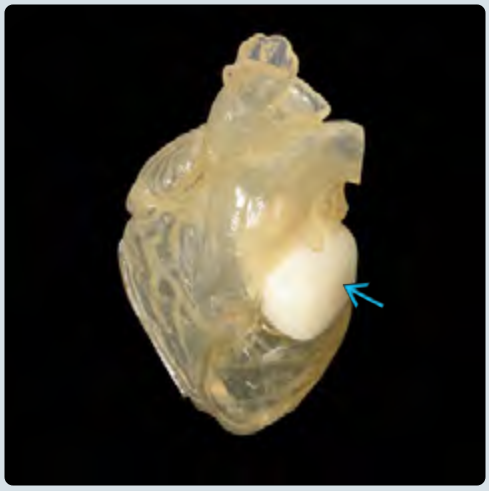
- Cardiac tumors in pediatrics rare
- Rhabdomyoma most common cardiac tumor in children
- 75% diagnosed before 1 year of age
- Natural history: Up to 93% show spontaneous regression; 70% regress by 4 years of age
- Surgical excision for minority of cases with refractory arrhythmias or hemodynamic compromise

(Left) *Graphic shows a partially exophytic rhabdomyoma in the apex of the left ventricle (LV).* **(Right)** *Axial T1 MR was performed in an asymptomatic 8-year-old girl with tuberous sclerosis complex (TSC) after a routine screening echocardiogram showed an intracardiac mass. MR shows a round, well-demarcated intraluminal mass originating from the free wall of the LV. It is slightly hyperintense to myocardium, a characteristic finding of rhabdomyomas.*



(Left) *Postnatal echocardiogram in the parasternal long axis shows a smaller (but still large) mass in the ventricular septum (as compared to fetal imaging, not shown). It is not causing obstruction of the LV outflow tract. Rhabdomyomas may grow during pregnancy but usually spontaneously regress postnatally.* **(Right)** *CT-derived 3D-printed heart model demonstrates a rhabdomyoma in the LV free wall of a teenager being considered for mass resection due to refractory ventricular tachycardia.*





TERMINOLOGY

Definitions

- Congenital cardiac hamartoma composed of abnormal myocytes

IMAGING

General Features

- Best diagnostic clue
  - Cardiac mass within or contiguous with myocardium
- Location
  - Interventricular septum > left or right ventricular free wall > > atrium
  - Multiple in up to 90% of cases
- Size
  - < 1 mm to 10 cm; most 3-4 cm
- Morphology
  - Well-circumscribed, nonencapsulated mass(es)
  - Intramural or exophytic
    - May involve entire wall & appear as wall thickening

Radiographic Findings

- Normal chest radiograph in small masses
- Cardiomegaly & signs of CHF in large masses

Echocardiographic Findings

- Often superior to MR for detection of small masses
- Homogeneous, hyperechoic mass involving myocardium
- No blood flow within mass
- Most often in interventricular septum but can be anywhere
- May appear as simple wall thickening
- Intraluminal portion of mass may move across adjacent valve during cardiac cycle

CT Findings

- NECT
  - Often hypodense compared with myocardium
- CECT
  - Intraluminal component may be assessed with contrast-enhanced studies

MR Findings

- T1WI
  - Iso- or mildly hyperintense to myocardium
- T2WI
  - Hyperintense to myocardium
  - No change with fat saturation (rules out lipoma)
- T1WI C+
  - Minimal initial enhancement (1st-pass perfusion)
  - Isointense to myocardium with late gadolinium enhancement (LGE)
- Cine sequences to differentiate tumor from contractile myocardium, evaluate hemodynamic effect of mass, & look for valvular leak

Imaging Recommendations

- Best imaging tool
  - Dedicated transthoracic echo in all cases
  - MR helpful for diagnostic uncertainty, large masses, & surgical planning
- Protocol advice
  - If cardiac mass identified
    - Look for additional masses
    - Assess location & quality of mass
  - Look for rhythm abnormalities
    - Premature atrial or ventricular contractions common
    - Supraventricular tachycardia
    - Sinus bradycardia
  - Look for signs of obstruction
    - Ventricular inflow or outflow obstruction
    - May manifest as valve regurgitation or stenosis
    - ↑ cardiac work to overcome obstruction → wall hypertrophy
  - Evaluate for other findings of tuberous sclerosis complex (TSC)
  - In fetus, monitor for signs of hydrops (poor function, effusions)

DIFFERENTIAL DIAGNOSIS

Fibroma

- Benign congenital cardiac neoplasm composed of fibroblasts & collagen
- 2nd most common cardiac neoplasm in pediatric population after rhabdomyoma
- Often arises from interventricular septum or left ventricular free wall
- MR: Isointense on T1WI, hypointense on T2WI

Teratoma

- Pericardial (not myocardial) tumor
- Exophytic growth (will not be in cardiac chamber)
- Pericardial effusion often present
- Contains all 3 germ cell layers → may be very heterogeneous on imaging with cystic, fatty, & calcified components

Lipoma

- Most arise from endocardial surface & protrude into chamber lumen
- Fat density/intensity on imaging studies allows for specific diagnosis

Myxoma

- Majority manifest in adulthood (4th-7th decades)
- 90% solitary & atrial in location
  - 75% in left atrium, 10-20% in right atrium
  - Predilection for interatrial septum adjacent to fossa ovalis

Papillary Fibroelastoma

- > 90% involve valves
- Typically small (< 15 mm)

Cardiac Malignancies

- Sarcomas account for most (with angiosarcoma being most common)
- Usually large masses with invasive features
- Pericardial & pleural effusion often present

PATHOLOGY

General Features

- Etiology

- Unknown but data suggests maternal hormones may play role in growth & development of fetal rhabdomyomas
  - Helps explain regression after delivery
- Genetics
  - Nearly 100% of patients with multiple & 50% with single rhabdomyomas have TSC
  - TSC: Autosomal dominant with variable expressivity
    - ~ 30% of cases inherited
    - Other cases due to new mutation
  - Caused by mutations in *TSC1* or *TSC2* genes
    - *TSC1* located on chromosome 9q
      - Encodes for hamartin protein
      - Complexes with tuberin to regulate cell cycle
    - *TSC2* located on chromosome 16p
      - Encodes for tuberin protein
      - Participates in normal brain development & cardiomyocyte differentiation
- Associated abnormalities
  - Other findings of TSC
    - Subependymal nodules & cortical/subcortical tubers
    - Subependymal giant cell astrocytoma
    - Pulmonary lymphangioleiomyomatosis
    - Renal angiomyolipomas & cysts
    - Retinal hamartomas
    - Ungual fibromas
- Pathophysiology
  - Mass may interfere with myocardial contraction
  - Exophytic masses frequently obstruct blood flow or cause valvular insufficiency

Gross Pathologic & Surgical Features

- Well-circumscribed, intramyocardial or exophytic mass(es)

Microscopic Features

- Large vacuolated myocytes
- Glycogen-rich vacuoles stretch perinuclear cytoplasm (spider cells)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Obstruction to blood flow → heart failure
  - Arrhythmias
  - Large intracavitary tumors causing turbulent flow → hemolytic anemia & thrombocytopenia
- May be seen prenatally
  - Generally incidental finding
  - Rarely presents with arrhythmia or hydrops
  - Can detect as early as 22-weeks gestation
    - May discover more masses as pregnancy progresses
    - Tend to ↑ in size prenatally & then regress after birth

Demographics

- Age
  - 75% diagnosed before 1 year of age
- Epidemiology
  - Cardiac tumors rare (1:30,000-1:100,000)
  - Rhabdomyoma most common pediatric cardiac tumor

Natural History & Prognosis

- Generally excellent with spontaneous regression in 70% of children by 4 years of age
- Poor prognosis for untreated large masses interfering with cardiac hemodynamics
  - Most respond well to surgical excision
  - Case reports of response to mTOR (mammalian target of rapamycin) inhibitor sirolimus
    - mTOR: Protein kinase that regulates cellular proliferation; used to treat subependymal giant cell tumors & angiomyolipomas

Treatment

- Surgical excision should be considered only for those with refractory arrhythmias or hemodynamic compromise
  - Partial resection of intraluminal component of large exophytic masses may be necessary
  - 3D printing from CT/MR data can build heart model with tumor location & extent for easy visualization; can assist with procedural planning
  - Attempts at electrophysiology testing & ablation around tumor focus have variable success rates; 3D printed models have been helpful
- Small intramural masses with no hemodynamic effect typically need no treatment or surgical excision

DIAGNOSTIC CHECKLIST

Consider

- Overall prognosis excellent
  - However, despite benign histology, rhabdomyomas may cause significant morbidity from obstruction to inflow or outflow, ventricular dysfunction, or arrhythmias

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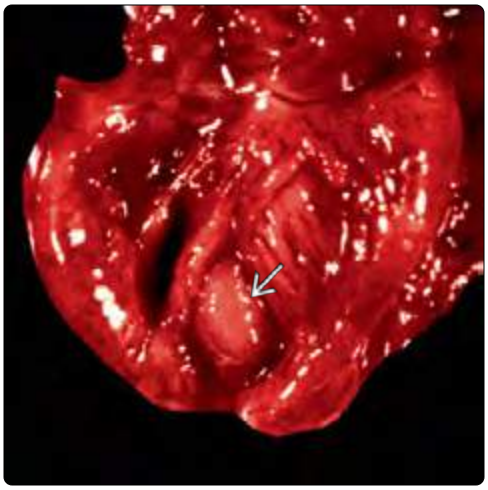
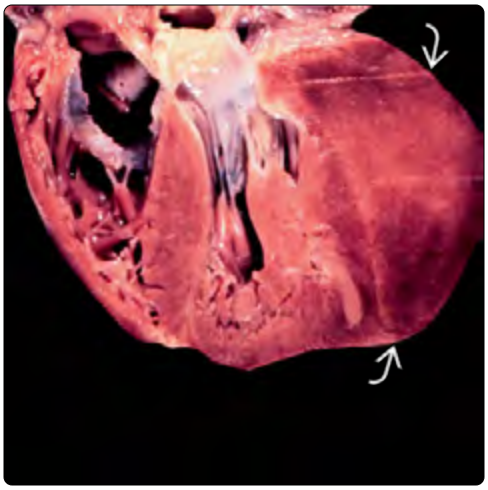
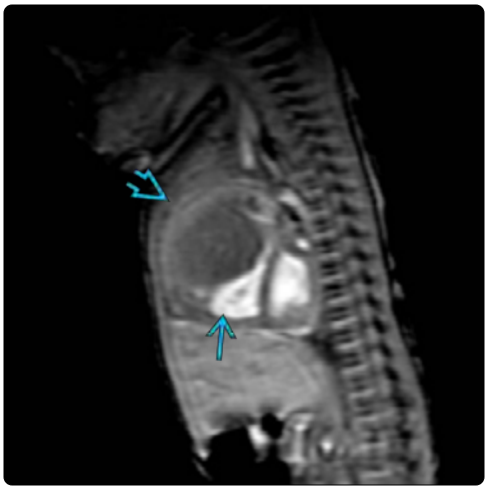
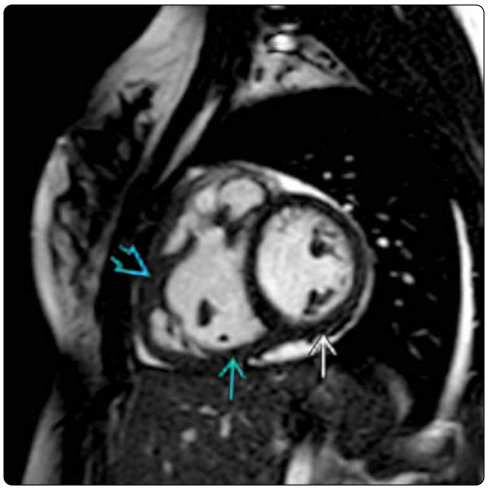


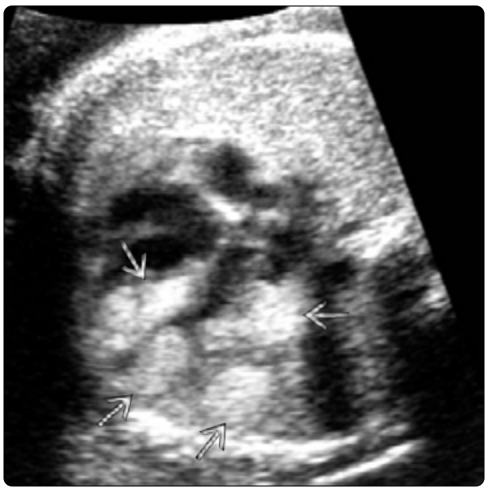


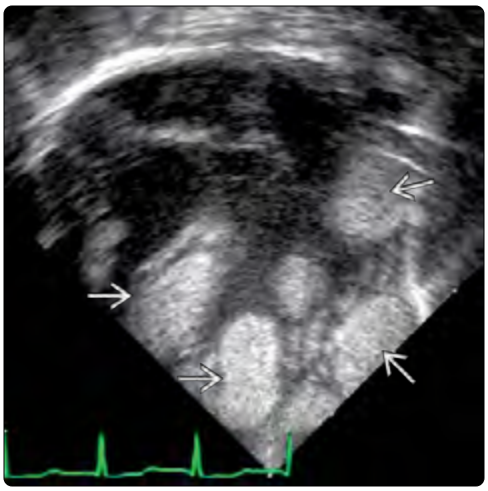
(Left) *Gross pathology of the heart shows a well-defined mass arising from the wall of the lateral ventricle. Histology confirmed a rhabdomyoma.* **(Right)** *Gross pathology shows a rhabdomyoma causing dramatic left ventricular wall thickening. Rhabdomyomas can vary widely in size, number, & morphology.*





(Left) *SSFP bright blood cine short-axis MR in a neonate demonstrates a large hypointense rhabdomyoma within the right ventricular wall.* **(Right)** *SSFP cine short-axis MR in same patient at 13 years of age demonstrates near complete resolution of the rhabdomyoma with minimal residual tumor. The right ventricle appears borderline dilated, & the LV appears normal.*





(Left) *Axial US through the fetal chest shows multiple, echogenic intracardiac masses involving both ventricles & the interventricular septum. Multiple rhabdomyomas are virtually diagnostic of TSC. Fetal MR can be used to evaluate the brain for the presence of subependymal nodules & cortical/subcortical tubers.* **(Right)** *Postnatal echocardiogram in the same patient shows persistence of the multiple rhabdomyomas. TSC was confirmed in this patient.*

Kawasaki Disease

KEY FACTS

TERMINOLOGY

- Inflammatory disease of small & medium-sized blood vessels of unknown etiology, mainly in young children
  - Widespread but characteristic manifestations
  - Coronary artery aneurysms most feared complication

IMAGING

- Chest radiography usually normal
- Echocardiography has sufficient sensitivity & specificity in detecting proximal coronary artery aneurysms
  - Remains 1st-line modality
- CTA can demonstrate aneurysms, stenoses, & Ca^{2+} of coronary or other arteries
- Cardiac MR protocol includes function, coronary artery imaging, 1st-pass perfusion, & late gadolinium enhancement for myocardial viability
- MRA with large field of view can show aneurysms of peripheral arteries

TOP DIFFERENTIAL DIAGNOSES

- Exanthematous infections: Viral or bacterial; allergies or hypersensitivity reactions; vasculitides

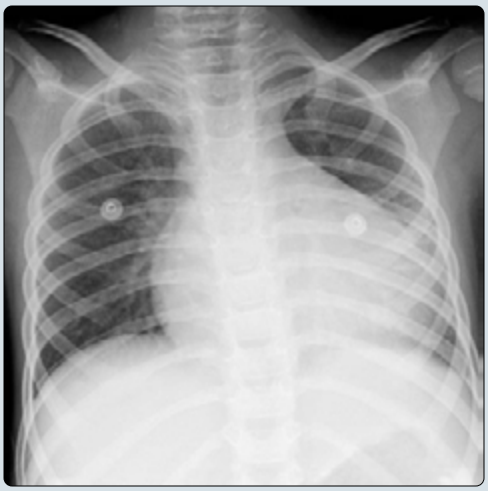
PATHOLOGY

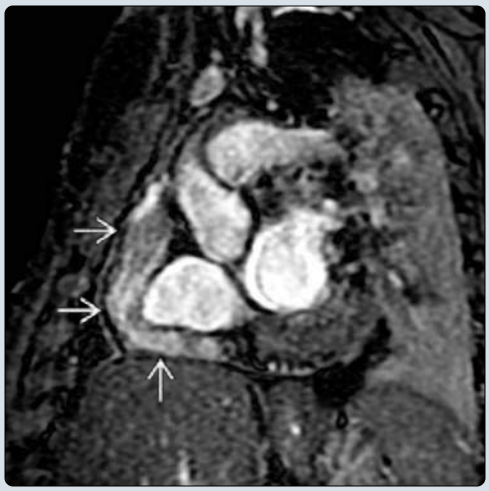
- Etiology unclear, but clinical & epidemiologic features suggest abnormal immune response to toxin or infection

CLINICAL ISSUES

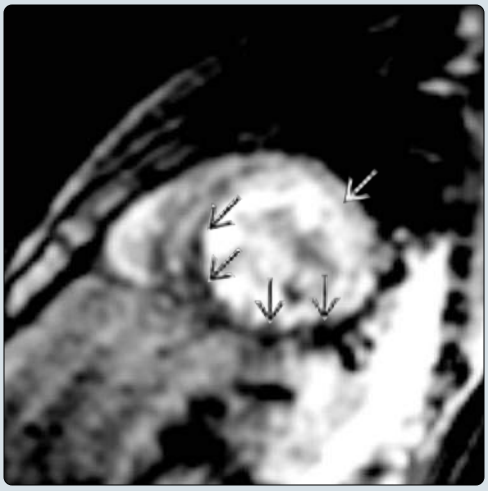
- Acute febrile phase (days 1-11): Fever for 5 days > 104°F, bilateral nonpurulent conjunctivitis & rash, hands & feet develop erythema/edema, tongue & oral mucosa become red & cracked, myocarditis (36%) & pericarditis (16%)
- Subacute phase (days 11-21): Thrombocytosis, desquamation of digits, aneurysms develop; fever resolved
- Convalescent phase (days 21-60): Symptoms resolved
- Chronic phase (> 60 days): Cardiac complications
- Favorable outcome with early recognition & treatment with intravenous gamma globulin, aspirin

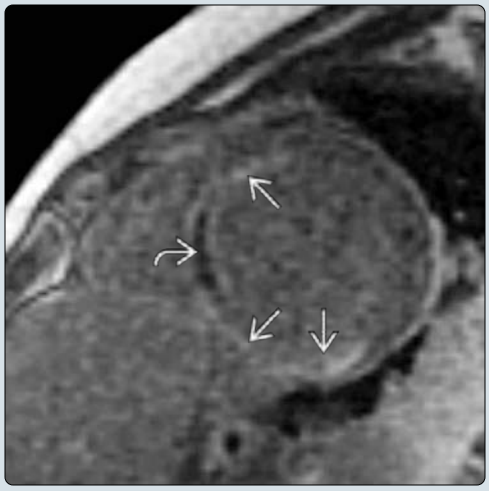
(Left) *AP radiograph of a 5-year-old boy with persistent fever shows an enlarged cardiac silhouette due to either cardiomegaly or a pericardial effusion. A chest radiograph obtained 6 days earlier (not shown) demonstrated a normal cardiac silhouette.* **(Right)** *Sagittal bright blood SSFP MR of the same patient shows a large fusiform aneurysm ➡ of the right coronary artery (RCA). This patient had already developed a myocardial infarction.*





(Left) *Short axis T2* GRE MR 1st pass perfusion image in the same patient shows a large area of nonenhancing myocardium ⇨. Note the normally enhancing myocardium ➡ for comparison.* **(Right)** *Short axis late gadolinium enhanced (LGE) MR shows enhancement ➡ in the same distribution of myocardium that showed no 1st-pass perfusion, confirming the distribution of ischemic myocardium. The septum ➡ also demonstrated a no-reflow phenomenon due to continued microvascular occlusion.*





TERMINOLOGY

Synonyms

- Mucocutaneous lymph node syndrome; acute febrile mucocutaneous syndrome

Definitions

- Inflammatory disease of small & medium-sized blood vessels of unknown etiology, occurring mainly in children
  - Manifests mainly with prolonged fever, nonsuppurative conjunctivitis, inflamed mucosal membranes of mouth & lips, cervical lymphadenopathy, maculoerythematous rash, & desquamation of hands & feet

IMAGING

General Features

- Best diagnostic clue
  - Multiple fusiform & saccular coronary aneurysms in severe cases; coronary artery ectasia in minor cases
- Location
  - Multisystem disease affecting skin, lymph nodes, mucous membranes, conjunctiva, myocardium, pericardium, coronary arteries, joints, bowel, gallbladder, kidney, urethra, & other sites

Radiographic Findings

- Chest radiography usually normal
- Occasionally, may have large cardiac silhouette due to pericardial effusion or cardiomegaly

CT Findings

- CTA
  - Can demonstrate aneurysms, stenoses, & Ca^{2+} of coronary or other arteries

MR Findings

- T2WI
  - Can show edema secondary to myocarditis in acute phase
    - Myocarditis in up to 50%
- T2* GRE
  - MR stress imaging with quantification of regional perfusion
- MRA
  - Accurately images coronary artery aneurysms, occlusions, & stenoses
  - Used to depict & follow other aneurysms of thorax & abdomen or peripheral involvement
- MR cine
  - Steady state free precession cine MR shows regional wall motion abnormalities
  - Can measure cardiac function with end diastolic volumes, systolic ejection, & ejection fraction
- Delayed enhancement
  - Late gadolinium enhancement can show infarcted myocardium

Ultrasonographic Findings

- Grayscale ultrasound
  - Lymphadenopathy usually nonsuppurative, unilateral, & in anterior triangle of neck
  - Gallbladder may be hydropic
  - ± nephromegaly
  - Aneurysms may be identified outside of chest

Echocardiographic Findings

- Sufficient sensitivity & specificity for proximal coronary artery ectasia & aneurysms → 1st-line modality
  - Sensitivity: 80-85%
  - Evaluates ventricular function, valvar function, & pericardial effusion

Angiographic Findings

- Conventional coronary angiography demonstrates aneurysm size & extent of stenosis
  - Aneurysms occur at bifurcating sites
  - Acute thrombotic occlusion of coronary artery may occur; thrombolytic therapy may be indicated

Nuclear Medicine Findings

- Thallium (Tl-201) or technetium (Tc-99m) sestamibi myocardial perfusion imaging (SPECT)
  - Pharmacological stress testing with dipyridamole or adenosine can demonstrate myocardial ischemia

Imaging Recommendations

- Best imaging tool
  - Echocardiography for initial & sequential studies
  - Cardiac MR in older children & adults for assessing function, myocardial ischemia, myocardial viability, & aneurysms
- Protocol advice
  - Comprehensive cardiac MR protocol includes function, coronary artery imaging, 1st-pass perfusion, & late gadolinium enhancement for myocardial viability
  - MRA with large field of view can show aneurysms of peripheral arteries

DIFFERENTIAL DIAGNOSIS

Exanthematous Infections: Viral or Bacterial

- Toxic shock syndrome has high fever, desquamation of hands & feet
- Rheumatic fever has skin rash, fever, arthritis, myocarditis, & pericarditis
- Mononucleosis has fever, lymphadenopathy, splenomegaly, & liver disease

Allergic or Hypersensitivity Reactions

- Drug reactions, Stevens-Johnson syndrome, erythema multiforme have systemic signs, rash, & fever but usually do not have cardiac involvement

Vasculitides

- Systemic lupus erythematosus, polyarteritis nodosa
- Takayasu may have medium to large vessel disease

PATHOLOGY

General Features

- Etiology
  - Remains elusive, but clinical & epidemiologic features suggest abnormal immune response to toxin or infection
- Laboratory evidence
  - C-reactive protein (CRP) elevated
  - White blood cell count elevated

- Thrombocytosis with marked elevation of platelet counts appearing in 2nd & 3rd weeks

Gross Pathologic & Surgical Features

- Generalized systemic vasculitis involving small & medium-sized arteries
- Late phase fibroblastic proliferation & active inflammation replaced by progressive fibrosis & scar

Microscopic Features

- Perivasculitis involving small vessels
- Larger vessels become secondarily inflamed, with aneurysm & thrombus formation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute febrile phase (days 1-11)
    - Temperature elevated > 104°F for 5 days
    - Bilateral nonpurulent conjunctivitis & rash
    - Hands & feet develop erythema & edema
    - Tongue & oral mucosa become red & cracked
    - Cervical lymphadenopathy, usually unilateral
    - Cardiac complications of myocarditis (36%) & pericarditis (16%)
  - Subacute phase (days 11-21): Fever has resolved
    - Persistent irritability, anorexia, conjunctivitis
    - Thrombocytosis develops
    - Desquamation of fingers & toes
    - Aneurysm may develop; greatest risk for sudden death
  - Convalescent phase (days 21-60): Symptoms of illness have disappeared
  - Chronic phase (> 60 days): Involves children with cardiac complications
- Other signs/symptoms
  - Dilation of gallbladder (hydrops) early in disease (15%)
  - Hepatic enlargement & jaundice can occur
  - Gastrointestinal complaints include diarrhea, vomiting, abdominal pain

Demographics

- Age
  - Peak incidence: 6 months to 2 years
  - Another peak occurs after 5 years of age
- Gender
  - M:F = 5:1
- Epidemiology
  - Japan: Incidence 50/100,000 children < 4 years of age (10x incidence in USA)
  - Also prevalent in other countries, affecting mainly Asian populations
  - Minor epidemic outbursts every 3-4 years with seasonal variation

Natural History & Prognosis

- Self-limited disease in majority of cases
- Favorable outcome with early recognition, treatment with intravenous gamma globulin, aspirin
- With development of coronary artery aneurysms
  - Thrombosis, arrhythmia, infarct, or delayed rupture
  - Persistent wall abnormalities after aneurysm regression
  - Chronic coronary insufficiency, premature atherosclerosis in < 4%
  - Death in > 1% due to arrhythmias

Treatment

- High-dose aspirin used as antiinflammatory agent early in disease until fever has decreased
- Low-dose aspirin used in children for 6-8 weeks & for prolonged period in children with confirmed aneurysms
- Intravenous gamma globulin (IVIg) given in acute phase to reduce coronary artery abnormalities
- Transcatheter coronary intervention if thrombosis occurs: Thrombolysis with tissue plasminogen activator (tPA)
- Long-term treatment directed by degree of coronary involvement
  - Coronary bypass surgery or cardiac transplantation may be necessary

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Masquerades as many common diseases in children, but distinct pattern & progression of acute illness striking

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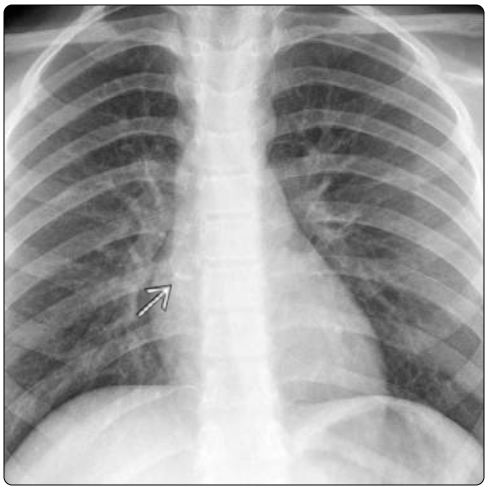
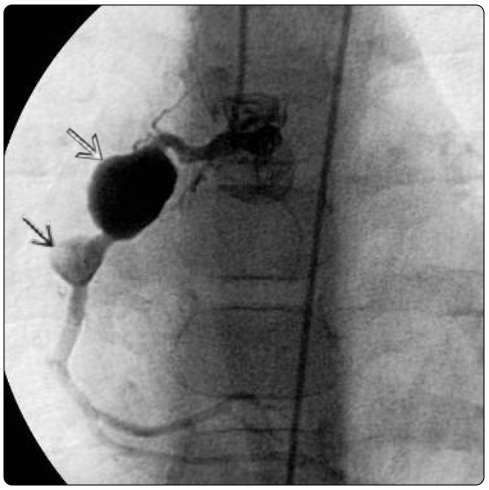
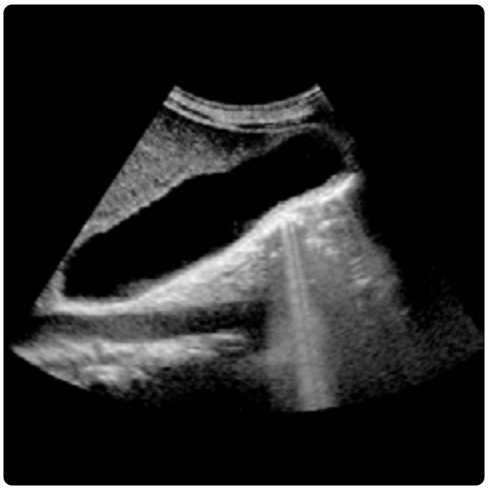
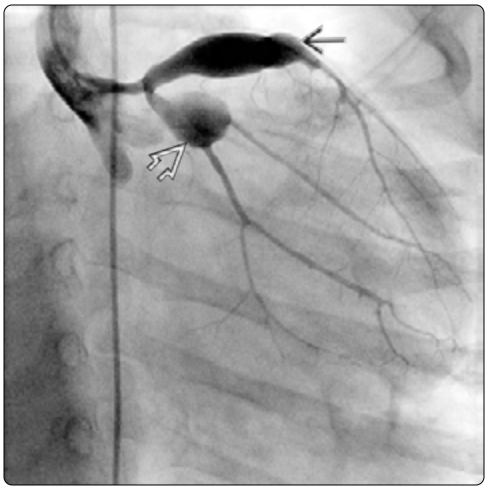


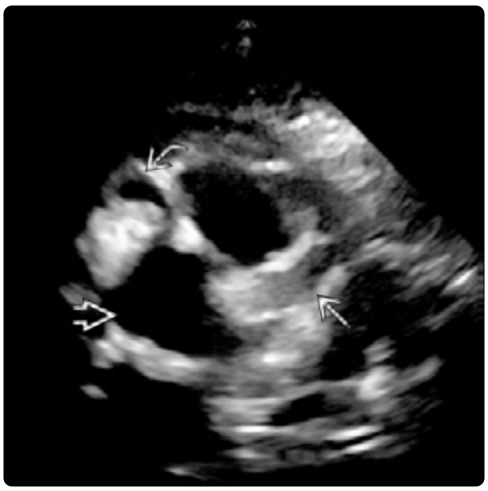


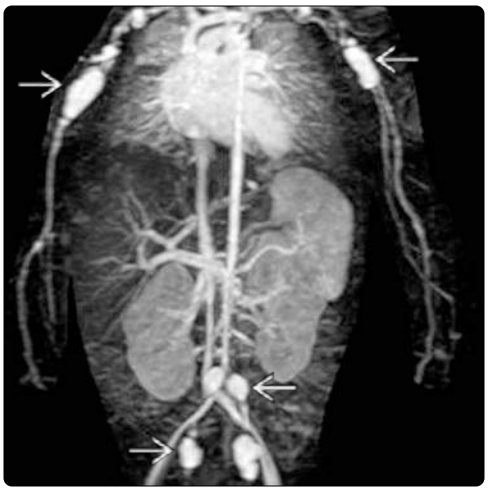
(Left) *PA radiograph shows a curvilinear Ca^{2+} ➡ projecting inferior to the right hilum. The patient's history revealed that the child had been diagnosed with Kawasaki disease at age 4 years, confirming that this abnormality is due to a calcified aneurysm.* **(Right)** *Oblique angiogram of a right coronary artery injection shows a large proximal aneurysm ➡ & a smaller more distal aneurysm, which may contain some thrombus ⇨. This patient eventually developed stenosis requiring stenting.*





(Left) *Oblique ultrasound of the right upper quadrant in a 5 year old with fever & pain shows an enlarged, hydropic gallbladder. This finding ultimately led to the diagnosis of Kawasaki disease.* **(Right)** *Oblique angiography of the left main coronary artery in a patient with Kawasaki disease shows fusiform aneurysmal dilation of the proximal left anterior descending coronary artery ⇨ & a bulbous aneurysmal dilation of the left circumflex coronary artery ➡ with 2 branches arising from the aneurysm.*





(Left) *Axial echocardiogram shows the aorta ➡ & origin of the RCA with an aneurysm ➡. A fusiform aneurysm ➡ of the left anterior descending coronary artery is also visualized. Echocardiography identifies ~ 85% of coronary artery aneurysms. (Courtesy P. Madueme, MD.)* **(Right)** *Coronal MRA in a child with Kawasaki disease shows multiple aneurysms ➡ of the peripheral arteries. This same MRA was also able to depict aneurysms of the coronary arteries (not shown). (Courtesy S. Yoo, MD.)*

KEY FACTS

TERMINOLOGY

- Acute rheumatic fever: Multisystem disease affecting heart, joints, skin, & brain 1-5 weeks following infection with group A β-hemolytic *Streptococcus*
  - Due to autoimmune response
  - 40% of acute rheumatic fever patients develop cardiac involvement (RHD)

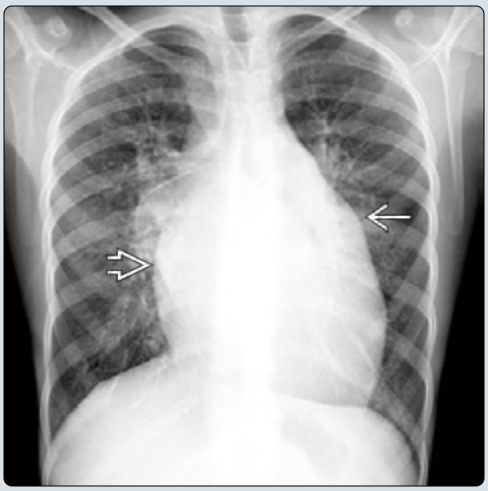
IMAGING

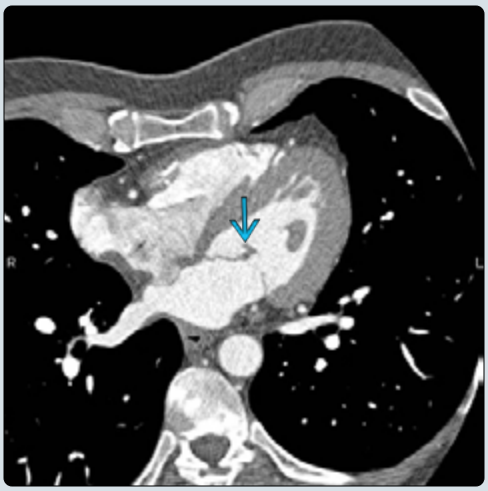
- Acute rheumatic fever: Large heart, left atrial enlargement, pulmonary edema (due to left ventricular dysfunction with mitral insufficiency)
  - "Rheumatic pneumonia" rarely seen
  - Pericardial effusion
- Chronic RHD: Ca^{2+} of valves, especially mitral or aortic
- Echocardiogram during acute disease quantitates degree of mitral insufficiency & left ventricular function
- Echocardiogram in chronic disease shows progression of valve stenosis with thickened leaflets, which calcify

CLINICAL ISSUES

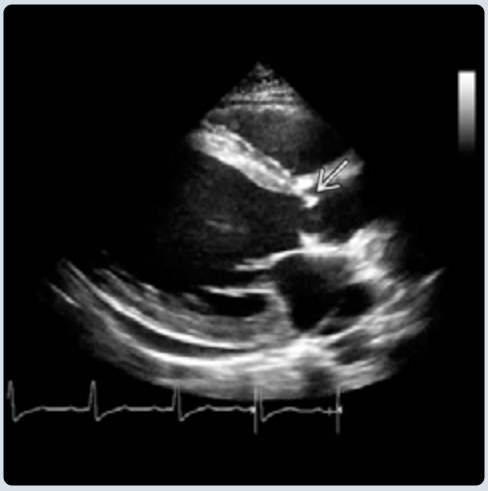
- Acute disease occurs in young children 3-15 years who present with streptococcal sore throat
  - RHD develops after 0.3% of such infections in USA
- Initial treatment: Therapy aimed at preventing acute rheumatic fever by treating group A streptococcal pharyngitis & eradicating reservoir for transmission
  - Disease has dramatically ↓ in USA where sore throat from *Streptococcus pyogenes* is treated
  - Disease now most common in overcrowded areas of world where infection can spread in dry, hot climate
    - High mortality rate in malnourished populations
- Jones criteria for rheumatic fever
  - Major: Carditis, polyarthritis, chorea, subcutaneous nodules, & erythema marginatum
  - Minor: Fever, arthralgia, elevated acute phase reactants, ↑ sedimentation rate

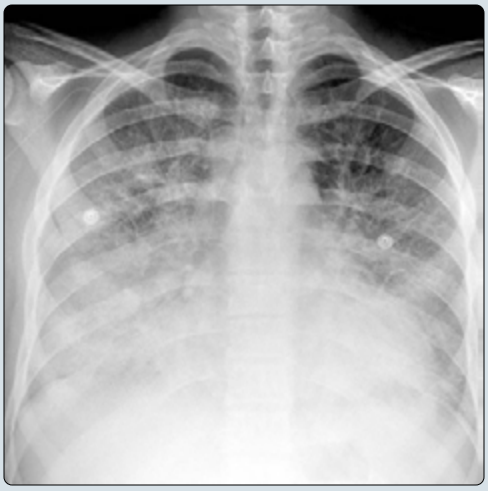
(Left) *PA radiograph in an adult with rheumatic heart disease (RHD) shows cardiomegaly with dilation of the left atrial appendage, creating a broad bump on the lateral margin of the heart below the level of the pulmonary artery. The left atrium splays the carina & creates a double shadow over the right heart border.* **(Right)** *Axial CECT in a young adult shows irregular thickening of the mitral valve due to RHD.*





(Left) *Left ventricular outflow tract (3 chamber) echocardiogram of a 17 year old with prior acute rheumatic fever shows the aortic valve to be thickened & destroyed with moderate to severe regurgitation (on Doppler, not shown). Vegetations were visualized, confirming endocarditis complicating RHD.* **(Right)** *AP radiograph in the same patient shows pulmonary edema due to progressive aortic insufficiency from acute bacterial endocarditis complicating rheumatic aortic valve disease.*





TERMINOLOGY

Abbreviations

- Rheumatic heart disease (RHD)

Definitions

- Acquired heart disease due to autoimmune response to prior group A β-hemolytic *Streptococcus* infection
- Part of multisystem disease affecting heart, joints, skin, & brain; occurs 1-5 weeks following infection

IMAGING

General Features

- Best diagnostic clue
  - Large heart, left atrial enlargement (LAE), pulmonary edema
    - Due to left ventricular dysfunction with mitral insufficiency
  - Pericardial inflammation & effusion
  - Multisystem involvement

Radiographic Findings

- Acute disease
  - Cardiomegaly & pulmonary congestion, pericardial effusion
  - LAE when mitral insufficiency or stenosis present
  - "Rheumatic pneumonia" rarely seen
- Chronic RHD
  - Ca^{2+} of valves, especially mitral or aortic
  - LAE with mitral regurgitation or stenosis

Echocardiographic Findings

- Acute disease
  - Quantitates degree of mitral insufficiency & left ventricular function
- Chronic
  - Progression of valve stenosis with thickened leaflets, which calcify
  - Fusion of commissures & chordae

MR Findings

- MR to assess cardiac function
  - Quantitates degree of valve insufficiency/stenosis

PATHOLOGY

General Features

- Etiology
  - Infection with group A *Streptococcus* is precipitating event
    - RHD develops after 0.3% of such infections in USA
  - May lead to acute rheumatic fever with inflammation of joints, heart, brain, connective tissue
    - Genetic susceptibility to rheumatic fever related to human leukocyte antigens
    - Immune reactions with cross-reactive antibody or cell-mediated immunity

Gross Pathologic & Surgical Features

- Inflammatory reaction involves connective or collagen tissue
- Acute carditis affects endocardium & myocardium
- Pericardial serositis may later calcify
  - Pericardium thickened & irregular
- Mitral valve involvement in 75% of cases
  - Verrucous lesions
- Arthritis does not affect cartilage, but synovial lining shows fibrinoid degeneration

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Cardiac symptoms
    - 39% of patients with rheumatic fever develop pancarditis with valve insufficiency
    - Pericardial friction rub: Pericardial involvement
  - Prominent noncardiac manifestations
    - Jones major criteria: Carditis, polyarthritis, chorea, subcutaneous nodules, & erythema marginatum
    - Jones minor criteria: Fever, arthralgia, elevated acute phase reactants, ↑ sedimentation rate
- Significant change in presentation over past 25 years
  - Disease has dramatically ↓ in USA where sore throat from *Streptococcus* is treated
  - Disease now most common in overcrowded, poor areas of world where *S. pyogenes* can spread in dry, hot climate

Demographics

- Age
  - Acute disease occurs in young children 3-15 years of age who present with streptococcal sore throat
  - Chronic disease can have multiple recurrences & progression
- Gender
  - Equal in numbers but prognosis worse for females

Natural History & Prognosis

- Group A *Streptococcus* is gram-positive coccus that colonizes skin & oral pharynx
  - Acute infection may precipitate acute rheumatic fever
    - Occurs 1-5 weeks later in 0.3%
  - Chronic sequelae of acute rheumatic fever
    - 40% of patients develop cardiac involvement
    - Mitral valve most severely affected in 65-75%, aortic valve in 25%
    - Varying degrees of arrhythmia, valve insufficiency, or ventricular dysfunction
- High mortality rate in malnourished populations
  - Mortality rate is ~ 0 in USA
  - 90,000 deaths worldwide each year

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Marfan Syndrome

KEY FACTS

TERMINOLOGY

- Inherited autosomal dominant connective tissue disorder due to *FBN1* mutation → abnormal fibrillin-1

IMAGING

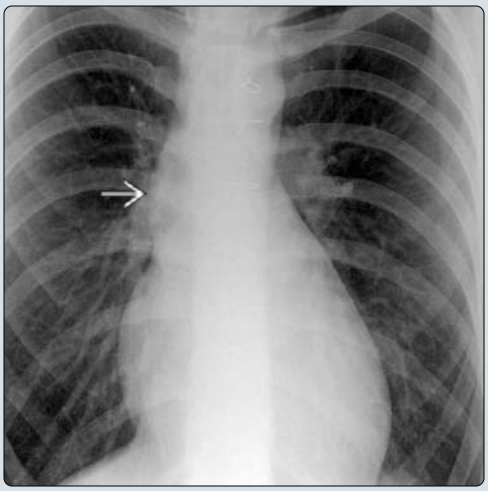
- Cardiovascular: Aortic root dilation at sinuses of Valsalva (75%), ascending aortic dissection, mitral valve prolapse [(MVP), 50-70%], main pulmonary artery dilation, mitral annulus Ca^{2+}, dilation of abdominal aorta
- Skeletal: Pectus excavatum, pectus carinatum, long arms & legs, arachnodactyly, joint hypermobility, scoliosis, thoracic lordosis, pes planus, protrusio acetabuli
- Pulmonary: Spontaneous pneumothorax, apical blebs
- Dural: Ectasia (enlarged nerve sleeves & posterior vertebral body scalloping)
- Postoperative complications: Pseudoaneurysm, aortic dissection, rupture
- Best imaging tools
  - CTA in acute setting to exclude dissection &/or rupture
  - Echocardiography or cardiac MR for routine follow-up of aortic root dilation & valvular disease

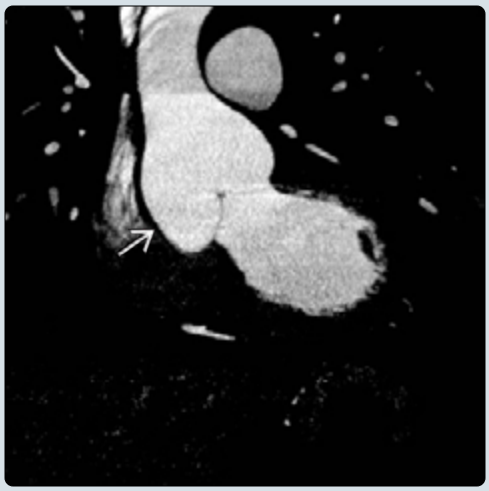
PATHOLOGY

- 2010 revised Ghent nosology for diagnosis
  - Based on family history, aortic root size/dissection, ectopia lentis, *FBN1* mutation, systemic score

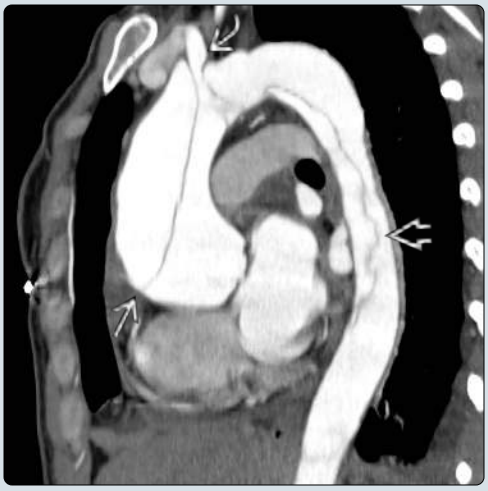
CLINICAL ISSUES

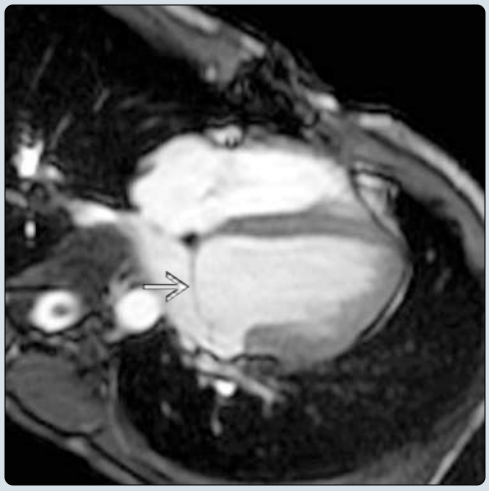
- 90% of deaths from cardiovascular complications (aortic dissection, congestive heart failure, & cardiac valve disease)
  - Progressive aortic dilation → highest risk of dissection
    - Chest pain radiating down back in Marfan syndrome patient → suspect aortic dissection
- Treatment
  - Medical: β-blocker or angiotensin II receptor blocker
  - Surgery: Prophylactic aortic root surgery when diameter at sinuses of Valsalva exceeds 5 cm





(Left) *Single frontal view of the chest in a patient with Marfan syndrome shows a dilated ascending aorta ➡ along the right side of the mediastinum.* **(Right)** *Coronal MR angiogram of the chest in a patient with Marfan syndrome shows aortic dilation at the sinuses of Valsalva ➡. This is commonly seen in patients with Marfan syndrome & causes soft tissue prominence along the right side of the mediastinum on the chest radiograph.*





(Left) *Sagittal chest CTA in a patient with Marfan syndrome demonstrates an extensive dissection of the aorta, which extends from the ascending aorta ➡ to the descending aorta ➡. Note the involvement of the brachiocephalic artery ➡.* **(Right)** *Four-chamber view from a cine "bright blood" SSFP cardiac MR demonstrates mitral valve prolapse with ballooning of the septal leaflet of the mitral valve ➡ into the left atrium.*

TERMINOLOGY

Definitions

- Inherited autosomal dominant connective tissue disorder due to *FBN1* mutation → abnormal fibrillin-1

IMAGING

General Features

- Best diagnostic clue
  - Aortic dissection in tall thin patient with pectus deformity, scoliosis, & long fingers
- Location
  - Cardiovascular: Aortic root dilation at level of sinuses of Valsalva (75%), ascending aortic dissection, mitral valve prolapse [(MVP) 50-70%], main pulmonary artery dilation, mitral annulus Ca^{2+}, dilation of abdominal aorta
  - Skeletal: Pectus excavatum, pectus carinatum, long arms & legs, arachnodactyly, joint hypermobility, scoliosis, thoracic lordosis, pes planus, protrusio acetabuli
  - Ocular: Ectopia lentis (50%), flat cornea, cataract, hypoplastic iris, nearsightedness, glaucoma (< 50 years of age), retinal detachment
  - Pulmonary: Spontaneous pneumothorax (5-10%), apical blebs
  - Skin: Stretch marks in absence of weight changes or pregnancy, recurrent hernia
  - Dural: Ectasia (widening & dilation of dural sac with enlarged nerve sleeves & posterior vertebral body scalloping)

Radiographic Findings

- Enlargement of ascending aorta creates right-sided border forming mediastinal prominence
- Enlarged main pulmonary artery
- Pectus excavatum or carinatum
- Apical bleb, spontaneous pneumothorax

CT Findings

- CTA
  - Dilation of ascending aorta (starts at sinuses of Valsalva)
  - Aortic dissection, dilated pulmonary artery, mass effect on heart or IVC from pectus excavatum
    - CTA fast & effective way to exclude aortic dissection in acute setting
    - Coronary CTA helpful in patients with dissection to evaluate coronary artery involvement
  - Apical blebs, pneumothorax
  - Posterior scalloping of vertebrae from dural ectasia

MR Findings

- Anatomic imaging: Ascending aortic aneurysm, dissection
- Functional cine "bright blood" imaging
  - Dephasing artifact from aortic & mitral regurgitation
  - MVP
  - Ventricular function best on short axis imaging
- Phase contrast imaging
  - Quantify aortic & mitral regurgitant fraction
  - Pulse wave velocity measurements to evaluate aortic stiffness

Imaging Recommendations

- Best imaging tool
  - CTA in acute setting to exclude dissection &/or rupture
  - Echocardiography or cardiac MR for routine follow-up of aortic root dilation & valvular disease

PATHOLOGY

General Features

- Genetics
  - Autosomal dominant
  - Mutation in *FBN1* gene located on chromosome 15q21.1
    - Mutation causes abnormal fibrillin (major substrate for microfibrils)

Staging, Grading, & Classification

- 2010 revised Ghent nosology for diagnosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Chest pain radiating down back from aortic dissection
  - Heart murmurs from aortic & mitral regurgitation
  - Unilateral chest pain from spontaneous pneumothorax
- Other signs/symptoms
  - Visual disturbances

Demographics

- Frequency: 2-3:10,000
- Diagnosis may be made in infancy or well into adulthood

Natural History & Prognosis

- 90% of deaths from cardiovascular complications (aortic dissection, congestive heart failure, & cardiac valve disease)
  - Progressive aortic dilation carries highest risk of aortic dissection
- Improved detection & treatment has helped prolong survival to nearly normal levels

Treatment

- Moderate restriction of physical activity
- β-blocker or angiotensin II receptor blocker
  - For prophylaxis of progressive aortic root dilation
  - Initiated at disease diagnosis
- Prophylactic aortic root surgery when diameter at sinuses of Valsalva exceeds 5 cm

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Loeys-Dietz Syndrome

KEY FACTS

TERMINOLOGY

- Autosomal dominant connective tissue disorder due to 1 of 5 genetic mutations affecting transforming growth factor β

IMAGING

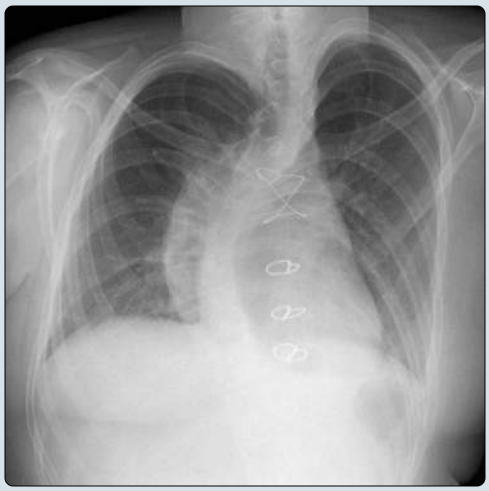
- 98% have aortic root aneurysms
- 20% have aneurysms in head & neck region
- Aneurysms also occur in coronary arteries, pulmonary arteries, & ductus arteriosus
- Recommendations
  - Baseline imaging
    - Echocardiography every 6-12 months
    - Screening CTA or MRA of head through pelvis every 2 years
  - For acute chest pain: Chest CTA for aortic dissection & aneurysms
  - For acute headache: Noncontrast head CT for intracranial hemorrhage; if positive, follow with head CTA

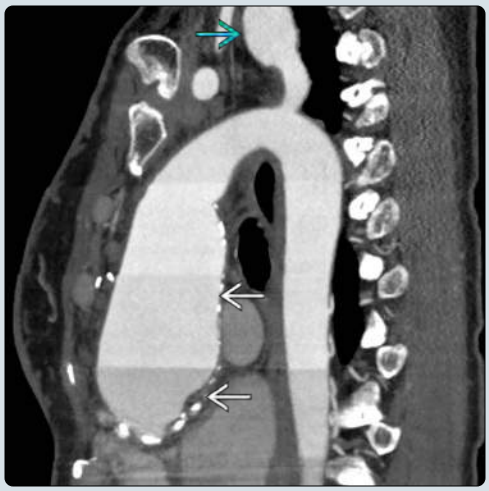
TOP DIFFERENTIAL DIAGNOSES

- Marfan syndrome
- Vascular Ehlers-Danlos syndrome

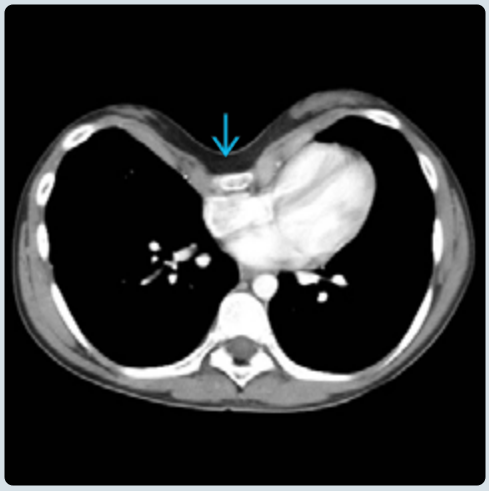
CLINICAL ISSUES

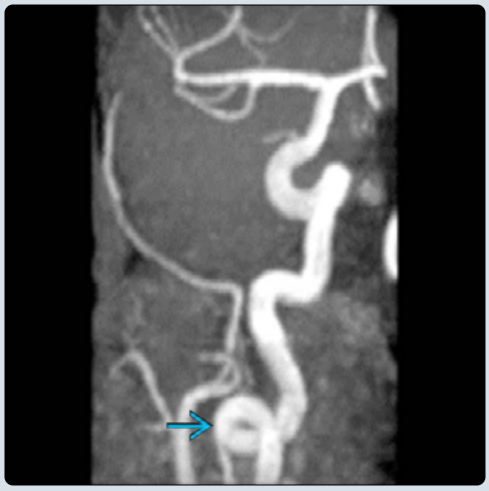
- Systemic involvement with craniofacial, cardiovascular, skeletal, skin, & nervous system abnormalities
- Characteristic clinical triad of hypertelorism, bifid uvula or cleft palate, & aortic aneurysms ± dissection & tortuous arteries
- Leading cause of death: Aortic dissection (67%)
  - ↑ risk for dissection or rupture at > 3.9 cm diameter
- Abdominal aortic dissection & cerebral hemorrhage from ruptured aneurysm account for 22% of deaths
- Preventative treatment with hypertension/heart rate control: β-blocker, angiotensin II receptor blocker





(Left) *Frontal view of the chest in a 15-year-old patient with Loeys-Dietz syndrome shows severe dextroscoliosis of the thoracic spine. Note the postoperative changes of a prior sternotomy for repair of the aortic root.* **(Right)** *Oblique sagittal CTA of a 15-year-old patient with Loeys-Dietz syndrome shows an aneurysm of the left subclavian artery ⇨. Note the ascending aortic aneurysm ➡ & postoperative changes.*





(Left) *Axial CECT of a 15-year-old patient with Loeys-Dietz syndrome shows a pectus excavatum deformity ⇨, which can also be seen in patients with Marfan syndrome.* **(Right)** *Anterior projection of a 3D TOF MRA in a 15-year-old patient with Loeys-Dietz syndrome shows tortuosity of the proximal right internal carotid artery ⇨, which is frequently seen in this syndrome.*

TERMINOLOGY

Definitions

- Autosomal dominant connective tissue disorder with aortic/arterial abnormalities

IMAGING

General Features

- Best diagnostic clue
  - Arterial tortuosity with aortic aneurysm & dissection
  - Hypertelorism
  - Bifid uvula or cleft palate
  - Instability or malformation of spine/neck
  - Club foot
  - White sclera of eye looks blue or gray
- Location
  - 98% have aortic root aneurysms
  - 20% have aneurysms in head & neck region
  - Aneurysms also occur in coronary arteries, pulmonary arteries, & ductus arteriosus

CT Findings

- CTA
  - Evaluation of aortic aneurysm & dissection; associated coronary artery, pulmonary artery, & ductus arteriosus aneurysms

MR Findings

- MRA
  - Evaluation of aortic aneurysm & dissection
    - Phase contrast to evaluate aortic valve regurgitation, which often accompanies aortic root aneurysm
  - Tortuosity of intracranial vessels

Echocardiographic Findings

- 1st-line evaluation of aortic root, myocardial function, valve morphology (including bicuspid aortic valve)

Imaging Recommendations

- Best imaging tool
  - For acute chest pain: CTA of chest for aortic dissection & aneurysms
  - For acute headache: Noncontrast head CT for acute intracranial hemorrhage; if positive, follow with head CTA
- Baseline imaging
  - Echocardiography + screening CTA or MRA of head, neck, chest, abdomen, & pelvis
- Follow-up imaging
  - Echocardiography every 6-12 months
  - CTA or MRA of head to pelvis at least every 2 years
    - Blood pool MR contrast agent helpful in screening large territories

DIFFERENTIAL DIAGNOSIS

Marfan Syndrome

- Dissection & aortic rupture tend to occur at larger diameter & later in life than patients with Loeys-Dietz syndrome
- Lacks tortuosity, aneurysms, & dissections beyond aorta
- Lens dislocation typical

Vascular Ehlers-Danlos Syndrome

- Aortic dissection & rupture without preceding dilation
  - Usually affect descending rather than proximal aorta
- Spontaneous carotid-cavernous fistula, bowel perforations

PATHOLOGY

General Features

- Connective tissue disorder from mutations in 1 of 5 genes

Staging, Grading, & Classification

- *TGFBR1* mutation causes Loeys-Dietz type 1
- *TGFBR2* mutation causes Loeys-Dietz type 2
- *SMAD3* mutation causes Loeys-Dietz type 3
- *TGFB2* mutation causes Loeys-Dietz type 4
- *TGFB3* mutation causes Loeys-Dietz type 5

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Characteristic clinical triad of hypertelorism, bifid uvula or cleft palate, & aortic aneurysms ± dissection & tortuous arteries
- Other signs/symptoms
  - Systemic involvement with craniofacial, cardiovascular, skeletal, skin, & nervous system abnormalities
    - Congenital heart problems such as patent ductus arteriosus, bicuspid aortic valve, or atrial septal defect
    - Scoliosis or kyphosis, pectus excavatum or carinatum deformity, camptodactyly, arachnodactyly, club foot
    - Translucent skin, joint hypermobility
    - Craniosynostosis, dural ectasia
    - Strabismus from weakened eye muscles

Natural History & Prognosis

- Leading cause of death: Aortic dissection (67%)
  - ↑ risk for dissection/rupture at aortic diameter > 3.9 cm
- Abdominal aortic dissection & cerebral hemorrhage from ruptured aneurysm account for 22% of deaths

Treatment

- Preventative treatment: Hypertension/heart rate control with β-blocker, angiotensin II receptor blocker

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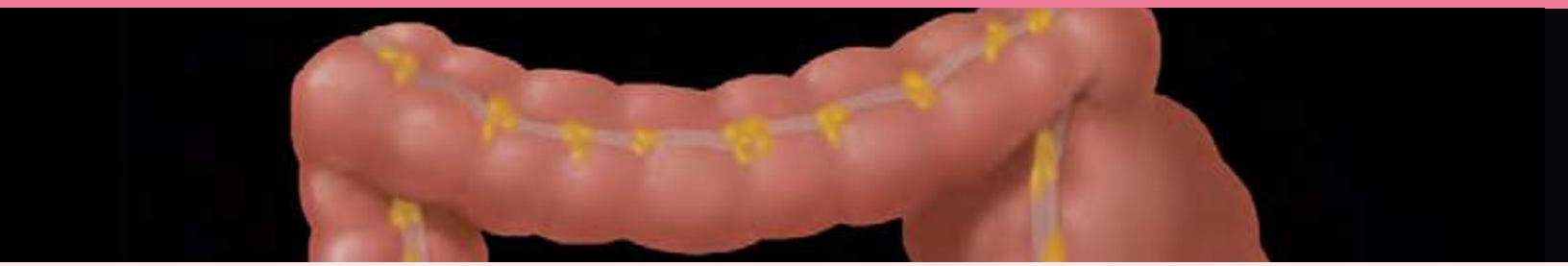
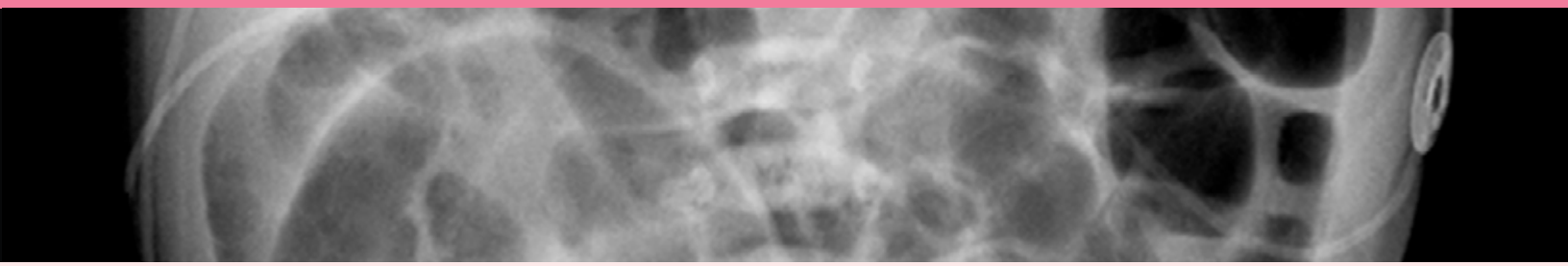
SECTION 4

Gastrointestinal





Imaging Techniques

Pediatric radiologists must have an understanding not only of the unique disorders that affect children, but also the expected range of normal appearances that occur from infancy to early adulthood. The radiologist must also be able to tailor the imaging study to meet a child's needs, which includes explaining the procedure in an age-appropriate manner, distracting the child so that imaging can be performed, &, when needed, immobilizing the child safely during the procedure. In addition, radiologists must remember that children are more sensitive to the effects of ionizing radiation than adults. This should be considered when deciding on the most appropriate imaging study to diagnose a suspected condition. If the selected test requires ionizing radiation, pediatric-specific protocols should be employed to keep the dose as low as reasonably achievable (the ALARA principle).

Many imaging modalities are available for diagnosing abdominal pathology in children. Depending on the patient age, presentation, & specific clinical question, each modality has unique advantages & disadvantages.

Radiography

Constipation, pain, vomiting/bowel obstruction, & necrotizing enterocolitis are common indications for obtaining pediatric abdominal radiographs. Modifications for children include a single view for suspected constipation & the decubitus view or cross-table lateral view to look for free air in infants.

Fluoroscopy

Methods to reduce radiation exposure to the patient include minimizing fluoroscopy time, pulsed fluoroscopy, fluoroscopic image hold or clips rather than digital spot exposures, small image intensifier-to-patient distance, appropriate collimation, & low-dose fluoroscopy settings for pediatric patients based on size.

Upper GI

Common pediatric indications for the upper GI series include bilious emesis, intractable nonbilious emesis, & difficulty swallowing. A single-contrast upper GI is typically employed for these investigations, though a double-contrast study is sometimes performed in teenagers when looking for esophagitis or gastric ulcers (i.e., studies requiring fine mucosal detail). Note that evaluating the position of the duodenojejunal junction (DJJ) is crucial, especially in young vomiting infants.

The typical single-contrast upper GI technique should include the following images: A left-side-down lateral view of the esophagus while swallowing, a supine frontal view of the esophagus (which can include a fluoroscopic clip of peristalsis), a right-side-down lateral view of the 1st pass of contrast into the duodenum as it courses posteriorly in the retroperitoneum & ascends toward the DJJ, & a straight supine frontal view of the duodenum during the 1st pass of contrast (documenting the normal DJJ to the left of the spine at or nearly at the same level as the duodenal bulb). A left posterior oblique (LPO) view is optional, which will show an overlapping bulb & DJJ if they are not seen on the right lateral view.

Contrast Enema

Common indications for a contrast enema include the failure of a newborn to pass meconium, newborn bilious emesis (if the radiograph suggests a distal obstruction), & chronic constipation.

Enema modifications for children include selecting a rectal catheter that is appropriate for size & the use of a water-soluble contrast instead of barium (with double contrast studies being rarely performed in children).

The typical single contrast enema technique includes the following images: An early left-side-down lateral view of the rectum through the splenic flexure (especially to evaluate the rectosigmoid ratio) while contrast is flowing, a frontal view of the rectum through the splenic flexure during active filling, an overhead frontal view of the abdomen when filled just barely to the cecum, & an overhead frontal view of the abdomen after spontaneous contrast evacuation. In newborns with a suspected distal bowel obstruction, reflux of contrast into the terminal ileum is helpful. If looking for a stricture after necrotizing enterocolitis, careful attention to subtle caliber change is required in real time during contrast flow, particularly if the colon overlaps at any point.

Enema Reduction of Intussusception

Various techniques can be used to reduce an ileocolic intussusception; however, air reduction is used in many hospitals. With this method, air is insufflated into the colon under fluoroscopic monitoring until gas refluxes into the terminal ileum & there is disappearance of the soft tissue mass of the intussusceptum. Important points for performing this technique include: An adequate seal must be created by the rectal tube at the anus to build effective intraluminal pressure, the sustained colonic pressure must be kept below 120 mmHg (with the pressure typically monitored by a Shiels device), & an 18-gauge IV-type cannula should be kept at the bedside to reduce a tension pneumoperitoneum if perforation occurs. Note that an enema reduction is contraindicated with peritoneal signs or pneumoperitoneum.

Abdominal US

US is a simple, cost-effective, noninvasive, & ionizing radiation-free method of imaging the pediatric abdomen.

Common indications for pediatric abdominal US include suspected hypertrophic pyloric stenosis (HPS), intussusception, appendicitis, a palpable mass or organomegaly, abdominal pain, hematuria, oliguria, & organ dysfunction suspected by laboratory values. US is also increasingly used for bowel abnormalities outside of intussusception.

Sonographic modifications for children include the selection of an appropriate transducer & distraction of the patient during scanning.

Abdominal CT

Common indications for pediatric abdominal CT include trauma, abdominal pain, suspected appendicitis with an equivocal US, complications of pancreatitis, & abscess. It should be noted, however, that, outside of trauma, alternative imaging strategies (including US & MR) are increasingly replacing CT due to the concerns of ionizing radiation.

Modifications for children include the use of weight/size-based protocols & automatic tube current modulation techniques.

The use of IV &/or oral contrast should be driven by the specific clinical indication.

Abdominal MR

MR imaging uses no ionizing radiation & provides excellent soft tissue contrast.

Common indications for pediatric abdominal MR include the further investigation of a mass, surveillance after the treatment of a cancer, assessment of inflammatory bowel disease & pancreatobiliary disorders, liver quantification (of iron, fat, &/or fibrosis), & utilization as an alternative modality for suspected appendicitis.

Specific indications will help drive the selection of the appropriate contrast agent & protocol. Diffusion-weighted imaging can be particularly helpful in increasing the conspicuity of inflammatory & neoplastic pathologies against collapsed normal bowel loops.

Modifications for children include the use of video goggles for distraction, pretest practice/simulation to prepare young patients for the noise & relatively small bore size, & the use of sedation or general anesthesia for most patients under 6 years of age.

Differential Diagnoses

Neonatal Bowel Obstruction
High intestinal obstruction (with radiographs showing a few dilated proximal loops with a paucity of distal bowel gas, ± bilious emesis) should be evaluated with an upper GI. Differential considerations in this setting will include malrotation & midgut volvulus, duodenal atresia, duodenal web, annular pancreas, & jejunal atresia.

Low intestinal obstruction (with radiographs showing many dilated loops, ± bilious emesis) should be evaluated with a water-soluble contrast enema. Differential considerations in this setting will include Hirschsprung disease, meconium plug/small left colon syndrome, meconium ileus, ileal atresia, & anorectal malformation.

Specific Neonatal Disorders
Esophageal atresia with a tracheoesophageal fistula (TEF) may show a distended proximal esophageal pouch containing the tip of an enteric tube. A proximal atresia with a distal TEF is the most common type, & air is typically seen in the stomach & intestines.

Necrotizing enterocolitis typically occurs in premature neonates & presents as fixed, unfolded, asymmetrically dilated bowel loops ± pneumatosis, portal venous gas, & pneumoperitoneum.

Neonatal abdominal masses in the GI tract include a duplication cyst, mesenteric cyst, obstructed bowel loop, meconium pseudocyst, choledochal cyst, hepatic hemangioma, hepatoblastoma, & mesenchymal hamartoma. Abdominal masses in the neonatal GU tract include hydronephrosis, mesoblastic nephroma, Wilms tumor, polycystic kidneys, neuroblastoma, bladder outlet obstruction, hydrocolpos, ovarian cyst, & teratoma.

Vomiting Infant
If bilious emesis is present, an emergent upper GI series is the study of choice to look for a proximal obstruction such as malrotation/midgut volvulus (even though bilious emesis in a neonate is more commonly due to a distal bowel obstruction). The diagnoses of malrotation & midgut volvulus are classically made on an upper GI, though they can potentially be made by other modalities as well.

If nonbilious emesis is present, US is an ideal test for the evaluation of suspected HPS. HPS is typically not found in newborns but in infants 2-12 weeks old with progressive nonbilious projectile vomiting.

Bowel Obstruction Beyond Neonates
The differential diagnosis for obstruction in this setting can be remembered by the mnemonic AAIIMM (appendicitis, adhesions, intussusception, inguinal hernia, Meckel diverticulum, malrotation/midgut volvulus).

Abdominal Tumors
Children are afflicted with a unique set of tumors. The most common abdominal malignancies affecting children are Wilms tumor, neuroblastoma, & hepatoblastoma. The remaining malignancies are all considered rare tumors.

Wilms tumor is the most common pediatric malignancy of the kidney. It invades vessels (i.e., the renal vein & inferior vena cava), uncommonly has Ca^{2+} (in approximately 15%), & rarely crosses midline.

Neuroblastoma is the most common extracranial solid tumor in children. It most commonly arises from the adrenal gland or paraspinal sympathetic ganglia. It often encases & displaces vessels, crosses the midline, & contains Ca^{2+} (~ 90%).

Hepatoblastoma is the most common pediatric hepatic malignancy. It is typically a large heterogeneous solid liver mass, usually presenting outside the newborn period. This lesion has Ca^{2+} in 50% of cases & typically causes an ↑ α-fetoprotein.

Pediatric Abdominal Trauma
Trauma is the leading cause of morbidity & mortality in children. Pediatric abdominal trauma is responsible for ~ 10% of trauma-related deaths, with 90% due to motor vehicle collisions, bicycle handlebar injuries, sports injuries, falls, & ATV accidents.

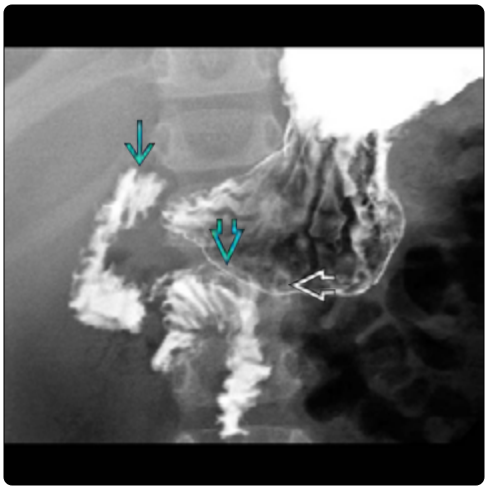
Children are at increased risk of abdominal traumatic injury as their visceral organs are proportionately larger compared to adult organs (leading to greater force per body surface area). Additionally, there is little fat to cushion direct blows.

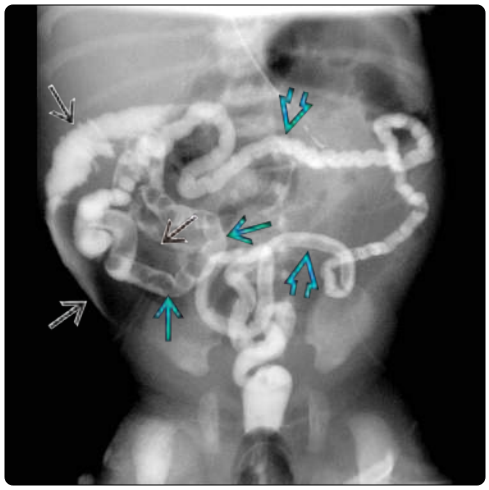
Approximately 60-80% of children with seat belt contusions have intraabdominal injuries in the plane of the seat belt. Rapid deceleration in these cases causes hyperflexion with compression of the abdominal viscera. Injuries of the small & large bowel, mesentery, liver, spleen, pancreas, kidneys, & lumbar spine may occur.

Abusive abdominal trauma is the 2nd most common form of fatal child abuse. The liver & spleen are the most commonly affected organs, but pancreatic or bowel trauma without explanation should also raise suspicion.

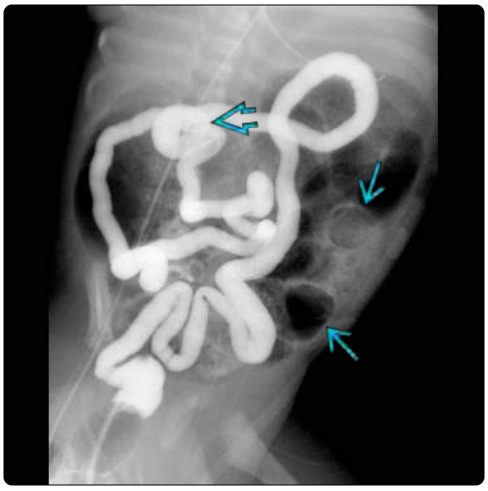
Hypoperfusion complex/shock bowel presents with diffuse wall thickening & hyperenhancement. Diminished calibers of the aorta & inferior vena cava are described but may not be seen due to fluid bolus before the CT scan. There may also be free fluid & abnormal solid organ enhancement.

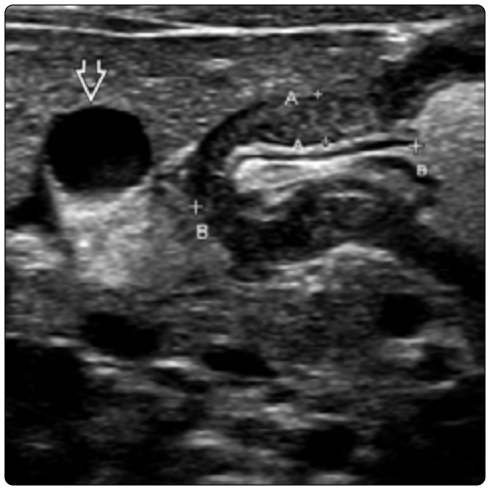
(Left) *Supine upper GI in a 3 year old with malrotation shows the duodenaljejunal junction ⇨ below the level of the duodenal bulb ⇨ & right of the left vertebral pedicle ⇨. This anomaly predisposes children to the surgical emergency of midgut volvulus.* **(Right)** *Supine contrast enema in a 1-day-old boy with bilious emesis & abdominal distention shows a microcolon ⇨ & filling defects of meconium ⇨ extending from the terminal ileum to the dilated loops ⇨ of mid ileum. The findings are consistent with meconium ileus.*



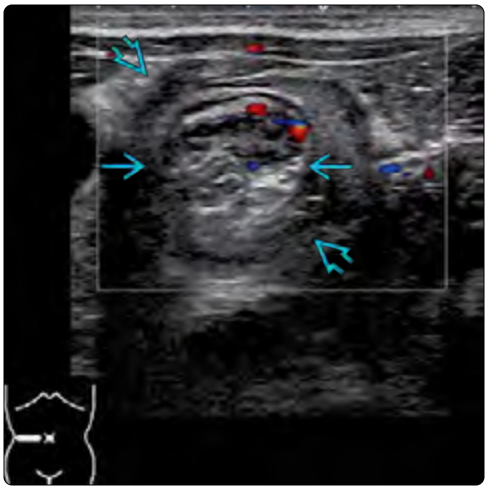


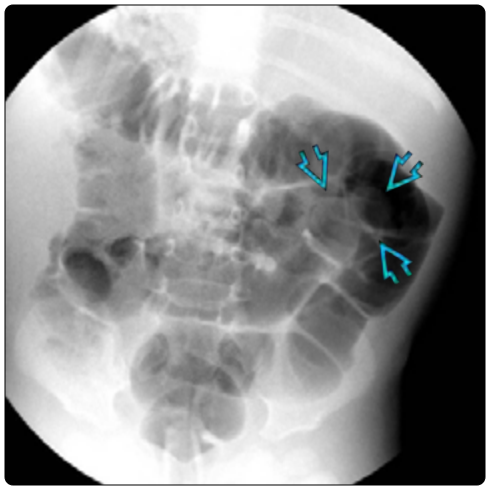
(Left) *Oblique contrast enema in a 2 day old with failure to pass meconium & progressive abdominal distention shows a microcolon. There is reflux of contrast into a blind-ending ileum ⇨, consistent with ileal atresia. Note the dilated air & meconium-filled bowel ⇨ proximally.* **(Right)** *Pyloric US in a 6-week-old boy with 2 days of projectile nonbilious emesis shows a thickened (A) & elongated (B) pylorus, consistent with hypertrophic pyloric stenosis. The correct plane of imaging often includes the gallbladder ⇨.*

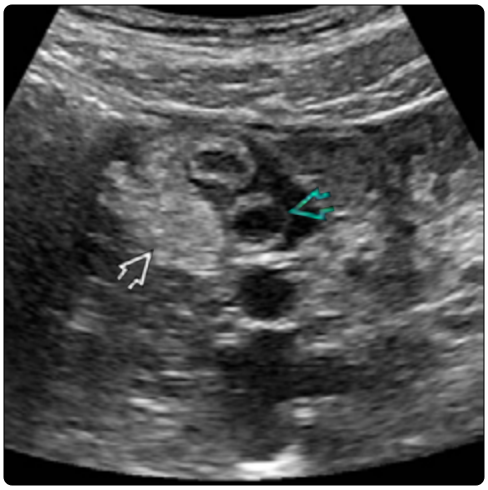


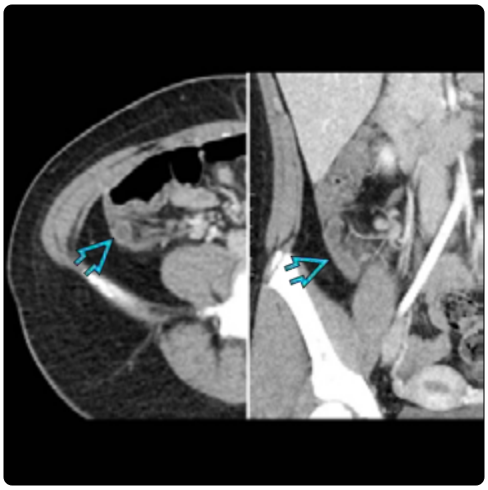


(Left) *Transverse US of the right lower quadrant in a 16-month-old boy with intermittent abdominal pain, bilious emesis, & bloody stools shows the target sign of an ileocolic intussusception. The concentric circles of the target sign are made up from the inner intussusceptum ⇨ & the outer intussuscipiens ⇨.* **(Right)** *Prone air-contrast enema performed in the same patient shows the intussusceptum ⇨ of the ileocolic intussusception. The reduction continues until there is free reflux of air into the small bowel (not shown).*

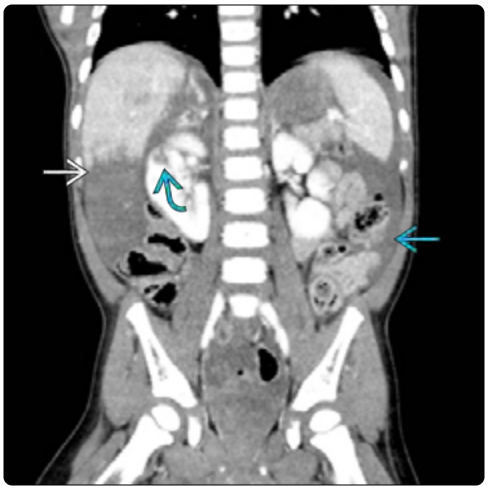


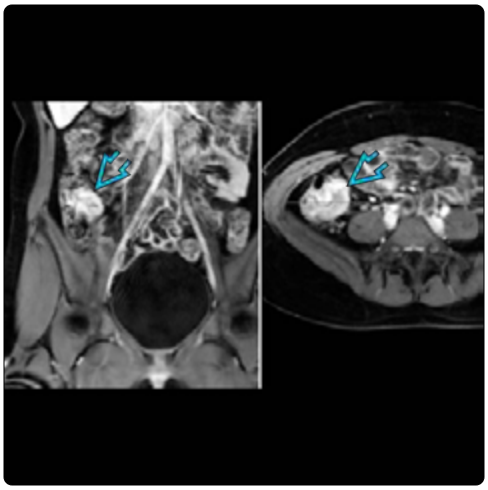




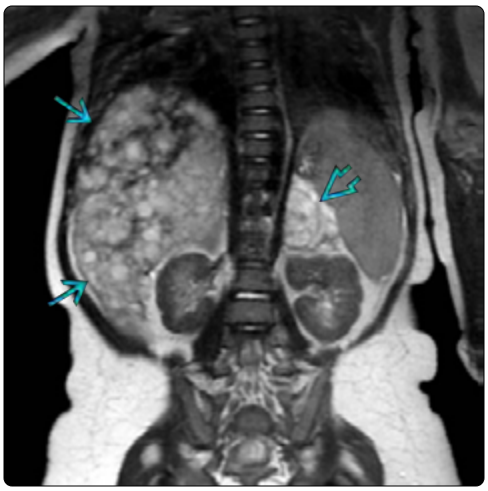


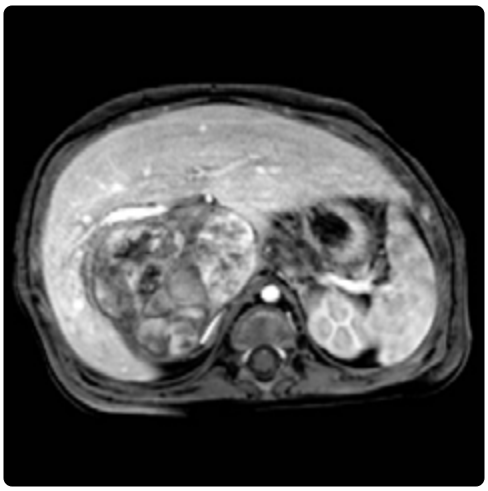
(Left) *Transverse US in an adolescent with appendicitis shows interruption of the appendiceal wall & induration/inflammation of the periappendiceal fat.* **(Right)** *Abdominal CECT images in an 11 year old with right lower quadrant pain, vomiting, & leukocytosis show a dilated retrocecal appendix with enhancement, adjacent fat stranding, & conal fascial thickening, consistent with acute appendicitis.*





(Left) *Abdominal CECT in a 3-year-old restrained boy involved in a motor vehicle collision shows lacerations of the liver & right kidney with hemoperitoneum.* **(Right)** *T1 C+ FS MR enterography images in a 15 year old with newly diagnosed Crohn disease show mucosal hyperenhancement & wall thickening of the terminal ileum, consistent with active inflammation.*





(Left) *Coronal T2 MR of the abdomen in a 4 month old with metastatic neuroblastoma shows a left adrenal mass & replacement of the hepatic parenchyma by metastases. The liver is the most common solid organ affected by neuroblastoma metastases.* **(Right)** *Axial T1 C+ FS MR in a 16 month old with hepatoblastoma shows a heterogeneously enhancing mass arising from the right lobe of the liver. Hepatoblastoma is the most common primary hepatic malignancy in children.*

Normal Variations of Duodenojejunal Junction Position

KEY FACTS

TERMINOLOGY

- Ligament of Treitz (LOT): Combination of muscle & fibrous tissue, very pliable
  - Normal LOT position implies normal bowel rotation & fixation not at risk of midgut volvulus

IMAGING

- LOT not directly depicted on any imaging study
- LOT position inferred by imaging depictions of duodenum, duodenojejunal junction (DJJ), & proximal jejunum
- Guidelines for normal position of duodenum & DJJ
  - Supine upper GI: DJJ at or leftward of left vertebral pedicle & nearly as craniad as duodenal bulb
  - Lateral upper GI: D2-D4 overlap posteriorly close to spine; DJJ superimposes on or adjacent to duodenal bulb
  - US/CECT/MR: D3 crosses midline to left between SMA & aorta; DJJ near same vertical level as duodenal bulb; SMA left & posterior of superior mesenteric vein

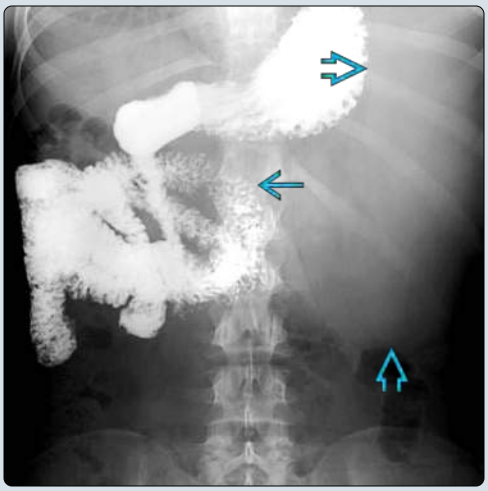
TOP DIFFERENTIAL DIAGNOSES

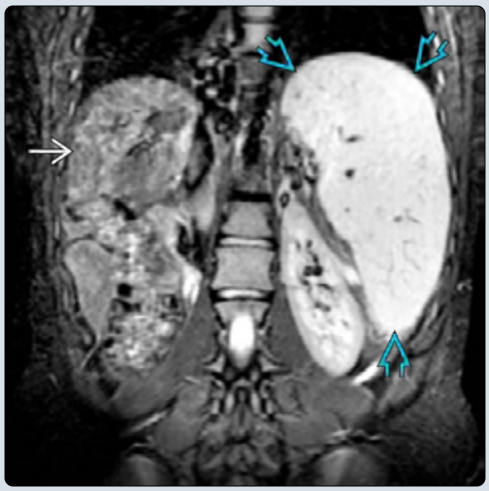
- True malrotation
- Artifactual displacement of normal DJJ
  - By mass or organomegaly
  - By bowel dilation
  - By enteric tube
- Redundant duodenum
- Duodenum inversum

CLINICAL ISSUES

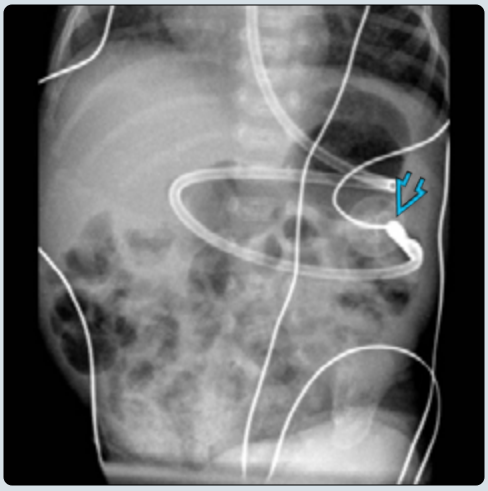
- If DJJ position unclear but suspicious for malrotation, convert study to small bowel follow-through
- Treatment: None for normal variations
  - Importance lies in distinguishing normal variant from malrotation, which can predispose to life-threatening midgut volvulus
    - Malrotation requires surgical Ladd procedure to prevent midgut volvulus

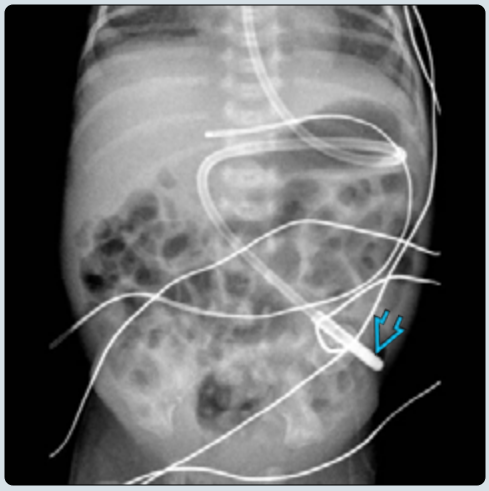
(Left) *Supine image from an upper GI shows an abnormal position of the duodenojejunal junction (DJJ) & proximal small bowel (SB) in the right upper quadrant, highly suggestive of malrotation. However, the child was not malrotated but had splenomegaly displacing the DJJ & SB.* **(Right)** *Coronal STIR MR in the same patient shows splenomegaly & a small, cirrhotic liver. The combination of the large spleen & small liver has shifted the proximal bowel into the right upper quadrant, mimicking malrotation.*





(Left) *AP radiograph in a 4-week-old infant with a nasojejunal feeding tube placed several days before this exam shows the tip of the tube at the laterally deviated DJJ.* **(Right)** *AP radiograph several hours later in the same infant shows that the tip of the tube (which denotes the position of the DJJ) has moved inferiorly, not due to malrotation but likely due to the torque of the tube & the pliability of the ligament of Treitz (LOT), which has both muscular & fibrous components.*





TERMINOLOGY

Abbreviations

- D1-D4: 1st through 4th duodenal segments

Definitions

- Ligament of Treitz (LOT): Combination of muscle & fibrous tissue; normal position implies normal midgut rotation & fixation not at risk of midgut volvulus
  - Upper portion of LOT (Hilfsmuskel) attached to diaphragmatic crus near esophageal hiatus; skeletal muscle & fibrous tissue component attaches to celiac axis
  - Lower portion of LOT (suspensory muscle of duodenum) inserts at duodenojejunal junction (DJJ) & has fibrous connection to celiac axis
  - Function
    - Involved in process of normal bowel rotation
    - With contraction, suspensory muscle of LOT widens angle of duodenojejunal flexure, allowing movement of intestinal contents

IMAGING

General Features

- Location
  - LOT not directly depicted on any imaging study
  - LOT position inferred by imaging depictions of duodenum, DJJ, & proximal jejunum
    - Normal DJJ suspended by LOT: Attached craniad by diaphragmatic crus near esophageal hiatus & DJJ in left upper quadrant of abdomen
- Morphology
  - LOT very pliable structure
    - Composed at least partly of muscle
    - Normal position of DJJ can be displaced by organomegaly, mass, adjacent bowel distention, or enteric tubes
  - Normal duodenum may have variant courses leading up to normal DJJ

Fluoroscopic Findings

- Upper GI
  - Supine image
    - DJJ at or leftward of left vertebral pedicle
    - DJJ nearly as craniad as duodenal bulb
  - Lateral image
    - D2-D4 course posteriorly, overlap anterior to spine
      - Cannot confirm true retroperitoneal position on upper GI
    - DJJ superimposes on or adjacent to (as craniad as) duodenal bulb

Ultrasonographic Findings

- Does not visualize exact DJJ position
- Normal midgut rotation implied if
  - D3 crosses midline to left between aorta & superior mesenteric artery (SMA)
    - Not seen with malrotation
  - Normal SMA to left & slightly posterior to superior mesenteric vein
    - Some false-positives & false-negatives

CT Findings

- Multiplanar CECT reconstructions can demonstrate normal retroperitoneal D3 course between aorta & SMA + appropriate height of DJJ in many cases

DIFFERENTIAL DIAGNOSIS

True Malrotation

- Abnormal DJJ position
- ± duodenal dilation/obstruction (Ladd bands or superimposed midgut volvulus)
- Cross-sectional imaging in difficult cases: Assess D3 course in relation to aorta & SMA
  - US reassuring but 100% reliability debated

Artifactual Displacement of DJJ

- By gastrojejunostomy or nasojejunal tube
- By adjacent solid or cystic abdominal mass
- By organomegaly (liver, spleen, kidney)
- By adjacent dilated viscus
  - Obstructed jejunal, ileal, or colonic loops
  - Marked gastric distention can displace DJJ caudally

Redundant/Wandering Duodenum

- D2-D4 segments may be long & redundant, ultimately passing to left of spine to normal DJJ position

Duodenum Inversum

- Duodenum descends & ascends right of spine before coursing left to normally located DJJ
  - If D3-D4 traverses left above L1 level across gastric body, likely abnormal but not typical malrotation
    - Susceptible to duodenal obstruction & compression by adjacent structures, not volvulus
  - If D3-D4 courses to left below L1 level to normal DJJ, likely lies between aorta & SMA with normal rotation

CLINICAL ISSUES

Treatment

- None for normal variations
- Importance lies in distinguishing from malrotation, which can predispose to life-threatening midgut volvulus

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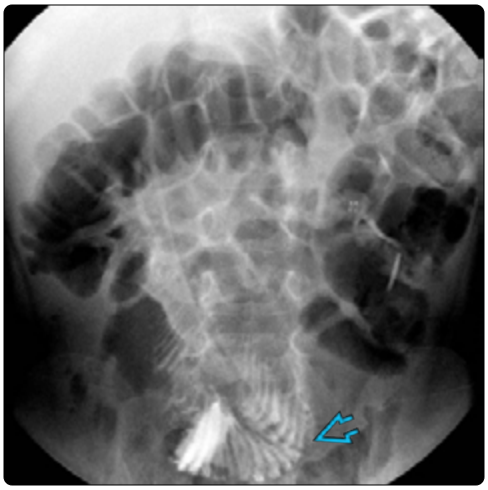
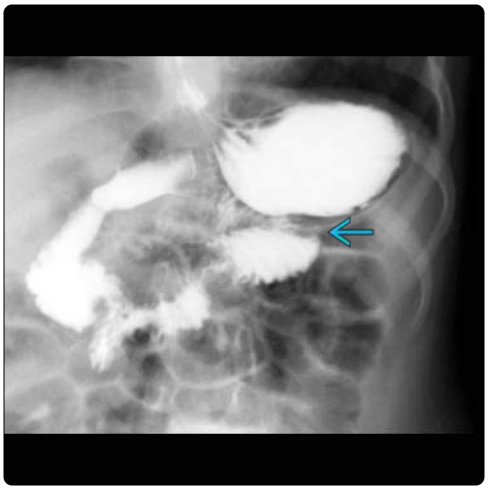
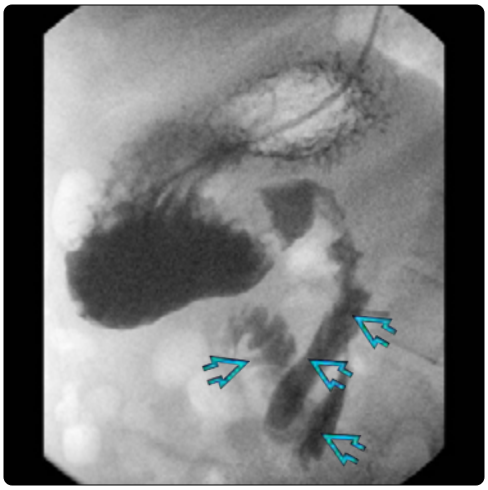
9. Long FR et al: Intestinal malrotation in children: tutorial on radiographic diagnosis in difficult cases. Radiology. 198(3):775-80, 1996

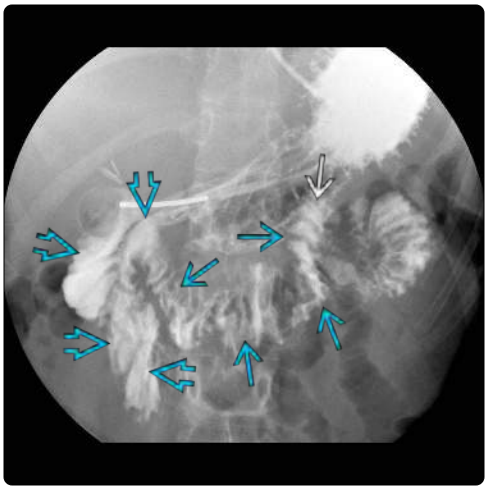
(Left) *Supine frontal radiograph during contrast injection to check the function of a gastrojejunostomy (GJ) tube shows the tip of the tube ⇨ at the DJJ, which is in the pelvis. One might conclude that the bowel must be malrotated to create this appearance.* **(Right)** *Supine upper GI image from the same patient prior to the GJ tube placement shows a normal location of the DJJ ⇨. Tubes frequently alter the DJJ position due to the pliability of the LOT, potentially displacing the DJJ anywhere in the abdomen.*



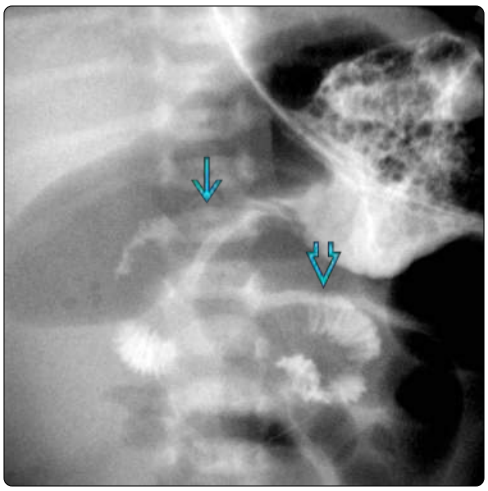


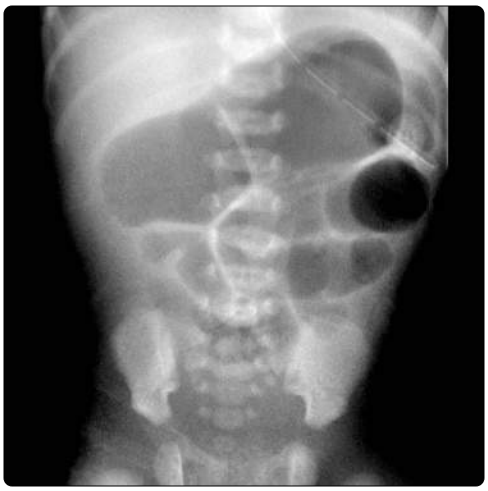
(Left) *Lateral upper GI image in a 17-year-old girl with abdominal pain & nausea shows significant redundancy of the retroperitoneal D2 & D3 segments ⇨.* **(Right)** *Supine frontal upper GI image in same patient shows that the D2 & D3 segments ⇨ are to the right of the spine, a configuration known as duodenum inversum. Note that the remainder of the duodenum is redundant as well but courses caudad & to the left ⇨ to reach a normal DJJ ➡.*

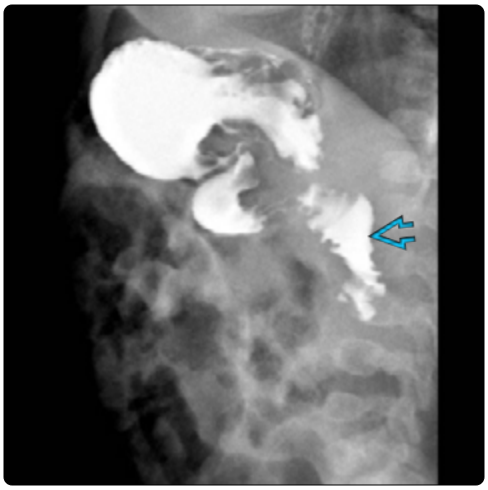


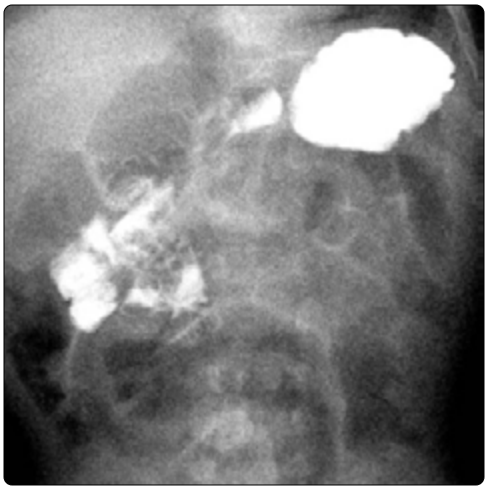


(Left) *Supine frontal upper GI image in a 2 day old with bilious emesis shows a low position of the DJJ ⇨ compared to the duodenal bulb ⇨. This appearance is likely related to the dilated, air-filled distal bowel loops seen in the background. Such dilated loops can displace the DJJ.* **(Right)** *Supine scout fluorograph in the same patient shows the multiple, dilated air-filled bowel loops of a congenital intestinal obstruction. Such loops can displace the DJJ & simulate malrotation.*

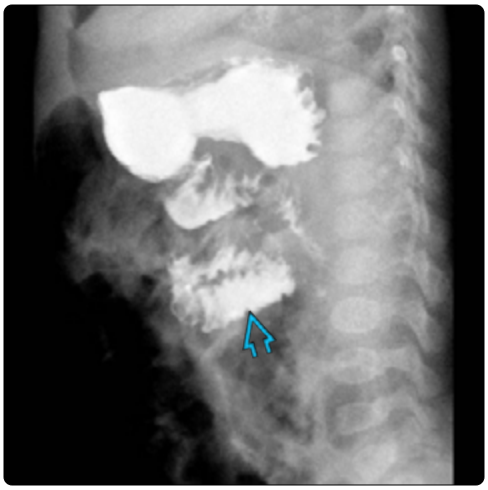


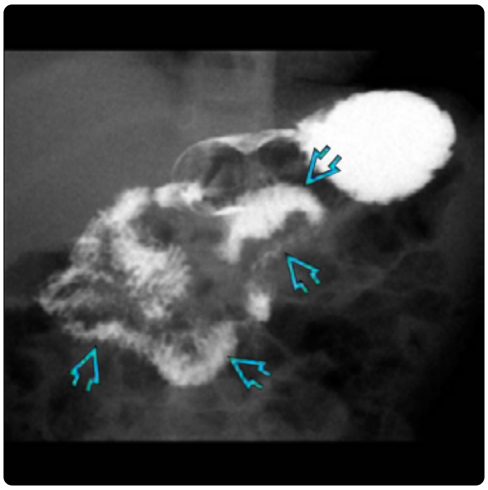




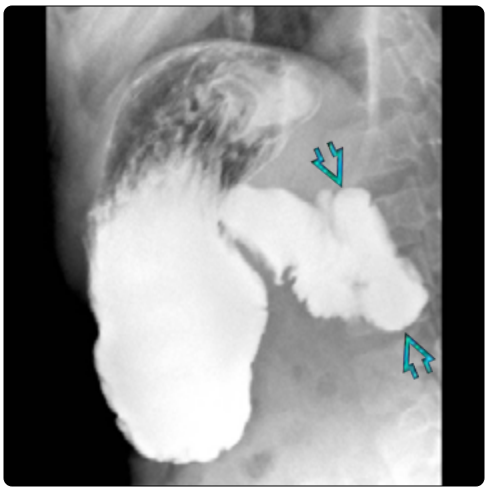


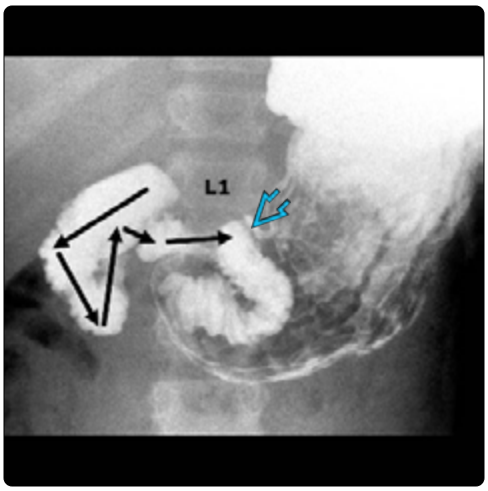
(Left) *Lateral upper GI image in a 2-month-old boy with failure to thrive & recurrent nonbilious vomiting shows the typical D2 retroperitoneal segment* ⇨. **(Right)** *Supine frontal upper GI image in the same patient shows that as the contrast progresses, multiple redundant loops of the duodenum are seen to the right of the spine, suggestive of malrotation without obstruction.*





(Left) *Lateral upper GI image taken later in the same study shows the more distal loops coursing anteriorly* ⇨, *suggesting an intraperitoneal location of segments D3 & D4.* **(Right)** *Final supine upper GI image in the same patient shows a loop of SB coursing craniad to the left* ⇨ *to the expected region of the LOT. The exam was converted to a SBFT & showed a high cecum near the hepatic flexure, concerning for malrotation. In the OR, the duodenum was retroperitoneal & normally fixed at the LOT, consistent with wandering duodenum.*





(Left) *Lateral upper GI image in a 9 year old with recurrent nonbilious emesis, nausea, & abdominal pain shows a normal retroperitoneal duodenum* ⇨. **(Right)** *Supine frontal upper GI image of the same patient shows a redundant course (denoted by black arrows) of the D2 & D3 segments to the right of the spine (duodenum inversum). The duodenum then courses to the left at the L2 level (likely between the aorta & SMA with the SMA takeoff usually at L1) to the DJJ* ⇨ *at the left pedicle, a normal variant course.*

Normal Variations of Cecal Position

KEY FACTS

TERMINOLOGY

- Slight mobility of normally fixed cecum, allowing variation in normal cecal position
- Clinical significance of cecal position
  - Often abnormal in malrotation
    - If adjacent to duodenum, suggests short mesenteric pedicle → ↑ risk of midgut volvulus (whether or not duodenal malrotation present)
  - If air-filled, essentially excludes ileocolic intussusception
    - Best seen with left side down decubitus position
  - In evaluation for appendicitis, appendix location follows cecum

IMAGING

- Best modality: Fluoroscopic or cross-sectional exam
- Fluoroscopy (contrast enema or SBFT)
  - Normal cecum can be slightly: High, low in pelvis, or medially directed
  - If sigmoid on right → craniad displacement of cecum
- Protocol advice
  - If questionably abnormal duodenal rotation on upper GI, get SBFT & delayed images until sure of cecal position
  - If abnormal cecal position suggested on SBFT, evaluate entire colon with delayed images
  - If cecum seen in abnormal position, look at prior studies
    - If previously normal, likely slightly mobile (variant)

TOP DIFFERENTIAL DIAGNOSES

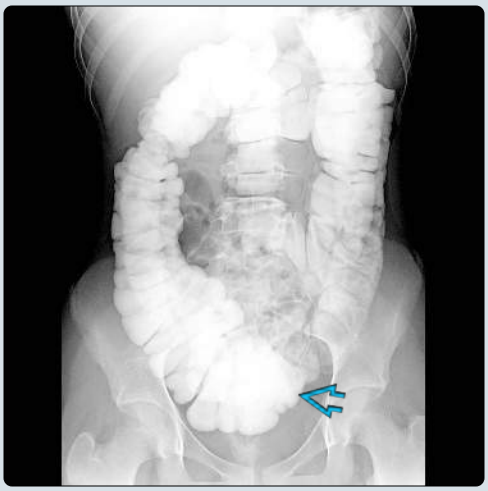
- Malrotation
- Internal hernia
- Postoperative appearance of Malone appendicostomy

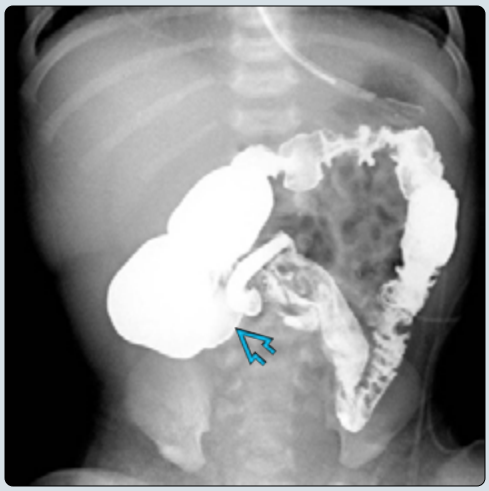
CLINICAL ISSUES

- Usually no clinical significance (other than differentiating from truly pathologic cecal position)

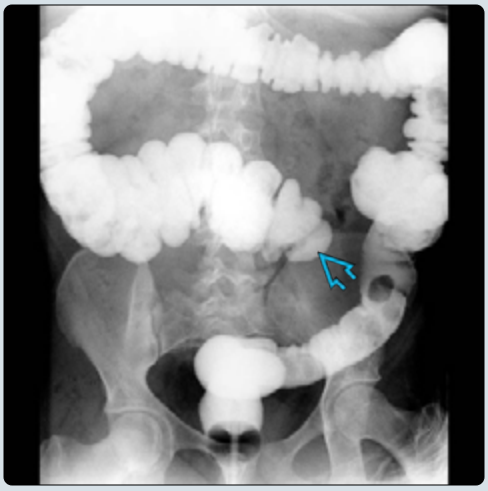
DIAGNOSTIC CHECKLIST

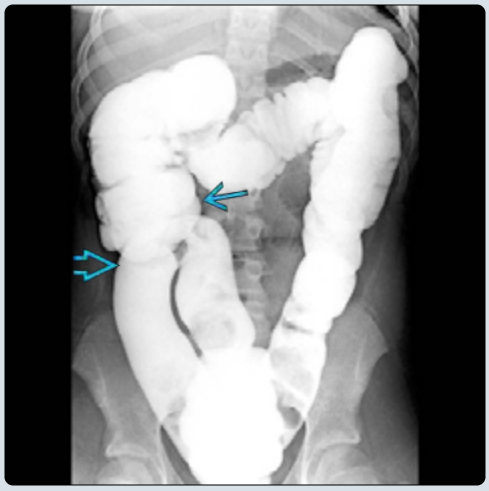
- If slightly high, low, or medially directed cecum but normal position of remaining right colon, consider normal variation





(Left) *Supine contrast enema of a teenager with a history of constipation shows the proximal colon in the deep pelvis with the cecal tip lying left of midline such that the appendix could potentially lie on the left rather than the right. This is a variation of normal cecal position.* **(Right)** *Delayed supine SBFT colonic image of a 4 week old shows a medially directed cecum that was noted to be mobile in comparison to a preceding contrast enema (not shown). The medial mobile cecum is within normal limits.*





(Left) *Supine contrast enema in a teenager with a history of an anorectal malformation & a Malone appendicostomy for bowel management shows surgical mobilization of the cecum medially to allow the appendix to reach the umbilicus.* **(Right)** *Supine contrast enema of a 12 year old with chronic constipation shows a long, redundant right-sided sigmoid colon with elevation of the cecum, a normal variant. On postevacuation images (not shown), the cecum was more caudally located (as expected).*

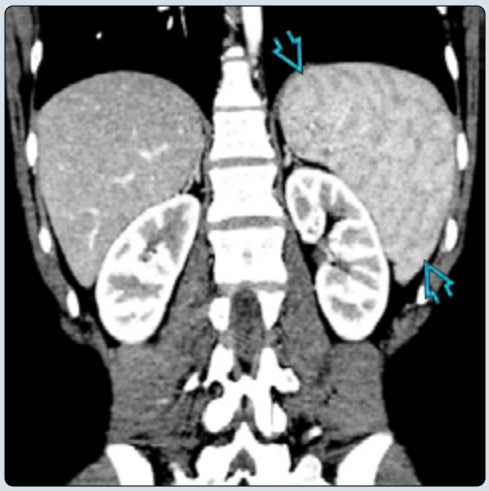
KEY FACTS

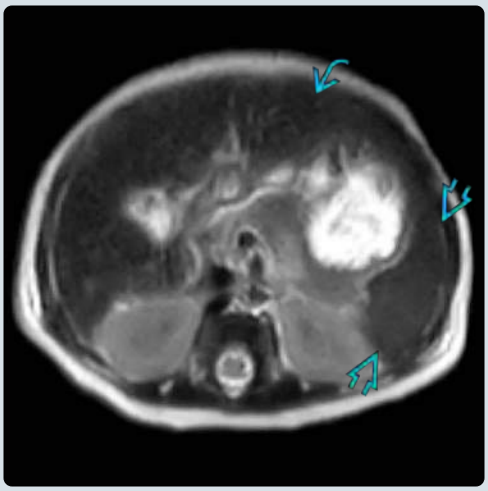
IMAGING

- Marked changes occur in appearance of normal spleen during early childhood
  - Related to evolving white pulp/red pulp volume ratios
- Normal T2 MR signal
  - Older child & adult: Hyperintense spleen vs. liver
  - Neonate: Spleen iso- or hypointense vs. liver
  - Normal adult appearance occurs by ~ 8 months of age
- CECT/MR
  - Transient patterns of heterogeneous enhancement occur when spleen imaged in arterial phase
    - Should become homogeneous on more delayed phases of imaging
  - Seen in most children during arterial phase
    - Less commonly encountered in children < 1 year old, also likely due to infantile white pulp to red pulp ratios
  - Patterns of splenic heterogeneity include
    - Archiform (alternating curvilinear & undulating bands of low & high attenuation/signal intensity)
    - "Zebra stripe"
    - Focal
    - Diffusely heterogeneous
- Ultrasound
  - Normal spleen homogeneous: Iso- to slightly hyperechoic relative to liver
    - Red pulp & white pulp not differentiated/visualized

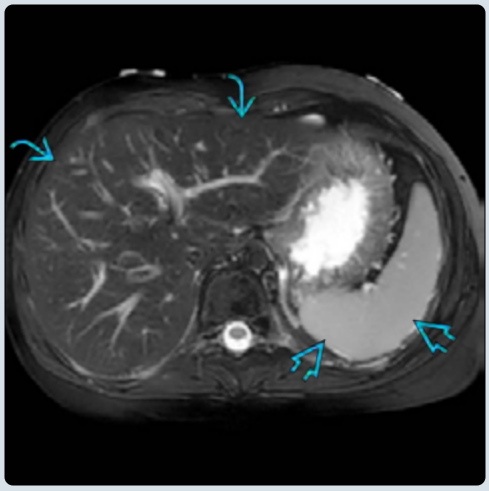
PATHOLOGY

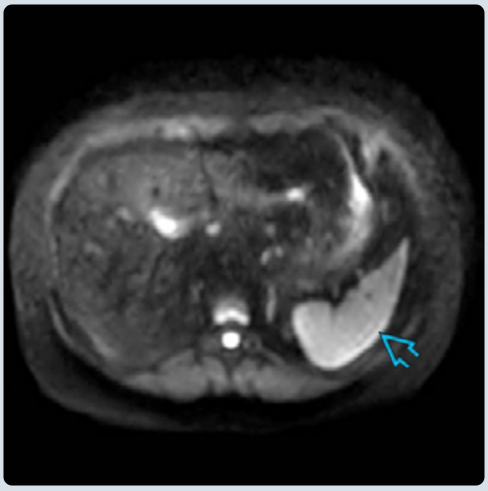
- Neonates & young infants have immature, small lymphoid follicles with relatively large red pulp volume
  - Accounts for changes in CT & MR appearances with age
- By ~ 7 months of age, white pulp to red pulp ratios have ↑, giving rise to adult appearance





(Left) *Coronal CECT in a 15-year-old boy demonstrates a normal transient pattern of heterogeneous splenic enhancement that is often seen when normal spleens (in children > 1 year of age) are imaged in the 1st minute after IV contrast injection (i.e., arterial or early phase).* **(Right)** *Axial T2 SSFSE MR in a 7-day-old girl being evaluated for an abdominal mass (not shown) demonstrates that the normal spleen is nearly isointense relative to the liver. This splenic appearance is due to a relatively high volume of red pulp at this age.*





(Left) *Axial T2 FS MR in a 9 year old undergoing MR enterography shows that the normal spleen is hyperintense relative to the liver. This change in appearance as compared to the neonate/young infant is due to an adult complement of white pulp.* **(Right)** *Axial diffusion-weighted MR in the same 9-year-old undergoing MR enterography shows that the normal spleen is mildly hyperintense.*

KEY FACTS

TERMINOLOGY

- Malrotation: Any abnormal rotation of small or large bowel, which rotate separately during development
- Rotation of duodenum/small bowel & cecum/large bowel are similar but differ in timing
  - Abnormalities of rotation may be either or both

IMAGING

- Fluoroscopic GI findings
  - D3 segment never crosses midline, often extends anteriorly on lateral view of upper GI
  - DJJ to right of left pedicle & below duodenal bulb on true frontal view of upper GI
  - Variable degrees of colonic malrotation with abnormal cecal position on enema or small bowel follow-through (SBFT)
- Cross-sectional imaging findings
  - Duodenal nonrotation: D3 segment of duodenum fails to pass between superior mesenteric artery (SMA) & aorta when crossing to left of midline
  - Reversal of normal SMA/superior mesenteric vein position (not reliable)
- Best imaging tool
  - Fluoroscopic upper GI vs. ultrasound debated

PATHOLOGY

- Numerous associated anomalies

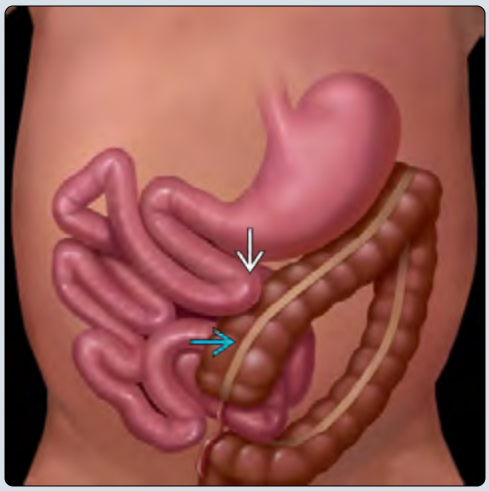
CLINICAL ISSUES

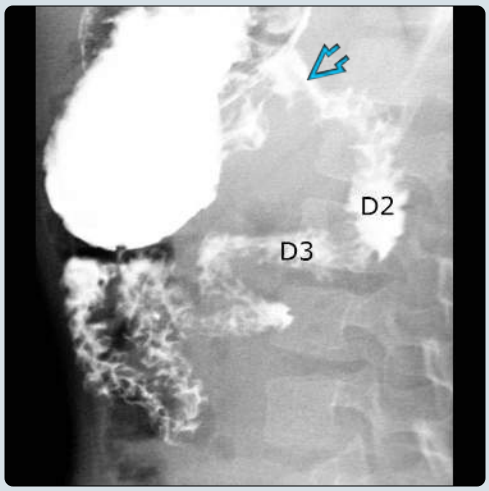
- Presentation in children: Nonbilious or bilious emesis, recurrent abdominal pain, poor weight gain, asymptomatic

DIAGNOSTIC CHECKLIST

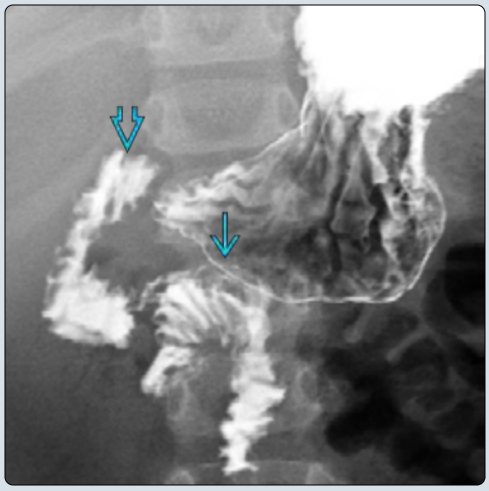
- If upper GI equivocal, obtain SBFT; document cecal position & estimate length of pedicle from DJJ to cecum (which determines risk for midgut volvulus)

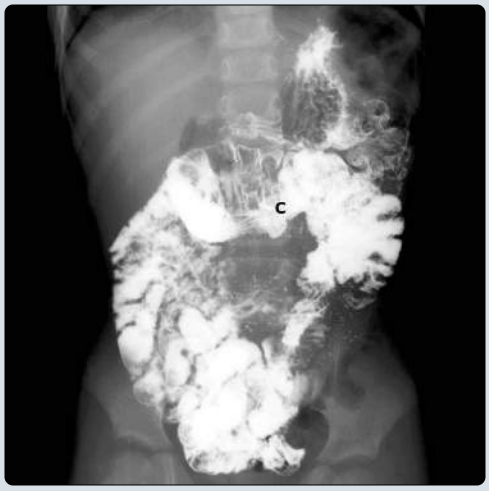
(Left) *Anterior graphic shows abnormal positions of the small & large bowel. The duodenojejunal junction (DJJ) lies low & midline, very close to the malpositioned cecum. This results in a short mesenteric fixation that predisposes to midgut volvulus.* **(Right)** *Lateral upper GI image in a 3-year-old child with a history of nonbilious vomiting shows an anterior, intraperitoneal course of the D3 segment with a low position of the DJJ below the duodenal bulb, indicating abnormal rotation.*





(Left) *Supine front view of the same patient shows a low DJJ (below the duodenal bulb) that fails to cross the midline, consistent with malrotation. Note: There is no twisting or dilation of the duodenum to suggest midgut volvulus or obstructing Ladd bands.* **(Right)** *Supine SBFT was continued in the same patient to determine the cecal position & estimate the length of the mesenteric pedicle. The cecum (C) is high & just left of the midline, suggesting a very short mesenteric pedicle that is at high risk of future midgut volvulus.*





TERMINOLOGY

Synonyms

- Malfixation

Definitions

- Malrotation: Varying degrees of abnormal positioning of small &/or large bowel due to abnormal rotation during development
  - Small & large bowel rotate separately in utero
  - Abnormalities of rotation may affect either or both
- Malfixation: Abnormal position or length of bowel fixation by mesentery, typically associated with malrotation
  - Predisposes to midgut volvulus (MV)

IMAGING

General Features

- Best diagnostic clue
  - Upper GI: Nonretroperitoneal position of duodenum + abnormal duodenojejunal junction (DJJ) position at or to right of midline
  - Cross-sectional imaging: Failure of D3 portion of duodenum to pass between aorta & superior mesenteric artery (SMA)
  - Enema or cross-sectional imaging: Abnormal configuration of colon
- Location
  - Duodenum & right colon
- Morphology
  - Abnormal rotation of duodenum, colon, or both; degree of abnormality quite variable (i.e., partial rotational anomalies)

Radiographic Findings

- Colon may be limited to left abdomen, small bowel in right abdomen

Fluoroscopic Findings

- Upper GI
  - D3 segment never crosses midline; often extends anteriorly on lateral view
  - DJJ to right of left pedicle on true frontal view
    - True frontal view
      - Right & left ribs are equal length
      - Base of heart in anatomic position
      - Vertebral pedicles appear symmetric
  - Jejunum often in right abdomen
  - ± abnormal cecal position
  - ± bowel obstruction due to Ladd (peritoneal fibrous) bands or MV
- Contrast enema
  - Variable degrees of colonic malrotation
    - High &/or midline cecum of partial rotation
    - Left-sided colon of nonrotation
    - Anything in between

CT Findings

- Malposition of intestine from expected location
  - Duodenal nonrotation: D3 segment fails to pass between SMA & aorta to cross left of midline
    - Duodenal partial rotation: D3 passes between SMA & aorta but abruptly turns right, coursing anterior or posterior to SMA
  - Nonrotated colon: Right colon in left lower abdomen
  - Partially nonrotated colon: Cecum in upper abdomen
- Reversal of normal SMA/super mesenteric vein (SMV) position (not reliable)
- ± bowel obstruction due to Ladd (peritoneal fibrous) bands or volvulus

MR Findings

- Similar to CT findings

Ultrasonographic Findings

- D3 segment of duodenum fails to pass between SMA & aorta to left of midline
- Reversal of normal SMA/SMV position (not reliable)
- Dilated proximal duodenum from Ladd bands or MV

Imaging Recommendations

- Best imaging tool
  - Fluoroscopic upper GI through DJJ
- Protocol advice
  - If upper GI is equivocal for malrotation, perform small bowel follow-through (SBFT)
    - Determine exact location of cecum; be confident, wait for adequate filling of cecum
    - Shorter distance of DJJ to cecum → shorter mesenteric attachment→ ↑ risk of volvulus
  - Normal position of DJJ can be displaced due to lax ligament of Treitz in infants
    - Adjacent dilated bowel of distal bowel obstruction
    - Adjacent masses, cysts, organomegaly
    - Enteric tube may distort duodenum

DIFFERENTIAL DIAGNOSIS

Duodenum Inversum

- Duodenum descends & ascends right of spine before coursing left to normally located DJJ
  - If D3-D4 traverses left above L1 level across gastric body, likely abnormal but not typical malrotation
    - Susceptible to duodenal obstruction & compression by adjacent structures, not volvulus

Redundant (Wandering) Duodenum

- D2-D4 segments may be long & redundant, ultimately passing to left of spine to normal DJJ position

PATHOLOGY

General Features

- Etiology
  - Failure of normal embryonic 270° counterclockwise rotation of midgut & colon, resulting in malposition of bowel to varying degrees
  - Ladd (peritoneal fibrous) bands attempt to fix abnormal duodenal &/or colonic positions
    - Potentially lead to extrinsic obstruction or volvulus
- Associated abnormalities
  - Malrotation almost always seen with left-sided Bochdalek congenital diaphragmatic hernia, gastroschisis, & omphalocele

- Malrotation commonly associated with duodenal atresia spectrum (including stenosis & web), small bowel atresia & stenosis, heterotaxia syndromes (asplenia & polysplenia), intestinal pseudoobstruction, Meckel diverticulum, Hirschsprung disease, biliary anomalies, anorectal malformation, intussusception, hypertrophic pyloric stenosis, absent kidney & ureter

Staging, Grading, & Classification

- Several types of malrotation
  - Complete nonrotation
  - Nonrotated duodenum & partially rotated colon
  - Isolated nonrotated duodenum
  - Partially rotated duodenum & colon
  - Isolated partially rotated duodenum
  - Isolated partially rotated colon
- Bottom line: Length of mesenteric fixation from DIJ to cecum determines risk for MV

Gross Pathologic & Surgical Features

- Ligament of Treitz: Actually muscle that can stretch
- Ladd bands can occur anywhere, frequently across D2 or D3 to liver hilum

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Children: Nonbilious or bilious emesis, recurrent abdominal pain, poor weight gain, or may be asymptomatic
  - Adults: Nonspecific → chronic vomiting, intermittent colicky abdominal pain, diarrhea
    - ± acute abdomen

Demographics

- Age
  - Majority present in 1st month of life
  - Vast majority present by 1st few years of life
  - Can present into adulthood
- 1/200 live births have asymptomatic rotational anomaly
- 1/6,000 live births have symptomatic malrotation
- M:F = 2:1
- > 33% associated with congenital anomaly
- Patients with congenital diaphragmatic hernia, gastroschisis, & omphalocele almost always are malrotated but rarely volvulize (due to postop adhesions)

Natural History & Prognosis

- Complications of malrotation
  - MV: Twisting of midgut about SMA → vascular occlusion & potential bowel ischemia
  - Bowel obstruction due to Ladd band: Obstruction can be anywhere but can mimic MV clinically & on fluoroscopic upper GI
  - Internal hernia: Rare, usually due to duodenal malrotation with normal colonic rotation
    - Sac-like mass of malfixed bowel herniates posterior to right colic vein into right upper quadrant

Treatment

- Ladd procedure
  - Untwist volvulus if present
  - Divide Ladd bands if present
  - Reposition small & large intestine into right & left abdomen, respectively
    - Postoperative adhesions expected to secure bowel in place
  - ± appendectomy
  - Laparoscopy vs. laparotomy
    - Laparoscopic Ladd procedure results in ↓ perioperative complications & faster recovery but may yield ↑ risk of postoperative volvulus compared to open approach (due to fewer adhesions)

DIAGNOSTIC CHECKLIST

Consider

- Normal variants of duodenal anatomy or secondary causes of displacement resulting in fluoroscopic false-positive for malrotation
- If upper GI equivocal for malrotation, obtain SBFT & document cecal position to estimate length of mesenteric pedicle of fixation (DJJ to cecum)

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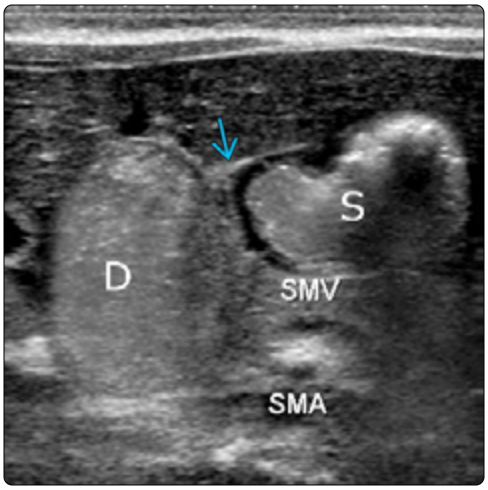
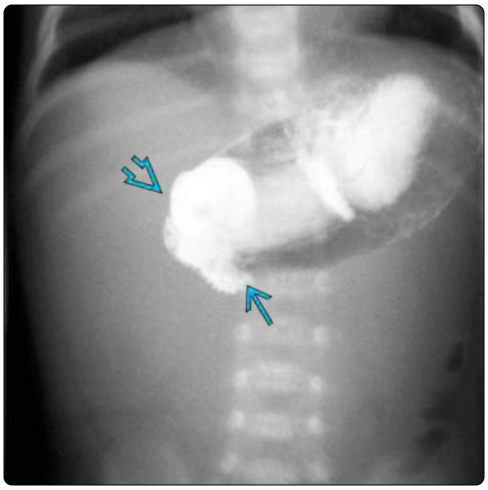
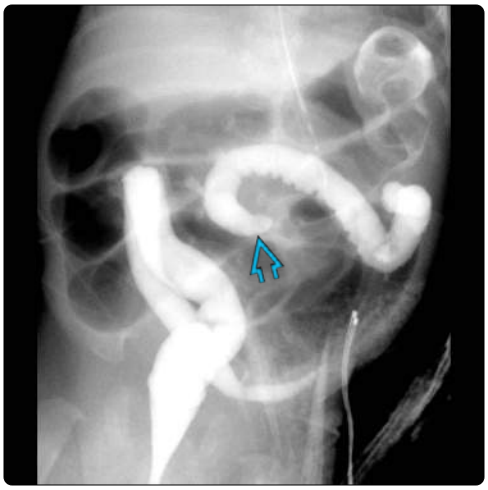
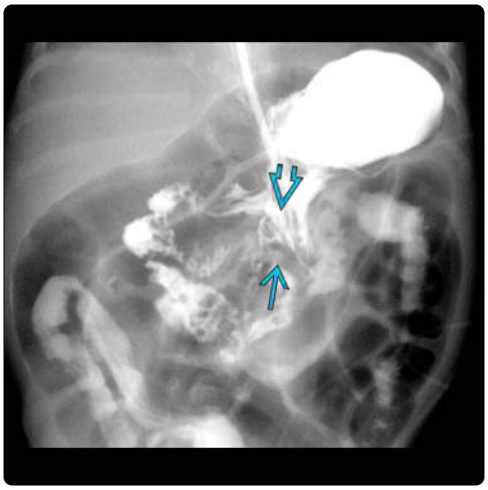


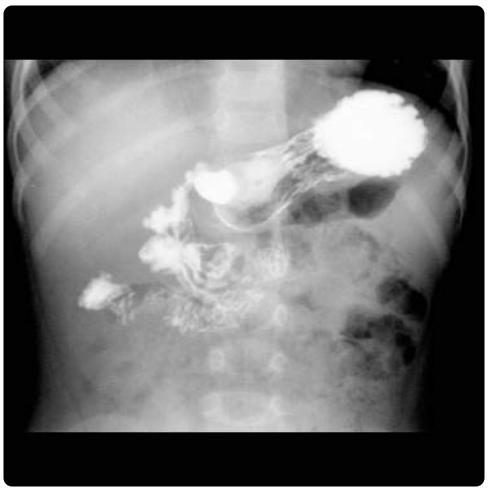


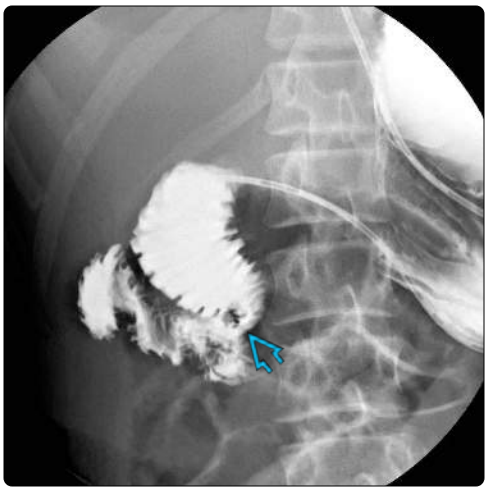
(Left) *Oblique pyloric ultrasound of an 18 day old with projectile nonbilious vomiting shows a dilated stomach (S) with a normal pylorus. There is a dilated duodenum (D), & the SMV lies directly anterior to the SMA, an abnormal configuration frequently seen in patients with malrotation.* **(Right)** *Supine upper GI in the same patient shows a dilated, obstructed duodenum up to a "beak" at D2-D3; this appearance represents midgut volvulus until proven otherwise (though only Ladd bands were found in this case).*





(Left) *Supine water-soluble contrast enema in a 5-week-old, former 31-week premature infant shows a high midline cecum with at least partial colonic malrotation. The small bowel dilation & small caliber colon may be due to an intervening stricture from prior necrotizing enterocolitis.* **(Right)** *Supine upper GI in the same patient shows normal duodenal rotation. If a line was drawn between the DJJ & cecum, the estimated mesenteric pedicle length would appear very short, increasing the risk of future volvulus.*





(Left) *Supine upper GI in a 2-year-old boy (who had malrotation corrected by a Ladd procedure at 6 days of age) presenting with abdominal pain & intermittent diarrhea shows malrotation without duodenal obstruction, a satisfactory postop appearance.* **(Right)** *Supine upper GI in a 13-year-old patient with new bilious emesis 6 days status post Ladd procedure for malrotation shows a dilated duodenum with a beak-like configuration worrisome for volvulus. In the OR, incompletely lysed bands were found.*

Midgut Volvulus

KEY FACTS

TERMINOLOGY

- Malrotation: Abnormal rotation & fixation of small bowel (SB) mesentery that can lead to complications
  - Bowel obstruction by Ladd bands
  - Midgut volvulus (MV) due to short mesenteric base, prone to twisting
- MV: Twisting of SB about superior mesenteric artery → bowel obstruction, ischemia/necrosis
- Ligament of Treitz: Suspends duodenojejunal junction (DJJ), defines normal duodenal rotation

IMAGING

- Most common radiographic appearance is normal
- Upper GI: Dilated duodenum to D2-D3 segment with corkscrew/spiral sign just beyond duodenal "beak"
- US or CT: Whirlpool sign

TOP DIFFERENTIAL DIAGNOSES

- Malrotation with obstructing Ladd band
- Spectrum of congenital duodenal obstruction
- Redundant duodenum

PATHOLOGY

- Potential ischemic or necrotic bowel

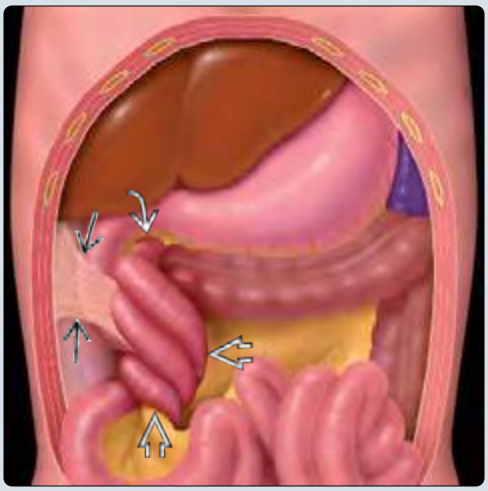
CLINICAL ISSUES

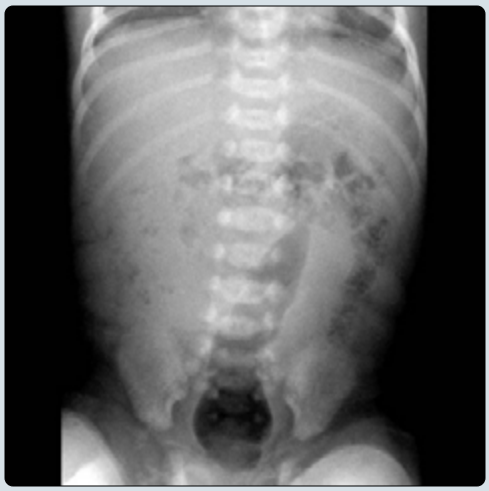
- Classic presentation: Infant with bilious vomiting
  - Requires emergent upper GI
- Treatment: Ladd procedure

DIAGNOSTIC CHECKLIST

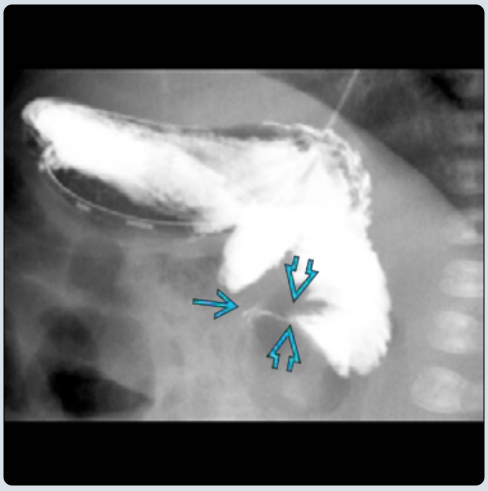
- Complete obstruction at D2-D3 with beak considered MV until proven otherwise
- Delayed diagnosis can lead to diffuse bowel necrosis or death

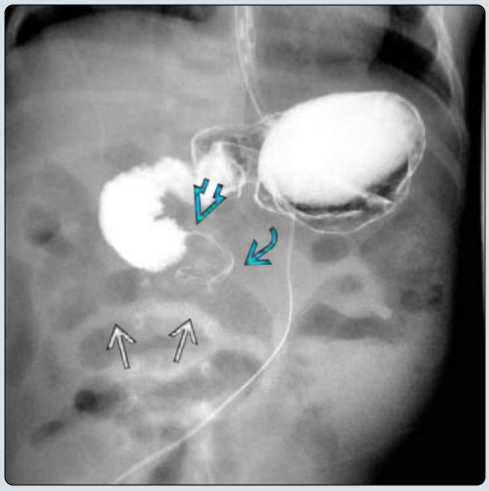
(Left) *Graphic shows a volvulus with twisted loops of proximal small bowel & a Ladd band. The cecum is malpositioned within the right upper quadrant.* **(Right)** *AP radiograph shows a nonspecific but nonobstructive bowel gas pattern in a patient with bilious emesis who was ultimately found to have midgut volvulus (MV) by upper GI. The most frequent radiographic gas pattern in MV is normal.*





(Left) *Lateral upper GI in a 3-day-old boy with bilious vomiting shows a dilated duodenum to D3 ending in a beak-like configuration with a wisp of contrast pointing anteriorly, highly suggestive of MV.* **(Right)** *Frontal upper GI image in the same patient (a few seconds later) shows duodenal dilation & partial duodenal obstruction at D3 with the corkscrew/spiral sign of MV. Thickened loops in this context suggest bowel ischemia.*





TERMINOLOGY

Definitions

- Malrotation: Abnormal rotation & fixation of small bowel (SB) mesentery that can lead to complications
  - Bowel obstruction by Ladd (peritoneal) bands
  - Midgut volvulus (MV) due to short mesenteric base prone to twisting
- MV: Abnormal twisting of SB about superior mesenteric artery (SMA) that can lead to bowel obstruction & bowel ischemia/necrosis
- Ligament of Treitz: Suspends duodenojejunal junction (DJJ), defines normal duodenal rotation
- Ladd band: Abnormal fibrous peritoneal bands that can cause duodenal obstruction
- Bilious vomiting: Green/yellow vomit typically from obstruction of duodenum distal to ampulla of Vater

IMAGING

General Features

- Best diagnostic clue
  - MV: Upper GI showing mildly to moderately dilated duodenum (usually through D2-D3 segment) with corkscrew or spiral sign at or distal to beak of obstruction
  - Whirlpool sign on US or CT: Wrapping of SB, its mesentery, & superior mesenteric vein (SMV) around SMA
  - Usually associated with malrotated bowel, either duodenal or colonic or both
- Morphology
  - Twisting of mesentery occurs about SMA, which can lead to venous obstruction, bowel wall ischemia, & necrosis
  - Ladd bands may cause bowel obstruction, especially of duodenum

Radiographic Findings

- Radiography
  - Most common early finding: Normal abdominal radiograph
  - Distended stomach & proximal duodenum with mild distal bowel gas very suggestive
    - **Not** marked longstanding dilation without distal gas, as seen in duodenal atresia
  - May show diffuse distal bowel distention/ileus from ischemia/necrosis
    - Such children often extremely ill
  - Rarely pneumatosis, portal venous gas, free intraperitoneal air

Fluoroscopic Findings

- Upper GI
  - Dilated duodenum to D2-D3, +/- "to-&-fro" motility due to obstruction
    - Degree of proximal duodenal dilation depends on chronicity
  - Often beaked appearance at level of twist, ± complete obstruction
  - Usually spiral/corkscrew appearance caudally, distal to beak
  - May see malrotation without MV
    - In patients with bilious emesis, this may reflect intermittent volvulus
- Contrast enema
  - Colon often nonrotated with cecum in upper midline abdomen ± obstruction of ileocecal region

Ultrasonographic Findings

- Proximal duodenum usually dilated
- Whirlpool sign of swirling vessels (SMV) & SB mesentery around SMA in clockwise fashion on grayscale & color Doppler
- SB may lack perfusion on color Doppler
- May see pneumatosis as foci of ↑ echogenicity with dirty shadowing within bowel wall circumferentially
- May see portal venous gas as punctate echogenic foci moving in portal vein(s) from liver hilum to periphery

CT Findings

- CECT
  - Whirlpool sign of swirling vessels (SMV) & SB mesentery around SMA
  - Potentially ↓ or no enhancement of SB due to obstruction of SMA (due to ischemia/necrosis)
  - May have SB distention due to ischemic ileus
  - Pneumatosis, portal venous gas, & rarely free peritoneal air present

Imaging Recommendations

- Best imaging tool
  - Infant with bilious vomiting → emergent upper GI
  - Small bowel follow-through (SBFT) or contrast enema if no volvulus seen but malrotation suspected to document position of cecum
    - Broadness of mesenteric base (DJJ-cecal distance) relates to potential risk of volvulus
- Protocol advice
  - In patients with high clinical suspicion of MV
    - Place nasogastric tube (if not already placed by clinicians)
    - Aspirate as much fluid & air from stomach as possible prior to instilling contrast
    - Inject 10 mL contrast into stomach in right lateral decubitus position
      - If not emptying into duodenum, inject small amounts of air to encourage gastric emptying
    - If volvulus seen, immediately notify referring clinicians
      - Longer time interval from diagnosis to operation makes intestinal ischemia & bowel loss more likely
    - Document duodenum in lateral & AP positions as per upper GI otherwise

DIFFERENTIAL DIAGNOSIS

Malrotation With Obstructing Ladd (Peritoneal Fibrous) Bands

- May be completely obstructive with beaking, mimicking MV
- Cannot distinguish from MV fluoroscopically if corkscrew sign not seen
  - US could distinguish by showing target/swirl sign of MV
- Must consider as tight MV & send for immediate surgical treatment

Spectrum of Congenital Duodenal Obstruction

- Duodenal atresia, duodenal stenosis, annular pancreas, duodenal web
  - Atresia has double bubble sign, marked duodenal dilation with **no** distal gas; usually D1-D2 obstructed
  - Stenosis or web usually has transition to normal distal duodenum & normal DJJ
    - Can mimic MV fluoroscopically if corkscrew sign not seen

Redundant Duodenum

- Duodenum may make several retroperitoneal loops prior to extending leftward across spine to normal DJJ
- **No** duodenal dilation or obstruction

PATHOLOGY

General Features

- Etiology
  - With normal rotation, DJJ positioned in left upper quadrant & cecum positioned in right lower quadrant
    - Results in long fixed mesenteric base between ligament of Treitz & cecum that keeps mesentery from twisting
  - If bowel malrotated, DJJ-cecal length (mesenteric base) is short, predisposing to twisting (volvulus)
    - Nonrotation contribution controversial; may be most common form in patients with volvulus (if both DJJ & cecum in midline with short pedicle)
    - Isolated duodenal or colonic malrotation may also predispose to MV
- Rarely, MV reported in setting of normal rotation; some of these cases may be segmental volvulus of ileum
- Malrotation may also be associated with duodenal obstruction from
  - Ladd bands (abnormal fibrous peritoneal bands)
  - Paraduodenal hernias

Microscopic Features

- Potential ischemic or necrotic bowel

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Classic presentation: Bilious vomiting in 1st month of life
    - However, can occur at any age, even in adulthood
- Other signs/symptoms
  - Acute abdominal pain
  - Vomiting, crampy abdominal pain
  - Failure to thrive
  - Patients may be asymptomatic or have atypical or chronic symptoms

Demographics

- Age
  - 39% present within first 10 days of life
  - > 90% present within first 3 months of life
  - Can occur at any age
- Gender
  - Slightly higher incidence in boys
- Epidemiology
  - 2.86/10,000 new births
  - Incidence inversely proportional to maternal age

Natural History & Prognosis

- Potential volvulus leading to bowel necrosis
- Possible MV is one of few true emergencies in pediatric GI

Treatment

- Surgical emergency
- Ladd procedure: Reduce volvulus, resect nonviable bowel, transect Ladd bands (if present), place SB in right & colon in left abdomen

DIAGNOSTIC CHECKLIST

Consider

- Delay in diagnosis can lead to diffuse bowel necrosis or death
- Infant with bilious vomiting → emergency upper GI
- Borderline cases of DJJ location (suspicious for malrotation) → SBFT or contrast enema to document location of cecum

Image Interpretation Pearls

- High-grade duodenal obstruction at D2-D3 with beaking (but no distal contrast passage) considered MV until proven otherwise
  - Should not be confused with classic double bubble sign of duodenal atresia in newborn
    - Markedly dilated round or ovoid proximal duodenum with no distal bowel gas
- If patient has MV by imaging, impression should read "midgut volvulus"
  - Must be communicated to clinician immediately

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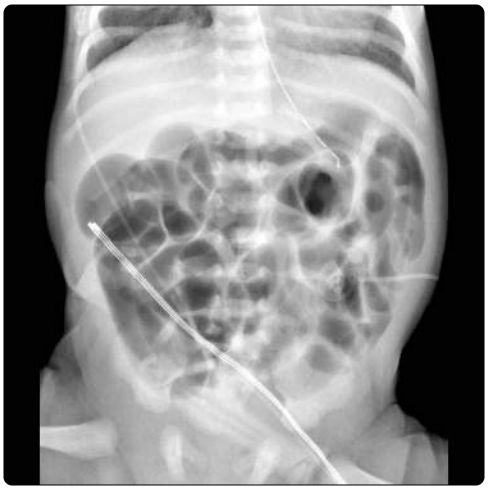
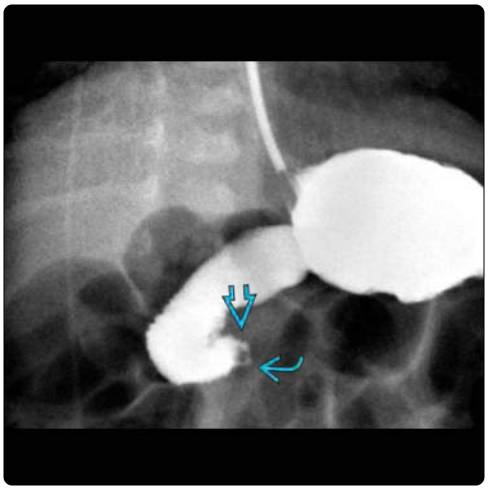
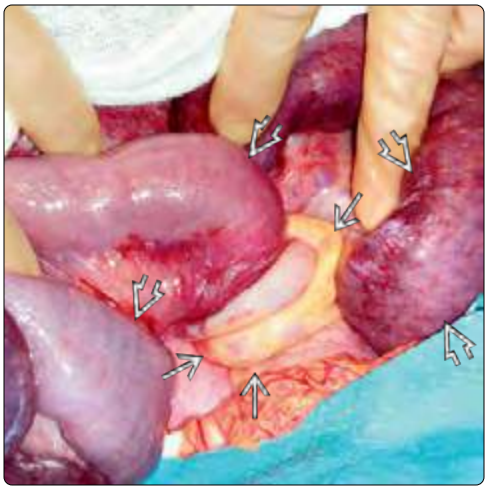
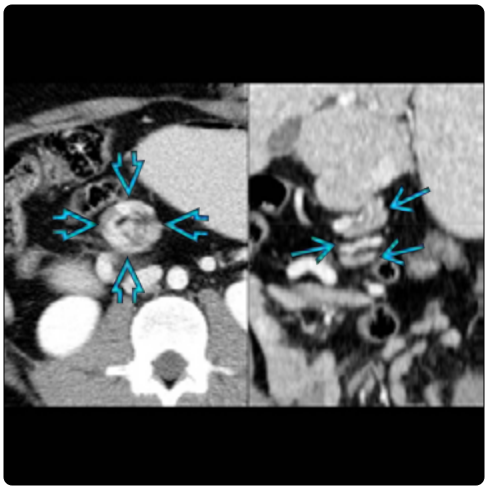


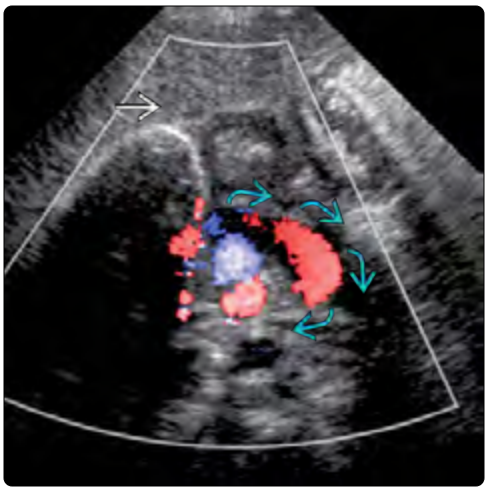


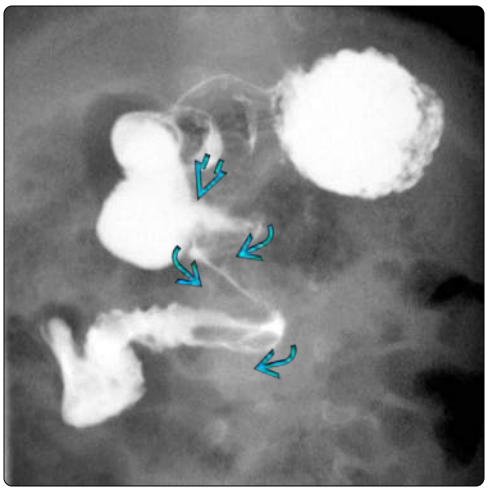
(Left) *Newborn AP radiograph on day 1 of life with bilious emesis shows multiple dilated air-filled bowel loops suggestive of distal obstruction. Prenatal imaging suggested a possible malrotation, raising concern that this bowel dilation could be due to an ileus of small bowel ischemia.* **(Right)** *Immediate postnatal upper GI in the same patient shows D3 beaking & a twist of MV. In the OR, there was diffuse small bowel necrosis, not unexpected in this rare case of in utero MV.*





(Left) *Intraoperative photograph in a child with malrotation & volvulus shows ischemic loops of dilated bowel. Note the twisted, volvulized bowel.* **(Right)** *Axial CECT of a 14-year-old boy with intermittent abdominal pain shows the swirl sign just caudal to the root of the superior mesenteric artery with spiraling vessels, mesentery, & bowel on the coronal reconstruction, consistent with MV.*





(Left) *Transverse US in this 24-day-old boy with recurrent vomiting shows swirling vessels at the mesenteric root, consistent with MV. This whirlpool sign showed complete encirclement of the SMA in a clockwise fashion. Note the dilated proximal duodenum. The surgeons requested an upper GI for confirmation.* **(Right)** *Frontal upper GI in the same patient shows a dilated duodenum to D2-D3 with a corkscrew or spiral sign of MV.*

Duodenal Atresia or Stenosis

KEY FACTS

TERMINOLOGY

- Most common upper intestinal obstruction in neonate
- Atresia: Congenital occlusion of intestinal lumen
- Stenosis: Fixed narrowing of intestinal lumen

IMAGING

- Newborn radiographic double bubble sign essentially diagnostic
  - Markedly dilated duodenum implies chronic in utero obstruction (with duodenal atresia as most common cause), especially with no distal gas
  - May not be seen on initial radiographs if stomach or duodenum decompressed by nasogastric tube or vomiting
- If duodenum mildly to moderately dilated with some distal gas → emergent UGI to exclude midgut volvulus, evaluate duodenal rotation

TOP DIFFERENTIAL DIAGNOSES

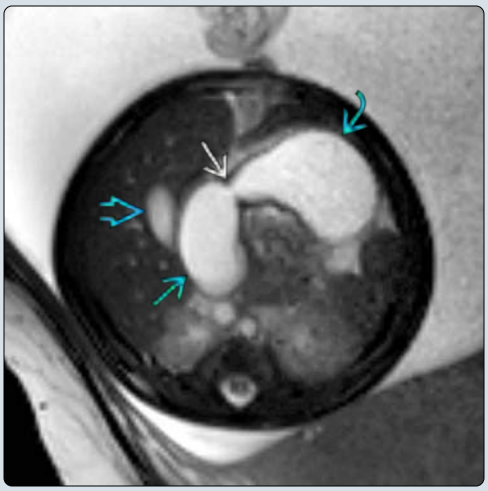
- Midgut volvulus
- Duodenal web
- Jejunal atresia
- Gastrointestinal duplication cyst
- Annular pancreas
- Hypertrophic pyloric stenosis

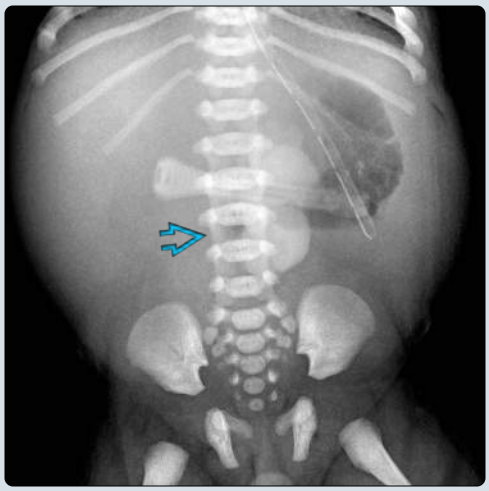
PATHOLOGY

- Associated anomalies in > 50% of patients
  - 30-46% have Down syndrome (trisomy 21)

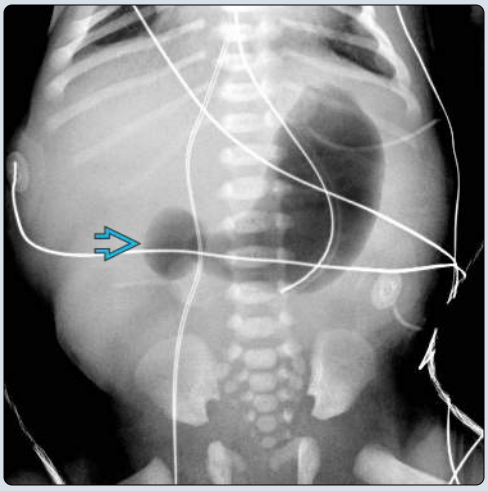
CLINICAL ISSUES

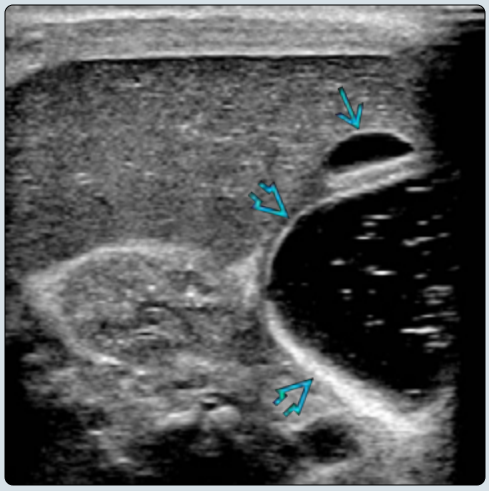
- Diagnosis often made prenatally by ultrasound
- With surgical treatment, survival rate > 90%
- Duodenoduodenostomy most common operation
- Postop complications: Megaduodenum, motility issues, adhesions





(Left) *Axial SSFSE T2 MR in a 36-week gestation fetus shows a double bubble sign of duodenal atresia with dilation of the stomach, pylorus, & duodenal bulb. The gallbladder (GB) is noted for reference. The bowel distal to D2 is decompressed.* **(Right)** *AP radiograph immediately after delivery shows a nasogastric tube within the stomach, which can obscure the classic double bubble sign if suction is applied, as seen here. There is only minimal air in the duodenal bulb. Note the lack distal bowel gas.*





(Left) *AP radiograph performed in the same patient on day 2 of life (after turning off the nasogastric suction) shows that air is starting to collect in the duodenal bulb with no distal gas, revealing a more classic double bubble appearance of duodenal atresia.* **(Right)** *Transverse US of the abdomen in the same patient shows a portion of the dilated, fluid-filled duodenum as well as a portion of the GB. Duodenal atresia was confirmed at surgery.*

TERMINOLOGY

Abbreviations

- Duodenal atresia (DA), duodenal stenosis (DS)

Definitions

- Atresia: Congenital occlusion of intestinal lumen
- Stenosis: Fixed narrowing of intestinal lumen
- Most common neonatal upper intestinal obstruction (40-50%)

IMAGING

General Features

- Best diagnostic clue
  - Double bubble sign
- Location
  - Usually 2nd duodenum (near ampulla of Vater)

Radiographic Findings

- Radiography
  - Double bubble: Classic
    - Gas-distended, dilated stomach + proximal duodenum without distal bowel gas
      - In this context, markedly dilated duodenum implies chronic in utero obstruction (DA as most common cause)
    - If dilated stomach + duodenum in newborn with distal gas, think of
      - Acute midgut volvulus (new onset since birth)
      - DA spectrum (stenosis, web, ± annular pancreas, etc.)
      - Variant biliary/pancreatic anatomy (collateral pathway around atretic segment)
    - If stomach suctioned by nasogastric tube (NGT) or vomiting, double bubble may not be seen on initial radiographs
  - Free air very uncommon
    - Rare gastric perforation; may be related to NGT placement

Fluoroscopic Findings

- Upper GI
  - Not usually performed in DA (as radiographs are typically diagnostic with no distal gas)
    - Can inject air through NGT to look for distal passage of air
  - Needed urgently with dilated duodenum in setting of distal bowel gas (particularly with less bulbous duodenal dilation)
    - Must exclude midgut volvulus
      - Can only truly be done by visualizing normal duodenal jejunal junction (DJJ)
    - DS: Focal or longer segment of fixed circumferential narrowing
    - DA with biliary/pancreatic duct variations: Biliary drainage above + below atresia → distal gas in bowel

MR Findings

- Fetal MR: Dilated, fluid-filled stomach + proximal duodenum

Ultrasonographic Findings

- Grayscale ultrasound
  - Dilated fluid- & gas-filled stomach & duodenum
  - Prenatal sonography
    - Fluid double bubble; polyhydramnios in 30-40%

Imaging Recommendations

- Best imaging tool
  - Radiographs for double bubble
  - Upper GI if distal gas present
- Protocol advice
  - If upper GI needed for diagnosis, place nasogastric tube
    - Aspirate stomach, then inject small amounts of barium in right lateral decubitus position
    - Small puffs of air to advance barium
  - Postoperative upper GI: Low-osmotic/isosmotic nonionic water-soluble contrast
    - Is there anastomotic leak, obstruction, additional web, malrotation?

DIFFERENTIAL DIAGNOSIS

Midgut Volvulus

- Proximal duodenum less dilated in acute volvulus than DA
  - Chronic in utero volvulus (rare) may cause greater duodenal dilation (than acute midgut volvulus), absent distal gas
    - ± US as useful adjunct exam
- Near-complete obstruction at D2-D3 of duodenum
- "Corkscrew" configuration of narrowed bowel beyond obstruction + malpositioned DJJ
- Ladd bands may also cause obstruction in malrotation

Duodenal Web

- Delayed clinical presentation; distal gas present
- ± windsock appearance of web in duodenum

Jejunal Atresia

- Dilated stomach + duodenum + several jejunal loops without distal gas
  - Triple or quadruple bubble classically

Gastrointestinal Duplication Cysts

- Extrinsic mass effect on bowel on upper GI
- Sonographic gut signature to cyst wall
  - With dilated stomach, may mimic double bubble on prenatal US

Annular Pancreas

- Circumferential narrowing of mid 2nd duodenum
- Distal gas present
- Usually associated DS at level of encircling pancreas

Hypertrophic Pyloric Stenosis

- Presents at 2-12 weeks of life (not 1st week)
- Projectile, nonbilious vomiting
- US: Thick, elongated, persistently closed pyloric muscle

Less Common Newborn Upper GI Obstructions

- Preduodenal portal vein, internal hernia, pyloric atresia

PATHOLOGY

General Features

- Etiology
  - Most accepted theory: Embryologic failure of duodenal recanalization in 12th gestational week
  - Unlike other intestinal atresias (due to vascular accidents)
- Genetics
  - Typically sporadic if isolated anomaly
  - Down syndrome (trisomy 21) in 30-46%
  - Feingold syndrome (autosomal dominant)
    - Hand/foot anomalies, microcephaly, tracheoesophageal fistula, esophageal/DA, short palpebral fissures, developmental delay
- Associated abnormalities
  - > 50% of patients with DA have other anomalies
  - 30-46% have Down syndrome (trisomy 21)
    - 11 pairs of ribs, macroglossia, flat acetabular angles, cardiomegaly with shunt vascularity
  - Malrotation: 28%
  - Annular pancreas: Up to 30%
  - 2nd duodenal web: 1-3%
  - Choledochal cyst, other biliary anomalies
  - Preduodenal portal vein
  - Esophageal atresia + tracheoesophageal fistula: Up to 18%
  - Imperforate anus
  - Cardiac defects: 20-30% (less frequent without trisomy 21 but more likely cyanotic)
  - Situs anomalies
  - Renal anomalies
  - VACTERL association

Staging, Grading, & Classification

- Type I DA (69-74%)
  - Intact intestinal wall & mesentery
  - Membranous luminal obstruction
- Type II DA (1-2%)
  - 2 blind ends separated by fibrous cord
- Type III DA (5-6%)
  - 2 blind ends without intervening cord
  - Biliary anomalies
- DS (18-23%)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Diagnosis often made prenatally by US
  - Bilious > > nonbilious (80:20) vomiting
    - Determined by site of atresia relative to ampulla of Vater
- Other signs/symptoms
  - Dehydration, weight loss, electrolyte imbalance

Demographics

- Age
  - Newborn (21-45% premature)
- Epidemiology
  - Incidence 1:5,000-10,000 live births

Natural History & Prognosis

- Untreated: Dehydration, severe electrolyte abnormalities, death
- With surgical treatment, survival rate > 90%
  - Mortality largely related to associated anomalies

Treatment

- Surgical repair
  - If radiographs diagnostic of DA (classic double bubble), surgical repair urgent but not emergent
  - In setting of distal gas, failure to demonstrate normal DJJ by upper GI requires emergent exploration (as midgut volvulus not excluded)
- Duodenoduodenostomy most common operation
  - Bypasses obstruction to preserve ampulla of Vater
  - Diamond-shaped vs. side-to-side anastomosis
  - ± tapering enteroplasty of dilated duodenum
  - Rubber catheter passed distally in duodenum, withdrawn with balloon inflated to exclude additional distal web (1-3%)
- Surgical repair may be delayed to 1st address
  - Electrolyte or fluid balance disturbances
  - Severe cardiac defects
  - Severe respiratory insufficiency
- Long-term complications
  - Megaduodenum, motility issues, adhesions

DIAGNOSTIC CHECKLIST

Consider

- How dilated is duodenum, could it be diagnosis other than DA; is there distal bowel gas?
- Are there other anomalies?

Image Interpretation Pearls

- True double bubble with no distal bowel gas essentially diagnostic for DA
- Upper GI needed if distal bowel gas present

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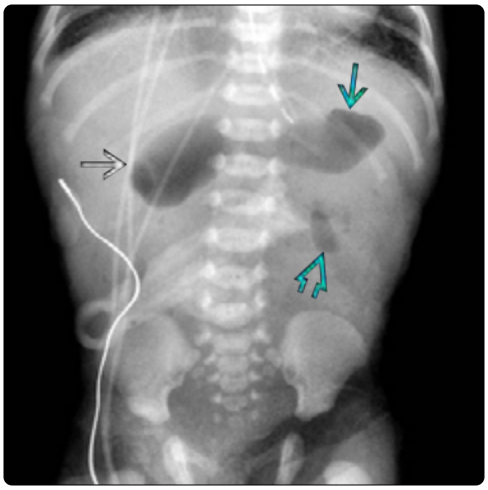
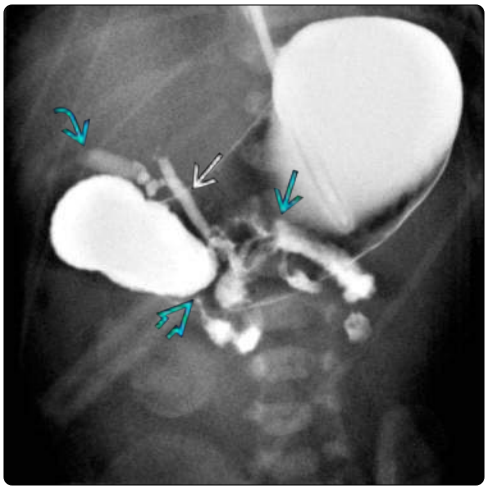
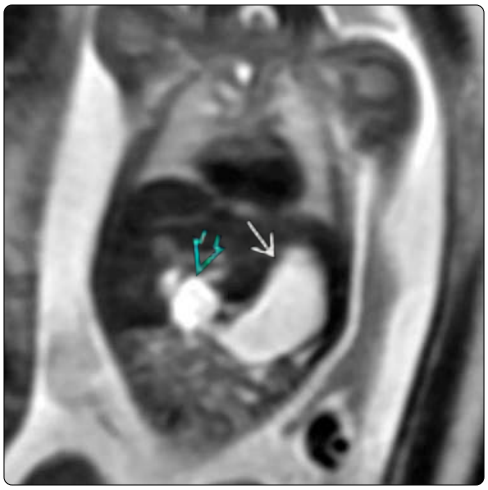
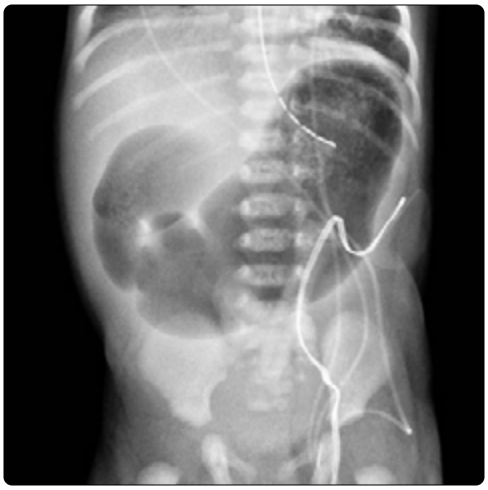


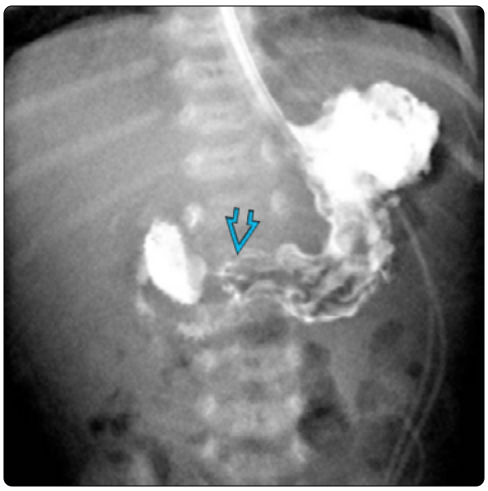


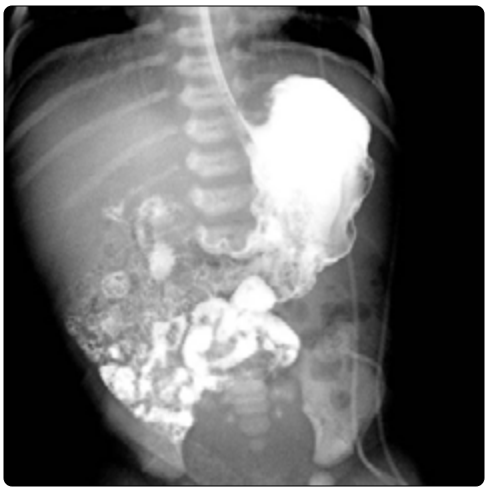
(Left) *AP radiograph in a full-term infant boy at 1 day old shows a dilated stomach & duodenum with a small amount of distal gas. The main surgical diagnostic considerations include midgut volvulus & duodenal stenosis or web. Immediate upper GI is indicated.* **(Right)** *Upper GI image in the same patient shows dilation of the duodenum to D2 with contrast passing through a stenotic orifice & the remaining duodenum to a normal duodenal jejunal junction. Note reflux into the common bile duct & GB.*





(Left) *Coronal T2 SSFSE MR in a 24-week-gestation fetus shows dilated stomach & duodenum with decompressed distal bowel, the classic double bubble of the duodenal atresia spectrum.* **(Right)** *AP radiograph of the same patient upon delivery shows a classic double bubble sign of duodenal atresia with no distal bowel gas. Duodenal atresia was confirmed surgically.*





(Left) *Supine frontal upper GI was performed in the same patient following operative repair of the duodenal atresia to exclude a leak. There was a nonretroperitoneal position of the duodenum (not shown) with the duodenojejunal junction in the midline, consistent with malrotation (which is associated with duodenal atresia).* **(Right)** *Frontal upper GI (converted to an SBFT) in the same patient shows most of small bowel on the right, confirming malrotation.*

Duodenal Web

KEY FACTS

TERMINOLOGY

- Incomplete diaphragm of duodenal lumen causing partial or intermittent complete duodenal obstruction
- Duodenal atresia spectrum, later clinical presentation

IMAGING

- 2nd to 4th portion duodenum
  - Usually adjacent to ampulla of Vater
- Aperture size determines degree of obstruction, age of presentation, imaging appearance
- Early presentation: Dilated stomach & proximal duodenum to D2/D3
- Late presentation: Thin, ballooned windsock in distal duodenum; variable duodenal caliber (depends on orifice size)

TOP DIFFERENTIAL DIAGNOSES

- Duodenal atresia
- Midgut volvulus
- Gastrointestinal duplication cysts
- Annular pancreas
- Superior mesenteric artery syndrome

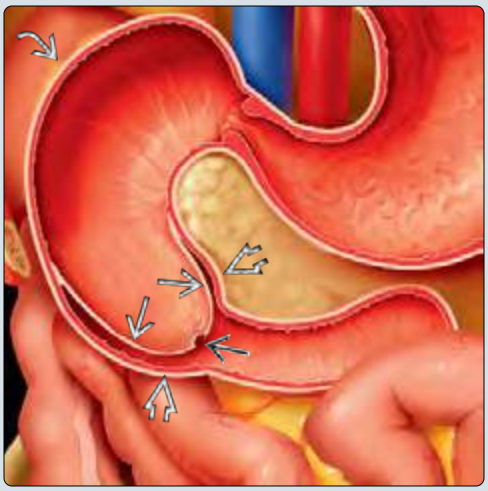
PATHOLOGY

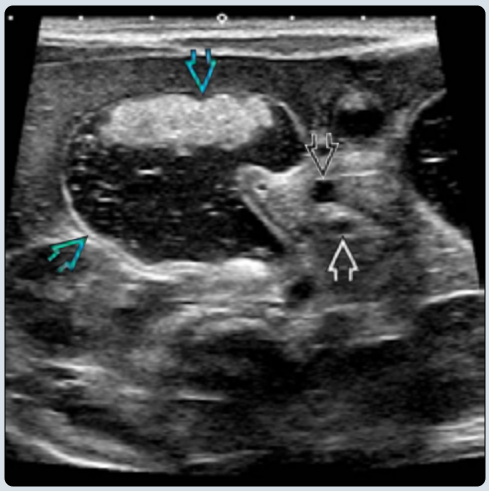
- Failed duodenal recanalization: Spectrum of duodenal atresia
- Associated anomalies
  - Down syndrome: 30%
  - Malrotation: 28%
  - Annular pancreas: 33%

CLINICAL ISSUES

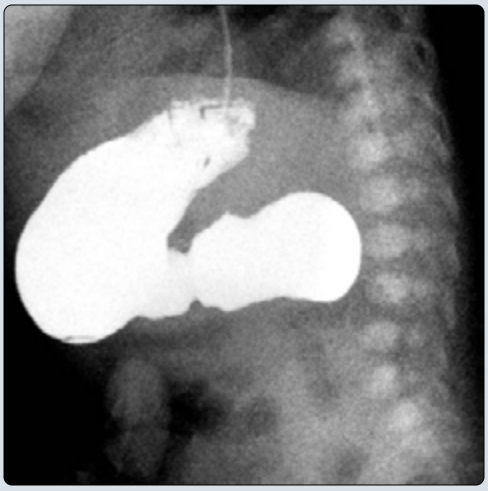
- Early presenters: Feeding intolerance, vomiting (bilious > nonbilious)
- Late presenters: Nausea, abdominal pain, progressive vomiting, acute pancreatitis
- Prognosis excellent with treatment
  - Surgical vs. endoscopic excision

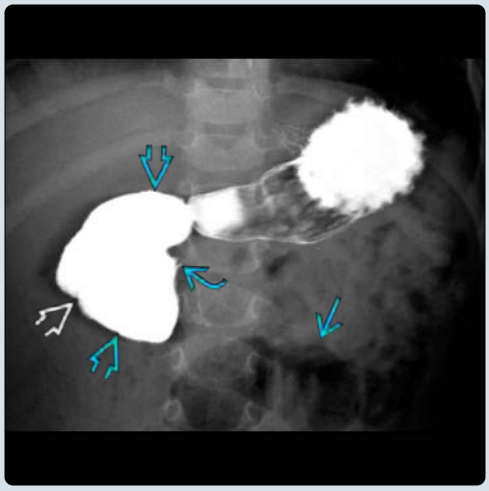
(Left) *Graphic shows a web with a windsock shape within the duodenal lumen with a pinhole opening distally. The proximal duodenum is moderately dilated from this partial obstruction.* **(Right)** *Transverse US performed for pyloric stenosis in a 3-week-old boy with recurrent emesis showed a normal pylorus. However, there is marked dilation of the duodenum to D2-D3 with no evidence of the swirl sign & a normal superior mesenteric artery/superior mesenteric vein relationship.*





(Left) *Lateral upper GI in the same patient immediately after the US shows no barium passing from the dilated proximal duodenum, but there is distal gas. The patient was brought straight to the operating room where a tight duodenal web was found.* **(Right)** *Supine upper GI shows a dilated duodenum with distal gas & a tiny orifice, as well as the dimple sign at the attachment of the proximal aspect of the web, which puckers when the web is stretched distally.*





TERMINOLOGY

Definitions

- Incomplete diaphragm of duodenal lumen causing high-grade duodenal obstruction
- Duodenal atresia (DA) spectrum, later clinical presentation

IMAGING

General Features

- Location
  - 2nd to 4th portion of duodenum
- Morphology
  - Aperture size determines degree of obstruction, age of presentation, imaging appearance

Radiographic Findings

- Radiography
  - Commonly mild, intermittent duodenal dilation with partial obstruction
  - Neonatal double bubble sign with distal gas (less common)

Fluoroscopic Findings

- Upper GI
  - Early presentation: Dilated stomach + proximal duodenum to D2; stenotic orifice
  - Late presentation: Thin, ballooned windsock distal duodenum; variable duodenal caliber (depends on orifice size)
  - Duodenal dimple
    - Real-time dimpling of duodenal wall contour at attachment of web to duodenal wall as nasoduodenal tube tip stretches contrast-filled web

Ultrasonographic Findings

- Grayscale ultrasound
  - Proximal duodenal dilation; fluid may outline thin web if fluid present distal to web

Imaging Recommendations

- Best imaging tool
  - Upper GI with barium
- Protocol advice
  - Webs rarely present in 1st days of life
    - True neonatal double bubble, no distal gas: Usually DA (no further imaging required)
    - Mild to moderate proximal duodenal dilation + distal gas → upper GI to exclude midgut volvulus

DIFFERENTIAL DIAGNOSIS

Duodenal Atresia

- Most common high neonatal bowel obstruction
- Classic, true double bubble, absent distal bowel

Midgut Volvulus

- Duodenum abnormally dilated in acute volvulus < DA
  - Chronic in utero volvulus (rare) may cause more duodenal dilation, absent distal gas
    - Ultrasound may be useful adjunct exam
- Near-complete obstruction at D2-D3 of duodenum
- Upper GI: Dilated duodenum up to twist at D2-D3
  - ± narrowed corkscrew beyond obstruction
  - If contrast obstructed at D2-D3, cannot differentiate from web
- Ladd bands from malrotation may also obstruct D2-D3

Gastrointestinal Duplication Cysts

- Round extrinsic mass (filling defect) on upper GI
- Sonographic cyst with gut signature

Annular Pancreas

- Circumferential narrowing of mid 2nd duodenum

Superior Mesenteric Artery Syndrome

- Older child/adult with compression of D3 between superior mesenteric artery & aorta
  - Rotating patient left lateral may allow contrast passage
- Predisposed by weight loss, scoliosis surgery (↓ mesenteric fat)

PATHOLOGY

General Features

- Etiology
  - Failed duodenal recanalization: Spectrum of DA
- Associated abnormalities
  - Down syndrome: 30%
  - Malrotation: 28%
  - Annular pancreas: 33%
  - Other intestinal atresias, biliary anomalies, pyloric stenosis, preduodenal portal vein; cardiac anomalies

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Early presenters: Late in 1st week of life & early infancy
    - Feeding intolerance
    - Emesis: 80% bilious, 20% nonbilious depends on (location of web relative to ampulla of Vater)
  - Late presenters: Childhood to adulthood
    - Nausea, abdominal pain, progressive emesis, acute pancreatitis

Demographics

- Epidemiology
  - Incidence 1:7,500-40,000

Treatment

- Prognosis excellent with treatment
  - Surgical or endoscopic resection of web
  - Duodenoduodenostomy if obstruction complete

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4. Dwek JR et al: The duodenal dimple: a specific fluoroscopic correlate to the duodenal web. Pediatr Radiol. 29(6):467-8, 1999

Pyloric Atresia

KEY FACTS

TERMINOLOGY

- Uncommon congenital gastric outlet obstruction
- Isolated vs. associated anomalies in 30-50%
  - Epidermolysis bullosa-pyloric atresia (EB-PA)
  - Hereditary multiple intestinal atresia or multiple intestinal atresia with immunodeficiency (HMIA/MIAI)

IMAGING

- Radiographs show "single bubble": Dilated gas-filled stomach in newborn with no bowel gas otherwise
  - ± skin erosions (EB-PA)
  - ± intestinal Ca^{2+} (HMIA/MIAI)
- Ultrasound may show abnormal, thin pyloric morphology + dilated gastric antrum = tennis racket appearance
  - Failure to visualize normal pyloric opening & antegrade passage of gastric contents into duodenal bulb

TOP DIFFERENTIAL DIAGNOSES

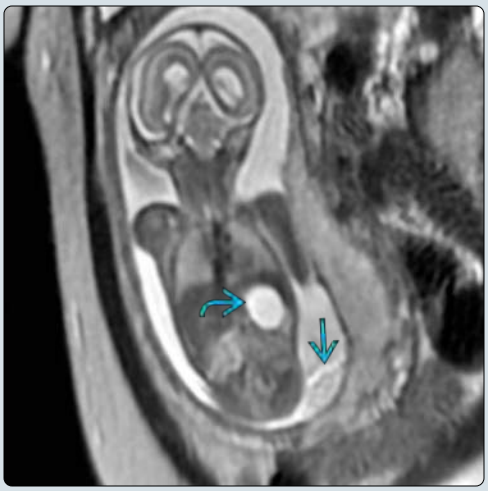
- Duodenal atresia
- Jejunal atresia
- Hypertrophic pyloric stenosis

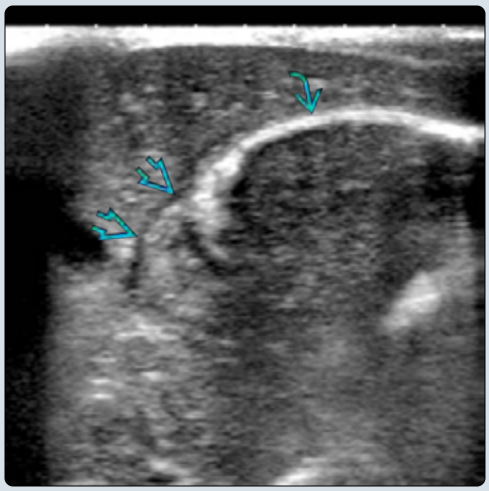
PATHOLOGY

- Atresia may be web, solid cord, or discontinuous blind ends
- In MIAI: Atresias extend from pylorus to rectum (not just superior mesenteric artery territory)
  - Severe immunodeficiency results in recurrent infections
- In EB-PA, clinical findings may include: Aplasia cutis; fusion/erosion/scarring with malformed protuberances (such as ears), small diameter orifices (such as nostrils), contractures

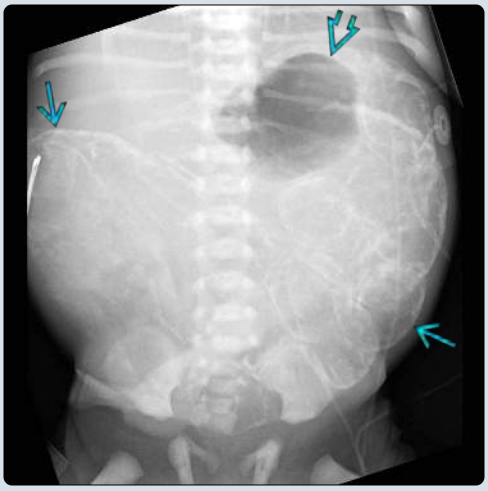
CLINICAL ISSUES

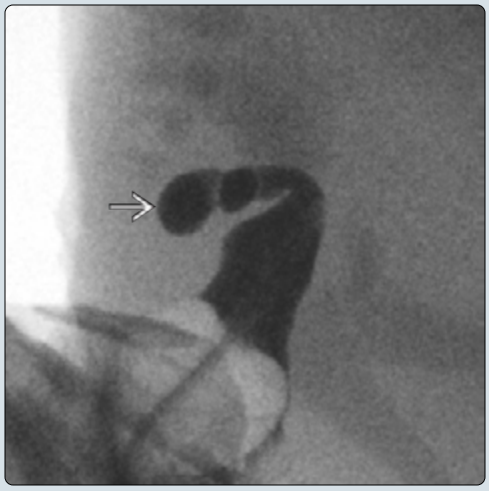
- Typical presentation for isolated PA: Nonbilious emesis
- PA has excellent prognosis in isolation; resection curative
- Mortality approaches 100% in EB-PA or HMIA/MIAI
  - Infections, malnutrition in both





(Left) *Coronal T2 SSFSE MR at 21 weeks gestation shows dilation of the stomach (persisting throughout the exam) with no dilation of the bowel. Particulate debris was seen lying dependently in the amniotic cavity on multiple sequences, raising concern for the epidermolysis bullosa-pyloric atresia (EB-PA) association.* **(Right)** *Transverse oblique ultrasound through the newborn's right upper quadrant shows gastric distention with an abnormally thin, elongated, & persistently closed pylorus. Skin biopsy confirmed EB-PA.*





(Left) *AP abdominal radiograph in a newborn shows extensive Ca^{2+} throughout dilated bowel loops (which were fluid-filled on prenatal ultrasound). The gas-filled stomach is moderately distended with no distal bowel gas identified.* **(Right)** *Lateral view from a water-soluble contrast enema in the same patient shows a small, blind-ending rectal pouch. Surgery confirmed numerous atresias from the pylorus to the rectum, consistent with MIAI/HMIA in this patient found to have immunodeficiency.*

TERMINOLOGY

Abbreviations

- Pyloric atresia (PA)
- Epidermolysis bullosa-pyloric atresia (EB-PA)
- Hereditary multiple intestinal atresia or multiple intestinal atresia with immunodeficiency (HMIA/MIAI)

Definitions

- Uncommon congenital gastric outlet obstruction

IMAGING

Radiographic Findings

- "Single bubble": Dilated gas-filled stomach in newborn with no bowel gas otherwise
  - ± skin erosions (EB-PA)
  - ± intestinal Ca^{2+} (HMIA/MIAI)

Ultrasonographic Findings

- Thinned, abnormal pyloric configuration + dilated gastric antrum = tennis racket appearance
- Failure to visualize normal pyloric opening & antegrade passage of gastric contents into duodenal bulb

DIFFERENTIAL DIAGNOSIS

Duodenal Atresia

- Classic "double bubble": Moderately to markedly dilated, bulbous, gas distended stomach & proximal duodenum
- No distal bowel gas

Jejunal Atresia

- "Triple or quadruple bubble": Dilated stomach & duodenum plus 1-2 dilated proximal jejunal loops
  - Bowel segment just proximal to obstruction typically most bulbous/dilated
- No distal bowel gas

Hypertrophic Pyloric Stenosis

- Not congenital; typically occurs at 2-12 weeks of age
- ± dilated stomach with vigorous peristalsis ("caterpillar"), decreased (but not absent) distal bowel gas
- Thickened pyloric muscle, elongated pyloric channel
  - Fails to open during ultrasound or upper GI

PATHOLOGY

General Features

- Associated abnormalities
  - 30-50% have additional anomalies, most commonly
    - EB-PA
      - Subtypes of EB include junctional, simplex, & dystrophic forms, depending on location of epidermal-dermal separation
      - EB-PA found with mutations of genes encoding plectin (*PLEC*) or α6-β4 integrin (*ITGA6* & *ITGB4*), both of which help maintain skin integrity
      - Due to skin fragility, clinical findings may include: Aplasia cutis; fusion/erosion/scarring with malformed protuberances (such as ears), small diameter orifices (such as nostrils), contractures
    - MIAI/HMIA
      - Atresias extending from pylorus to rectum (not just superior mesenteric artery territory)
      - Frequently have long occluded segments with characteristic sieve-like appearance at histology (due to multiple lumina on single cross section)
      - Severe immunodeficiency results in multiple infections

Staging, Grading, & Classification

- Pyloric atresia types
  - Type A (most common): Obstructing membrane
  - Type B: Solid cord
  - Type C (least common): Separated blind ends

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - With isolated PA: Nonbilious emesis
    - May be mistaken for gastroesophageal reflux

Demographics

- Epidemiology
  - 1% of gastrointestinal atresias

Natural History & Prognosis

- PA has excellent prognosis in isolation
- Mortality approaches 100% in EB-PA or MIAI/HMIA

Treatment

- Isolated PA: Surgery depends on type of obstruction (ranges from web excision to gastroduodenostomy)
- EB-PA
  - Prevent blistering & superimposed infections; ensure adequate nutrition
  - Monitor for numerous long-term EB complications
- MIAI/HMIA
  - Poor gut function despite resection of atresias
  - Antimicrobials for recurrent severe infections due to immunodeficiency
  - Small bowel transplant ± stem cell transplant may be curative

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- With pre-/postnatal imaging findings suggesting congenital gastric outlet obstruction, look for clues of anomalies associated with PA

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5. Tomá P et al: Pyloric atresia: report of two cases (one associated with epidermolysis bullosa and one associated with multiple intestinal atresias). Pediatr Radiol. 32(8):552-5, 2002

Jejunoileal Atresia

KEY FACTS

TERMINOLOGY

- Congenital occlusion of jejunal or ileal lumen
  - Ranges from focal membrane to long-segment intestinal + mesenteric gap

IMAGING

- Site of obstruction (atresia) determines radiographic, fluoroscopic patterns
- Proximal jejunal atresia
  - Dilated stomach + duodenum + 1-2 loops of jejunum; microcolon less likely on enema
- Midjejunal to distal ileal atresia
  - Numerous dilated loops; enema shows microcolon
- Protocol advice
  - Water-soluble contrast enema (low osmolality, nearly iso-osmotic to body fluids)
    - Avoids fluid shifts into or out of bowel
    - Barium not used (may impede meconium passage)
  - If microcolon, reflux into small bowel up to dilated loops (if possible)

TOP DIFFERENTIAL DIAGNOSES

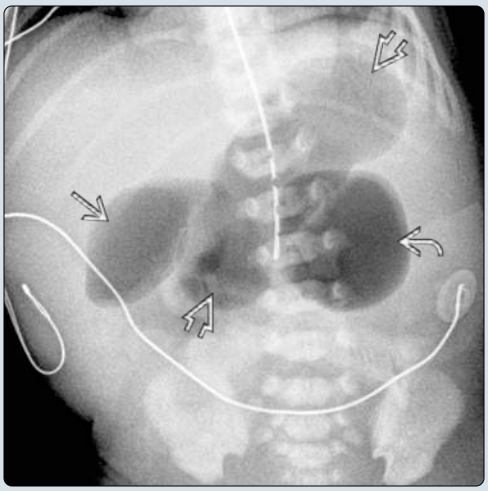
- Meconium ileus
- Meconium plug syndrome/small left colon
- Hirschsprung disease
- Anorectal malformation
- Inguinal hernia
- Necrotizing enterocolitis

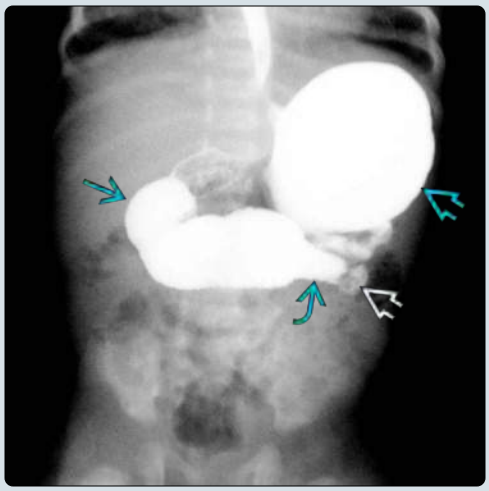
PATHOLOGY

- Associated anomalies in 10-52% of cases
  - Gastroschisis (up to 20%), meconium ileus/cystic fibrosis (up to 10%), malrotation, volvulus

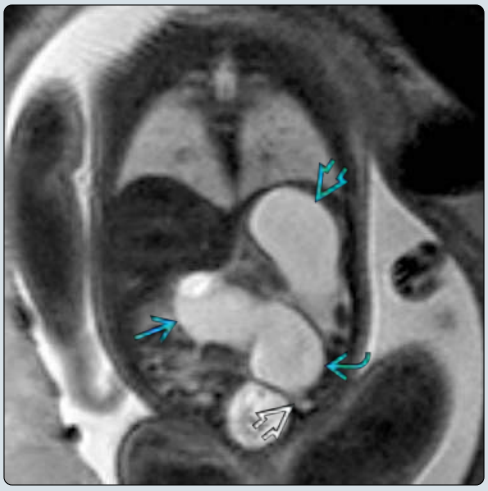
CLINICAL ISSUES

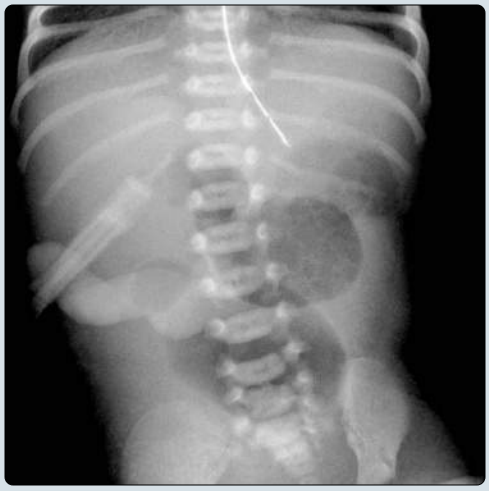
- Prognosis dependent on amount of residual functional bowel post repair + associated anomalies
- Mortality: 11-14%





(Left) *AP radiograph shows the classic triple bubble sign of jejunal atresia due to a dilated stomach, duodenum, & proximal jejunum with no distal bowel gas.* **(Right)** *Supine UGI in a 13-day-old girl with several days of projectile yellow emesis shows dilation of the stomach, duodenum, & initial jejunal segment with transition to a normal caliber jejunum just beyond the duodenojejunal junction, most consistent with jejunal stenosis (which was found at surgery).*





(Left) *Coronal T2 SSFSE fetal MR at 30-weeks gestation shows the triple bubble sign of jejunal atresia with a dilated, fluid-filled stomach, duodenum, & proximal jejunum. Note the abrupt blind end of the atresia.* **(Right)** *Radiograph in a 1-day-old newborn shows 3 or 4 air- & fluid-filled dilated bowel loops without other distal gas. Bilious material was aspirated from the Replogle tube. These findings are consistent with a proximal jejunal atresia, in agreement with fetal MR.*

TERMINOLOGY

Definitions

- Congenital occlusion of jejunal or ileal lumen
- Ranges from focal membrane to long-segment intestinal atresia + mesenteric gap
- Stenosis, incomplete web: Forme fruste of atresia

IMAGING

General Features

- Best diagnostic clue
  - Contrast enema showing microcolon, with reflux of contrast into blind-ending ileum or jejunum
- Location
  - Jejunum to distal ileum
- Morphology
  - Site of obstruction (atresia) determines radiographic, fluoroscopic patterns
    - Proximal jejunal atresia (classic high/proximal neonatal intestinal obstruction)
      - Dilated stomach + duodenum + 1-2 loops of jejunum; microcolon less likely on enema
    - Midjejunal to distal ileal atresia (classic low/distal neonatal intestinal obstruction)
      - Numerous dilated bowel loops; enema shows microcolon
    - Multiple or long-segment intestinal atresias may have mixed imaging features
  - Appearance of microcolon on contrast enema related to timing & level of obstruction
    - Smaller caliber colon with earlier or distal obstructions
      - Unused colon receiving little succus entericus
    - Near-normal to normal caliber colon with later or more proximal obstructions
      - Colonic succus entericus accumulation less impaired

Radiographic Findings

- Radiography
  - Jejunal atresia: Proximal
    - Triple bubble sign
      - Dilated stomach + duodenum + 1 (or a few) proximal jejunal loop(s) without distal bowel gas
  - Ileal atresia
    - Multiple dilated bowel loops
      - Difficult to tell small bowel from colon in neonate
  - Soft tissue mass, ascites, peritoneal Ca^{2+} suggest complicated obstruction
    - Perforation ± pseudocyst
    - Free intraperitoneal air uncommon
  - Intraluminal Ca^{2+} in multiple atresias
  - Stenosis or web may have distal bowel gas beyond dilated loops

Fluoroscopic Findings

- Upper GI
  - Jejunal atresia
    - Dilated stomach, duodenum, proximal jejunum
    - Duodenal-jejunal junction may be displaced by dilated bowel loops in neonate (due to ↑ laxity of ligament of Treitz) despite normal rotation
  - Ileal atresia: Normal (in absence of multiple atresias)
- Water-soluble contrast enema (WSCE)
  - Jejunal atresia
    - Proximal atresia → normal or mildly small
    - Distal atresia → variable degrees of small colon
    - Long segment or multiple atresias → microcolon
  - Ileal atresia
    - Microcolon
    - Abrupt blind end to contrast flow at atresia
      - Contrast fails to reach dilated proximal bowel
    - Ileum distal to atresia also small in caliber

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal or postnatal
    - Dilated, fluid-filled bowel loops
      - Echogenic dilated loops prenatally may favor cystic fibrosis or gastroschisis ± atresia
    - Peritoneal Ca^{2+}, ascites (complex > simple), pseudocyst → in utero perforation (meconium peritonitis)
    - Intraluminal Ca^{2+} possible in multiple intestinal atresias
      - Must also consider anorectal malformation with rectourinary fistula or total colonic Hirschsprung disease

MR Findings

- Depending on gestational age, fetal MR better than prenatal US for detecting level of obstruction
  - Distribution of T1 bright meconium can evaluate length, caliber, & position of colon relative to abnormal small bowel loops

Imaging Recommendations

- Best imaging tool
  - For clinical + radiographic distal/low obstruction: WSCE
  - For clinical + radiographic proximal/high obstruction: Upper GI series (UGI)
    - For true triple or quadruple bubble with marked dilation & no distal gas, fluoroscopy may not be required prior to surgery
    - ± enema to exclude distal atresia preoperatively
  - If answer to obstruction site not provided by 1st of these exams, be ready to perform 2nd
- Protocol advice
  - Enema performed with low-osmolality water-soluble contrast (nearly iso-osmotic to body fluids)
    - Avoids fluid shifts into or out of bowel
    - Barium not used (may impede meconium passage)
  - If microcolon, reflux contrast into small bowel (SB) until contrast reaches dilated loops or atresia

DIFFERENTIAL DIAGNOSIS

Meconium Ileus

- Microcolon on WSCE
- Meconium pellets obstructing distal ileum on enema

- Refulxed contrast outlines ileal filling defects before passing into dilated loops

Small Left Colon/Meconium Plug Syndrome

- Small-caliber left colon up to splenic flexure on WSCE
- Normal to dilated proximal colon
- ± meconium plugs in small left colon

Hirschsprung Disease

- Rectosigmoid ratio < 1 with transition zone on WSCE
- Most of proximal colon dilated
  - Except small colon in total colonic Hirschsprung disease

Anorectal Malformation

- No normal anal opening

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome

- Rare, often fatal disease with marked bladder dilation
- More common in female patients (4:1)

Colonic Atresia

- 1 or 2 loops dilated out of proportion to rest on radiograph
- Small distal colon with proximal blind ending on WSCE

Inguinal Hernia

- Dilated bowel with asymmetric inguinoscrotal fold
  - Gas in scrotum diagnostic

Necrotizing Enterocolitis

- Premature infants
- Uncommon in first 1-2 days of life
- Bowel separation, unchanging bowel loops, pneumatosis, portal venous gas, pneumoperitoneum; no enema

PATHOLOGY

General Features

- Etiology
  - Intrauterine vascular accident → necrosis with segmental stenosis/resorption
- Genetics
  - French Canadian ancestry in hereditary multiple intestinal atresias with immunodeficiency
    - Specific types of atresia (often pyloric & sieve-like colon)
  - Autosomal recessive inheritance in some type 3b
- Associated abnormalities
  - 10-52% of jejunoileal atresia cases
    - Predisposing gastrointestinal anomalies
      - Gastroschisis (up to 20%), meconium ileus/cystic fibrosis (up to 10%), malrotation, volvulus
    - Various cardiac, genitourinary, brain anomalies

Staging, Grading, & Classification

- Atresia: Surgical grading system
  - Type 1: Membranous atresia, web/stenosis
    - No mesenteric defect, bowel not short
  - Type 2: Blind ends separated by fibrous cord
    - No mesenteric defect, bowel not short
  - Type 3a: Blind ends with complete disconnection
    - V-shaped mesenteric gap, bowel short
  - Type 3b: Apple peel or Christmas tree deformity
    - Large mesenteric defect, bowel short
  - Type 4: Multiple small bowel atresias
    - 6-32% of jejunoileal atresias

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Distal ileal atresia: Failure to pass meconium + abdominal distention, bilious emesis
  - Jejunal or proximal ileal atresia: Bilious emesis
  - Stenosis or web: Early vs. delayed presentation with intermittent emesis, failure to thrive

Demographics

- Age
  - Atresia presents in utero to first 1-2 days of life
- Epidemiology
  - 1:3,000-5,000 live births

Natural History & Prognosis

- Prognosis dependent on amount of residual functional bowel post repair + associated anomalies
  - 40 cm of normal bowel length considered functionally adequate to avoid treatment for short gut syndrome
- Mortality: 11-14%

Treatment

- Full resuscitation prior to surgical correction (unless perforation or volvulus)
- Surgical resection of atretic segment with anastomosis
  - Tapering vs. resection of very dilated proximal segment, depending on residual bowel length
- Complications: Short gut syndrome (14%), dysmotility, adhesions

DIAGNOSTIC CHECKLIST

Consider

- Bilious emesis can occur in neonatal distal or proximal obstructions
  - Radiograph determines initial fluoroscopic study in neonatal obstruction
    - WSCE 1st study in distal, UGI 1st study in proximal

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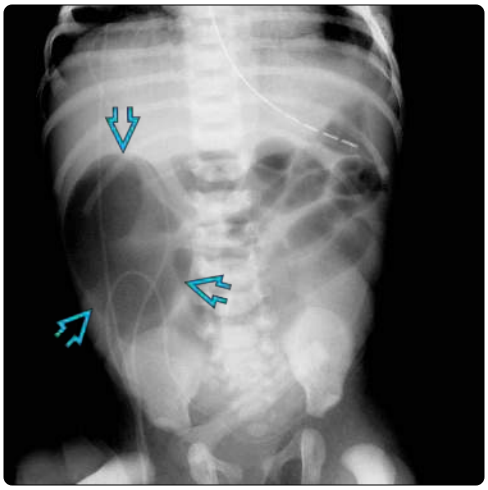
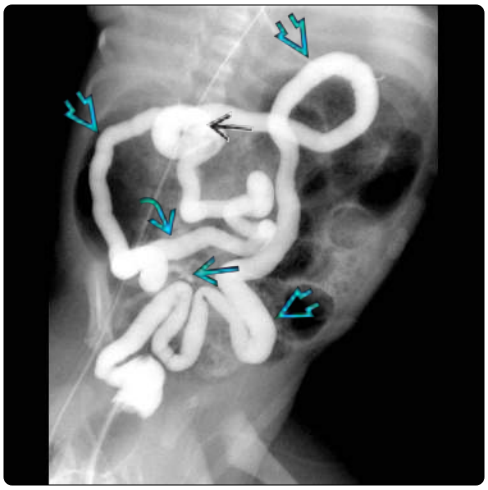
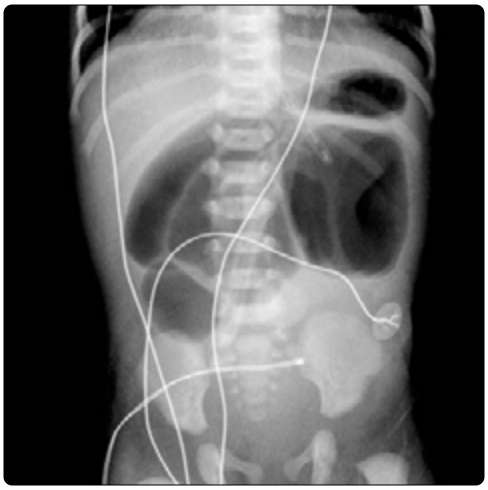
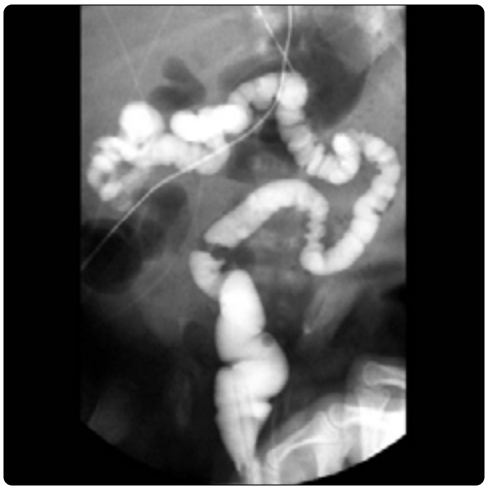


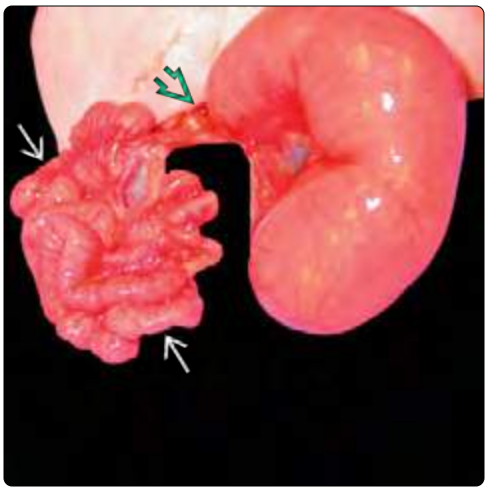


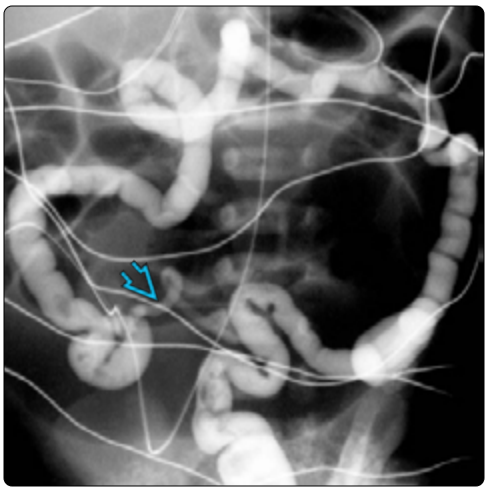
(Left) *AP radiograph in a 1-day-old term boy with abdominal distention & no meconium passage is shown. There are multiple dilated bowel loops, consistent with distal obstruction. A dominant dilated loop suggests there may be a distal ileal or colonic atresia, among the other diagnostic possibilities.* **(Right)** *Water-soluble contrast enema (WSCE) in the same patient shows the microcolon with reflux into the appendix & terminal ileum, which curls & ends blindly, consistent with ileal atresia.*





(Left) *AP radiograph of a 1-day-old boy with bilious emesis shows several moderately dilated loops of bowel, suggestive of an upper bowel obstruction, likely jejunal atresia.* **(Right)** *WSCE in the same patient shows a microcolon, suggesting that there may be an additional distal atresia or a long-segment atresia. Surgery confirmed the former.*





(Left) *Surgical photograph of same patient shows the dilated jejunum to the atretic segment; the distal bowel is decompressed. When the bowel was inspected in the OR, distal ileal atresia was noted, explaining the microcolon.* **(Right)** *WSCE in a 2 day old shows a microcolon with reflux into appendix. However, contrast would not reflux into the terminal ileum; this enema is indeterminate. Considerations include meconium ileus, ileal atresia, & total colonic Hirschsprung disease. At surgery, ileal atresia was found.*

Colonic Atresia

KEY FACTS

TERMINOLOGY

- Congenital colonic obstruction due to variable forms of interruption: Membrane (type I), fibrous cord (II), or complete separation with mesenteric defect (III)

IMAGING

- Radiography: Multiple dilated air-filled bowel loops ± 1 disproportionately dilated loop of atretic proximal colon
- Contrast enema: Distal microcolon with abrupt termination to retrograde contrast flow proximally (at colonic blind end)
  - Contrast does not fill cecum/terminal ileum or dilated bowel loops
  - Atresia may affect any segment of colon

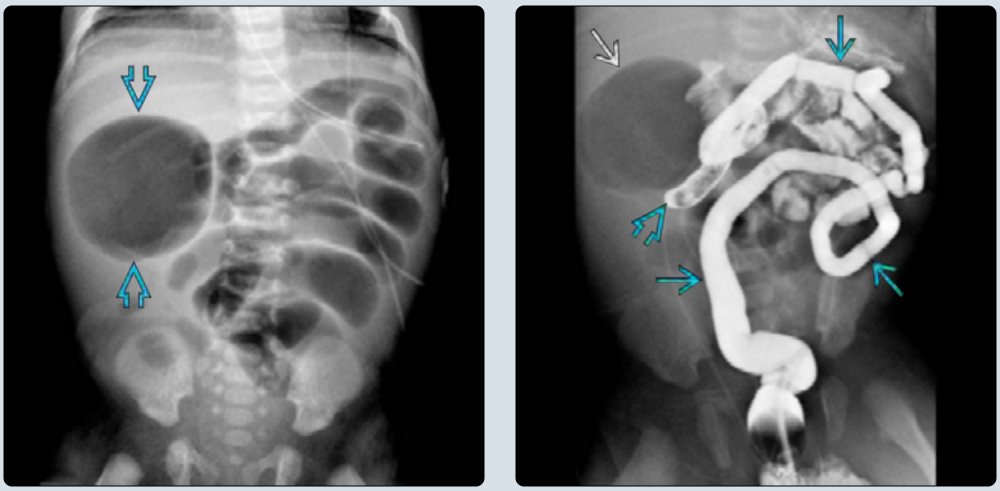
TOP DIFFERENTIAL DIAGNOSES

- Hirschsprung disease
- Meconium plug syndrome/neonatal small left colon
- Jejunoileal atresia
- Meconium ileus

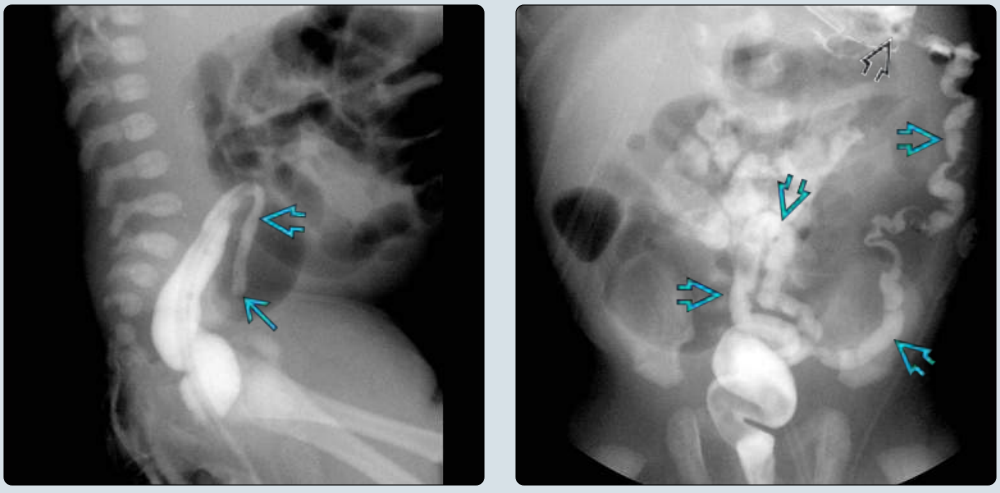
CLINICAL ISSUES

- Presents similar to other congenital distal bowel obstructions
  - Newborn with failure to pass meconium, abdominal distention, ± bilious emesis
    - If initial abdominal radiograph shows distal bowel obstruction → water-soluble contrast enema
    - History of bilious emesis frequent in distal obstructions; should not necessarily divert work-up to upper GI 1st (to rule out midgut volvulus) if patient clinically/radiographically suggestive of distal obstruction
- Least common of all intestinal atresias (1.8-15%)
  - Very rare; incidence ~ 1:20,000 live births
- Treatment: Surgery to eliminate bowel obstruction & establish intestinal continuity

(Left) *AP supine abdominal radiograph of a 1-day-old female full-term infant with bilious emesis shows multiple dilated gas-filled bowel loops in the left abdomen with a single disproportionately dilated loop in the right upper quadrant, an appearance often seen in colonic atresia.* **(Right)** *Supine contrast enema in the same patient (after a normal upper GI) shows a microcolon with a blind end. The adjacent large dilated loop was due to an atretic proximal colon.*



(Left) *Left-side down lateral contrast enema in a newborn with failure to pass meconium shows that the sigmoid colon is tiny & blind ending, consistent with colonic atresia at the sigmoid level.* **(Right)** *Supine contrast enema in a neonate with bilious emesis (& a normal preceding upper GI) shows a microcolon with a blind end at the distal transverse colon, consistent with colonic atresia.*



KEY FACTS

TERMINOLOGY

- Intestinal atresias multifocal in 6-32%; 2 distinct diseases
  - Multiple intestinal atresia (MIA)
    - Atresias of types I, II, IIIa, IIIb
    - Affects mid-duodenum to mid-distal transverse colon
      - Due to vascular insult to superior mesenteric artery territory (midgut)
  - Hereditary multiple intestinal atresia (HMIA) or multiple intestinal atresia with immunodeficiency (MIAI)
    - Atresias of types I & II only
    - Affects pylorus to rectum, including long segments of occlusion
      - Multiple proposed etiologies
    - Develop severe infections from immunodeficiency

IMAGING

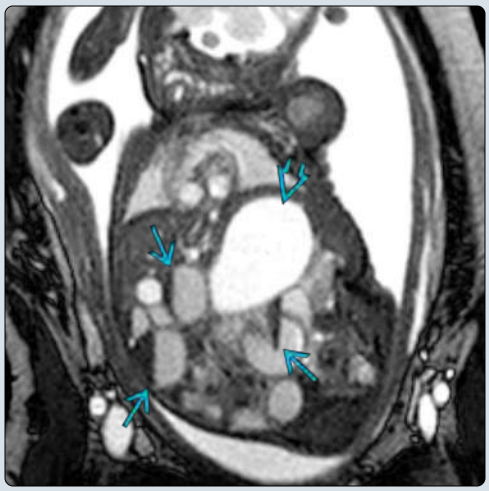
- No bowel gas beyond most proximal obstruction
- Ca^{2+} develop in chronically obstructed, dilated closed loops of intestine
- Enema typically shows small unused colonic segments distal to obstructions
- Ultrasound shows fluid-filled bowel ± echogenic Ca^{2+}, wall thickening; ± biliary dilation

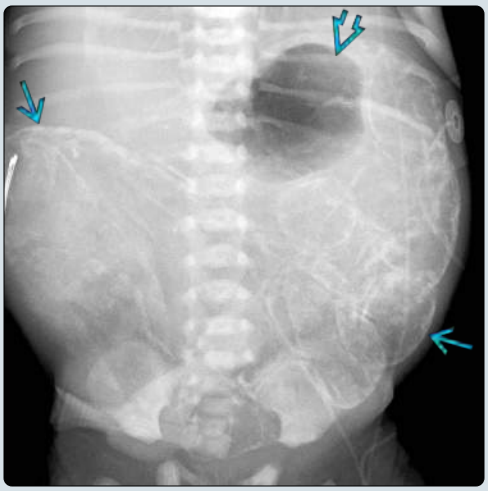
PATHOLOGY

- Histology of HMIA/MIAI: Characteristic "sieve-like" occluded segments (multiple lumina in single cross section)

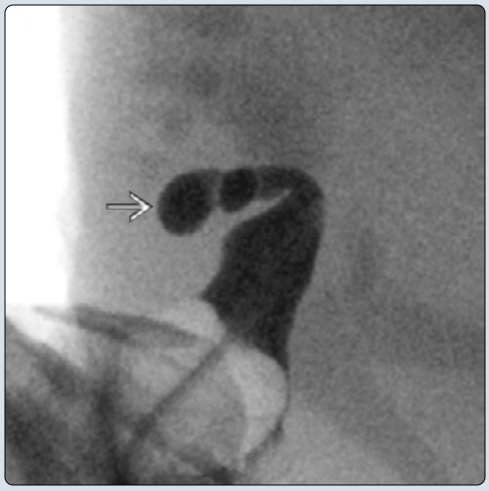
CLINICAL ISSUES

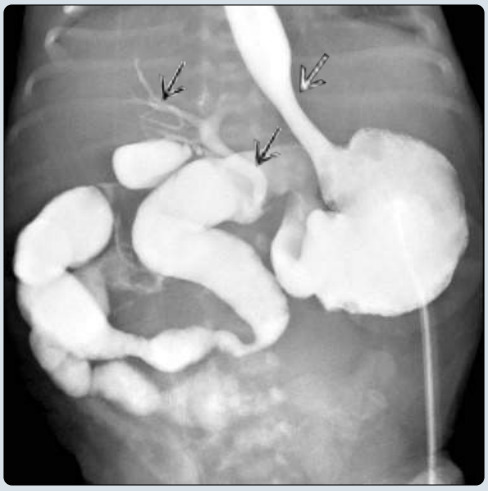
- MIA: Operative repair frequently curative; low mortality unless associated with gastroschisis or short gut syndrome
- HMIA/MIAI: Despite resection of atretic segments, patients have very high morbidity/mortality
  - Longstanding gut & biliary dysfunction
  - Immunocompromise with severe infections (similar to severe combined immunodeficiency or SCID)
    - Intestinal transplant may be curative with high level of immune system reconstitution by donor gut





(Left) *Coronal SSFP MR of a 33-week-gestation fetus shows numerous dilated bowel loops containing fluid-fluid levels. The distended stomach is of different signal intensity than the dilated bowel, which can suggest multiple intestinal atresias. No colonic meconium was seen on T1 MR.* **(Right)** *AP radiograph of the abdomen hours after birth shows extensive Ca^{2+} throughout the dilated bowel loops. The gas-filled stomach is moderately distended with no distal bowel gas identified.*





(Left) *Lateral view of a water-soluble contrast enema (with catheter balloon pressed externally against the anus) shows a small, blind-ending rectal pouch. Surgery confirmed numerous atresias at the pylorus, small bowel, & colon. This patient developed infections from a severe immunodeficiency & was diagnosed with hereditary multiple intestinal atresias.* **(Right)** *After resection of the atresias (same patient), upper GI (with contrast injected through the gastrostomy tube) shows free reflux into the esophagus & bile ducts.*

Meconium Ileus

KEY FACTS

TERMINOLOGY

- Neonatal obstruction of distal ileum due to abnormally thick, tenacious meconium
- Up to 90% of meconium ileus (MI) patients have cystic fibrosis (CF)
- MI is presenting illness in 10-20% of CF patients

IMAGING

- Microcolon + meconium-filled terminal ileum on water-soluble contrast enema (WSCE)
- Complicated MI
  - Soft tissue mass or gasless abdomen
  - ± intrauterine perforation (Ca^{2+} of meconium peritonitis)

TOP DIFFERENTIAL DIAGNOSES

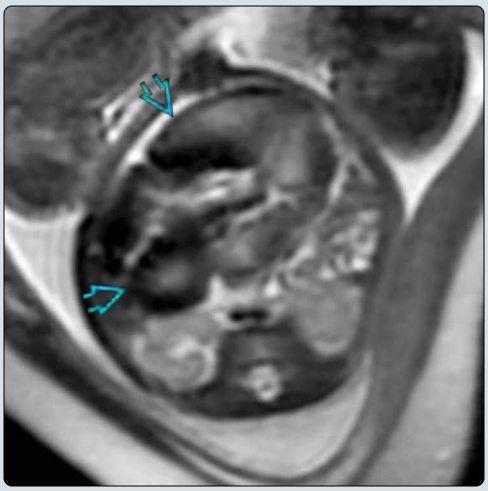
- Ileal atresia
- Hirschsprung disease
- Meconium plug syndrome
- Colonic atresia
- Megacystis-microcolon-intestinal hypoperistalsis syndrome
- Anorectal malformation
- Midgut volvulus

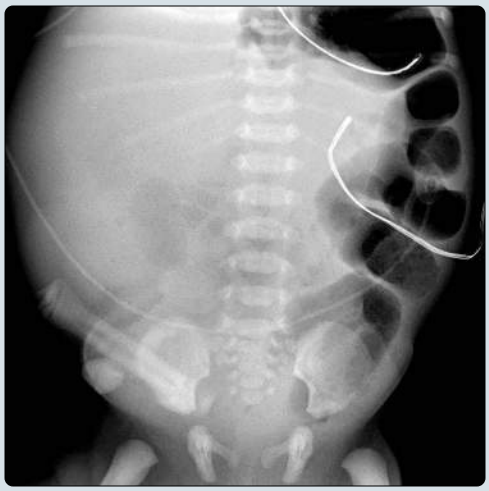
PATHOLOGY

- Mutation of *CFTR* gene (chromosome 7)
- Uncomplicated MI: 50%
- Complicated MI: 50%
  - Segmental volvulus, atresia, necrosis, ± perforation with meconium peritonitis

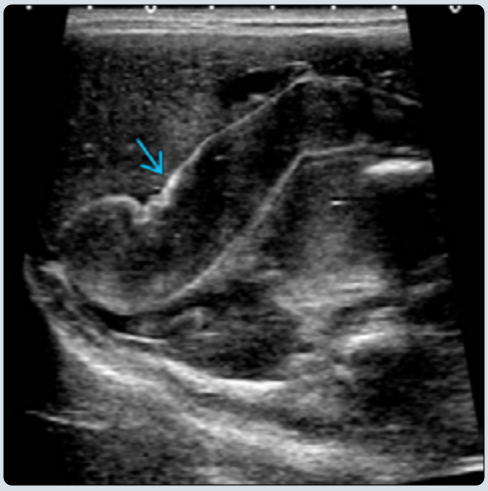
CLINICAL ISSUES

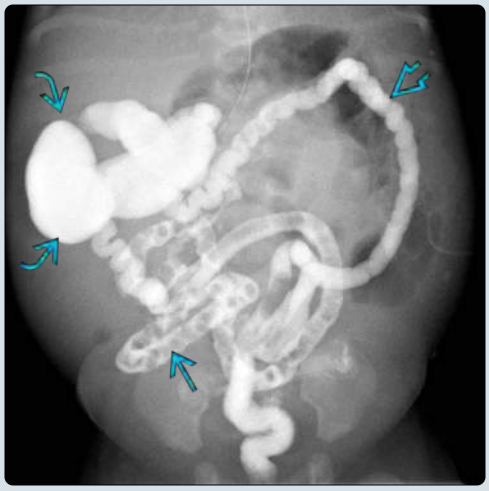
- CF incidence: 1:3,000 Caucasian live births (much less common in other races)
- Most common signs/symptoms: Failure to pass meconium, abdominal distention, bilious emesis
- Serial hyperosmotic WSCE vs. surgery for treatment of uncomplicated MI
  - Surgery often required in complicated MI





(Left) *Axial SSFSE T2 fetal MR at 33-weeks gestation shows dilated small bowel (SB) filled with intermediate to low signal intensity meconium. A tiny-caliber colon was best seen on sagittal images (not shown), consistent with an SB obstruction. The mother was noted to be a carrier of the cystic fibrosis (CF) gene.* **(Right)** *Radiograph of the same patient several hours after birth shows a large soft tissue mass displacing dilated bowel towards the left, worrisome for a complicated meconium ileus (MI).*





(Left) *US immediately following the radiograph in the same patient shows dilated, debris-filled bowel with echogenic walls consistent with bowel obstruction. Ascites was scattered in the abdomen. However, no pseudocyst or other sign of perforation was seen.* **(Right)** *WSCE in same patient on the same day shows a microcolon without significant meconium. Reflux into the terminal ileum (TI) shows obstructing meconium pellets before reaching dilated SB. The findings confirm complicated MI.*

TERMINOLOGY

Abbreviations

- Meconium ileus (MI)

Definitions

- Neonatal obstruction of distal ileum due to abnormally thick, tenacious meconium
  - Up to 90% of MI patients have cystic fibrosis (CF)
  - Presenting illness in 10-20% of CF newborns

IMAGING

General Features

- Best diagnostic clue
  - Microcolon + meconium-filled terminal ileum (TI) on water-soluble contrast enema (WSCE)
- Size
  - Microcolon (small unused colon due to impaired passage of succus entericus to colon in utero); dilation of proximal small bowel

Radiographic Findings

- Radiography
  - Uncomplicated (or simple) MI
    - Multiple dilated bowel loops
    - ± bubbly lucencies in right lower quadrant
      - Air mixed in meconium
    - Few if any air-fluid levels (sticky meconium)
  - Complicated (or complex) MI
    - Soft tissue mass or gasless abdomen
    - ± intrauterine perforation (meconium peritonitis)
      - Pseudocyst, peritoneal-lining Ca^{2+}
    - Ultrasound &/or enema for further evaluation
      - Enema treatment often fails (requiring surgery)

Fluoroscopic Findings

- Low osmolar WSCE for diagnosis; higher osmolar WSCE for therapy
  - "Smallest of microcolons" (often)
  - Contrast refluxed into TI outlines meconium pellets ("pearls on string")
    - Pellets may obstruct contrast passage proximally (mimicking ileal atresia)
    - Often not much colonic meconium
  - Switching to higher osmolar WSCE can be therapeutic in uncomplicated MI
    - Up to 80% success in experienced hands

MR Findings

- Fetal MR
  - Microcolon containing little T1-bright meconium in later gestation
  - T1-bright signal often seen in dilated, obstructed small bowel loops

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatally
    - Dilated, echogenic bowel
    - Echogenic ascites, peritoneal Ca^{2+}, pseudocyst from in utero perforation
  - Postnatally
    - Dilated, thick-walled small bowel loops containing heterogeneous meconium
      - Echogenic foci of gas in meconium
    - Echogenic ascites, peritoneal Ca^{2+}, pseudocyst from in utero perforation

Imaging Recommendations

- Best imaging tool
  - WSCE
- Protocol advice
  - With neonatal distal bowel obstruction on radiographs → WSCE for further investigation
    - Barium not used (may impede meconium passage)
    - Nonballoon tip catheter used (in general) due to possible risk of rectal injury
      - If using balloon-tipped Foley, inflate balloon outside & insert tip into rectum with balloon held up against anus to prevent leakage
      - Alternatively, carefully inflate balloon with small amount of contrast in rectum (under fluoroscopy to prevent rectal injury by overinflating)
      - Enema contrast gravity infusion vs. gentle pulsing of contrast by syringe injection
    - If microcolon seen, attempt reflux into ileum
      - Visualization of meconium pellets diagnostic
    - Switch to higher osmolality (600-900 mOsm) contrast for therapeutic enema
      - Reflux contrast up to dilated bowel if possible
      - Active fluid resuscitation during enema to balance fluid shifts into bowel: 1.5x maintenance
      - Have surgical team check electrolytes after each therapeutic enema
    - Serial enemas may be required to relieve meconium obstruction; usually 1 per day
    - Complications of enema
      - Perforation
      - Dehydration/intravascular volume depletion/hypotension
      - Electrolyte imbalances
      - Segmental volvulus of dilated proximal bowel if no meconium clearance
  - If bowel obstruction with central mass on radiographs (suggesting complicated MI), consider ultrasound ± enema
  - If enema normal (uncommon with numerous dilated loops), consider upper GI
    - Exclude midgut volvulus causing ischemic ileus

DIFFERENTIAL DIAGNOSIS

Ileal Atresia

- Microcolon on WSCE
- Enema contrast refluxing into ileum stops abruptly at atresia
- Opacified distal ileum may be small without meconium

Hirschsprung Disease

- Microcolon in some cases of total colonic Hirschsprung disease (HD) on WSCE
- In more common setting of short-segment HD
  - Low rectosigmoid (R:S) ratio (< 1); serrated mucosa

- ± meconium plugs in colon above spasmodic rectum

Small Left Colon/Meconium Plug Syndrome

- Small distal colon; transition at splenic flexure on WSCE
- Nonobstructing meconium plugs in colon (not TI)
- Enema usually curative of functional obstruction
- Not associated with CF

Colonic Atresia

- Microcolon up to level of atresia on WSCE
- Colon proximal to atresia dilated out of proportion to small bowel

Anorectal Malformation

- Distal obstruction without normal anus on exam

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome

- Microcolon on WSCE
- Very dilated bladder
- M:F = 1:4; often fatal

Midgut Volvulus

- Bowel loops may be dilated from ischemic ileus
- Upper GI most reliable diagnostic tool

PATHOLOGY

General Features

- Genetics
  - CF: Autosomal recessive
  - Mutation of *CFTR* gene (chromosome 7)
    - > 900 mutations; ΔF508 mutation most common overall & with MI
    - Faulty chloride transport across epithelium → ↓ water in luminal contents → dehydrated, thick secretions
      - Obstruction of glands & ducts
      - Pancreas, intestines, lungs most affected
    - Thick meconium → distal ileal obstruction
- Associated abnormalities
  - CF manifestations beyond neonatal period
    - Lung disease
    - Exocrine pancreas failure, biliary disease, chronic appendiceal dilation without inflammation, distal intestinal obstruction syndrome (DIOS)

Staging, Grading, & Classification

- Uncomplicated MI: 50%
- Complicated MI: 50%
  - Segmental volvulus
  - Atresia
  - Necrosis
  - Perforation with meconium peritonitis
    - Ascites, peritoneal surface Ca^{2+}, pseudocyst (may be large)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Failure to pass meconium, abdominal distention, bilious emesis

Demographics

- Age
  - Newborn
- Gender
  - M = F
- Epidemiology
  - CF: 1:3,000 Caucasian live births (much less common in other races)
  - Up to 90% of patients with MI have CF
    - Some reports of up to 50% of MI patients without CF
      - Complicated MI rate may be higher
      - Prematurity + low birth weights more likely
  - 10-20% of CF patients present with MI

Natural History & Prognosis

- MI patients classically have worse lung function, nutritional status, & survival vs. other CF patients
  - More recent papers show no difference
  - Earlier diagnosis of CF may be beneficial
- Recurrent bowel issues in CF
  - DIOS ("MI equivalent")
    - Older children with obstruction from thick stool in TI
  - Chronic constipation (may be 1st CF manifestation)

Treatment

- Uncomplicated MI: Serial hyperosmotic, water-soluble enemas vs. surgery
  - Therapeutic enema success up to 83% (with experience)
    - Reported success rates vary widely (5-83%)
  - Perforation rate of enema 1-3%
    - Greatest with injection & use of rectal balloon
  - Surgery for patient decompensation, failed enemas, or perforation
    - Enterotomy, meconium removal, primary anastomosis vs. temporary enterostomy
- Complicated MI: Surgery
  - Resect abnormal bowel, meconium removal, primary anastomosis vs. temporary enterostomy
- Testing for CF

DIAGNOSTIC CHECKLIST

Consider

- Causes of distal obstruction & microcolon
- Diluted hyperosmolar WSCE for simple MI therapy
- Enema rarely curative in complicated MI (requires surgery)

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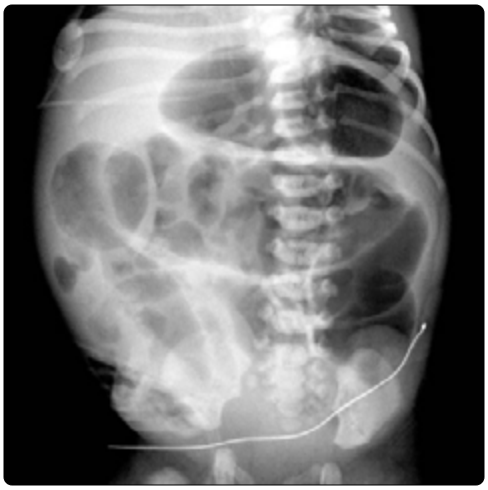
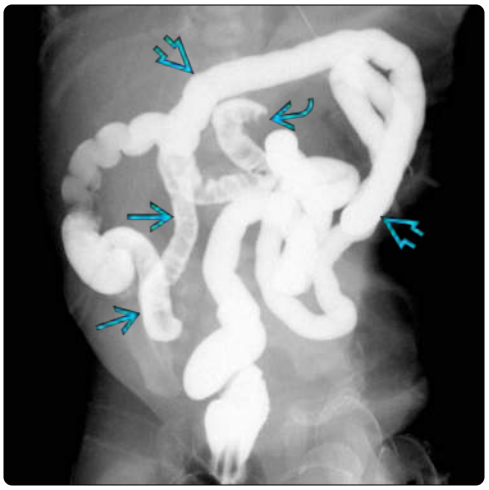
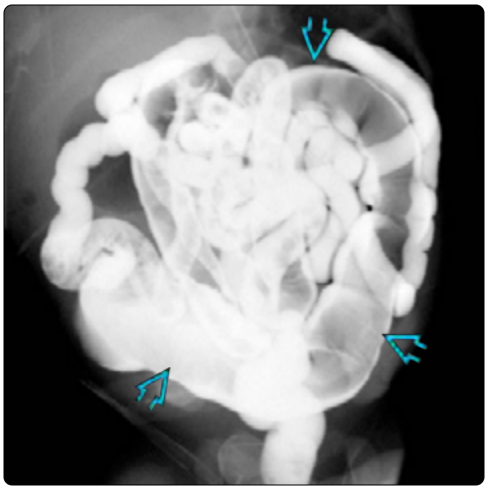
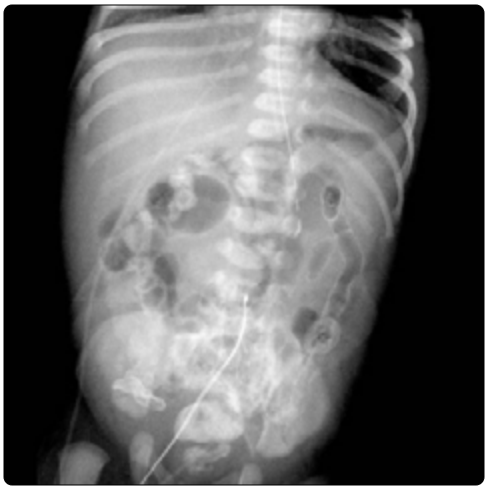


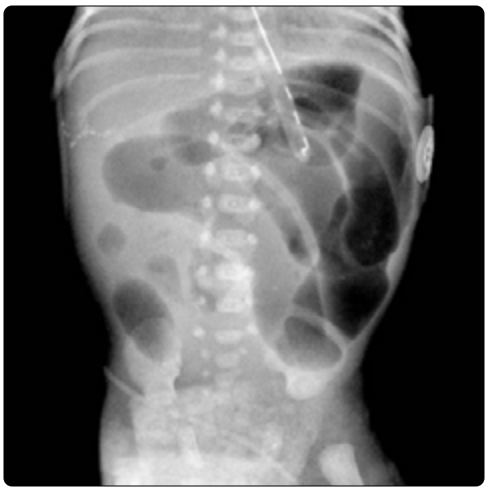


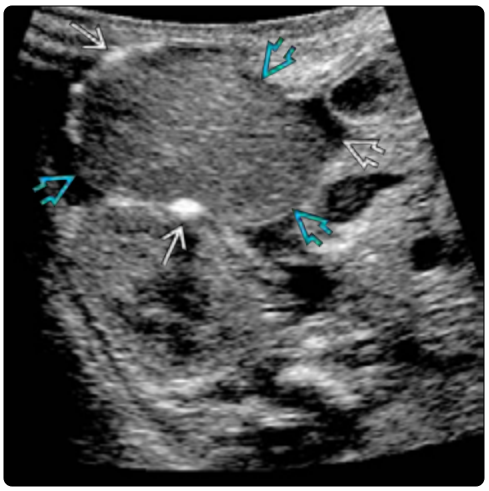
(Left) *AP radiograph in a full-term newborn with failure to pass meconium (& whose parents are CF gene carriers) shows a distal bowel obstruction without soft tissue mass effect or signs of meconium peritonitis. With this history, MI is the primary diagnostic consideration.* **(Right)** *Supine WSCE in the same patient shows a microcolon & reflux of contrast into meconium-filled TI, most consistent with meconium ileus. Contrast did not reach dilated bowel.*





(Left) *A 2nd supine WSCE was performed in the same patient. There is now reflux of the high osmolality 1:1 diluted Gastroview (950 mOsm) into multiple dilated bowel loops proximal to the obstructing meconium.* **(Right)** *AP abdominal radiograph in the same patient 1 day after the 2nd enema shows much less dilation of the bowel proximal to the previously obstructed TI. There is mild residual contrast in the persistently small, unused colon. This is considered a successful treatment enema for MI.*





(Left) *Newborn AP radiograph shows calcification in the right upper quadrant (RUQ) (suggestive of meconium peritonitis) plus dilated bowel loops of a distal bowel obstruction. Both parents were CF gene carriers.* **(Right)** *US shows a round fluid collection in the RUQ with punctate echogenic foci in the wall, likely corresponding to the calcification seen on the radiograph. There is also a small amount of ascites. The WSCE (not shown) confirmed a microcolon with impacted TI pellets, giving a diagnosis of complicated MI.*

Neonatal Small Left Colon

KEY FACTS

TERMINOLOGY

- Transient functional colonic obstruction of newborn
  - Retained colonic plugs of normal meconium are secondary to functional obstruction in this scenario (not an underlying cause of mechanical obstruction)
- Synonyms: Functional immaturity of colon, meconium plug syndrome

IMAGING

- Numerous dilated loops of bowel on newborn radiograph
  - Difficult to radiographically distinguish small vs. large bowel in neonate
- Water-soluble contrast enema (WSCE)
  - Small-caliber left colon (sigmoid + descending segments) up to splenic flexure with abrupt or gradual transition to normal/mildly dilated transverse colon
    - Normal rectosigmoid ratio
  - Scattered meconium filling defects in colon
    - May be proximal or distal to level of caliber change

TOP DIFFERENTIAL DIAGNOSES

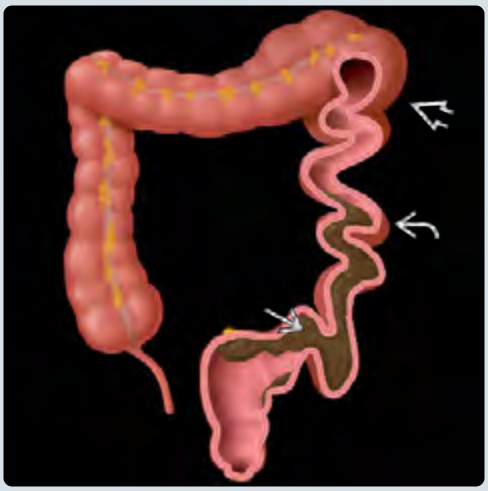
- Hirschsprung disease
- Ileal atresia
- Meconium ileus
- Anorectal malformation
- Ileus secondary to midgut volvulus or necrotizing enterocolitis

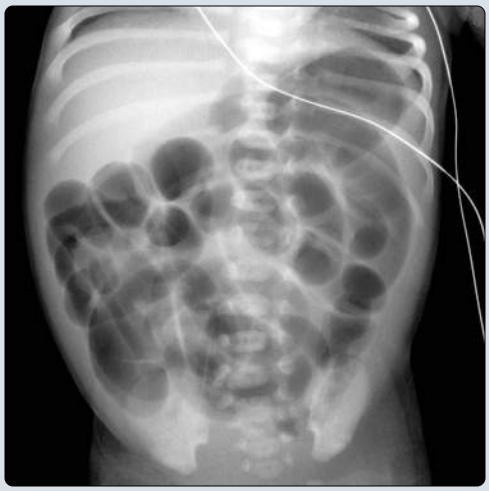
PATHOLOGY

- ↑ incidence in infants of diabetic mothers or mothers who received magnesium sulfate

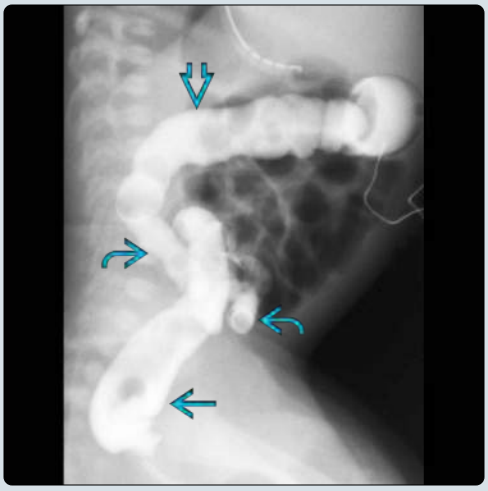
CLINICAL ISSUES

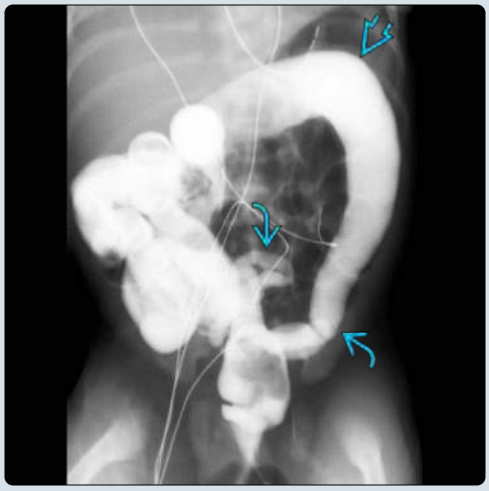
- Presents with abdominal distention, delayed passage of meconium, bilious emesis
- Temporary phenomenon: Usually resolves within several days (hastened by enemas or rectal stimulation)
- Rectal biopsy to exclude Hirschsprung disease if symptoms persist; some advocate biopsy for all





(Left) *Frontal graphic shows meconium plug syndrome with a small left colon up to the splenic flexure. A plug of meconium is shown in the small sigmoid colon, but this normal meconium does not cause the obstruction (unlike meconium ileus).* **(Right)** *AP radiograph of the abdomen in a term neonate with abdominal distention & emesis shows diffuse dilation of bowel but no rectal gas, suggesting a distal intestinal obstruction. The patient's mother had received magnesium late in pregnancy.*





(Left) *Lateral water-soluble contrast enema in the same patient shows a normal-caliber rectum with small sigmoid & descending colonic segments. There is a gradual transition to a normal-caliber distal transverse colon.* **(Right)** *Supine frontal image from the same contrast enema shows the gradual transition from small sigmoid & descending colonic segments to a normal splenic flexure. Meconium was passed with stimulation, & biopsy was negative for Hirschsprung disease, consistent with functional immaturity.*

TERMINOLOGY

Synonyms

- Meconium plug syndrome (MPS)
- Functional immaturity of colon

Definitions

- Transient functional obstruction of newborn colon
  - Retained colonic meconium merely secondary finding (not primary cause of mechanical obstruction)

IMAGING

Radiographic Findings

- Multiple dilated loops of bowel in newborn (i.e., neonatal distal bowel obstruction)
  - Note that dilated large & small bowel loops (from any cause) cannot be reliably differentiated in neonates by morphology or size

Fluoroscopic Findings

- Contrast enema
  - Rectosigmoid ratio normal (> 1)
    - Rectum of relatively normal caliber
  - Small-caliber left colon (sigmoid & descending segments) up to splenic flexure
  - Abrupt or gradual transition to normal/mildly dilated proximal colon
  - Multiple filling defects of normal meconium scattered throughout colon, often present in narrowed segment
    - May be displaced proximally during enema
  - Meconium plugs frequently pass during enema
  - Enema often therapeutic

DIFFERENTIAL DIAGNOSIS

Hirschsprung Disease

- Rectal caliber smaller than sigmoid ± serrated mucosa/irregular contractions
- Distal aganglionic segment small in caliber up to transition
  - Long-segment Hirschsprung disease (HD) with splenic flexure transition zone may mimic functional immaturity

Ileal Atresia

- Microcolon
- Contrast refluxed into ileum stops abruptly at atresia
- Cannot opacify proximal dilated small bowel

Meconium Ileus

- Microcolon
- Enema contrast outlines obstructive meconium pellets in distal ileum
- Contrast may or may not reach dilated small bowel proximal to impacted meconium
- Most have cystic fibrosis (CF)

Anorectal Malformation

- Distal obstruction with no normal anus present

Colonic Atresia

- Distal obstruction with disproportionately dilated loop
- Microcolon to level of atresia on enema

Midgut Volvulus

- Bowel loops may be diffusely dilated due to ischemic ileus in ill neonate
- Upper GI study of choice in ill neonate with bilious emesis

PATHOLOGY

General Features

- Etiology
  - Unclear; probably due to immature ganglion cells or hormonal receptors resulting in transient functional obstruction of colon
  - ↑ incidence with
    - Infants of diabetic mothers (40-50% of MPS)
    - Infants of mothers who receive magnesium sulfate
  - Plugs of normal meconium do not cause obstruction in this diagnosis (unlike abnormal meconium in CF)
- Genetics
  - Rare familial reports
  - Few reports with CF controversial (unlike established CF-meconium ileus link)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal distention
  - Delayed passage of meconium (> 24-48 hours)
  - Bilious emesis

Natural History & Prognosis

- Temporary phenomenon: Usually resolves within few days
- Excellent prognosis

Treatment

- Condition resolves over time, hastened by rectal stimulation
  - Often resolves after diagnostic contrast enema
- Suction rectal biopsy to exclude HD if symptoms persist
  - Some advocate biopsy regardless of symptom resolution

DIAGNOSTIC CHECKLIST

Consider

- Long-segment HD may have similar appearance
  - Obtain rectal biopsy if functional obstruction persists after characteristic enema

Image Interpretation Pearls

- Usually normal rectosigmoid ratio (> 1)
- Small left colon ± scattered meconium plugs
- Transition point from small to normal/dilated colon at splenic flexure

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Hirschsprung Disease

KEY FACTS

TERMINOLOGY

- Congenital disorder of enteric nervous system: Absence of ganglion cells in intestinal myenteric & submucosal plexus
  - Lack of peristalsis → functional bowel obstruction
- Aganglionic segment extends retrograde from anus for variable length with gradual transition to normal innervation

IMAGING

- Newborn radiograph: Numerous loops of dilated bowel
- Radiograph outside neonatal period: Large stool burden with variable colonic dilation
- Contrast enema especially useful in evaluating neonate with distal bowel obstruction; findings suggestive of Hirschsprung disease (HD) include
  - Rectosigmoid ratio < 1
  - Transition zone from small distal colon to dilated proximal colon

TOP DIFFERENTIAL DIAGNOSES

- Neonatal small left colon (meconium plug syndrome)
- Ileal atresia
- Meconium ileus
- Anorectal malformation
- Milk allergy colitis

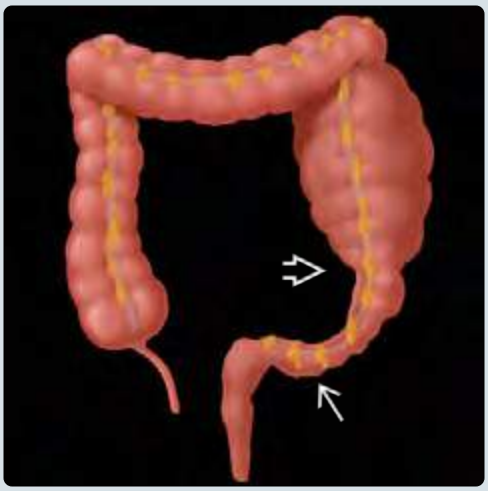
CLINICAL ISSUES

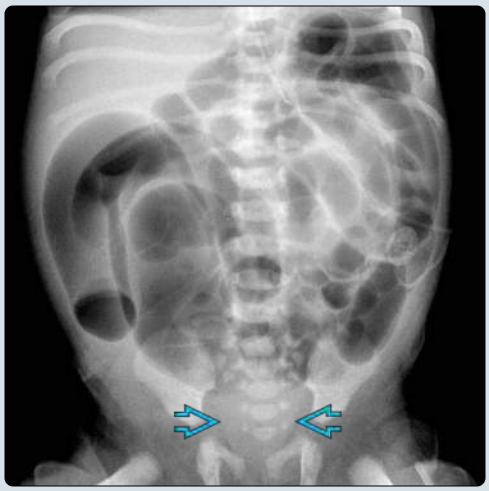
- Neonate: Failure to pass meconium, abdominal distention, bilious emesis, enterocolitis
- Older child: Constipation since birth, enterocolitis
- Diagnosis by rectal biopsy
- Treatment: Resect affected colon & pullthrough normal bowel to anus

DIAGNOSTIC CHECKLIST

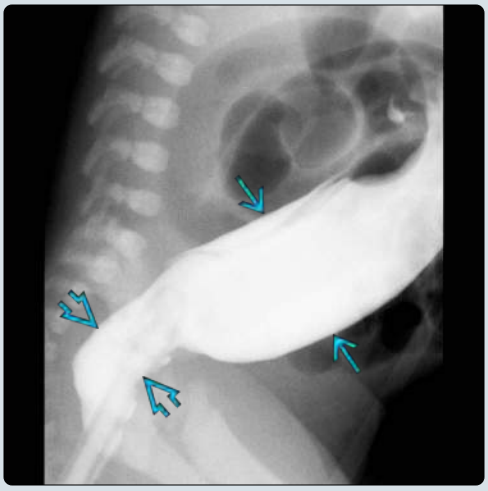
- **Contrast enema cannot rule out HD** (but can suggest HD or other diagnoses)
- Frank colitis in term newborn: HD until proven otherwise

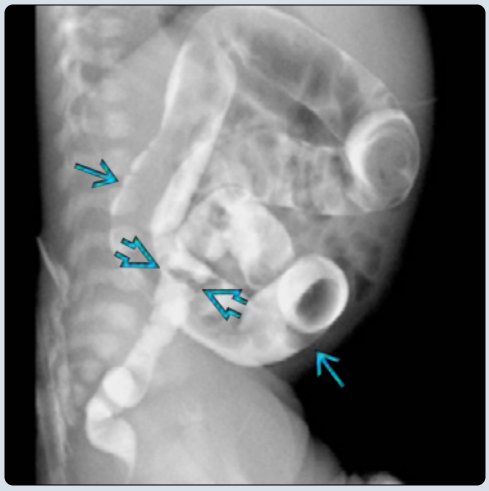
(Left) *Graphic shows a narrow caliber distal colon with a transition to a dilated proximal colon at the sigmoid-descending colonic junction, characteristic of short-segment Hirschsprung disease (HD).* **(Right)** *AP radiograph of a 2-day-old boy with failure to pass meconium shows multiple dilated loops of bowel, suggesting a distal bowel obstruction. No air is seen in the rectum. One cannot differentiate small bowel (SB) from colonic loops on this exam. A contrast enema is required to elucidate the etiology of obstruction.*





(Left) *Lateral view of a water-soluble contrast enema (WSCE) in the same patient shows a small caliber rectum relative to the dilated sigmoid, suggesting short-segment HD. This was confirmed by rectal biopsy.* **(Right)** *Lateral WSCE in a 2 day old with a history of failure to pass meconium & bilious emesis (as well as radiographs suggestive of a distal bowel obstruction) shows a small caliber rectum with a sigmoid transition to a dilated, meconium-filled colon, suggesting HD. Biopsy was confirmatory.*





TERMINOLOGY

Abbreviations

- Hirschsprung disease (HD)

Synonyms

- Colonic aganglionosis

Definitions

- Congenital anomaly of enteric nervous system
  - Absence of ganglion cells in myenteric & submucosal plexus of intestine
    - Lack of peristalsis → functional bowel obstruction
  - Aganglionic segment extends retrograde from anus for variable length with gradual transition to innervated colon
    - Rectosigmoid: Short-segment HD (70-80%)
    - Proximal to rectosigmoid: Long-segment HD (15-25%)
    - Entire colon: Total colonic HD (4-13%)
    - Colon & small bowel (SB): Total intestinal HD (very rare)
    - Just above anorectal verge: Ultrashort-segment HD (very rare)

IMAGING

General Features

- Best diagnostic clue
  - Rectosigmoid ratio < 1 on contrast enema
    - Requires well-distended lateral view from rectum to splenic flexure
- Morphology
  - Small caliber of aganglionic distal colon
  - Dilation of innervated proximal colon above transition

Radiographic Findings

- In newborn
  - Numerous dilated bowel loops suggesting distal obstruction
  - ± irregular, thickened bowel wall with enterocolitis
    - Rarely pneumatosis
  - Rarely intraluminal ileal Ca^{2+} in total colonic HD
- Outside neonatal period
  - Large stool burden with varying degrees of dilation
  - Paucity of gas with thick colonic wall: Question enterocolitis
  - Rarely, free air from perforation in 1st year of life
    - Perforation usually at proximal colon or appendix

Fluoroscopic Findings

- Contrast enema
  - Short- or long-segment HD
    - Rectosigmoid ratio classically < 1 (but not always)
    - Transition zone: Level at which small distal colon becomes dilated proximal colon
    - ± sawtooth pattern of distal mucosa (spasm)
    - ± irregular mucosa/thickened wall of enterocolitis
      - Cobblestone appearance
    - Delayed (> 24-48 hours) evacuation of contrast
    - Near normal enema in ultrashort-segment HD
  - Total colonic HD
    - May appear normal vs. microcolon vs. shortened length with round flexures (question mark- or comma-shaped)
    - Rectum often same caliber as rest of colon (rectosigmoid ratio ≤ 1)
    - SB proximal to colon may be dilated

Imaging Recommendations

- Best imaging tool
  - Water-soluble contrast enema (WSCE)
- Protocol advice
  - With clinical + radiographic suspicion for neonatal distal bowel obstruction → WSCE
    - Low osmolality water-soluble contrast
    - Lateral & AP views of rectum up to splenic flexure on early filling images
      - Compare rectum to sigmoid + more proximal colon
      - Normal rectosigmoid ratio > 1
    - WSCE stopped once transition zone clear
    - If WSCE normal, consider upper GI to exclude midgut volvulus (MV), as ischemic ileus can mimic obstruction

DIFFERENTIAL DIAGNOSIS

Neonatal Distal Intestinal Obstruction With Small Distal Colon

- Neonatal small left colon (meconium plug syndrome)
  - Nonpathologic transient functional obstruction
  - WSCE: Rectosigmoid ratio usually > 1, sharp transition at splenic flexure
  - ± rectal biopsy to exclude long segment HD
- Colonic atresia
  - WSCE: Blind-ending small caliber distal colon
  - Dilated proximal colon (larger compared to dilated SB)

Neonatal Distal Intestinal Obstruction With Microcolon

- Ileal atresia
  - WSCE: Blind end of contrast column in ileum
- Meconium ileus
  - WSCE: Contrast outlines meconium pellets obstructing terminal ileum & may extend proximally into dilated SB
- Megacystis microcolon intestinal hypoperistalsis syndrome
  - WSCE: Tiniest microcolon
  - Dilated floppy bladder, most common in females

Neonatal Distal Intestinal Obstruction With Normal Colon

- Omphalomesenteric duct remnant
  - Various forms: Fibrous band, Meckel diverticulum
  - WSCE: May see medialization or beaking of cecum
- Inguinal hernia
  - May visualize gas in inguinal canal or scrotum
- Midgut volvulus
  - Ischemic ileus in ill neonate mimics distal obstruction
  - Consider upper GI series first in ill neonate with bilious emesis
- Necrotizing enterocolitis
  - Prematurity most common etiology by far
  - Ischemic ileus mimics distal obstruction
  - Not typical in first few days after birth

Enterocolitis in Infant

- Milk allergy colitis
  - First weeks of life, usually with formula feeds
  - WSCE: Rectosigmoid ratio < 1 ± colitis
  - Rectal biopsy: Eosinophils, normal ganglion cells

Intraluminal Calcifications

- Anorectal malformation
- Multiple intestinal atresias
- Total colonic HD

PATHOLOGY

General Features

- Etiology
  - Precise mechanism unknown for sure
    - Defect in 1st-trimester migration of enteric ganglia
    - Normal migration but abnormal survival of ganglia
- Genetics
  - Most cases sporadic
  - Numerous genetic mutations found
  - Familial HD in 8-10%
- Associated abnormalities
  - 5-32% of HD overall
    - Down syndrome (7-15%)
    - Neurocristopathy syndromes
    - Congenital central hypoventilation ("Ondine curse")
    - Colonic atresia
    - Other isolated anomalies

Microscopic Features

- Diagnosis by rectal biopsy
  - Suction biopsy: Bedside, less reliable
  - Full-thickness biopsy: In operating room, definitive
- Absent ganglia in myenteric & submucosal plexus
- Hypertrophic nerve fibers (acetylcholinesterase positive)
- Disease generally contiguous without skip areas
  - Reported cases of segmental HD
    - May lead to incorrect diagnosis or inadequate resection/failed pullthrough
- Radiologic-pathologic correlation of transition zone site
  - Short-segment (or low transition zone) HD: 75% correlation
  - Long-segment (or high transition zone) HD: 25% correlation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Failure to pass meconium 24-48 hours of life (60-90%)
  - Abdominal distention (63-91%)
  - Bilious vomiting (19-37%)
  - Enterocolitis (5-44%)

Demographics

- 90% diagnosed in newborn period
- 10% diagnosed later, rarely adolescent or adult
- M > F = 4:1 (long-segment & total colonic HD → 1:1)
- 1:5,000 live newborns

Natural History & Prognosis

- Untreated HD may lead to constipation, enterocolitis, toxic megacolon, sepsis, death
- Treated: Up to 40% with chronic soiling, constipation
  - Must assess for failure of pull-through procedure
  - Poorer outcome: Down syndrome, total colonic HD

Treatment

- Resection of aganglionic colon
- Pullthrough of normal bowel to anus
- Complications
  - Early: Leak, infection, obstruction
  - Late: Bowel adhesion/obstruction, stricture, enterocolitis, constipation, incontinence
- Common etiologies for failed pullthrough
  - Soave cuff, Duhamel pouch, ischemic stricture, residual HD, twisted pullthrough

DIAGNOSTIC CHECKLIST

Consider

- **Contrast enema (CE) cannot rule out HD**
  - With clinical suspicion, must biopsy
- CE most useful in assessing distal obstruction in neonates
  - Only up to 80% sensitive for HD in newborns

Image Interpretation Pearls

- Rectosigmoid ratio < 1 & transition zone on WSCE → HD
- Frank colitis in term newborn: HD until proven otherwise

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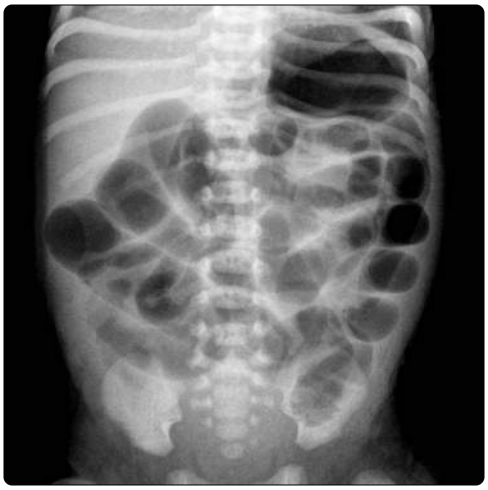
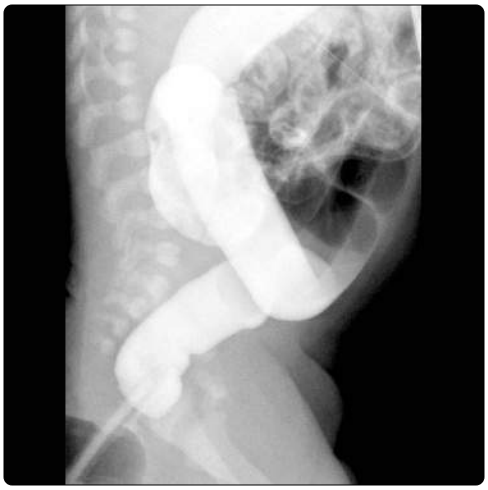
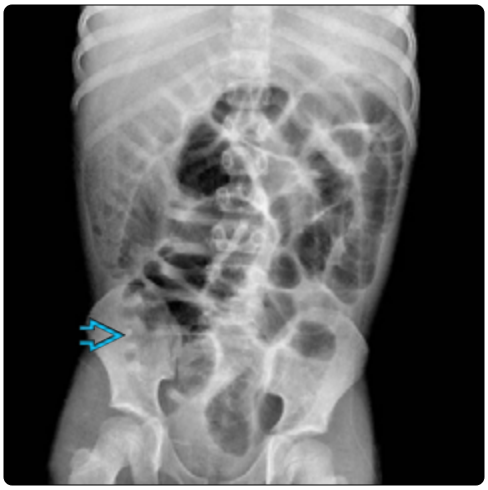
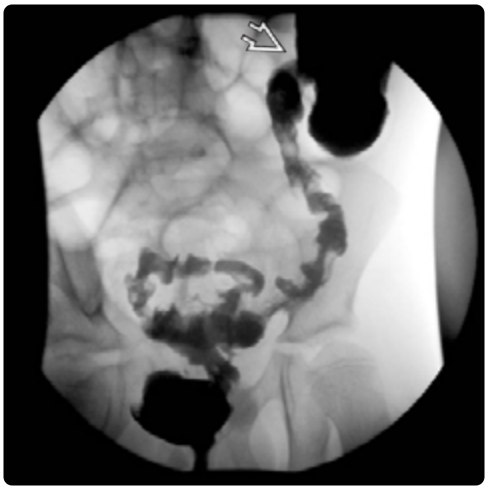


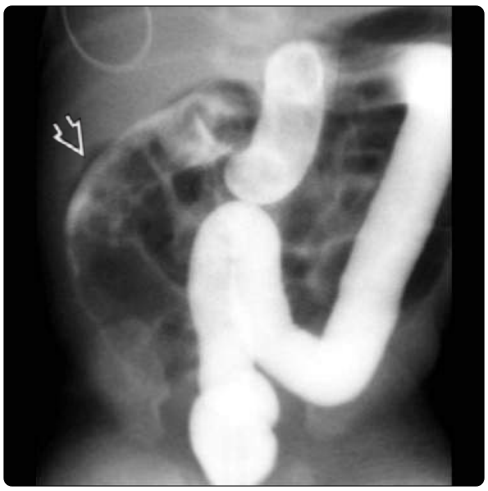


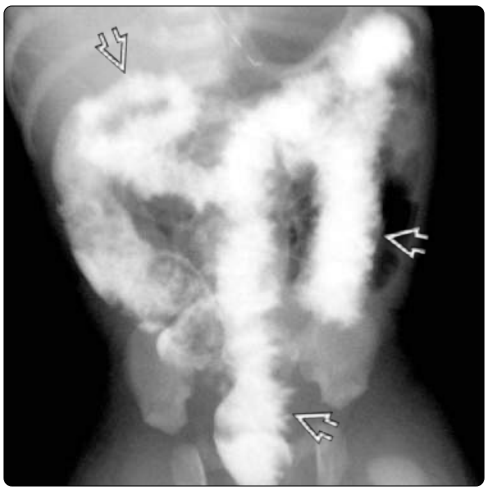
(Left) *AP radiograph of a 2 day old with abdominal distention & failure to pass meconium shows multiple air-filled bowel loops diffusely, suggesting a distal intestinal obstruction. The next study should be a WSCE to elucidate the etiology of this distal bowel obstruction.* **(Right)** *Lateral view of a WSCE in the same patient shows the rectum to be slightly smaller in caliber as compared to sigmoid & proximal colon without an abrupt transition. However, this appearance raised suspicion for HD, & biopsy was confirmatory.*





(Left) *AP radiograph in an 8 year old with a long history of constipation and multiple episodes of colitis shows little stool after a clean out. There are dilated colonic & SB loops with possible wall-thickening* ⇨. **(Right)** *Frontal view from a WSCE in the same patient shows a fairly distended rectum. However, the colon was narrow & spastic almost to the splenic flexure* ⇨, *consistent with long-segment HD, which was confirmed at biopsy.*





(Left) *Frontal view from a WSCE in a newborn with total colonic HD shows a mildly small caliber colon, which is fairly uniform throughout. The course of the colon is shortened with rounding of the flexures* ⇨. **(Right)** *Frontal view of a WSCE in a newborn with bilious emesis shows severe mucosal irregularity* ⇨ *throughout the colon, suggesting colitis in HD. Biopsies showed total intestinal aganglionosis.*

KEY FACTS

IMAGING

- Neonatal clinical exam + AP abdominal radiograph
  - Many dilated bowel loops, ± rectal gas, ± bowel Ca^{2+}
  - Prone XTL view if fistula not clinically evident (~ 5%)
- Renal, spine US; pelvic US for females with cloaca
  - Urgent drainage of hydrocolpos required to prevent rupture & alleviate bladder/ureteral obstruction
- Evaluate additional anomalies (VACTERL or syndromes)
- Delayed distal colostogram for classification of ARM & preoperative planning: Must visualize fistula (~ 95%)
  - Perfect lateral view (superimposed femoral heads)
  - If bladder fills, fill until voiding; image distal colonic segment, bladder, urethra
- ± pelvic MR prior to, during, &/or after operative repair

TOP DIFFERENTIAL DIAGNOSES

- Neonatal distal bowel obstruction (with normal anus)
  - Meconium plug syndrome, Hirschsprung disease, meconium ileus, jejunoileal atresia
- Neonatal abdominal Ca^{2+}
  - In bowel: Total Hirschsprung, multiple intestinal atresia

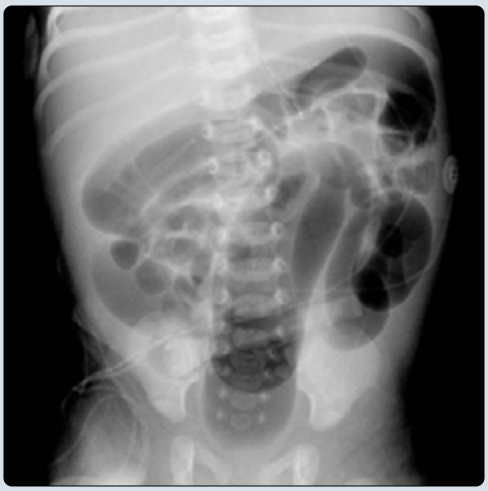
PATHOLOGY

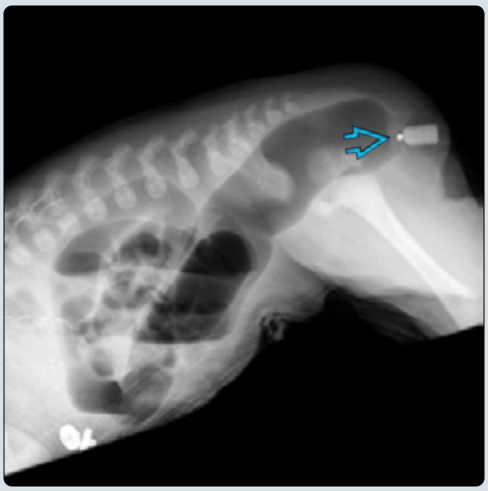
- Imperforate anus
  - Rectoperineal fistula (male or female)
  - Rectovestibular fistula: 25% of female ARMs
  - Rectourethral fistula: 50% of male ARMs
  - Rectobladder neck fistula: 10% of male ARMs
  - No fistula: 5% of ARMs (male or female)
  - Cloacal malformation: Only females
- Rectal atresia or stenosis: 1% of ARMs

CLINICAL ISSUES

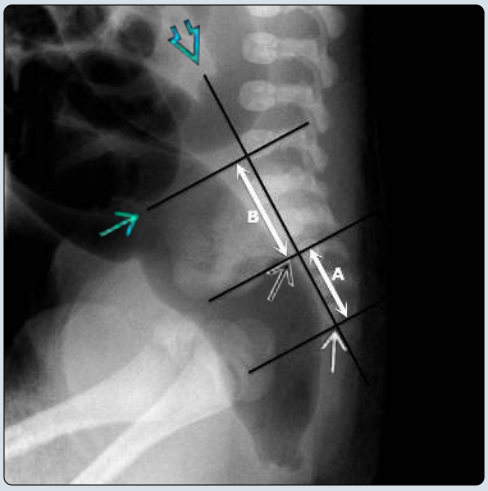
- Goals: Maximize continence of feces & urine, sexual function
- Most ARMs require
  - Diverting colostomy within days of birth
  - PSARP for definitive repair months later

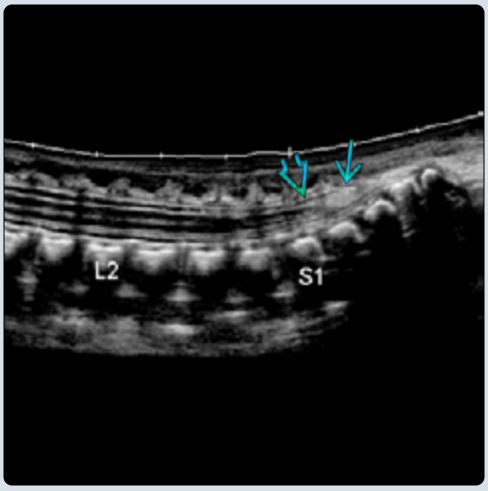
(Left) *AP radiograph in a newborn (24 hours old) boy with imperforate anus & no clinical signs of a fistula (no meconium per urethra or perineum) shows multiple dilated bowel loops to the pelvis with a dilated rectum.* **(Right)** *Prone cross-table lateral view with a BB on the anus in the same patient shows the rectal pouch adjacent to the anus, a type of low anorectal malformation (ARM) unlikely to have a rectourinary fistula that can be repaired as a neonate without colostomy.*





(Left) *Lateral radiograph of the same patient was taken to calculate the sacral ratio (A/B). The axis is drawn along the sacrum, then lines are drawn perpendicular to the sacral axis at the iliac crest, greater sciatic notch, & tip of the ossified sacrum/coccyx.* **(Right)** *Spinal US in a 2 day old with a history of imperforate anus who had meconium per the urethra (clinical evidence of a rectourinary fistula) shows a low conus at the S1 level with an echogenic terminus, which may be a filar lipoma.*





TERMINOLOGY

Synonyms

- Imperforate anus

Definitions

- Anorectal malformation (ARM): Spectrum of congenital anomalies related to abnormal development of anorectal canal
- Diagnosis of ARM made by clinical exam
- Imaging used to identify associated anomalies, plan operations, & evaluate postoperative complications
- Evolution of ARM classification: Traditionally as high, intermediate, or low (relative to pubococcygeal line)
  - Current (Peña or Krickenbeck): By specific malformation with therapeutic & prognostic implications
- Cloaca: Subtype of ARM in girls; rectum, vagina, & bladder converge into common channel → single perineal orifice
- Definitive repair as neonate vs. divided colostomy with delayed distal colostogram & definitive repair
- Posterior sagittal anorectoplasty (PSARP): Definitive surgical reconstruction for most ARMs

IMAGING

General Features

- Best diagnostic clue
  - Multiple dilated bowel loops in newborn without normal external anus
- Morphology
  - Rectal opening outside anal sphincter on perineum vs. no rectal opening on perineum

Radiographic Findings

- Radiography
  - Multiple dilated bowel loops of distal obstruction in newborn
  - Ca^{2+} → enteroliths (intraluminal Ca^{2+})
    - Mixing of urine & meconium via rectourinary fistula
    - Meconium peritonitis rare in ARM; only seen if ARM has no fistula
  - Prone cross-table lateral (XTL) view of abdomen > 24 hours old: Evaluates pouch to anus distance
    - In ~ 5% of ARM, fistula **not** clinically apparent to help direct operative management
    - Rectal pouch close to anal region → neonatal repair; if not → diverting colostomy/delayed repair
  - ± hemisacrum/scimitar sacrum (different from truncated sacrum): Defect represents presacral mass
    - Teratoma, anterior meningocele, mixed lesion
  - Sacral ratio predicts potential for fecal continence; > 0.6 = normal; < 0.4 = ↓ probability of continence
  - ± esophageal atresia (EA), tracheoesophageal fistula (TEF), congenital heart disease
  - ± vertebral or radial ray anomalies

Fluoroscopic Findings

- Voiding cystourethrogram
  - Normal; vesicoureteral reflux (VUR); ± bladder thickening, trabecula, neurogenic bladder; fistula from bladder neck or urethra → rectum in males, reflux from urethra to vagina &/or rectum in female cloaca
- Fistulagram
  - a.k.a. augmented pressure distal colostogram using Foley catheter
    - Water-soluble contrast injection of mucous fistula
  - To be performed several weeks to months after colostomy, prior to definitive repair
    - Must opacify rectal pouch & rectourinary fistula
  - Lateral view (superimposed femoral heads) showing
    - Distal colonic segment length, fistula to bladder or urethra in boys, common channel in girls
      - Most boys (95%) have rectourinary fistula (except Down syndrome: 95% have **no** fistula)
      - **Adequate distention needed to see fistula**
      - If bladder fills, fill until voiding; image distal colonic segment, bladder, urethra
    - Document relationship of rectal pouch to sacrum & anal sphincter (marked by BB)
    - Is presacral mass displacing rectal wall?
- ± cloacagram in females (usually performed prior to definitive repair)
  - Contrast injection of distal colostomy
  - Contrast into bladder &/or vagina via cloaca catheter
  - 3D imaging by rotational fluoroscopy, CT, or MR

MR Findings

- Prenatal: Absent T1-bright meconium posterior/inferior to bladder > 20-weeks gestation
- Postnatal
  - Prerepair pelvic MR: Anal sphincter + pelvic floor musculature, rectal pouch, presacral mass, other GU anomalies
  - Intraoperative pelvic MR: Guide rectum through sphincter → novel approach
  - Postrepair pelvic MR (poor functional outcome): Look for misplaced rectum, entrapped fat, posterior urethral diverticulum
- Spine MR: Tethered cord, spinal dysraphism, sacral agenesis/dysgenesis

Ultrasonographic Findings

- Grayscale ultrasound
  - Perineal US: Assess distance of rectal pouch → perineum; not reliable
  - Assess neonate for renal + spine anomalies
  - Assess for hydrocolpos in newborn cloaca → if positive, urgent drainage required to relieve secondary bladder/ureteral obstruction

Imaging Recommendations

- Best imaging tool
  - Neonate: Clinical exam + AP & prone XTL radiographs; renal, spine, & pelvic US (especially pelvis in cloaca)
  - Infant after colostomy: Distal colostogram for operative planning

DIFFERENTIAL DIAGNOSIS

Neonatal Distal Bowel Obstruction (Normal Anus)

- Small left colon/meconium plug syndrome
- Hirschsprung disease
- Meconium ileus
- Jejunoileal atresia

Abdominal Calcifications in Newborn

- Intraluminal Ca^{2+}: ARM, total colonic Hirschsprung disease, multiple intestinal atresia
- Meconium peritonitis: Peritoneal Ca^{2+} due to in utero bowel perforation
- Visceral: TORCH, prior hemorrhage
- Mass: Teratoma, neuroblastoma, congenital hemangioma
- Vascular: Prior thrombosis

PATHOLOGY

General Features

- Etiology
  - Abnormal separation of GU system from hindgut
- Genetics
  - No responsible gene identified; ↑ risk if sibling has ARM
  - Syndromic cases
    - Trisomy 21 (Down), Currarino (ARM, sacral deformity, presacral mass), VACTERL, OEIS, caudal regression, Pallister-Hall, Townes-Brocks
- Associated abnormalities
  - GU (50% of ARMs): Solitary/horseshoe kidney, VUR, bicornuate/didelphys uterus, hydrocolpos (50% of cloacas), hemivaginas
  - Spine: Tethered cord (25% of ARM, ↑ with more complex ARM), myelomeningocele, sacral anomalies (most affected bony structure)
  - Cardiac: Tetralogy of Fallot, ventricular septal defect
  - GI: EA ± TEF, duodenal atresia, Hirschsprung disease

Gross Pathologic & Surgical Features

- Imperforate anus
  - Rectoperineal fistula (male or female) → **no** colostomy → PSARP as newborn
    - Rectum opens to small stenotic orifice anterior to normal anal sphincter complex; normal sacrum, muscles, sphincter
  - Rectovestibular fistula: 25% of female ARMs → **no** colostomy → PSARP as newborn
    - Rectum opens between hymen & perineal skin; most patients have good sacrum, sphincter
  - Rectourethral fistula: 50% of male ARMs → divided colostomy
    - Fistula to bulbar urethra: Normal/near normal sacrum, muscles → delayed PSARP
    - Fistula to prostatic urethra: ± deficient sacrum, muscles → delayed PSARP ± lap procedure
  - Rectobladder neck fistula: 10% of male ARMs → divided colostomy → PSARP + lap procedure
    - Only true supralevator malformation; poor sacrum, sphincter, flat bottom (poorly developed gluteal crease)
  - No fistula: 5% of all ARM patients (male or female) with normal chromosomes → divided colostomy → PSARP
    - Rectum usually ≤ 2-cm deep to perineum (if rectum very close to skin on XTL → ± PSARP as neonate)
    - Usually good sacrum, sphincter
    - Frequent in Down syndrome (95%)
  - Rectal atresia or stenosis: 1% of ARMs (male or female) → different operative technique → colostomy
    - Atresia between normal anus & rectum; normal sacrum & sphincter
- Cloaca: Female with single perineal orifice (genitalia frequently ambiguous)
  - Common channel drains rectum, vagina, urethra
  - Hydrocolpos in up to 50%
    - May cause urinary obstruction at distal ureters/bladder → requires neonatal drainage
    - Undrained hydrocolpos → renal insufficiency, vaginal perforation, sepsis, even death
  - Common channel length
    - < 3 cm: Divided colostomy → delayed PSARP, good prognosis
    - > 3 cm: Divided colostomy → delayed PSARP + lap procedure, poorer prognosis (more complex malformation, more associated anomalies)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Absent/misplaced rectal opening, abdominal distention
- Other signs/symptoms
  - Meconium per vestibule, urethra, or abnormal perineal opening; flat bottom, poorly developed gluteal crease

Treatment

- Goals: Maximize continence of feces & urine, sexual function
- Most ARMs require diverting colostomy within days of birth, PSARP for definitive repair months later
  - + laparotomy/laparoscopy for bladder neck & some prostatic fistulas + long common channel cloaca
  - Subsequent proactive constipation prevention
- Prognosis dependent on type of malformation, sacral ratio, spine anomalies, meticulous surgical technique
  - 75% of ARMs will have voluntary bowel movements
  - 25% of ARM patients suffer incontinence, receive bowel management enemas daily to keep clean

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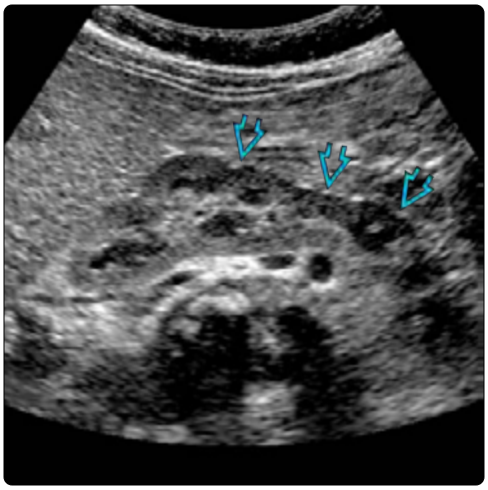
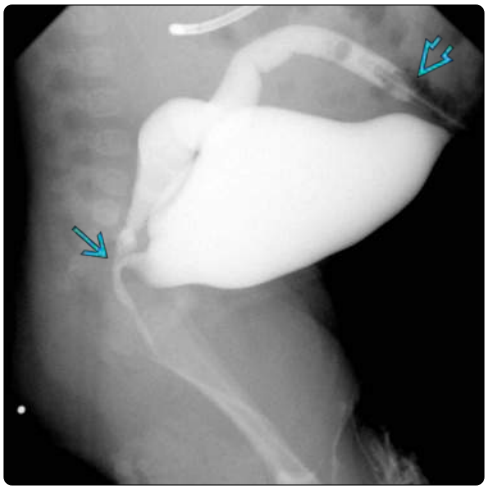
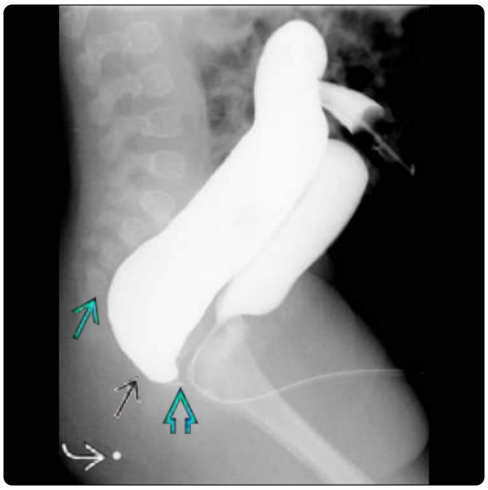
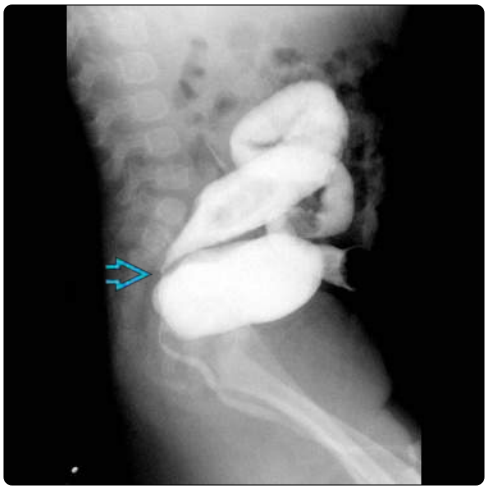


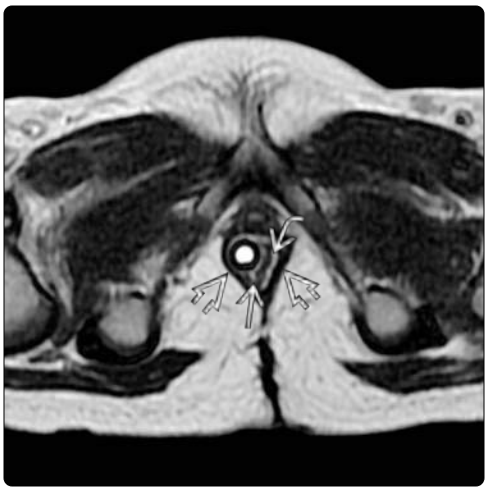


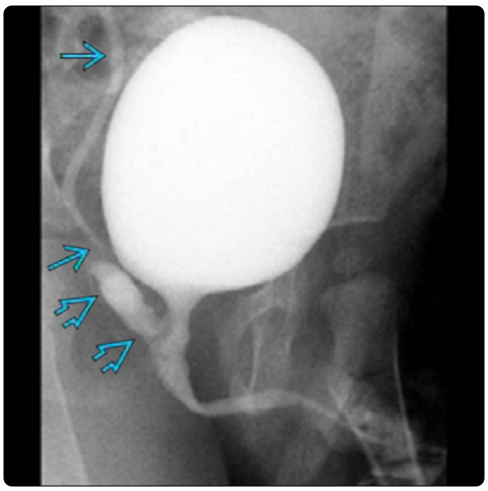
(Left) *Transverse US in a 3-day-old boy with imperforate anus shows renal tissue crossing anterior to the spine & great vessels, consistent with a horseshoe kidney.* **(Right)** *Colostogram in 3-month-old boy with an ARM shows a Foley balloon inflated in the distal colonic segment. Contrast injection shows a rectoprostatic urethral fistula located anterior to the spine, which will require laparoscopy as well as a posterior sagittal anorectoplasty (PSARP).*





(Left) *Colostogram in a 3-month-old boy with an ARM shows a rectobulbar urethral fistula with a rectal pouch well below the tip of spine. The distal colonic segment is adequate in length to bring the pouch to the anal region by a PSARP.* **(Right)** *Colostogram in a 2 month old with an ARM & poorly developed gluteal crease shows a rectobladder neck fistula. The rectal pouch lies above the spine tip at S3 & will only be reached by a laparoscopy or open abdominal approach prior to PSARP.*





(Left) *Axial T2 MR in a repaired ARM patient shows asymmetric positioning of the rectum (containing a T2 bright catheter) within the asymmetric sphincter complex. Fat is seen between the left rectal wall & sphincter muscle.* **(Right)** *Oblique voiding cystourethrogram of a 9-month-old boy after PSARP for a prostatic fistula ARM shows a posterior urethral diverticulum (or remnant of the original fistula), which can be a source of infection, stone formation, & dysuria. Note the right vesicoureteral reflux.*

Meconium Peritonitis

KEY FACTS

TERMINOLOGY

- Meconium peritonitis (MP): Chemical peritonitis from in utero bowel perforation with leakage of sterile meconium & digestive enzymes
- Meconium cyst: Meconium + fluid contained by fibrous membranes in setting of MP
- Meconium pseudocyst: Meconium + fluid contained by membranes partly derived from muscular layer of intestine

IMAGING

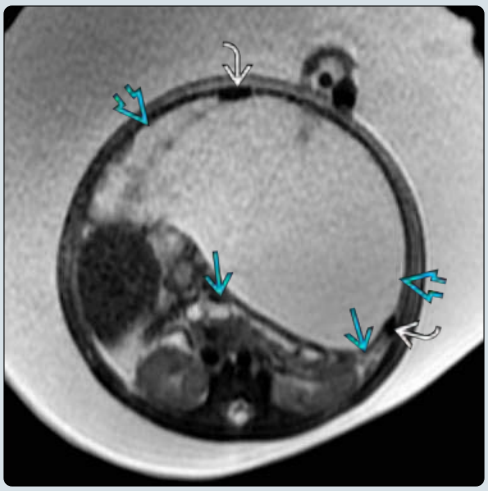
- Best diagnostic clue
  - Newborn abdominal radiograph showing linear, curvilinear, or punctate Ca^{2+} on peritoneal surfaces &/or cyst/mass causing bowel displacement
  - Pre- or postnatal US or prenatal MR showing peritoneal Ca^{2+}, complex ascites, dilated bowel, &/or cyst/pseudocyst
- Fluoroscopic contrast study can limit differential diagnosis

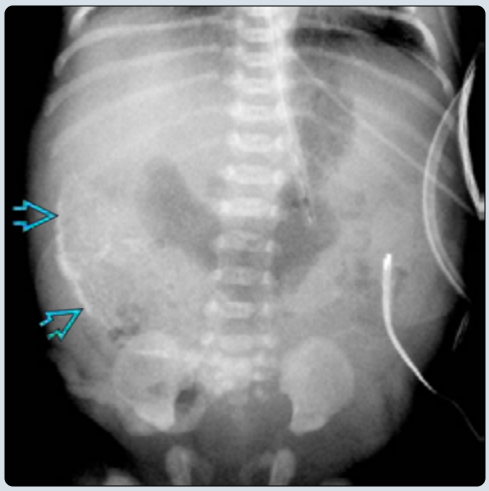
TOP DIFFERENTIAL DIAGNOSES

- Ileal atresia
- Meconium ileus
- Midgut volvulus
  - Much less common cause of MP than distal obstructions

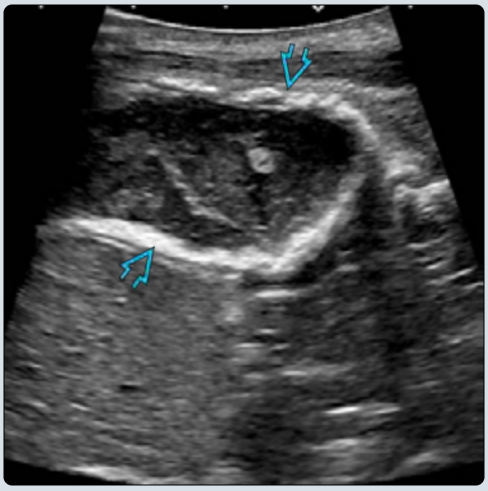
CLINICAL ISSUES

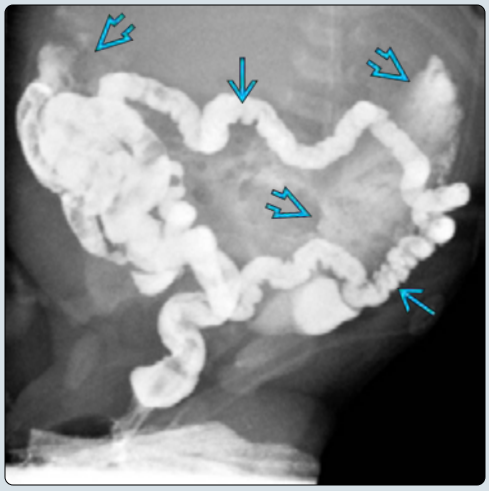
- Prenatal diagnosis key to direct perinatal therapy
- Neonatal surgery required in 50-90%
- No treatment required in asymptomatic patients with normal bowel function (i.e., healed perforation in utero)
- 2-20% mortality even in modern era





(Left) *Axial SSFSE T2 fetal MR shows a large cystic lesion of the abdomen, which contains some low-signal debris, possibly meconium, & displaces decompressed bowel. Punctate low signal foci suggest calcifications raising the possibility of meconium peritonitis (MP).* **(Right)** *Newborn radiograph performed in the same patient delivered at 31-weeks gestation shows peritoneal or cyst wall calcification, consistent with MP. No free intraperitoneal air is seen.*





(Left) *Abdominal US in the same patient shows a complex fluid collection with a thick, echogenic, calcified wall, correlating with the preceding radiograph. The bowel was decompressed (not shown).* **(Right)** *Supine WSCE was performed in the same patient to assess continuity of bowel in hopes of conservative initial management. Contrast first opacifies a microcolon & small bowel before leaking into the meconium pseudocyst. Three unexplained sites of perforation were found in the OR, but no atresia or thick meconium was seen.*

TERMINOLOGY

Definitions

- Meconium peritonitis (MP): Chemical peritonitis resulting from in utero bowel perforation that leaks sterile meconium & digestive enzymes into peritoneal cavity
- Meconium cyst: Meconium + fluid contained by fibrous membranes in setting of MP
- Meconium pseudocyst: Meconium + fluid contained by membranes partly derived from muscular layer of intestine

IMAGING

General Features

- Best diagnostic clue
  - Newborn abdominal radiograph showing linear, curvilinear, or punctate Ca^{2+} along peritoneal surfaces &/or cyst wall
    - Ca^{2+} begins within hours of meconium spillage
  - Pre- or postnatal US with peritoneal Ca^{2+}, complex ascites, dilated bowel, &/or cyst

Radiographic Findings

- Radiography
  - Abdominal soft tissue mass (cyst/pseudocyst)
    - May displace bowel loops
    - May have rim or internal Ca^{2+}
    - May cause gasless abdomen
  - Amorphous, linear, or curvilinear Ca^{2+} along peritoneal surfaces
    - Parietal layer lining abdominal cavity
    - Visceral layer along organ surfaces (liver, etc.)
    - Calcified meconium in bowel loops (enteroliths) may be difficult to differentiate from MP on radiographs
      - Differentials: Total colonic aganglionosis, anorectal malformation, multiple bowel atresias
  - ± centralized bowel loops due to ascites
  - Multiple dilated bowel loops of bowel obstruction; 2 possible etiologies
    - In utero obstruction → perforation
    - Spontaneously sealed in utero perforation ± adhesions → obstruction
  - Free intraperitoneal air uncommon by most reports
    - Site of perforation often sealed in utero
  - Ca^{2+} in scrotum (meconium orchitis)
    - Due to intraperitoneal Ca^{2+} passing through patent processus vaginalis

Ultrasonographic Findings

- Grayscale ultrasound
  - Pre- or postnatal
    - Hyperechoic punctate, linear echogenic foci along abdominal surfaces ± shadowing
      - Prenatal US more sensitive than postnatal radiographs
    - Ascites: Complex fluid (from meconium) very suggestive
    - Dilated bowel
    - Cystic mass ± wall nodularity, Ca^{2+}, debris
      - Mass effect may mimic abdominal tumor
    - Ca^{2+}, fluid (hydrocele) in scrotum

MR Findings

- Prenatal MR: Similar findings to prenatal US
  - Fetal ascites or cystic collection
    - Complex ± bright T1 signal debris (meconium) vs. simple fluid
  - Dilated bowel loops
  - ± polyhydramnios

Fluoroscopic Findings

- Upper GI
  - Can be normal or show variety of anomalies
  - Malrotation ± midgut volvulus
  - Duodenal jejunal junction displaced by adjacent dilated bowel loops or cyst/pseudocyst despite normal rotation
    - ↑ laxity of ligament (actually muscle) of Treitz in newborn
  - Small bowel obstruction (SBO) frequent
- Contrast enema
  - Microcolon (small colon)
    - Strongly suggests distal SBO
    - Most commonly due to
      - Ileal atresia: Contrast stops at blind-ending TI
      - Meconium ileus: Numerous filling defects in TI
  - Normal colon
    - Possible proximal or late SBO
  - Colonic obstruction uncommon (i.e., MP uncommonly seen with Hirschsprung disease, colonic atresia)

Imaging Recommendations

- Best imaging tool
  - Prenatal &/or postnatal sonography
  - Neonatal abdominal radiographs
- Protocol advice
  - Fluoroscopic contrast study can limit differential diagnosis
    - In face of distal obstruction, water-soluble contrast enema most helpful
      - Usually meconium Ileus vs. ileal atresia

DIFFERENTIAL DIAGNOSIS

Neonatal Distal Bowel Obstruction (± MP)

- Ileal atresia
- Meconium ileus
- Hirschsprung disease
- Small left colon/meconium plug syndrome
- Anorectal malformation
- Malrotation
  - Midgut volvulus
    - Much less common cause of MP than distal obstructions
    - Ischemic ileus may mimic distal obstruction due to multiple dilated loops
  - Congenital bands

Neonatal Abdominal Calcifications

- Enteroliths
  - Anorectal malformation
  - Multiple intestinal atresias
  - Total colonic Hirschsprung disease
- Liver parenchyma

- TORCH infections
- Vascular Ca^{2+}
- Mass
  - Sacrococcygeal teratoma
  - Neuroblastoma
  - Hepatoblastoma
  - Congenital hemangioma

PATHOLOGY

General Features

- Etiology
  - In utero bowel perforation most commonly due to
    - Meconium ileus
      - 8-40% have meconium ileus with cystic fibrosis
    - Jejunal/ileal atresia or stenosis
    - Volvulus
      - Malrotation with midgut volvulus
      - Other segmental volvulus
  - In utero bowel perforation less commonly due to
    - Intussusception, vascular accident, congenital bands, internal hernia, Hirschsprung disease, colonic atresia, cloaca
  - Cause unknown in 25-50% of cases
  - Meconium & digestive enzymes leak into peritoneal cavity
  - Intense chemical peritonitis + inflammatory response
  - Granulomatous response + Ca^{2+} within hours-days

Gross Pathologic & Surgical Features

- Subtypes at surgery
  - Fibroadhesive: Sealed off perforation → bowel obstruction
  - Cystic or pseudocystic (fibrous vs. partially muscular/bowel wall): Perforation does not seal off → inflammation → cystic cavity
  - Generalized: Late in utero perforation → more fibrinous than fibrous

Microscopic Features

- Peritoneal squames, bile pigment, fibrosis, chronic inflammation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prenatal: Polyhydramnios, bowel obstruction, Ca^{2+}, abdominal cyst, ascites
  - Postnatal: Abdominal distention, mass, failure to pass meconium, bilious emesis, respiratory distress

Demographics

- Age
  - 2nd-trimester gestational age through birth
- Epidemiology
  - 1:35,000 live births

Natural History & Prognosis

- Majority of cases do not resolve spontaneously in utero
- Prenatal US prediction for neonatal surgery
  - Isolated Ca^{2+}: No surgery required
  - Ca^{2+} & 1 additional finding (ascites or pseudocyst or bowel dilation): > 50% require surgery
  - Ca^{2+} & 2-3 additional findings: 80-100% require surgery
- No relation between prenatal diagnosis or type of MP & outcome
- Massive ascites or cyst may lead to hydrops, pulmonary hypoplasia in utero
  - Fetal paracentesis to ↓ fluid
  - Urinary trypsin inhibitor may ↓ inflammation
- 2-20% mortality in modern era
- Predicting factors for morbidity & mortality
  - Postnatal circulation deficiency, serum CRP, persistent ascites, persistent pulmonary hypertension

Treatment

- None in asymptomatic patients with normal bowel function (i.e., healed perforation)
- Neonatal surgery required in 50-90%
  - Abdominal drainage
  - Relieve bowel obstruction
  - Resection of perforated &/or necrotic bowel
  - Anastomosis vs. temporizing bowel diversion with delayed anastomosis
- Cyst vs. pseudocyst may be important distinction to direct surgical treatment in recent series
  - Pseudocyst: Primary bowel anastomosis
  - Cyst: Bowel diversion with delayed anastomosis
- Complications: Sepsis, adhesions, short gut syndrome

DIAGNOSTIC CHECKLIST

Consider

- Most common causes of in utero bowel perforation
  - Complicated meconium ileus (cystic fibrosis)
  - Intestinal atresia

Image Interpretation Pearls

- Pre- or postnatal sonography: Polyhydramnios, abdominal cyst, ascites, peritoneal Ca^{2+}, dilated bowel
- Postnatal radiography: Ca^{2+} of peritoneal lining diagnostic
  - Evaluate if Ca^{2+} could be in other locations
    - Intestinal lumen (enteroliths)
    - Hepatic, splenic parenchyma
    - Intraabdominal neoplasm

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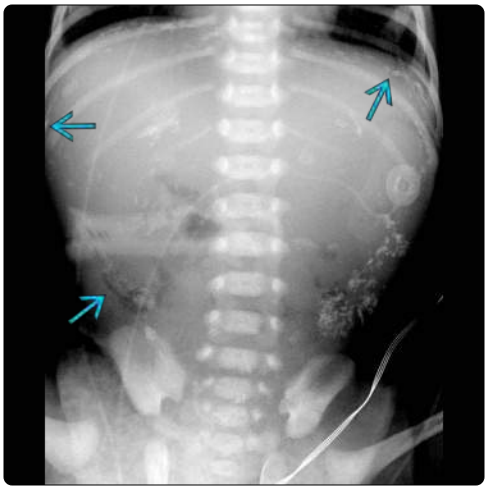
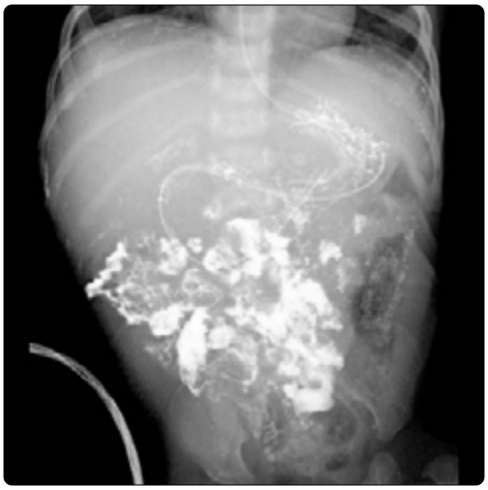
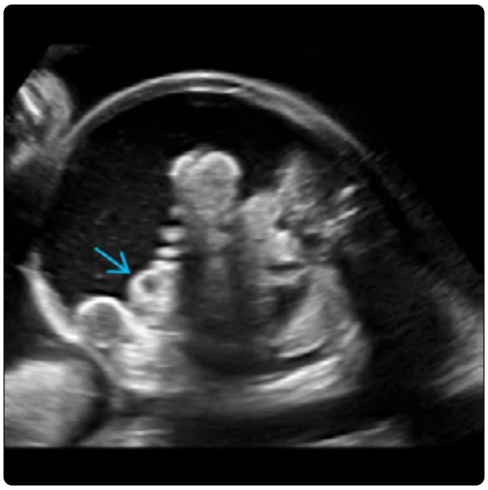
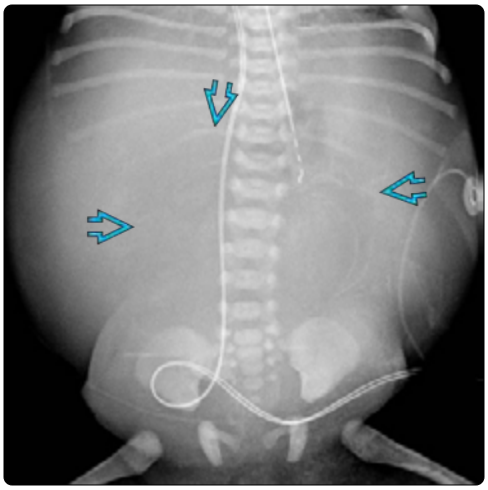


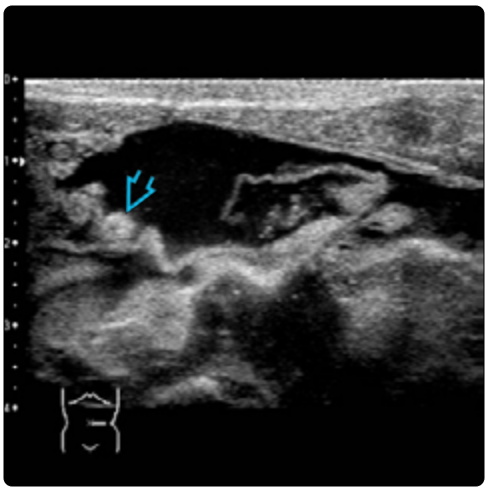


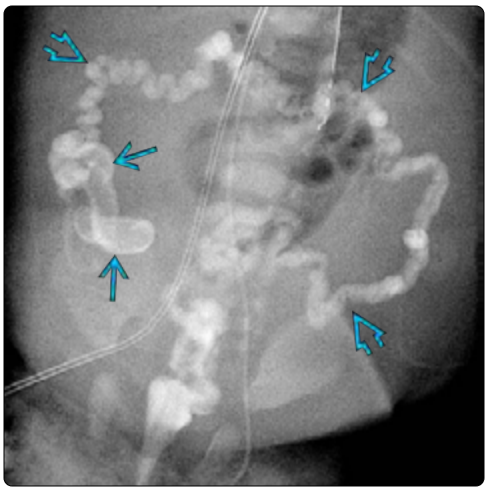
(Left) *AP radiograph in a 1-day-old girl with prenatal concern for MP shows amorphous calcification on peritoneal surfaces throughout the abdomen, which is consistent with MP. A gasless abdomen is suspicious for a meconium cyst, although US showed normal bowel & no cyst.* **(Right)** *Fluoroscopic upper GI-small bowel follow-through in the same patient showed continuous bowel & no obstruction. MP with no cystic mass, ascites, or obstruction equals a healed in utero perforation.*





(Left) *Prenatal US shows complex ascites & somewhat thickened echogenic bowel loops, suggestive of MP. Echogenic bowel is nonspecific but can be seen in cystic fibrosis.* **(Right)** *AP radiograph in the same patient at delivery shows a gasless abdomen with curvilinear & amorphous subtle calcification in the abdomen, suggestive of MP. The main diagnostic considerations include a bowel atresia or complicated meconium ileus, which is usually a manifestation of cystic fibrosis (CF).*





(Left) *US in the same patient confirms the presence of complex ascites suggestive of MP (as well as echogenic bowel, as seen on the prenatal US).* **(Right)** *Supine WSCE in the same patient shows a microcolon devoid of significant meconium. The normal caliber terminal ileum is filled with meconium, consistent with meconium ileus, a common cause of MP. Genetic testing confirmed CF in this neonate.*

Necrotizing Enterocolitis

KEY FACTS

TERMINOLOGY

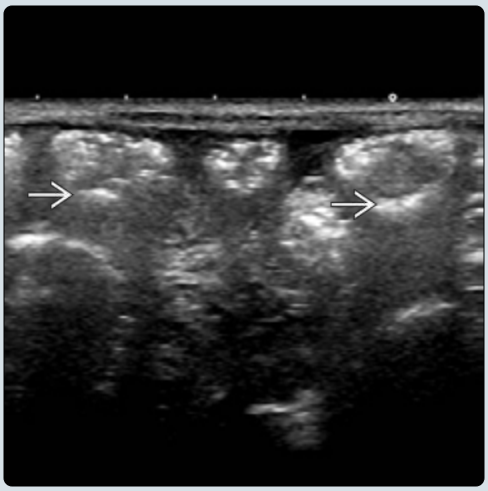
- Necrotizing enterocolitis (NEC): Life-threatening condition of neonatal GI tract characterized by inflammation, ischemia, & translocation of bacteria into bowel wall

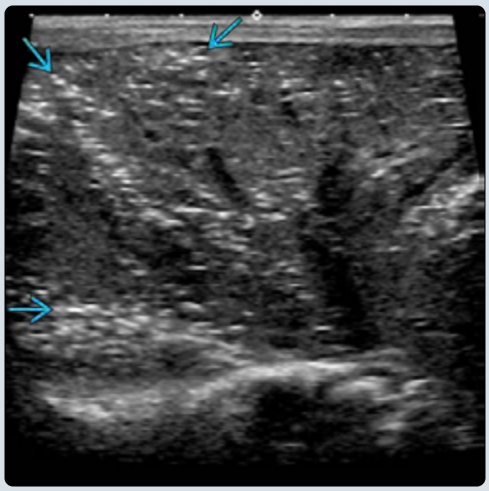
IMAGING

- Diagnosis based on clinical & imaging findings
- Mainstay of imaging for suspected NEC: Radiography
  - Findings range from nonspecific (paucity of bowel gas) to suggestive (thickened, dilated bowel loops) to diagnostic [pneumatosis, portal venous gas (PVG), & free peritoneal gas]
  - Duke Abdominal Assessment Scale for radiographs
    - Standard lexicon for reporting NEC findings
    - Strong intraobserver & interobserver agreement
    - ↑ scores correlate with need for surgery
- Ultrasound excellent adjunct
  - Radiographically occult necrosis requiring surgery suggested by absent vascularity + ↓ peristalsis
  - Additional findings include focal fluid collections, echogenic ascites, ↑ bowel wall echogenicity, intramural gas, bowel wall thickening or thinning, PVG, & free air
- Contrast enema not used acutely; useful to localize strictures after treatment of acute episode

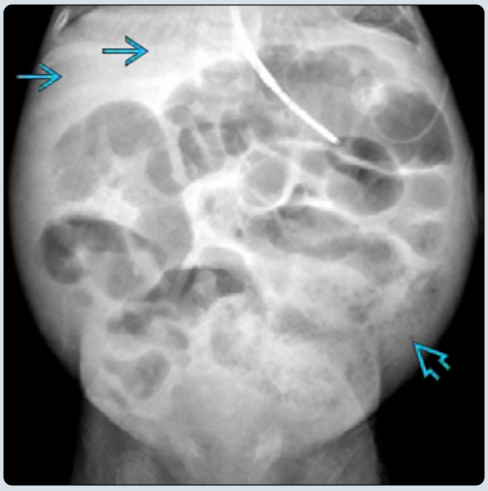
CLINICAL ISSUES

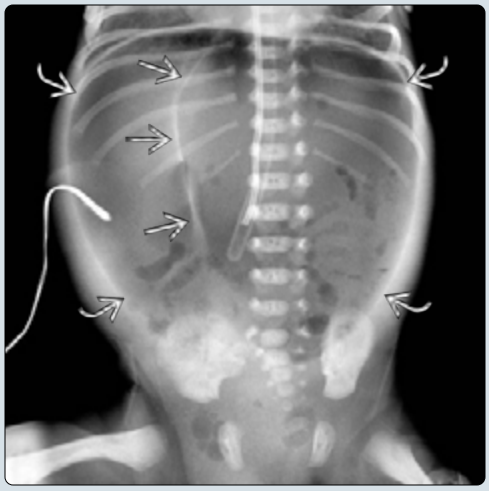
- Most common in very low birth weight (< 1,500 g) premature infants 2-3 weeks after delivery
  - 10% in term infants (usually with underlying diseases)
- Typical history: Feeding intolerance with emesis, ↑ gastric residuals, bloody stools
- Other frequent clinical findings include abdominal distention &/or discoloration, apnea & bradycardia, lethargy, temperature instability
- Treatment: IV nutrition + antibiotics ± surgery
- Overall mortality 10-50%
  - Death secondary to sepsis from bowel perforation
- Delayed bowel strictures in 10-20% of survivors





(Left) *Longitudinal US of the abdominal left lower quadrant (LLQ) in a former 34-week premature infant shows numerous bowel loops with circumferential nodular echogenicity to their walls. Many of the echogenic foci demonstrate "dirty shadowing" posteriorly, typical of pneumatosis.* **(Right)** *Transverse US of the liver in the same patient shows numerous branching linear & nodular echogenic foci in the parenchyma, particularly peripherally, typical of portal venous gas (PVG).*





(Left) *Subsequent supine AP radiograph (same patient) shows bubbly lucencies of pneumatosis in the LLQ. Branching foci of PVG are seen in the liver. This patient was managed conservatively for medical NEC.* **(Right)** *Abdominal radiograph in a premature infant with NEC shows a large amount of pneumoperitoneum causing abnormal lucency throughout the abdomen. The falciform ligament is outlined by air; this appearance resembles the laces of an American-type football (the football sign).*

TERMINOLOGY

Definitions

- Necrotizing enterocolitis (NEC): Poorly understood but life-threatening condition of neonatal GI tract characterized by inflammation, ischemia, & translocation of bacteria into bowel wall
- Very low birth weight (VLBW) infant: < 1,500 g
- Extremely low birth weight (ELBW) infant: < 1,000 g

IMAGING

General Features

- Best diagnostic clue
  - Dilated bowel loops with pneumatosis (intramural bowel gas), portal venous gas (PVG), & free intraperitoneal air
- Location
  - Affected bowel most commonly in right lower quadrant
    - Ascending colon, terminal ileum
  - May occur anywhere from stomach to rectum

Radiographic Findings

- Nonspecific findings
  - Paucity of bowel gas
  - Loss of normal mosaic pattern of polygon-shaped bowel loops throughout abdomen
- Suggestive findings
  - Asymmetric bowel dilation
  - Fixed, "unfolded" bowel loops on serial radiographs
    - May be normal caliber or dilated
  - Separation of bowel loops
    - Due to bowel wall thickening or intervening collapsed or fluid-filled bowel or free fluid
- Definitive findings
  - Pneumatosis (50-75% of patients)
    - Bubbly (submucosal) or curvilinear (subserosal) lucencies
    - Mimics formed stool (not typically seen in unfed infants)
  - PVG
    - Branching lucencies over liver
    - Greater peripheral extension than biliary gas (with biliary gas being uncommon in newborns)
  - Free intraperitoneal air
    - Overall ↑ upper abdominal lucency on supine radiograph with liver appearing less opaque than heart
    - Football sign: Lucent, distended abdomen with gas outlining vertical falciform ligament on supine radiograph
    - Cupola sign: Gas under midline diaphragm on supine radiograph (with well-defined superior, but not inferior, borders)
    - Rigler sign: Gas outlining both sides of bowel wall
    - Crescentic lucency overlying right hepatic margin on left lateral decubitus view or anterior hepatic margin on cross-table lateral view
    - Triangles of lucency anteriorly on cross-table lateral view or peripherally on supine AP view

Ultrasonographic Findings

- Grayscale ultrasound
  - Focal fluid collections, echogenic ascites, aperistaltic bowel with thickening or thinning, & ↑ echogenicity of bowel wall all suggest NEC
  - Pneumatosis: Nodular echogenicities around entire circumference of bowel wall ± "dirty shadowing" posteriorly
  - PVG: Branching linear & punctate echogenicities in liver periphery; may see echogenic foci coursing through portal veins real time
  - Free intraperitoneal air: Linear echogenicities with "dirty shadowing" immediately deep to peritoneal surface
- Color Doppler
  - ↓ vascularity indicates necrosis/perforation
  - ↑ vascularity may be seen in earlier stages
  - ↑ peak systolic velocity & resistive index in superior mesenteric artery

Fluoroscopic Findings

- Acute: Enema contraindicated in acute NEC
- Chronic: Strictures create obstruction in "older" NICU babies; causative NEC episode not always clinically apparent
  - Single or multiple strictures found 4-8 weeks after NEC
  - Radiologically discovered strictures usually left-sided while most surgically discovered strictures found in right colon (sometimes even small bowel)

Imaging Recommendations

- Best imaging tool
  - Serial radiography: Fixed, dilated loops predictive of bowel necrosis
  - Sonography up to 100% sensitive, 95% specific for bowel necrosis
    - Primarily by absent vascularity + ↓ peristalsis
    - Other helpful findings: Bowel wall thickening, parietal peritoneal thickening, echogenic free fluid, & focal fluid collections

DIFFERENTIAL DIAGNOSIS

Newborn Bowel Obstruction

- Congenital bowel obstructions (e.g., atresias, meconium ileus) present within first 1-3 days after delivery, often with "bilious emesis" or "failure to pass stool"

Newborn Bowel Perforation

- Idiopathic/spontaneous most common

Enterocolitis of Term Infants

- Milk allergy, cyanotic heart disease, maternal cocaine use, Hirschsprung disease (especially total colonic)

Nonspecific Gaseous Distention of Bowel

- Continuous positive airway pressure
- Bag ventilation at intubation

Hypertrophic Pyloric Stenosis

- Rarely associated with benign gastric pneumatosis
- Usually healthy term infants presenting with projectile vomiting at 2-12 weeks of life

Benign Pneumatosis in Older Children

- Often with complex medical history, including bone marrow transplant & steroid therapy
- Predominately colonic; may cause free air

PATHOLOGY

General Features

- Etiology
  - Different mechanisms in different newborn groups
    - Very premature: Dysregulated inflammatory response to stasis of feeds, incomplete or abnormal GI microbial colonization, & immunologic immaturity
    - Term & late preterm: Likely from hypoxia-ischemia
  - Multifactorial etiology in all patients
- Associated abnormalities
  - Lung disease of prematurity
  - Congenital heart disease
  - Intracranial hemorrhage, periventricular leukomalacia

Staging, Grading, & Classification

- Duke Abdominal Assessment Scale: Standardized lexicon for reporting findings in newborns with suspected NEC
  - High intra- & interobserver agreement
  - Higher scores strongly correlate with disease severity/need for surgical intervention
    - 0: Normal gas pattern
    - 1: Mild bowel distention
    - 2: Moderate distention (or normal with nonspecific bubbly lucencies)
    - 3: Focal moderate distention of bowel loops
    - 4: Separation or focal thickening of bowel loops
    - 5: Featureless or multiple separated bowel loops
    - 6: Possible pneumatosis with other abnormal findings
    - 7: Fixed or persistent dilation of bowel loops
    - 8: Pneumatosis (highly probable or definite)
    - 9: PVG
    - 10: Pneumoperitoneum

Gross Pathologic & Surgical Features

- Dilated, gray, hemorrhagic, friable bowel

Microscopic Features

- Mucosal coagulation, ulceration, submucosal hemorrhage, submucosal or subserosal gas (pneumatosis intestinalis), PVG, mesenteric gas
- Histopathologic features include coagulative & hemorrhagic necrosis, inflammation, bacterial overgrowth

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Signs of feeding intolerance (emesis, ↑ gastric residuals, bloody stools)
  - Abdomen may become distended, discolored, erythematous, &/or shiny
  - Apnea & bradycardia, lethargy, temperature instability
- Other signs/symptoms
  - 1/3 have fulminant course with bowel perforation
  - 1/3 develop septic shock

Demographics

- Age
  - Premature VLBW or ELBW newborns have highest incidence (90% of cases)
    - Affects 3-13% < 1,500 g; rate in ELBW 3x that of VLBW
    - Onset of NEC peaks at 29-31 weeks postmenstrual age, especially 2-3 weeks after delivery
  - Term newborns make up 10% of cases
    - Earlier age onset of NEC (1-3 days after delivery)
    - Often have 1 or more risk factors
- Epidemiology
  - Overall incidence: 1 per 1,000 live births
    - Varies by nursery/intensive care unit
    - Outbreaks frequently follow epidemic pattern; no single causative infectious agent known

Natural History & Prognosis

- Overall mortality 10-50%
  - Death secondary to sepsis from bowel perforation
- Morbidity in survivors includes: Delayed bowel strictures (10-20%), short gut syndrome, neurodevelopmental delays

Treatment

- Prevention: Probiotics ↓ NEC incidence & mortality in VLBW infants
- When NEC suspected: IV nutrition + antibiotics
- Indications for surgery: Clinical ± radiologic findings
  - Free air considered absolute indication
  - Clinical deterioration, bowel necrosis

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- NEC can be associated with adhesions, so intraperitoneal gas may be loculated rather than "free"
  - Strongly consider left lateral decubitus &/or cross-table lateral images to look for intraperitoneal gas

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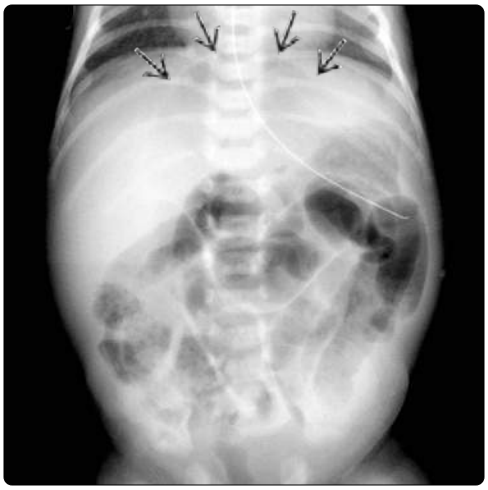
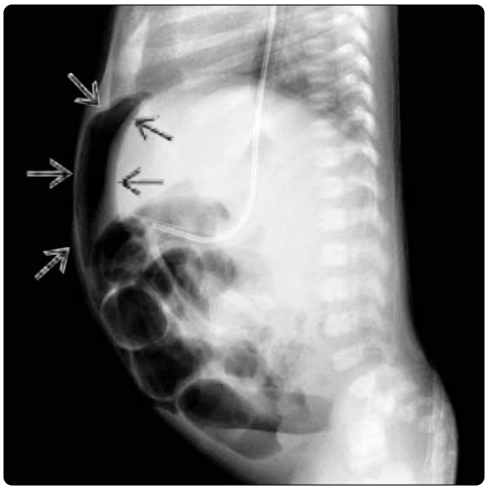
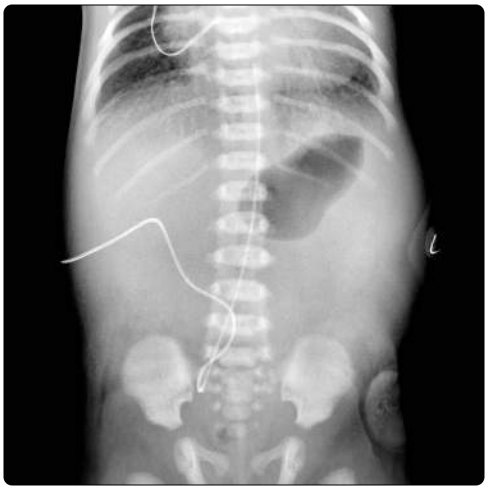
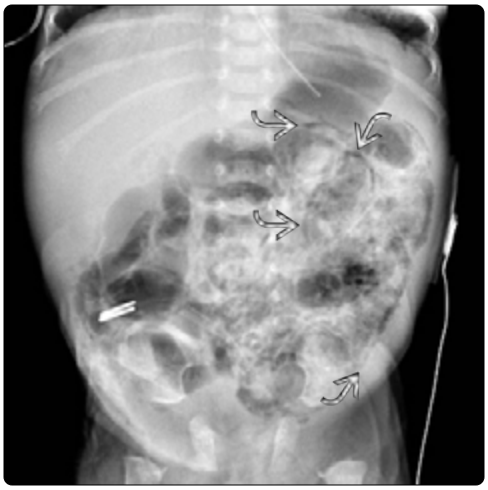


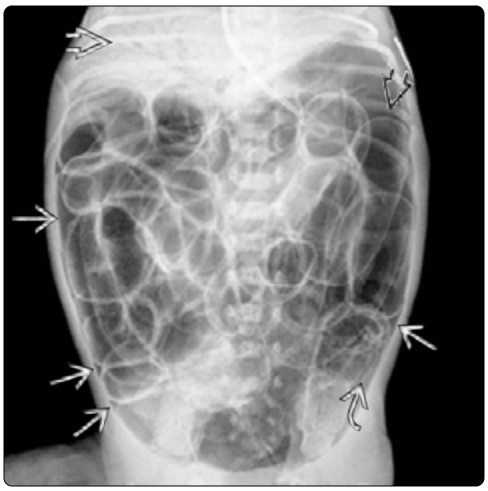


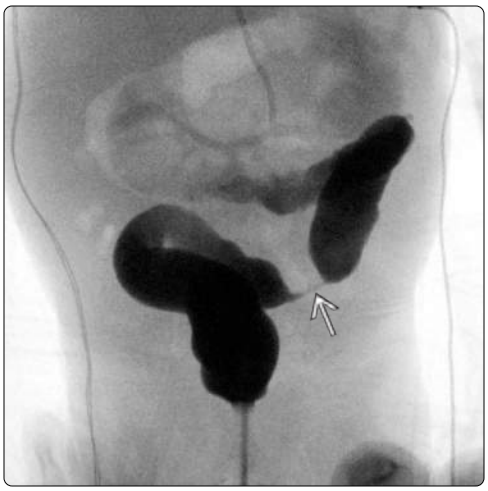
(Left) *Frontal radiograph of a premature infant with chorioamnionitis & NEC shows a cupola sign on supine imaging. The pneumoperitoneum is seen as a subtle, rounded lucency ⇨ below the midline diaphragm.* **(Right)** *A cross-table lateral radiograph was obtained in the same child & confirms a moderate amount of free intraperitoneal gas ⇨. Cross-table images deliver more radiation but can be a problem-solving tool if perforation is suspected. Note the persistently dilated bowel on all images.*





(Left) *Supine radiograph in a term infant with aortic coarctation, feeding intolerance, bloody stools, & spells of apnea/bradycardia shows a nearly gasless abdomen, a nonspecific finding of NEC.* **(Right)** *Supine radiograph shows a premature infant with NEC who has widespread pneumatosis intestinalis with curvilinear & bubbly lucencies seen in most of the intestinal walls ⇨, especially on the left.*





(Left) *Supine radiograph shows a dramatic case of NEC with PVG ⇨, pneumatosis ⇨, & free air causing numerous small, triangular lucencies ⇨. Also note a Rigler sign (with gas outlining both sides of the bowel wall ⇨), indicating pneumoperitoneum.* **(Right)** *An 8-week-old former premature infant with a history of NEC had signs of obstruction when feedings were reinstated. This frontal water-soluble contrast enema image reveals a short-segment, high-grade stenosis ⇨ of the distal colon from a NEC-associated stricture.*

KEY FACTS

TERMINOLOGY

- Gastroesophageal reflux (GER): Retrograde flow of gastric contents into esophagus
- GER disease (GERD): GER causes clinical symptoms or tissue damage

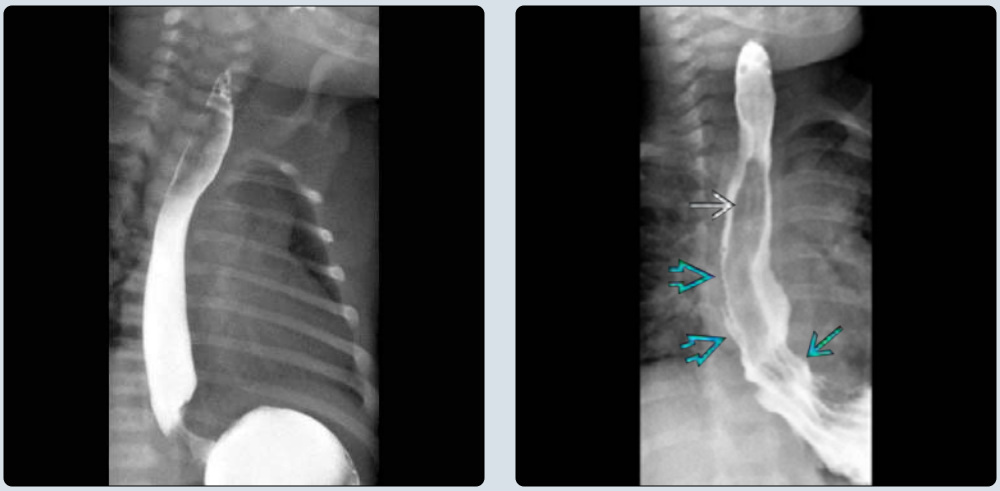
IMAGING

- Retrograde passage of gastric contents into esophagus
  - Can see on US, fluoroscopic upper GI, radionuclide scintigraphy
- Reflux esophagitis (GERD)
  - Best seen fluoroscopically
  - Esophageal dysmotility often earliest sign
  - Distal esophageal mucosal irregularity/thickening/stricture
  - Barrett esophagus (rare in children)
- Upper GI to exclude anatomic/functional abnormalities, not to identify GER
  - Esophageal dysmotility
  - Hiatal hernia
  - Gastric outlet/duodenal obstruction
  - Malrotation
- Radionuclide scintigraphy: Most sensitive imaging test to detect GER

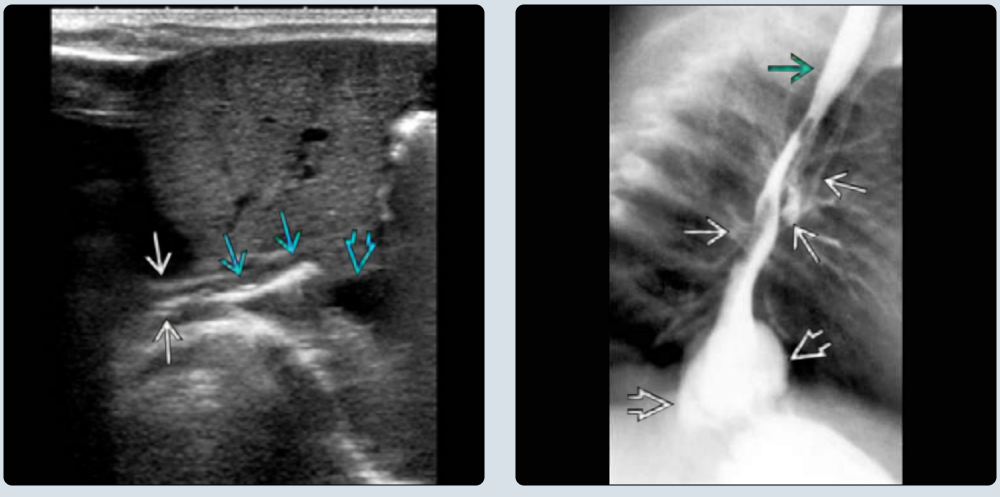
CLINICAL ISSUES

- Symptoms
  - Recurrent vomiting/regurgitation
  - Poor weight gain/failure to thrive
  - Excessive irritability
  - Respiratory symptoms (e.g., wheezing, cough)
- Epidemiology
  - Most infantile GER resolves by age 1-2 years
  - ~ 4% have persistent GER, 1% require surgical treatment
  - GER: 50% at 4 months, ↓ to 5-10% at 12 months
- Treatment
  - Nonsurgical: ~ 99% of refluxers
  - Surgical: Fundoplication

(Left) *Upper GI of a 3-day-old girl with severe regurgitation shows almost no peristalsis of an otherwise normal, completely distended esophagus. Hypomotility is the earliest finding in reflux esophagitis & gastroesophageal reflux disease (GERD).* **(Right)** *Upper GI in a 7 month old with worsening GER shows a mild hiatal hernia (gastric folds above diaphragm ⇨) with thickened folds of the distal esophagus ⇨, consistent with reflux esophagitis & dysmotility. Note the air ➡ within the esophagus.*



(Left) *Longitudinal US of the gastroesophageal junction in a 20 day old with recurrent nonbilious vomiting shows a column of echogenic air ⇨, which in real-time scanning was moving from the fluid-filled stomach ⇨ in a retrograde direction into the esophagus ➡.* **(Right)** *Lateral upper GI image in a patient with a hiatal hernia ➡ demonstrates significant reflux of contrast ⇨, which the patient aspirated ➡, requiring placement of an enteric suction tube into the esophagus to prevent further aspiration.*



TERMINOLOGY

Abbreviations

- Gastroesophageal reflux (GER), GER disease (GERD)

Definitions

- GER: Retrograde flow of gastric contents into esophagus
  - Physiologic phenomenon, especially in young infants
- GERD: GER causes clinical symptoms or tissue damage

IMAGING

General Features

- Best diagnostic clue
  - Retrograde passage of contrast across gastroesophageal junction (GEJ) into esophagus
- Morphology
  - Normal GEJ or associated hiatal hernia, usually sliding type

Radiographic Findings

- Radiography
  - Usually normal; rarely shows round retrocardiac lucency (hiatal hernia)

Fluoroscopic Findings

- Upper GI
  - Retrograde flow of contrast into esophagus
  - Reflux esophagitis (GERD)
    - Esophageal dysmotility often earliest sign
    - Distal esophageal mucosal irregularity/thickening/stricture
    - Barrett esophagus (rare in children)
  - Other potential findings with GER
    - Hiatal hernia, usually sliding type
    - Malrotation: Incidental in 4%
    - Gastric outlet/duodenal obstruction
      - Pyloric stenosis, gastric antral/duodenal web, etc.; delayed gastric emptying

Ultrasonographic Findings

- Retrograde flow of gastric contents at GEJ

Nuclear Medicine Findings

- Tc-99m sulfur colloid
  - Activity above GEJ during period of observation

Imaging Recommendations

- Best imaging tool
  - Tc-99m labeled sulfur colloid meal
  - Fluoroscopic upper GI evaluation for anatomic abnormality of GI tract
- Protocol advice
  - Upper GI to evaluate anatomy, not confirm reflux
    - Looking for GERD & GER causes: Evaluate swallowing & esophageal motility, no provocative maneuvers or prolonged monitoring
  - Radionuclide scintigraphy: Most sensitive imaging test for GER

DIFFERENTIAL DIAGNOSIS

Gastric/Duodenal Obstruction

- Pyloric stenosis, antral web, duodenal stenosis/web

Malrotation

- GER can be presenting symptom of malrotation without volvulus

Achalasia

- Childhood to adolescence

Esophagitis

- Many causes other than reflux

Esophageal Atresia (Repaired)

- Anastomotic stricture or GER not uncommon after repair

PATHOLOGY

General Features

- Etiology
  - Potentially many physiologic & anatomic factors
    - Suboptimal angle of His

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Infants: Recurrent vomiting/regurgitation, excessive irritability, feeding difficulty, poor weight gain/failure to thrive, sleep disturbances
  - Children: Poorly localized abdominal or chest pain, heartburn, recurrent vomiting/regurgitation, dysphagia, respiratory symptoms (e.g., wheezing, cough)

Natural History & Prognosis

- Most infantile GER resolves by age 1-2 years
- GER: 50% at 4 months, ↓ to 5-10% at 12 months
- ~ 4% have persistent GER, 1% require surgical treatment
- No treatment → pneumonia, asthma, laryngitis, otitis media, tooth decay
- Barrett esophagus, not usually seen in children

Treatment

- Nonsurgical: ~ 99% of refluxers
  - Lifestyle adjustment, diet/formula alteration
  - Prokinetic agents, antacids, & proton-pump inhibitors
- Surgical: ~ 1% of all refluxing patients
  - Fundoplication

DIAGNOSTIC CHECKLIST

Consider

- Potential anatomic causes of GER

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Hypertrophic Pyloric Stenosis

KEY FACTS

TERMINOLOGY

- Hypertrophic pyloric stenosis (HPS): Idiopathic pyloric muscle thickening in young infants → progressive gastric outlet obstruction

IMAGING

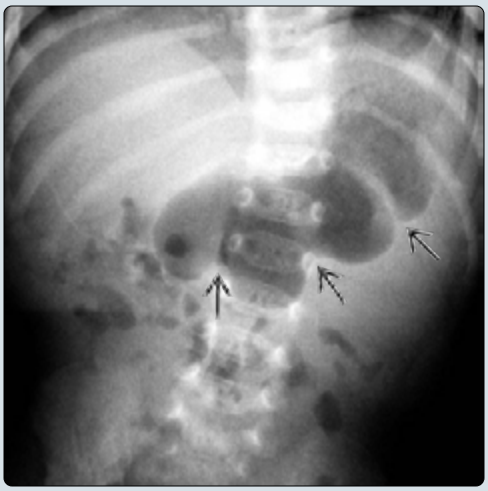
- Near complete gastric outlet obstruction due to abnormally elongated & thickened pyloric muscle
  - Pylorus fails to relax/open → minimal gastric emptying → gastric overdistention → emesis
- Ultrasound shows hypertrophied circumferential hypoechoic muscle & elongated pyloric canal filled with echogenic mucosa
  - Commonly accepted sonographic criteria for HPS
    - Pyloric channel length > 15-16 mm
    - Single wall thickness of pyloric muscle > 3 mm
    - Failure of thickened pylorus to change during exam
      - May still see trickle of passage of gastric contents
- Upper GI shows minimal barium traversing narrowed & elongated pyloric channel, mass effect on gastric antrum & duodenum by hypertrophied muscle, hyperperistaltic gastric contractions, & gastroesophageal reflux/emesis

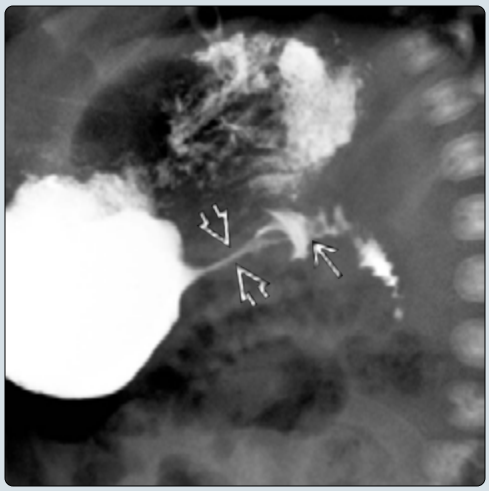
CLINICAL ISSUES

- Typically seen in infants 2-12 weeks old with progressive nonbilious projectile vomiting
  - Feedings previously tolerated (i.e., HPS not congenital)
- Incidence of 2.5-3 per 1,000 infants; M:F = 5:1
  - HPS in 1/5 infants imaged for vomiting
    - Gastroesophageal reflux > > HPS
- Surgical treatment: Pyloromyotomy
- Nonsurgical alternative (rarely employed): Medications & frequent small feedings

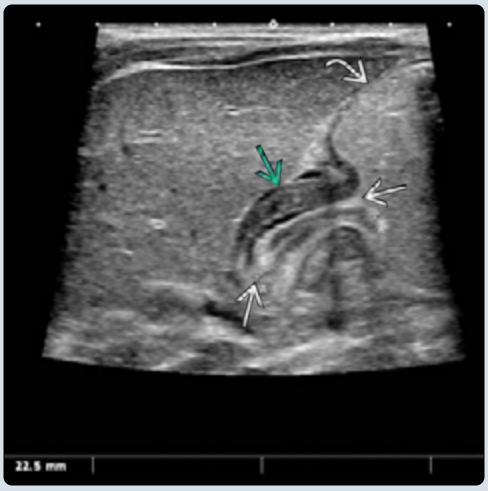
DIAGNOSTIC CHECKLIST

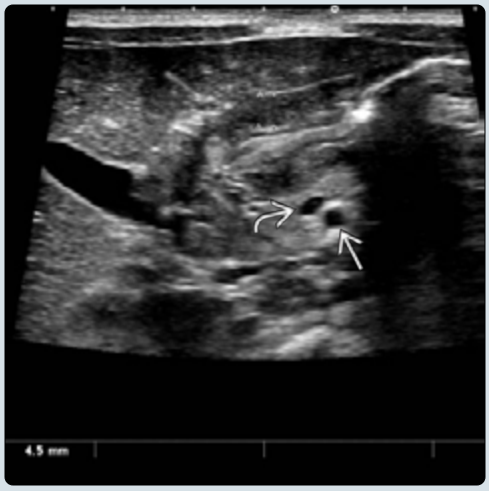
- Pylorospasm mimics HPS (but typically transient)
- Avoid overdistending stomach during imaging exam (which displaces/obscures) pylorus





(Left) *AP radiograph in a vomiting patient with hypertrophic pyloric stenosis (HPS) shows hyperperistalsis of a gas-filled stomach as muscular contractions try to push gastric contents through the narrowed, hypertrophied pylorus.* **(Right)** *Lateral upper GI shows a thin "string" of barium extending through the narrowed, elongated pyloric channel, opacifying the duodenal bulb. The thickened pyloric muscle creates rounded indentations on the gastric antrum & base of the duodenal bulb.*





(Left) *Transverse oblique ultrasound in a 3-week-old boy with HPS shows elongation of the pyloric channel at 22.5 mm in length. The hypoechoic muscle is thickened, & there is a large amount of retained formula in the stomach.* **(Right)** *Transverse oblique ultrasound in a different baby with HPS shows thickening of the pyloric muscle at 4.5 mm between the cursors. Note the normal relationship of the SMA & SMV (which suggests normal rotation of the midgut).*

TERMINOLOGY

Abbreviations

- Pyloric stenosis, hypertrophic pyloric stenosis (HPS)

Definitions

- Idiopathic pyloric muscle thickening → progressive gastric outlet obstruction
- Typically seen in infants 2-12 weeks old with progressive nonbilious projectile vomiting

IMAGING

General Features

- Best diagnostic clue
  - Near complete gastric outlet obstruction due to elongated & thickened pyloric muscle
  - Ultrasound reveals hypertrophied muscle & ↓ gastric emptying on dynamic exam
  - Upper GI shows minimal contrast traversing narrowed & elongated pyloric channel with mass effect on antrum & duodenal bulb from thickened pyloric muscle
- Location
  - Near gallbladder in RUQ by US

Radiographic Findings

- Radiography
  - Overdistended stomach with ↓ distal bowel gas
    - Enlarged stomach containing gas & retained ingested material displaces adjacent bowel
  - Stomach may be collapsed if infant has recently vomited
  - Benign gastric pneumatosis & portal venous gas rarely reported in association with HPS

Fluoroscopic Findings

- Overdistended "caterpillar stomach": Undulations of wall due to exaggerated gastric motility (hyperperistalsis)
- Tram-track or string sign of contrast within narrowed pyloric channel
- Shoulders of thickened pyloric muscle create impressions on distal antrum
- "Beak" where contrast encounters narrowed pyloric canal
- Contrast in duodenal bulb (covering hypertrophied muscle impressions at base of duodenal bulb) + contrast in narrowed & elongated pyloric channel: Mushroom sign

Ultrasonographic Findings

- Grayscale ultrasound
  - Threshold for abnormal measurements of pyloric muscle & channel length vary by study
    - In general, ↑ threshold measurements ↑ specificity but ↓ sensitivity
  - Commonly accepted threshold values for HPS
    - Single wall thickness of hypoechoic pyloric muscle > 3 mm
    - Pyloric channel length > 15-16 mm
    - Pyloric diameter > 15 mm
  - Hypertrophied & redundant echogenic mucosal lining
  - Gastric hyperperistalsis & persistently obliterated pyloric lumen on dynamic exam
  - If duodenal bulb easily identified & distended with fluid, diagnosis of HPS unlikely
- Color Doppler
  - ↑ flow in pyloric muscle & mucosa with HPS

Imaging Recommendations

- Best imaging tool
  - Ultrasound when HPS suspected
    - Excellent sensitivity & specificity; no ionizing radiation
  - Barium upper GI with atypical history (including bilious emesis)
- Protocol advice
  - Begin ultrasound scan with patient rolled onto right side in order to pool gastric fluids in antrum
  - Administer glucose water in small amounts
    - Avoid overdistending stomach, which will displace pyloric channel posteriorly or into right lower quadrant
  - Watch for pyloric channel to open with thinning of muscle & passage of fluid through channel
  - Vigorous gastric contractions of body & antrum that do not open pylorus suggest diagnosis of HPS
  - Formula or glucose water will "swirl" against thickened pylorus with each wave of gastric peristalsis in HPS

DIFFERENTIAL DIAGNOSIS

Pylorospasm

- Typically seen in irritable infants; resolves with time → wait & reimage
- Rarely as thick or elongated as true HPS

Gastroesophageal Reflux

- Cause of vomiting in 2/3 infants referred to radiology
- Presumed diagnosis when pyloric ultrasound normal

Malrotation With Midgut Volvulus

- Emesis classically bilious (greenish) from bile
- Surgical emergency due to risk of midgut ischemia
- Mildly dilated proximal duodenum with abrupt beaking near junction of 2nd-3rd duodenum
- Distal contrast passage shows "corkscrew" or swirling contrast in typically narrowed bowel

Gastric Bezoar

- Caused by accumulation of undigested matter in stomach
  - Trichobezoar: Composed of hair (& sometimes nails)
  - Phytobezoar: Composed of plant or vegetable fiber
- Contrast seen in interstices of large filling defect on upper GI

Other Causes of Gastric Outlet Obstruction

- Duodenal stenosis or antral web
- Antral polyps, antritis, annular pancreas, choledochocele, right upper quadrant mass
- Pyloric atresia

PATHOLOGY

General Features

- Etiology
  - Idiopathic hypertrophy of circular muscle bundles in pylorus
  - May be prostaglandin or erythromycin induced, neural mediated, familial

- Slightly ↑ incidence in preterm infants vs. full term
  - Presents at later chronological age in preemies
- Higher incidence in mothers < 20 years old, nulliparous, smokers, & formula-fed babies
- Associated with erythromycin (macrolide) exposure prenatally & postnatally via breast milk
- Higher incidence of HPS in patients with cystic fibrosis
- Genetics
  - Tends to run in families, not truly inherited
  - Discordant incidence among monozygotic twins favors environmental factors over genetic predisposition
- Associated abnormalities
  - Eosinophilic gastritis, prostaglandin-induced antral mucosal hyperplasia, hypergastrinemia, nasoenteric tubes, erythromycin
- Abnormal muscle tone/electrophysiology of gastroduodenal junction shown in HPS

Gross Pathologic & Surgical Features

- Hypertrophy of circular muscular layers of pylorus
- Thickening of mucosa in antrum & pylorus to ~ 1/3 diameter of pylorus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Progressive vomiting in infant who previously tolerated feedings
  - Palpable "olive" on physical exam: 97% specific in experienced hands
    - Less common finding due to earlier imaging diagnosis
- Other signs/symptoms
  - Weight loss
  - Acidosis & dehydration if prolonged course

Demographics

- Age
  - 2-12 weeks or even later in premature infants
  - Peak age of 5 weeks; 80% by 8 weeks
- Gender
  - M:F = 5:1
- Ethnicity
  - Slightly more common in Caucasians
- Epidemiology
  - Incidence of 2.5-3 per 1,000 infants
  - Vomiting caused by HPS in 1/5 infants referred for imaging

Natural History & Prognosis

- Weight loss & parental concerns typically prompt imaging
- Gradual progression with spontaneous remission after weeks
- Excellent prognosis following surgery or conservative medical management
  - No significant GI disturbances seen in German study of infants treated surgically or medically 16-26 years after diagnosis

Treatment

- Surgical: Pyloromyotomy
  - Splits thickened muscle longitudinally (without breaching mucosa), opening channel
  - Laparoscopic pyloromyotomy & open procedures have equal success rates
- Nonsurgical alternative: Medications & frequent small feedings
  - Requires several weeks before resuming normal feeding without medications
  - Medications used include Botox, IV atropine
  - Withdrawal of prostaglandins after corrective cardiac surgery → muscular hypertrophy regresses
- Complications
  - Failed surgery due to inadequate pyloromyotomy
  - Gastroduodenal myotomy or enterotomy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Pylorospasm mimics HPS transiently
- Avoid overdistending stomach, which displaces pylorus

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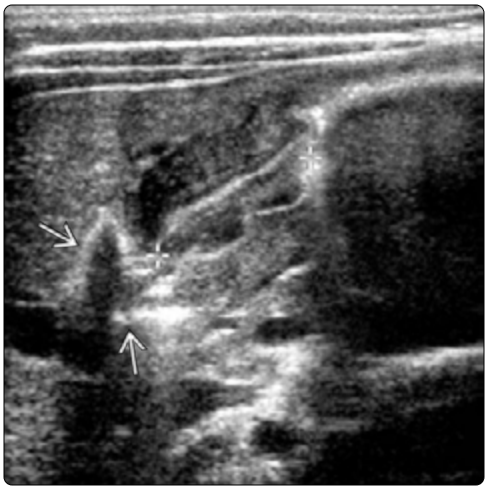
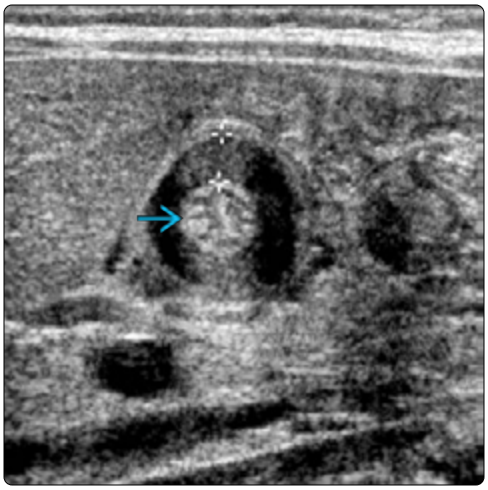
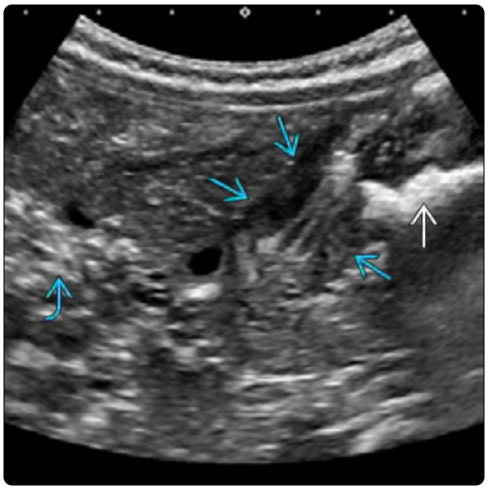
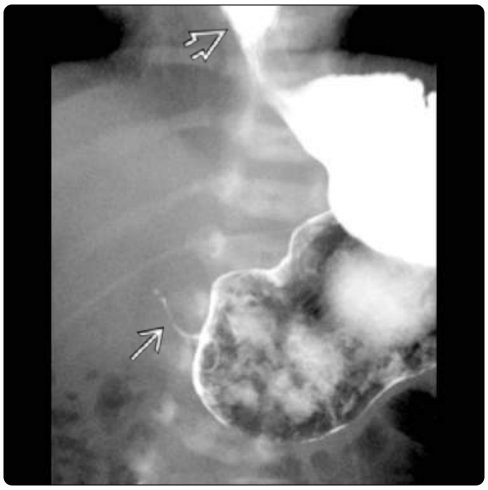


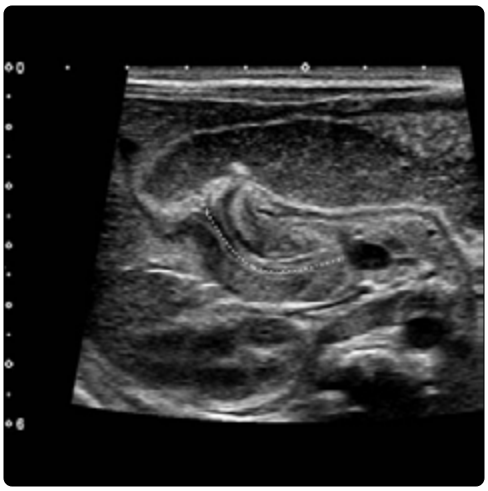


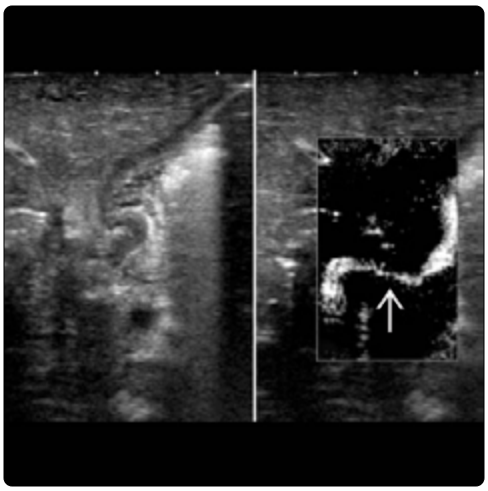
(Left) *Transverse oblique ultrasound shows cursors measuring the length of the pyloric channel in a baby with HPS. In this infant, enough fluid has left the stomach to distend the duodenal bulb, which is a useful landmark.* **(Right)** *Sonographic cross section of a hypertrophied pylorus shows the doughnut sign of HPS, consisting of a circumferential, thickened, hypoechoic muscle (between cursors) + the central, redundant, echogenic mucosa.*





(Left) *Transverse ultrasound shows a thick & elongated pylorus of HPS. There is abnormal echogenicity with "dirty shadowing" along the posterior gastric wall, consistent with pneumatosis. Branching linear echogenicities in the liver are due to portal venous gas.* **(Right)** *Delayed frontal upper GI shows a string sign of barium passing through a narrowed pylorus with reflux into the distal esophagus. Demonstrating barium in a tight pyloric channel requires patience & intermittent fluoroscopy.*





(Left) *Transverse ultrasound after a glucose-water feeding in an infant with HPS shows how feeding can overdistend the stomach & push the pylorus posteriorly. The pylorus is thickened with the channel elongated at 27.6 mm.* **(Right)** *This postprandial ultrasound shows how gas mixed with formula can interfere with visualization of structures (L). The insert (R) shows a type of Doppler optimized for slow flow (SMI), which shows a thin trickle of fluid passing through the hypertrophied channel.*

Gastric Volvulus

KEY FACTS

TERMINOLOGY

- Twisting of all or part of stomach on its axes at least 180°
- Organoaxial volvulus (OAV): Rotation along long axis
- Mesenteroaxial volvulus (MAV): Rotation along short axis

IMAGING

- Radiographic findings
  - Round gas-distended viscus under &/or above left hemidiaphragm that decompresses with nasogastric (NG) tube
  - Upright image may show 2 air-fluid levels
- Upper GI vs. CECT to confirm diagnosis
  - OAV
    - Horizontal stomach with pylorus directed inferiorly
    - Greater curvature superior to lesser curvature
  - MAV
    - Vertical stomach with pylorus superior to fundus
    - Pylorus close to or overlaps GE junction

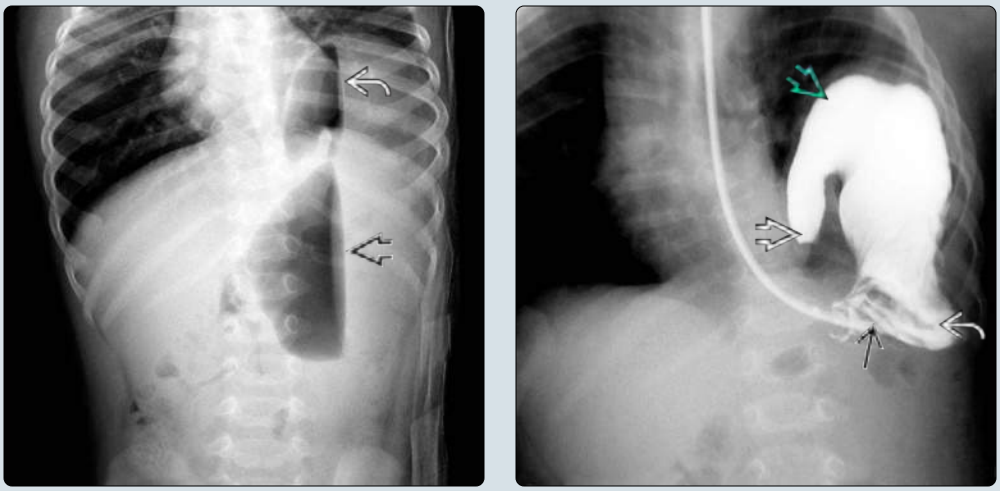
PATHOLOGY

- Primary volvulus: Absent, failed, or lax ligamentous fixation; chronic > acute
- Secondary volvulus: Disorder of gastric anatomy/function, often with diaphragm anomalies; acute > chronic

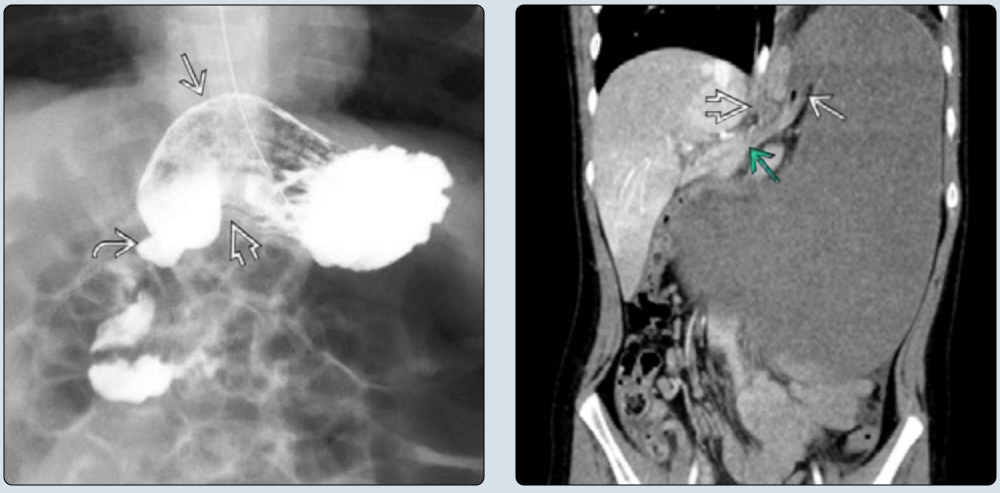
CLINICAL ISSUES

- 58% present within 1st year of life
- Symptoms: Retching, nonbilious emesis, pain, & distention
  - Respiratory distress & cyanosis also in acute setting
  - Failure to thrive & colic also in chronic setting
  - Complete Borchardt triad uncommon (unproductive retching, epigastric distention, inability to pass NG tube)
- Treatment
  - Resuscitation required in 23-60% of acute presentations
  - Gastric decompression with NG tube
  - Open or laparoscopic reduction + gastropexy &/or gastrostomy tube
  - Repair of associated defects in secondary volvulus

(Left) *Left side down decubitus radiograph of a 6-month-old patient with vomiting shows 2 large air-fluid levels in the left lower hemithorax & left upper quadrant of the abdomen, respectively.* **(Right)** *Frontal upper GI image in the same patient shows the stomach lying largely in the left hemithorax. The stomach is inverted with the antrum & gastric outlet lying above the fundus. Note the twisting of rugal folds in the cardia. Acute mesenteroaxial volvulus was found at surgery with a chronic diaphragmatic hernia.*



(Left) *Frontal upper GI image in a 4 month old with ↓ oral intake shows that the greater curvature of the stomach lies superior to the lesser curvature. The pylorus is directed inferiorly. The findings are consistent with an organoaxial volvulus.* **(Right)** *Coronal CECT in an 11 year old with vomiting shows a narrow pylorus above the level of the GE junction. The proximal duodenum is stretched. The abdominal contents extend abnormally into the lower left hemithorax. A mesenteroaxial gastric volvulus was found at surgery.*



TERMINOLOGY

Definitions

- Twisting of all or part of stomach on its long or short axis by at least 180°
- Organoaxial volvulus (OAV): Twisting along gastric long axis [i.e., line drawn from gastroesophageal (GE) junction to pylorus]
- Mesenteroaxial volvulus (MAV): Twisting along short axis (perpendicular to long axis)
- Mixed volvulus: Rotation along both long & short axes
- Twisting of stomach on axes < 180°: Gastric torsion

IMAGING

Radiographic Findings

- Round gas-distended viscus under &/or above diaphragm
  - ↓ in size after gastric decompression by nasogastric (NG) tube
- Stomach lies horizontally (OAV) or vertically (MAV)
- Upright image may show 2 air-fluid levels: One at fundus & one at antrum
- Inability to pass or abnormal course of NG tube
- + elevation of left hemidiaphragm
- ± paucity of bowel gas distally

Fluoroscopic Findings

- Upper GI series
  - OAV
    - Horizontal stomach with pylorus at level of gastric cardia; pylorus directed inferiorly
    - Greater curvature superior to lesser curvature
  - MAV
    - Vertical stomach with pylorus superior to fundus
    - Pylorus close to or overlaps GE junction
  - Beaking of contrast at sites of twisting
  - Partial or complete obstruction of contrast passage into or out of stomach

CT Findings

- Findings similar to upper GI series

MR Findings

- Fetal MR of congenital diaphragmatic hernia may show volvulus of intrathoracic stomach ± polyhydramnios

Imaging Recommendations

- Upper GI series vs. CECT

DIFFERENTIAL DIAGNOSIS

Air-Filled Thoracic Mass ± Air-Fluid Levels

- Sliding or paraesophageal hiatal hernia
- Postoperative anatomy (e.g., esophageal atresia repair with gastric pull-up vs. colonic interposition)
- Bronchopleural fistula
- Hydropneumothorax
- Congenital pulmonary airway malformation

Massively Dilated Abdominal Viscus

- Hypertrophic pyloric stenosis
- Toxic megacolon ± Hirschsprung disease
- Cecal or sigmoid volvulus

PATHOLOGY

General Features

- Stomach relatively fixed at ends: GE junction at esophageal hiatus, pylorus attached to retroperitoneal duodenum
- Stomach also normally fixed by ligaments: Gastrophrenic, gastrosplenic, gastrocolic, gastrohepatic
- Primary volvulus
  - Absent, failed, or lax ligamentous fixation
  - 74% of chronic volvulus
- Secondary volvulus
  - Disorder of gastric anatomy/function (distention, peptic ulcer, neoplasm, prior gastric surgery) or abnormality of adjacent organ (diaphragmatic hernia or eventration, splenic anomalies, intestinal malrotation)
  - 69% of acute volvulus, commonly with diaphragm anomalies

Gross Pathologic & Surgical Features

- Acute: OAV 54%, MAV 41%, mixed 2%
- Chronic: OAV 85%, MAV 10%, mixed 3%

CLINICAL ISSUES

Presentation

- 58% present within 1st year of life
- Acute, chronic, or acute-on-chronic symptoms include
  - Retching, nonbilious emesis, abdominal pain, & distention
  - Respiratory distress & cyanosis also seen acutely
  - Failure to thrive & colic also seen chronically
  - Hematemesis in late stage secondary to ischemia
- Complete Borchardt triad uncommon (unproductive retching, epigastric distention, inability to pass NG tube)

Natural History & Prognosis

- Complications: Obstruction, ischemia, perforation
- Mortality: Acute 7.1%, chronic 2.7%
- In large review, 1 in 8 chronic volvulus cases developed acute symptoms with 60% requiring resuscitation

Treatment

- Resuscitation required in 23-60% of acute presentations
- Gastric decompression with NG tube
- Open or laparoscopic reduction + gastropexy &/or gastrostomy tube
- Repair of associated defects in secondary volvulus
  - Controversial if gastropexy required after repair of associated anomaly

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Ingested Coins

KEY FACTS

IMAGING

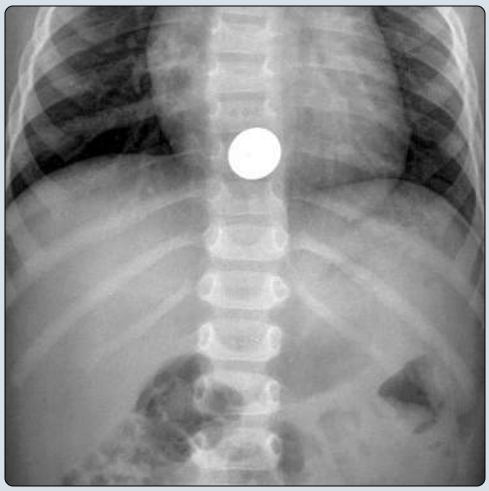
- Disc-shaped metallic density without circumferential beveled edge/step-off
- Most common sites of impaction
  - Upper esophagus at thoracic inlet
  - Midesophagus at aortic arch impression
  - Lower esophageal sphincter at gastroesophageal junction
- Other sites of impaction include pylorus, duodenum, ileocecal valve
- Imaging recommendations
  - Screening frontal radiographs of neck including nasopharynx through pelvis ± lateral upper airway
  - Lateral radiograph if foreign body identified

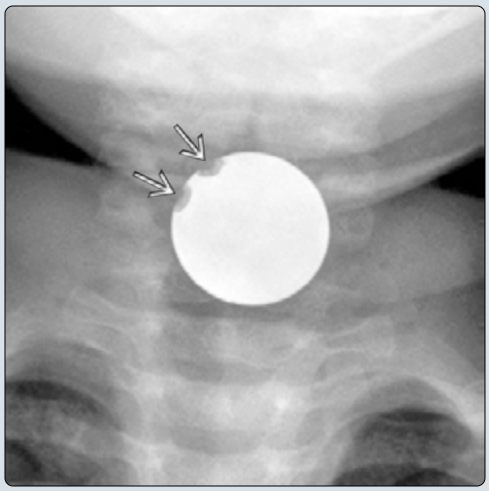
TOP DIFFERENTIAL DIAGNOSES

- Button battery ingestion
- Magnet ingestion
- Aspirated coin in trachea

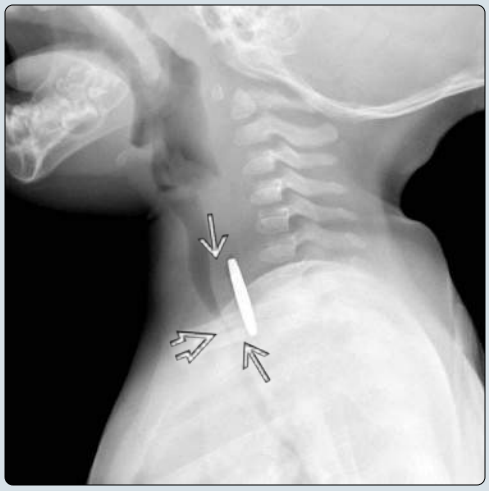
CLINICAL ISSUES

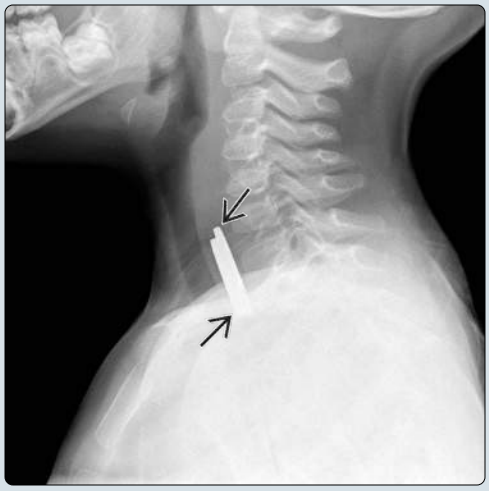
- Majority of ingestions by children < 5 years old
- Most common symptoms
  - Asymptomatic: Witnessed ingestion or incidental finding
  - Symptomatic: Drooling, chest/neck pain, vomiting, dysphagia, cough, respiratory distress, stridor
- Treatment of esophageal coins
  - Symptomatic: Urgent endoscopic removal
  - Asymptomatic: Endoscopic removal within 24 hours; repeat radiograph prior to endoscopy
- Treatment of coins in stomach
  - Monitor stools for passage; repeat radiograph in 2 weeks
  - Endoscopic removal if not passed in 2-4 weeks; repeat radiograph prior to endoscopy
- Treatment of small bowel coins
  - Observation
  - Enteroscopy/surgical removal if symptomatic





(Left) *Frontal radiograph of the lower chest in a 2 year old with emesis shows a 19.5-mm metallic disc, consistent with a coin, projecting over the distal esophagus. The disc was found to be a dime at endoscopy. Radiographs cannot accurately identify coin types based on size.* **(Right)** *AP radiograph shows a coin lodged in the cervical esophagus. Note that the coin is wider than the trachea, indicative of an esophageal location. The erosions ⇨ are due to gastroesophageal reflux reaching a zinc-based coin.*





(Left) *Lateral radiograph demonstrates a metallic coin in the proximal esophagus at the thoracic inlet. There is soft tissue swelling ⇨ between the coin and the trachea with associated tracheal narrowing ⇨. These findings suggest a more chronic foreign body that may be difficult to remove.* **(Right)** *Lateral airway radiograph in a 2 year old after a witnessed ingestion shows 2 directly apposed coins ⇨ projecting over the upper esophagus at the thoracic inlet. This appearance can mimic the edge step-off of a button battery.*

IMAGING

General Features

- Location
  - Most common sites of impaction
    - Upper esophagus at thoracic inlet
    - Midesophagus at aortic arch impression
    - Lower esophageal sphincter (LES) at gastroesophageal junction
  - Other sites of impaction include pylorus, duodenum, ileocecal valve
- Size
  - United States coins
    - Penny (1 cent): 19 mm
    - Nickel (5 cents): 21 mm
    - Dime (10 cents): 18 mm
    - Quarter (25 cents): 25 mm
  - Radiographic measurements may not allow accurate coin identification due to magnification & small differences in coin diameters

Radiographic Findings

- Disc-shaped metallic foreign body without circumferential beveled edge/step-off/halo
- Typically seen en face (oriented in coronal plane) on frontal radiograph
- Rarely oriented in sagittal plane on frontal radiograph
- May exert mass effect on trachea secondary to size of coin or associated soft tissue swelling

Imaging Recommendations

- Ingested foreign body screening radiographs: Frontal views to include neck through pelvis ± lateral upper airway
- Lateral radiograph if foreign body identified
  - Helps confirm location
  - Helps determine type of foreign body

DIFFERENTIAL DIAGNOSIS

Button Battery Ingestion

- Disc-shaped metallic foreign body
  - Lateral view: Circumferential beveled edge/step-off
  - Frontal view: Circumferential halo or double ring sign
- Esophageal impaction requires emergent removal

Magnet Ingestion

- Metallic densities of variable shapes & sizes
- Multiple magnets or magnets with other metallic foreign bodies may attract through different bowel loops → fistulization, perforation, obstruction

Other Metallic Foreign Bodies

- Buttons, jewelry, toy parts

Aspirated Coin in Trachea

- Classic teaching that sagittally oriented coin on frontal radiograph likely in trachea: Incorrect
  - Coin impacted in esophagus much more likely regardless of orientation (coronal or sagittal)
  - Coin may lie in coronal or sagittal plane in trachea or esophagus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic: Witnessed ingestion or incidental finding
  - Symptomatic: Drooling, chest/neck pain, vomiting, dysphagia, cough, respiratory distress, stridor

Demographics

- Majority of patients < 5 years old
- Penny ingestions most common: 44%

Natural History & Prognosis

- 25-30% of esophageal coins pass spontaneously
  - Likelihood of spontaneous passage correlates with location in esophagus: Proximal 14%, middle 43%, distal 67%
- Complications uncommon
  - Esophageal stricture, perforation, aortoesophageal or tracheoesophageal fistulas
  - Post-1982 copper-plated zinc penny: 97.5% zinc
    - Gastric acid may erode coin margins
    - Gastric acid reacts with zinc to form zinc-chloride
    - Ingestion of large number may cause zinc toxicity

Treatment

- North American Society for Pediatric Gastroenterology, Hepatology, & Nutrition guidelines
  - Esophageal coin
    - Symptomatic: Urgent endoscopic removal
    - Asymptomatic: Endoscopic removal within 24 hours; repeat radiograph prior to endoscopy to look for interval progression
  - Gastric coin
    - Monitor stools for passage; repeat radiograph in 2 weeks
    - Endoscopic removal if not passed within 2-4 weeks
    - Repeat radiograph prior to endoscopy
  - Small bowel coin
    - Clinical observation
    - Enteroscopy/surgical removal if symptomatic
- Use of glucagon (to relax LES) controversial: May cause vomiting & aspiration
- Fluoroscopic-guided retraction with Foley catheter may result in acute airway obstruction if coin moves into airway
- Pushing coin into stomach by dilator (bougienage) does not allow esophagus to be evaluated

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Ingested Button Batteries

KEY FACTS

TERMINOLOGY

- Ingestion of disc-shaped battery, typically by young child
  - Increasingly of more injurious lithium cell type
- Esophagus particularly susceptible to injury by lodged battery with potentially catastrophic consequences

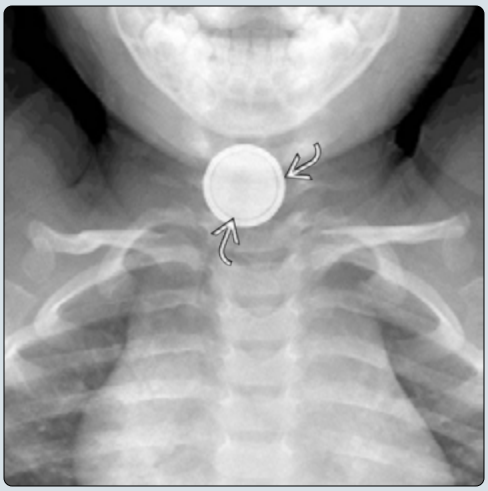
IMAGING

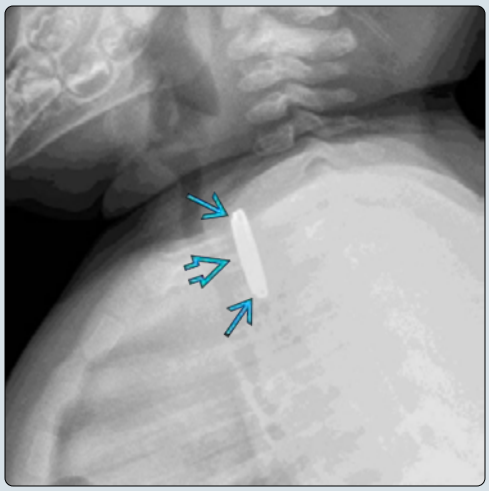
- Frontal radiograph: Margin shows double halo/ring en face
- Lateral radiograph: Rim of step-off/beveled edge
  - Negative pole (narrower side): Site of anticipated most severe injury
- North American Society for Pediatric Gastroenterology, Hepatology, & Nutrition imaging guidelines
  - Radiographic coverage from nasopharynx to anus
    - Lateral view at least at site of confirmed foreign body
  - After emergent removal of esophageal battery
    - CTA/MR if esophageal injury present to evaluate proximity/involvement of vascular structures
      - CTA safest & most efficient vessel assessment
    - Esophagram to exclude leak prior to advancing diet
  - Battery distal to esophagus: Management varies based on battery size & patient age

CLINICAL ISSUES

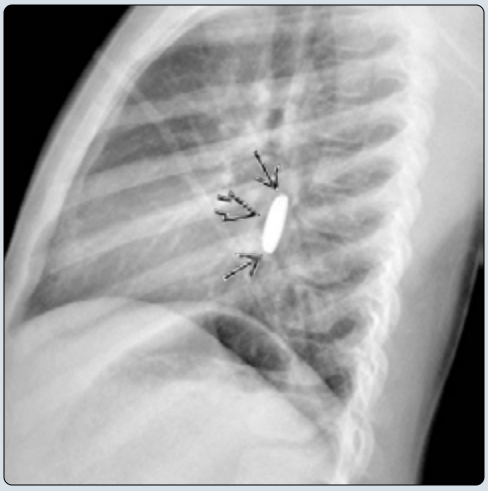
- Caustic injury due to hydroxide radical production in tissues adjacent to negative pole
- Unwitnessed ingestion more likely to present in delayed fashion with nonspecific symptoms: Vomiting, difficulty feeding, cough, chest or abdominal pain, drooling, stridor
- ↑ risk of major complications: Unwitnessed ingestion, size ≥ 20 mm (majority lithium; radiographs overestimate size), age < 5 years old, multiple batteries ingested
- Complications include tracheoesophageal fistula, esophageal perforation, esophageal stricture, vocal cord paralysis, aortoenteric fistula (high fatality rate)
- Injury evolves weeks after removal, potentially resulting in delayed aortoenteric fistula

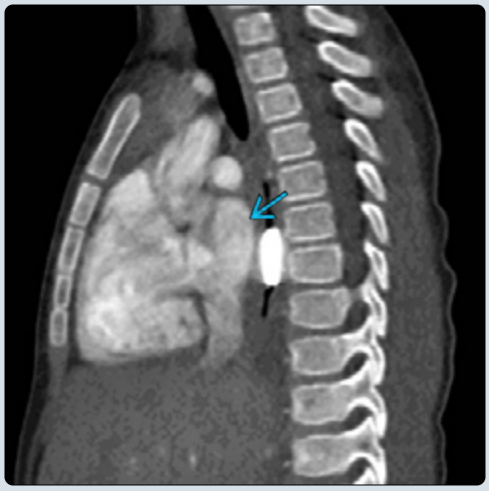
(Left) *Frontal radiograph of a 1 year old with acute cough, gagging, & emesis (but no witnessed ingestion) shows a 23-mm diameter disc-shaped foreign body with a double ring/halo ➡ appearance in the proximal esophagus, consistent with a button battery.* **(Right)** *Lateral radiograph of a 10 month old with a witnessed foreign body ingestion & subsequent drooling & emesis shows the circumferential edge step-off ⇨ typical of a button battery. The negative pole is the narrower anterior side ⇨.*





(Left) *Lateral radiograph from a 7 year old after the known ingestion of a button battery (> 20 mm in diameter > 12 hours prior to presentation) shows that the button battery is impacted in the mid-distal esophagus. Beveled edges are seen ⇨ with the negative pole ⇨ directed anteriorly toward the left atrium.* **(Right)** *Sagittal CECT from the same patient shows the esophageal battery posterior to the left atrium ⇨. There is soft tissue thickening (inflammation) without evidence of fistula.*





TERMINOLOGY

Definitions

- Ingestion of disc-shaped battery, typically by young child
- Esophagus particularly susceptible to injury by lodged battery with potentially catastrophic consequences

IMAGING

General Features

- Most common sites of impaction in esophagus
  - Upper esophagus at thoracic inlet
  - Mid esophagus at aortic arch
  - Lower esophageal sphincter at GE junction

Radiographic Findings

- Metallic density disc of variable size
  - Size ≥ 20 mm: ↑ risk for worse outcome (but radiograph overestimates size)
- Frontal view: Margin shows double halo/ring en face
- Lateral view: Rim of step-off/beveled edge in tangent
  - Step-off may not be visible on new thinner batteries
  - Negative pole: Narrower side on lateral view; site most likely for most severe injury
  - Soft tissue edema surrounding battery may cause anterior tracheal displacement or narrowing
- Complications may show mediastinal widening (from edema or fluid collections) &/or gas
- Metallic fragments may remain after battery removal

CT

- CTA to look for aortic/vascular injury
  - Wall irregularity/outpouching or wall edema
  - Active extravasation
  - Mediastinal edema/fluid collections
- No established findings predictive of subsequent vascular catastrophes at this time

Esophagram

- Irregularity/edema of mucosa with luminal narrowing
- Leak of contrast into mediastinum or trachea
- Long-term stricturing

Imaging Recommendations

- Initial "foreign body screening" frontal radiographs to include neck through pelvis
- Lateral radiographs of
  - Upper airway to include nasopharynx
  - Other site if foreign body identified
    - Distinguish battery from coin & identify negative pole
- Follow-up imaging as per North American Society for Pediatric Gastroenterology, Hepatology, & Nutrition (NASPGHAN) guidelines
  - CTA safest & most efficient tool to assess vessels

DIFFERENTIAL DIAGNOSIS

Coin Ingestion

- Flat metallic disc (no beveled edge or double ring sign)

Magnet Ingestion

- > 1 magnet or 1 magnet + other metallic foreign body may cause bowel injury by attraction through bowel walls

PATHOLOGY

General Features

- Lithium cell → caustic injury after impaction in esophagus
  - Mucosa contacts both poles, completes circuit
  - Electrolytic current generates hydroxide radicals in tissue adjacent to negative pole → ↑ pH
  - Injury evolves weeks after removal
- Nonlithium types injure mainly via alkaline leakage

CLINICAL ISSUES

Presentation

- Ingestion may be witnessed (often without symptoms) or unwitnessed with symptoms: Vomiting, difficulty feeding, cough, chest or abdominal pain, drooling, stridor

Natural History & Prognosis

- Unwitnessed ingestion accounts for 92% of associated fatalities & 56% of major outcome cases
- ↑ risk of major complications: Size ≥ 20 mm (majority lithium), age < 5 years old, multiple batteries ingested
- Major complications
  - Tracheoesophageal fistula in 48%
  - Esophageal perforation in 23%
  - Esophageal stricture in 38%
  - Vocal cord paralysis in 10%
  - Aortoenteric fistula in 46% of fatal cases (1977-2015)
    - Can occur weeks after battery removal

Treatment

- NASPGHAN guidelines based on battery location & size, clinical stability, & patient age
  - Esophageal & stable: Immediate endoscopic removal
  - Esophageal & unstable/active bleeding: Immediate endoscopic removal in OR with surgery present
    - CTA or MR to evaluate involvement/proximity of aorta
    - Negative CTA, MR → esophagram to exclude leak
    - Injury close to aorta (≤ 3 mm) → serial CTA or MR every 5-7 days until injury recedes
  - Distal to esophagus, ≥ 20 mm, < 5 years old
    - Assess for esophageal injury + endoscopic removal within 24-48 hours
    - If esophageal injury → CTA, MR
  - Distal to esophagus, < 20 mm, &/or ≥ 5 years old
    - May consider outpatient observation with repeat radiograph (48 hours ≥ 20 mm or 10-14 days < 20 mm)
    - Endoscopic removal if GI symptoms develop or battery not passed at time of repeat radiograph

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Ingested Multiple Magnets

KEY FACTS

TERMINOLOGY

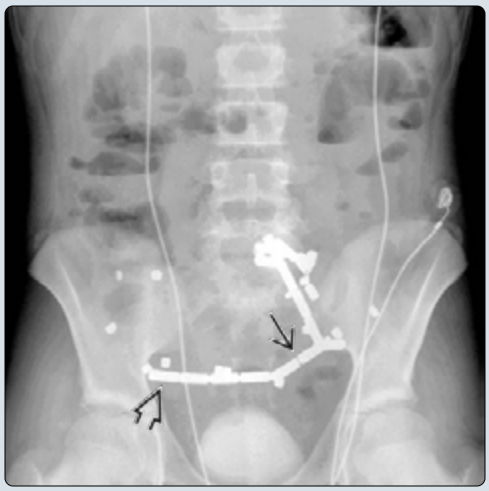
- Ingestion of multiple magnets or single magnet + additional metallic foreign bodies
  - Potential for significant bowel complications
- Rare-earth magnets 5-10x stronger than traditional magnets

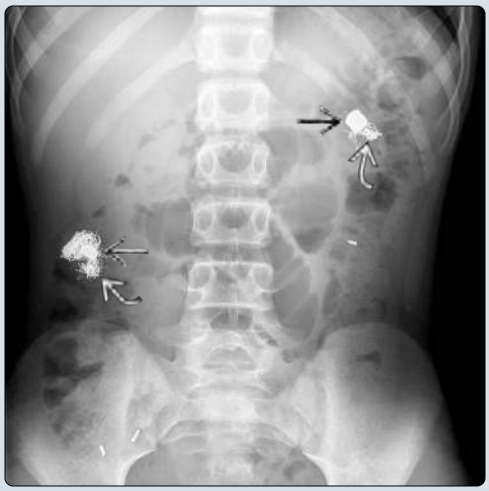
IMAGING

- Metallic density foreign bodies; shapes variable
  - Multiple attracted "stacked" magnets may simulate single cylindrical foreign body
- Magnets attract through bowel walls
- Entrapment of interposed bowel wall suggested with
  - Gap between otherwise closely apposed magnets or magnet & adjacent metallic foreign body
  - Failure of magnet to move on sequential radiographs
- Abnormal bowel gas patterns from complications
  - Ulceration, perforation, fistulae, obstruction, volvulus
- Foreign body ingestion radiographic series includes
  - Frontal views of neck through anus, ± lateral view of upper airway
  - Lateral views if foreign body identified

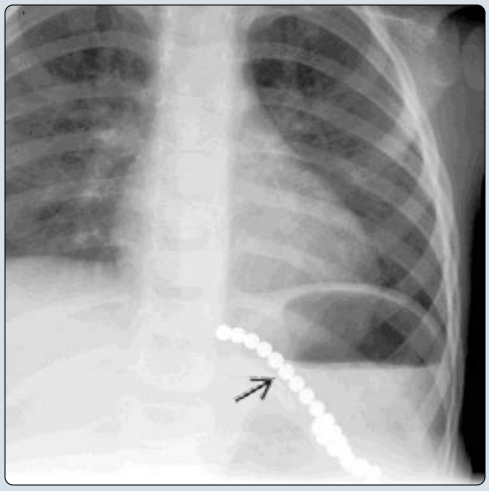
CLINICAL ISSUES

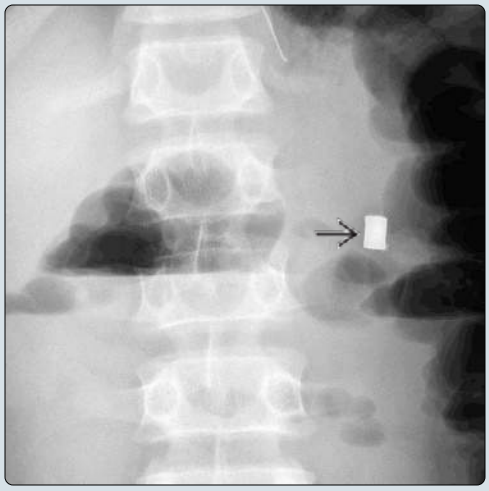
- Management depends on number of magnets, location in GI tract, & symptoms
  - Single magnet: Remove vs. follow radiographic passage
  - Multiple magnets or magnet + metal in esophagus/stomach: Endoscopic or surgical removal
  - Multiple magnets or magnet + metal beyond stomach
    - With symptoms &/or abnormal bowel gas pattern: Operative exploration with magnet removal
    - Without symptoms or abnormal bowel gas pattern: Consider careful, frequent clinical & radiographic evaluation until passage vs. removal





(Left) *Frontal upright abdominal radiograph of a 12-year-old autistic boy with abdominal pain & bilious emesis shows multiple rod-shaped metallic foreign bodies, most of which appear linked. Small gaps between the magnets may represent entrapped intervening bowel. Numerous small bowel perforations & fistulae were noted at surgery.* **(Right)** *Frontal radiograph shows 2 clusters of magnets, which attracted additional ingested metallic foreign bodies. Associated bowel perforations were present at surgery.*





(Left) *Frontal upright chest radiograph in an asymptomatic 2 year old suspected to have swallowed magnets shows a chain of magnets projecting over the stomach. The magnets were removed endoscopically.* **(Right)** *Frontal upright abdominal radiograph from an asymptomatic 13 year old who swallowed 6 magnets shows a stack of metallic foreign bodies in the left abdomen. Jejunal & colonic perforations due to the magnets attracting across bowel walls were found at surgery.*

TERMINOLOGY

Synonyms

- Postgastric magnetopathy

Definitions

- Ingestion of multiple magnets or single magnet + additional metallic foreign bodies
  - Potential for significant bowel complications due to attraction through bowel loops
- Newer rare-earth magnets composed of iron, boron, & neodymium: 5-10x stronger than traditional magnets

IMAGING

Radiographic Findings

- Metallic density foreign bodies; shapes variable
  - Rods, discs, spheres/ball bearings, others
- May have larger nonradiopaque component
- Multiple attracted "stacked" magnets may simulate single cylindrical foreign body
  - Magnification helps delineate individual components
- Entrapment of interposed bowel wall suggested with
  - Gap between otherwise closely apposed magnets or magnet & adjacent metallic foreign body
  - Failure of magnet to move on sequential radiographs
- Abnormal bowel gas patterns from complications
  - Ulceration, perforation, fistulae, obstruction, volvulus

Imaging Recommendations

- NASPGHAN guidelines recommend imaging if
  - Known magnet ingestion
  - Unexplained GI symptoms with rare-earth magnets in environment
- Screening foreign body ingestion radiographs
  - Frontal views from neck through anus, ± lateral view of upper airway
  - Lateral view if foreign body identified
    - Assists in localization & characterization (such as attached metal or magnets not seen on 1 view)

DIFFERENTIAL DIAGNOSIS

Coin Ingestion

- Most common ingested radiopaque foreign body
- Metallic disc without beveled edge

Button Battery Ingestion

- Metallic disc with beveled edge on lateral view, double ring on frontal view
- Esophageal location requires emergent removal

PATHOLOGY

General Features

- Magnets attract to each other through bowel walls
  - Powerful rare-earth magnets attract through up to 6 bowel walls
    - Rare-earth magnets banned from toys in 2009
    - Now commonly sold as adult desk toys, faux piercings
- Produce pressure ulcers, ischemic injury, fistulae, necrosis, perforations, &/or obstruction
  - Mucosal ulcerations may occur in < 8 hours

CLINICAL ISSUES

Presentation

- 54.7% < 5 years old
- Suspicion of magnet ingestion critical: Only 1% of cases between 2000-2012 had witnessed ingestion [US Consumer Product Safety Commission (CPSC) data]
- Common symptoms (single center study, 56 cases): None 57.1%, abdominal pain/vomiting 32.1%, choking 10.7%

Natural History & Prognosis

- CPSC data: 72 cases in United States (2000-2012)
  - No adverse effect: 33%, > 1 perforation & necrosis: 34%, 1 perforation: 6%, ulcer: 5%, fistula: 3%, volvulus: 2%
  - Surgery: 70%, endoscopic removal: 8%, passed naturally: 21%, death: 1%

Treatment

- NASPGHAN published guideline algorithm
  - Single magnet
    - Esophagus/stomach: Remove if risk of more ingestions or consider outpatient serial radiographs until passage ensured
    - Beyond stomach: Removal if possible or outpatient serial radiographs until passage
  - > 1 magnet or 1 magnet + metal in esophagus/stomach
    - If < 12 hours: Pediatric GI consult for endoscopic removal
    - If > 12 hours: Consult surgery prior to endoscopic removal; surgical removal if endoscopy unsuccessful
  - > 1 magnet or 1 magnet + metal beyond stomach
    - Consult pediatric gastroenterologist & surgery
    - Symptomatic: Surgical removal
    - Asymptomatic & no obstruction/perforation on imaging
      - Entero/colonoscopic removal **or** serial radiographs
      - Radiographs every 4-6 hours in emergency department
      - Progression on radiographs: Confirm passage (may educate parents & continue as outpatient)
      - No progression on radiographs: Admit → continue serial radiographs every 8-12 hours if asymptomatic **or** surgical/endoscopic removal
  - Parent education: No other metal or magnets near child

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KEY FACTS

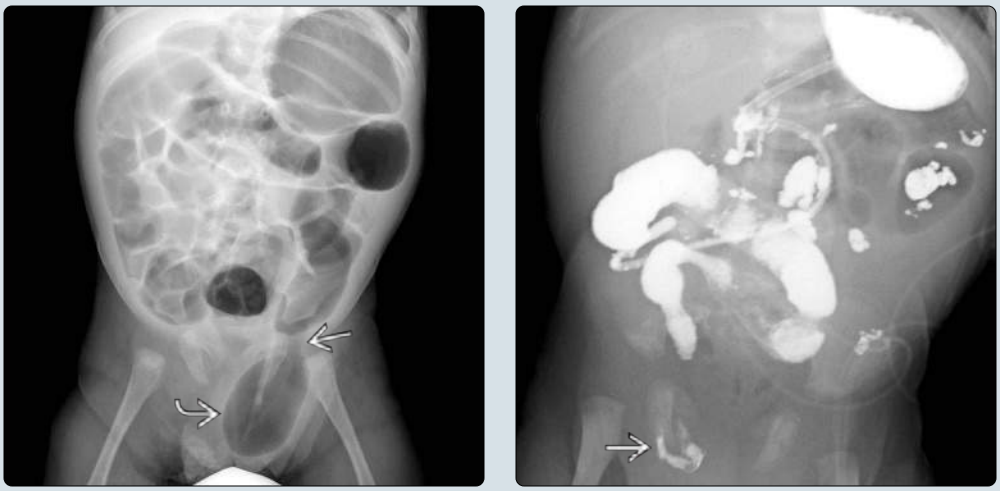
TERMINOLOGY

- Hernia: Protrusion of contents from normally encasing body cavity through normal or abnormal opening
- Inguinal hernia: Protrusion of abdominal contents through defect in inguinal region
  - Indirect inguinal hernia: Contents protrude into open deep inguinal ring, extend through patent processus vaginalis, & exit superficial inguinal ring
    - 15% of inguinal hernias bilateral
    - In females, ovary may herniate through canal of Nuck
  - Direct inguinal hernia: Contents pass through wall of inguinal canal (due to weak abdominal musculature), exiting superficial inguinal ring
- Umbilical hernia: Contents extend into open umbilical ring
- Femoral hernia: Contents extend through femoral ring
- Littre hernia: Contains Meckel diverticulum; site varies
- Amyand hernia: Inguinal hernia contains appendix ± inflammation
- Internal hernia: Extends through defect in abdominal cavity
- Traumatic: Extends through posttraumatic abdominal wall defect
- Incarcerated hernia: Contents cannot be reduced without special maneuvers, sedation, anesthesia, or surgery
- Strangulated hernia: Contents ischemic due to compression by hernia channel

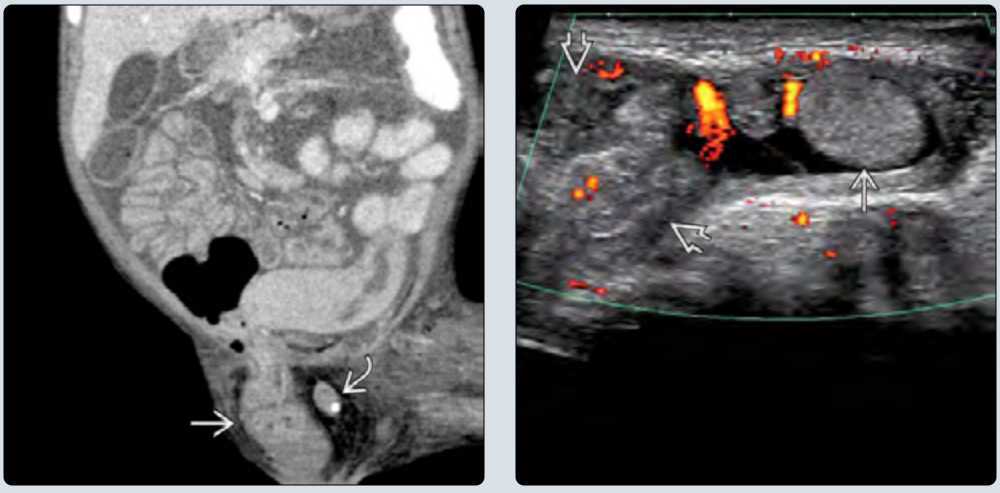
IMAGING

- US excellent modality to investigate palpable abnormality
  - Can characterize hernia contents & evaluate blood flow to contents & adjacent tissues
    - Bowel: Peristalsis, swirling contents, gut signature
  - Standing &/or Valsalva maneuver help reproduce reduced hernia to confirm diagnosis
- Obstructive hernia: Dilated bowel loop enters abdominal wall defect, decompressed bowel loop exits hernia defect
- Signs of strangulation: Bowel wall thickening &/or ↓ enhancement, engorged vasa recta, mesenteric stranding/fluid

(Left) *AP radiograph in a neonate with abdominal distention shows a loop of gas-filled bowel extending into the left hemiscrotum ➡ through the left inguinal canal ➡. The dilation of proximal bowel loops in the abdomen suggests an associated obstruction.* **(Right)** *Frontal image from a small bowel follow-through in a neonate shows a loop of small bowel ➡ extending into the right inguinal canal. The bowel upstream is not dilated. Inguinal hernias are more common in premature infants due to a patent processus vaginalis.*



(Left) *Coronal CECT in a 1-year-old boy shows the cecum ➡ & appendix ➡ herniating into the right hemiscrotum. The appendix contains a fecalith but was not inflamed. When an inguinal hernia contains the appendix, it is termed an Amyand hernia.* **(Right)** *Longitudinal power Doppler US in a 1-month-old boy shows bowel ➡ herniating through the inguinal canal. There is ischemia of the ipsilateral testis ➡ from spermatic cord compression, a phenomenon limited to neonates.*



TERMINOLOGY

Definitions

- Hernia: Protrusion of contents from normally encasing body cavity through normal or abnormal opening
  - Inguinal hernia: Protrusion of abdominal contents through defect in inguinal region
    - Indirect inguinal hernia: Protrusion of abdominal contents into open deep inguinal ring, through patent processus vaginalis, exiting superficial inguinal ring
      - Canal of Nuck: Term for patent processus vaginalis in females; extends into labia majoris
    - Direct inguinal hernia: Abdominal contents pass through wall of inguinal canal (due to weak abdominal musculature), exiting superficial inguinal ring
  - Umbilical hernia: Protrusion of abdominal contents through open umbilical ring
  - Femoral hernia: Protrusion of abdominal contents through femoral ring
  - Littre hernia: Hernia contains Meckel diverticulum
  - Amyand hernia: Inguinal hernia containing appendix
  - Internal hernia: Hernia through fossa or foramen within abdominal cavity
    - Defect associated with internal hernia can be congenital or acquired
  - Traumatic: Hernia through traumatic abdominal wall defect
- Incarcerated hernia: Hernia in which contents cannot be reduced without special maneuvers, sedation, anesthesia, or surgery
- Strangulated hernia: Hernia in which contents become ischemic due to vascular compression by hernia channel

Associations

- Indirect inguinal hernia: Prematurity
- Umbilical hernia: Prematurity, Down syndrome, Beckwith-Wiedemann syndrome
- Acquired internal hernia: Abdominal surgery requiring Roux-en-Y reconstruction
- Recent repair of large congenital hernia: Omphalocele, gastroschisis, diaphragmatic hernia
  - ↑ intraabdominal pressure status post reduction of abdominal contents + defect repair can lead to recurrent or new hernias

IMAGING

General Features

- Best diagnostic clue
  - Protrusion of abdominal contents through defect in abdominal wall
- Location
  - Inguinal hernia: Extends to labia or scrotum
    - 60% of inguinal hernias occur on right side
    - 15% of inguinal hernias bilateral
  - Umbilical hernia: Extends into umbilicus
  - Femoral hernia: Extends into femoral ring, lateral to lacunar ligament
    - More common on right side
  - Littre hernia: Occurs with inguinal (50%), umbilical (30%), & femoral (20%) hernias
  - Traumatic hernia: Right lower abdomen, lateral to rectus sheath
  - Left paraduodenal hernia: To left of inferior mesenteric vein & 4th duodenum within left mesocolon
- Size
  - Abdominal wall defect can vary in size from < 5 mm to > 10 cm depending on location, size of patient, & type of hernia

Radiographic Findings

- Signs of obstruction with dilated air-filled loops of bowel & multiple air-fluid levels
- Inguinal hernia: Soft tissue or air-filled mass extending beyond pelvis inferiorly
- Umbilical hernia: Round soft tissue or air-filled mass within umbilicus
  - Best seen on cross-table lateral view

Fluoroscopic Findings

- Small bowel follow-through
  - Contrast entering loop of bowel within hernia
  - Obstructive hernia: Dilated loop of bowel entering abdominal wall defect & decompressed loop of bowel exiting hernia defect

CT Findings

- Protrusion of abdominal contents through defect in abdominal wall
- Obstructive hernia: Dilated loop of bowel entering abdominal wall defect & decompressed loop of bowel exiting hernia defect
- Signs of impending strangulation: Free fluid within hernia, bowel wall thickening, or luminal dilation
- Signs of strangulation: Bowel wall thickening, ↓ enhancement of bowel wall, engorged vasa recta, & mesenteric stranding
- Internal hernia: Cluster of small bowel loops, stretched mesenteric vessels
  - Left paraduodenal hernia: Bowel between stomach & pancreas or transverse colon
    - Bowel may appear to be contained by sac
    - Superior/anterior displacement of inferior mesenteric vein
- Traumatic hernia: Defect in abdominal musculature with soft tissue stranding ± injury of abdominal organs

Ultrasonographic Findings

- Inguinal hernia: Abdominal contents entering scrotum or labia
  - Confirm bowel presence with peristalsis, visualization of swirling bowel contents, gut signature in bowel wall
  - Use color Doppler to confirm blood flow to bowel
  - In females, may see herniated ovary
  - In male neonates, may see ↓ to absent color Doppler flow to ipsilateral testis

Imaging Recommendations

- Best imaging tool
  - Diagnosis often based on clinical exam
  - US used to diagnose etiology of unknown abdominal bulges

- Standing or Valsalva maneuver help reproduce reduced hernia (by forcing abdominal contents through abdominal defect)
- Can characterize hernia contents
- Can evaluate blood supply to hernia contents & adjacent tissues compressed by hernia

- CT useful to diagnose etiology of obstruction or abdominal pain

DIFFERENTIAL DIAGNOSIS

Hydrocele

- Can communicate through inguinal hernia defect
- May be confined to spermatic cord or extend into scrotum
  - Fluctuant mass of inguinal canal or scrotum
  - ↑ transillumination
  - ↑ in size late in day or during viral infection

Omphalocele

- Congenital midline abdominal wall defect into base of umbilicus
- Herniated contents covered by sac

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Hernia: Painless easily reducible bulge
  - Incarcerated hernia: Hernia not easily reduced
  - Obstructed hernia: Vomiting, abdominal distension
  - Strangulated hernia: Pain, peritonitis, shock

Demographics

- Age
  - Inguinal hernia: Occurs at any age; ↑ incidence in premature infants
  - Femoral hernia: Most commonly occurs 5-10 years
  - Umbilical hernia: Most common in infants & toddlers
    - 75% of infants < 1,500 g have umbilical hernia
  - Traumatic hernia: Mean age = 9.5 years
- Gender
  - Inguinal hernia: 9-10x more common in males
  - Femoral hernia: During childhood, slightly more common in males
    - In adults, femoral hernias more common in females
  - Traumatic hernia: More common in males due to high-risk activities
- Ethnicity
  - Umbilical hernias more common in children of African descent
- Epidemiology
  - Indirect inguinal hernias
    - Inguinal hernias in 0.8-4.4% of children
      - 13% of infants born < 32 weeks & 30% of infants weighing < 1,000 g will have inguinal hernia
    - Risk for inguinal hernia becoming incarcerated ↑ in early infancy
      - 10-30% become incarcerated in premature infants
      - Up to 15% become incarcerated in older children
    - Inguinal hernia repair 1 of most frequently performed surgeries in children
    - Incarcerated inguinal hernia 2nd most common cause of bowel obstruction after postsurgical adhesions
  - Direct inguinal hernia
    - Accounts for 2-5% of groin hernias in children
  - Femoral hernias
    - Rare in children
    - Accounts for 0.1-1.1% of groin hernias
    - Become incarcerated in 15-20% of patients
    - Majority contain properitoneal fat
  - Umbilical hernia
    - 10-20% of all infants born with umbilical hernia
  - Littre hernia
    - 0.3-0.8% of all hernias contain Meckel diverticulum
  - Amyand hernia
    - Appendix found in inguinal hernia in 1% at operation
    - Accompanied by acute appendicitis in 0.1%
  - Internal hernia
    - Left paraduodenal hernia most common type of congenital internal hernia
  - Traumatic hernia
    - May be caused by bicycle handle bar injury

Natural History & Prognosis

- Indirect inguinal hernia occurs with patent processus vaginalis
  - 40% of patent processus vaginalis close during 1st months of life
  - 20% of boys have patent processus vaginalis at 2 years of age

Treatment

- Surgery performed to treat most types of hernia due to risk of incarceration
- Umbilical hernia has low risk of incarceration
  - Most umbilical hernias spontaneously close by age 5
  - Surgery performed to close large hernias or hernias that fail to spontaneously close
- Complications of inguinal hernia repair include recurrence, infection, testicular atrophy, injury to vas deferens, & infertility

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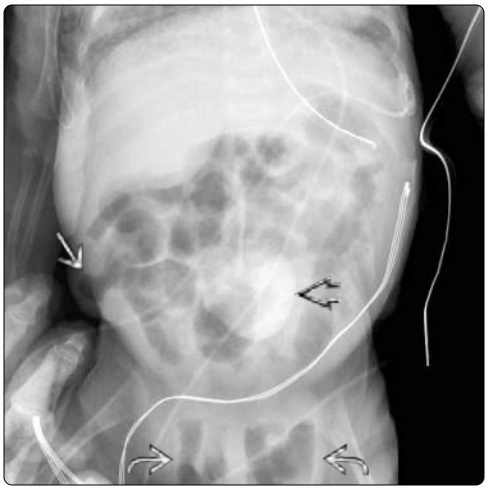
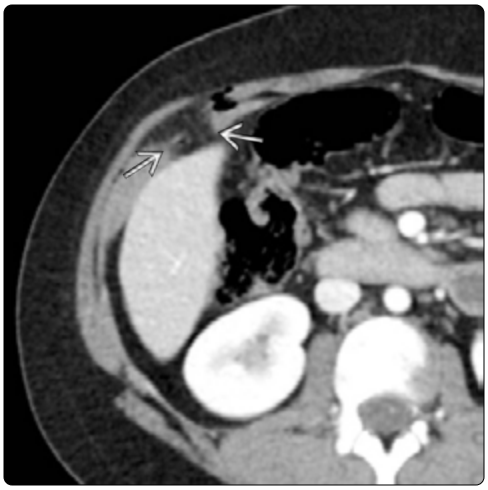
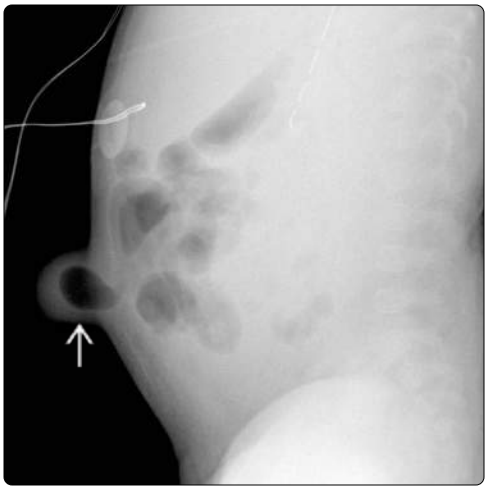
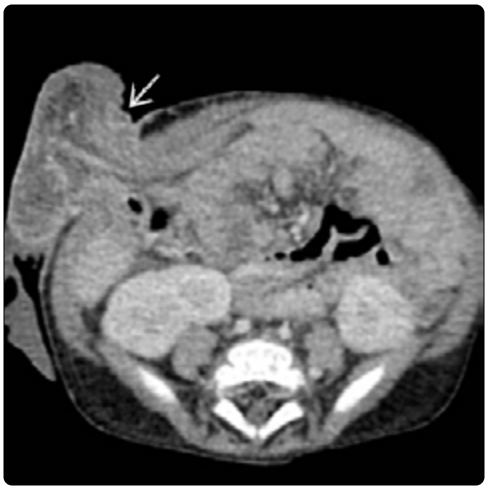


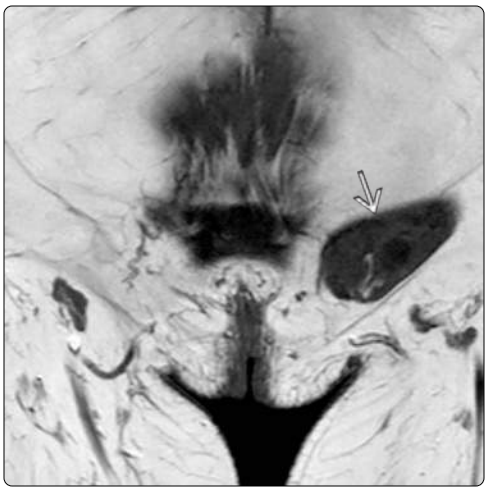


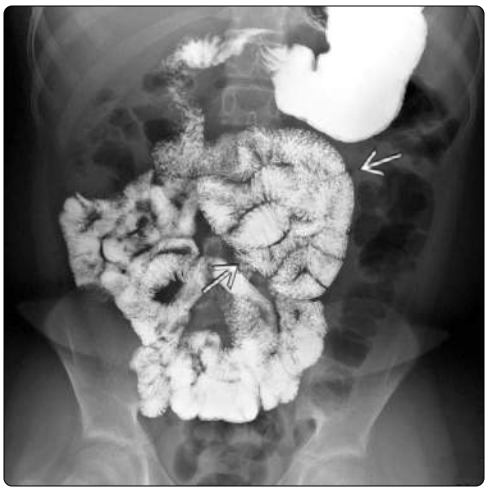
(Left) *AP radiograph in a premature infant shows 4 distinct abdominal hernias: Umbilical hernia, bilateral inguinal hernias, & a lateral abdominal wall hernia at the site of prior surgery. Inguinal & umbilical hernias are more common in premature infants.* **(Right)** *Axial CECT of the abdomen in an 8-year-old boy status post bicycle handlebar injury shows a defect of the abdominal wall musculature with herniation of omental fat.*





(Left) *Cross-table lateral radiograph shows an air-filled loop of bowel within a small umbilical hernia. Umbilical hernias are common in newborns. They typically spontaneously resolve by 5 years of age.* **(Right)** *Axial CECT in a young child with a history of a right lower quadrant ileostomy shows a large parastomal hernia containing bowel.*





(Left) *Coronal T1 MR in a young adult female shows the left ovary extending through the left inguinal canal (a hernia of the canal of Nuck).* **(Right)** *Small bowel follow-through in an adolescent male shows a cluster of small bowel loops in the left upper quadrant, consistent with a left paraduodenal internal hernia. This is the most common type of congenital internal hernia.*

Omphalocele

KEY FACTS

TERMINOLOGY

- Midline abdominal wall defect (AWD) with visceral extrusion into umbilical cord base
- AWD < 5 cm: Minor or small; > 5 cm: Giant

IMAGING

- Extruded abdominal contents contained by round membranous sac
  - Umbilical cord inserts on sac
- Small bowel & liver more frequently herniated than spleen, stomach, bladder, colon, or gonads
- Bowel malrotation in virtually all cases
  - Postrepair risk of midgut volvulus higher than gastroschisis as sac prevents bowel inflammation that incites "protective" adhesions
- Prenatal US/MR defines AWD location, size, covering, contents, & associated anomalies
- Postnatal radiographs demonstrate sac, which may contain gas-filled bowel loops

TOP DIFFERENTIAL DIAGNOSES

- Gastroschisis
- Umbilical hernia
- Bladder exstrophy
- Limb-body wall complex

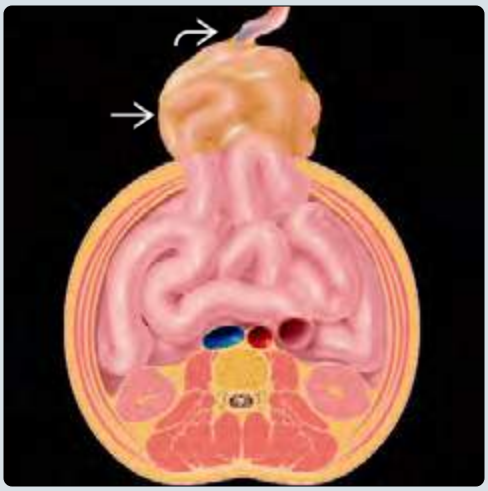
PATHOLOGY

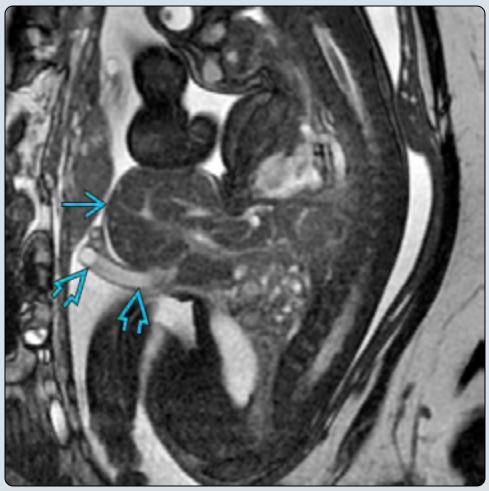
- High rates of associated structural & chromosomal anomalies: Beckwith-Wiedemann syndrome, OEIS complex, pentalogy of Cantrell, trisomy 18 > 21, 13; many others

CLINICAL ISSUES

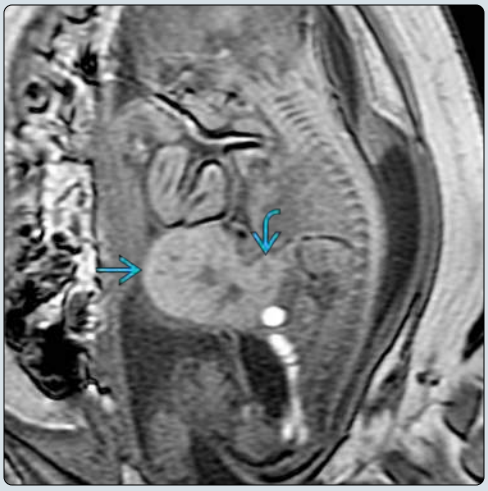
- Repair technique & timing depends on size, contents, patient stability, & other anomalies
- With isolated omphalocele, survival 75-95%
- Poor prognosis with
  - Associated structural &/or chromosomal abnormalities
  - Omphalocele rupture
  - Pulmonary hypoplasia

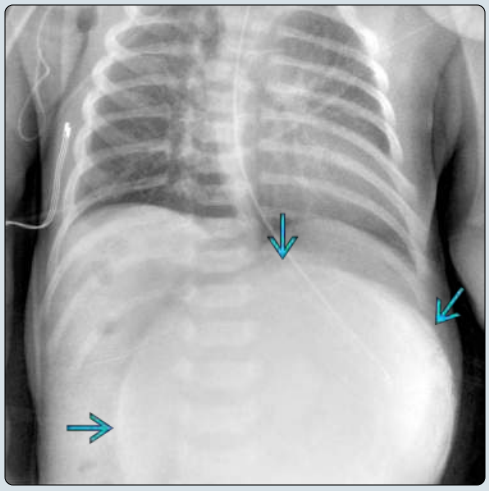
(Left) *Axial graphic shows a midline abdominal wall defect (AWD) with herniation of the small bowel into a sac. The umbilical cord inserts directly on the membranous sac.* **(Right)** *Sagittal SSFP MR in a 34-week gestation fetus shows a large midline AWD with the liver herniated into a sac. The umbilical cord attaches to the inferior aspect of the sac. The lung volumes (not shown) were mildly low in this fetus; for poorly understood reasons, patients with giant omphalocele may develop pulmonary hypoplasia.*





(Left) *Sagittal T1 MR in the same fetus depicts the extraabdominal & intraabdominal portions of the liver.* **(Right)** *Supine AP radiograph in the same patient immediately after delivery shows a large, round opacity projecting over the abdomen. No bowel gas is seen in the mass, typical of a giant omphalocele containing only liver. No pneumothorax or bell-shaped chest is noted to suggest significant pulmonary hypoplasia.*





TERMINOLOGY

Synonyms

- Exomphalos

Definitions

- Midline abdominal wall defect (AWD) with visceral extrusion into umbilical cord base

IMAGING

General Features

- Best diagnostic clue
  - Extruded abdominal contents contained by sac
    - Small bowel & liver more frequently herniated than spleen, stomach, bladder, colon, or gonads
  - Umbilical cord inserts on sac
- Size
  - AWD < 5 cm: Minor or small
    - Chromosomal abnormalities more likely
    - Higher rate of intestinal, brain anomalies
  - AWD > 5 cm: Giant
    - Usually contains liver
    - Higher rate of cardiac, renal, pulmonary anomalies

Radiographic Findings

- Round mass protruding from abdominal midline
  - Gas-containing bowel may be seen in sac
- Giant omphalocele may lead to pulmonary hypoplasia
  - Pneumothorax, bell-shaped chest
- Ruptured omphalocele
  - Extruded bowel loops ± other viscera without sac
- Wide variety of associated anomalies

Fluoroscopic Findings

- Bowel malrotation in virtually all cases
  - Normal fixation never occurs for herniated bowel
  - Postrepair adhesions not as high as gastroschisis (as covered/unexposed bowel not inflamed)
    - Risk of midgut volvulus may be greater

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal scans
    - Transient physiologic bowel herniation into umbilical cord before 12 weeks gestation
    - Omphalocele confirmed with
      - Herniation of stomach or liver (not physiologic)
      - Bowel in umbilical cord > 12 weeks
- Pulsed Doppler
  - Postnatal abdominal Doppler may be useful during reduction of abdominal viscera to evaluate flow
    - Abnormal high-resistance visceral waveforms seen with ↑ abdominal pressures

MR Findings

- Fetal MR useful to
  - Characterize sac contents + associated anomalies
  - Calculate lung volumes to predict pulmonary hypoplasia

Imaging Recommendations

- Best imaging tool
  - Prenatal US detects & characterizes AWD (location, size, contents, covering)

DIFFERENTIAL DIAGNOSIS

Gastroschisis

- Small AWD to right (rarely left) of umbilical cord insertion
- No membrane covering herniated bowel loops
  - Chronic exposure to amniotic fluid → inflammation → dysmotility, adhesions
- Extruded contents limited to bowel, stomach
  - Liver involvement extremely rare
- Additional anomalies in < 20%
  - Intestinal atresias most common

Umbilical Hernia

- Small (< 2 cm) skin-covered midline AWD
- May contain bowel or omentum

Bladder Exstrophy

- Lower AWD with superficial soft tissue mass (exposed bladder surface), wide pubic symphysis
- No normal bladder visualized prenatally despite normal amniotic fluid

Limb-Body Wall Complex

- Lethal malformation complex
  - Large AWD with extruded abdominal viscera fused to placenta or uterine wall
  - Scoliosis, limb abnormalities, short umbilical cord

PATHOLOGY

General Features

- Genetics
  - Most cases sporadic
  - Chromosomal abnormalities in 30-50%
    - Trisomy 18 most common
    - Less likely with larger, liver-containing AWD
- Associated abnormalities
  - Additional structural anomalies: Up to 77%
    - More likely overall if omphalocele contains liver
    - Additional intestinal anomalies (especially Meckel diverticulum, intestinal atresias) & brain anomalies more likely in small omphaloceles
  - Genitourinary anomalies: Up to 40%
    - Bladder exstrophy, cloacal exstrophy
    - Ureteropelvic junction obstruction, renal ectopia, solitary kidney, cryptorchidism
    - Prune-belly syndrome
  - Gastrointestinal anomalies: Up to 40%
    - Tracheoesophageal fistula, imperforate anus, absent gallbladder, enteric duplication, intestinal atresia, Meckel diverticulum
  - Respiratory insufficiency
    - Pulmonary hypoplasia with giant omphaloceles
      - Impaired diaphragm, thoracic cage development
      - Oligohydramnios if fluid accumulates in omphalocele sac from bladder anomaly
  - Congenital heart disease: Up to 50%
    - Septal defects, transposition, ectopia, tetralogy of Fallot, absent inferior vena cava

- Musculoskeletal anomalies: Up to 42%
  - Clubfoot, polydactyly, limb deficiency, rib anomalies
  - Scoliosis, vertebral abnormalities
- Central nervous system (CNS) anomalies: Up to 33%
  - Encephalocele, holoprosencephaly, cerebellar hypoplasia, myelomeningocele, anencephaly
- Specific associated syndromes
  - Beckwith-Wiedemann syndrome in 5-10%
    - Omphalocele, macroglossia, visceromegaly, hypoglycemia, embryonal tumors (hepatoblastoma, Wilms tumor)
  - OEIS complex (1:200,000 live births)
    - **O**mphalocele, **e**xstrophy of cloaca/bladder, **i**mperforate anus, **s**pinal defects
  - Pentalogy of Cantrell
    - Omphalocele, ectopia cordis, & adjacent defects of diaphragm, pericardium, sternum

- Embryology
  - Failure of central migration of lateral mesodermal body folds in early gestation

Gross Pathologic & Surgical Features

- Omphalocele membrane composed of peritoneum + amnion + intervening Wharton jelly
- Covered AWD protects bowel from inflammation, dilation
- Larger AWD makes in utero vascular insults to bowel unlikely

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Detected by prenatal ultrasound
- Other signs/symptoms
  - ↑ maternal serum α-fetoprotein (70%)

Demographics

- Epidemiology
  - Incidence: 2-4.4/10,000
    - Multigestation:singleton pregnancies = 3:1

Natural History & Prognosis

- Premature birth in up to 42%
- Survival 75-95% if normal chromosomes & no other anomalies
- Overall survival beyond neonatal period
  - Small/minor: Up to 92%; giant: Up to 67%
- Poor prognosis with
  - Associated structural or chromosomal abnormalities: Mortality 80-100%
  - Omphalocele rupture
  - Pulmonary hypoplasia

Treatment

- Fetus: Amniocentesis for karyotype; no intervention
- Birth: Delivery at tertiary facility
  - C-section for giant omphalocele to prevent dystocia, rupture
  - Respiratory support in giant omphalocele patients with pulmonary hypoplasia → pneumothorax, respiratory distress
  - Sac covered to prevent fluid & heat loss, stabilized to prevent rupture
    - Urgent repair not indicated unless sac ruptures
- Repair goal: Return viscera to abdomen with skin & fascial coverage
  - Abdominal compartment syndrome (ACS) develops if viscera returned too quickly, causing ↑ intraabdominal pressures with impaired visceral blood flow, hypotension, respiratory distress (↓ diaphragm motion)
- Small/minor omphalocele: Initial direct repair
  - If bladder pressure ↑ intraoperatively by visceral reduction, then temporizing measures required to prevent ACS
    - Synthetic graft; skin-only closure; silo reduction
- Giant: Treatment controversial, more complex
  - Staged: Gradual reduction by silo with gravity vs. synthetic graft + increasing compression
  - Nonoperative (best for unstable patients): Topical agents promote epithelialization of sac (over ~ 3 months), which gradually incorporates to abdominal cavity
    - Subsequent hernia repair required
- Postrepair issues
  - Pulmonary dysfunction
  - Inguinal or ventral hernia
  - Gastroesophageal reflux
    - May account for associated feeding intolerance as bowel functions normally
  - Complications of malrotation
    - Midgut volvulus risk > repaired gastroschisis patients due to lack of bowel inflammation (which incites "protective" adhesions)

DIAGNOSTIC CHECKLIST

Consider

- Omphalocele often associated with structural &/or chromosomal anomalies

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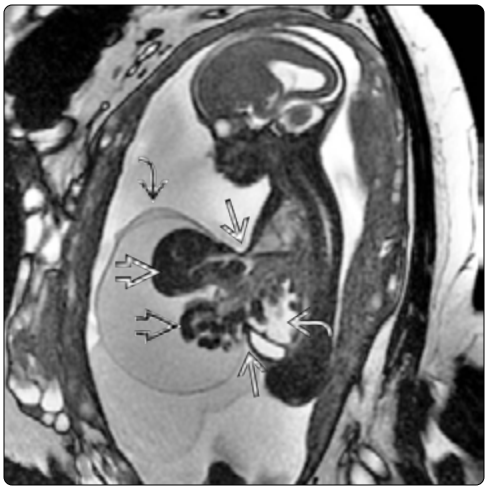
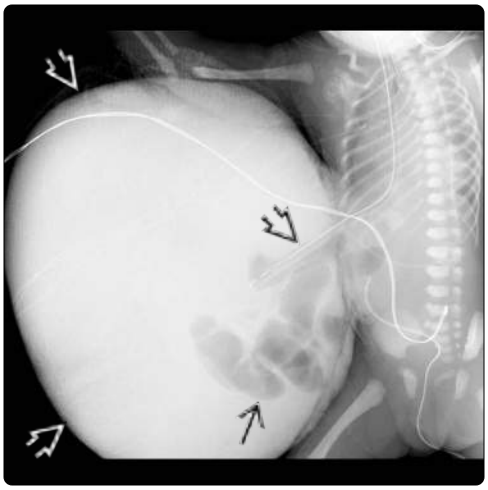
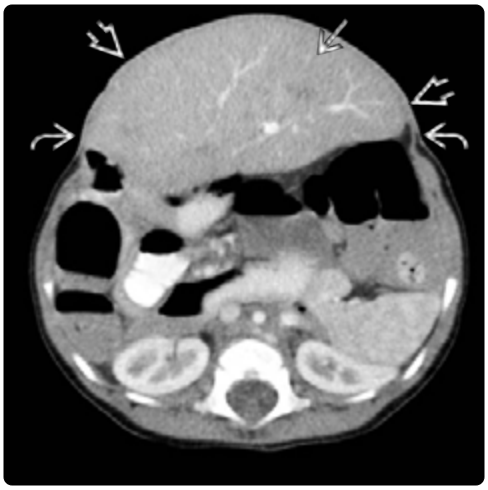
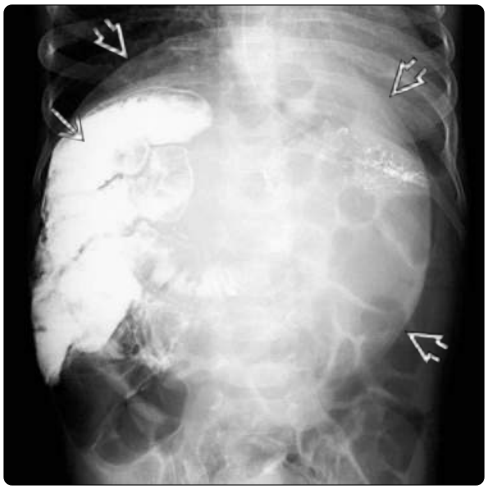


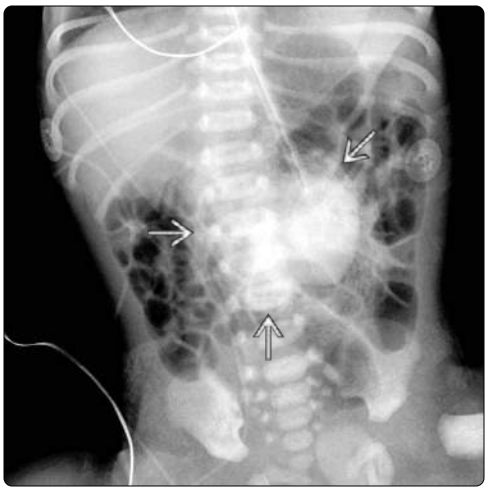


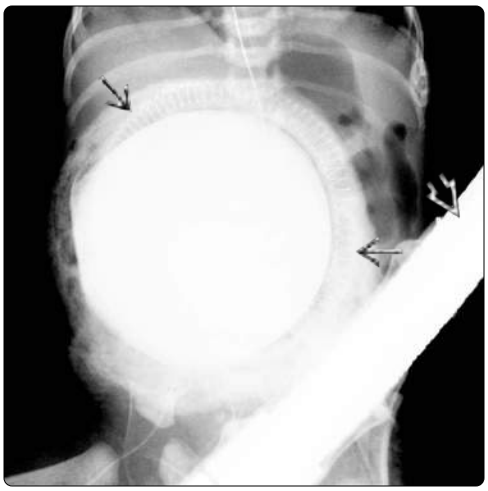
(Left) *Sagittal SSFP MR in a 23-week gestation fetus shows a large midline AWD with herniation of the entire liver & much of the bowel into a large sac, consistent with a giant omphalocele. Ascites is noted in the fetal abdomen.* **(Right)** *Oblique radiograph of the same patient after delivery shows the giant omphalocele sac containing the decompressed stomach & multiple gas-distended small bowel loops.*





(Left) *Axial CECT of an 18 month old shows a giant omphalocele that has been closed by epithelialization (without fascia or muscle). There is continued protrusion of the liver into the abdominal wall defect (which is covered by skin).* **(Right)** *Frontal upper GI series 10-minute image from the same patient shows proximal small bowel loops in the right abdomen, typical of abnormal bowel fixation (i.e., malrotation) in these patients. The large abdominal protuberance is clearly visible.*





(Left) *AP radiograph in a newborn shows a lobular density overlying the central abdomen at the expected site of the umbilicus, corresponding to a small omphalocele. This protuberant mass is larger than the rounded opacity seen with an umbilical hernia.* **(Right)** *AP radiograph shows a silo enclosing a ruptured omphalocele. Note the characteristic appearances of the intraabdominal metallic coiled spring at the base of the silo as well as the overlying bar from which the silo bag is suspended.*

Gastroschisis

KEY FACTS

TERMINOLOGY

- Congenital abdominal wall defect (AWD) lateral (usually to right) of normal umbilicus
  - Bowel herniates freely into amniotic cavity without sac
- Simple gastroschisis (majority): No bowel complications
- Complex gastroschisis (10-20%): Presence of bowel atresia, volvulus, necrosis, &/or perforation

IMAGING

- Prenatal: Dilated intraabdominal bowel in complex cases
- Postnatal: Upper GI & small bowel follow-through after repair shows malrotation ± dysmotile dilated loops, obstruction, short gut

TOP DIFFERENTIAL DIAGNOSES

- Omphalocele
- Limb-body wall complex
- Cloacal exstrophy
- Amniotic band syndrome

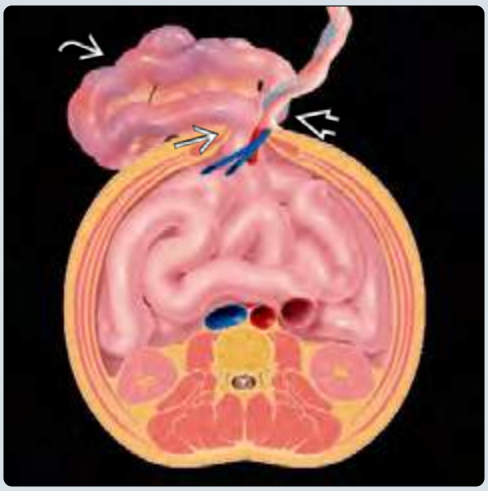
PATHOLOGY

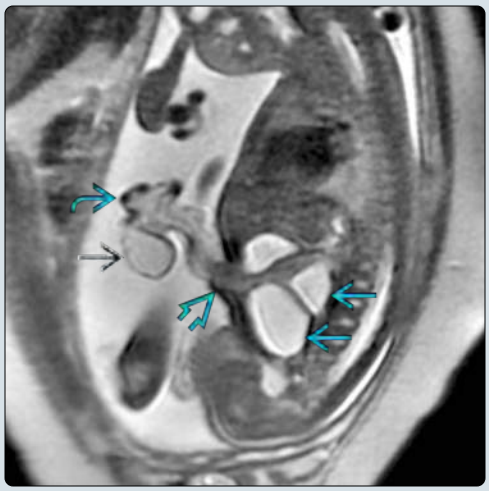
- Additional anomalies in up to 20%
  - Intestinal atresias (jejunoileal > colonic) most common
- AWD may cause prenatal bowel injury
  - Chronic irritation by exposure to amniotic fluid → dysmotility, adhesions
  - Constriction at AWD → vascular injury with atresia, volvulus, necrosis, perforation
- Postrepair bowel insults
  - Necrotizing enterocolitis, hernias, obstruction by adhesions
  - Not at risk of midgut volvulus despite malrotation

CLINICAL ISSUES

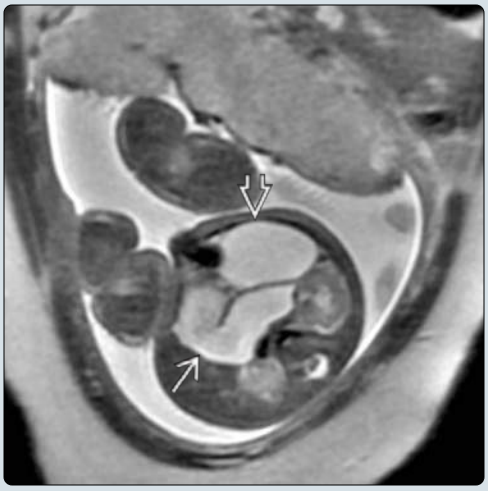
- At delivery, herniated bowel inflamed in ~ 33%
- Treatments include immediate primary repair vs. gradual bowel reduction with preformed silo to prevent compartment syndrome
- Overall infant survival > 90% (but ↓ in complex forms)

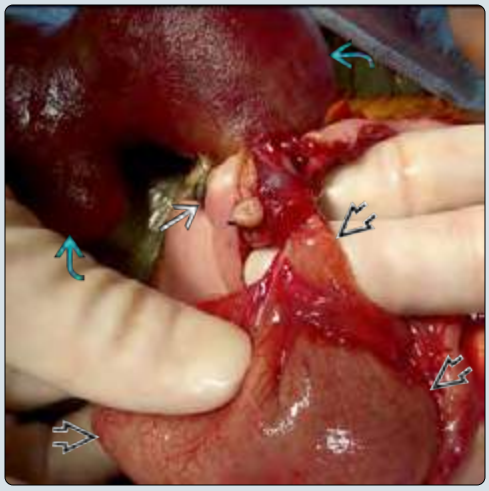
(Left) *Axial graphic shows a small abdominal wall defect (AWD) ➡ to the right of the umbilicus ➡ with herniation of bowel loops ➡. No covering membrane is seen. These features are typical of gastroschisis.* **(Right)** *Sagittal SSFSE T2 MR in a 31-week gestation fetus shows a small AWD ➡ right of the umbilical cord insertion (not shown). Small-caliber meconium-containing bowel ➡ protrudes with a cystic lesion ➡ into the amniotic fluid. Several dilated proximal small bowel loops ➡ remain in the abdomen.*





(Left) *Axial SSFSE T2 MR in the same patient shows dilated intraabdominal duodenum ➡ & proximal jejunum ➡.* **(Right)** *Intraoperative photograph in the same neonate shows a dilated atretic jejunum ➡ extending up to the AWD, which is right of the umbilicus ➡. A cystic mass of necrotic bowel ➡ protruded from the defect, consistent with a closed gastroschisis.*





TERMINOLOGY

Definitions

- Gastroschisis: Congenital abdominal wall defect (AWD) lateral to normal umbilicus
  - AWD almost always right of umbilical cord insertion
  - Bowel herniates into amniotic cavity without covering membrane
- Simple gastroschisis: Without bowel complications
- Complex gastroschisis: Presence of bowel atresia, volvulus, necrosis, &/or perforation in ~ 10-20% of cases

IMAGING

General Features

- Best diagnostic clue
  - Prenatal ultrasound: AWD to right of umbilical cord with herniation of free-floating loops of bowel
    - Herniated viscera include
      - Small bowel (almost always)
      - Less frequently: Large bowel, stomach, gonads
      - Liver very rarely reported

Radiographic Findings

- Bowel loops protrude from AWD without covering sac
  - Bowel may be dilated & thick walled
- Preformed silo: Intraabdominal metallic spring coil at base of overlying dense bag that contains herniated bowel
  - Only if utilizing gradual (rather than primary) reduction of bowel into abdomen
- Pneumatosis, portal venous gas, free air if necrotizing enterocolitis (NEC) develops

Fluoroscopic Findings

- Upper GI series + small bowel follow-through to demonstrate intestinal complications after repair
  - Malrotation (expected in all cases)
  - Dysmotility with dilated bowel loops → delayed small bowel transit time
  - Obstruction due to adhesions
  - Short gut with history of complicated gastroschisis & necrosis requiring resection

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal US
    - Herniated bowel loops free floating in amniotic fluid without covering membrane
    - Bowel wall may be thickened, echogenic, nodular
    - Significance of bowel dilation debated; intra- or extraabdominal loops may be dilated
      - Considerations include closing gastroschisis, volvulus, atresia, ischemia, chronic inflammation from amniotic fluid exposure
      - Definition of dilation controversial: Ranges from > 6 mm to > 25 mm
    - Oligohydramnios more frequent than polyhydramnios
    - Intrauterine growth restriction (IUGR)
      - Overestimated by ↓ abdominal circumference due to bowel extrusion
    - Predictors of "complex gastroschisis" or "adverse neonatal outcome": Intraabdominal bowel dilation, polyhydramnios, IUGR; ± extraabdominal bowel dilation, gastric dilation

MR Findings

- Fetal MR may be useful to evaluate associated anomalies

Imaging Recommendations

- Best imaging tool
  - Prenatal ultrasound

DIFFERENTIAL DIAGNOSIS

Omphalocele

- Viscera herniated into base of umbilical cord with covering membranous sac
- Always contains bowel; often contains liver, other organs
- Ruptured omphalocele may mimic gastroschisis
- > 50% with associated malformations, chromosomal abnormalities

Limb-Body Wall Complex

- Lethal malformation with large AWD
- Herniated viscera not covered by membrane
- Fetus fixed to placenta
- Abnormally short umbilical cord
- Scoliosis, limb anomalies

Cloacal Exstrophy

- Low AWD with nonvisualization of bladder
- Exposed bladder plates divided by everted bowel
  - Prolapsed ileum resembles "elephant trunk"
- Low omphalocele forms upper part of AWD

Physiologic Gut Herniation

- Gut should not extend > 1 cm into cord
- Herniation always midline
- Bowel returns to abdomen by 12 weeks gestation

Amniotic Band Syndrome

- Multiple body parts affected (especially limbs, head)
- Random "slash" defects
- May visualize bands with extremity constriction ± distal edema/amputations

PATHOLOGY

General Features

- Etiology
  - Numerous theories
    - Incomplete lateral fold closure
    - Mesenchymal defect
    - Abnormal involution of right umbilical vein
    - Omphalomesenteric (vitelline) artery occlusion with necrosis at right side of umbilical ring
    - In utero rupture of umbilical hernia
  - Possible teratogens: Aspirin, pseudoephedrine, acetaminophen, cocaine, smoking, others
- Genetics
  - No known genetic basis for gastroschisis
    - Chromosomal abnormalities in < 2%
  - Familial cases uncommon

- 2-5% recurrence risk for siblings
- Associated abnormalities
  - Additional anomalies in up to 20%
    - Intestinal atresias 5-20% (jejunoileal > colonic)
    - Hydronephrosis, bladder herniation into AWD
    - Congenital cardiac defects
    - Limb hypoplasia, arthrogryposis
    - Cryptorchidism

Gross Pathologic & Surgical Features

- Defect usually < 4 cm
  - Tight, constricted defect, usually right of umbilicus
- In utero bowel injury mechanisms from AWD
  - Chronic irritation by exposure to amniotic fluid
    - Bowel thickening, coating by inflammatory fibrin peel
      - Dysmotility, ↑ risk of adhesions
    - Protein loss by bowel to amniotic fluid → IUGR
  - Constriction at AWD → "complex gastroschisis" (10-20%)
    - Vascular injury with necrosis, atresia, volvulus, perforation
      - Closed gastroschisis: AWD closes around prolapsed gut causing midgut infarction ± resorption (vanishing midgut); abdominal wall may be normal
    - ↑ rates of necrotizing enterocolitis, short gut
- Post-repair bowel insult
  - Necrotizing enterocolitis
  - Malrotation with adhesions
    - No significant risk of midgut volvulus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Recognized during prenatal US in 98% of cases in developed countries
    - ↑ α-fetoprotein in maternal serum (95%)
  - At delivery, herniated bowel inflamed with fibrin coating ("peel") in ~ 33%; bowel rarely necrotic

Demographics

- Gender
  - M:F = nearly 1:1
- Epidemiology
  - ↑ prevalence; up to 5:10,000 births
  - Maternal risk factors: Young age, low socioeconomic status, primigravida, poor nutrition, smoking, vasoconstrictive agents

Natural History & Prognosis

- Prematurity in up to 60%
- Overall infant survival > 90% (but ↓ in complex forms)
  - Sepsis most common cause of death
- 10-15% with persistent disability
  - Inguinal hernias due to ↑ intraabdominal pressure
  - Short-gut syndrome with malabsorption
  - Motility disorders
    - 10% incidence of hypoperistalsis syndrome
    - 50% have gastroesophageal reflux
  - Small bowel obstruction from adhesions post repair
    - 27% in 1st year; 37% by 10 years
  - Chronic abdominal pain

Treatment

- Antenatal
  - Close monitoring in 3rd trimester due to ↑ risk of intrauterine demise
  - Delivery at tertiary care center
  - Early delivery not routinely recommended
  - Without other obstetric indications, cesarean section has no advantage over vaginal delivery
- Postnatal
  - Protection of herniated bowel, IV fluid replacement & nutrition, IV antibiotics, thermoregulation
  - Prior to reduction, bowel must be inspected for atresias
  - Repair of AWD: Controversy over best method
    - Operative vs. sutureless umbilical cord flap
      - Sutureless method only if bowel not inflamed
    - Primary closure
      - Possible if intraabdominal pressure < 20 mm Hg (measured in stomach or bladder) when external contents returned to abdominal cavity
      - Excessive intraabdominal pressure (abdominal compartment syndrome) → hypotension, renal failure, bowel necrosis, respiratory compromise
    - Staged closure
      - Preformed silo provides gradual reduction of bowel to abdomen over 1st week of life
  - Rates of reported sepsis, NEC, reoperation, days on ventilator, & days until enteral feedings vary for each closure method depending on study
  - Total parenteral nutrition required until intestinal function returns

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Prenatally: Consider ruptured omphalocele if liver herniated into defect without covering membrane (alters prognosis)
- Postnatally: Many possible causes for vomiting + dilated bowel in repaired gastroschisis patients

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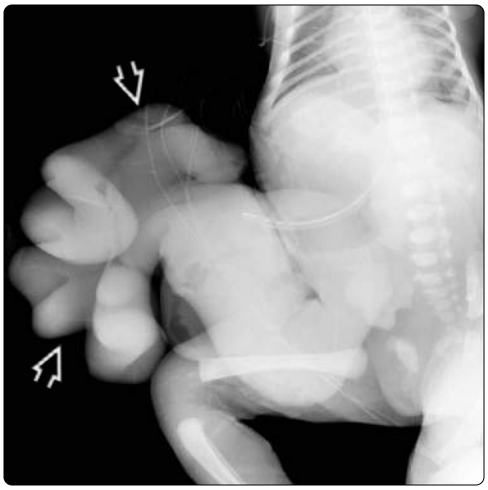
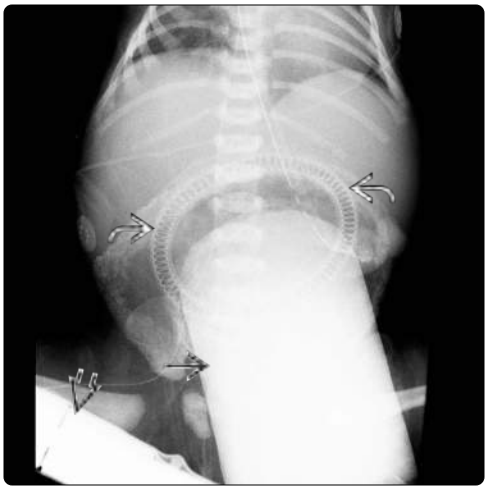
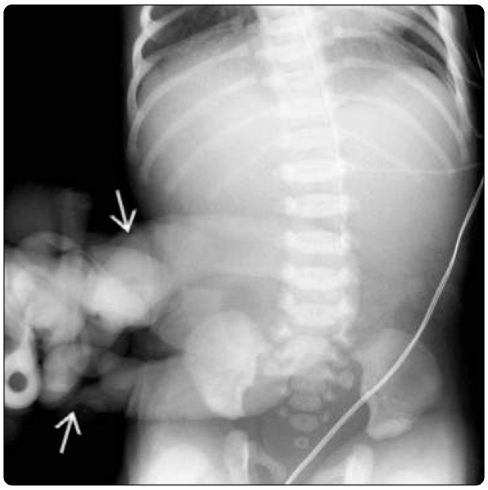
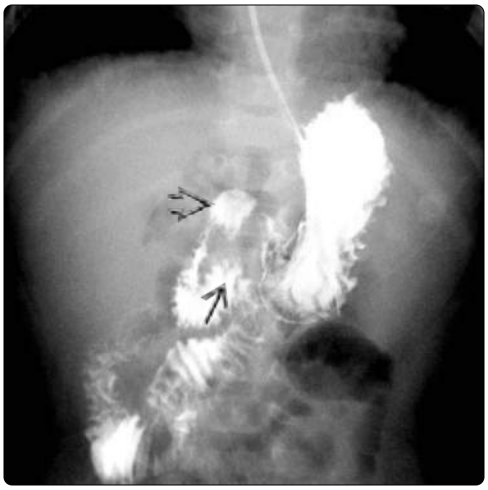


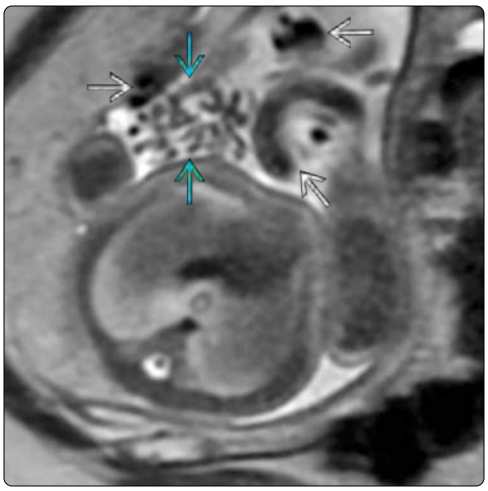


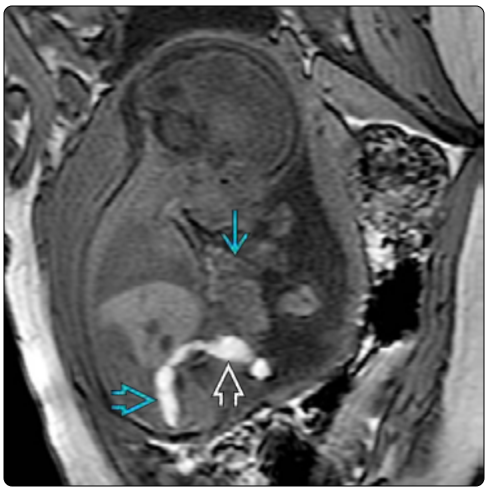
(Left) *Oblique radiograph in a newborn shows numerous dilated bowel loops ⇨ herniated from the abdominal cavity without a covering membrane.* **(Right)** *AP radiograph in the same patient shows containment of the herniated bowel loops within a silo bag ⇨ for gradual reduction into the peritoneal cavity. Note the characteristic metallic spring ⇨ within the AWD as well as the overlying bar ⇨ from which the bowel-containing silo is suspended.*





(Left) *AP radiograph in a newborn with gastroschisis shows numerous bowel loops ⇨ herniated from the abdominal cavity without a covering membrane.* **(Right)** *Frontal upper GI image in the same patient after repair of the gastroschisis shows that the duodenojejunal junction ⇨ is abnormally positioned right of the midline & below the duodenal bulb ⇨. This is consistent with malrotation, which is typical in gastroschisis as the herniated bowel does not undergo normal fixation in utero.*





(Left) *Axial SSFSE T2 MR in a 30-week gestation fetus shows numerous loops of uncovered bowel ⇨ herniated into the amniotic cavity. The loops lie between segments of the umbilical cord ⇨ & originated from an AWD at the right of the cord insertion (not shown). No dilated bowel is seen.* **(Right)** *Sagittal T1 MR in the same fetus with gastroschisis shows herniated meconium-containing sigmoid colon ⇨ coursing back into the abdomen with a normal rectal appearance noted ⇨. Herniated small bowel ⇨ is also seen superiorly.*

KEY FACTS

TERMINOLOGY

- Cloacal exstrophy: Complex abdominal wall defect (AWD) with exposed, abnormally persistent connection of genital, urinary, & intestinal tracts
- OEIS: **O**mphalocele, cloacal **e**xstrophy, **i**mperforate anus, **s**pinal anomalies

IMAGING

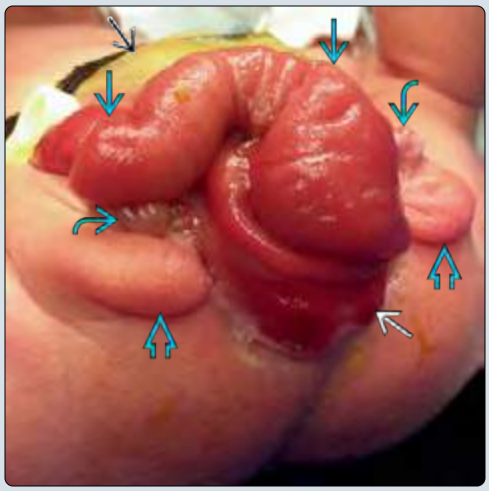
- Prenatal/prerepair findings
  - Absence of urinary bladder with normal to ↓ amniotic fluid volume (depending on renal/ureteral anomalies)
    - Protuberant exposed hemibladder plates at lower abdominal wall
    - Stenotic ureteral orifices may lead to hydroureteronephrosis
    - Additional genitourinary abnormalities: Various renal anomalies, hemiuteri/hemivaginas, ambiguous, &/or bifid genitals
  - Everted cecum splits bladder plates
    - ± elephant trunk appearance of prolapsed terminal ileum below umbilical cord
    - Short, blind-ending microcolon remains internal
      - Lacks normal T1-bright meconium signal
      - Imperforate anus noted clinically
  - Omphalocele above bladder plates; typically low & small
  - Splayed anterior pelvic bones with abducted thighs
  - Neural tube defects most commonly closed, not open
    - Terminal myelocystocele, lipomyelomeningocele
  - ± lower limb anomalies

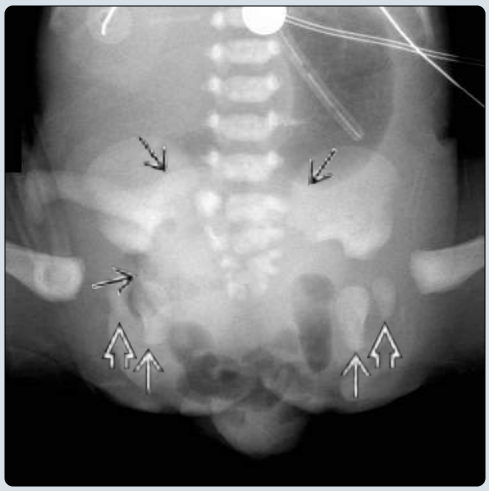
TOP DIFFERENTIAL DIAGNOSES

- Covered cloacal exstrophy
- Bladder exstrophy
- Limb body wall complex

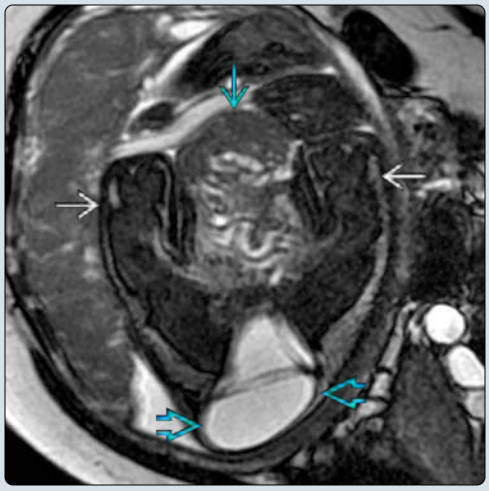
CLINICAL ISSUES

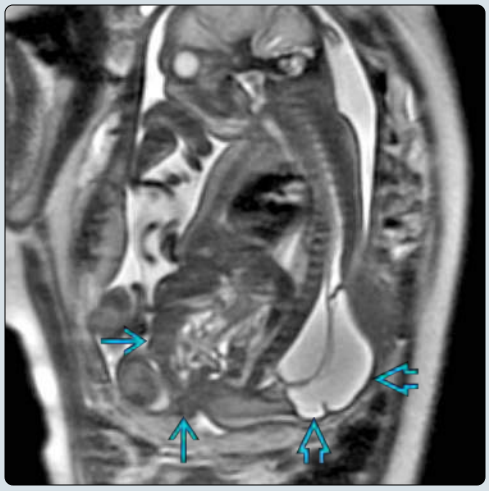
- Survival near 100%; long-term function/morbidity related to severity of individual anomalies





(Left) *Photograph of a newborn boy with cloacal exstrophy shows a bifid scrotum, bifid penis, prolapsed terminal ileum, everted cecum, & omphalocele membrane. No anal opening is visualized. The everted bladder plates & ureteral orifices are not well seen in this image.* **(Right)** *AP radiograph in the same patient shows the splayed ischial & pubic rami & low omphalocele. The sacrum is dysplastic.*





(Left) *Axial SSFP MR in a 31-week gestation fetus shows a low-lying, bowel-filled omphalocele between the fetal thighs. A complex skin-covered spinal dysraphic defect is noted. No urinary bladder is seen.* **(Right)** *Sagittal T2 SSFSE MR in the same fetus shows the low omphalocele & skin-covered spinal dysraphism (which was a variant of a terminal myelocystocele at neonatal surgical repair). No urinary bladder is seen, although the amniotic fluid volume is normal, typical of cloacal exstrophy.*

KEY FACTS

TERMINOLOGY

- Rare lethal malformation complex with large abdominal wall defect (AWD), limb anomalies, &/or craniofacial anomalies

IMAGING

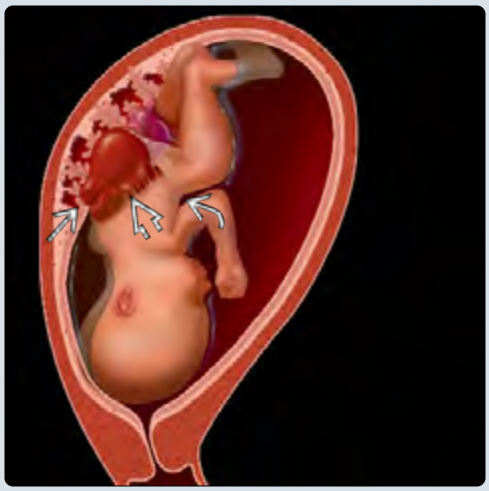
- Best imaging clue: Prenatal US/MR showing distorted fetus with AWD, fixation of extruded viscera to placenta, severe scoliosis, various limb anomalies, & short umbilical cord
- Spectrum of anomalies
  - Large AWD, may involve thorax
    - Herniated viscera include liver, bowel, stomach, bladder, ± kidney, spleen
    - Extruded organs not contained by membrane
    - Viscera often attached to placenta or uterine wall
  - Short or absent umbilical cord with 2 vessels
  - Severe scoliosis
  - Deformed, hypoplastic, or absent limb(s)
  - Persistent extraembryonic coelomic cavity
  - Craniofacial anomalies considered as diagnostic component by some definitions
    - Encephalocele, exencephaly, facial clefts
  - Many other associated anomalies reported
- Limited ex utero imaging: Typically autopsy radiographs

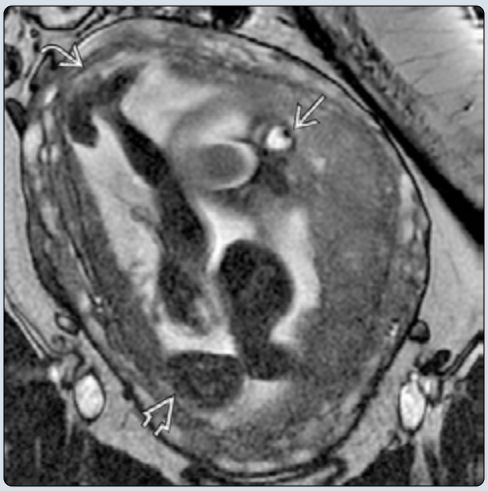
TOP DIFFERENTIAL DIAGNOSES

- Omphalocele
- Gastroschisis
- Amniotic band syndrome
- Pentalogy of Cantrell
- OEIS complex

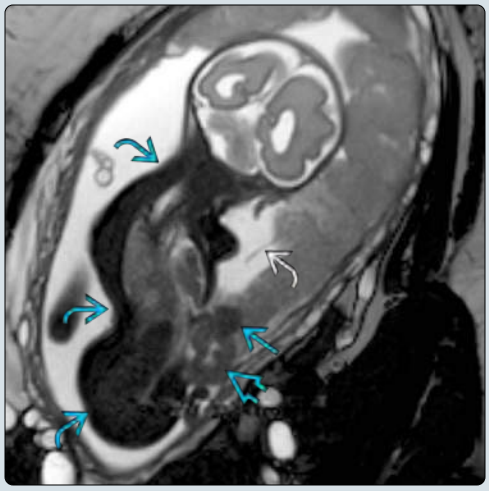
PATHOLOGY

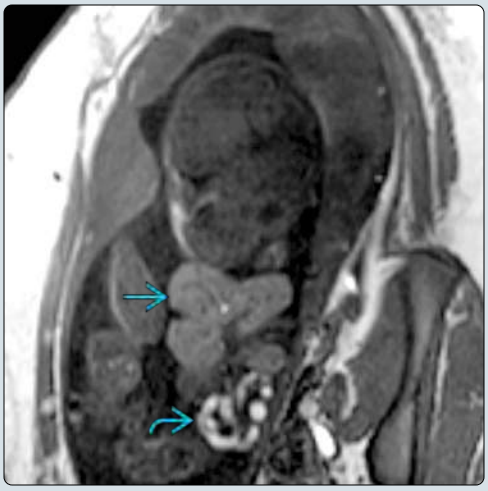
- Some classify 2 distinct LBWC phenotypes
  - Type 1: "Placento-cranial"
  - Type 2: "Placento-abdominal"





(Left) *Graphic shows a large body wall defect with fixation of the abdominal viscera to the placenta. No free-floating umbilical cord is seen. Attachment of the fetal body to the placenta results in trunk distortion & scoliosis.* **(Right)** *Sagittal oblique SSFP MR in a 23-week-gestation fetus shows a fixed 180° flexion & rotation of the lower extremities relative to the face due to a severe scoliosis. The herniated liver was partially adherent to the placenta in this fetus with limb-body wall complex (LBWC).*





(Left) *Coronal oblique SSFP MR in a 26-week-gestation fetus shows a fixed scoliotic curvature with extrusion of abdominal viscera & fixation of the left kidney & spleen to the placenta. An amniotic band was also noted.* **(Right)** *Coronal T1 MR in the same fetus shows extrusion of the entire liver & much of the meconium-containing bowel through a large abdominal wall defect into the amniotic cavity. LBWC was confirmed in this fetus that also had a short umbilical cord & limb anomalies (not shown).*

Appendicitis

KEY FACTS

TERMINOLOGY

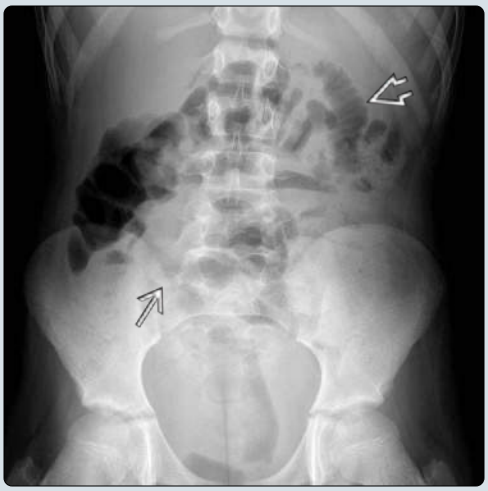
- Acute obstruction of appendiceal lumen → distention → ↑ intraluminal pressure → venous obstruction → ischemia → superimposed infection of appendiceal wall → eventual perforation

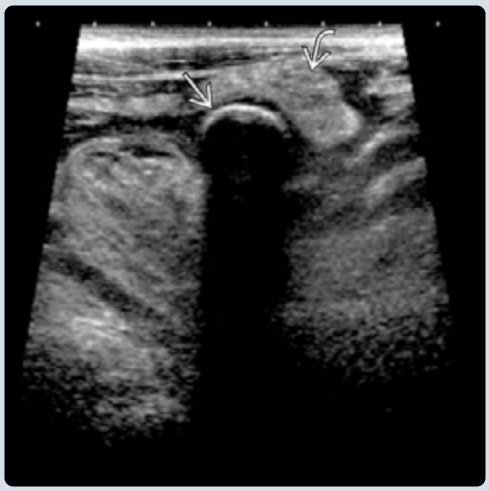
IMAGING

- US 1st-line modality in child with RLQ pain
  - Noncompressible, dilated, tubular blind-ending structure (appendix) in RLQ with induration of surrounding fat
    - ± echogenic, shadowing appendicolith
  - US diameter of appendix ≥ 6 mm (during compression) suggestive of acute appendicitis
- CT diameter of appendix > 6 mm found in 40% of normal patients; look for other inflammatory features
  - Appendiceal wall thickening & hyperenhancement
  - Periappendiceal inflammation with fat stranding & mild poorly defined fluid
  - RLQ focal ileus
- MR findings similar to CECT
- Features of perforation
  - Discontinuity of appendiceal wall
  - Appendicolith surrounded by inflammatory change but no well-defined appendix
  - Adjacent bowel wall thickening, moderate free fluid, & localized complex collections
- Staged approach gaining traction: US/MR > US/CT

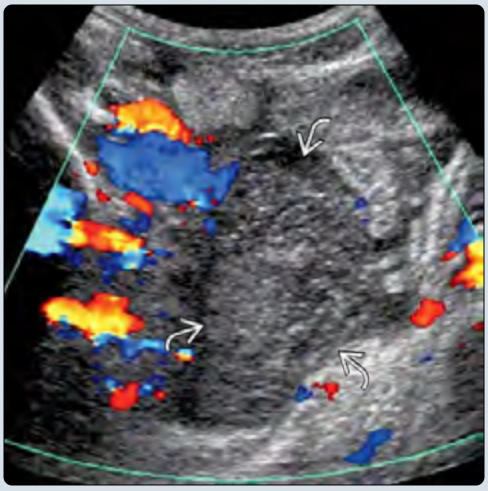
CLINICAL ISSUES

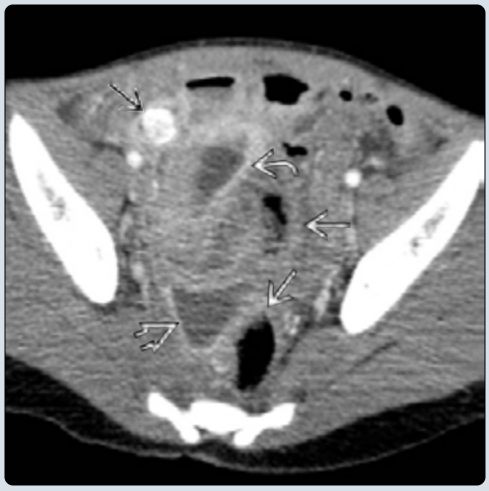
- Classic presentation: RLQ pain, anorexia, nausea, & vomiting
  - Periumbilical pain migrating to RLQ over 12-24 hours
  - Tenderness at McBurney point
  - Fever, guarding, rebound tenderness
- Clinical presentation nonspecific in up to 1/3 of patients
  - Especially young children
- Typically benign course if classic history leads to prompt surgery
- Morbidity & mortality ↑ with perforation





(Left) *AP radiograph in a 9 year old with right lower quadrant (RLQ) pain & vomiting shows an ovoid RLQ calcification ➡ suggesting an appendicolith. A few mildly prominent small bowel loops ➡ with air-fluid levels are noted in the left upper quadrant (LUQ), concerning for focal ileus.* **(Right)** *Transverse RLQ ultrasound in the same patient shows the round echogenic appendicolith ➡ with complete posterior acoustic shadowing (typical of calcification). Thickening & increased echogenicity of the surrounding fat ➡ is noted.*





(Left) *Transverse ultrasound in this same patient shows a poorly defined, heterogeneous RLQ fluid collection ➡ concerning for abscess.* **(Right)** *Axial CECT in the same patient shows the appendicolith ➡ & a thick-walled rim-enhancing abscess ➡ with surrounding inflammation. A similar posterior collection ➡ was also seen along the rectum. Adjacent bowel loops are displaced with wall thickening ➡. These findings are typical of perforated appendicitis.*

TERMINOLOGY

Definitions

- Acute obstruction of appendiceal lumen → distention → ↑ intraluminal pressure → venous obstruction → ischemia → superimposed infection of appendiceal wall → eventual perforation

IMAGING

General Features

- Best diagnostic clue
  - Noncompressible, dilated, tubular blind-ending structure (appendix) in right lower quadrant (RLQ) with induration of surrounding fat on ultrasound (US)
- Location
  - Various RLQ positions: Pelvic (draped over iliac vessels), retrocecal, subcecal, preileal, postileal
  - Underlying anomalies can lead to unusual locations
    - Other abdominal quadrants in setting of malrotation or situs inversus
    - Scrotum in setting of inguinal hernia (Amyand hernia)
    - Colonic lumen in setting of ileocolic intussusception (where inflamed appendix = pathologic lead point)
- Size
  - Mean diameter of normal pediatric appendix
    - ~ 4 mm on US, ~ 5.7 mm on CT, 5-6 mm on MR
  - US diameter of appendix ≥ 6 mm (during compression) suggests acute appendicitis
  - CT appendix diameter > 6 mm in 40% of normal patients
  - Differences in normal appendiceal diameters between US, CT, & MR may be attributable to mild degree of compression normally applied during routine US
- Morphology
  - Normal
    - Smooth continuous wall with "gut signature" on US: Echogenic mucosa & serosa with intervening hypoechoic muscular layer
    - Lies dependently, minimally undulating, often collapsed, may be coiled
  - Abnormal
    - Distended noncompressible lumen with taut or straight appendiceal configuration
    - Involvement of entire appendix vs. distal segment (tip)

Radiographic Findings

- Radiography
  - ± calcified appendicolith
  - Air-fluid levels with paucity of bowel gas in RLQ
  - Splinting/scoliosis
  - Loss of right psoas margin
  - Study may be normal
  - Abnormalities more likely with perforation
    - More diffuse bowel dilation with obstruction or ileus
    - RLQ extraluminal gas
    - Displacement of bowel loops from RLQ by abscess
    - Thickening of adjacent bowel wall

Ultrasonographic Findings

- Grayscale ultrasound
  - Noncompressible blind-ending tubular structure (extending from cecum) ≥ 6 mm in diameter
  - Echogenic appendicolith with posterior acoustic shadowing
  - Echogenic thickened periappendiceal fat (may be best independent predictor)
  - Free fluid in RLQ/pelvis (may be normal finding if small, anechoic, & isolated) vs. localized complex phlegmon or abscess (suggesting perforation)
  - US findings most suggestive of perforation include dilated or thick-walled adjacent bowel, loss of appendiceal wall integrity, fluid in at least 2 locations, complex fluid, & discrete abscess
  - Assessment may be limited by rigid abdomen, overlying bowel gas, or gas in abscess
- Color Doppler
  - ± hyperemia of appendiceal wall

CT Findings

- CECT
  - Dilated appendix (> 6-8 mm)
  - Appendiceal wall thickening & hyperenhancement
  - Periappendiceal inflammation with fat stranding & mild poorly defined fluid
  - Lack of appendiceal filling by enteric contrast despite oral or rectal contrast in cecum
  - Calcified appendicolith
  - RLQ lymphadenopathy
  - RLQ focal ileus
  - With perforation
    - Discontinuity of appendiceal wall
    - Appendicolith surrounded by inflammatory change without well-defined appendix
    - Localized phlegmon or fluid collections in RLQ or dependent pelvis (cul-de-sac) ± rim enhancement to suggest abscess
    - Extraluminal gas
    - Adjacent bowel wall thickening
    - Small bowel obstruction or diffuse ileus
    - Moderate free fluid & peritoneal enhancement with generalized peritonitis
  - Negative predictive value of normal CT with nonvisualized pediatric appendix ~ 99%

MR Findings

- Attractive alternative to CT due to lack of ionizing radiation
- Appendicitis features similar to CT
- Published appendix protocols vary regarding IV contrast administration; sensitivity/specificity > 94% regardless

Imaging Recommendations

- Best imaging tool
  - Staged approach gaining traction with US as 1st modality in patient with RLQ pain
    - US accurate in experienced hands
    - No ionizing radiation exposure
    - Negative predictive value > 90% with visualized normal appendix or nonvisualization of appendix without secondary findings of inflammation
  - Staged US/CT with contrast: Sensitivity ~ 99%, specificity > 90%; negative appendectomy rate of 8.1%

- Staged US/MR with contrast: Sensitivity ~ 100%, specificity ~ 99%; negative appendectomy rate of 1.4%
- Protocol advice
  - US with high-frequency transducer & graded compression (to displace overlying bowel)
    - Must look in typical & variant locations
      - Start scanning where patient endorses pain
    - Must visualize entire appendix
  - MR to include fast multiplanar T2-weighted sequences (e.g., SSFSE) ± FS
  - CECT often requested to determine extent of fluid collections prior to drainage procedure if perforation & fluid detected by US

DIFFERENTIAL DIAGNOSIS

Ovarian Pathology

- Hemorrhagic cyst, ovarian torsion, tuboovarian abscess
- Consider pelvic US

Meckel Diverticulum

- May become inflamed &/or cause obstruction

Inflammatory Bowel Disease

- Crohn disease & ulcerative colitis may primarily involve appendix (even in isolation)
- Bowel abnormalities usually more extensive
  - Not expected with nonperforated appendicitis

Omental Infarct

- Focal infarction of omental fat: Nonsurgical disease
- Focal inflammatory changes in anterior RLQ near colon

Cystic Fibrosis

- Average noninflamed appendix in CF > 8 mm, may be noncompressible
- No periappendiceal inflammation

Mucocele

- Dilated fluid-filled appendix

Fecal Impaction

- Appendiceal lumen distended by fecal material
- Nontender, no periappendiceal inflammation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - RLQ abdominal pain, anorexia, nausea, & vomiting
- Other signs/symptoms
  - Classic symptoms in older children without perforation
    - Periumbilical pain migrating to RLQ over 12-24 hours
    - Tenderness at McBurney point
    - Fever, guarding, rebound tenderness
  - Clinical presentation nonspecific in up to 1/3 of patients
    - Delay of diagnosis, higher perforation rate
    - More common in younger children

Demographics

- Age
  - Any age; ↑ incidence from ages 5-15 years
- Epidemiology
  - Most common reason for emergent surgery in children
  - 60,000-80,000 children treated in USA every year

Natural History & Prognosis

- Typically benign course if classic history leads to prompt surgery
- Morbidity & mortality ↑ with perforation
  - Up to 40% perforated at presentation; risk ↑ with delay in diagnosis
  - Adjacent abscesses form over days
- Delayed abscesses present weeks to years later with retained infected appendicoliths
  - Appendicolith can migrate/erode to extraperitoneal sites

Treatment

- Appendectomy by laparoscopy for early appendicitis
  - Limited data to support IV antibiotics without surgery in some cases of nonperforated appendicitis
- Perforated appendicitis with abscesses managed with IV antibiotics, percutaneous drainage, delayed appendectomy
  - Must remove appendicolith to prevent future abscess
- Open laparotomy for peritoneal washout if frank peritonitis

DIAGNOSTIC CHECKLIST

Consider

- US excellent modality for initial evaluation of RLQ pain
- MR seeing ↑ use, particularly in staged work-up
- CT in limited circumstances

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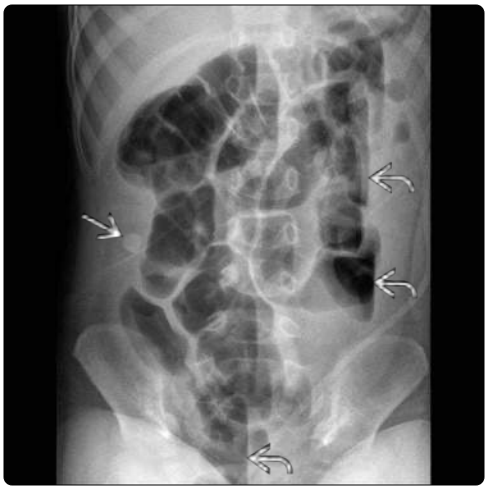
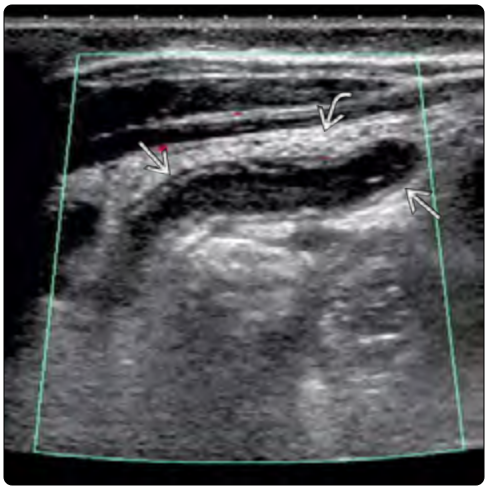
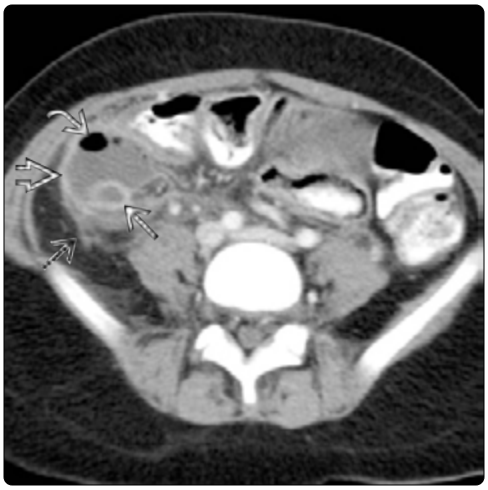
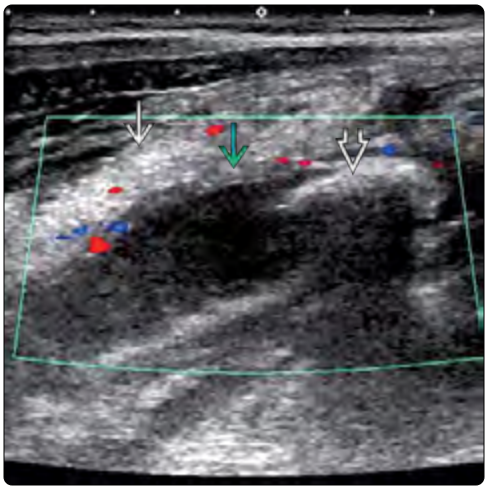


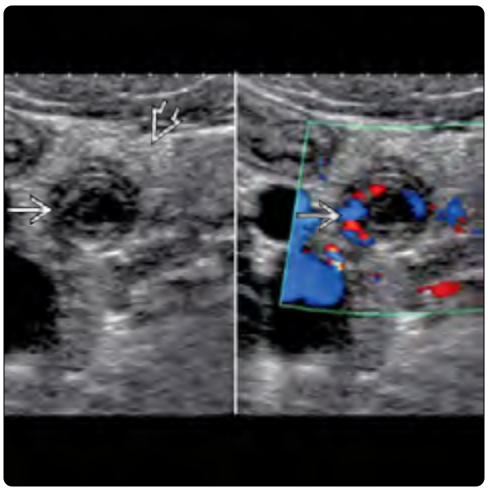


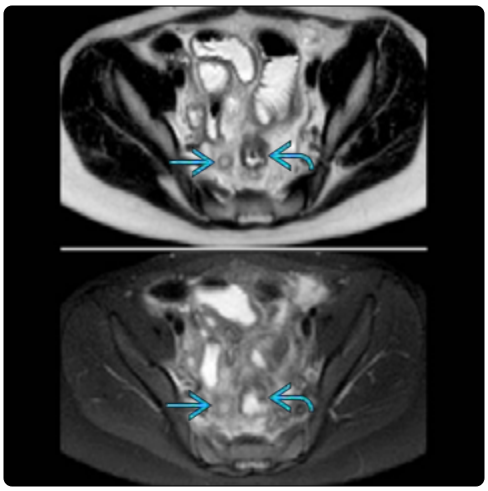
(Left) *Left side down decubitus abdominal radiograph in a 2 year old with RLQ pain & fever shows a large appendicolith ➡ with medial displacement of adjacent bowel loops. The bowel is mildly dilated diffusely & contains numerous air-fluid levels ➡, suggesting ileus.* **(Right)** *Longitudinal ultrasound of the RLQ in the same patient shows dilation (12 mm) of a noncompressible appendix ➡ with thickening & increased echogenicity of the periappendiceal fat ➡, typical of acute appendicitis.*





(Left) *Axial CECT in a 10 year old shows a dilated, hyperenhancing appendix ➡ with a surrounding rim-enhancing collection ➡ (which contains foci of gas ➡) & adjacent inflammatory stranding ⇨, consistent with perforated appendicitis & abscess.* **(Right)** *Color Doppler ultrasound in a 6 year old with pain & fever shows a dilated (16 mm) appendix with a thick & irregular wall ⇨, echogenic appendicolith ➡ with posterior shadowing, & indurated periappendiceal fat ➡. Appendiceal perforation was found at surgery.*





(Left) *Transverse grayscale & color Doppler ultrasound in a 9 year old with pain show a dilated appendix with a thickened, irregular & hyperemic wall ➡. The periappendiceal fat is indurated ➡ in this patient with acute appendicitis.* **(Right)** *Axial T2 SSFSE (top) & FS T2 (bottom) MR images in an 8 year old with pain show a dilated (9 mm), thick-walled appendix ⇨ with surrounding fat edema. Adjacent bowel-wall thickening ⇨ & a localized fluid collection (not shown) confirmed perforated appendicitis.*

Ileocolic Intussusception

KEY FACTS

TERMINOLOGY

- Invagination of distal small bowel (intussusceptum) into colon (intussuscipiens) in telescope-like manner

IMAGING

- US: Best diagnostic modality if clinically suspected
  - Round mass with target sign in right abdomen
  - Mean diameter of 2.6 cm (vs. 1.5 cm for purely small bowel intussusceptions)
  - Sweeping transducer proximal & distal shows relationship to small & large intestine
  - May see entrapped lymph nodes, appendix, other pathologic lead points (such as duplication cyst)
  - Entrapped fluid: ↑ failure rate of enema reduction
  - ↓ vascularity associated with ↑ likelihood of bowel necrosis & ↑ failure rate of reduction
- Radiography: Often abnormal, not always perceived
  - Paucity of right abdominal colonic gas ± round mass
    - ± fat density (from entrapped mesentery) in mass
  - Crescent sign: Curvilinear mass-gas interface
  - Lateralization of ileum to expected cecal location
  - ± small bowel obstruction
- Air enema reduction: Rush of air into small bowel → success

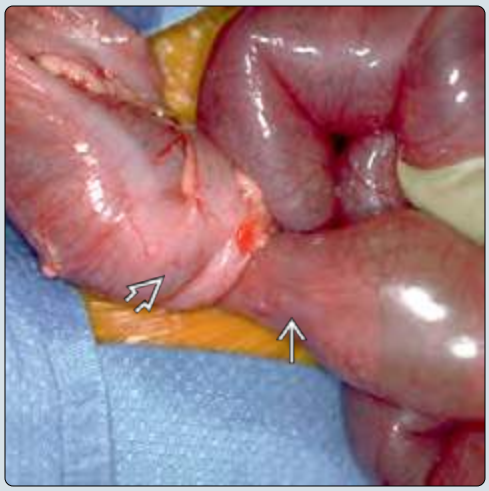
PATHOLOGY

- ~ 90% idiopathic (due to lymphoid hypertrophy)
  - May be preceded by viral illness
- ~ 5-10% from pathologic lead points

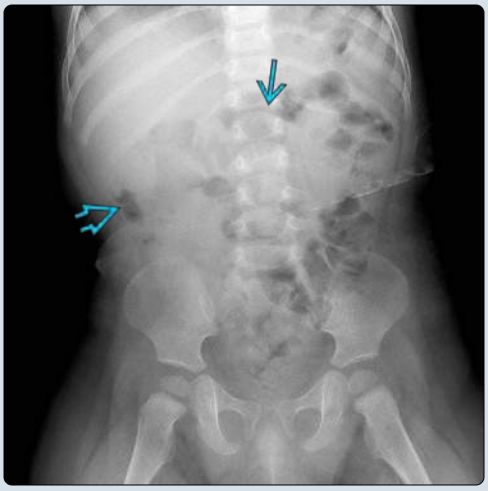
CLINICAL ISSUES

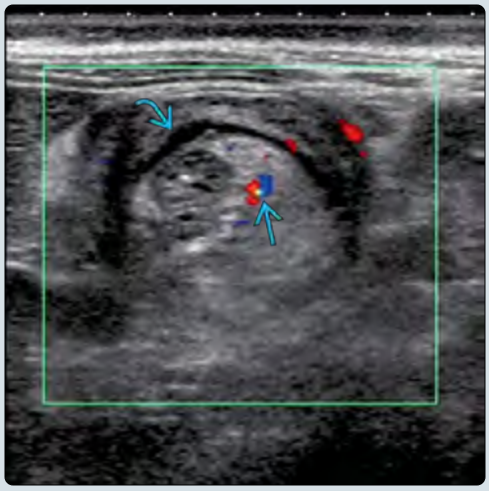
- Most common from ages 3 months to 3 years, presents with alternating lethargy & irritability, "currant jelly" stools
- Medical urgency: Bowel can infarct if not reduced
- Treat with enema reduction: Air enema under fluoroscopic guidance; hydrostatic with US guidance gaining acceptance
- Intussusception recurs after reduction in ~ 5-15%
- Surgery reserved for cases of enema reduction failure or when enema contraindicated

(Left) *Coronal graphic shows an ileocolic (IC) intussusception with the terminal ileum invaginating into the cecum & ascending colon. Note the vascular congestion of the intussusceptum.* **(Right)** *Photograph from an intraoperative IC intussusception reduction (after a failed enema reduction attempt) shows the distal ileum ➔ invaginating into the cecum ➔.*



(Left) *AP radiograph in a 12-month-old inconsolable male with recurrent abdominal pain shows a paucity of colonic gas in the right abdomen. There is a crescent sign ➔ (with a rim of colonic gas outlining the intussusceptum). A small bowel loop is seen in the far right abdomen ➔ (from the lateralization of the ileum).* **(Right)** *Transverse US of the right abdomen (in the same patient) shows a 3-cm diameter target sign with internal blood flow ➔, echogenic mesentery, small bowel, & fluid ➔ in this IC intussusception.*





TERMINOLOGY

Definitions

- Invagination of distal small bowel (intussusceptum) into colon (intussuscipiens) in telescope-like manner

IMAGING

General Features

- Best diagnostic clue
  - US: Round target sign in right abdomen at expected level of colon
  - Radiography: Paucity of colonic gas in right abdomen + crescent sign in colon + lateralization of ileum
- Location
  - Always involves proximal colon, extends distally to variable degrees
- Size
  - Mean diameter of 2.6 cm (vs. 1.5 cm for purely small bowel intussusceptions)

Radiographic Findings

- Paucity of colonic gas in right abdomen suggestive in right clinical setting
- Round mass in right abdomen: May see layers of fat density (from intussuscepted mesentery)
- Crescent sign: Curvilinear gas outlining rounded soft tissue mass (intussusceptum) within right or transverse colon
- ± small bowel obstruction
- Pneumoperitoneum before air enema extremely rare

Ultrasonographic Findings

- Grayscale ultrasound
  - Target sign: Cross section of intussusception shows round mass with alternating concentric rings of hyper- & hypoechogenicity representing bowel walls & mesenteric fat
    - Mean diameter of 2.6 cm
    - Sweeping transducer proximal & distal shows relationship to small & large intestine
      - May see entrapped lymph nodes, appendix, other pathologic lead points (such as intestinal duplication cyst)
  - Pseudokidney or sandwich sign: Longitudinal or oblique image showing ovoid mass with alternating layers of hyper- & hypoechogenicity
  - May not be located in right lower quadrant (RLQ) if intussusception has progressed distally (or in setting of underlying malrotation)
    - Need to scan all 4 quadrants
  - Trapped fluid within intussusception → ↑ failure rate of enema reduction
- Color Doppler
  - ↓ internal vascular flow → ↑ likelihood of bowel necrosis, ↑ failure rate of enema reduction

Fluoroscopic Findings

- Air-contrast enema
  - Apex of intussusceptum projects as round soft tissue mass against distal colonic air column
  - Multilobular intussusceptum in right colon/cecum suggests irreducibility
  - Dissection sign: Air surrounding length of intussusceptum predicts irreducibility
- Liquid contrast enema
  - Apex of intussusceptum projects as round soft tissue/lucent defect against distal colonic positive contrast column
  - Dissection or coiled spring sign: Contrast surrounding length of intussusceptum predicts irreducibility

CT Findings

- CECT
  - Incidentally seen on abdominal CT performed for nonspecific abdominal pain when intussusception not suspected
    - Not primary modality for diagnosis
  - Abdominal mass with alternating rings of high & low attenuation (target or sandwich sign)
  - May not be located in RLQ if intussusception has progressed distally

Imaging Recommendations

- Best imaging tool
  - US for diagnosis
  - Air enema for treatment

DIFFERENTIAL DIAGNOSIS

Appendicitis

- May present with similar symptoms of abdominal pain; typically older age group
- Identification of appendicolith helpful
- In cases of perforation, inflammatory collection can mimic soft tissue mass

Ovarian Torsion

- Can present as fussiness or pain in young child
- US may show avascular mass with peripheral cysts in midline with only 1 normal ovary visualized

Gastroenteritis

- Radiographs shows multiple air-fluid levels within mildly distended bowel loops
- Air-fluid levels throughout colon suggest gastroenteritis

Meckel Diverticulum

- May become inflamed, cause obstruction, cause GI bleed
- May serve as lead point for intussusception

PATHOLOGY

General Features

- Etiology
  - ~ 90% idiopathic (likely secondary to reactive lymphoid hyperplasia)
    - May be preceded by viral illness
  - ~ 5-10% caused by pathologic lead points in children
    - Meckel diverticulum > duplication cyst > polyp > lymphoma

Gross Pathologic & Surgical Features

- Telescoping of terminal ileum & ileocecal valve into cecum or ascending colon

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Alternating lethargy & irritability
  - Colic or "intermittent fussiness"
  - Palpable right-sided abdominal mass
- Other signs/symptoms
  - Bloody diarrhea ("red currant jelly" stools classic)
  - Crampy abdominal pain
  - Vomiting, may be bilious

Demographics

- Age
  - Classic: 3 months to 3 years
  - If < 3 months or > 3 years, question pathologic lead point
- Gender
  - M:F = 3:2
- Epidemiology
  - Most common cause of pediatric small bowel obstruction
  - ~ 56 per 100,000 children annually
  - Seasonal occurrence (classically winter, spring) with viral illnesses

Natural History & Prognosis

- Medical urgency: Bowel can infarct if not reduced
  - Bowel necrosis → perforation → peritonitis, shock, & death
- May spontaneously reduce
- Intussusception recurs after successful reduction in ~ 5-15%
  - Most recurrences within 48-72 hours

Treatment

- Imaging-guided pressure reduction treatment of choice: Air insufflation with fluoroscopic guidance most common
  - Liquid contrast under fluoroscopy less frequent
  - US-guided hydrostatic reduction gaining acceptance
- Contraindications: Peritonitis (relative), pneumoperitoneum
  - In developed countries, pneumoperitoneum prior to reduction attempt rare
    - Likely due to prompt presentation to medical care facility vs. developing countries
- US findings associated with ↓ success rate of enema reduction (but not contraindications)
  - Trapped fluid within intussusception, ↓ intussusceptum vascularity, presence of pathologic lead points, small bowel obstruction, younger age, rectal bleeding, prolonged duration of symptoms (> 24-72 hours)
- Preparation guidelines: Adequate hydration, IV access, physical examination, surgery consultation
- Air enema guidelines
  - Good rectal seal without leak
  - Maximum of 120 mm Hg sustained colonic pressure at rest (greater pressure spikes usually seen during crying or Valsalva)
- Intussusception encountered as round mass that moves retrograde toward cecum with air insufflation
- Intussusceptum most likely to get "stuck" at ileocecal valve
- Success: Rush of gas into small bowel + resolution of soft tissue mass
  - Edematous ileocecal valve may protrude into cecum, mimicking persistent intussusception: Follow clinically or by US
- If mass progresses on initial attempts but does not reduce beyond ileocecal valve, period of ~ 60 minutes may ↓ edema & ↑ chance of success
  - Repeat attempts with interval of 1 hour of waiting acceptable as long as patient stable & head of intussusception progressing more toward cecum with each attempt
  - Recurrences typically treated by enema up to 3x prior to considering surgical exploration for potential pathological lead point
- Success rates ~ 80% with enema reduction
- Risk of perforation 0.5-1.0%
- Surgery reserved for cases of enema reduction failure or when enema contraindicated

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Radiographs: Often abnormal, but signs of intussusception not always perceived
  - Lateralized ileum or redundant sigmoid may mimic air in cecum with false exclusion of intussusception
    - Sigmoid colon in RLQ on radiographs 43% of time
  - Left side down decubitus radiograph can help evaluate cecum (& exclude free air)
- US: Best test if intussusception suspected clinically

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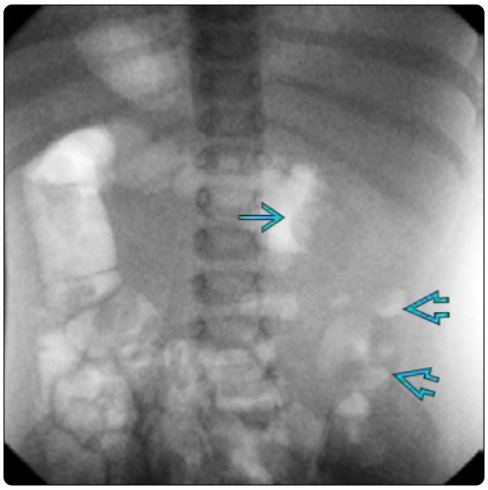
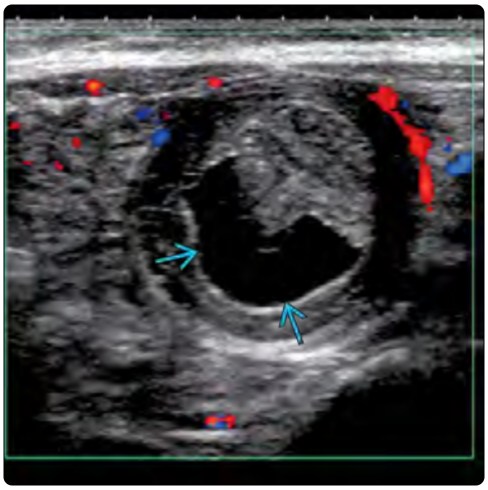
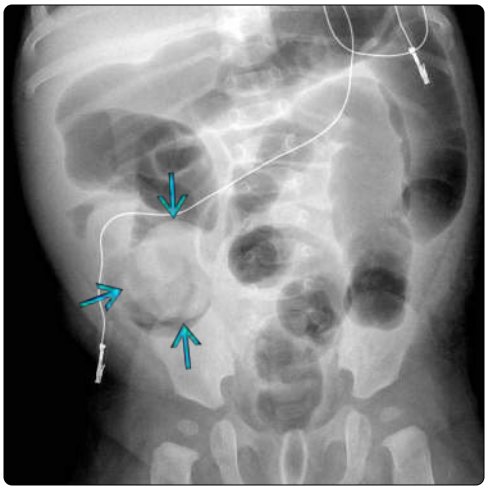
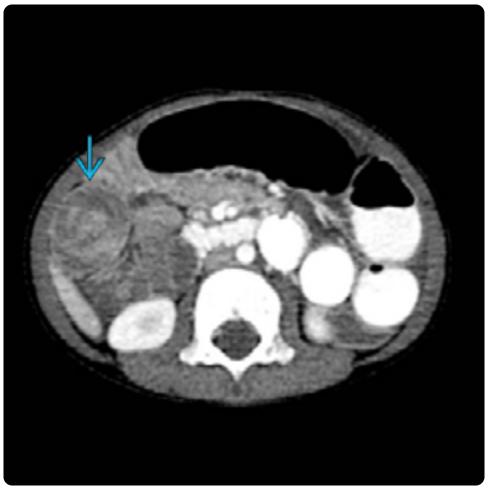


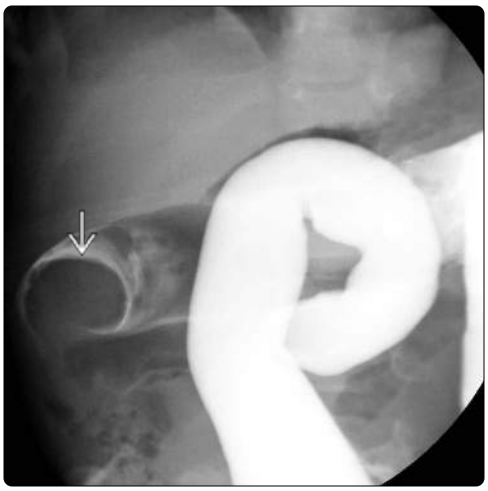


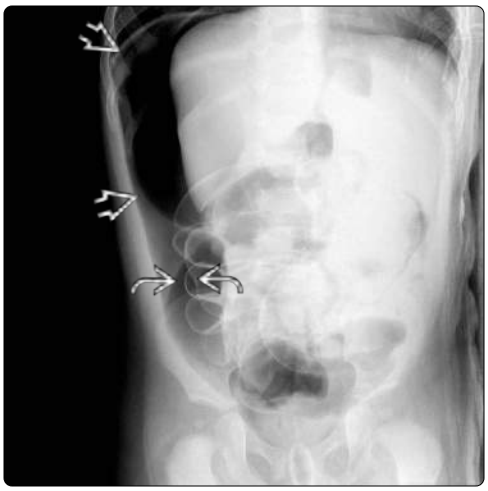
(Left) *Air enema performed prone in a patient with IC intussusception shows the head of the intussusceptum near the hepatic flexure. Note the air-filled small bowel loops drawn distally to the right lower quadrant (the lateralization of ileum sign of IC intussusception).* **(Right)** *Transverse color Doppler US in a 4 month old with lethargy & recurrent abdominal pain shows trapped fluid & no blood flow within the intussusceptum, suggesting the IC intussusception will be more difficult to reduce nonsurgically.*





(Left) *Supine AP radiograph in a 5 month old after multiple air enema reduction attempts shows a large multilobular residual soft tissue mass at the cecum. A multilobular configuration may suggest nonreducibility of the IC intussusception by enema.* **(Right)** *Axial CECT of a 3-year-old child with a small bowel obstruction & clinical findings suggestive of appendicitis shows an enhancing target sign in the right abdomen, consistent with an IC intussusception.*





(Left) *Supine image from a contrast enema reduction on a 3 month old shows the IC intussusception as a lucent filling defect outlined by small amounts of instilled contrast in the hepatic flexure.* **(Right)** *Left lateral decubitus radiograph in a 7 month old immediately after an unsuccessful air enema intussusception reduction attempt shows pneumoperitoneum consistent with bowel perforation. Note the Rigler sign (with gas outlining both sides of the bowel wall).*

Meckel Diverticulum

KEY FACTS

TERMINOLOGY

- Most common omphalomesenteric duct remnant
- Presents with bleeding, inflammation, intussusception, bowel obstruction, or perforation
  - ~ 65% of symptomatic Meckel diverticula contain ectopic gastric mucosa
- Rule of 2s: 2% of general population, found within 2 feet of ileocecal valve, most have symptoms < 2 years of age

IMAGING

- Classic imaging appearance (in patient with GI bleeding): Focal persistent accumulation of radiotracer in right lower quadrant on nuclear pertechnetate scan
  - Coincident with & isointense to gastric uptake
  - ↑ in visibility with time
- Other modalities (US, CT, MR)
  - Blind-ending tubular structure may be inconspicuous
  - May present as cyst, even with "gut signature"
  - With inflammation, findings similar to appendicitis
    - Thick-walled tubular structure, hyperemic bowel loops
    - Rare perforation
  - Intraluminal mass as lead point in intussusception
  - May see bowel obstruction without obvious cause

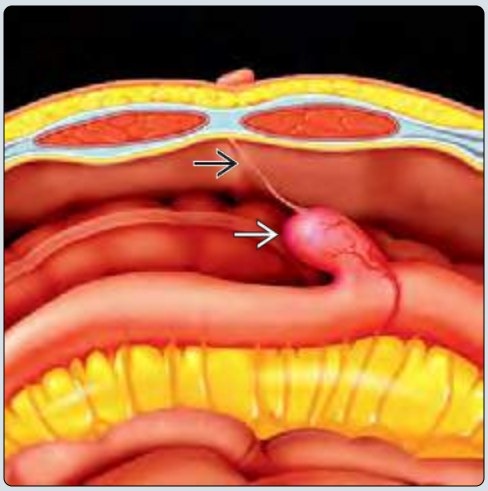
TOP DIFFERENTIAL DIAGNOSES

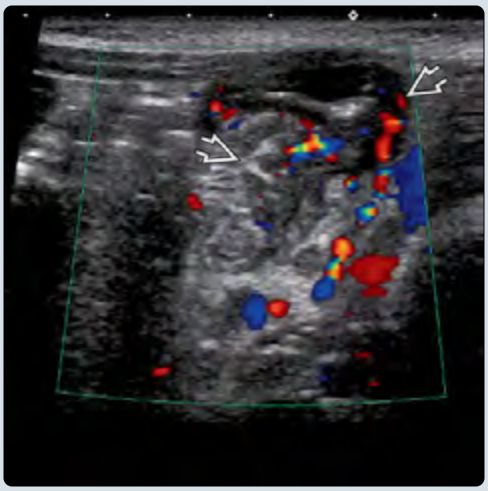
- GI bleeding with positive pertechnetate scan
  - Strong accumulation in GI duplication cyst
  - Hyperemia of Crohn disease, appendicitis, or vascular lesion may cause mild accumulation
- Small bowel obstruction
  - Appendicitis, adhesions, intussusception, inguinal hernia, malrotation (mnemonic of AAIIMM)
- Right lower quadrant inflammation
  - Appendicitis, Crohn disease, omental infarct, mesenteric adenitis, ovarian torsion

CLINICAL ISSUES

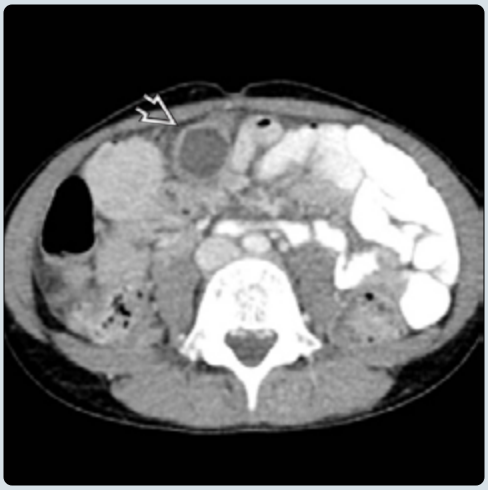
- Treated surgically

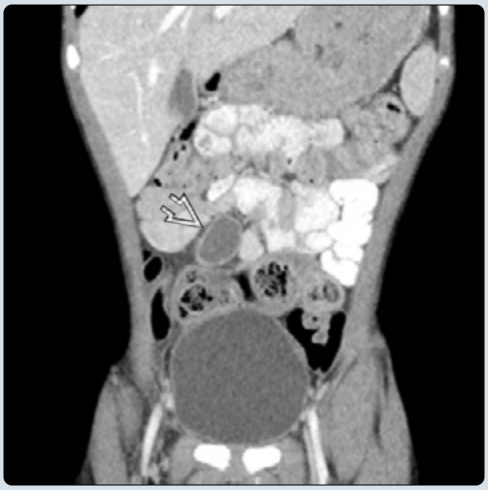
(Left) *Axial graphic shows an inflamed Meckel diverticulum ⇨ growing off of the antimesenteric border of the intestine with the obliterated remnant of the omphalomesenteric duct ⇨ extending from its tip.* **(Right)** *Longitudinal US in a 14-year-old boy with right lower quadrant (RLQ) pain demonstrated a normal appendix (not shown) plus a thickened, hyperemic tubular structure ⇨ just above the bladder. An inflamed Meckel diverticulum was removed at surgery.*





(Left) *Axial CECT in a 10-year-old boy with vague, intermittent abdominal pain shows a rim-enhancing cystic lesion ⇨ just deep to the umbilicus. This cyst did not appear to communicate with bowel.* **(Right)** *Coronal reformatted CECT in the same boy shows mild inflammation surrounding the cystic lesion ⇨, which was found to be a Meckel diverticulum distended with secretions at surgery.*





TERMINOLOGY

Definitions

- Remnant of omphalomesenteric duct
- Presents with bleeding (when containing ectopic gastric mucosa), inflammation, intussusception, bowel obstruction, or perforation
- Rule of 2s
  - Incidence: 2% of general population
  - Found within 2 feet of ileocecal valve
  - Most have clinical symptoms before 2 years of age

IMAGING

General Features

- Best diagnostic clue
  - Radiologic appearance varies by type of presentation
  - Classic imaging appearance on nuclear pertechnetate scan in patient with GI bleeding
    - Persistent, focal accumulation of radiotracer in right lower quadrant (RLQ) due to ectopic gastric mucosa in Meckel diverticulum
      - Coincident with & isointense to gastric uptake
      - Increasingly visible with time
- Location
  - RLQ or midline/periumbilical in location
  - Most within 2 feet of ileocecal valve
- Size
  - Variable; often small if uninflamed, large if inflamed or intussuscepted

Radiographic Findings

- Abdominal films may be normal or show
  - RLQ mass
  - Displacement of bowel loops
  - Small bowel obstruction
  - Enteroliths & ingested foreign bodies reported

Fluoroscopic Findings

- Often normal; variable communication with bowel lumen
- Contrast studies may show indirect evidence of mass & inflammatory changes in adjacent bowel
- May serve as lead point for intussusception
  - Often not reducible with air enema

Ultrasonographic Findings

- Grayscale ultrasound
  - Thick-walled tubular structure or hyperemic bowel loops in RLQ
  - Heterogeneous echotexture mass in RLQ, may mimic appendicitis
    - Look for normal appendix
  - Meckel diverticulum may present as cyst, even with "gut signature"
    - Walls more heterogeneous & thick with inflammation
  - Intraluminal mass as lead point in intussusception, "inverted Meckel"
- Color Doppler
  - Hyperemia related to inflammatory process

CT Findings

- CECT
  - Incidentally, may be collapsed & blind-ending or appear cystic with trapped secretions
  - If inflamed, similar to appendicitis: Thick-walled blind-ending structure near cecum with surrounding inflammation
    - If perforated, may see abscess & free air
    - Normal appendix clue to diagnosis
  - May cause small bowel obstruction with no clear explanation on imaging
  - If intussuscepted, may be occult
  - If bleeding, CTA may show ↑ flow locally

MR Findings

- Follows CECT findings; enterography may help visualize

Nuclear Medicine Findings

- Tc-99m pertechnetate scan
  - Most specific test for Meckel diverticulum: ~ 90% accuracy
  - Pertechnetate accumulates in mucous cells in acidic environment
    - Ectopic gastric mucosa of most Meckel diverticula
  - Diverticulum typically does not communicate with bowel lumen, so radiotracer does not appear to move downstream in bowel unless there is active bleeding
  - Pharmacologic enhancement of pertechnetate scans by using
    - Pentagastrin subcutaneously
    - Ranitidine or cimetidine, oral or intravenous
    - Glucagon intramuscularly
  - However, given high sensitivity otherwise, additional medications may be reserved for repeat studies in patients with high clinical suspicion of Meckel diverticular disease & normal initial scans
  - False-negative pertechnetate scans
    - Lack of any or sufficient gastric mucosa to localize radiotracer
      - ~ 65% of symptomatic Meckel diverticula have ectopic gastric mucosa
    - Secondary ischemia due to volvulus or intussusception
    - ~ 1/2 of scans repeated for high clinical suspicion are positive on 2nd study

Imaging Recommendations

- Best imaging tool
  - Tc-99m pertechnetate scan for GI bleeding
  - US or CECT for other presentations
- Protocol advice
  - Lateral & postvoid images should be obtained prior to conclusion of pertechnetate scan to look for Meckel diverticulum behind urinary bladder

DIFFERENTIAL DIAGNOSIS

GI Bleeding With Positive Pertechnetate Scan

- Strong accumulation in GI duplication cyst
- Hyperemia of Crohn disease, appendicitis, or vascular lesion may cause mild accumulation

Small Bowel Obstruction

- Appendicitis, adhesions, intussusception, inguinal hernia, malrotation (mnemonic of AAIIHM)
  - With Meckel diverticulum → AAIIMM

RLQ Inflammation

- Appendicitis, Crohn disease, omental infarct, mesenteric adenitis, ovarian torsion

PATHOLOGY

General Features

- Omphalomesenteric duct remnant (OMDR) found in 2-3% of autopsy series
  - Connection between yolk sac & primitive digestive tract in early fetal life
  - Meckel diverticulum most common end of spectrum of OMDRs, which also include umbilicoileal fistula, umbilical sinus or cyst, fibrous cord connecting ileum to umbilicus
- Small percentage of Meckel diverticula become symptomatic, typically due to presence of ectopic gastric mucosa
  - Rarely, diverticulum contains rests of pancreatic tissue

Microscopic Features

- Composed of same layers as adjacent small bowel but with addition of heterotopic gastric or pancreatic rests
- Risk of cancer: Malignant carcinoid, adenocarcinoma in older patients

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - GI bleeding: Bleeding occult or frank
- Other signs/symptoms
  - Abdominal pain with small bowel obstruction, intussusception, volvulus (including torsion of diverticulum), bowel perforation
    - Perforations reported from ingested fish bone, phytobezoar, button battery, etc.

Demographics

- Age
  - Most become symptomatic before 2 years of age
    - 60% come to medical attention before 10 years of age, with remainder of cases manifesting in adolescence & adulthood
    - Older patients more likely to present with intussusception or small bowel obstruction than with GI bleeding
- Gender
  - M = F in true incidence
  - Bleeding & other symptoms/complications are more common in male patients

Treatment

- Surgical resection; incidental appendectomy usually also performed
- Meckel diverticula generally removed when found incidentally on imaging or in operating room
  - ↑ detection on double balloon (push pull) endoscopy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Tc-99m pertechnetate scan: Positive in Meckel diverticula containing gastric mucosa
- Consider OMDR in small bowel obstruction otherwise unexplained on imaging & history

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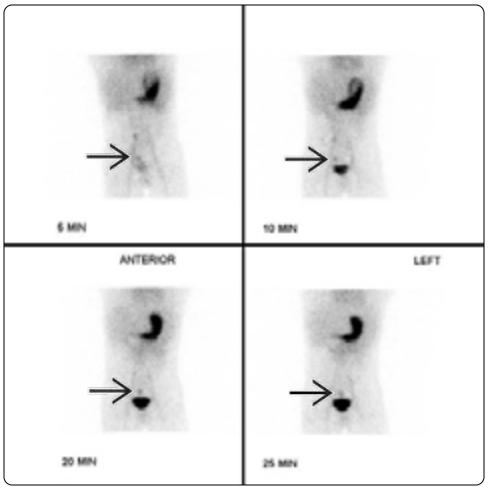
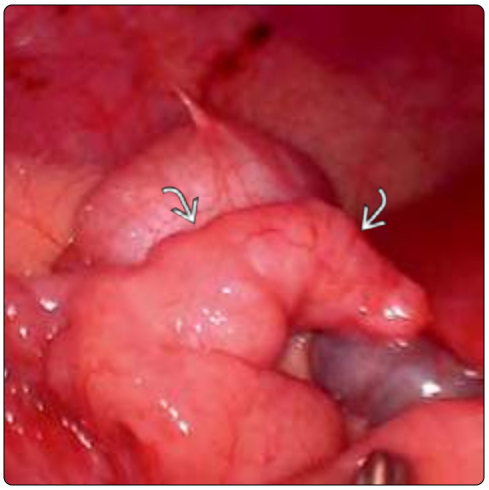
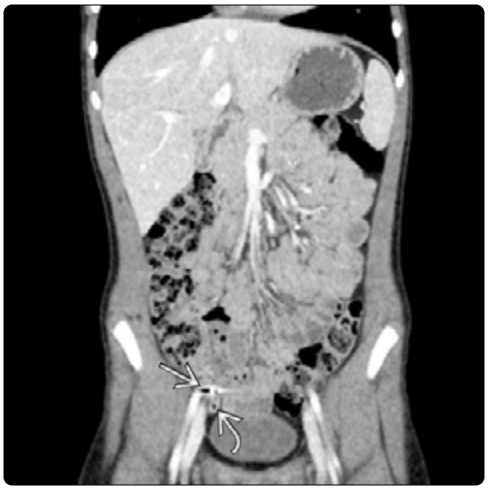
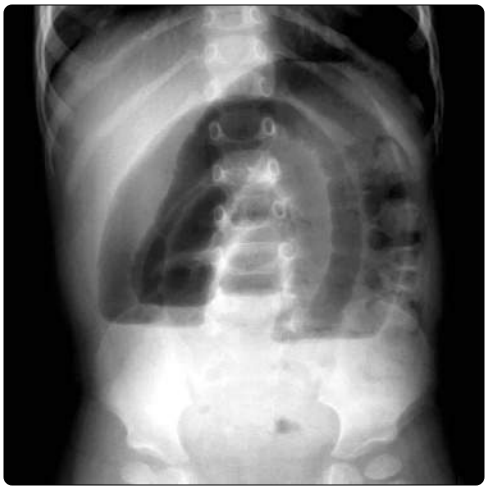


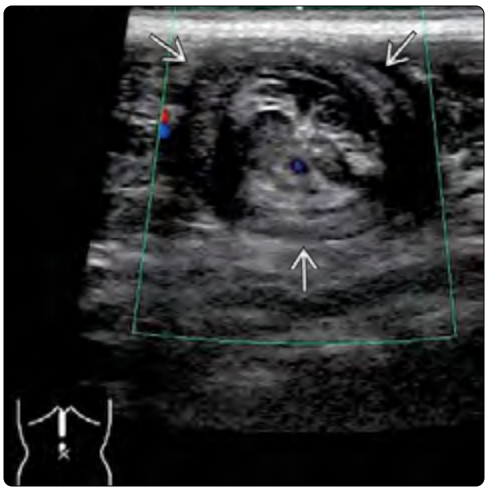


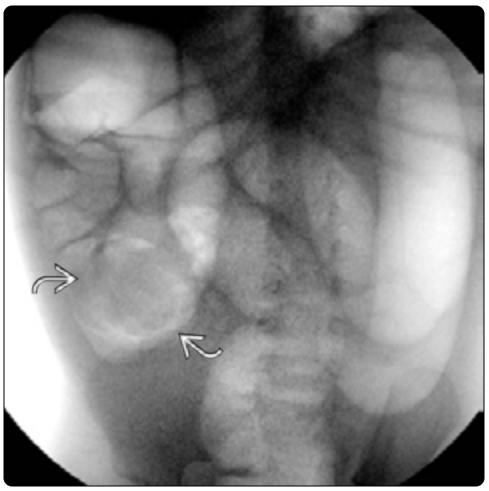
(Left) *Anterior images of a 9 year old with intermittent bloody stools show a persistent focus of radiotracer accumulation ⇨ above the urinary bladder, surgically confirmed to be Meckel diverticulum containing gastric mucosa.* **(Right)** *Intraoperative photograph in a patient with a history of GI bleeding shows an inflamed, blind-ending Meckel diverticulum ⇨.*





(Left) *Coronal CECT in a 10-year-old child with mild abdominal pain & no prior surgical history shows a metal artifact ⇨ in the RLQ, just superior to a normal appendix ⇨. At surgery, an ingested metal BB was found in a mildly inflamed Meckel diverticulum (which was resected).* **(Right)** *AP radiograph in a young child shows dilated small bowel loops with differential air-fluid levels & a paucity of colonic gas, consistent with a small bowel obstruction. An obstructing omphalomesenteric duct remnant was found at surgery.*





(Left) *Longitudinal color Doppler US of the upper abdomen in a 19 month old with intermittent fussiness & drawing up of his legs shows concentric layers of fat & bowel wall ⇨, consistent with the target sign of intussusception. There is minimal blood flow centrally within the intussusceptum.* **(Right)** *Air reduction enema in the same patient shows a persistent, nonreducible mass ⇨ at the ileocecal valve, which was found to be an inverted Meckel diverticulum in the operating room.*

Colonic Volvulus

KEY FACTS

TERMINOLOGY

- Colonic volvulus: Twisting of mobile segment of colon → obstruction
  - Frequency: Sigmoid > cecum > transverse colon

IMAGING

- Radiographs: Upper abdominal dilated loop of unusual size & shape for small bowel
  - Cecal volvulus often rounded
  - Sigmoid volvulus often bean-shaped (1/3 of cases)
  - Paucity of distal colonic gas
- Water-soluble contrast enema (WSCE): Bird's beak shape to proximal contrast at site of colonic twisting
  - Contrast fails to pass proximal to twist
    - Does not opacify dilated loop
- CECT: Whirl sign of twisted mesocolon encircling vascular pedicle that serves affected colonic segment
  - May have ↓ enhancement of involved segment due to vascular compromise
    - ± bowel wall thickening, pneumatosis, mesenteric edema, free or trapped fluid
- Protocol advice: If initial radiographs worrisome for colonic volvulus → WSCE for definitive diagnosis; if pneumatosis & ill patient → CECT

TOP DIFFERENTIAL DIAGNOSES

- Ileosigmoid knot
- Toxic megacolon
- Colonic pseudoobstruction

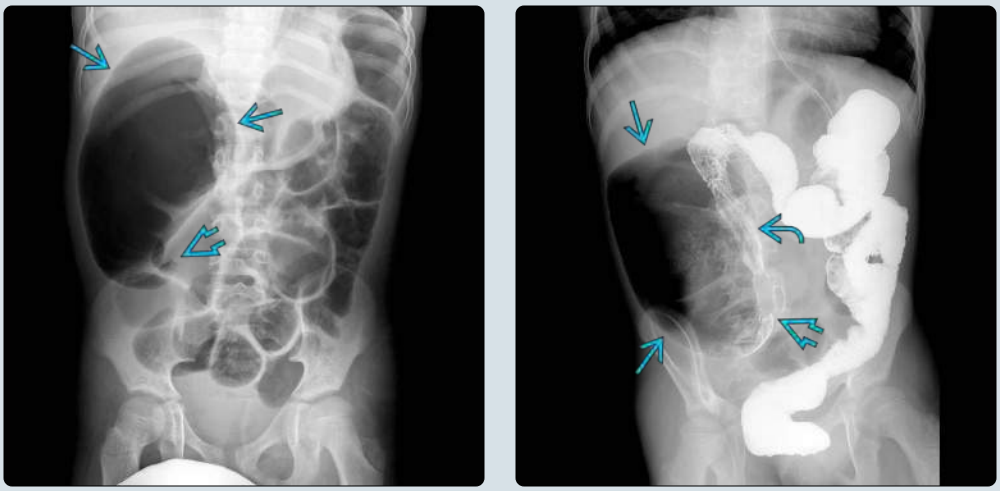
CLINICAL ISSUES

- Nonspecific abdominal distention & pain, vomiting

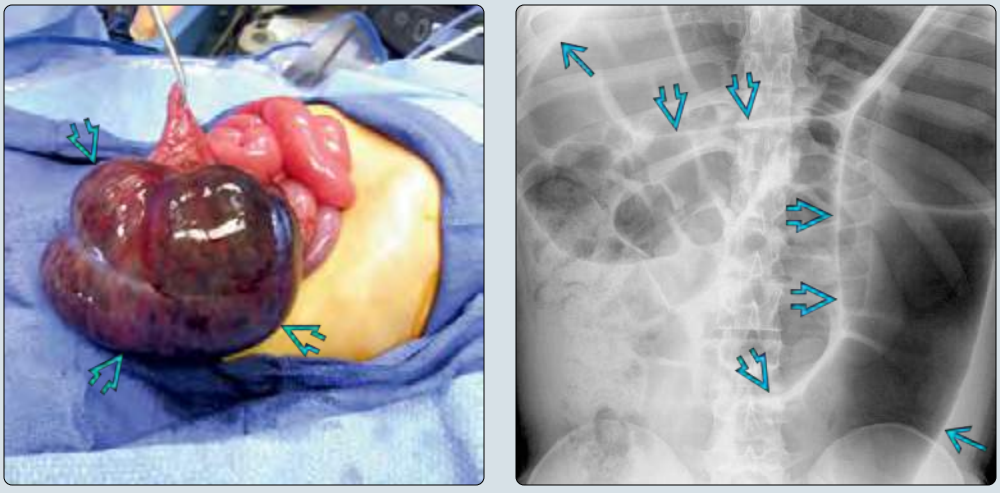
DIAGNOSTIC CHECKLIST

- Predisposed patients often have chronic constipation &/or chronically dilated bowel loops
- Suspicion for volvulus requires careful assessment of new radiograph for changes compared to prior radiographs that showed chronic bowel dilation

(Left) *AP radiograph in a 12-year-old developmentally delayed boy with acute abdominal pain shows a very dilated bowel loop in the right upper quadrant. The loop has a beak configuration at its inferior aspect, highly suggestive of a colonic volvulus. A contrast enema was recommended.* **(Right)** *Frontal radiograph of the same patient after a water-soluble contrast enema shows narrowing of the ascending colon, which ends proximally in a twist, leading to upstream dilation in this cecal volvulus.*



(Left) *Surgical photograph of a 2 year old with cerebral palsy & acute abdominal pain who had a preceding enema (not shown) worrisome for cecal volvulus shows a large necrotic cecum, which was extracted from the left upper quadrant & resected.* **(Right)** *AP radiograph of a 14 year old with acute abdominal pain shows a dilated closed loop of sigmoid colon. The centrally apposed colonic walls create the coffee bean sign. This appearance can be seen in almost 1/3 of sigmoid volvulus cases (as in this patient).*



TERMINOLOGY

Synonyms

- Cecal volvulus, sigmoid volvulus, transverse colon volvulus

Definitions

- Colonic volvulus: Twisting of mobile segment of colon
- Very rare in children

IMAGING

General Features

- Best diagnostic clue
  - Radiographs: Upper abdominal dilated loop of unusual size & shape for small bowel
    - Cecal volvulus often rounded
    - Sigmoid volvulus often bean-shaped
  - Water-soluble contrast enema (WSCE): Bird's beak shape to proximal contrast at site of colonic twisting
- Location
  - Sigmoid colon > cecum > transverse colon

Radiographic Findings

- Cecal volvulus: Abnormally positioned, focally dilated & rounded cecum
  - ± small bowel air-fluid levels & paucity of distal colonic gas
- Transverse colon volvulus: Gas-distended central colonic segment with paucity of distal colonic gas
- Sigmoid volvulus: Distended, twisted, ovoid closed loop; characteristic coffee bean sign in up to 1/3 of cases

Fluoroscopic Findings

- Contrast enema
  - Bird's beak shape to proximal contrast head at site of twisting on WSCE
  - Contrast fails to pass proximal to twist to opacify dilated loop

CT Findings

- CECT
  - Whirl sign: Twisted mesocolon encircling vascular pedicle that serves affected colonic segment
  - Dilated & obstructed segment of colon
    - Transverse & sigmoid segments usually show closed loop obstruction
    - Cecum shows twisted tubular segment
  - Colon distal to obstruction decompressed
  - May have ↓ enhancement of involved segment due to vascular compromise
    - ± bowel thickening, pneumatosis, mesenteric edema, free or trapped fluid

Imaging Recommendations

- Best imaging tool
  - WSCE
- Protocol advice
  - If radiographs suggest colonic obstruction with possible volvulus → WSCE for definitive diagnosis
  - If pneumatosis present, patient ill-appearing, or diagnosis not suggested on radiography, consider CECT

DIFFERENTIAL DIAGNOSIS

Ileosigmoid Knot

- Ileum & sigmoid become entangled → knot → vascular compromise
- Acute abdominal pain & rapid shock

Colonic Pseudoobstruction

- Acutely or chronically dilated large > small bowel without identifiable mechanical obstruction

Toxic Megacolon

- Ill patient with dilated, thick-walled colon
- Infectious causes or ulcerative colitis classic

PATHOLOGY

General Features

- Etiology
  - Predisposition in redundant & dilated mobile segments
  - ↑ risk with ↑ colonic distention, constipation, prior surgery, or malrotation
- Associated abnormalities
  - Sigmoid volvulus
    - Hirschsprung disease (18%), omphalomesenteric abnormalities, bowel malrotation, anal stenosis, chronic constipation, postoperative adhesions, prune-belly syndrome, & developmental delay

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Nonspecific abdominal distention & pain
- Other signs/symptoms
  - Vomiting, constipation, diarrhea

Treatment

- Endoscopic reduction success: Sigmoid > > cecal volvulus
- Even with endoscopic detorsion, affected colonic segment typically resected to prevent recurrence
  - High recurrence rate with pexy procedures

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Predisposed patients often have chronic constipation &/or chronically dilated bowel loops
- Suspicion for volvulus requires careful assessment of new radiograph for changes compared to prior radiographs that showed chronic bowel dilation

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1. Garel C et al: Diagnosis of pediatric gastric, small-bowel and colonic volvulus. Pediatr Radiol. 46(1):130-8, 2016
2. Perrot L et al: Management of the colonic volvulus in 2016. J Visc Surg. ePub, 2016
3. Colinet S et al: Presentation and endoscopic management of sigmoid volvulus in children. Eur J Pediatr. 174(7):965-9, 2015
4. Chen MH et al: Transverse colon volvulus presenting as 'inverted' coffee-bean sign. Arch Dis Child. 97(2):123, 2012

KEY FACTS

TERMINOLOGY

- Herniation of bowel through mesenteric defect, congenital band, or normal anatomic opening
- Transmesenteric hernia: Congenital mesenteric defect, usually near ileocecal valve or ligament of Treitz
  - Most common in children
- Acquired: Postsurgical (iatrogenic) or posttraumatic
- Other types of internal hernias more common in adults
  - Foramen of Winslow, paraduodenal, pericecal, transomental

IMAGING

- Best imaging modality: CECT
  - Dilated small bowel loops often localized to 1 region of abdomen
  - Closed loop obstruction suggested by decompressed distal & proximal bowel loops
  - Vascular compromise/ischemia suggested with
    - Mesenteric edema, free fluid (nonspecific)
    - ↓ bowel wall enhancement, pneumatosis (more specific)
- Finding exact location of obstruction less important than recognizing signs of vascular compromise

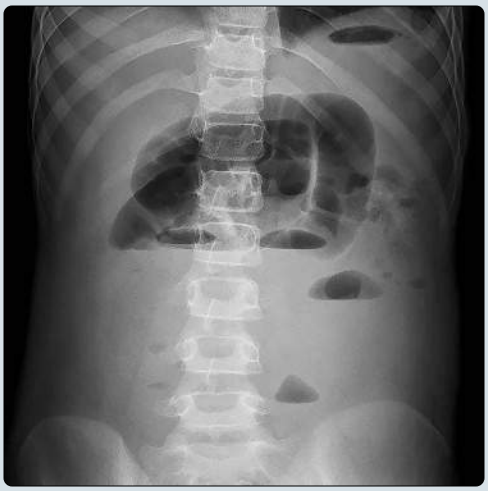
TOP DIFFERENTIAL DIAGNOSES

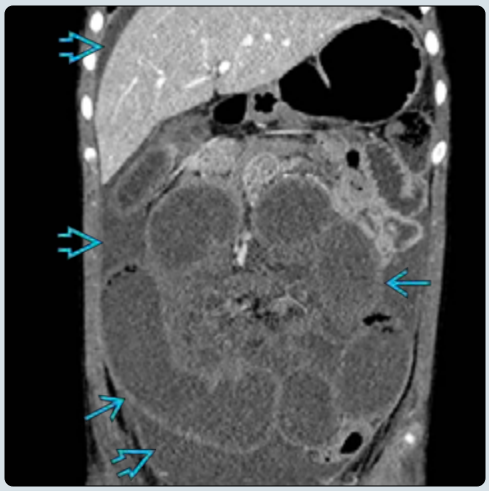
- Obstruction from other causes (AAIIMM mnemonic)
- Ileus
- Bowel injury

CLINICAL ISSUES

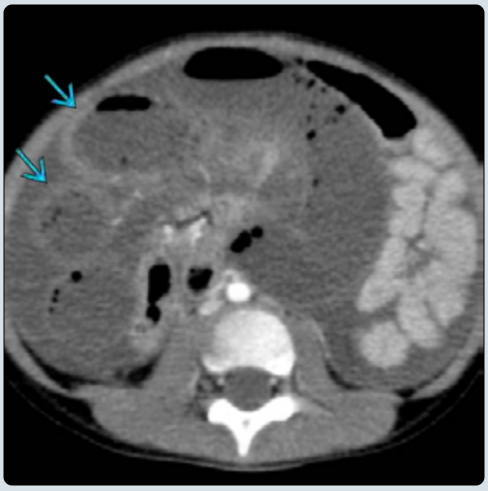
- Presents with vomiting, distention, & other clinical signs of small bowel obstruction
  - Symptoms of obstruction occasionally intermittent as bowel moves in & out of hernia
  - Signs of shock if vascular compromise & bowel necrosis present
- Surgical treatment: Reduction of hernia, resection of necrotic bowel, & repair of underlying mesenteric defect (if present)

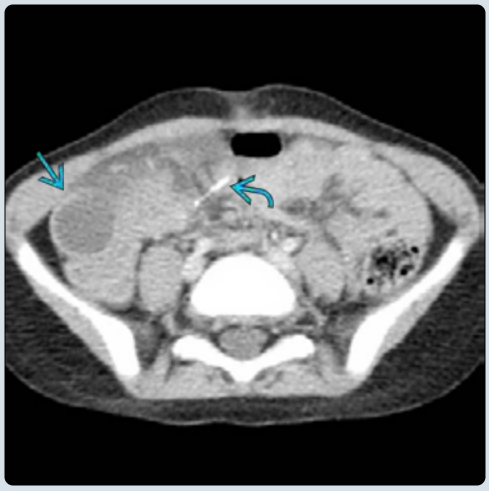
(Left) *Upright abdominal radiograph in a 7-year-old boy with vomiting shows dilated small bowel loops with air-fluid levels, consistent with a bowel obstruction.* **(Right)** *Coronal CECT in the same patient shows dilated small bowel loops with hypoenhancing walls & extensive intraabdominal free fluid, consistent with a bowel obstruction complicated by ischemia. A congenital mesenteric defect with internal hernia was found at surgery.*





(Left) *Axial CECT in a 4-year-old girl with abdominal distention demonstrates free fluid & dilated small bowel loops (with wall hypoenhancement) clustered in the right lower quadrant. These findings suggest ischemic bowel, & a transmesenteric internal hernia was found at surgery.* **(Right)** *Axial CECT in a 5 year old with pain & vomiting after appendectomy shows a suture line with adjacent edematous mesenteric fat & a dilated small bowel loop due to an iatrogenic internal hernia.*





KEY FACTS

TERMINOLOGY

- Twisting of bowel segment (ileum > jejunum) resulting in obstruction ± ischemia
- Segmental small bowel volvulus usually due to lead point of mass lesion or adhesive band, not malrotation

IMAGING

- Best clue: Dilated small bowel loops tapering into twist/swirl of collapsed bowel & vessels
- Radiographs
  - Dilated bowel loops with air-fluid levels
  - ± pneumatosis, pneumoperitoneum
- UGI/SBFT or water-soluble contrast enema
  - Spiral appearance of bowel loops at transition point, indicating volvulus
- CECT or ultrasound
  - Dilated proximal small bowel
  - Tapering of dilated bowel into vertical "whirlpool" or "swirl" of collapsed small bowel & vessels
    - Appearance accentuated by scrolling/sweeping superior to inferior through transition zone
  - Bowel wall ischemia suggested by bowel wall thickening, ↓ enhancement (CT) or color flow (Doppler), mesenteric edema, ascites, pneumatosis, pneumoperitoneum

PATHOLOGY

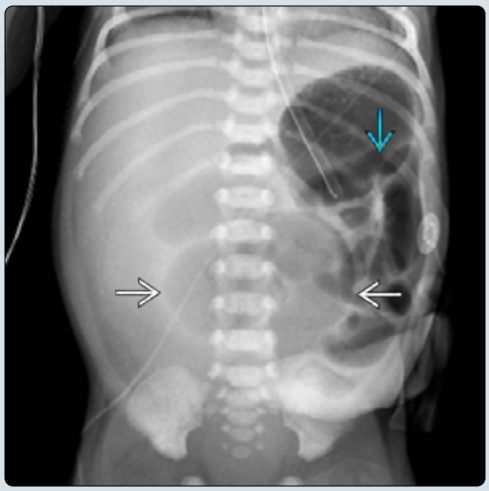
- Underlying lead points: Congenital band, meconium ileus, duplication cyst, Meckel diverticulum, lipoma, lymphatic malformation, postoperative adhesion, foreign body (e.g., ingested magnets)

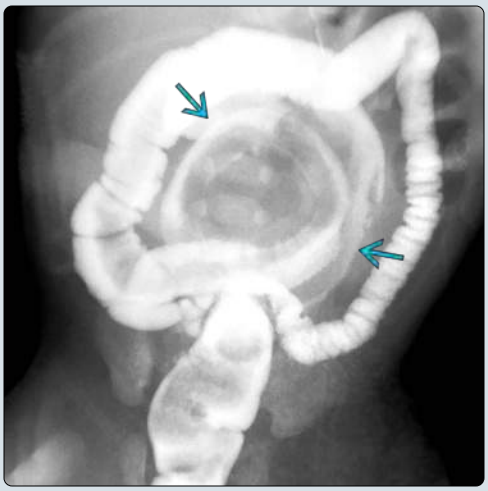
CLINICAL ISSUES

- Presentation: Abdominal pain, distention, nausea/vomiting
- Treatment: Surgical reduction (untwisting) of volvulus ± resection of necrotic bowel & lead point

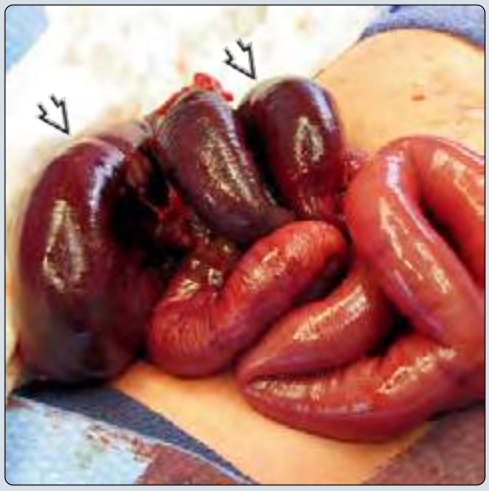
DIAGNOSTIC CHECKLIST

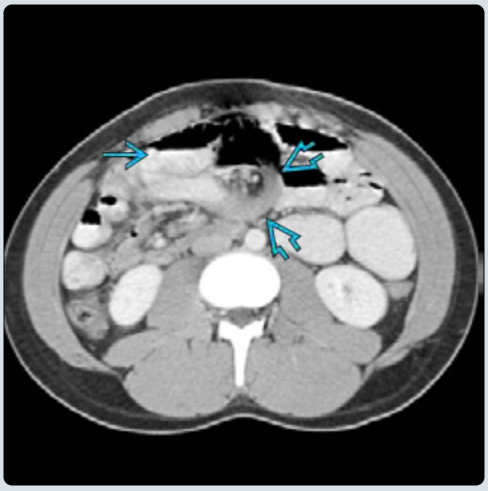
- Look for lead point & signs of vascular compromise





(Left) *Supine abdominal radiograph in a 3-day-old girl with vomiting shows dilated, gas-filled small bowel loops being displaced into the left abdomen ⇨ by a cluster of mildly lucent dilated bowel loops in the central abdomen ⇨. There is no bowel gas in the right abdomen. These features suggest a distal obstruction.* **(Right)** *Water-soluble contrast enema in the same patient shows swirling of opacified distal small bowel loops in the midabdomen ⇨. Surgery confirmed an ileal volvulus around a congenital band.*





(Left) *Intraoperative photograph in the same 3-day-old girl shows twisting of the necrotic dilated small bowel loops ⇨. This ileal volvulus was due to a congenital band.* **(Right)** *Axial CECT in a 17-year-old boy with a history of neonatal repair of a proximal intestinal atresia, now presenting with vomiting, shows tapering & twisting/swirling ⇨ of small bowel loops at the transition point from dilated ⇨ to decompressed small intestine. At surgery, the volvulus was found to be secondary to an adhesion.*

Hepatoblastoma

KEY FACTS

TERMINOLOGY

- Malignant embryonal hepatic tumor

IMAGING

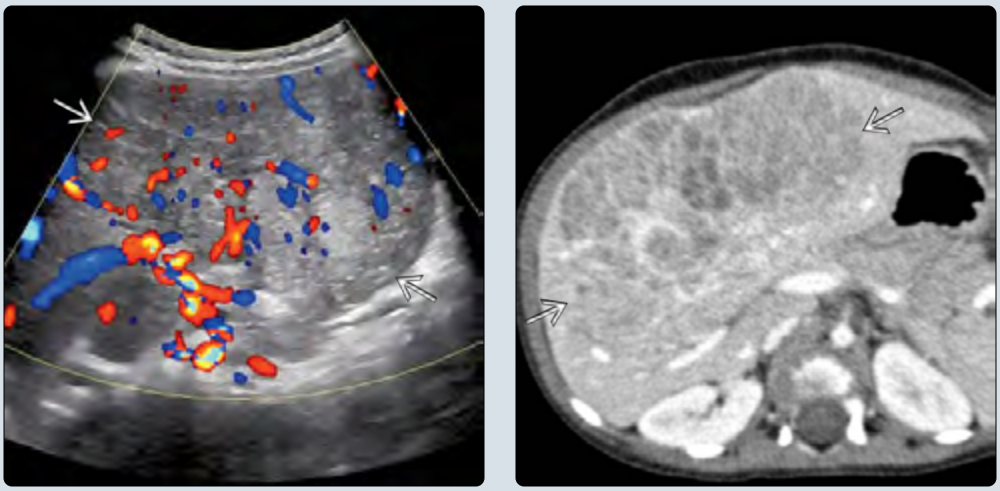
- Large, well-defined, solid but heterogeneous liver mass in child < 4 years of age with high α-fetoprotein (AFP) levels
  - Heterogeneity due to hemorrhage, necrosis, Ca^{2+}, or mixed histologies
- Single round, lobulated mass (80%), usually > 10 cm
  - May be multifocal (20%)
- Displacement/compression/effacement of adjacent vessels vs. true vascular invasion
- Usually enhances less than normal liver on all phases
- Chest CT to evaluate for pulmonary metastases
  - Metastases in 20% at presentation

PATHOLOGY

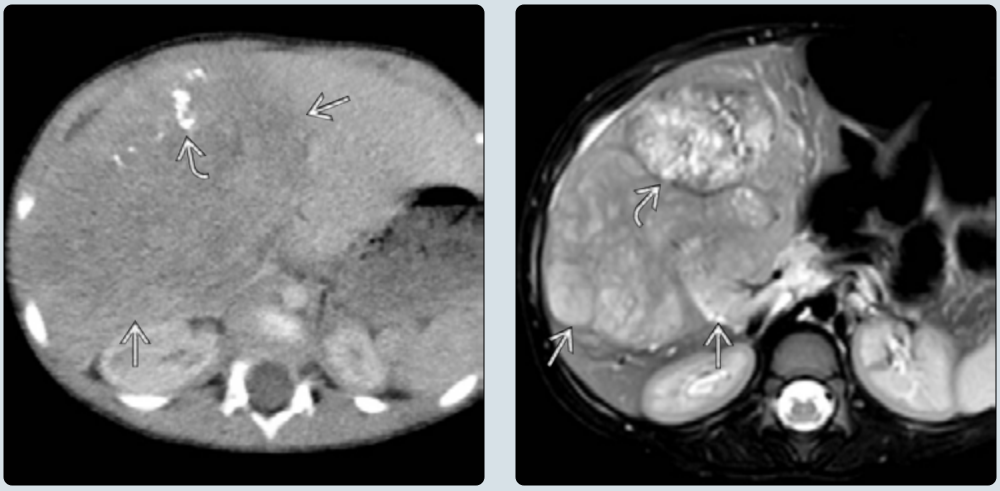
- Epithelial type more common than mixed epithelial & mesenchymal type
  - Fetal histology: Best prognosis
  - Small cell undifferentiated histology: Worst prognosis
- **Pret**reatment **ext**ent of disease (PRETEXT) image-based staging system evaluates number of contiguous sectors free of tumor

CLINICAL ISSUES

- Most common primary liver tumor of childhood
- 3rd most common pediatric abdominal malignancy
- Presents as painless abdominal mass or hepatomegaly
- Diagnosed in young children; median age: 19 months
  - Only 5-10% occur in children > 4 years of age
  - Only 4% congenital
- Surgical resection ± neoadjuvant chemotherapy
  - Resection alone can be curative
  - Primary transplant if unresectable after neoadjuvant therapy
- Posttherapy AFP levels serve as tumor marker for surveillance



(Left) *Transverse color Doppler US of the liver in a 1 year old with hepatoblastoma shows a heterogeneously hyperechoic, moderately vascular mass in the liver.* **(Right)** *Axial CECT in the same patient shows the mass occupying segments 4b & 5. Assuming that the tumor is confined to these 2 central sectors, this would represent a pretreatment extent of disease (PRETEXT) III tumor (as 3 sectors would need to be resected in order to completely excise the tumor).*



(Left) *Axial CECT in a 2 year old shows a hypodense mass in the liver. The mass has a small area of calcification. Calcification is present in up to 50% of hepatoblastomas.* **(Right)** *Axial T2 FS MR in the same patient shows a heterogeneous mass in the inferior liver. The mass is of intermediate signal intensity overall but is mildly hyperintense compared to the background liver. A hypointense septation & adjacent foci of increased T2 signal are noted anteriorly.*

TERMINOLOGY

Definitions

- Malignant embryonal hepatic tumor

IMAGING

General Features

- Best diagnostic clue
  - Large, well-defined, solid, heterogeneous liver mass in child < 4 years of age with high α-fetoprotein (AFP) levels
    - Heterogeneity due to hemorrhage, necrosis, or mixed histologies; Ca^{2+} in 50%
- Location
  - More common in right lobe of liver
- Size
  - Usually > 10 cm
- Morphology
  - Single round, lobulated mass (80%)
    - May be multifocal (20%)
  - Displaces, compresses, & effaces adjacent hepatic structures (vessels, falciform ligament)
  - May invade vessels (especially portal veins)
    - Tumor thrombus favored over bland thrombus if
      - Vessel dilated
      - Thrombus contiguous with mass
      - Thrombus shows enhancement with contrast or internal arterial flow on color Doppler ultrasound
  - Large, unusual veins may surround tumor

Radiographic Findings

- Homogeneous soft tissue mass in right upper quadrant
  - Displaces bowel inferiorly
  - May mimic generalized hepatomegaly
- Ca^{2+} uncommonly seen on radiographs

CT Findings

- NECT
  - Not recommended for liver mass evaluation
- CECT
  - Well-defined, heterogeneous mass
  - Most enhance < background liver
    - Multiphase CT not recommended due to radiation
    - ± arterial phase enhancement in rim & septa

MR Findings

- T1WI
  - Generally hypointense to normal liver
  - High signal intensity foci of intratumoral hemorrhage
- T2WI
  - Often of heterogeneous intermediate signal intensity but usually hyperintense to normal liver
    - ± hypointense fibrous bands, hemorrhage
    - ± internal hyperintense cystic/necrotic foci
- T1WI C+ FS
  - Heterogeneous enhancement, usually less than background liver on all phases
  - Hepatocyte phase (≥ 20-minute delay) using hepatocyte-specific contrast agents useful for
    - Characterizing tumor
    - Establishing relationship to biliary system & vessels
    - Detecting multifocal lesions

Ultrasonographic Findings

- Grayscale ultrasound
  - Solid, well-defined mass
  - Usually hyperechoic to liver, often heterogeneous
    - Acoustic shadowing due to echogenic Ca^{2+}
    - Hypoechoic/anechoic foci from necrosis, hemorrhage
  - May have spoke-wheel appearance
- Pulsed Doppler
  - Portal or hepatic venous tumor thrombus may show arterial waveforms in venous lumen
- Color Doppler
  - Variable amount of disorganized vascularity in tumor
    - Often hypovascular

Imaging Recommendations

- Best imaging tool
  - Ultrasound best initial imaging modality in child with palpable abdominal mass
    - Determines organ of origin, helping to protocol subsequent cross-sectional imaging study
    - ± visualization of vessel involvement
  - MR with hepatocyte specific contrast agent favored over CECT due to improved detail & lack of radiation
  - Chest CT to look for pulmonary metastases

DIFFERENTIAL DIAGNOSIS

Hepatocellular Carcinoma

- Most common malignant hepatic tumor in older children
  - Pediatric differs from adult hepatocellular carcinoma (HCC)
    - Underlying liver disease in 30-50%
    - Associated with glycogen storage diseases, tyrosinemia, hemochromatosis, α-1 antitrypsin deficiency, biliary atresia, hepatitis B
- AFP levels elevated < hepatoblastoma

Mesenchymal Hamartoma

- Benign hepatic tumor, patients usually < 2 years of age
- Large multicystic or mixed solid-cystic mass

Undifferentiated Embryonal Sarcoma

- Rare, usually 6-10 years of age, normal AFP
- May arise from mesenchymal hamartoma
- Large mass appears solid on US but cystic at CT/MR due to myxoid stroma

Hepatic Hemangioma

- Can be congenital or infantile type
- Congenital: Typically large & solitary, may cause heart failure with shunting
  - Detected in perinatal period
  - Heterogeneous mass with vigorous peripheral enhancement but central areas of persistent nonenhancement
  - Often regresses in 1st few months of life
- Infantile type: Multifocal or diffuse, small to moderate size
  - Develop in 1st weeks to months of life
  - Gradual complete enhancement (periphery → center)
  - Slowly regress after 6-24 months

Hepatic Metastases

- Most commonly caused by neuroblastoma or Wilms tumor
- Delayed hepatocyte phase MR may be most sensitive

PATHOLOGY

General Features

- Etiology
  - Unknown
  - Risk factors & predisposing conditions [with risk compared to population in surveillance, epidemiology & end result (SEER) database]
    - Very low birth weight (20x risk)
    - Beckwith-Wiedemann syndrome (2,280x risk)
    - Familial adenomatous polyposis (847x risk)

Staging, Grading, & Classification

- **Pret**reatment **ext**ent of disease (PRETEXT) staging system
  - Image-based system to guide surgical management
  - Evaluates number of contiguous sectors (combination of Couinaud segments) free of tumor
    - PRETEXT I: 3 contiguous sectors free of tumor
    - PRETEXT II: 2 contiguous sectors free of tumor
    - PRETEXT III: 1 sector free of tumor
    - PRETEXT IV: All sectors involved with tumor
  - PRETEXT number specifies how many sectors would need to be resected to entirely remove tumor
  - Modifiers for vascular invasion (portal veins & hepatic veins), multifocality, metastases, extrahepatic spread, & tumor rupture
- POST-TEXT (**postt**reatment **ext**ent) obtained after every 2 chemotherapy cycles to assess neoadjuvant response

Microscopic Features

- Multiple histologic subtypes
  - 2 main classes: Epithelial vs. mixed epithelial & mesenchymal
    - Epithelial type more common
      - Pure fetal histology has best prognosis; does not require chemotherapy
      - Small cell undifferentiated histology has worst prognosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless abdominal mass
  - Hepatomegaly
  - Markedly elevated AFP levels (90%)

Demographics

- Age
  - Diagnosed in young children
    - Median age at diagnosis: 19 months
    - 4% congenital
    - Only 5-10% occur beyond 4 years of age
- Gender
  - 60% male
- Epidemiology
  - Most common primary liver tumor of childhood
    - 91% of hepatic malignancies < 5 years of age
  - Incidence
    - 10.5 cases per million in children < 1 year of age
    - 5.2 cases per million in children 1-4 years of age
  - 3rd most common pediatric abdominal malignancy after Wilms tumor & neuroblastoma

Natural History & Prognosis

- Event-free survival/overall survival
  - Standard risk: Up to 83%/95%
    - Standard risk: Tumor confined to liver, ≤ 3 sectors
  - High risk: Up to 65%/69%
    - High risk: Tumor in 4 sectors, metastatic disease, low AFP levels
- Metastatic disease in 20% at diagnosis
  - Lung >> bone, brain
- Poor prognostic factors
  - Vascular involvement, multifocal tumor, tumor rupture, metastases, low AFP level (< 100 ng/ml), very high AFP (> 1 million ng/ml), ≥ 3 years of age, PRETEXT III or IV

Treatment

- Surgical resection can be curative
  - Goal to remove tumor as soon as feasible
  - Up to 60% unresectable initially
- Chemotherapy
  - Neoadjuvant therapy improves resectability
- Liver transplantation for unresectable tumor
  - 75-87% disease-free survival for primary transplant
- COG guidelines by PRETEXT/POST-TEXT staging
  - Upfront resection: PRETEXT I/II
  - Resection after neoadjuvant therapy: POST-TEXT II/III without venous invasion
  - Complex resection vs. transplant after neoadjuvant therapy: POST-TEXT IV or POST-TEXT III + venous invasion
- Neoadjuvant therapy given to all patients in some protocols
- Posttherapy AFP serves as surveillance tumor marker

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Large, well-defined, heterogeneous liver mass in infant
  - May have vascular invasion
    - Can be difficult initially to distinguish tumor invasion beyond sector vs. compression/displacement of adjacent structures
- CT of chest to evaluate for pulmonary metastases

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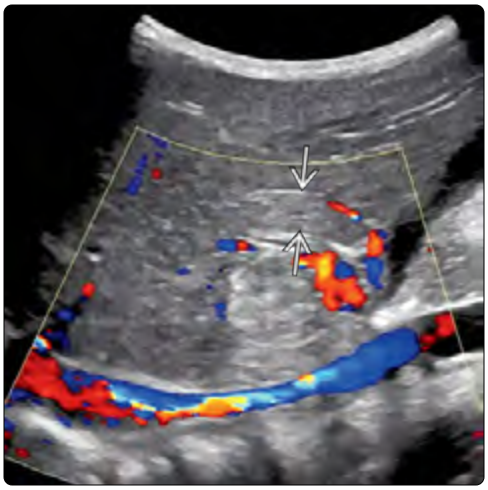
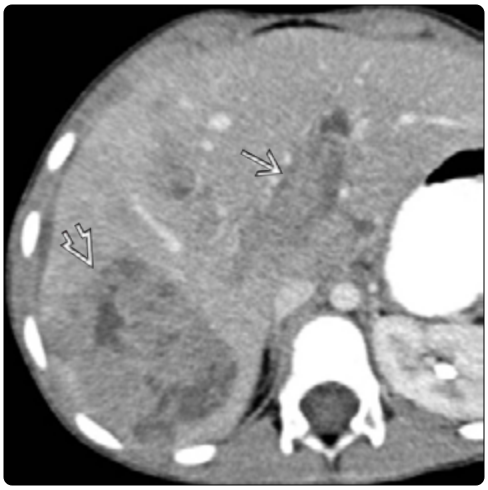
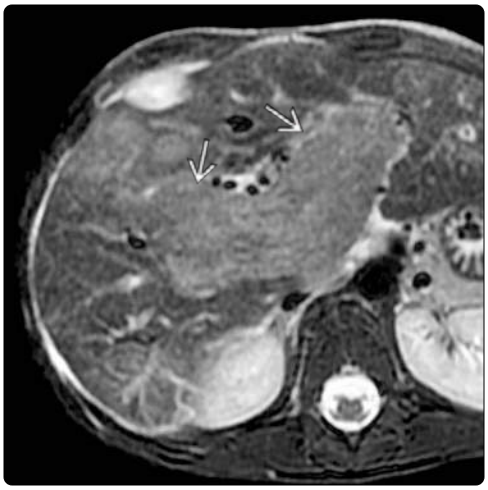
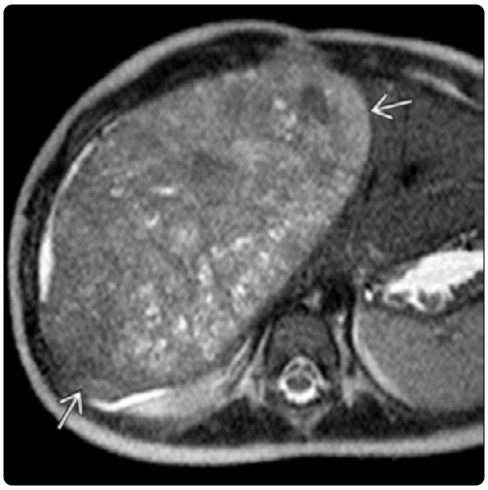


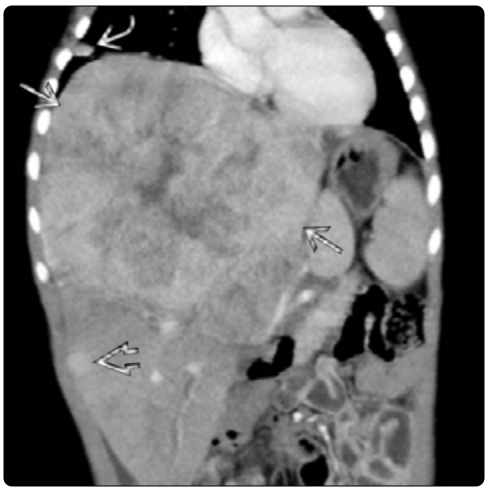


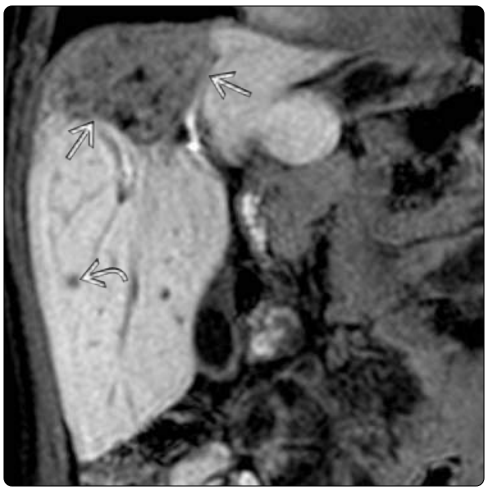
(Left) *Longitudinal color Doppler US in a 6 year old with hepatoblastoma shows tumor thrombus in the main portal vein. Tumor thrombus expands vessels, is echogenic, & can have internal Doppler flow (with arterial flow inside a venous thrombus being particularly suggestive).* **(Right)** *Axial CECT in the same patient shows tumor thrombus in the main portal vein & extending into the right & left portal veins. The primary tumor is partially imaged in the right lobe of the liver.*





(Left) *Axial T2 FS MR in the same patient shows the tumor thrombus extending into the right & left portal veins. Vascular involvement is a poor prognostic factor in children with hepatoblastoma.* **(Right)** *Axial T2 MR in a 2 year old with hepatoblastoma shows a heterogeneous mass in the right lobe of the liver. Assuming that this mass only involves the right lobe, this tumor would be considered a PRETEXT II tumor (as there are 2 contiguous uninvolved sectors of the left lobe).*





(Left) *Coronal CECT in a 4 year old with hepatoblastoma shows multifocal disease. The primary tumor is the large heterogeneous mass in the superior liver. A smaller lesion is visible inferiorly. Up to 20% of patients present with multifocal disease. A pulmonary metastasis is also visible in the right lung base.* **(Right)** *Coronal T1 C+ FS MR (in the delayed hepatocyte phase) of the same patient after 2 cycles of chemotherapy shows a marked interval decrease in the size of the primary tumor. A 2nd lesion remains visible.*

Hepatic Hemangiomas, Infantile and Congenital

KEY FACTS

TERMINOLOGY

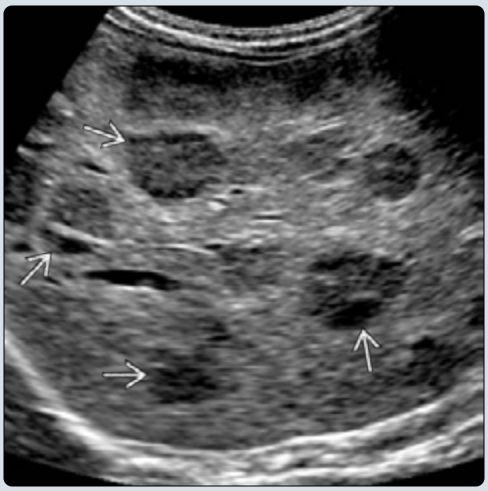
- Hemangioma: Benign endothelial neoplasm of neonates/infants in soft tissues or viscera (especially liver)
  - **Not** hemangioendothelioma (more aggressive tumor)
  - **Not** cavernous hemangioma (venous malformation)
- Congenital hemangioma (CH): Found in perinatal period, does not proliferate beyond birth; stains GLUT1 negative
  - Rapidly involuting subtype more common in liver than noninvoluting
- Infantile hemangioma (IH): Develops in 1st few weeks of life with characteristic proliferating & involuting phases; stains GLUT1 positive

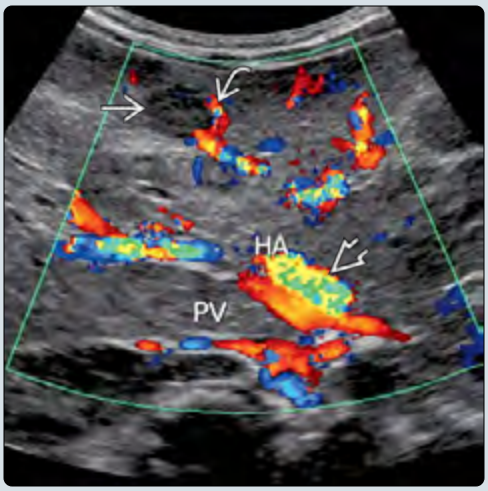
IMAGING

- Pediatric hepatic hemangiomas considered as 3 types
  - Focal (CH): Solitary large heterogeneous mass
  - Multifocal (IHs): Multiple small to moderate homogeneous masses
  - Diffuse (IHs): Liver enlarged & replaced by masses
- Focal hepatic CH: T2 heterogeneity; early peripheral enhancement + persistent foci of central nonenhancement
- Multifocal/diffuse IHs: T2 hyperintense lesions with gradual complete central filling after early peripheral enhancement
  - Often found in infants with ≥ 5 cutaneous IHs
- CH & IH often have enlarged feeding arteries/draining veins

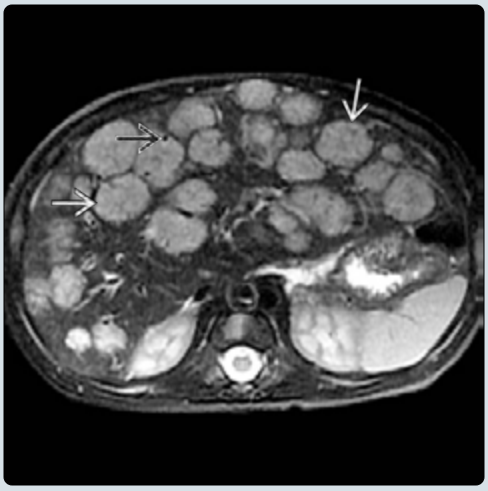
CLINICAL ISSUES

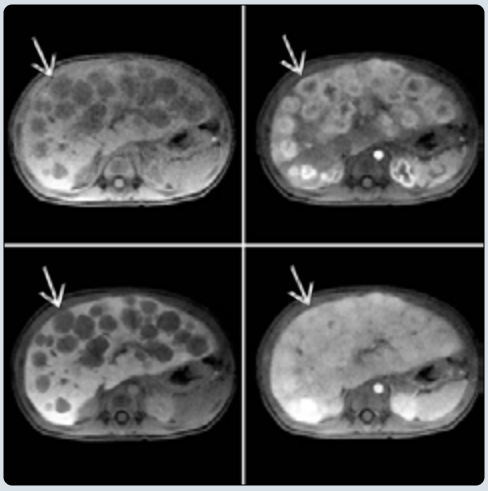
- Hepatic CH & IHs may be asymptomatic or can present with hepatomegaly, heart failure
- CH may have anemia & mild, transient consumptive coagulopathy (**not** Kasabach-Merritt phenomenon)
- Diffuse IHs may also present with hypothyroidism, liver failure, abdominal compartment syndrome
- Treatment required if complications develop (< 10%)
  - CH: Embolization, resection
  - IHs: Medical therapies initially (propranolol, steroids; rarely vincristine); embolization, liver transplant in extreme cases





(Left) *Longitudinal ultrasound in a 7 month old with a history of cutaneous infantile hemangiomas (IHs) shows multiple, round well-circumscribed, hypoechoic masses throughout the liver.* **(Right)** *Transverse color Doppler ultrasound in the same patient shows increased vascularity along the hypoechoic masses. Note the enlarged hepatic artery with color aliasing (as compared to the normal color Doppler appearance of the portal vein) due to undersampling of high-velocity flow supplying the lesions.*





(Left) *Axial T2 FS MR in this patient shows the typical appearance of IHs: Multiple, round, well-circumscribed small- to moderate-sized hyperintense (but not fluid-bright) lesions, some of which contain flow voids.* **(Right)** *Axial dynamic T1 C+ FS MR images with a hepatocyte-specific agent in (clockwise from top left) precontrast, arterial, portal venous, & delayed hepatocyte phases show a typical appearance of IH with early peripheral enhancement, complete venous filling, & washout.*

TERMINOLOGY

Synonyms

- **Not** hemangioendothelioma (more aggressive vascular tumor not typically found in infantile liver)
- **Not** cavernous hemangioma (venous malformation not typically found in infantile liver)

Definitions

- Hemangioma: Benign endothelial neoplasm occurring in soft tissues or viscera (especially liver: Most common benign hepatic tumor of infants)
  - 70% of 2009 publications used term hemangioma incorrectly
- Congenital hemangioma (CH): Found in perinatal period, does not proliferate beyond birth
  - Rapidly involuting (RICH) vs. noninvoluting (NICH) subtypes
- Infantile hemangioma (IH)
  - Not present at birth
  - Proliferating phase: Lesion(s) develop during 1st few weeks of life with rapid growth over next few months
  - Involuting phase: Slow regression of lesion(s) over years, usually beginning by 12 months of age

IMAGING

General Features

- Best diagnostic clue
  - CH: Large solitary heterogeneous hypervascular hepatic mass in fetus/newborn
  - IHs: Multiple small to moderate T2 hyperintense hepatic lesions with progressive peripheral to central enhancement in infant with ≥ 5 cutaneous IHs
- Morphology
  - Hepatic hemangiomas of young children characterized as focal, multifocal, or diffuse
    - Focal hepatic hemangioma = true CH (usually RICH)
      - Large, solitary, well-defined mass
      - Multifocal CHs very rare
    - Multifocal/diffuse hepatic hemangiomas = IHs
      - Multifocal IHs (most common): Multiple small to moderate-sized round lesions
      - Diffuse IHs: Liver enlarged & nearly completely replaced by lesions
  - Enlarged feeding arteries/draining veins may be seen with either CH or IHs

Radiographic Findings

- Radiography
  - Hepatomegaly or right upper quadrant mass
    - Depends on size/number of lesions
  - Ca^{2+} can be seen in ~ 15% of CH cases
  - ± heart failure with cardiomegaly, pulmonary edema

CT Findings

- NECT
  - Well-defined hypodense mass/masses
  - CH: ± hyperdense foci of hemorrhage, Ca^{2+}
- CECT
  - Typical enhancement patterns
    - CH: Increasingly confluent early nodular peripheral enhancement on dynamic imaging; large central areas do not fill in/enhance over time
    - IHs: Early peripheral enhancement with gradual complete central fill in

MR Findings

- T1WI
  - Hypointense mass/masses compared to unaffected liver
  - CH: Hyperintense foci of hemorrhage
- T2WI
  - CH: Very heterogeneous internally with mixed hyperintense/hypointense foci
  - IHs: Relatively homogeneous, hyperintense lesions compared to unaffected liver (but not as bright as fluid); small septa; scattered prominent flow voids
- T1WI C+
  - Enhancement patterns similar to CT
  - Washout expected with hepatocyte-specific agents
- MRA/MRV
  - ± enlargement of
    - Upper abdominal aorta, celiac axis, hepatic arteries
    - Hepatic veins, inferior vena cava
    - Arterial & venous branches along periphery of lesions (especially CH)

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal ultrasound
    - CH: Heterogeneous mass ± ↑ vascularity, hydrops
    - IHs: Not present
  - Postnatal ultrasound
    - CH: Solitary heterogeneous mass
    - IHs: Multiple round well-defined lesions of variable echogenicity, most frequently hypoechoic & homogeneous
- Color Doppler
  - CH: ↑ peripheral vascularity ± arteriovenous shunts; little to no central vascularity
  - IHs: Variable ↑ vascularity within/along lesions; low resistance arterial waveforms during proliferative phase
  - Hepatic arteries & veins frequently enlarged

Imaging Recommendations

- Best imaging tool
  - US best screening tool for palpable abdominal mass, hepatomegaly, or infant with multiple cutaneous IHs
  - MR for further characterization, extent determination

DIFFERENTIAL DIAGNOSIS

Hepatoblastoma

- Usually large well-defined, solitary mass
- Uncommon in newborns
- Rarely hypervascular; usually hypoenhancing
  - Peripheral enhancement not typical
- Elevated α-fetoprotein (AFP)

Neuroblastoma Metastasis

- Multiple masses vs. diffuse liver heterogeneity & enlargement
- Adrenal or paraspinal mass typically present

Mesenchymal Hamartoma

- Solitary liver mass of young children
- May be multicystic or solid & cystic
- Not hypervascular

Umbilical Catheter Complication

- Vascular/hepatic perforation by umbilical venous catheter may lead to hemorrhage &/or parenchymal infusion of fluid

Arteriovenous Malformation

- Very uncommon in pediatric liver
- Tangles of large flow voids without solid enhancing tissue

Venous Malformation

- Uncommon in pediatric liver outside of syndromes
- Lobulated fluid-signal intensity lesions ± layering fluid-fluid levels, phleboliths
- Gradual puddling of contrast into lesion over time

PATHOLOGY

Microscopic Features

- Clusters of thin-walled capillaries lined by plump endothelium without cellular atypia
- IH
  - Densely packed capillaries with little intervening stroma
  - Cellular proliferation present during early stages
  - GLUT1-positive endothelium
  - Fatty infiltration not usually seen with involution in hepatic IHs (unlike soft tissue IHs)
- CH
  - Varying sized lobules of capillaries surrounded by dense myxoid stroma & large malformed vessels
  - Foci of hemorrhage, necrosis, fibrosis, extramedullary hematopoiesis, Ca^{2+}
  - GLUT1-negative endothelium

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Hepatic CH, IHs may be asymptomatic
  - CH, IHs can present with hepatomegaly, heart failure
- Other signs/symptoms
  - CH may have anemia & mild, transient consumptive coagulopathy
    - **Not** Kasabach-Merritt phenomenon (occurs in Kaposiform hemangioendothelioma or tufted angioma)
  - Diffuse IHs may also present with
    - Hypothyroidism (due to production of enzyme type 3 iodothyronine deiodinase)
      - 100% of diffuse IHs vs. 21% of multifocal IHs
    - Liver failure
    - Abdominal compartment syndrome
  - Cutaneous IH found in
    - 50-75% of multifocal/diffuse hepatic IHs
    - 15% of focal hepatic CH
  - AFP not typically elevated by CH or IHs
    - AFP normally high in newborns but ↓ over 1st months of life

Demographics

- Age
  - CH usually detected in perinatal period
  - IHs develop in 1st few weeks of life
    - More common in premature infants
- Gender
  - CH: M:F = 1:1; IH: M:F = 1:3
- Ethnicity
  - IH much more common in Caucasians

Natural History & Prognosis

- Focal hepatic CH
  - Can enlarge from hemorrhage
  - RICH will mostly involute by 14 months of age
  - Mortality widely variable in literature
- Multifocal/diffuse IHs
  - May grow after detection, depending on age
  - After 1st year of life, gradually involute over years
  - Mortality: 38% for diffuse IHs, 9% for multifocal IHs; median age of death = 115 days

Treatment

- Treatment required if complications develop
  - < 10% of pediatric hepatic hemangiomas require therapy
- CH: Embolization, resection; medical therapies not proven effective
- IHs: Medical therapies initially (propranolol, steroids; rarely vincristine); embolization, liver transplant in extreme cases

DIAGNOSTIC CHECKLIST

Consider

- Liver US screening in patients with ≥ 5 cutaneous IHs
- Biopsy of hepatic lesions should be performed if diagnosis unclear by combination of clinical history/exam, initial imaging, & lesion behavior over appropriate timeframe
  - Safely performed percutaneously in experienced hands

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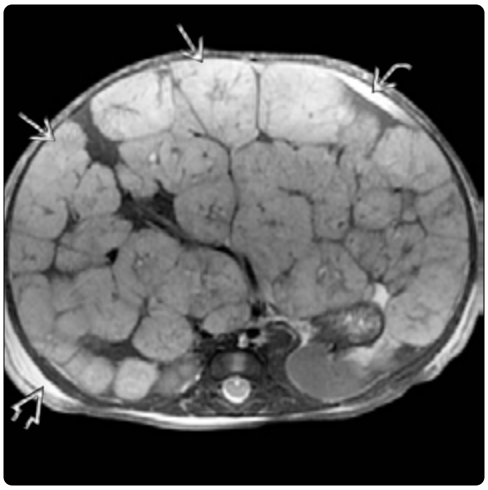
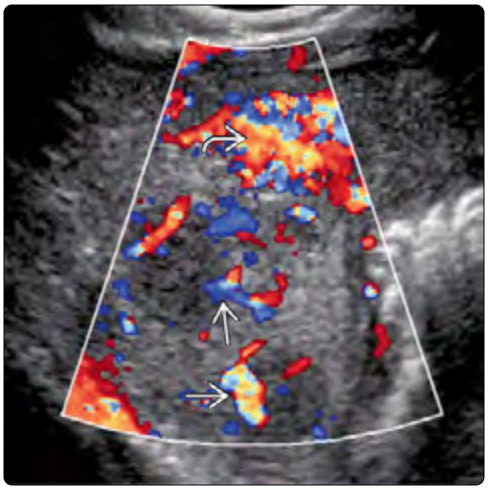
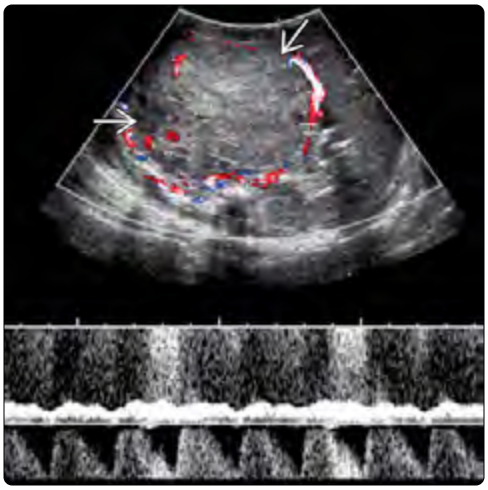
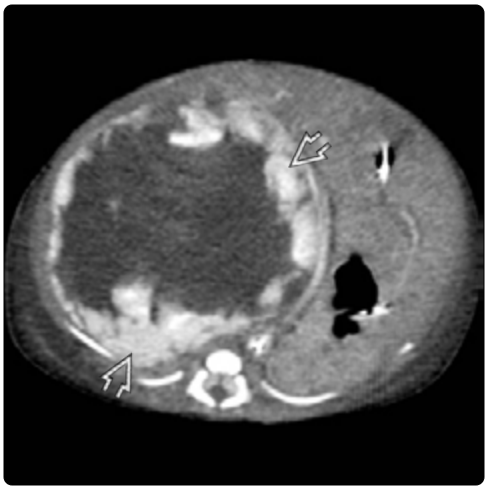


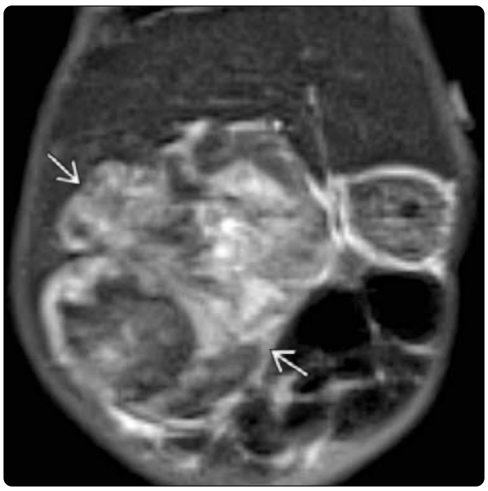


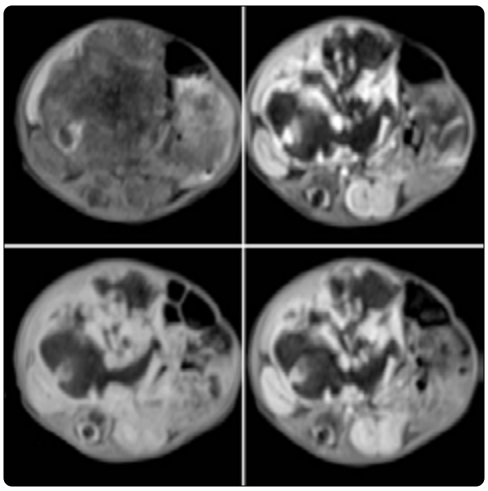
(Left) *Axial T2 FS MR in a patient with cutaneous IHs & abdominal distention shows near total replacement of the hepatic parenchyma by innumerable hyperintense lesions. Note the ascites & body wall edema secondary to liver failure.* **(Right)** *Transverse color Doppler ultrasound in a 2 month old with cutaneous IHs shows variable vascularity within the hypoechoic hepatic lesions, ranging from moderate peripheral vascularity to diffusely increased internal flow.*





(Left) *Pulsed Doppler ultrasound in a newborn with hydrops shows a large, solid, mildly heterogeneous mass in the right hepatic lobe with increased peripheral vascularity. Interrogation of an enlarged artery shows elevated velocities wrapping around the scale.* **(Right)** *Axial CECT shows irregular heterogeneous enhancement of the lesion periphery on arterial phase. Central portions of the mass did not enhance on delayed images (not shown), typical of a focal hepatic congenital hemangioma (CH).*





(Left) *Coronal T2 FS MR in a 4 day old with a palpable abnormality shows a large exophytic mass arising from the inferior right hepatic lobe. The lesion shows marked internal heterogeneity.* **(Right)** *Axial T1 C+ FS MR images of the same patient at (clockwise from top left) precontrast, 90-second, 180-second, & 5-minute times show irregular peripheral enhancement with large areas of persistent central nonenhancement, typical of a focal CH of the liver.*

Hepatic Mesenchymal Hamartoma

KEY FACTS

TERMINOLOGY

- Mesenchymal hamartoma (MH): Benign liver tumor of infants due to primitive mesenchymal proliferation

IMAGING

- Spectrum from mostly cystic to mostly solid masses
  - Amount of solid stromal tissue determines appearance
  - Cysts found (to some degree) in up to 85%
    - Septations may be thin or thick
- Typically unifocal; rarely multifocal or diffuse
  - Right hepatic lobe: 75%
- Up to 30 cm in size; mean: 16 cm
- Ca^{2+}, hemorrhage uncommon

TOP DIFFERENTIAL DIAGNOSES

- Hepatoblastoma
- Congenital hemangioma (CH)
- Umbilical venous catheter extravasation
- Undifferentiated embryonal sarcoma (UES)
- Mesenteric lymphatic malformation

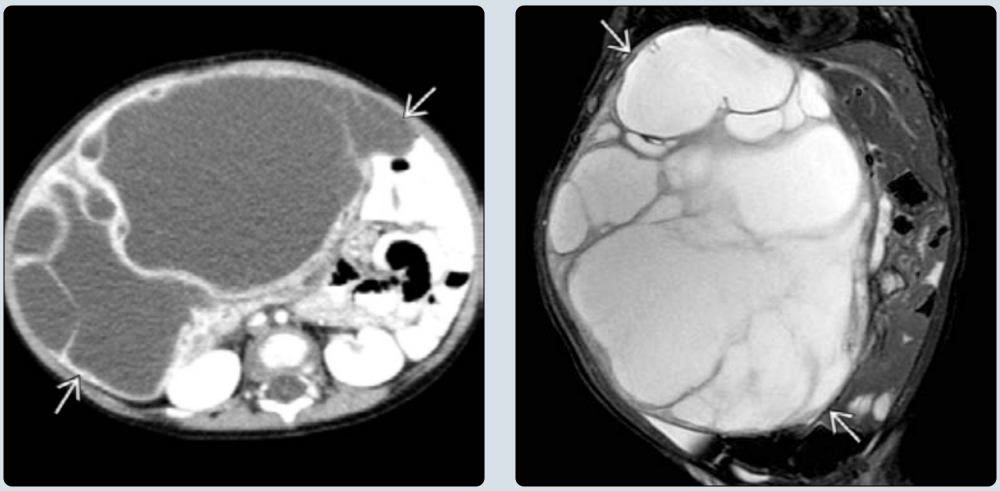
CLINICAL ISSUES

- Typically < 2 years old; 5% > 5 years old
- Grow slowly or rapidly in perinatal period
  - Prenatal: May cause polyhydramnios, hydrops
  - Postnatal: May cause respiratory distress
- Subsequent stabilization vs. partial regression
- > 90% long-term survival
- Complete excision due to rare malignant potential
  - Reports of UES arising in background MH
- Cyst aspiration may be required emergently (pre- or postnatally) to alleviate compression

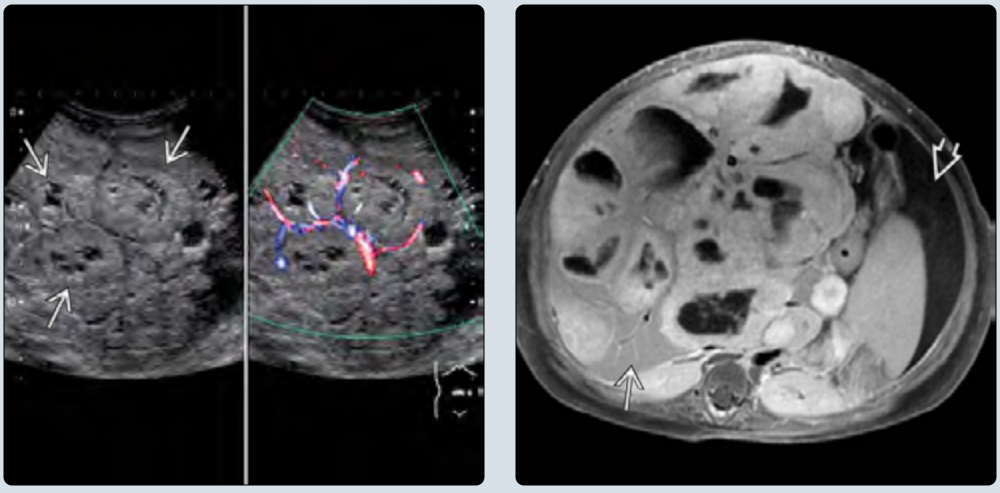
DIAGNOSTIC CHECKLIST

- Most likely hepatic neoplasms in young children
  - Largely cystic in neonate/infant → MH
  - Solid, hypovascular in infant/toddler → hepatoblastoma
  - Solid, heterogeneous, hypervascular in newborn → CH

(Left) *Axial CECT of the abdomen in a 9-month-old patient shows a large multicystic mass ➡ extending inferiorly from the right hepatic lobe. Resection confirmed a mesenchymal hamartoma.* **(Right)** *Coronal T2 FS MR in a 6-month-old patient shows a large multicystic mass ➡ replacing the right lobe of the liver & filling much of the abdomen, displacing the bowel to the left. Mesenchymal hamartoma was confirmed upon surgical resection.*



(Left) *Transverse grayscale & color Doppler ultrasounds in a 6 month old with new onset liver failure show numerous mixed solid & cystic masses ➡ filling the visible liver & splaying the hepatic vessels. Scant internal vascularity is seen in the lesions.* **(Right)** *Axial T1 C+ FS MR in the same patient shows heterogeneously enhancing masses replacing all but a small volume of normal liver parenchyma ➡. Note the ascites ➡ secondary to liver failure. A liver transplant was ultimately required for diffuse mesenchymal hamartomas.*



TERMINOLOGY

Abbreviations

- Mesenchymal hamartoma (MH)

Definitions

- Benign liver tumor of infants due to primitive mesenchymal proliferation

IMAGING

General Features

- Best diagnostic clue
  - Large multicystic liver mass in child < 2 years of age
- Location
  - Right hepatic lobe: 75%; pedunculated: 20%
  - Typically unifocal; rarely multifocal or diffuse
- Size
  - Up to 30 cm; mean: 16 cm
- Morphology
  - Spectrum from mostly cystic to mostly solid
    - Cysts found (to some degree) in up to 85%
      - Variable cyst sizes: < 1 cm to > 15 cm
    - Septations may be thin or thick
  - Amount of solid stromal tissue determines appearance
  - Ca^{2+}, hemorrhage uncommon

Radiographic Findings

- Hepatomegaly or noncalcified RUQ mass displacing bowel

CT Findings

- Fluid-attenuation cystic components
- Enhancing septations, solid components

MR Findings

- Cysts: T1 signal variable; high T2 signal intensity (fluid); no central enhancement after contrast
- Septations/stromal components: T1 & T2 intermediate to low signal intensity; variable enhancement after contrast

Ultrasonographic Findings

- Anechoic cysts, echogenic septations
  - Mobile septations & hyperechoic nodules suggestive
- Swiss cheese or sieve appearance: Multiple cysts scattered throughout solid tissue
- Typically show little blood flow by Doppler exam

DIFFERENTIAL DIAGNOSIS

Hepatoblastoma

- Usually solid, heterogeneous; Ca^{2+} in 50%
- Markedly elevated AFP: 90%
- < 5 years of age but congenital lesions uncommon

Congenital Hemangioma

- Large heterogeneous hypervascular mass in newborn liver
- Early progressive peripheral enhancement; patchy, often large, central foci of nonenhancement on delayed images
- ± high-output heart failure

Umbilical Venous Catheter Extravasation

- Irregular fluid collection in neonatal hepatic parenchyma adjacent to malpositioned umbilical venous catheter

Undifferentiated Embryonal Sarcoma

- Overlap of imaging & histology with MH
- Older age group (6-10 years)

Mesenteric Lymphatic Malformation

- Infiltrating vs. well-defined, low-flow vascular anomaly
- Numerous cysts & septations of varying size
- Fluid-fluid levels frequent due to internal hemorrhage

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal distension, gradual vs. rapid
- Other signs/symptoms
  - Palpable RUQ mass, vomiting, anorexia
  - Respiratory distress (when compressing diaphragm)
  - AFP may be mildly elevated; usually normal
  - Antenatal: Polyhydramnios, hydrops; ↑ maternal AFP, β-HCG

Natural History & Prognosis

- Grows slowly or rapidly in perinatal period
- Subsequent stabilization vs. partial regression
- Rare malignant transformation to undifferentiated embryonal sarcoma (UES)
- > 90% long-term survival

Treatment

- Complete excision due to rare malignant potential
- Incomplete resection with cyst marsupialization (suboptimal) vs. liver transplant for unresectable lesions
- Cyst aspiration may be required emergently (pre- or postnatally) to alleviate compression

DIAGNOSTIC CHECKLIST

Consider

- Most likely hepatic neoplasms in young children
  - Largely cystic in neonate/infant → MH
  - Solid, hypovascular in infant/toddler→ hepatoblastoma
  - Solid, heterogeneous, hypervascular in newborn → CH
- In neonate with recent UVC infusion & newly discovered "cystic hepatic mass," consider extravasation

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Focal Nodular Hyperplasia

KEY FACTS

TERMINOLOGY

- Benign epithelial tumor composed of hepatocytes, Kupffer cells, abnormal bile ducts, & vessels
  - Central scar present in 50-70%

IMAGING

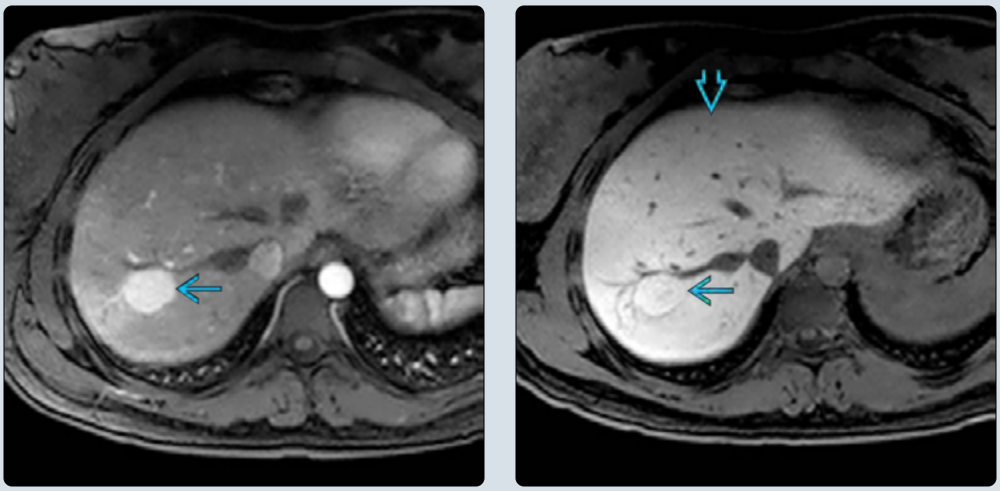
- Well-defined round or ovoid mass with smooth or mildly lobulated margins
- CECT: Homogeneous arterial enhancement ± hypoenhancing central scar
- MR with hepatobiliary phase contrast agent (best tool)
  - T1WI: Iso- to slightly hypointense; ± hypointense scar
  - T2WI: Slightly hyperintense; ± hyperintense scar
  - Arterial phase: Hyperenhancing mass ± hypoenhancing central scar
  - Venous phase: Iso- to slight hyperenhancement of mass ± hyperenhancing central scar
  - Delayed hepatobiliary phase: Mild hyperenhancement relative to normally enhancing background liver
    - Usually homogeneous (can be heterogeneous)
- US: Can be ↑, ↓, or ↔ echogenicity relative to liver
  - Doppler shows spoke-wheel pattern" with hypervascular central scar & radiating vessels extending to periphery of mass

PATHOLOGY

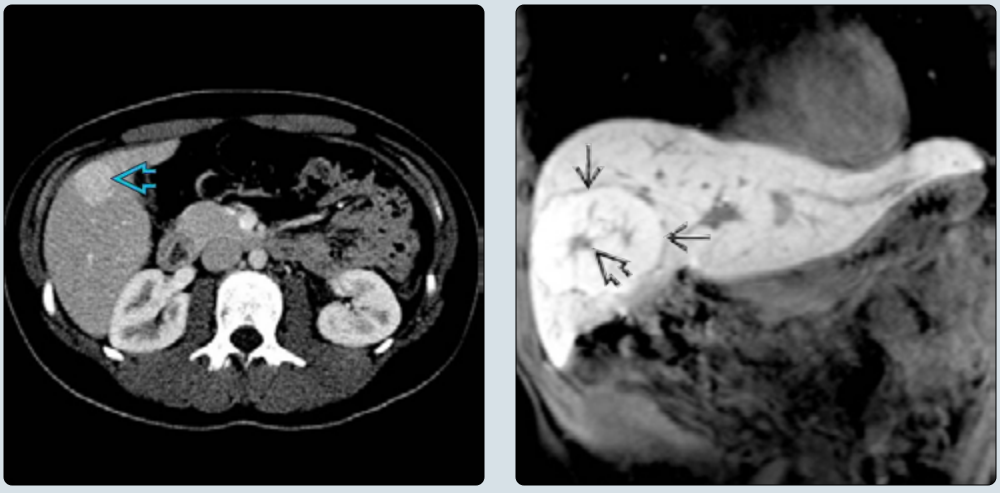
- Hyperplastic response to preexisting vascular malformation within central scar
- ↑ prevalence after chemotherapy &/or stem cell transplant
  - Often multiple lesions, smaller with no central scar

CLINICAL ISSUES

- 2% of primary hepatic tumors in children
- Often asymptomatic; occasionally causes palpable mass &/or vague abdominal pain
- No known malignant potential
- Managed conservatively in asymptomatic patients & surgically in symptomatic patients



(Left) *Arterial phase T1 C+ FS MR in a 16-year-old girl with an incidentally discovered liver mass demonstrates a homogeneously hyperenhancing lesion typical of focal nodular hyperplasia (FNH).* **(Right)** *Twenty-minute delayed MR after the injection of a hepatocyte specific contrast agent in the same patient shows normal homogeneous enhancement of the background liver parenchyma. The mass demonstrates mild homogeneous hyperenhancement relative to the liver, consistent with FNH.*



(Left) *Axial CECT in a 14-year-old girl shows a homogeneous liver mass with mild hyperenhancement. On the subsequent multiphase MR with a hepatocyte-specific contrast agent, the lesion was consistent with FNH.* **(Right)** *Twenty-minute delayed coronal T1 C+ FS MR using a hepatocyte specific contrast agent in a patient with a hepatic lesion demonstrates hyperenhancement of the mass relative to the surrounding normally enhanced liver parenchyma. There is a nonenhancing central scar typical of FNH.*

TERMINOLOGY

Abbreviations

- Focal nodular hyperplasia (FNH)

Definitions

- Benign epithelial tumor composed of hepatocytes, abnormal bile ducts, & vascular structures
- 2% of primary hepatic tumors in children

IMAGING

General Features

- Best diagnostic clue
  - Homogeneous, arterially enhancing mass on CT or MR with delayed enhancement of central scar
  - Central scar present in 50-70%
    - Scar more likely in larger lesions
- Morphology
  - Well-defined round or ovoid mass
  - Smooth vs. mildly lobulated margins

CT Findings

- CECT
  - Arterial phase
    - Hyperenhancing hepatic lesion ± ↓ attenuation central scar
  - Portal venous & delayed phases
    - Becomes isoattenuating relative to normal liver
    - Enhancing central scar (if present)
  - May be impossible to distinguish from normal liver on NECT or postcontrast portal venous & delayed phases

MR Findings

- T1WI
  - Iso- to hypointense lesion; ± hypointense central scar
- T2WI
  - Iso- to slightly hyperintense; ± hyperintense central scar
- T1WI C+
  - Arterial phase
    - ↑ enhancement relative to normal liver; ± ↓ enhancing scar
  - Portal venous phase
    - Becomes isointense to liver; ± ↑ enhancing scar
  - Hepatobiliary phase (e.g., gadoxetate disodium)
    - Most common: Mildly ↑ enhancement of mass relative to normal liver; ± ↓ enhancing scar
    - Different patterns of enhancement reported: Homogeneous, heterogeneous, peripheral, isointense to liver
      - Rare ↓ enhancement has been described

Ultrasonographic Findings

- Grayscale ultrasound
  - Iso-, hypo-, or hyperechoic mass ± central scar
- Color Doppler
  - Spoke-wheel pattern with hypervascular central scar & radiating vessels extending to periphery

Imaging Recommendations

- Best imaging tool
  - Dynamic MR with hepatobiliary contrast agent

DIFFERENTIAL DIAGNOSIS

Fibrolamellar Hepatocellular Carcinoma

- Central scar hypointense on both T1WI & T2WI MR

Hepatocellular Carcinoma

- ± underlying liver disease in children
- Typically ↓ enhancement on hepatobiliary phase MR

Hepatic Adenoma

- ± intracellular lipid with ↓ signal on opposed-phase MR
- Rare hepatobiliary phase MR enhancement
  - Can be indistinguishable from FNH

PATHOLOGY

General Features

- Hyperplastic response to preexisting vascular malformation within central scar
  - Leads to proliferation of functional hepatocytes, vascular structures, & abnormal bile ducts
    - Abnormal bile ducts → hepatobiliary phase hyperenhancement (↓ clearance of contrast agent)
- ↑ prevalence after chemotherapy &/or stem cell transplant
  - Lesions more likely multiple, smaller, no central scar

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often asymptomatic; occasionally causes abdominal pain due to mass effect
- Other signs/symptoms
  - Normal lab values, including α-fetoprotein

Natural History & Prognosis

- No known malignant potential

Treatment

- Managed conservatively in asymptomatic patients & surgically in symptomatic patients

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Homogeneous arterially-enhancing mass on CT or MR, ± central scar
- ↑ enhancement relative to normal liver with delayed hepatobiliary phase contrast MR

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Hepatic Adenoma

KEY FACTS

TERMINOLOGY

- Benign neoplasm arising from hepatocytes
- May hemorrhage/rupture; rare malignant potential (~ 5%)
- Pathologic subtypes
  - Inflammatory ~ 35-50%
  - Hepatocyte nuclear factor 1α (*HNF1α*) mutated ~ 35-40%
    - "L-FABP negative"
  - β-catenin activated ~ 10-19%
    - ↑ malignant potential
  - Undifferentiated ~ 5-10%

IMAGING

- Heterogeneously enhancing on arterial phase
  - May contain fat, hemorrhage, necrosis, Ca^{2+}
- Subtypes may have different MR features
  - Inflammatory: Peripheral ↑ on T2WI, ↑ arterial enhancement
  - *HNF1α:* ↓ on T1 opposed-phase (due to ↑ lipid), ↑ to ↔ arterial enhancement
  - β-catenin & undifferentiated subtypes: Variable features
- Hepatobiliary phase MR contrast agents may help differentiate from FNH
  - Adenomas rarely retain contrast on hepatobiliary phase
- US: Heterogeneous, ± ↑ echogenicity due to lipid or hemorrhage
- CT: Arterial phase → heterogeneous hyperenhancement
  - ↑ attenuation subcapsular or peritoneal fluid if tumor rupture/hemorrhage occurs

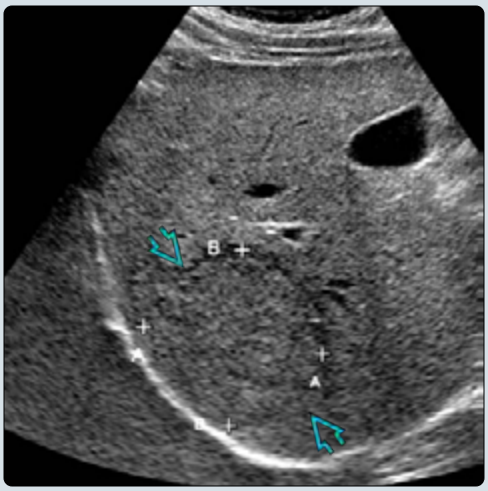
TOP DIFFERENTIAL DIAGNOSES

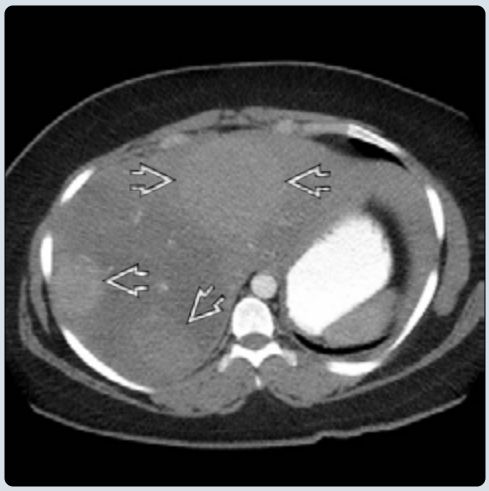
- Focal nodular hyperplasia
- Hepatocellular carcinoma

CLINICAL ISSUES

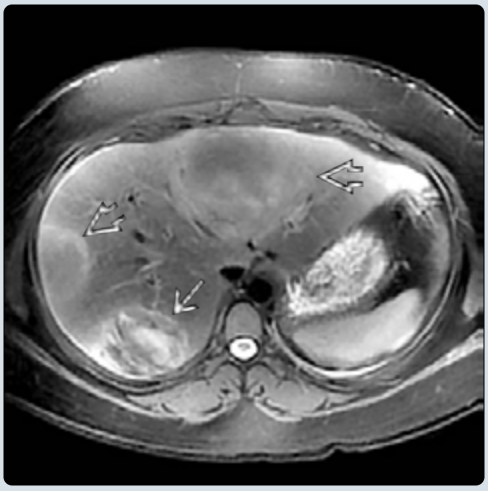
- Adenomas more likely with oral contraceptive use, steroid use, glycogen storage disease
- May regress if oral contraceptives discontinued
- Surgical excision if large or symptomatic

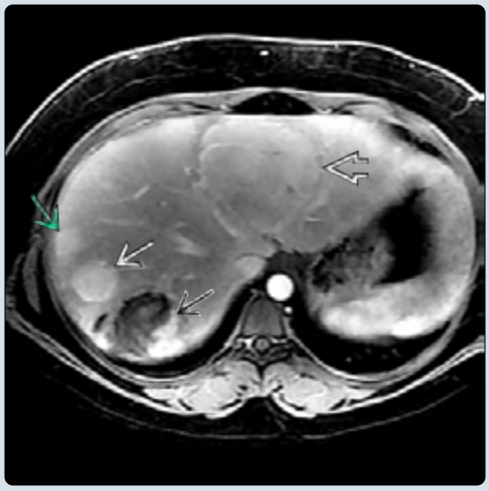
(Left) *Longitudinal ultrasound in a teenager with Fanconi anemia on androgen therapy shows an isoechoic but relatively well-defined round mass in the right lobe of the liver. Biopsy confirmed a hepatic adenoma.* **(Right)** *Axial arterial phase CECT in a 16-year-old girl with vague abdominal pain shows 3 liver masses with mild early enhancement, later found to be hepatic adenomas at biopsy.*





(Left) *Axial T2 FS MR in the same 16-year-old girl shows 2 of the adenomas to be mildly hyperintense. The 3rd mass is heterogeneous with hypointense foci due to prior hemorrhage.* **(Right)** *Arterial phase T1 C+ FS MR in the same 16-year-old girl shows mild enhancement in the largest adenoma, homogeneous enhancement in a smaller adenoma, & heterogeneous enhancement with low signal in a 3rd adenoma. Additional smaller enhancing adenomas are also present.*





TERMINOLOGY

Definitions

- Benign neoplasm arising from hepatocytes
- Risk of rupture/hemorrhage; rare malignant potential
- Different subtypes with different imaging features & clinical significance
  - Inflammatory ~ 30-50%
  - Hepatocyte nuclear factor 1α (*HNF1α*) mutated ~ 35-40%
    - Also known as "L-FABP negative"
    - More frequent fat content
  - β-catenin activated ~ 10-19%
    - ↑ malignant potential
  - Undifferentiated ~ 5-10%

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous, arterial-phase enhancing liver mass in young female on oral contraceptives

CT Findings

- NECT
  - Typically ↓ attenuation compared to liver
  - ± ↑ attenuation due to Ca^{2+} or hemorrhage
  - ↑ attenuation subcapsular or peritoneal fluid if tumor rupture/hemorrhage occurs
- CECT
  - Arterial phase → heterogeneous hyperenhancement
  - Venous phase → ± heterogeneous enhancement

MR Findings

- Most common MR appearance
  - ↓ to ↔ T1WI, mildly ↑ T2WI
    - ± ↑ T1WI with hemorrhage , ± ↓ T2WI due to hemosiderin
  - ± signal loss on T1 opposed-phase images due to lipid
  - ↑ arterial enhancement, ± heterogeneity
- Different subtypes may have some differentiating features; however, there is overlap
  - *HNF1α* subtype
    - ↓ signal on T1 opposed-phase due to intracellular lipid
    - Moderate arterial enhancement
  - Inflammatory subtype
    - Intracellular lipid less common than *HNF1α*
    - Heterogeneously hyperintense on T2WI
      - ± peripheral rim of ↑ T2WI signal
    - ↑ arterial enhancement, ↔ venous phase enhancement
  - β-catenin activated & undifferentiated subtypes
    - Variable, often similar to inflammatory
      - ± ↑ heterogeneity
- Hepatobiliary phase MR contrast agent
  - Typically hypoenhancing on hepatobiliary phase
  - Rarely can be iso- to hyperenhancing
    - Most common with β-catenin subtype

Ultrasonographic Findings

- Heterogeneous well-defined mass, may be hyperechoic due to lipid content or hemorrhage
- ± complex ascites if rupture/hemorrhage occurs

Imaging Recommendations

- Best imaging tool
  - Multiphasic MR including opposed-phase T1WI
  - Hepatobiliary MR contrast agent can help distinguish from focal nodular hyperplasia (FNH) in most cases

DIFFERENTIAL DIAGNOSIS

Focal Nodular Hyperplasia

- Homogeneous arterial enhancement ± central scar
- No intracellular lipid
- Iso- or mildly hyperintense to liver on hepatocyte phase with hepatobiliary contrast agent

Hepatocellular Carcinoma (HCC)

- Can be indistinguishable from hepatic adenoma on imaging

Hypervascular Metastasis

- Uncommon in children
- No intracellular lipid

PATHOLOGY

General Features

- Associated with oral contraceptives, exogenous steroids, glycogen storage diseases

Gross Pathologic & Surgical Features

- ± fat, necrosis, & hemorrhage
- 70-80% solitary

Microscopic Features

- Composed of hepatocytes arranged in sheets & cords with rare Kupffer cells

CLINICAL ISSUES

Natural History & Prognosis

- ↑ risk of hemorrhage with ↑ size of mass
- Risk of malignant transformation to HCC ~5%
  - ↑ risk with β-catenin subtype

Treatment

- Discontinuation of oral contraceptives can lead to ↓ in size or even complete regression
- Surgical excision if large or symptomatic

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KEY FACTS

TERMINOLOGY

- Hepatocellular carcinoma (HCC): Malignant tumor of hepatocytes
- Hepatocyte-specific contrast agent (HSCA): MR contrast agent with substantial hepatic uptake & biliary excretion

IMAGING

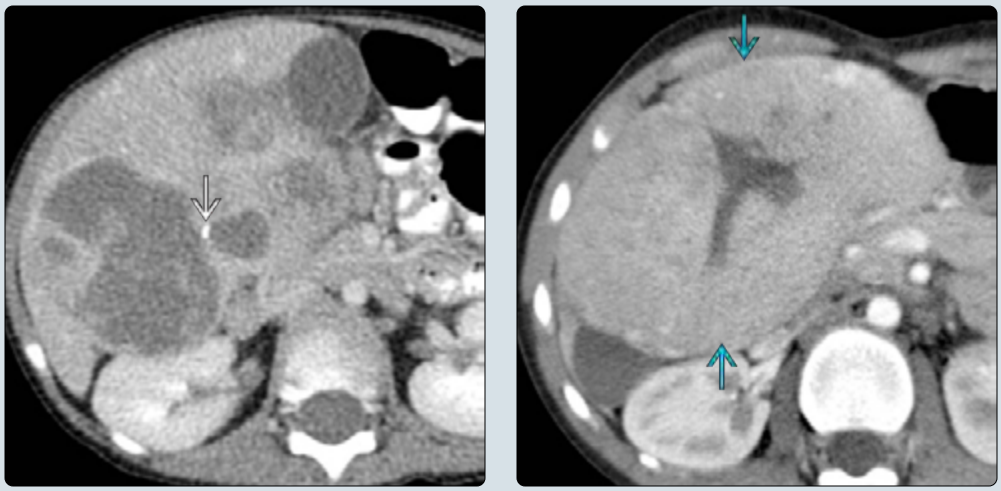
- US: Smaller HCCs often hypoechoic; larger HCCs often heterogeneous in echogenicity
- CT & MR
  - Variable precontrast CT attenuation/MR signal intensity
    - ± fat, fibrosis, Ca^{2+}, necrosis, & hemorrhage
  - Typically hyperenhancing on arterial phase
  - Rapid washout on portal venous & delayed phases
    - HSCA MR: Typically no delayed hepatocyte phase contrast retention
- Venous invasion common
  - Enhancing thrombus on CT/MR
  - ± arterial waveforms in thrombus on spectral Doppler

TOP DIFFERENTIAL DIAGNOSES

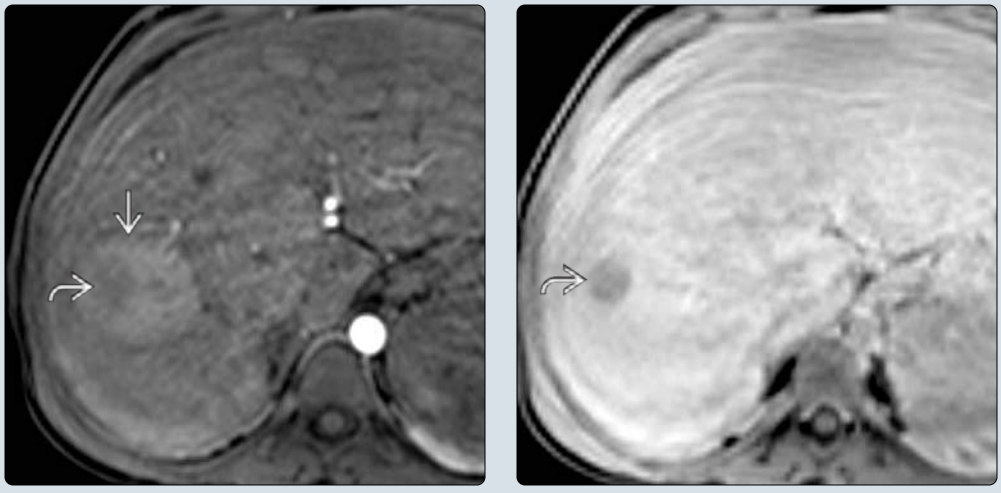
- Hepatic adenoma
- Focal nodular hyperplasia
- Fibrolamellar HCC
- Undifferentiated embryonal sarcoma
- Hepatoblastoma

CLINICAL ISSUES

- 2nd most common primary hepatic malignancy in children after hepatoblastoma
- Typical age: 10-19 years
- Preexisting liver disease in < 50% of patients
- Best therapy: Complete resection
  - 2/3 unresectable at diagnosis due to multifocality, vascular invasion, size, or metastases (25-50%)
    - May become resectable with neoadjuvant therapy
    - Transplant may be curative for localized disease
- Overall survival rate of ~ 10-30%



(Left) *Axial CECT of the liver in a child shows multiple predominantly hypodense hepatic lesions consistent with multifocal hepatocellular carcinoma (HCC). There is a small calcification within 1 of the lesions.* **(Right)** *Axial CECT of the liver in an adolescent with a history of biliary atresia status post Kasai procedure shows a large mass occupying the entirety of the inferior liver. Biliary atresia is a condition that predisposes children to HCC.*



(Left) *Axial T1 C+ FS MR (imaged in the arterial phase) in an adolescent with a-1 antitrypsin deficiency shows a slightly hyperintense mass in the right lobe of the liver. The mass has a concerning nodule-in-nodule appearance with a small hypointense area.* **(Right)** *Axial T1 C+ FS MR (imaged in the delayed phase after hepatocyte-specific contrast agent administration) in the same patient shows no retention of contrast in the smaller, central lesion. This lesion was HCC arising within a dysplastic nodule.*

TERMINOLOGY

Definitions

- Hepatocellular carcinoma (HCC): Malignant tumor of hepatocytes
- Hepatocyte-specific contrast agent (HSCA): MR contrast agent with substantial hepatic uptake & biliary excretion

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous, hypervascular mass in older children
  - Shows washout of contrast on delayed CT & MR
- Size
  - Typically larger at diagnosis than in adults
  - ~ 80% > 4 cm at diagnosis
- Morphology
  - Solitary, multinodular, & diffuse infiltrative patterns
- Signs of vascular invasion
  - Contiguity of tumor & vessel
  - Expansion of vessel diameter
  - Intravascular tumor shows arterial flow & enhancement

CT Findings

- CECT
  - Hyperenhancing mass on arterial-phase imaging
  - Rapid washout on portal venous & delayed-phase images

MR Findings

- Variable signal intensity depending on presence of fat, fibrosis, Ca^{2+}, necrosis, & hemorrhage
- T1WI
  - Iso- or hypointense, though can have hyperintense signal if fat or hemorrhage present
- T2WI
  - Slightly hyperintense
- T1WI C+
  - Heterogeneous hyperenhancement during arterial phase with washout on portal venous & delayed phases
  - Typically hypointense on hepatocyte phase with HSCA

Ultrasonographic Findings

- Grayscale ultrasound
  - Smaller HCCs usually hypoechoic
  - Larger HCCs tend to be heterogeneous
- Color Doppler
  - Mass shows moderate to high internal vascularity
  - Helpful in evaluating portal & hepatic venous invasion
    - Arterial waveforms in lumen of vein confirm tumor thrombus rather than bland thrombus

Imaging Recommendations

- Best imaging tool
  - Multiphase MR saves ionizing radiation exposure from multiphase CT
  - HSCA MR best for satellite lesions

DIFFERENTIAL DIAGNOSIS

Hepatic Adenoma

- May be indistinguishable from HCC on imaging
- Adenoma favored in clinical scenario of adolescent girl on oral contraceptives or steroids

Focal Nodular Hyperplasia

- Arterial phase enhancement plus central scar on CT & MR
- Retains contrast on delayed hepatocyte phase

Fibrolamellar Hepatocellular Carcinoma

- HCC variant in adolescents without underlying liver disease
- α-fetoprotein (AFP) level typically normal
- Scar in 60-75% (uncommon in conventional HCC)
- Ca^{2+} in 35-55% (more common than conventional HCC)

Undifferentiated Embryonal Sarcoma

- Rare; typically 6-10 years of age
- AFP normal
- Appears more cystic at CT/MR due to myxoid stroma

Hepatoblastoma

- Young children (median age: 19 months) with large solid liver mass & ↑ AFP
- Hyperenhancement uncommon

PATHOLOGY

General Features

- Preexisting liver disease in < 50% of patients
  - Cirrhosis, biliary atresia, glycogen storage diseases, α-1 antitrypsin, Wilson disease, hepatitis B/C infection

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal mass, ↑ AFP ~ 70%

Demographics

- 25% of primary pediatric hepatic malignancies
  - 2nd most common after hepatoblastoma
  - Higher incidence in areas with endemic hepatitis B
- M:F= 1.5:1.0
- Typical age: 10-19 years

Natural History & Prognosis

- Overall survival rate of ~ 10-30%

Treatment

- Best therapy: Complete resection
  - 2/3 unresectable at diagnosis due to multifocality, large size, vascular invasion, &/or metastases
    - Metastases present at diagnosis in 25-50%: Lung, abdominal lymph nodes, bone most common
- Additional options: Chemotherapy, chemoembolization, radiofrequency ablation, & transplantation

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Undifferentiated Embryonal Sarcoma

KEY FACTS

TERMINOLOGY

- Malignant hepatic mesenchymal tumor with undifferentiated cells

IMAGING

- Well-circumscribed solitary liver mass in 6-10 year old child
- Typically > 10 cm in size; mean: 14 cm
- Mass appears cystic on CT/MR but solid on ultrasound due to myxoid components
- Septations & mural nodules may be present
- Central hyperdense foci & fluid-debris levels suggest hemorrhage
- Minimal enhancement during arterial phase
- ± peripheral enhancement on delayed phases
- Central hyperdense foci & fluid-debris levels suggest hemorrhage; Ca^{2+} uncommon

TOP DIFFERENTIAL DIAGNOSES

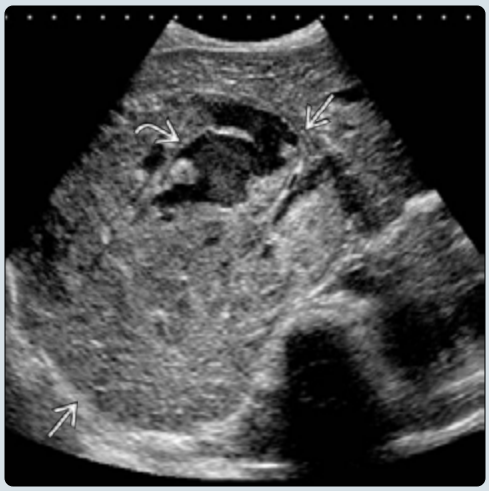
- Hepatoblastoma
- Mesenchymal hamartoma
- Hepatocellular carcinoma
- Hepatic pyogenic abscess

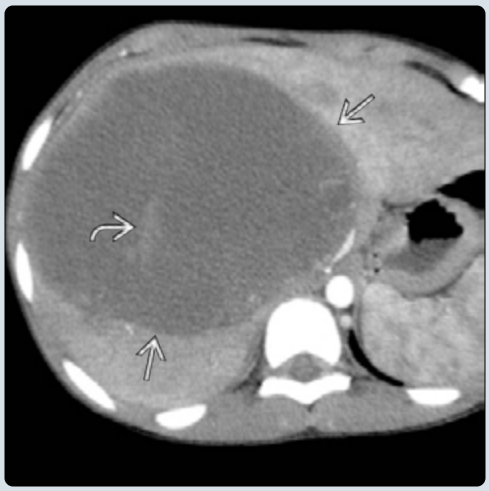
PATHOLOGY

- Thought to arise from dedifferentiation of mesenchymal hamartoma

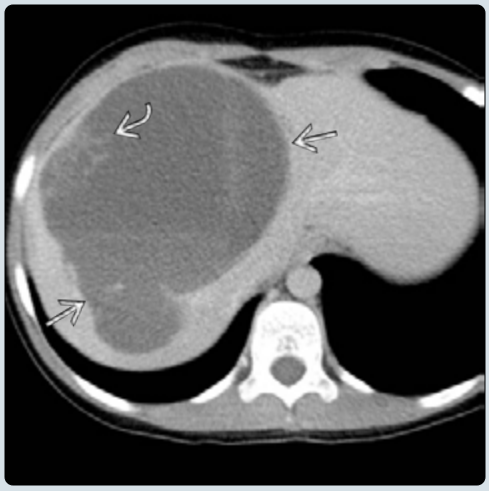
CLINICAL ISSUES

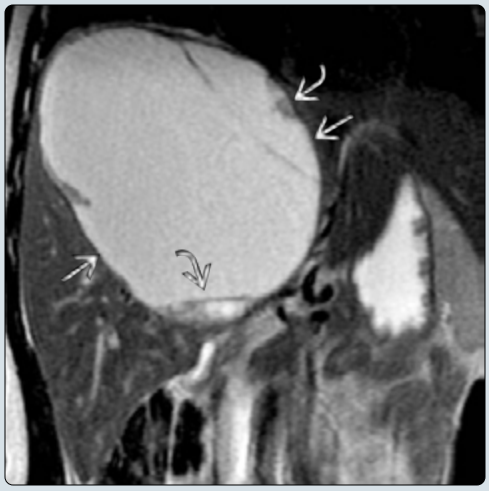
- 3rd most common pediatric hepatic malignancy after hepatoblastoma & hepatocellular carcinoma
- Most frequently occurs between 6-10 years of age
- Most commonly presents with abdominal mass ± abdominal pain; normal AFP levels
  - Fever, weight loss, anorexia, vomiting, diarrhea, lethargy, constipation, respiratory distress less common
- Survival rates ↑ from ~ 30% to > 70% over past 25 years
- Multimodal therapy (including chemotherapy & resection or transplant) key for ↑ survival





(Left) *Transverse ultrasound of the liver in a 10 year old with undifferentiated embryonal sarcoma (UES) shows a heterogeneous, predominately solid mass in the right lobe. The mass has a small irregular cystic focus due to necrosis.* **(Right)** *Axial CECT of the liver in the same child with UES shows a cystic-appearing mass in the right lobe. The mass has subtle, poorly defined foci of increased density.*





(Left) *Delayed phase axial CECT of the liver in a patient with UES shows fine septations & peripheral nodularity to the cystic-appearing right hepatic mass.* **(Right)** *Coronal T2 MR in a patient with UES shows a cystic-appearing mass with fine septations & peripheral nodularity. UES frequently appears cystic by CT & MR due to its myxoid components.*

TERMINOLOGY

Definitions

- Malignant hepatic mesenchymal tumor with undifferentiated cells

IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed solitary liver mass in child 6-10 years of age
  - Mass appears cystic on CT/MR but solid on ultrasound due to myxoid components
- Size
  - > 10 cm; mean: 14 cm

CT Findings

- Mass appears cystic with near fluid attenuation
- Septations & mural nodules may be present
- Central hyperdense foci & fluid-debris levels suggest hemorrhage; Ca^{2+} uncommon
- Minimal enhancement during arterial phase
- May have peripheral enhancement on delayed phases

MR Findings

- Mass follows fluid signal intensity on T1 & T2
- Fibrous pseudocapsule gives hypointense rim
- Internal hemorrhage causes foci of ↑T1 & ↓T2 signal
- Heterogeneous enhancement in peripheral & solid portions of mass
- Does not retain contrast during hepatocyte phase (if hepatocyte specific contrast agent used)

Ultrasonographic Findings

- Isoechoic to hyperechoic solid but heterogeneous mass
- Small anechoic spaces correspond to foci of necrosis, hemorrhage, & cystic degeneration

Imaging Recommendations

- Best imaging tool
  - Ultrasound localizes mass to liver & confirms solid nature of tumor
  - CT/MR used to evaluate extent of mass

DIFFERENTIAL DIAGNOSIS

Hepatoblastoma

- Most common primary liver malignancy in children
- Median age of detection: 19 months
- Solid mass with rare cystic components
- More common in premature infants, Beckwith-Wiedemann syndrome, & familial adenomatous polyposis

Mesenchymal Hamartoma

- 2nd most common benign liver mass in young children (after congenital & infantile hemangiomas)
- Most common < 2 years of age
- Classically multicystic but may be largely/entirely solid
- Possible rare dedifferentiation to undifferentiated embryonal sarcoma

Hepatocellular Carcinoma

- More common in children > 10 years of age
  - Most common liver malignancy in adolescents
- Only 30-50% of pediatric patients have underlying liver disease

Hepatic Pyogenic Abscess

- Uncommon in children; patients typically ill
- Thick, irregular rim enhancement + surrounding edema

PATHOLOGY

General Features

- Etiology
  - Thought to arise from mesenchymal hamartoma
    - Malignant dedifferentiation due to translocation in long arm of chromosome 19

Gross Pathologic & Surgical Features

- Heterogeneous mass with glistening gray-white appearance alternating with gelatinous foci
  - Dark brown foci due to hemorrhage
  - Yellow regions of necrosis

Microscopic Features

- Fibrous pseudocapsule containing cords & clusters of hepatocytes
- Mass has sarcomatous cells with myxoid background

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal mass ± abdominal pain; AFP levels normal
- Other signs/symptoms
  - Fever, weight loss, anorexia, vomiting, diarrhea, lethargy, constipation, respiratory distress

Demographics

- Accounts for 9-13% of pediatric liver tumors
  - 3rd most common pediatric hepatic malignancy after hepatoblastoma & hepatocellular carcinoma
- Most common between 6-10 years of age

Natural History & Prognosis

- Survival rates ↑ from ~ 30% to > 70% over past 25 years

Treatment

- Multimodal therapy (including chemotherapy & resection or transplant) key for ↑ survival

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Biliary Atresia

KEY FACTS

TERMINOLOGY

- Biliary atresia (BA): Absent or severely deficient extrahepatic biliary tree

IMAGING

- US to exclude other causes of neonatal jaundice; characteristic findings can be very suggestive of BA
  - Absent or abnormal small irregular gallbladder in vast majority (gallbladder ghost sign)
  - Echogenic fibrous tissue (triangular cord sign) anterior to portal vein at site of obliterated extrahepatic biliary duct
  - No biliary ductal dilation
  - Liver echotexture typically normal
- Hepatobiliary scan: No radiotracer excretion into intestines; gallbladder visible in up to 25%
- Intraoperative cholangiogram: No biliary enteric channel

TOP DIFFERENTIAL DIAGNOSES

- Neonatal hepatitis
- Bile-plug syndrome
- Alagille syndrome
- Choledochal malformation

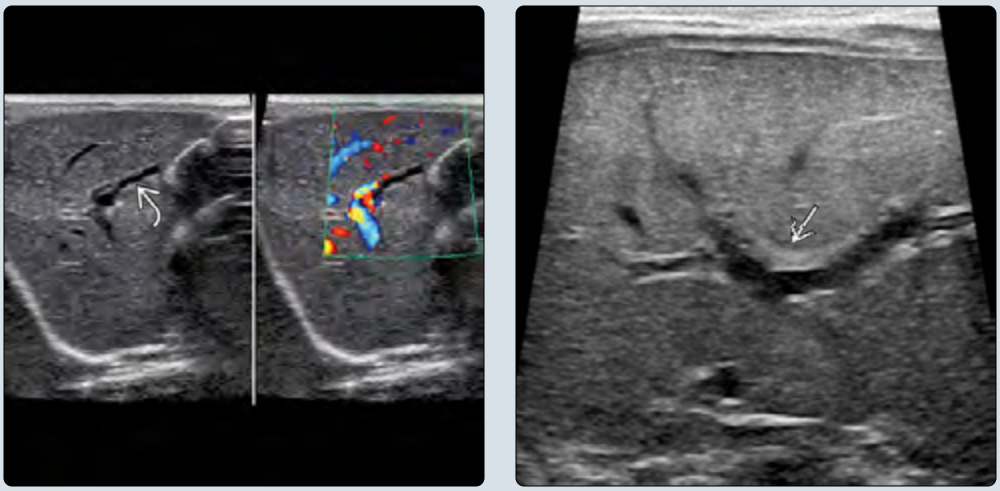
PATHOLOGY

- Preduodenal portal vein, interrupted inferior vena cava, situs anomalies, congenital heart disease, & polysplenia (or asplenia) in up to 15% (BASM syndrome)

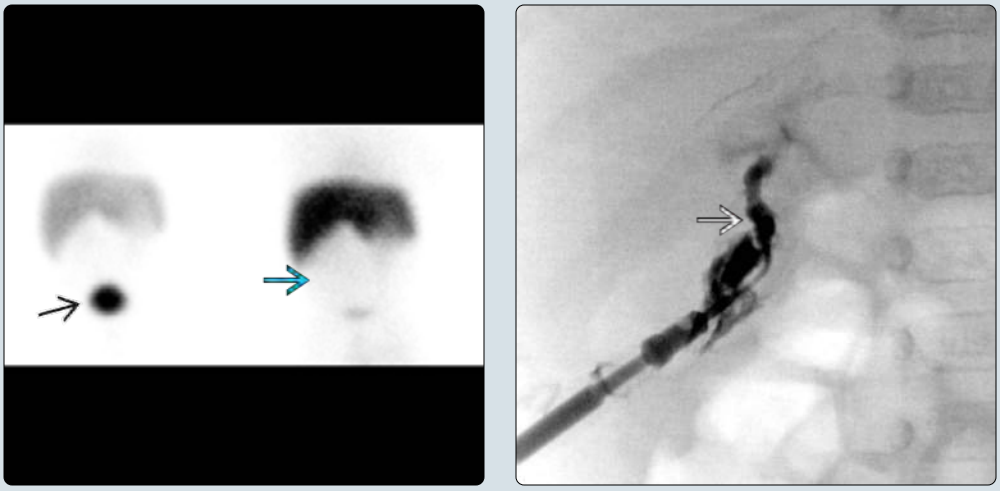
CLINICAL ISSUES

- Neonatal jaundice with conjugated (direct) hyperbilirubinemia
  - Affects 1 in 10,000-13,000 newborn infants
- Initially, hepatocytes function well with gradual deterioration
- Kasai portoenterostomy temporarily effective in 90% if performed < 2 months of age; ↓ to < 50% if > 3 months
- Liver transplant ultimately required in most by adulthood, even with prompt Kasai
  - Due to gradual fibrosis & portal hypertension

(Left) *Split-screen grayscale & color Doppler US show a very small & irregularly shaped gallbladder ➡ along the inferior margin of the liver in a newborn with jaundice, despite adequate fasting. This crenulated appearance is sometimes called the ghost gallbladder.* **(Right)** *Transverse US shows echogenic tissue ➡ at the expected location of the common bile duct anterior to the portal vein, the triangular cord sign. This sign & the ghost gallbladder strongly suggest biliary atresia (BA).*



(Left) *Hepatobiliary scintigraphy in a jaundiced infant shows no intestinal excretion of mebrofenin. The hepatic radiotracer is slowly excreted by the kidneys into the urinary bladder ⇨. A 24-hour delayed image also shows no intestinal radiotracer ⇨, typical of BA.* **(Right)** *Intraoperative cholangiogram through a hypoplastic gallbladder shows a short segment of common hepatic duct ➡ (but no intestinal drainage), confirming BA. A portoenterostomy (Kasai procedure) followed.*



TERMINOLOGY

Abbreviations

- Biliary atresia (BA)

Definitions

- Absent or severely deficient extrahepatic biliary tree

IMAGING

General Features

- Best diagnostic clue
  - US shows small & irregular or absent gallbladder with obliterated bile duct anterior to portal vein
  - Hepatobiliary scan shows lack of radiotracer excretion into intestines
  - Intraoperative cholangiogram shows no biliary enteric channel
- Morphology
  - Often confused clinically with neonatal hepatitis (common nonsurgical disease)
  - Vast majority of biliary atresia cases show abnormal or absent gallbladder

Ultrasonographic Findings

- Grayscale ultrasound
  - Liver echotexture typically normal, though liver may be enlarged
  - Gallbladder typically absent or abnormal
    - Small (< 19-25 mm in length) with irregular shape & discontinuous echogenic wall: Gallbladder ghost triad
    - Improves sonographic accuracy for BA
  - Extrahepatic bile ducts usually not visible in BA
    - Echogenic obliterated/fibrotic remnant of common bile duct (CBD) anterior to portal vein: Triangular cord sign
    - Dilated bile ducts (intra- or extrahepatic) usually not present
      - Rare cystic BA forms
  - Associated anomalies of BA-splenic malformation (BASM) syndrome
    - Polysplenia
    - Preduodenal portal vein
    - Transverse liver (showing continuity of portal veins in leftward hepatic tissue)
    - Interrupted inferior vena cava (IVC)
    - Cardiac anomalies
  - Acoustic radiation force impulse (ARFI) technique shows promise of differentiating BA from other causes of neonatal jaundice based on liver stiffness
  - Post-Kasai procedure, liver often shows gradual fibrosis/cirrhosis with portal hypertension, splenomegaly, & varices
- Color Doppler
  - Useful to demonstrate main portal vein & hepatic artery when searching for echogenic triangular cord
  - ± ↑ subcapsular flow

MR Findings

- MRCP
  - Limited utility for BA
    - Complete biliary tree only visible in 60-75% of normal patients < 3 months old
    - 90% sensitivity, 77% specificity, 82% accuracy
- T1WI, T2WI
  - Post-Kasai procedure, liver often shows gradual fibrosis/cirrhosis with portal hypertension, splenomegaly, & varices

Nuclear Medicine Findings

- Hepatobiliary scan
  - Tc-99m disofenin (DISIDA) & mebrofenin (BRIDA) have highest hepatic extraction rates & shortest transit times of hepatobiliary radiotracers
  - Pretreat with oral phenobarbital (5 mg/kg/day in divided doses x 5 days)
    - Potent inducer of hepatic microsomal enzymes, enhances biliary excretion & improves scintigraphic accuracy
    - Ursodeoxycholic acid has also been used as choleretic prescintigraphy (20 mg/kg every 12 hours for 48-72 hours) with good results
  - Hepatocyte uptake & extraction of radiotracer from blood pool usually preserved in first 2-3 months of life, later deteriorates
  - Lack of excretion into intestines on 24-hour delayed images highly suggestive of BA or other extrahepatic occlusion
    - Excretion into intestines effectively excludes BA
  - Visualization of gallbladder not helpful; seen in 25%
  - High sensitivity (~ 100%), but 87% specificity, 91% accuracy

Other Modality Findings

- ERCP invasive, requires general anesthesia, uses ionizing radiation, & has significant morbidity (1-7%) & failure (3-14%) rates
- Analysis of duodenal drainage difficult to perform, not yet standardized; sensitivity of ~ 97%, specificity of ~ 93%

Imaging Recommendations

- Best imaging tool
  - US performed 1st
    - Traditionally to exclude other causes of jaundice
    - Combination of characteristic findings can strongly suggest BA
  - If gallbladder seen by US, some surgeons proceed directly to intraoperative cholangiogram with liver biopsy; if patent ducts not demonstrated by cholangiogram, definitive surgery performed
  - Hepatobiliary scintigraphy requires 5 days pretreatment with phenobarbital for optimal accuracy

DIFFERENTIAL DIAGNOSIS

Neonatal Hepatitis

- Very common entity; usually self-limited medical disease, though can be caused by hepatitis A, hepatitis B, cytomegalovirus, rubella, toxoplasmosis, α-1-antitrypsin deficiency, familial recurrent cholestasis, or other metabolic disorders

Bile-Plug Syndrome

- Due to cystic fibrosis, dehydration, sepsis, hemolytic disorders, or total parenteral nutrition

Alagille Syndrome

- Intrahepatic ducts sparse & malformed
- Pulmonic stenosis, characteristic facies, butterfly vertebra, CNS arterial anomalies

Choledochal Malformation

- 5 subtypes of localized dilation of extrahepatic biliary tree
- ± dilated intrahepatic ducts

PATHOLOGY

General Features

- Etiology
  - Hypoplastic, atretic, or fibrosed extrahepatic ducts that worsen in perinatal period
  - Possibly result of prenatal biliary duct inflammation of unknown etiology
  - Proposed mechanism involves initial virus-induced, progressive T-cell-mediated inflammatory obliteration of bile ducts
- Associated abnormalities
  - Preduodenal portal vein, interrupted IVC, congenital heart disease, situs anomalies, & polysplenia (or asplenia) in BASM (10-15%)
  - Another 10-25% have other anomalies, most commonly involving abdomen & genitourinary tract

Gross Pathologic & Surgical Features

- Absent extrahepatic ducts; cirrhosis if diagnosis delayed
- ~ 12% of BA patients have patent proximal ducts & can have simple reanastomosis surgery; 88% require Kasai procedure

Microscopic Features

- Periportal fibrosis, proliferation of small intrahepatic ducts, absent extrahepatic bile ducts, absence of multinucleated giant cells in liver

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Progressive conjugated (direct) hyperbilirubinemia in neonatal period
    - Bilirubin unconjugated in sepsis, hepatitis, & metabolic hepatocellular diseases

Demographics

- Affects 1 in 10,000-13,000 newborn infants
- Jaundice evident in immediate perinatal period
- No gender or racial predilection

Natural History & Prognosis

- Prompt diagnosis crucial to surgical success
  - Initially, hepatocyte function preserved; hepatocyte function gradually deteriorates
- Kasai portoenterostomy temporarily effective in 90% if performed < 2 months of age; ↓ to < 50% if > 3 months of age
  - Poor prognosis if total bilirubin not < 2 mg/dL within 3 months of Kasai procedure
  - Steroid treatment post Kasai hastens clearance of jaundice; long-term effects unclear
  - 4-year survival rate after Kasai portoenterostomy: ~ 40%
- Most patients ultimately require liver transplantation
  - < 18% who have prompt Kasai procedures avoid liver transplantation ≥ 20 years later
  - BA most common reason for pediatric liver transplant

Treatment

- Kasai portoenterostomy
  - Intestinal loop anastomosed to dissected surface of porta hepatis
- Liver transplant

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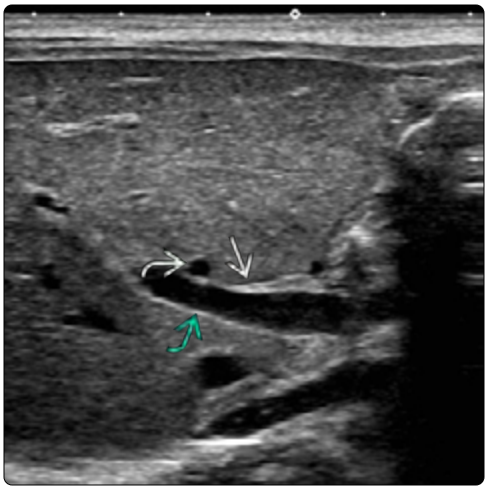
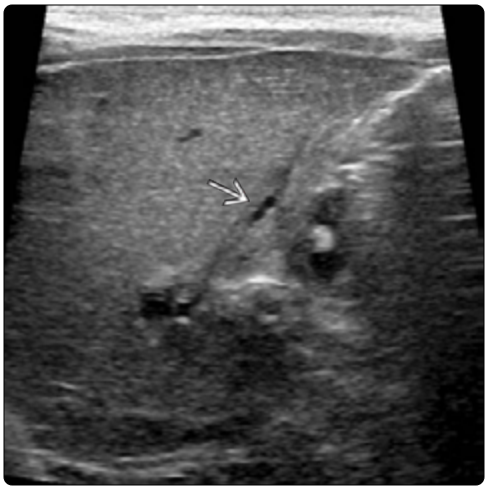
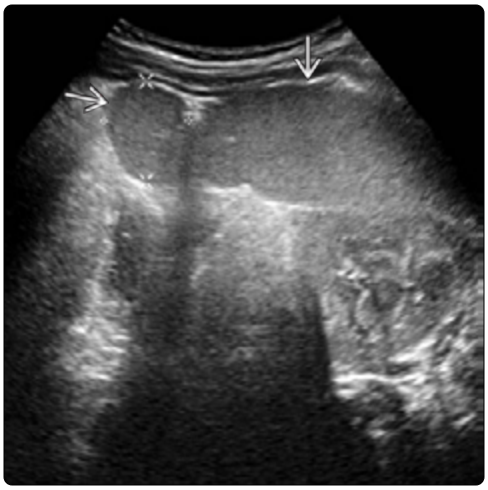
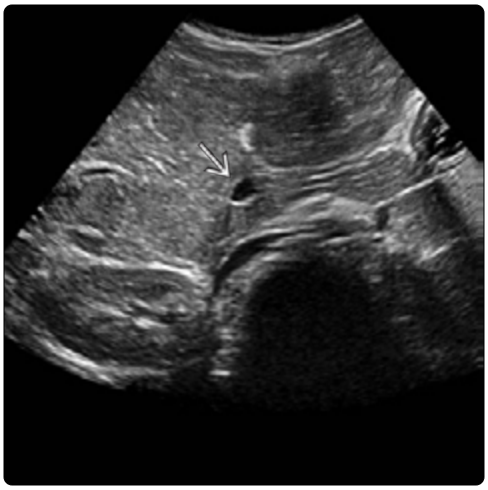


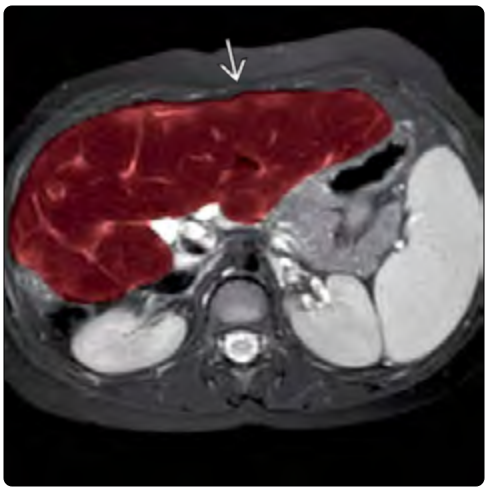


(Left) *Transverse US of the porta hepatis in a 20 day old shows a normal portal vein & hepatic artery but only echogenic fibrotic tissue where the common bile duct should be (triangular cord sign).* **(Right)** *Transverse US in a jaundiced infant shows a small, irregularly shaped gallbladder despite an adequate fast. In addition to the gallbladder ghost, this patient also had a triangular cord sign of BA.*





(Left) *Oblique US in a newborn with jaundice shows at least 2 spleens in the left upper quadrant, a finding that should prompt investigation for other BA-associated anomalies as these features could be seen in biliary atresia splenic malformation (BASM) syndrome.* **(Right)** *Paramidline US shows a preduodenal portal vein along the liver margin in the same patient with BA & polysplenia, further suggesting BASM syndrome.*



(Left) *Axial T2 FS MR in the same child 9 years status post Kasai procedure shows a mildly lobulated liver contour suggesting fibrosis/cirrhosis. The red shading is due to post processing in order to determine the liver volume.* **(Right)** *Intraoperative photograph during a Kasai procedure shows the discolored liver (related to chronic cholestasis), tiny spider angiomata, & lack of a common bile duct in the porta hepatis in this patient with BA.*

Choledochal Cyst

KEY FACTS

TERMINOLOGY

- Choledochal cyst: Spectrum of malformations involving extrahepatic & intrahepatic bile ducts
- Etiology may be due to pancreaticobiliary maljunction
  - Pancreatic duct joins common bile duct (CBD) proximal to sphincter of Oddi → biliary reflux of pancreatic enzymes
- High risk (in long term) of cholangiocarcinoma if choledochal malformation not resected

IMAGING

- US in child with jaundice & elevated liver enzymes
  - Round or tubular cystic right upper quadrant mass separate from gallbladder
    - CBD dilation > 10 mm very suggestive in child
- MRCP for detailed preoperative assessment of ductal anatomy & pancreaticobiliary maljunction
  - Delayed scan with hepatobiliary contrast agent may confirm connection of cyst to biliary tree

TOP DIFFERENTIAL DIAGNOSES

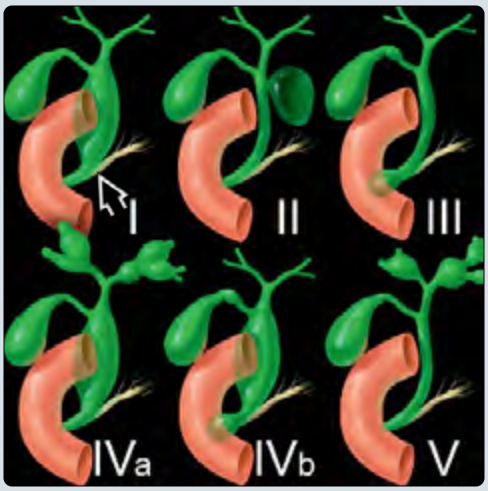
- Primary sclerosing cholangitis
- Obstructing choledocholithiasis
- Gastrointestinal duplication cyst
- Pancreatic pseudocyst

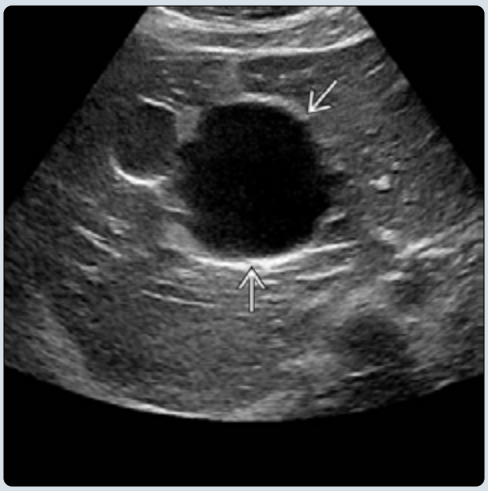
PATHOLOGY

- Classification modified by Todani in 1977
  - Type I cyst: Segmental or diffuse fusiform dilation of CBD; most common variety (75-95% of cases)
  - Type II cyst: Diverticulum of duct
  - Type III cyst: Choledochocele protruding into duodenum
  - Type IV: Discontinuous extrahepatic bile duct cysts, isolated (type IVb) or Caroli type (type IVa)
  - Type V: Intrahepatic cystic dilations (Caroli disease)

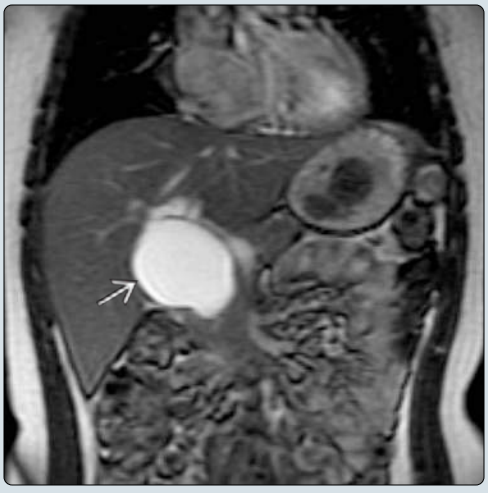
CLINICAL ISSUES

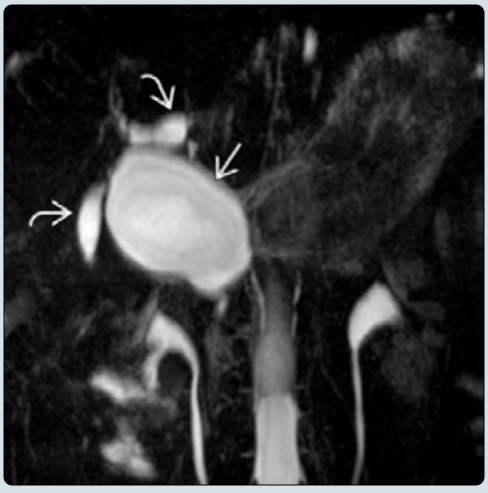
- General treatment: Cyst resection & biliary diversion/portoenterostomy





(Left) *Graphic shows various types of choledochal malformations. Note the anomalous pancreaticobiliary junction ➔ with the pancreatic duct inserting into the common bile duct proximal to the sphincter of Oddi. Characteristics of the types are described in the chapter text.* **(Right)** *Transverse US of the liver in a 9 year old shows a large simple-appearing cyst ➔ adjacent to the gallbladder.*





(Left) *Coronal SSFP MR of the same child shows the elliptical cyst ➔ in the porta hepatis extending into the pancreatic head, typical of a choledochal cyst.* **(Right)** *Coronal volumetric image from an MRCP in the same patient shows a severely enlarged common hepatic duct ➔ & focally dilated central intrahepatic bile ducts ➔ in this case of a type IVa choledochal cyst.*

TERMINOLOGY

Synonyms

- Choledochal malformations, common bile duct (CBD) cyst or diverticulum, choledochocele

Definitions

- Choledochal cyst: Spectrum of malformations involving extrahepatic & intrahepatic bile ducts
- Etiology may be due to pancreaticobiliary maljunction
  - Pancreatic duct joins CBD proximal to sphincter of Oddi → biliary reflux of pancreatic enzymes
- ↑ cholangiocarcinoma risk in patients with choledochal malformations

IMAGING

General Features

- Best diagnostic clue
  - Marked dilation of biliary tree, focal or diffuse
- Location
  - May involve intrahepatic bile ducts, extrahepatic ducts, or both
- Size
  - Bile duct dilation > 10 mm very suggestive in child
- Morphology
  - Ductal dilation may be continuous or multifocal

Ultrasonographic Findings

- Grayscale ultrasound
  - Round or tubular cystic right upper quadrant lesion separate from gallbladder
  - ± intrahepatic ductal dilation
- Color Doppler
  - Shows lack of flow in anechoic round/tubular mass, confirming cyst rather than abnormal vessel
  - Useful to demonstrate position & displacement of adjacent vessels

MR Findings

- T2WI, MR cholangiogram to show
  - Anatomy of dilated ducts & variants
  - Pancreaticobiliary maljunction
- T1 C+ with hepatocyte-specific contrast agent
  - Delayed images can confirm biliary connection of cyst

Nuclear Medicine Findings

- Hepatobiliary scan shows early photopenia at hilum with delayed filling
- Can help show bile leak after resection

Other Modality Findings

- ERCP & PTC usually reserved for difficult, complex cases or intervention

Imaging Recommendations

- Best imaging tool
  - US in child with jaundice & elevated liver enzymes
    - Shows dilated biliary tree & extent of ductal involvement
  - MRCP for detailed preoperative assessment of ductal anatomy
    - Has replaced preoperative percutaneous cholangiogram
  - Hepatobiliary nuclear scans for functional evaluation

DIFFERENTIAL DIAGNOSIS

Primary Sclerosing Cholangitis

- Irregular beading with dilation & stenosis of bile ducts
- Look for findings of ulcerative colitis

Obstructing Choledocholithiasis

- Stone disease may cause obstruction at several levels
- Ductal dilation typically < 10 mm

Gastrointestinal Duplication Cyst

- Duodenal cyst can involve porta hepatis
- Look for "gut signature" & peristalsis of cyst

Pancreatic Pseudocyst

- Round pancreatic head collection can mimic dilated CBD
- History/findings of recent pancreatitis

Caroli Disease

- Localized saccular ectasia of intrahepatic bile ducts
- Central dot sign of encased portal radicles

PATHOLOGY

General Features

- Etiology
  - Primary theory: Anomalous junction of common biliary & pancreatic ducts → conduit for mixing (& reflux) of pancreatic enzymes & bile
    - Dilation & structural weakness of CBD (with destruction of elastic fibers) after reflux of pancreatic secretions in animal models
    - Unclear that this leads to marked duct dilation
  - Additional theories: ↓ in ganglion cells in narrow portion of bile duct causing ↑ intraluminal pressure; preceding viral infection; familial pattern of inheritance; failure of recanalization
- Genetics
  - Autosomal recessive polycystic kidney disease often associated with Caroli syndrome
    - Ciliopathy underlies these disorders; likely different etiology from remaining choledochal malformations

Staging, Grading, & Classification

- Classification modified by Todani in 1977
  - Type I cyst: Segmental or diffuse fusiform dilation of CBD; most common variety (75-95% of cases)
  - Type II cyst: Diverticulum of duct, usually protruding from lateral wall
  - Type III cyst: Choledochocele, most often of duodenal wall (protruding into duodenal lumen)
  - Type IV: Multiple discontinuous extrahepatic bile duct cysts, isolated (type IVb) or with Caroli-type intrahepatic biliary cysts (type IVa)
  - Type V: Multifocal cystic dilations of intrahepatic bile ducts (Caroli disease)
    - Caroli syndrome (large & small bile duct ectasia with congenital hepatic fibrosis) > > Caroli disease (large bile duct ectasia only)

- Additional proposed modifications
  - Type ID: Dilated cystic duct + dilated CBD & CHD
  - Type VI: Isolated dilation of cystic duct
- Intrahepatic ductal dilation preoperatively may resolve postoperatively, changing cyst classification

Gross Pathologic & Surgical Features

- Range in diameter from few centimeters to > 15 cm
- Cyst wall thickened, fibrotic, & occasionally calcified in adults

Microscopic Features

- Varying degrees of chronic inflammation
- Biliary epithelium lining cyst often intact in infants
- Type III cysts (choledochocele) often lined by duodenal mucosa but occasionally have biliary epithelium
- Goblet-cell metaplasia & epithelial dysplasia with nuclear hyperchromasia, irregularity, & loss of polarity have been described
  - May play role in subsequent development of carcinoma

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prolonged neonatal cholestasis, so-called infantile obstructive cholangiopathy
  - Infants: Jaundice, acholic stools, hepatomegaly, & palpable abdominal mass
  - Adults: Upper abdominal pain, jaundice, cholangitis, & pancreatitis

Demographics

- Age
  - 2/3 of all choledochal malformations diagnosed before 10 years of age
  - Only 20% diagnosed in adults
- Gender
  - M:F = 1:3-4
- Epidemiology
  - More common in Asian than Western countries
    - 1 in 100,000-150,000 live births in USA vs. 1 in 1,000 live births in Japan
    - ~ 1/3 of all reported cases occur in Japanese patients

Natural History & Prognosis

- Low-grade biliary obstruction may develop cirrhosis & portal hypertension
- Complications: Bile duct perforation, biliary stone formation, bacterial cholangitis with subsequent hepatic abscess & sepsis, biliary strictures, low-grade biliary obstruction → cirrhosis & portal hypertension, development of bile duct carcinomas
  - Prevalence of cancer (adenocarcinoma) in choledochal cysts: 2-18% (5-35x ↑ risk)
  - Both Caroli disease & Caroli syndrome associated with risk of cholangiocarcinoma (100x that of general population)

Treatment

- Type I: Complete surgical excision + biliary drainage procedure, typically Roux-en-Y choledochojejunostomy
  - Excision with hepaticoduodenostomy has higher rate of bile reflux/gastritis
- Type II: Usually excised entirely; defect in CBD closed primarily over T tube
  - Type II usually diverticula of bile duct
- Type III: Choledochocele with diameter < 3 cm may be approached endoscopically with sphincterotomy
  - Cysts > 3 cm often associated with some degree of duodenal obstruction → excised surgically using transduodenal approach
- Type IV: Dilated extrahepatic duct completely excised + biliary-enteric drainage procedure
  - No surgery specifically directed at intrahepatic ductal disease
- Type V: Caroli disease, when limited to single hepatic lobe (usually left), may be resected
  - Liver transplantation necessary in diffuse disease with liver failure
- Internal & external drainage, without cyst excision → unacceptably high rate of cholangitis, does not alter malignant potential
- After resection of choledochal cysts
  - < 0.5% develop cancer before 18 years
  - 11% develop cancer as adult; mean age: 42 years

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Bile duct measuring > 10 mm in childhood nearly always due to choledochal malformation

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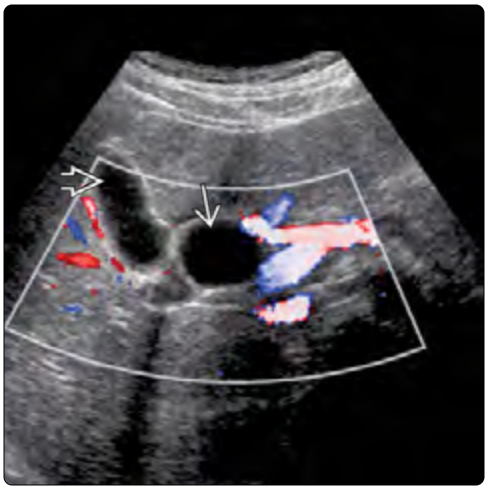
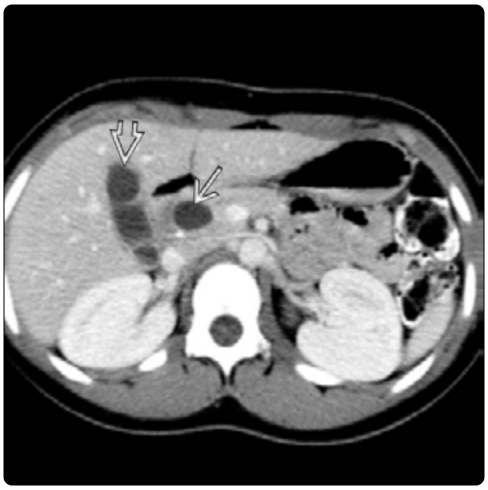
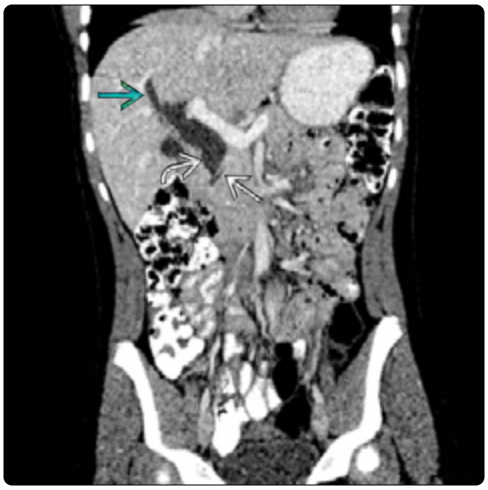
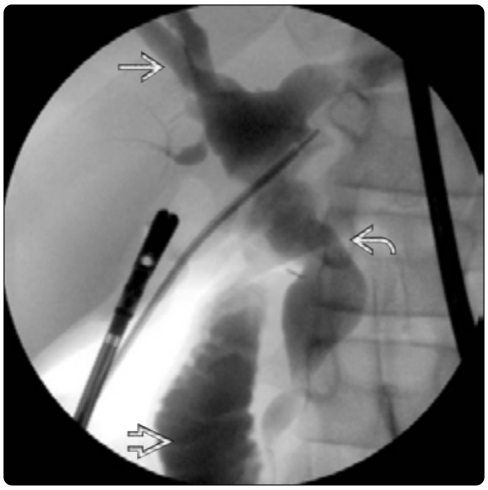


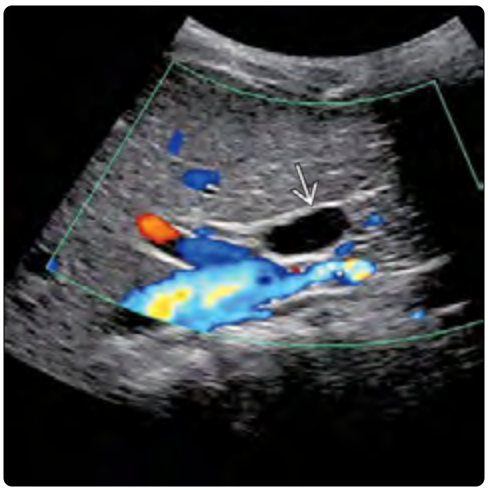


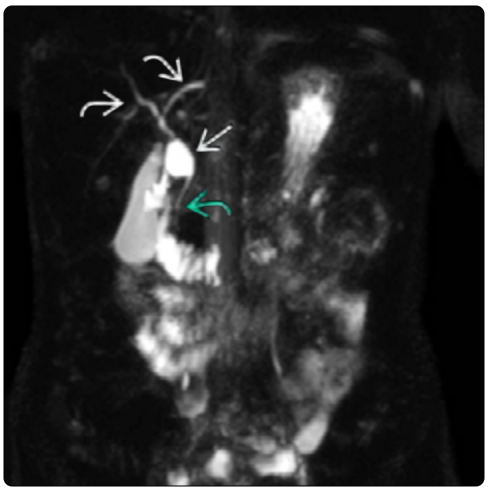
(Left) *Transverse color Doppler US through the right upper quadrant in a 12 year old shows a normal gallbladder ⇨ with diffuse, marked dilation of the common bile duct ⇨.* **(Right)** *Axial CECT in the same patient shows marked dilation of the common bile duct ⇨ with a normal gallbladder ⇨. Mild intrahepatic ductal dilation was also noted (not shown).*





(Left) *Coronal CECT in the same patient shows dilation of the central intrahepatic ⇨ as well as common hepatic & common bile ⇨ ducts extending to the pancreas. A short segment of dilated pancreatic duct ⇨ is also seen.* **(Right)** *Intraoperative cholangiogram in the same patient shows moderate intrahepatic ⇨ & marked extrahepatic ⇨ ductal dilation. However, the biliary tree still drained into the duodenum ⇨. This patient had a type I choledochal malformation repaired surgically.*





(Left) *Color Doppler US in an 8-year-old girl shows focal dilation of the common bile duct ⇨ but no intrahepatic biliary dilation.* **(Right)** *Coronal volumetric MRCP in the same girl shows a focally dilated common bile duct ⇨ tapering to normal caliber ⇨ as it enters the duodenum. The intrahepatic bile ducts ⇨ are normal in caliber.*

Caroli Disease

KEY FACTS

TERMINOLOGY

- Congenital hepatic fibrosis (CHF): Fibrotic tissue between portal tracts with persistent ductal plate structure of intralobular ducts
- Caroli disease (CD): Saccular dilation of large intrahepatic bile ducts (IHBD)
- Caroli syndrome (CS): CHF + CD

IMAGING

- Multiple intrahepatic cystic foci connected to biliary tree
- Central dot sign: Cross section of dilated IHBD surrounding portal vein radicle
- Hepatic involvement can be segmental, lobar, or diffuse

TOP DIFFERENTIAL DIAGNOSES

- Choledochal cyst
- Primary sclerosing cholangitis

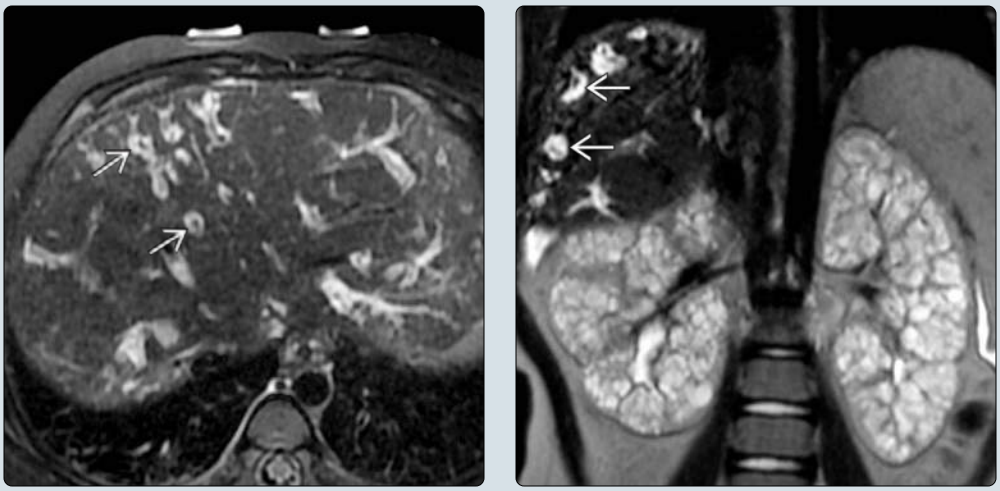
PATHOLOGY

- Autosomal recessive inheritance
- CD, CS, & autosomal recessive polycystic kidney disease caused by impaired ciliary function (ciliopathy)

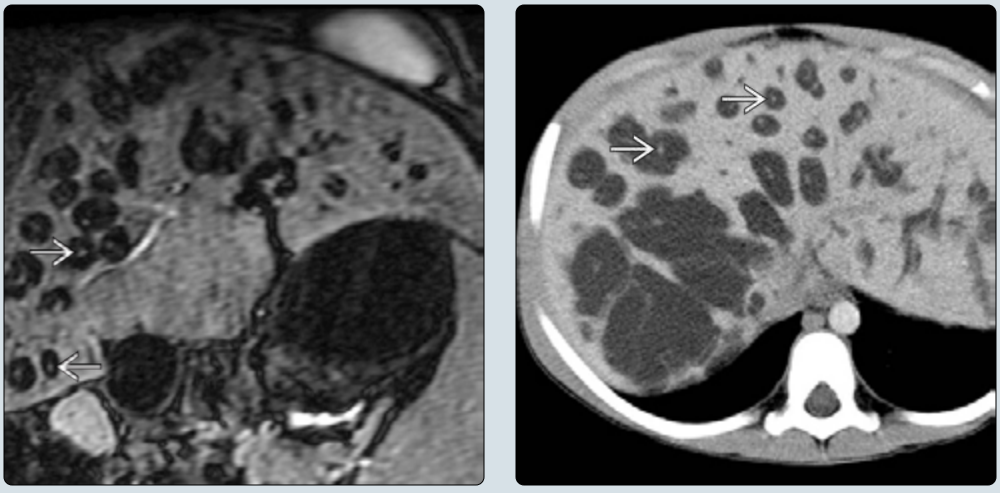
CLINICAL ISSUES

- Rare (1:1 million of general population)
- Patients can be diagnosed at any age
- Symptoms/complications
  - RUQ pain, fever, & jaundice
  - Stone formation (95%), recurrent cholangitis
  - Hepatic fibrosis with portal hypertension → hepatosplenomegaly & gastrointestinal tract bleeding
  - Cholangiocarcinoma ultimately develops in 7% of patients (100x ↑ risk compared to general population)
- Treatment
  - Ursodeoxycholic acid to ↓ bile duct stones; broad-spectrum antibiotics for cholangitis
  - Biliary decompression: External drainage & biliary-enteric anastomosis effective but with ↑ risk of infection
  - Resection ranging from segmentectomy to transplant

(Left) *Axial T2 FS MR shows dilated intrahepatic bile ducts extending to the liver periphery. Several of the ducts have a visible central dot of low signal intensity ➡ due to encased portal radicles. This appearance is typical of Caroli disease.* **(Right)** *Coronal T2 MR shows dilated intrahepatic bile ducts at the periphery of the liver. The dilated ducts have a central dot of low signal intensity ➡ consistent with entrapped portal radicles. The kidneys are markedly enlarged & hyperintense due to innumerable small cysts, also typical of Caroli disease.*



(Left) *Coronal T1 MR in the early arterial phase after contrast injection shows dilated intrahepatic bile ducts with an enhancing central dot ➡. The central dot represents the portal radicle surrounded by a dilated intrahepatic bile duct.* **(Right)** *Axial CECT shows dilated round to ovoid intrahepatic bile ducts. The central dot sign ➡ of Caroli disease is visible at numerous sites on this image.*



TERMINOLOGY

Definitions

- Congenital hepatic fibrosis (CHF): Presence of fibrotic tissue between portal tracts with persistent ductal plate structure of intralobular ducts
- Caroli disease (CD): Saccular dilation of large intrahepatic bile ducts (IHBD)
- Caroli syndrome (CS): CHF + CD

IMAGING

General Features

- Best diagnostic clue
  - Central dot sign: Portal radicle encased by dilated IHBD
  - Cholangiography confirms direct communication of segmental saccular ductal ectasia with IHBDs
- Location
  - Hepatic involvement can be segmental, lobar, or diffuse
  - Common bile duct classically spared; dilation (in up to 50%) due to cholangitis, stones
- Size
  - Varying diameters of cystic-appearing dilated IHBDs in same patient (ranging from millimeters to centimeters)

MR Findings

- T2WI
  - Numerous "cysts" scattered throughout liver
  - Small, round, hypointense central dots visible in dilated, hyperintense cystic-appearing IHBDs
    - Due to flow void of portal radicle in IHBD cross section
    - ± larger biliary filling defects from stones, sludge
  - Renal involvement
    - Enlarged hyperintense kidneys
      - Due to diffuse medullary or corticomedullary cysts/tubular ectasia
- T1WI C+
  - Central dot of enhancement (of portal vein radicle) surrounded by dilated IHBD
  - Heterogeneous enhancement of liver parenchyma from hepatic fibrosis
- MRCP
  - Direct communication of segmental saccular ductal ectasia with IHBDs
  - ± intraluminal filling defects of sludge or calculi within dilated IHBDs

Ultrasonographic Findings

- Grayscale ultrasound
  - Numerous "cysts" scattered throughout liver ± visible biliary communication
  - Intraluminal portal vein sign = central dot sign
    - Portal radicle surrounded by cystic-appearing IHBD
  - Intraductal bridging septa: Echogenic septa completely or incompletely traversing dilated IHBDs
- Color Doppler
  - Flow within central dot (portal vein radicle)

DIFFERENTIAL DIAGNOSIS

Choledochal Cyst

- Extrahepatic bile ducts affected in 90%
- Type IV: Cystic dilation of common bile duct ± intrahepatic ducts (type IVa has intrahepatic biliary cysts)
- Not inherited; no renal disease

Primary Sclerosing Cholangitis

- Dilation of both IHBD & extrahepatic bile ducts with multiple irregular strictures
- ± history of ulcerative colitis

PATHOLOGY

General Features

- Etiology
  - CD, CS, & autosomal recessive polycystic kidney disease caused by impaired ciliary function (ciliopathy)
    - Ciliopathy during fetal development leads to ductal plate malformation
- Genetics
  - Autosomal recessive

Staging, Grading, & Classification

- Todani classification of choledochal cysts included CD as type V; CD no longer considered as choledochal cyst

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Intermittent abdominal/right upper quadrant pain
- Other signs/symptoms
  - Recurrent cholangitis, fever, & jaundice
  - Hepatic fibrosis with portal hypertension → hepatosplenomegaly & GI bleeding
  - Renal failure may dominate clinical picture in infants
- Complications
  - Stone formation (95%), recurrent cholangitis, end-stage liver disease
  - Cholangiocarcinoma (ultimately) in 7% of patients
    - 100x ↑ risk compared to general population

Demographics

- Age
  - Patients can be diagnosed at any age
  - Patients diagnosed in infancy have more severe disease
- Epidemiology
  - Rare; 1:1 million of general population

Treatment

- Ursodeoxycholic acid (to ↓ biliary stones) & broad-spectrum antibiotics (for cholangitis)
- Biliary decompression: External drainage & biliary-enteric anastomosis effective but with ↑ risk of infection
- Resection ranging from segmentectomy to transplant

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KEY FACTS

TERMINOLOGY

- Cirrhosis: Liver disease characterized by bridging fibrosis
  - Variety of underlying disorders may lead to cirrhosis

IMAGING

- Nodular contour of liver, easiest to see along free margin
- US: Liver has coarsened echotexture
- MR: Lace-like ↓ T1 signal of fibrosis
  - MR can detect underlying deposition (fat, iron, etc.)
  - Hepatocyte-specific contrast agents allow characterization of nodules
- MR or US elastography can determine liver stiffness as marker of fibrosis
- Progressive fibrosis leads to ↓ hepatic size
- Signs of portal hypertension: Ascites, splenomegaly, varices

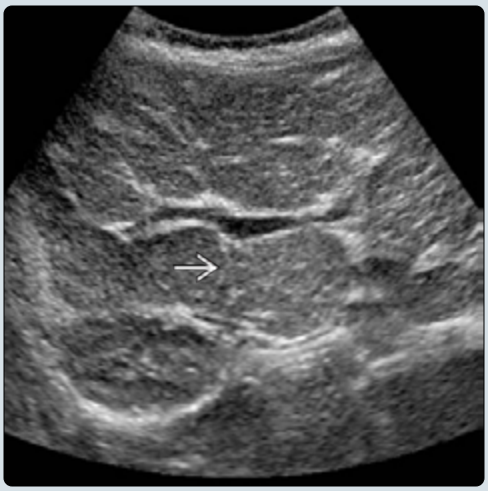
TOP DIFFERENTIAL DIAGNOSES

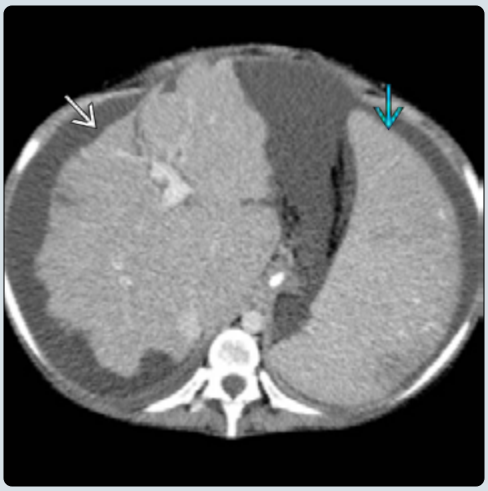
- Biliary atresia
- Choledochal cyst
- Alagille syndrome
- Progressive familial intrahepatic cholestasis
- Viral hepatitis
- α-1 antitrypsin deficiency
- Cystic fibrosis
- Wilson disease
- Nonalcoholic fatty liver disease
- Autoimmune hepatitis
- Primary sclerosing cholangitis
- Total parenteral nutrition cholestasis
- Post-Fontan physiology

CLINICAL ISSUES

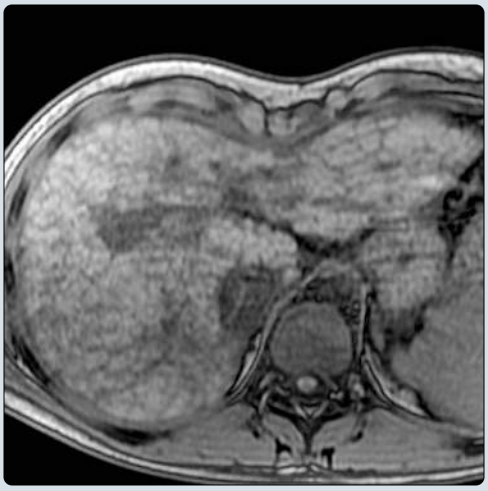
- Symptoms & treatment depend on underlying condition
- Early stages of fibrosis can be reversed by treating underlying condition
- Treatment for end-stage liver disease: Liver transplant
- Cirrhotic patients typically screened every 6 months for hepatocellular carcinoma

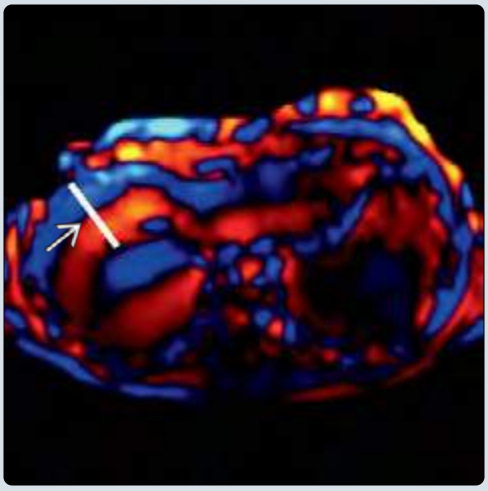
(Left) *Transverse ultrasound shows a markedly abnormal appearance of the liver with a macronodular contour & coarsened echotexture. Note the fine echogenic regions of fibrosis.* **(Right)** *Axial CECT shows a small, cirrhotic liver with a macronodular contour. There are signs of portal hypertension, including ascites & splenomegaly.*





(Left) *Axial T1 opposed-phase MR in an adolescent with autoimmune hepatitis shows an abnormal lace-like pattern of fibrosis throughout the liver.* **(Right)** *Axial MR elastogram of the liver shows abnormal increased thickness of the sound wave traversing the liver. The liver stiffness was measured at 4.92 kPa in this patient (with advanced fibrosis > 2.71 kPa).*





TERMINOLOGY

Definitions

- Cirrhosis: Liver disease characterized by bridging fibrosis

IMAGING

General Features

- Best diagnostic clue
  - Nodular contour of liver
  - Signs of portal hypertension: Ascites, splenomegaly, varices

CT Findings

- ↓ in liver size with progressive fibrosis
- Hepatic arterial phase of enhancement most useful for detecting hepatocellular carcinoma

MR Findings

- Fibrosis may appear as lace-like ↓ T1 signal
- Elastography can determine liver stiffness
- Hepatocyte-specific contrast agent: Characterizes nodules
  - Must look for development of hepatocellular carcinoma
- Detects other causes of fibrosis such as fat or iron deposition

Ultrasonographic Findings

- Liver has coarsened echotexture
- Nodular contour easier to see on free margin
- Doppler US currently best study to evaluate direction & amplitude of portal flow

Imaging Recommendations

- Best imaging tool
  - MR provides complete evaluation: Fat or iron deposition, liver stiffness, vascular supply, & tumor screening

CLINICAL ISSUES

Treatment

- Depends on underlying condition
- Early stages of fibrosis can be reversed by treating underlying condition
- Treatment for end-stage liver disease: Liver transplant

SELECTED REFERENCES

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Common Causes of Cirrhosis in Children

Disease	Key Facts
Biliary Obstruction	
Biliary atresia	Presents in 1st weeks of life; early diagnosis essential; differentiated from neonatal hepatitis by biopsy ± HIDA scan; ultrasound can be suggestive
Choledochal cyst	Types classified via Todani classification system
Intrahepatic Cholestasis	
Alagille syndrome	Bile ducts malformed & reduced in number; associated with tetralogy of Fallot, vertebral anomalies, & abnormal facies
Progressive familial intrahepatic cholestasis	Autosomal recessive disorder of intrahepatic cholestasis; presents in neonatal period
Genetic	
α-1 antitrypsin deficiency	Liver disease occurs in 10-15%; onset of lung disease usually between 20-50 years of age
Cystic fibrosis	Liver disease occurs in 25%; caused by plugging of bile ducts
Wilson disease	Caused by defect in copper transport; copper accumulates in liver, brain, & eyes
Other	
Nonalcoholic fatty liver disease	Associated with obesity & metabolic syndrome; can measure excess fat content in liver with MR
Hepatitis B	Most common cause of viral cirrhosis in children & adolescents
Autoimmune hepatitis	Hepatocellular inflammation of unknown cause; elevated levels of serum IgG
Primary sclerosing cholangitis	Associated with inflammatory bowel disease (particularly ulcerative colitis)
Total parenteral nutrition (TPN)	TPN-associated cholestasis can occur rapidly; more common in infants after bowel resection
Post-Fontan physiology	Caused by chronic passive venous congestion & volume overload

KEY FACTS

TERMINOLOGY

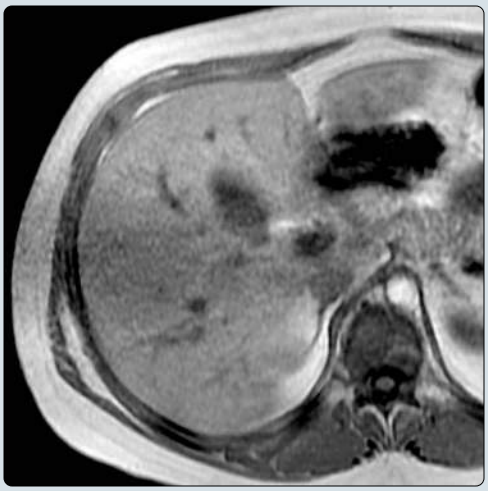
- Simple steatosis: > 5% of hepatocytes with fat infiltration
- Nonalcoholic fatty liver disease (NAFLD): Steatosis with metabolic syndrome &/or abnormal liver function enzymes
- Nonalcoholic steatohepatitis (NASH): Abnormal hepatic fat content with inflammatory activity or fibrosis

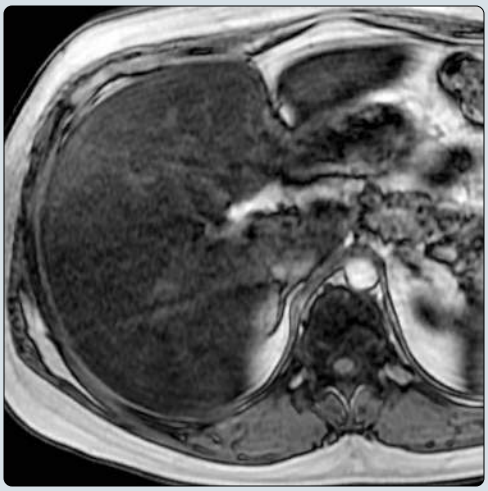
IMAGING

- NECT: ↓ liver attenuation relative to spleen
  - Normal liver 8-10 Hounsfield units (HU) > spleen
- CECT: Difficult to assess steatosis due to variable enhancement of liver & spleen depending on phase
  - Venous/delayed images: Liver HU ≥ 35 lower than spleen
- MR: Only imaging test that can quantify hepatic fat
  - Multiple methods to measure hepatic fat content
    - MR spectroscopy
    - Chemical shift imaging
      - Signal dropout on opposed-phase vs. in-phase T1
  - Steatosis defined as fat fraction > 5%
- Ultrasound shows diffusely ↑ hepatic echogenicity
  - Liver much more echogenic than right kidney
  - Poor through transmission of sound waves
    - ↓visualization of echogenic portal triads & right hemidiaphragm

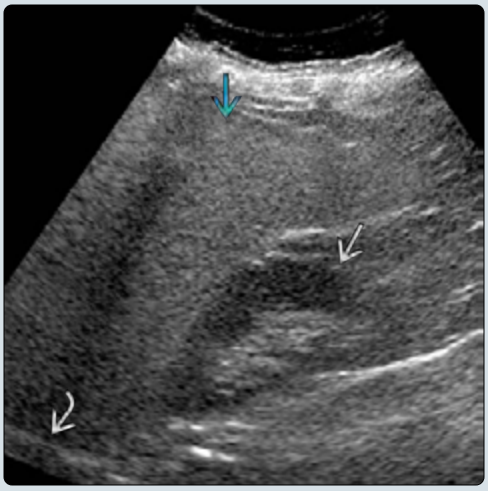
CLINICAL ISSUES

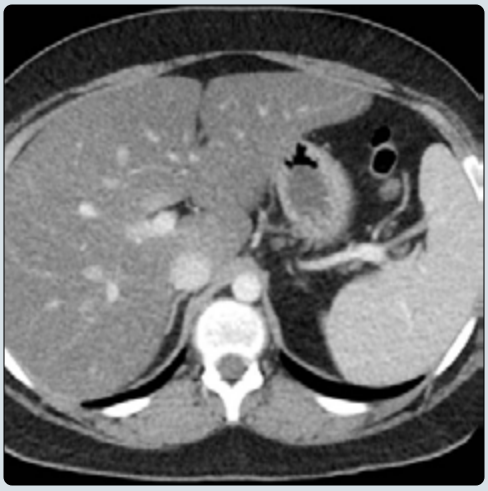
- Most common cause of chronic liver disease in children
  - NAFLD present in 38-40% of obese patients
  - Most commonly presents between 11-13 years of age
    - Obesity, abdominal pain, fatigue, hepatomegaly
- Risk factors
  - Modifiable: Obesity, sedentary lifestyle, high intake of sugar-sweetened beverages, sleep apnea
  - Nonmodifiable: Male, Hispanic origin, family history, parental obesity, low birth weight
- Thought to represent progression from simple steatosis → NAFLD → NASH → cirrhosis
- Treatment: Weight loss with dietary modification & exercise





(Left) *Axial T1 MR in-phase image of the liver in an adolescent with nonalcoholic steatohepatitis (NASH) shows a normal contour & signal intensity of the liver.* **(Right)** *Axial T1 MR opposed-phase image in the same patient shows considerable loss of signal (i.e., dropout) of the liver, typical of steatosis. In this case, the fat-fraction was measured at 30%. There are multiple methods to detect & quantify hepatic fat content via MR, including MR spectroscopy & chemical shift imaging.*





(Left) *Ultrasound of the liver in the same patient shows diffusely increased echogenicity of the liver as compared to the right kidney. In addition, there is poor through transmission of the sound beam with nonvisualization of the portal triads & poor visualization of the right hemidiaphragm.* **(Right)** *Axial CECT in the same patient shows the liver to have diffusely lower attenuation than the spleen. The liver measured 46 Hounsfield units (HU), while the spleen measured 125 HU, consistent with hepatic steatosis.*

TERMINOLOGY

Definitions

- Simple steatosis: Fatty infiltration of > 5% of hepatocytes
- Nonalcoholic fatty liver disease (NAFLD): Steatosis with metabolic syndrome &/or abnormal liver function tests
- Nonalcoholic steatohepatitis (NASH): Abnormal hepatic fat content with inflammatory activity or fibrosis

IMAGING

General Features

- Best diagnostic clue
  - Fatty infiltration of hepatic parenchyma, demonstrable by variety of modalities/techniques

CT Findings

- NECT
  - ↓ attenuation of liver compared to spleen
    - Attenuation inversely proportional to fat content
    - Normal liver ~ 8-10 Hounsfield units (HU) > spleen
- CECT
  - Difficult to assess due to variable appearances of liver & spleen depending on phase of enhancement
  - Venous or delayed-phase images can suggest steatosis if liver HU ≥ 35 lower than spleen

MR Findings

- Steatosis defined as MR fat fraction > 5%
- Multiple methods to measure hepatic fat content
- MR spectroscopy
  - Considered imaging gold standard for quantification
    - Measures protons in acyl groups of liver tissue triglycerides
    - Able to measure fat content across spectrum of severity & confounding diseases
      - Not susceptible to effects from fibrosis, iron, or glycogen
  - Rarely performed in most clinical practices
- Chemical shift imaging
  - Spin-echo (Dixon) technique
    - By altering TE, creates in-phase & opposed-phase images based on differing precession frequencies of protons in fat vs. water
      - Signal of fat & water protons additive for in-phase
      - Signal cancellation for opposed-phase (if both fat & water protons present in same voxel)
  - Modified Dixon technique: GRE images to ↓ scan time
  - Multipoint Dixon technique: Adds more echo series, allowing for better fat quantification
  - Proton density fat fractionation
    - Uses spoiled gradient-recalled echo images
- MR elastography
  - ↑ liver stiffness > 2.74 kPa associated with hepatic steatosis

Ultrasonographic Findings

- Diffusely ↑ hepatic echogenicity
  - Liver much more echogenic than right kidney
- Poor through transmission of sound waves
  - Poor visualization of echogenic portal triads
  - Poor visualization of echogenic right hemidiaphragm

Imaging Recommendations

- Best imaging tool
  - MR currently only modality that can quantify hepatic fat
  - Chemical shift imaging through Dixon technique available on most MR scanners

DIFFERENTIAL DIAGNOSIS

Cirrhosis

- End result of various chronic liver diseases → shrunken, nodular morphology with bridging fibrosis at histology

Cystic Fibrosis

- Steatosis occurs in 23-75% of patients, potentially due to malnutrition, oxidative stress, insulin resistance

PATHOLOGY

Staging, Grading, & Classification

- Histologic fat content measures % of cell containing fat
- MR fat fraction measures % of liver tissue made up of fat

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Obesity (NAFLD present in 38-40%)
  - Vague abdominal pain, irritability, fatigue
  - Hepatomegaly

Demographics

- Age
  - Most commonly presents between 11-13 years
  - Onset may be associated with puberty
- Gender
  - More common in males
- Ethnicity
  - Affects Hispanics > Caucasians > African Americans
- Epidemiology
  - Most common cause of chronic liver disease in children
  - Prevalence of 8-10% in general population
- Risk factors
  - Modifiable risk factors: Obesity, sedentary lifestyle, high intake of sugar-sweetened beverages, sleep apnea
  - Nonmodifiable risk factors: Male, Hispanic origin, family history, parental obesity, low birth weight

Natural History & Prognosis

- Thought to represent progression from simple steatosis → NAFLD → NASH → cirrhosis
  - Unknown why some patients progress & others do not
  - Progression to NAFLD or NASH does not guarantee further progression of disease

Treatment

- Weight loss with dietary modification & exercise

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Hepatic Venoocclusive Disease

KEY FACTS

TERMINOLOGY

- Therapy-induced occlusion of hepatic sinusoids → clinical triad of jaundice, ascites, & painful hepatomegaly
- Typically occurs after stem cell transplantation or certain tumor chemotherapy regimens

IMAGING

- Ultrasound with Doppler
  - Hepatomegaly ± heterogeneously ↑ echogenicity of liver parenchyma
  - Ascites, gallbladder wall thickening
  - Reversed or to & fro flow in portal veins
  - High-resistance hepatic arterial flow
- CECT/MR
  - Small-caliber hepatic veins
  - Ascites, heterogeneous liver parenchyma

TOP DIFFERENTIAL DIAGNOSES

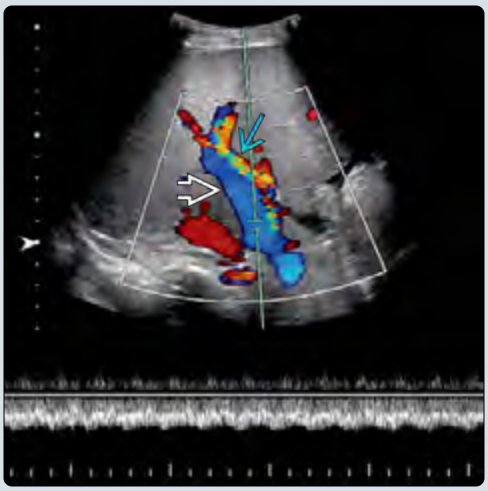
- Graft-vs.-host disease
- Infection

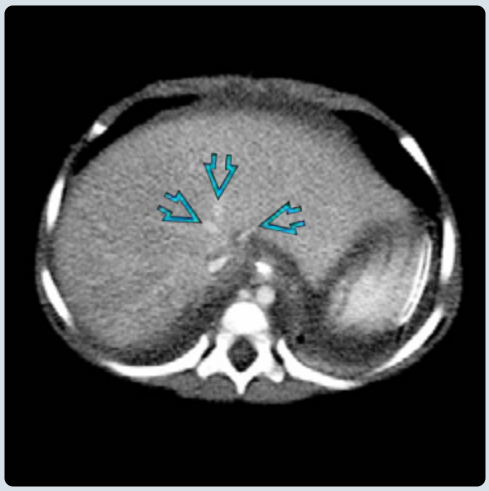
PATHOLOGY

- Toxic metabolite of chemotherapy &/or radiation therapy leads to small-vessel endothelial damage, venous occlusion, & postsinusoidal portal hypertension

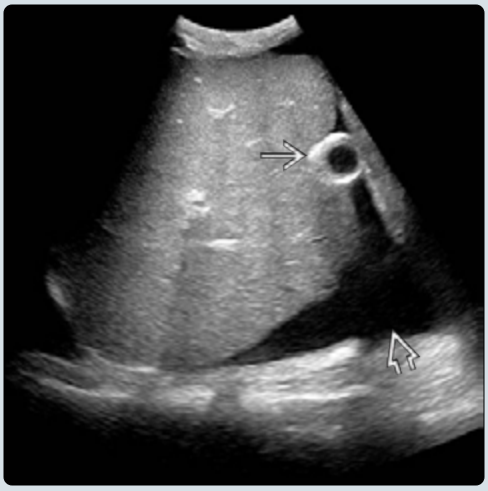
CLINICAL ISSUES

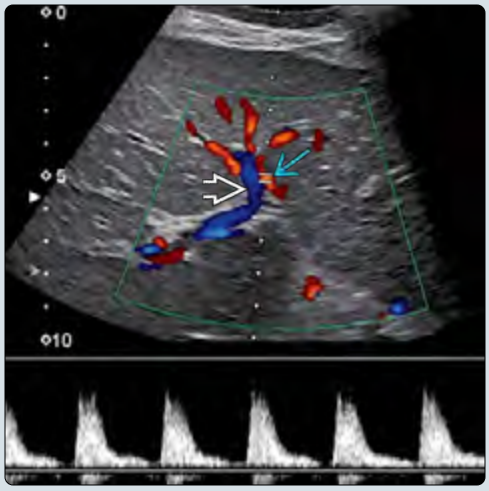
- Presents with jaundice, ascites, painful hepatomegaly
- Laboratory evaluation shows elevated bilirubin ± thrombocytopenia, evidence of hepatic synthetic dysfunction
- Graded clinically as mild, moderate, or severe
  - Mild: Resolves without specific treatment
  - Moderate: Progressive, but no multiorgan failure
  - Severe: Progressive with multiorgan failure & high mortality rate (> 80%)
- Treatment: Supportive care, withdrawal of hepatotoxic therapy, ± anticoagulation
  - Defibrotide: Experimental; promising in clinical trials





(Left) *Pulsed Doppler ultrasound in a 2-year-old girl receiving chemotherapy for neuroblastoma & now presenting with new onset of ascites & elevated bilirubin shows that flow in the main portal vein is reversed (hepatofugal flow) ⇨, typical of hepatic venoocclusive disease. Note the normal hepatopetal flow in the hepatic artery ⇨.* **(Right)** *Axial CECT in the same 2-year-old girl undergoing chemotherapy for neuroblastoma shows small caliber but patent hepatic veins ⇨, typical of hepatic venoocclusive disease.*





(Left) *Grayscale ultrasound in a 13-year-old girl on chemotherapy with newly diagnosed hepatic venoocclusive disease shows ascites ⇨ & marked thickening of the gallbladder wall ⇨. The portal venous flow was reversed (not shown).* **(Right)** *Transverse pulsed Doppler ultrasound in a 5-year-old patient status post stem cell transplantation shows reversed portal venous flow ⇨ with high-resistance waveforms in the adjacent hepatic artery ⇨, suggesting hepatic venoocclusive disease.*

TERMINOLOGY

Synonyms

- Sinusoidal obstruction syndrome

Definitions

- Therapy-induced occlusion of hepatic sinusoids, → clinical triad of jaundice, ascites, & painful hepatomegaly
- Onset within 30 days of stem cell transplantation (SCT)
  - Secondary to myeloablative chemotherapy &/or irradiation preceding SCT
- May also occur with specific tumor chemotherapy regimens

IMAGING

General Features

- Best diagnostic clue
  - Reversed portal vein flow, hepatomegaly, & ascites status post SCT or chemotherapy

Radiographic Findings

- Displaced bowel loops due to hepatomegaly & ascites

CT Findings

- CECT
  - Heterogeneous liver parenchyma
  - Small caliber hepatic veins
    - < 3 mm in diameter
  - Ascites, gallbladder wall thickening

Ultrasonographic Findings

- Grayscale ultrasound
  - Nonspecific hepatomegaly
  - Signs of fluid overload
    - Ascites, gallbladder wall thickening
  - ± splenomegaly
- Pulsed Doppler
  - Reversed or to & fro flow in portal vein
    - May only affect single segmental vein early (before clinical symptoms)
  - Elevated resistive indices in hepatic artery (> 0.75)
- Color Doppler
  - Reversed portal vein flow
  - Small, but patent, hepatic veins
  - Recanalized paraumbilical vein

Imaging Recommendations

- Best imaging tool
  - Ultrasound with Doppler
- Protocol advice
  - Imaging abnormalities may precede clinical manifestations
  - Baseline imaging prior to SCT should be considered

DIFFERENTIAL DIAGNOSIS

Graft-Vs.-Host Disease

- Affects skin, bowel, liver
- Featureless, ribbon-like bowel

Infection

- Viral pathogens, particularly cytomegalovirus
- No specific imaging findings

PATHOLOGY

General Features

- Etiology
  - Damage to small vessels usually due to toxic metabolite of intensive chemotherapy or radiation therapy
    - Leads to release of inflammatory factors & cytokines
    - Ultimately results in endothelial damage, small vein occlusion, & postsinusoidal portal hypertension

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Jaundice, ascites, painful hepatomegaly
- Other signs/symptoms
  - Weight gain due to fluid overload
  - Laboratory abnormalities
    - Elevated bilirubin
    - Thrombocytopenia
    - Abnormal protein C & other clotting factors

Demographics

- Age
  - Occurs in children & adults
- Epidemiology
  - Affects 8-14% of SCT patients; up to 60% of high risk

Natural History & Prognosis

- Graded clinically as mild, moderate, or severe
  - Mild: Resolves without specific treatment
  - Moderate: Progressive, but without multiorgan failure
  - Severe: Progressive disease with multiorgan failure
    - High mortality rate (> 80%)

Treatment

- Supportive care, withdrawal of hepatotoxic therapy, steroids, ± anticoagulation
- Defibrotide: Experimental medication, promising in trials

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Abernethy Malformation

KEY FACTS

TERMINOLOGY

- Congenital malformation characterized by diversion of portal venous blood flow around liver, most commonly into inferior vena cava (IVC)
  - Type 1: Absence of portal vein with extrahepatic portosystemic shunt
  - Type 2: Intact but hypoplastic portal vein with extrahepatic portosystemic shunt

IMAGING

- Absent or hypoplastic portal vein without findings of portal vein thrombosis or portal hypertension
- Extrahepatic portosystemic shunt, typically draining to IVC
- Small liver with multiple nodules

TOP DIFFERENTIAL DIAGNOSES

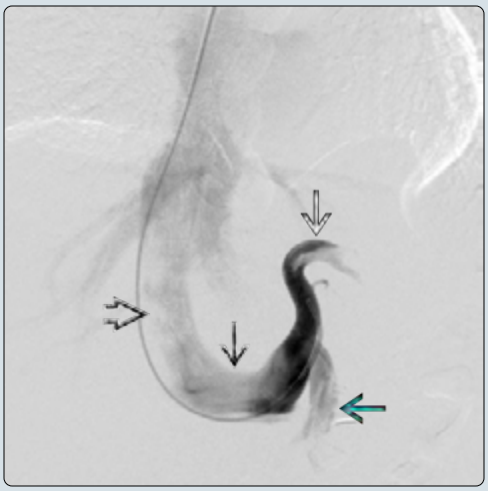
- Portal vein thrombosis
- Intrahepatic portosystemic shunt

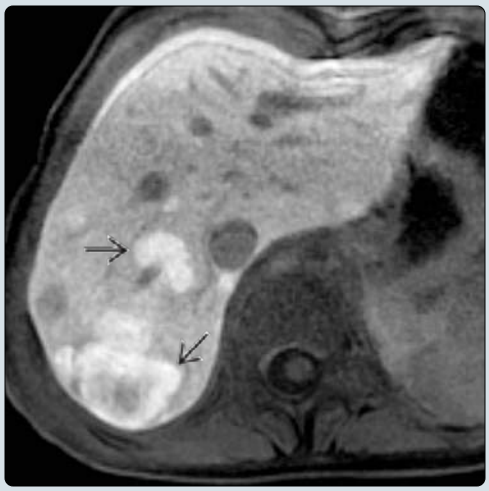
PATHOLOGY

- Hepatic lesions present in ~ 50% of patients
  - Most common: Focal nodular hyperplasia, nodular regenerative hyperplasia, hepatocellular adenoma
  - Rare reports of malignant degeneration to hepatoblastoma or hepatocellular carcinoma
- Congenital heart disease present in ~ 15% of patients

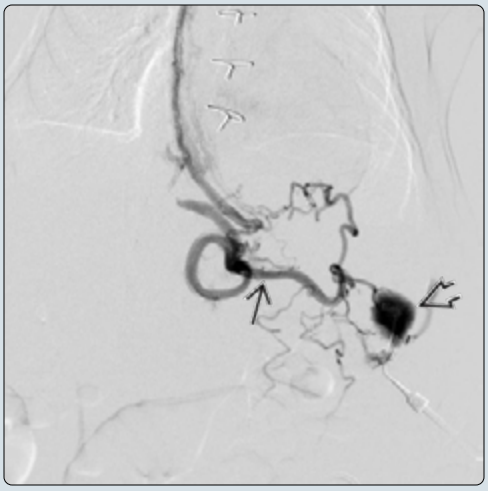
CLINICAL ISSUES

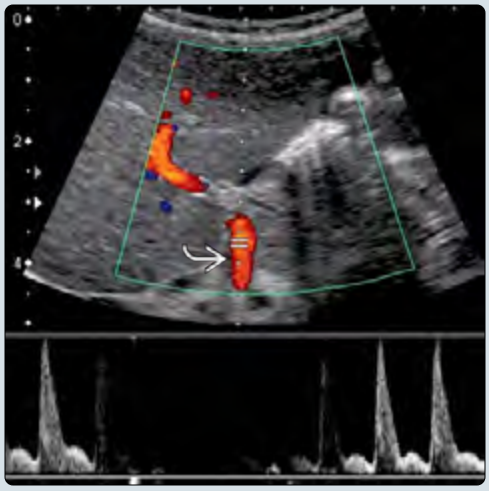
- 3:1 = F:M ratio in type 1 Abernethy malformation; no gender predilection in type 2
- Type 1 9x more common than type 2
  - ↑ detection may be due to more symptomatic disease
- May be diagnosed in utero or up to 7th decade of life
  - Type 1 malformations typically present at younger age with hepatic dysfunction, failure to thrive
  - Risks of hepatic encephalopathy & hepatopulmonary syndrome ↑ with age





(Left) *Digital subtraction angiography (DSA) in a patient with a type 1 Abernethy malformation shows contrast opacifying the splenic vein, superior mesenteric vein, & shunt vessel. The shunt connects directly to the inferior vena cava (IVC).* **(Right)** *Delayed axial T1 MR of the liver after injection of a hepatocyte-specific contrast agent in a patient with Abernethy malformation demonstrates multiple liver lesions. At biopsy, these lesions were shown to represent adenomas & hepatocellular carcinoma.*





(Left) *DSA during a direct splenic injection shows a tortuous splenic vein, numerous small collateral vessels, & no portal vein, typical of a type 1 Abernethy malformation.* **(Right)** *Pulsed Doppler ultrasound at the hepatic hilum shows absence of the portal vein with an enlarged hepatic artery. No hilar collateral veins are seen, typical of an Abernethy malformation rather than portal vein occlusion.*

TERMINOLOGY

Synonyms

- Congenital absence of portal vein, congenital extrahepatic portosystemic shunt

Definitions

- Congenital malformation characterized by diversion of portal venous blood flow around liver, more commonly into inferior vena cava (IVC) than renal, iliac, or azygous veins
  - Type 1: Absence of portal vein with extrahepatic portosystemic shunt
  - Type 2: Intact but hypoplastic portal vein with extrahepatic portosystemic shunt

IMAGING

General Features

- Best diagnostic clue
  - Absent or hypoplastic portal vein without findings of portal vein thrombosis or portal hypertension
    - Type 1: Absent portal vein with shunt vessel draining into IVC
    - Type 2: Hypoplastic portal vein with partial extrahepatic drainage into IVC
  - Liver findings: Small liver with multiple nodules

CT Findings

- CECT
  - Venous phase shows portosystemic shunt ± small portal vein
  - Coronal plane & maximum intensity projection images useful to evaluate shunt & portal system

MR Findings

- T1WI C+ FS
  - Hepatocyte-specific contrast agent helpful to delineate nodules & identify malignant degeneration
- MRV
  - Blood pool contrast agent may be useful to evaluate portosystemic shunt

Ultrasonographic Findings

- Grayscale ultrasound
  - Absent portal vein with ↑ periportal echogenicity (related to fibrosis) in expected region of portal vein
  - Decreased liver size for age
- Color Doppler
  - No portal venous flow
  - No hilar venous collaterals (to suggest portal vein occlusion)
  - Enlarged hepatic artery
  - Extrahepatic shunt; Doppler helps determine flow direction

Angiographic Findings

- Best technique to identify & measure portosystemic shunt
- Portal system may be imaged via
  - IVC access of shunt
  - Direct splenic injection
  - Direct percutaneous hepatic portography (type 2 only)
  - Indirect mesenteric portography (during mesenteric arteriography)

Imaging Recommendations

- Protocol advice
  - Maximum-intensity projection images useful to show small vessels or shunt vessels
  - MR blood pool contrast agent may be useful to image liver & vasculature in multiple planes

DIFFERENTIAL DIAGNOSIS

Portal Vein Thrombosis

- Filling defect with absence of portal venous flow (US) or enhancement (CECT or MR)
- Cavernous transformation with numerous small hilar venous collaterals
- Secondary findings of portal hypertension

Intrahepatic Portosystemic Shunt

- Abnormal connection between branches of portal vein & hepatic veins or IVC

PATHOLOGY

General Features

- Associated abnormalities
  - Hepatic lesions present in ~ 50% of patients
    - Most common: Focal nodular hyperplasia, nodular regenerative hyperplasia, adenoma
    - Rare reports of malignant degeneration to hepatoblastoma or hepatocellular carcinoma
  - Congenital heart disease in ~ 15% of patients

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Nonspecific liver dysfunction including failure to thrive
- Other signs/symptoms
  - Metabolic disturbances (↑ serum ammonia & galactose)

Demographics

- Age
  - May be diagnosed in utero or up to 7th decade of life
- Gender
  - 3:1 = F:M ratio in type 1 Abernethy malformation
  - No gender predilection in type 2
- Epidemiology
  - Type 1 9x more common than type 2
    - ↑ detection may be due to worse symptoms

Natural History & Prognosis

- Risks for hepatic encephalopathy & hepatopulmonary syndrome ↑ with age
- Liver lesions at risk for malignant transformation

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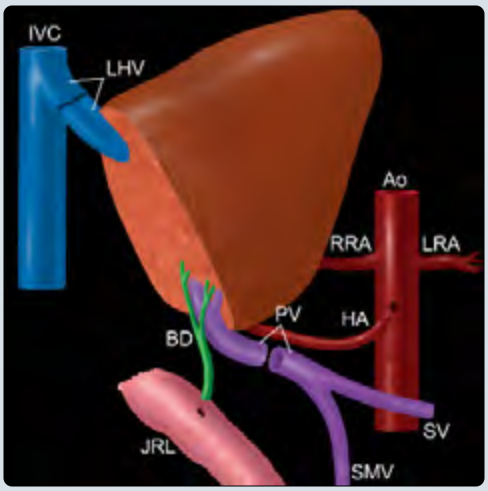
KEY FACTS

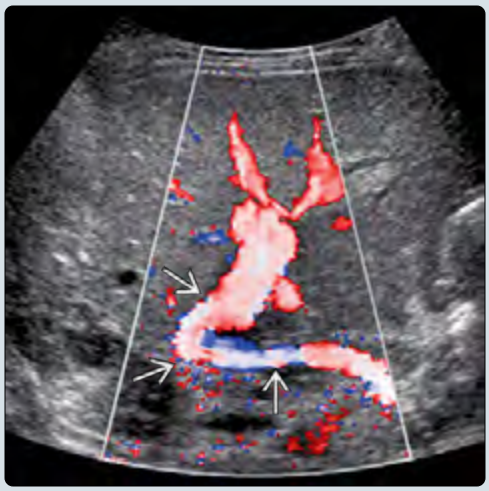
TERMINOLOGY

- Segmental liver transplant
  - In children, left lobe or lateral segment of left lobe of liver usually transplanted
  - Developed to ↑ supply of liver transplants for children
    - Usually adult donor, cadaveric or living
- Vascular complications
  - Hepatic arterial thrombosis (HAT) or stenosis
  - Hepatic artery pseudoaneurysm at infrarenal anastomosis with aorta
  - Portal vein stenosis or thrombosis
  - Hepatic vein stenosis
  - Anastomotic bleeding
- Biliary complications
  - Biliary stenosis or leak
- Extrahepatic fluid collection
  - Hematoma, seroma, bile leak, abscess
  - Usually found soon after surgery
- Posttransplant lymphoproliferative disorder (PTLD)
- Infection
- Organ rejection

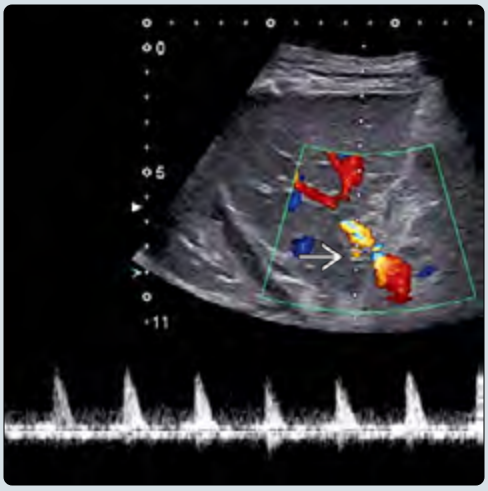
CLINICAL ISSUES

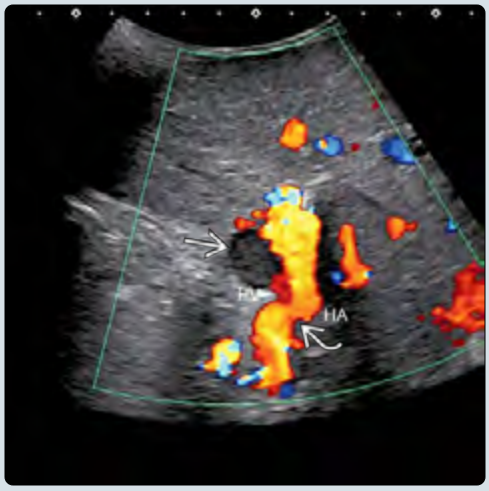
- Treatment options
  - Vascular complications
    - Vasodilator for arterial spasm early
    - Balloon dilation ± stent for narrowing
    - Thrombectomy for early HAT
    - Surgical revision
    - Retransplant
  - Biliary complications
    - Balloon dilation
    - ± placement of percutaneous biliary drain
    - Surgery
  - PTLD
    - Reduce immunosuppression
    - ± chemotherapy





(Left) *Graphic shows segmental left lobe liver transplant anatomy: Bile duct (BD), jejunal Roux loop (JRL), portal vein (PV), superior mesenteric vein (SMV), splenic vein (SV), hepatic artery (HA), aorta (Ao), right & left renal arteries (RRA & LRA), inferior vena cava (IVC), left hepatic vein (LHV).* **(Right)** *Transverse color Doppler US shows the typical curved course of the main PV ➔ & HA following segmental liver transplant. Note how the neoporta is located along 1 side of the liver rather than in the center.*





(Left) *Pulsed Doppler ultrasound of HA edema/vasospasm immediately after transplant shows a small-caliber HA ➔ with a high resistive index (RI). Elevated RIs can be seen with downstream arterial narrowing/thrombus or parenchymal edema. Sampling distal to a focus of narrowing may give a parvus et tardus waveform (not shown).* **(Right)** *Transverse color Doppler US image shows acute PV thrombosis with hypoechoic clot ➔ in the main PV & robust HA flow ➔ next to the clot.*

TERMINOLOGY

Synonyms

- Segmental liver transplant = partial liver transplant, split liver transplant, reduced-size liver transplant

Definitions

- Segmental liver transplant
  - In children, left hepatic lobe or lateral segment of left lobe usually transplanted
  - Developed to ↑ supply of liver transplants for children
  - Usually adult donor, cadaveric or living
  - Combined with small bowl transplant in select cases
  - Combined with kidney transplant in select cases
- Vascular complications
  - Hepatic artery (HA) vasospasm (immediately postop)
  - Hepatic arterial thrombosis (HAT) or stenosis
  - HA pseudoaneurysm at infrarenal anastomosis with aorta
  - Portal vein stenosis (PVS) & thrombosis
  - Hepatic vein stenosis (HVS)
  - Anastomotic bleeding
- Biliary complications
  - Anastomotic: Biliary stenosis or leak
  - Nonanastomotic
    - Intrahepatic biliary stenosis → dilation
    - Biloma
    - Intraductal sludge or stone
- Extrahepatic fluid collection
  - Hematoma, seroma, bile leak, abscess
  - Usually found soon after surgery
- Posttransplant lymphoproliferative disease (PTLD)
- Infection
  - Abscess from hematologic spread
  - Cholangitis via ascending route
- Organ rejection

IMAGING

Ultrasonographic Findings

- Grayscale ultrasound
  - HAT: Echogenic clot within artery
  - HA pseudoaneurysm: Round anechoic structure near HA anastomosis with aorta
  - PVS: Narrowing, usually at anastomosis, poststenotic dilation
  - PV thrombosis: Echogenic clot in PV lumen
  - HVS: Distended HVs
  - Biliary complications: Biliary dilation or collection
  - PTLD: Adenopathy, mass(es) within abdomen
- Color Doppler
  - HA spasm/edema/stenosis: Hours to days after transplant
    - Tardus parvus waveform distal to narrowing
    - Elevated peak systolic velocity in narrowing
    - High resistive index (RI) proximal to narrowing
      - Graft edema may also cause ↑ RI
  - HAT: Absent or reversed flow in HA
    - ↑ RI proximal to clot
    - Distal tardus parvus waveform (if any distal flow)
    - Abundant portal venous flow
  - HA pseudoaneurysm: Color flow filling dilated HA lumen
  - PVS: Focal narrowing
  - PV thrombosis: Absent flow or reversed flow
    - May see abundant HA flow in response
  - HVS: Dampened atrial pulsations
    - May normally be dampened early from edema
  - Arteriovenous fistula: Turbulent flow with spectral broadening, low RI arteries, arterialization of veins
    - Usually longer term after biopsy

CT Findings

- CECT
  - HAT or stenosis
    - Peripheral wedge-shaped low-attenuation regions
    - Unopacified HA
    - May lead to biliary necrosis with biloma or biliary dilation due to strictures
  - HA pseudoaneurysm at anastomosis to infrarenal aorta
  - PVS & thrombosis
    - Stenosis: Focal narrowing, ± poststenotic dilation
      - Note that adult donor PV often larger
    - Thrombosis: Unopacified PV, portosystemic shunts, splenomegaly, ascites
      - May be residua of pretransplant portal hypertension
  - HVS
    - Distended HVs
    - Congested liver with delayed enhancement
  - Biliary strictures from necrosis → dilation
  - Extrahepatic fluid collection
    - Hematoma, bile, abscess, or seroma
    - Aspiration required for determination of contents
  - PTLD: Wide range of appearances, may be adenopathy or masses almost anywhere in body
  - Infection: Fluid collections, solid organ lesions, dilated bile ducts (cholangitis)
  - Organ rejection: Nonspecific, biopsy required
- CTA
  - Similar to CECT but better detail of vascular structures

MR Findings

- T1WI
  - Hepatocyte-specific contrast agent can confirm biliary leak on delayed images
- MRA
  - HAT: Signal loss beyond thrombus due to absent flow
  - HA pseudoaneurysm: High signal focal enlargement of artery near anastomosis with aorta
- MRCP
  - Biliary dilation: High signal dilated bile ducts

Angiographic Findings

- DSA
  - HAT: Cut-off of HA
  - HA stenosis: Focal narrowing of artery, commonly at anastomosis
  - HA pseudoaneurysm: Focal dilation of HA, usually near aorta
  - HVS: Narrowing at HV anastomosis with atrium

Nonvascular Interventions

- Transhepatic cholangiography
  - Biliary strictures: Anastomotic or nonanastomotic → biliary dilation proximal to stricture
  - Biliary sludge/stones: Intraluminal filling defects → proximal biliary dilation

Imaging Recommendations

- Best imaging tool
  - Vasculature & biliary system
    - 1st-line screening: Ultrasound with Doppler
    - If possible arterial abnormality, may consider CTA
    - HAT: Angiogram with intervention
    - PVS/thrombosis: Percutaneous transhepatic portogram
    - HVS: Venogram
    - Biliary stenosis: Percutaneous transhepatic cholangiogram (PTC)
  - Biloma or fluid collection: MR vs. CECT
    - MR with hepatobiliary contrast agent can confirm biliary leak on delayed images
  - PTLD or infection: CECT, MR, or PET

PATHOLOGY

General Features

- Etiology
  - Biliary complications: Depends on surgical reconstruction technique, length of cold ischemia time, immunological reactions, HAT, CMV infection, ABO blood group incompatibility
  - Biliary strictures: ABO incompatibility, HAT (ischemia to bile ducts)
  - PTLD: Epstein-Barr virus (EBV) related in 90%, usually EBV(-) recipient who develops EBV infection after transplant

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depends on complication
    - HAT: Elevated liver function tests (LFTs)
    - PVS: GI bleeding from portosystemic collaterals, splenomegaly, ascites
    - Biliary strictures: Jaundice, cholangitis
    - PTLD: Nonspecific & variable, EBV seroconversion
    - Infection: Fever

Demographics

- Epidemiology
  - HAT
    - Leading cause of graft loss/retransplant (4-26%)
    - Interposition grafts or anastomosis with recipient aorta may ↓ HAT incidence when arteries < 3 mm
    - ↑ risk in pediatrics due to small arteries
    - Children do better than adults because of higher rate of recruitment of collateral arteries
  - PV thrombosis
    - High risk in segmental liver transplant because of short PV segment from donor
    - Higher risk in patients with ↓ PV flow because of splenectomy, portosystemic collaterals
    - Usually develops slowly after liver transplant → signs of portal venous hypertension
  - HVS/thrombosis or inferior vena cava (IVC) obstruction
    - Most often at anastomosis with IVC/atrium
    - Can be due to twisting when transplant moves/shifts
  - Anastomotic bleeding
    - Occurs early after transplant & usually requires surgical repair
  - Biliary complications
    - Most frequent liver transplantation complication
    - Higher incidence in segmental liver transplants
    - May lead to chronic graft failure, biliary cirrhosis
  - PTLD: More common in children than adults due to higher rate of EBV(-) before transplantation
    - 1-20% incidence
  - Infection: Leading cause of death (43%)

Treatment

- Vascular complications
  - HA vasospasm: Vasodilators
  - HA stenosis: Balloon dilation ± stent
    - Surgical repair if balloon resistant
  - HAT: Thrombectomy if early
    - May require retransplantation
  - PVS: Percutaneous transhepatic portogram → balloon dilation ± stent; surgical revision if necessary
  - HVS: Balloon dilation ± stent, or surgery
- Biliary complications
  - Anastomotic biliary stricture
    - Balloon dilation ± placement of percutaneous biliary drain or surgery
    - Stents have variable long-term patency
  - Nonanastomotic biliary stricture
    - Percutaneous biliary drain & balloon dilation
  - Biliary obstruction
    - 20% have no biliary dilation showing obstruction with PTC
    - Thus, if patient jaundiced with ↑ LFTs → PTC & possible percutaneous drain
  - Biloma: Drain if infected
  - Biliary leak: Surgery if large; heals with drain if small
  - Biliary sludge/stones: Remove percutaneously ± drain
- PTLD: Reduce immunosuppression ± chemotherapy
- Infection: Antibiotics, antifungals, ± drainage

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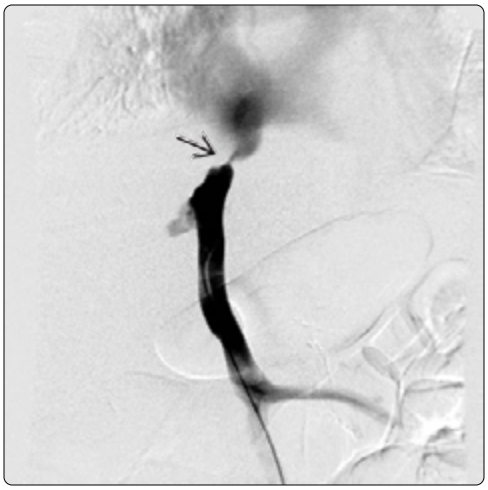
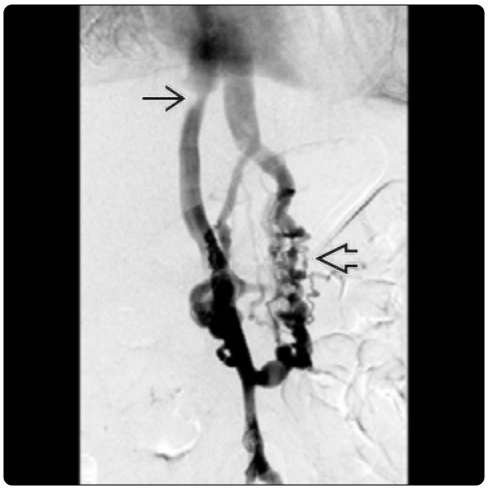
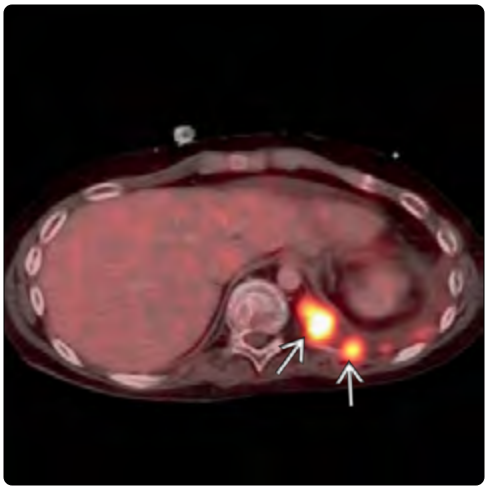
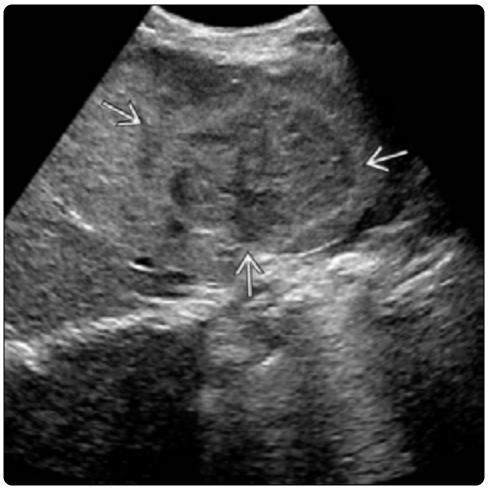


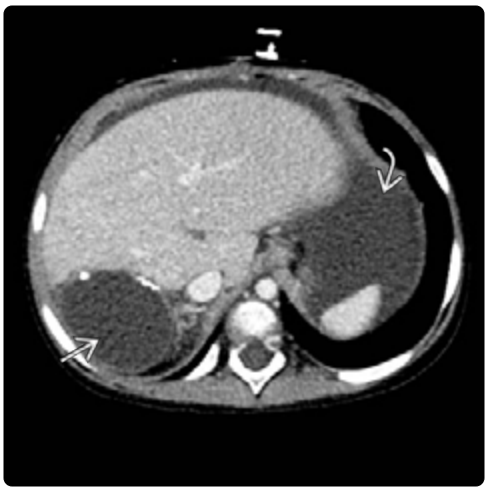


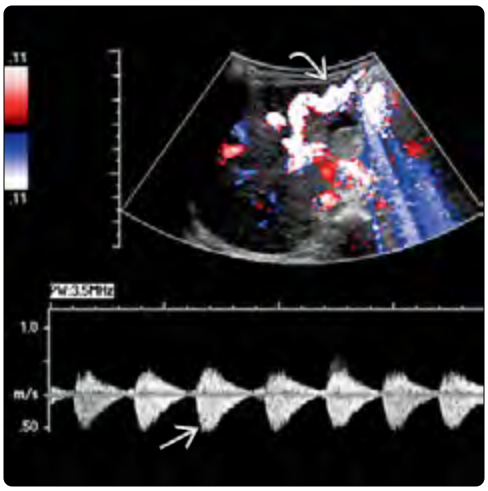
(Left) *Frontal DSA of the IVC shows a jet of contrast ⇨ due to stenosis at the anastomosis between the intrahepatic cava & the right atrium. This patient had declining hepatic function & recurrent GI bleeding due to varices.* **(Right)** *Frontal DSA in the same patient following balloon venoplasty of the stricture shows improved flow into the right atrium & with less narrowing at the IVC-right atrial anastomosis ⇨. Note the prominent varices in a left paraspinal location ⇨.*





(Left) *Axial FDG PET/CT image through the lung base shows multiple FDG avid soft tissue lesions along the pleura ➡ in this case of PTLD. Adenopathy & masses from PTLD can occur anywhere in the body.* **(Right)** *Transverse US shows a heterogeneous mass ➡ with a well-defined wall in the left aspect of the liver transplant in a patient with fever. Mild internal swirling of debris was noted real time. Peripheral but no internal vascularity was seen on color Doppler US (not shown). This abscess was drained percutaneously.*





(Left) *Axial CECT through a liver transplant shows a rounded low-density collection ➡ along the staple line in addition to abundant ascites ➡ in this case of bile leak/biloma.* **(Right)** *Transverse pulsed Doppler US shows turbulent flow (by both color ➡ & pulsed Doppler waveform ➡) in a tortuous vessel extending to the anterior capsule, consistent with an arteriovenous fistula, likely secondary to a prior percutaneous liver biopsy.*

Wandering Spleen

KEY FACTS

TERMINOLOGY

- Increased mobility of spleen due to abnormal ligamentous attachments (congenitally absent vs. increased laxity)
  - Long vascular pedicle predisposes to acute or chronic intermittent torsion ± infarction

IMAGING

- Absence of normal spleen in LUQ
- Enlarged spleniform mass in mid-lower abdomen
- With torsion: Twisted hilar vessels, fat edema, & ascites
  - CT: Heterogeneous vs. globally decreased splenic enhancement; ± capsular/rim enhancement
  - US: Hypovascular, heterogeneous splenic parenchyma

TOP DIFFERENTIAL DIAGNOSES

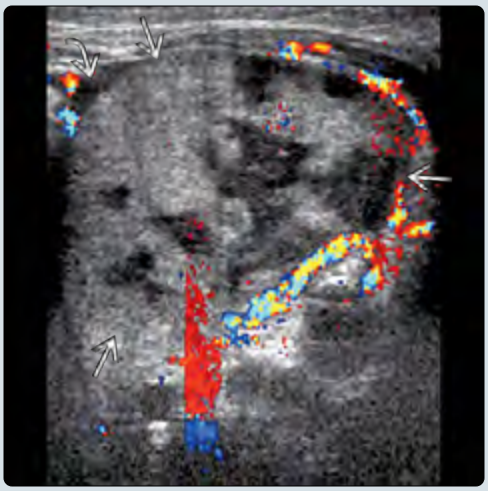
- Sickle cell disease
- Heterotaxy syndromes
- Lymphoma
- Splenic trauma
- Mononucleosis

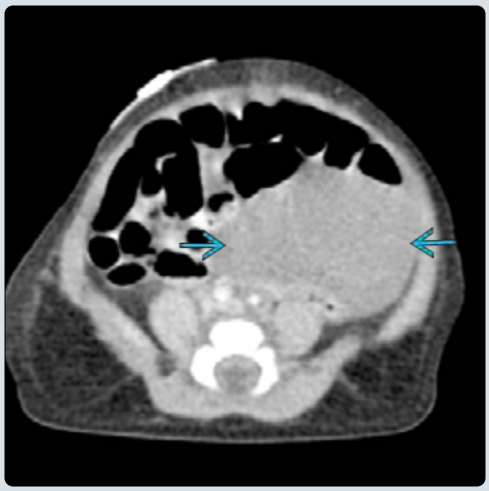
PATHOLOGY

- Due to abnormal splenic fixation by gastrosplenic, splenophrenic, splenorenal, splenocolic ligaments
  - Congenital absence of ligaments (affects young children)
  - Acquired laxity (affects women of reproductive age)
- Associated abnormalities include gastric volvulus, diaphragmatic hernia, prune-belly syndrome

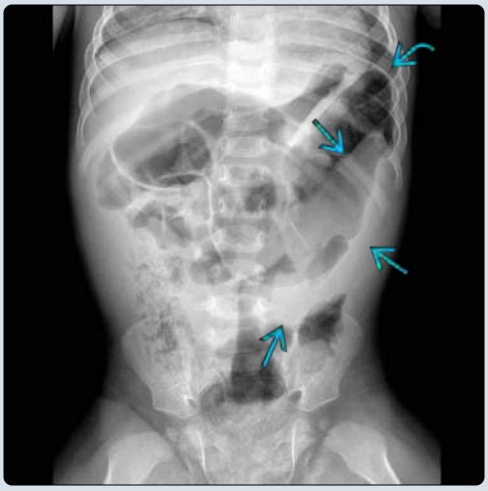
CLINICAL ISSUES

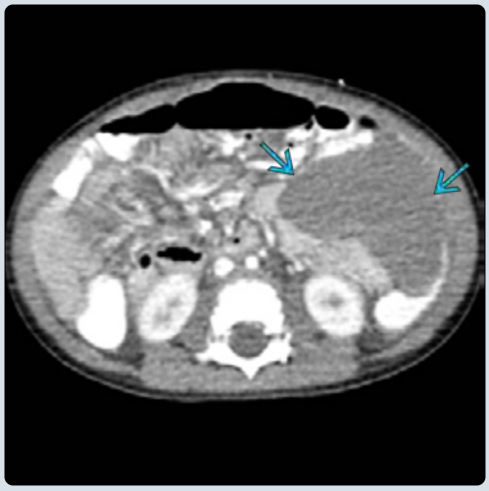
- Presentation
  - Pain, vomiting, palpable mobile abdominal mass
  - Asymptomatic (15%)
  - Rarely: Bowel obstruction, pancreatitis, bleeding varices
- Treatment
  - Without infarction: Detorsion + splenopexy
  - With infarction: Splenectomy + vaccinations, antibiotic prophylaxis for asplenia





(Left) *Transverse color Doppler ultrasound in a newborn with a palpable abdominal mass shows a spleniform mass in the mid to lower left abdomen with heterogeneous internal echotexture. No internal vascular flow is seen in the mass. Mild ascites is noted. A normal spleen was not found in the LUQ.* **(Right)** *Axial CECT of the same patient shows no significant enhancement of the spleniform left abdominal mass. A torsed & infarcted wandering spleen was surgically removed.*





(Left) *AP supine radiograph in an 18 month old with fussiness & abdominal distention shows absence of a normal splenic shadow in the abdominal LUQ. There is displacement of bowel gas by an enlarged spleniform mass in the mid left abdomen.* **(Right)** *Axial CECT in the same patient shows no enhancement of a displaced & abnormally rotated, enlarged spleen, consistent with torsion & ischemia/infarction. The spleen was freely mobile & entangled in omentum at surgery.*

TERMINOLOGY

Synonyms

- Ectopic spleen, splenoptosis

Definitions

- Increased mobility of spleen due to abnormal ligamentous attachments (absence vs. increased laxity)
  - Long vascular pedicle predisposes to acute or chronic intermittent torsion ± infarction

IMAGING

General Features

- Best diagnostic clue
  - Hypoenhancing/hypovascular spleniform abdominal mass in mid to lower abdomen
  - Absence of normal spleen in high LUQ/hypochondrium

Radiographic Findings

- Splenic shadow may be enlarged or absent from LUQ
- ± midlower abdominal mass displacing bowel

CT Findings

- Absence of splenic tissue in normal high posterior LUQ
- Displaced, enlarged spleniform mass
- Elongated vascular pedicle
  - Chronically enlarged splenic vein ± adjacent varices
- With torsion
  - Whirl or corkscrew sign of twisted vascular pedicle
    - Swirling lucent & dense bands at hilum due to vessels surrounded by thickened folds of peritoneum + fat
  - Heterogeneous enhancement of splenic parenchyma from congestion or infarcts vs. global nonenhancement
    - May have preserved capsular/rim enhancement
  - Ascites + perisplenic fat edema

Ultrasonographic Findings

- Absence of splenic tissue in normal posterior LUQ
- Displaced spleniform mass with variable echogenicity
  - Heterogeneous from congestion or infarcts
- Internal vascular flow variable
  - Lack of flow confirms torsion + ischemia
- Vascular pedicle
  - Chronically dilated, tortuous vessels
  - Twisting, narrowing ± thrombosis in torsion

Imaging Recommendations

- Best imaging tool
  - Ultrasound excellent screening modality in children with abdominal pain &/or mass
    - Altering patient position (to right decubitus) during scan can demonstrate mobility of wandering spleen
  - CT historically more accurate in setting of torsion

DIFFERENTIAL DIAGNOSIS

Sickle Cell Disease

- Splenic infarction
  - Heterogeneous spleen acutely
  - Calcified small spleen chronically
- Splenic sequestration
  - Acutely enlarged, heterogeneous spleen in young patients due to blood trapping by vasoocclusion

Heterotaxy Syndromes

- Spleen may be absent (asplenia), malpositioned, or of unusual configuration (polysplenia)

Lymphoma

- Enlarged spleen with round hypoenhancing masses

Splenic Trauma

- Broad imaging spectrum from isolated laceration to shattered, devascularized spleen

Mononucleosis

- Nonspecific splenomegaly + adenopathy
- Fever, pharyngitis, fatigue

PATHOLOGY

General Features

- Etiology
  - Abnormal splenic fixation by gastrosplenic, splenophrenic, splenorenal, splenocolic ligaments
    - Congenital absence (affects young children)
    - Acquired laxity (affects women of reproductive age)
- Associated abnormalities include gastric volvulus, diaphragmatic hernia, prune-belly syndrome

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Without torsion: Palpable mobile mass or asymptomatic
  - With torsion: Abdominal pain, palpable mass
- Other signs/symptoms
  - Fever, vomiting ± bowel obstruction, pancreatitis (due to pancreatic tail involvement), bleeding varices

Demographics

- Gender
  - M > F in young children; F > M in reproductive age

Natural History & Prognosis

- Acute torsion (up to 65%): Splenic infarction, peritonitis; potentially fatal
- Chronic complications
  - Intermittent torsion: Recurrent pain, splenomegaly (rupture risk from trauma), varices
  - Functional or iatrogenic asplenia: Infections, sepsis

Treatment

- Without infarction: Detorsion + splenopexy
- With infarction: Splenectomy + vaccinations, antibiotic prophylaxis for asplenia

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Splenic Infarct

KEY FACTS

IMAGING

- CECT/MR
  - Early: Ill-defined, mottled area(s) of ↓ enhancement relative to normal spleen
  - Late: Wedge-shaped, peripheral area(s) of ↓ enhancement
  - Diffuse infarction: Global nonenhancement ± preserved cortical rim of enhancement from capsular arteries
  - ± perisplenic fluid & inflammatory changes
- Ultrasound with Doppler
  - Peripheral, wedge-shaped hypoechoic foci ± internal parallel echogenic lines (bright band sign)
  - Absent flow in infarcted parenchyma

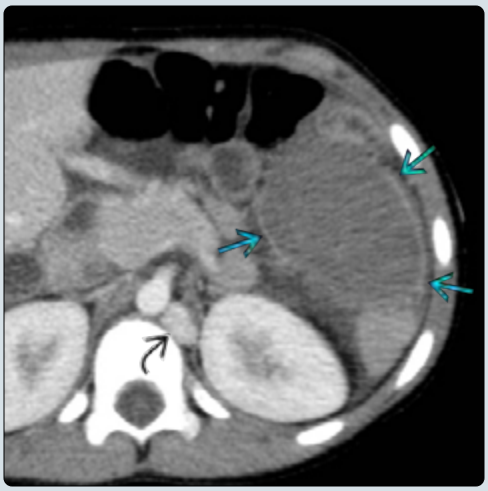
PATHOLOGY

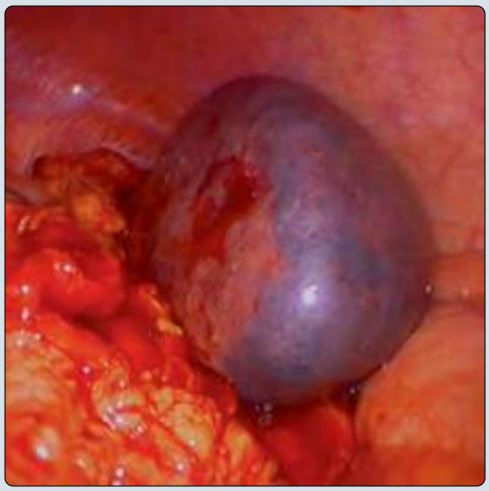
- Segmental vs. diffuse infarction
  - Segmental due to end-artery configuration of splenic arteries with ↓ collateral circulation
  - Diffuse due to twisting or occlusion of main splenic vessels
- Etiologies include
  - Hematologic: Sickle cell disease, myeloproliferative disorders, leukemia/lymphoma, hypercoagulable conditions
  - Embolic: Atrial fibrillation, endocarditis
  - Torsion: Wandering spleen, polysplenia
  - Vasculitis
  - Splenomegaly: Portal hypertension, infection, Gaucher disease

CLINICAL ISSUES

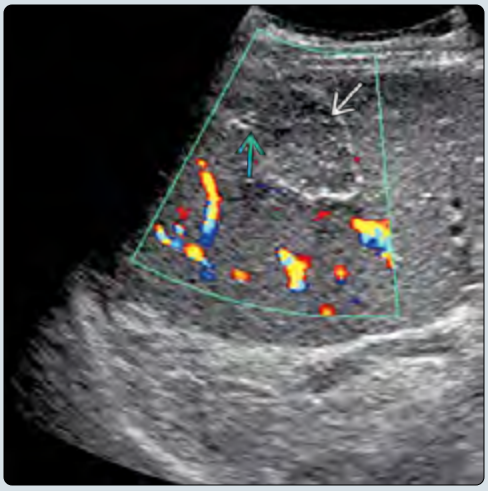
- Presents with left upper quadrant pain ± fever
- May resolve with focus of scar ± Ca^{2+} or cyst
- Complications include rupture, hemorrhage, & abscess formation
- Typically managed conservatively unless complications develop

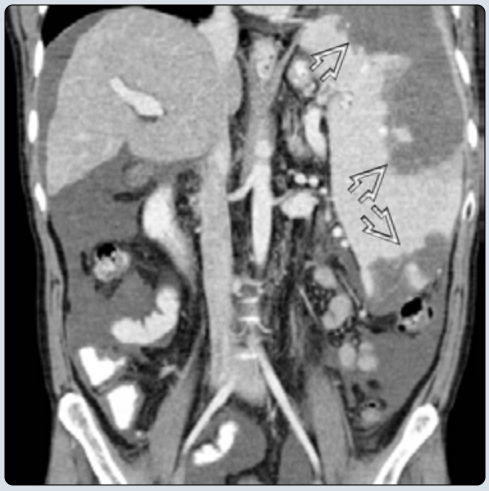
(Left) *Axial CECT in a 5-year-old girl with situs ambiguous & polysplenia who presented with abdominal pain shows a nonenhancing spleen with a rim sign of enhancement ⇨, typical of a splenic infarction. Note the left-sided inferior vena cava ⇨.* **(Right)** *Intraoperative photograph in the same 5-year-old girl shows an engorged, infarcted spleen.*





(Left) *Longitudinal oblique color Doppler ultrasound in a 17-year-old bone marrow transplant patient shows a well-demarcated, wedge-shaped, peripheral focus of avascular decreased echogenicity ➡ within the spleen, consistent with infarct. Note the echogenic parallel lines (bright band sign) ⇨ in this infarct.* **(Right)** *Coronal CECT in the same patient shows multifocal, well-demarcated peripheral areas of hypoattenuation ➡ in the enlarged spleen, consistent with infarcts.*





TERMINOLOGY

Definitions

- Segmental or diffuse ischemia &/or necrosis caused by vascular occlusion

IMAGING

General Features

- Best diagnostic clue
  - Peripheral, wedge-shaped, hypoenhancing foci on CECT/MR
  - Heterogeneous, hypoechoic, peripheral avascular foci on ultrasound with Doppler
    - ± bright band sign: Thin linear echogenic foci within hypoechoic infarct
- Location
  - Peripheral, segmental or diffuse type
- Size
  - Variable, can involve entire spleen
- Morphology
  - Typically wedge-shaped
    - Can be spherical/round or "peripheral band"
  - Well-demarcated nearly straight borders

CT Findings

- Variable appearance depending on stage
  - Early: Ill-defined, mottled area(s) of ↓ enhancement relative to normal spleen
  - Late: Wedge-shaped, peripheral area(s) of ↓ enhancement
  - Diffuse infarction: Global nonenhancement ± rim of cortical enhancement from capsular arteries
- ± associated vascular filling defects
- ± perisplenic fluid & inflammatory change
- ± splenomegaly

MR Findings

- Wedge-shaped, peripheral focus of nonenhancement
  - ↓ T1, ↑ T2 signal due to edema
- ↑ poorly defined edema/fluid in surrounding tissues

Ultrasonographic Findings

- Grayscale ultrasound
  - Peripheral, well-demarcated region of ↓ echogenicity
    - May see small linear parallel echogenic foci: Bright band sign
  - ± splenomegaly
- Color Doppler
  - Absent flow in infarcted areas

DIFFERENTIAL DIAGNOSIS

Laceration

- Irregular linear foci of hypoenhancement + surrounding fluid in trauma setting

Transient Heterogeneous Enhancement Pattern

- Corrugated, archiform alternating bands of ↑ & ↓ enhancement during arterial phase postcontrast CT/MR

Lymphoma

- Focal masses throughout spleen ± splenomegaly, adjacent adenopathy

Preserved Splenic Tissue in Chronic Infarction

- Round hypoechoic mass(es) of residual functioning splenic tissue in otherwise echogenic chronically infarcted spleen
- Normal internal vascularity on ultrasound
- + uptake on nuclear medicine sulfur colloid scan
- Associated with sickle cell disease

PATHOLOGY

General Features

- Etiology
  - Hematologic: Sickle cell disease, myeloproliferative disorders, leukemia/lymphoma, hypercoagulable conditions
  - Embolic: Atrial fibrillation, endocarditis
  - Torsion: Wandering spleen, polysplenia
  - Vasculitis
  - Splenomegaly: Portal hypertension, infection, Gaucher disease

Staging, Grading, & Classification

- Segmental infarction
  - Occurs due to end-artery configuration of splenic arteries with ↓ collateral circulation
- Diffuse infarction
  - Due to occlusion or twisting of main splenic vessels

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Left upper quadrant pain
  - ± fever & constitutional symptoms

Natural History & Prognosis

- Variable; often requires no treatment
- May resolve completely leaving focus of scar ± Ca^{2+}
- May resolve & leave acquired cyst
- Complications: Rupture, abscess, intraperitoneal hemorrhage
- Functional asplenia may result from extensive infarctions
  - Predisposed to overwhelming post splenectomy infection with encapsulated organisms
  - Vaccination for *Streptococcus pneumoniae*, *Haemophilus influenzae*, meningococcus; ± antibiotic prophylaxis

Treatment

- Typically no treatment unless complications develop

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Splenic Cysts

KEY FACTS

IMAGING

- Best clue: Well-circumscribed, round splenic mass
  - Follows fluid characteristics on all modalities
  - "Claw" of splayed splenic tissue along margin of larger cysts (confirming spleen as organ of origin)
- Typically solitary & unilocular
- Can occasionally have thin rim of Ca^{2+}
- US: Typically suffices to demonstrate cystic nature
  - Anechoic/hypoechoic with ↑ through transmission
  - Can have low-level internal echoes
  - No internal flow on color/power Doppler US
- No enhancement on CECT or MR C+
  - Typically hypointense on precontrast T1 MR unless proteinaceous or hemorrhagic contents

TOP DIFFERENTIAL DIAGNOSES

- Splenic lymphoma
- Splenic infection
- Splenic infarct
- Splenic perfusion artifact
- Intrasplenic pancreatic pseudocyst

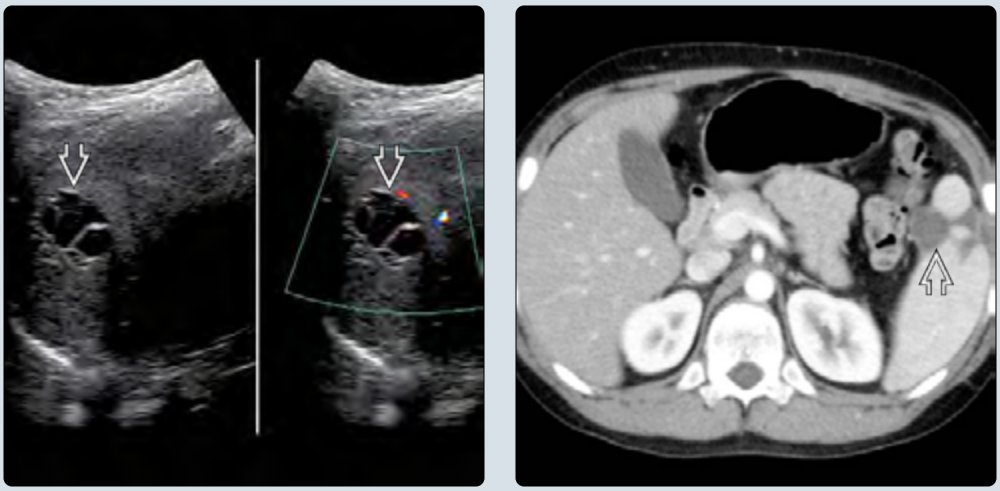
PATHOLOGY

- Congenital (true) cysts either epidermoid or mesothelial
  - ± elevation of serum CEA & CA 19-9
- Acquired cysts (comprise 80% of splenic cysts)
  - Posttraumatic
  - Postinfarction
  - Infectious/inflammatory (pyogenic, parasitic, fungal)
  - Vascular malformation (venolymphatic)
    - May be congenital but not visible early

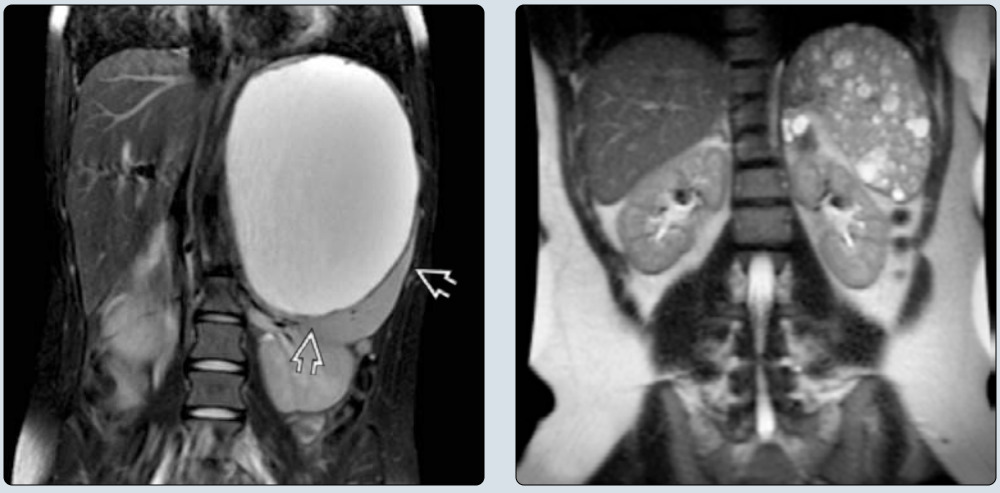
CLINICAL ISSUES

- Most commonly incidental
- Complications may include hemorrhage, rupture, infection
- Distinction between congenital (true) & acquired cysts not possible by imaging

(Left) *Longitudinal grayscale & color Doppler US images show an incidentally detected, mildly complex splenic cyst with septations in a 10-year-old girl. Pathology confirmed a benign epidermoid cyst.* **(Right)** *Axial CECT in a teenager who was involved in a motor vehicle collision several weeks prior shows a hypoattenuating rounded mass in the location of a prior laceration, consistent with a posttraumatic cyst.*



(Left) *Coronal T2 FS MR shows a "claw" of splenic tissue splayed along the inferior margin of a large cyst. The claw sign can help determine the organ of origin for a large mass. This splenic cyst was benign & posttraumatic.* **(Right)** *Coronal SSFSE T2 MR shows innumerable, small, cystic-appearing lymphatic malformations throughout the spleen. Patients with generalized lymphatic anomaly frequently have numerous cystic bone & splenic lesions in addition to soft tissue malformations & possibly pleural disease.*



IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed, round mass with simple fluid characteristics
  - Distinction between congenital (true) & acquired cysts not possible by imaging
- Size
  - Variable

Ultrasonographic Findings

- Grayscale ultrasound
  - Well-circumscribed, rounded mass
  - Typically anechoic but can have low-level internal echoes
  - ↑ posterior acoustic enhancement
  - Thin wall
- Color Doppler
  - No internal color flow

CT Findings

- Well-circumscribed, water attenuation mass
- Typically solitary & unilocular
- No enhancement
- Can occasionally have thin rim of Ca^{2+}

MR Findings

- Well-circumscribed, rounded mass
- Typically hypointense on T1WI unless proteinaceous or hemorrhagic material present internally
- Hyperintense on T2WI
- No enhancement
- No restricted diffusion (unless prior hemorrhage)

Imaging Recommendations

- Best imaging tool
  - US typically suffices to demonstrate cystic nature of splenic mass
  - Further characterization with any imaging modality not typically possible

DIFFERENTIAL DIAGNOSIS

Splenic Lymphoma

- Can be solitary, multifocal, or diffuse
- Typically higher density/echogenicity than cysts
- Look for associated lymphadenopathy

Splenic Infection

- Numerous tiny lesions typical for fungal microabscesses
- Look for lesions in liver, kidneys, & lungs

Splenic Infarct

- Typically well-demarcated, peripheral, wedge-shaped foci of ↓ attenuation/echogenicity
- Higher attenuation/density than cysts
- Lacks internal blood flow
- May resolve completely or evolve into acquired cyst

Splenic Perfusion Artifact

- Focal areas of ↓ attenuation on early phase of CECT
  - Classically archiform waves of ↑ & ↓ enhancement
- Resolves on delayed phase (typically not necessary)
- US normal

Intrasplenic Pancreatic Pseudocyst

- May result from tail pancreatitis
- Patient typically has clinical &/or imaging evidence of recent pancreatitis

PATHOLOGY

General Features

- Congenital (true) cysts = epidermoid or mesothelial cysts
  - Differentiated by type of epithelial lining
  - Thought to arise from defect in mesothelial migration
- Acquired cysts (comprising 80% of splenic cysts)
  - Posttraumatic
  - Postinfarction
  - Inflammatory/infectious (pyogenic, parasitic, fungal)
  - Vascular (venolymphatic)
    - May be congenital but not visible early

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Typically asymptomatic & incidentally identified
- Other signs/symptoms
  - Left upper quadrant pain
  - Palpable mass
  - Splenomegaly
  - ± elevated serum CEA or CA 19-9

Natural History & Prognosis

- Complications may include hemorrhage, rupture, infection

Treatment

- Small & asymptomatic masses typically require no treatment
- Larger or symptomatic masses may undergo surgical excision or splenectomy

DIAGNOSTIC CHECKLIST

Consider

- Infectious & neoplastic processes should be excluded
  - Splenic neoplasms uncommon in children (outside of lymphoma)

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Cat-Scratch Disease

KEY FACTS

TERMINOLOGY

- Spectrum of clinical manifestations resulting from *Bartonella henselae* inoculation of skin by cat scratch or bite
  - Typical cat-scratch disease (CSD) includes tender regional adenopathy + fever 1-3 weeks after inoculation
    - Papule at scratch site may be 1st clue
  - Self-limited in most patients; dissemination in 5-14%

IMAGING

- Numerous small hypoechoic (US), hypodense (CT) splenic &/or hepatic lesions of variable size
- ± splenomegaly, rarely complicated by rupture
- Residual granulomas may calcify

TOP DIFFERENTIAL DIAGNOSES

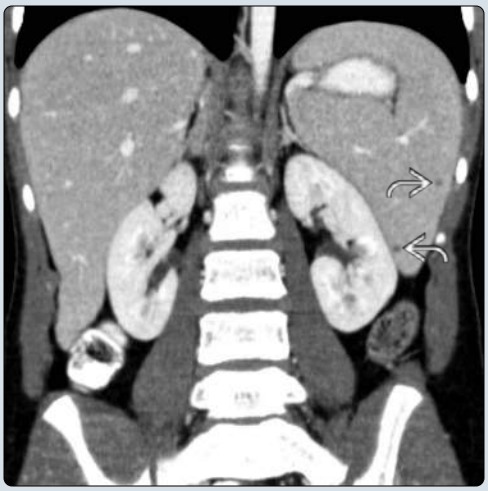
- Splenic lymphoma
- Splenic microabscesses
- Lymphangiomatosis
- Other granulomatous diseases

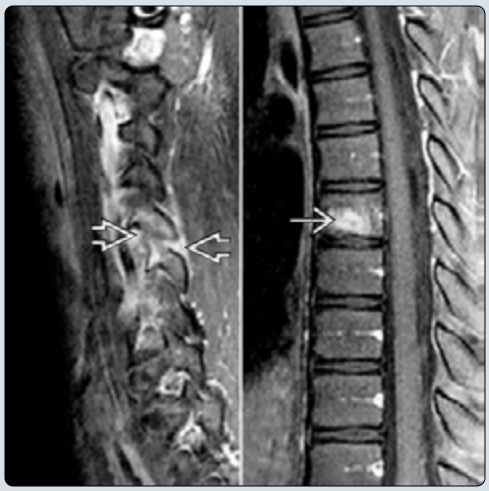
CLINICAL ISSUES

- Typical CSD: Single lymph node involved in up to 85%
  - Axillary, epitrochlear > head/neck > groin, other nodes
- Disseminated disease manifestations include hepatic &/or splenic involvement, conjunctivitis, retinitis, encephalopathy, osteomyelitis, endocarditis, pneumonia
  - Adenopathy occurs in only 55% of splenic cases
- Diagnosis made by combination of cat exposure history, positive serum antibody titers or DNA PCR, hepatic/splenic lesions, positive biopsy
- Treatment
  - Most typical CSD cases in immunocompetent hosts resolve within months without antibiotics
  - Antibiotic therapy for systemic disease &/or immunocompromised states

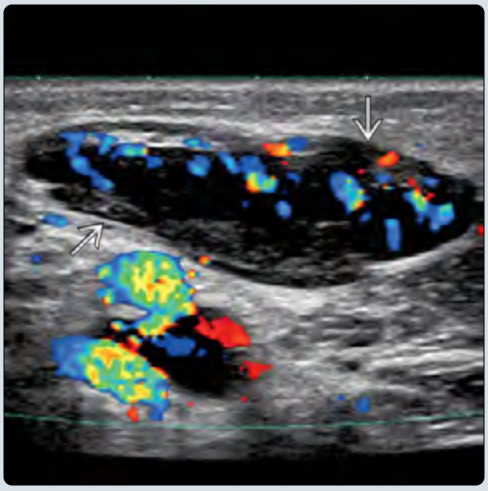
DIAGNOSTIC CHECKLIST

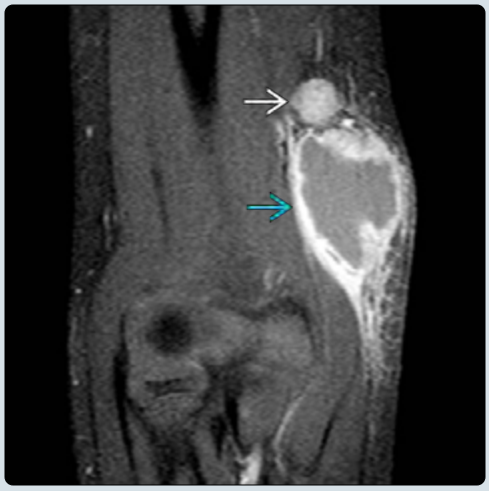
- Multifocal splenic lesions → nonspecific; recent cat exposure ± focal adenopathy should raise suspicion for CSD





(Left) *Abdominal coronal CECT in an 11 year old with 1 month of fevers + back & neck pain shows numerous tiny, hypodense lesions throughout the spleen.* **(Right)** *In the same patient, sagittal T1 C+ FS MR images of the cervical (L) & thoracic (R) spine show abnormal enhancement from osteomyelitis of the C5 & C6 vertebral elements (centered at the facet joint) & the T7 vertebral body. Excisional biopsy of enlarged inguinal lymph nodes showed cat-scratch disease.*





(Left) *Transverse color Doppler ultrasound shows an enlarged & hyperemic inguinal lymph node in a teenager with seizures. The patient had positive Bartonella serologies.* **(Right)** *Coronal T1 C+ FS MR in an 8-year-old patient with arm swelling shows a suppurative medial epitrochlear lymph node with thick irregular wall & central necrosis/fluid. Additional adjacent adenopathy is noted. The patient had a history of kitten exposure & showed positive Bartonella IgM & IgG titers.*

TERMINOLOGY

Abbreviations

- Cat-scratch disease (CSD)

Definitions

- Spectrum of clinical manifestations resulting from *Bartonella henselae* inoculation of skin by cat scratch or bite
  - Most common clinical presentation ("typical CSD")
    - Tender regional adenopathy + fever 1-3 weeks after inoculation; papule at scratch site may be 1st clue
  - Self-limited in most patients
    - Disseminated disease in 5-14%

IMAGING

General Features

- Best diagnostic clue
  - Splenic ± hepatic lesions in setting of focal extremity adenopathy & recent cat exposure
- Morphology
  - Numerous small hypoechoic (US), hypodense (CT) splenic lesions of variable size
  - Lesions show ↑ metabolic activity on FDG-PET
  - ± splenomegaly, rarely complicated by rupture
  - Residual granulomas may calcify

Imaging Recommendations

- Best imaging tool
  - Ultrasound
- Protocol advice
  - Clinical history important as many pathologies involving spleen have similar imaging appearance

DIFFERENTIAL DIAGNOSIS

Splenic Lymphoma

- Hypoechoic/hypodense round masses of variable size
- Multifocal/confluent adenopathy ± other visceral lesions

Splenic Microabscesses

- Fungal infection (most commonly *Candida*) with numerous tiny hypoechoic lesions
  - ± central echogenic foci (target or bull's-eye sign)

Lymphangiomatosis

- Numerous small hypoechoic/hypodense splenic lesions in setting of cystic soft tissue &/or bony lymphatic malformations

Other Granulomatous Diseases

- Hypoechoic splenic lesions from tuberculosis, sarcoid, histoplasmosis, Wegener granulomatosis

PATHOLOGY

General Features

- Associated abnormalities
  - Lymphadenopathy (only in 55% of splenic cases), conjunctivitis, retinitis, encephalopathy, osteomyelitis, endocarditis, pneumonia

Microscopic Features

- Necrotizing granulomas + microabscesses
- Organism detection by
  - Positive Warthin-Starry silver stain
  - Immunohistochemical antibody stain
  - Polymerase chain reaction (PCR)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Erythematous papule at inoculation site, regional adenopathy, fever ("typical CSD": Up to 95% of cases)
    - Adenopathy 1-3 weeks post inoculation
      - Axillary, epitrochlear > head/neck > groin, other
      - Single node involved in up to 85% of patients
- Other signs/symptoms
  - Aches, malaise, abdominal pain, fever of unknown origin
- Clinical profile
  - Positive serologic titers for IgM, IgG
  - Elevated erythrocyte sedimentation rate

Demographics

- Age
  - Up to 87% of cases < 18 years old
- Epidemiology
  - 22,000 cases of CSD per year in United States
  - 50-87% report scratch by cat

Natural History & Prognosis

- Diagnosis made by combination of clinical, laboratory, &/or imaging criteria
  - Hepatosplenic lesions by imaging, cat or flea contact, positive antibody serology, positive biopsy, sterile pus aspiration

Treatment

- Most typical CSD cases in immunocompetent hosts resolve within months without antibiotics
  - Suppurative nodes require drainage in 10%
- Antibiotic therapy for systemic disease &/or immunocompromised state

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Multifocal splenic lesions → nonspecific: Clinical history of recent cat exposure ± focal adenopathy should raise suspicion for CSD

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Pancreatoblastoma

KEY FACTS

TERMINOLOGY

- Primary exocrine epithelial malignancy of pancreas occurring in young children

IMAGING

- Most commonly arises in pancreatic head
- Mean size: 7-18 cm
  - May be difficult to determine organ of origin due to size
- Well-circumscribed mass with smooth border
- Heterogeneous appearance due to mixed cystic & solid foci
  - Predominantly hypoechoic (US), hypoenhancing (CECT)
- Speckled Ca^{2+} common

TOP DIFFERENTIAL DIAGNOSES

- Pancreatic pseudocyst
- Solid pseudopapillary neoplasm
- Pancreatic neuroendocrine tumor
- Non-Hodgkin lymphoma

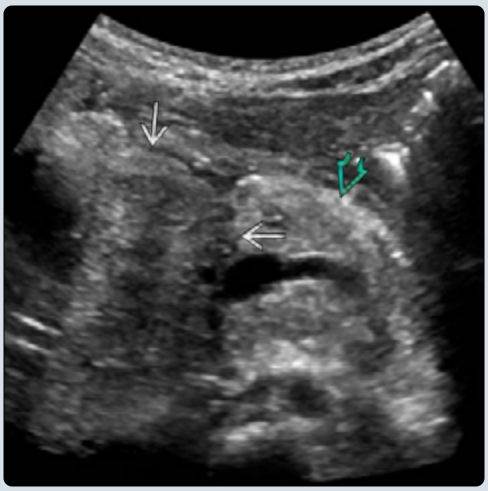
PATHOLOGY

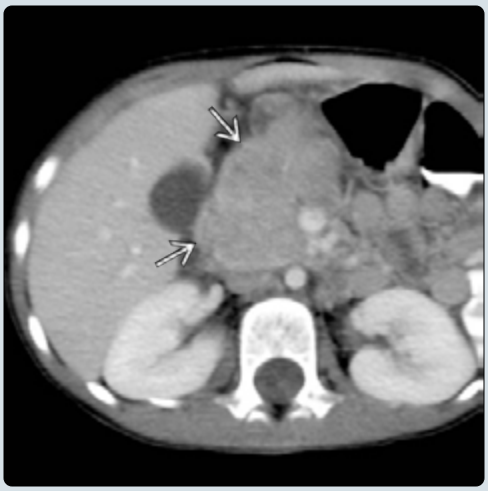
- ↑ incidence in patients with Beckwith-Wiedemann syndrome & familial adenomatous polyposis

CLINICAL ISSUES

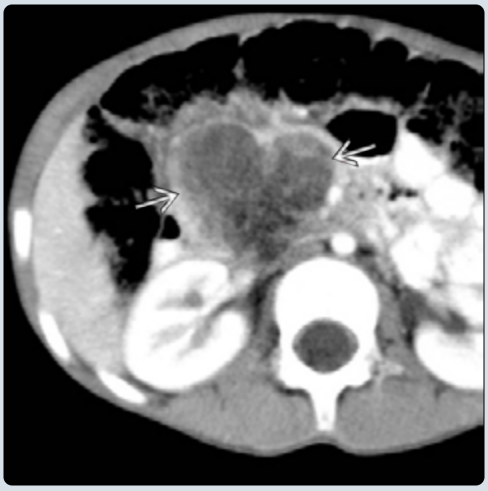
- Most common malignant pancreatic tumor of young children
- Mean age at presentation: 5 years
- 1.3-2.7x more common in male patients
- More common in Asians
- Most commonly presents as asymptomatic abdominal mass
- Elevated α-fetoprotein in 33-70% of patients
- Metastases present in 1/3 at diagnosis
  - Liver > lungs, lymph nodes
- Best treated with surgical resection ± chemotherapy
  - Recurrence common

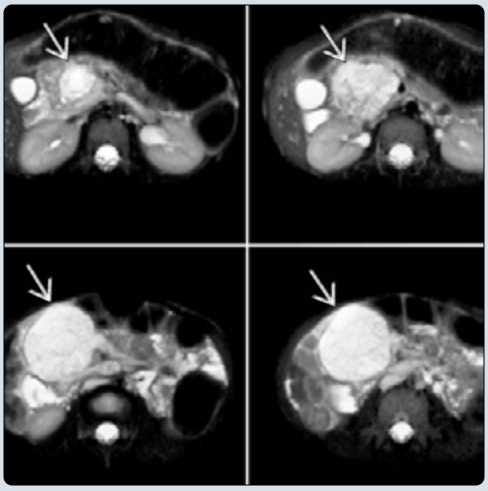
(Left) *Transverse ultrasound of the pancreas in a child with pancreatoblastoma shows a large lobulated hypoechoic mass arising from the pancreatic head.* **(Right)** *Axial CECT in the same patient shows a lobulated enhancing mass arising from the head of the pancreas, which is the most common site of origin. Pancreatoblastoma usually presents in the 1st decade of life as an asymptomatic abdominal mass.*





(Left) *Axial CECT in a child with pancreatoblastoma shows a heterogeneously enhancing but predominantly hypodense mass arising from the head of the pancreas. Heterogeneous enhancement is typical of pancreatoblastoma.* **(Right)** *Axial T2 FS MRs in the same child show a lobulated hyperintense mass arising from the head of the pancreas. The internal signal intensity suggests components of septated fluid.*





TERMINOLOGY

Definitions

- Primary exocrine epithelial malignancy of pancreas occurring in young children

IMAGING

General Features

- Best diagnostic clue
  - Large, heterogeneous mass of pancreas in child
- Location
  - 40-50% arise in pancreatic head
  - 13-25% arise in pancreatic body
  - 13-37% arise in pancreatic tail
- Size
  - Large tumors; average: 7-18 cm in diameter
- Morphology
  - Well-circumscribed, large mass of pancreas
    - May be difficult to determine organ of origin

CT Findings

- CECT
  - Well-circumscribed mass with smooth border
  - Heterogeneous attenuation of mixed cystic & solid foci
    - Cystic areas represent necrosis
    - Overall hypodense vs. normally enhancing pancreas
  - Multiloculated with enhancing septa
  - Small punctate Ca^{2+} common

MR Findings

- T1WI: Well-circumscribed, iso- to hypointense mass
  - Areas of lower signal correspond to necrosis
  - Areas of higher signal correspond to hemorrhage
- T2WI: Heterogeneously hyperintense mass
- T1WI C+: Enhances < adjacent pancreas
  - Rim may enhance to greater degree than remainder of pancreas

Ultrasonographic Findings

- Well-circumscribed mass with heterogeneous echogenicity
  - Cystic & solid components
    - Cystic foci hypoechoic with hyperechoic septa

DIFFERENTIAL DIAGNOSIS

Pancreatic Pseudocyst

- Collection of fluid, tissue, debris, pancreatic enzymes, & blood surrounded by fibrous capsule
- Gradually develops 4 weeks after onset of acute pancreatitis

Solid Pseudopapillary Neoplasm

- Most common pancreatic neoplasm in pediatric population
- Occurs in adolescent to young adult female patients
- Large, well-defined, solid mass of pancreas with variable cystic components
- Usually benign

Pancreatic Neuroendocrine Tumor

- Rare tumor in children, may be nonfunctioning
  - Insulinoma most common in children
- Usually hypervascular
- Associated with von Hippel-Lindau, multiple endocrine neoplasia type 1

Non-Hodgkin Lymphoma

- Typically smaller & uniformly hypodense solid lesion
- Pancreatic involvement typically occurs in setting of other nodal &/or visceral involvement
  - Adjacent adenopathy common

PATHOLOGY

General Features

- Associated abnormalities
  - Beckwith-Wiedemann syndrome
    - Patients present earlier in life
    - More likely to have cystic tumor
  - Familial adenomatous polyposis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic large abdominal mass
- Other signs/symptoms
  - Abdominal pain, fatigue, lethargy, weight loss, anorexia, diarrhea, & vomiting
  - Elevated α-fetoprotein in 33-70% of patients

Demographics

- Age
  - Typically occurs in 1st decade of life
    - Mean age at presentation: 5 years
- Gender
  - 1.3-2.7x more common in male patients
- Ethnicity
  - More common in Asians
- Epidemiology
  - Most common malignant pancreatic tumor of young children

Natural History & Prognosis

- Metastases in 1/3 of patients at presentation
  - Liver > lungs, lymph nodes
- Recurrence common after resection

Treatment

- Best treated with surgical resection
- Adjuvant/neoadjuvant chemotherapy often used

DIAGNOSTIC CHECKLIST

Consider

- Pancreatoblastoma in child with large pancreatic mass

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Solid Pseudopapillary Neoplasm

KEY FACTS

TERMINOLOGY

- Synonyms: Solid-cystic pancreatic tumor, Frantz tumor, papillary cystic tumor, solid & papillary neoplasm, solid & cystic acinar cell tumor, papillary epithelial neoplasm

IMAGING

- Large, well-defined, solid mass of pancreas with variable cystic components
  - Mean size: 6-10 cm
- Central areas of ↑ T1 & heterogeneous T2 signal due to hemorrhage or necrosis
- Peripheral low signal due to fibrous capsule or compressed pancreatic tissue
- Eggshell Ca^{2+} may be present in 1/3
- Studies mixed regarding most common pancreatic site of origin (e.g., body & tail vs. equal distribution throughout)

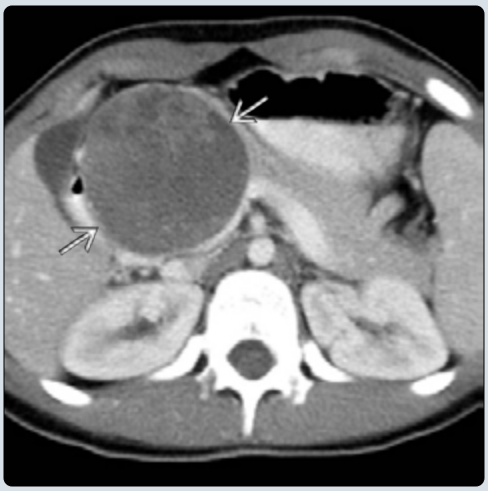
TOP DIFFERENTIAL DIAGNOSES

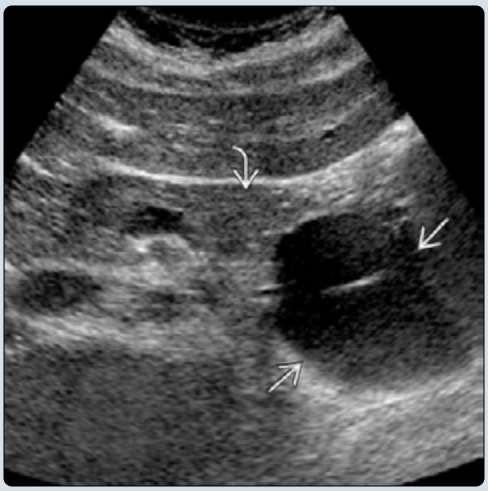
- Pancreatic pseudocyst
- Pancreatoblastoma
- Pancreatic adenocarcinoma

CLINICAL ISSUES

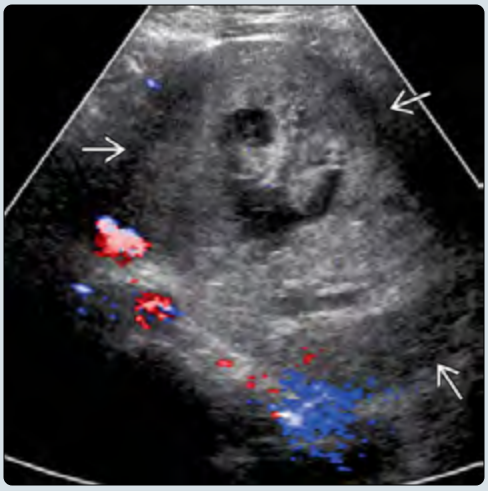
- Mean age: 26 years old
- 9x more common in females
- Accounts for 1-3% of all pancreatic tumors
  - Most common primary pancreatic neoplasm in pediatric population
- Patients most commonly present with large epigastric mass
  - Can present with chronic abdominal pain or acute abdominal pain after cyst rupture
- Usually benign tumor
  - > 95% overall survival
  - Local spread & metastases to liver reported in 10-20%
- Treatment: Surgical resection
  - Can rarely have local recurrence after resection

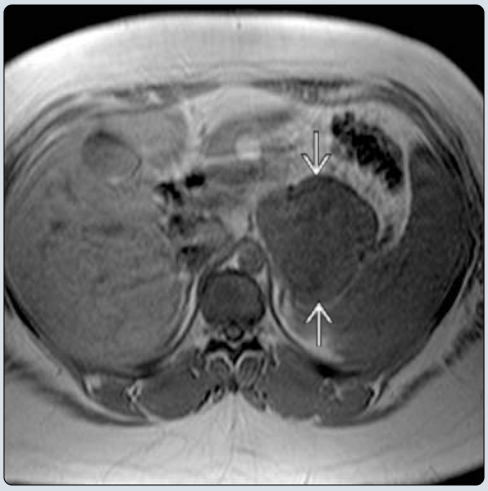
(Left) *Axial CECT in an adolescent female with a solid pseudopapillary neoplasm (SPN) shows a well-defined, hypodense, round mass ➔ arising from the pancreas. SPNs occur almost exclusively in women in their 2nd or 3rd decade of life.* **(Right)** *Transverse abdominal ultrasound in a young female with an SPN shows a cystic mass ➔ arising from the tail of the pancreas ➔. SPNs are variable in their appearance with differing solid & cystic components.*





(Left) *Longitudinal ultrasound of an adolescent female with an SPN shows a large, heterogeneous mass ➔ of the tail of the pancreas. The mass is mostly solid with a small central cystic component. There is no appreciable internal blood flow. SPNs often present as a large mass with a mean size at presentation of 6-10 cm.* **(Right)** *Axial T1 MR in a young adult female with a SPN shows a hypointense mass ➔ arising from the tail of the pancreas. On MR, SPNs can show mildly increased signal related to hemorrhage.*





TERMINOLOGY

Synonyms

- Solid-cystic pancreatic tumor, Frantz tumor, papillary cystic tumor, solid & papillary neoplasm, solid & cystic acinar cell tumor, papillary epithelial neoplasm

Definitions

- Exocrine tumor of pancreas

IMAGING

General Features

- Best diagnostic clue
  - Solid & cystic pancreatic mass in young woman
- Location
  - Studies mixed regarding most common site of origin in pancreas (e.g., body & tail vs. equal distribution throughout)
- Size
  - Mean: 6-10 cm
- Morphology
  - Well-defined pancreatic mass with variable cystic components

Radiographic Findings

- Soft tissue mass in upper abdomen displacing bowel

CT Findings

- Large, well-defined, solid mass of pancreas with variable cystic components
- Eggshell Ca^{2+} may be present in 1/3

MR Findings

- T1WI
  - Large hypointense mass of pancreas
  - Central areas of ↑ signal due to hemorrhage or necrosis
  - Peripheral ↓ signal due to fibrous capsule or compressed pancreatic tissue
- T2WI
  - Heterogeneous signal due to hemorrhage & necrosis
- T1WI C+
  - Vast majority show enhancing components

Ultrasonographic Findings

- Large pancreatic mass with thick capsule
- Mixed cystic & solid components
- Echogenic Ca^{2+} at periphery

Nuclear Medicine Findings

- PET/CT: Various patterns of FDG uptake depending on degree of cystic change

DIFFERENTIAL DIAGNOSIS

Pancreatic Pseudocyst

- Collection of fluid, tissue, debris, pancreatic enzymes, & blood surrounded by fibrous capsule
- Gradually develops 4 weeks after onset of acute pancreatitis

Pancreatoblastoma

- Most common malignant primary pancreatic tumor in young children
- Mean age of presentation: 5 years
- ~ 2x more common in male patients
- Associated with Beckwith-Wiedemann syndrome

Pancreatic Adenocarcinoma

- Extremely rare in pediatric population
- Usually originates from pancreatic duct
- Most common in head of pancreas → double duct sign of dilated biliary & pancreatic ducts

PATHOLOGY

Gross Pathologic & Surgical Features

- Pancreatic tumor with solid outer capsule
- Has solid & pseudopapillary structures, ↑ vascularization, & cellular degeneration

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Patients most commonly present with large epigastric mass
- Other signs/symptoms
  - Chronic abdominal pain, abdominal distension, back pain, vomiting
  - Acute abdominal pain after cyst rupture

Demographics

- Age
  - Mean age: 26 years
- Gender
  - 9x more common in females
- Ethnicity
  - More common in Asians
- Epidemiology
  - Accounts for 1-3% of all pancreatic tumors
  - Most common primary pancreatic neoplasm in pediatric population

Natural History & Prognosis

- Usually benign tumor
  - > 95% overall survival
  - Local spread & metastases to liver in 10-20%

Treatment

- Surgical resection
  - Can rarely have local recurrence after resection

DIAGNOSTIC CHECKLIST

Consider

- Solid & cystic pancreatic mass in young woman

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Pancreas Divisum

KEY FACTS

TERMINOLOGY

- Incomplete fusion of dorsal & ventral pancreatic ducts with persistence of 2 separate ductal systems

IMAGING

- Separate pancreatic ducts seen draining dorsal & ventral pancreas on CECT, MRCP, or ERCP
  - Secretin may be used to ↑ diameter of pancreatic ducts
- Short ventral duct of Wirsung drains to major papilla
  - Ventral duct does not cross midline

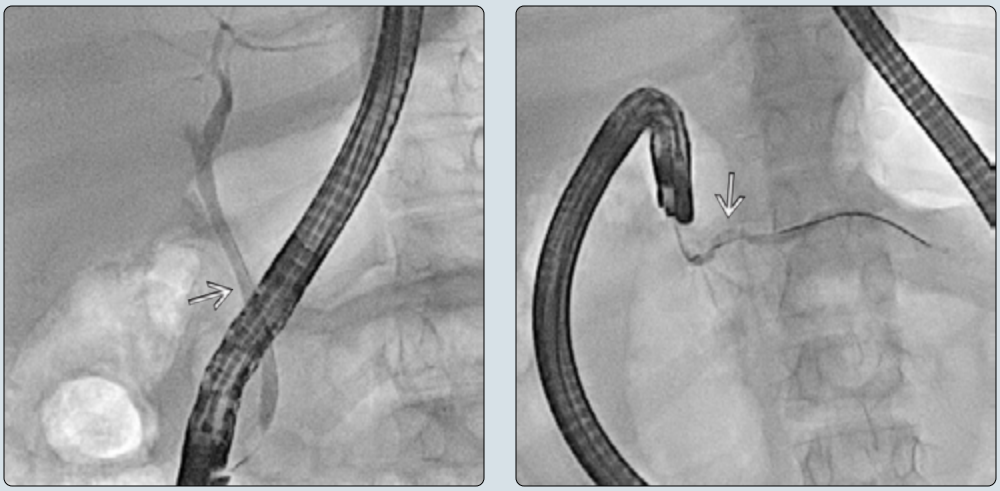
PATHOLOGY

- Dorsal anlage forms pancreatic body & tail
- Ventral anlage forms head of pancreas & uncinate process
- When dorsal & ventral ducts fail to fuse, dorsal duct drains majority of pancreas through minor papilla
  - Superior & anterior to major papilla, which drains duct of Wirsung & common bile duct
- Proposed etiology for pancreatitis
  - Small diameter of minor papilla causes relative obstruction to drainage of large volume from pancreatic body & tail
- Types of divisum
  - Type 1: Classic pancreas divisum with failure of fusion of dorsal & ventral ducts
  - Type 2: Absence of ventral duct of Wirsung
  - Type 3: Incomplete divisum with small caliber communication between ducts

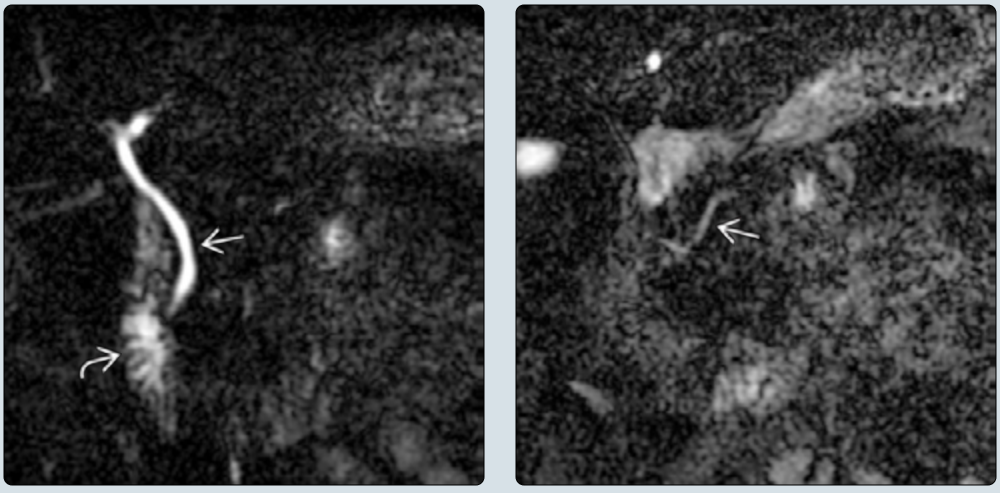
CLINICAL ISSUES

- Most common congenital variant of pancreatic ductal development
  - Occurs in 5-10% of general population
- Most patients asymptomatic
  - Children uncommonly present with pancreatitis
  - Pancreatitis more common between 30-50 years of age
- Most symptomatic patients respond to sphincterotomy of minor papilla

(Left) *ERCP in a patient with chronic pancreatitis & pancreas divisum shows cannulation of the major papilla. Contrast is present in the common bile duct ➡. The pancreatic duct is not visible.* **(Right)** *ERCP in the same patient after cannulation of the minor papilla shows contrast in the dorsal pancreatic duct ➡. Note the different position of the ERCP scope. The minor papilla is located superior & anterior to the major papilla.*



(Left) *Coronal MRCP in an adolescent with chronic pancreatitis & pancreas divisum shows fluid in the common bile duct ➡ & the duodenum ➡. The pancreatic duct is not visible.* **(Right)** *Coronal MRCP in the same patient shows fluid signal in the pancreatic duct ➡. Because the ventral pancreatic duct is never seen, this configuration is typical of a type 2 pancreas divisum.*



TERMINOLOGY

Definitions

- Incomplete fusion of dorsal & ventral pancreatic ducts with persistence of 2 separate ductal systems

IMAGING

General Features

- Best diagnostic clue
  - Separate pancreatic ducts draining dorsal & ventral pancreas

CT Findings

- CECT
  - Separate fluid-attenuation pancreatic ducts seen draining dorsal & ventral pancreas
    - May be difficult to see in children as pancreatic ducts usually not dilated
  - May see 2 separate pancreatic moieties separated by fatty cleft

MR Findings

- MRCP
  - Separate T2 hyperintense pancreatic ducts seen draining dorsal & ventral pancreas
    - Secretin may be used to ↑ diameter of pancreatic ducts
  - Duct of Santorini drains to minor papilla, superior & anterior to major papilla
  - Duct of Wirsung joins common bile duct & drains at major papilla

Ultrasonographic Findings

- Grayscale ultrasound
  - 2 separate pancreatic ducts that do not communicate
- Endoscopic ultrasound
  - Cannot demonstrate continuity of pancreatic duct in uncinate process & duct in body & tail

Nonvascular Interventions

- ERCP
  - Cannulation of major papilla reveals short ventral duct of Wirsung
    - Ventral duct ranges from 1-4 cm in length
    - Ventral duct does not cross midline
  - Cannulation of minor papilla can be difficult
    - Located superior & anterior to major papilla

Imaging Recommendations

- Best imaging tool
  - ERCP or MRCP: Defines pancreatic ductal anatomy

DIFFERENTIAL DIAGNOSIS

Annular Pancreas

- Result of incomplete separation of dorsal & ventral pancreatic anlage
- Pancreatic tissue encircles 2nd portion of duodenum
- Pancreatic duct encircles duodenum

Dorsal Agenesis of Pancreas

- Absence of pancreatic body or tail
- ERCP appearance of pancreatic duct identical to pancreas divisum
- Associated with polysplenia

PATHOLOGY

General Features

- Etiology
  - Dorsal & ventral pancreatic anlagen normally rotate & fuse during 6th-8th weeks of fetal development
    - Ventral anlage forms head of pancreas & uncinate process
    - Dorsal anlage forms pancreatic body & tail
  - When 2 ducts fail to fuse, dorsal duct remains & drains majority of pancreas through minor papilla
  - Proposed etiology for pancreatitis
    - Small diameter of minor papilla causes relative obstruction to drainage of large volume from pancreatic body & tail
- Types
  - Type 1: Classic pancreas divisum with failure of fusion of ducts
  - Type 2: Absence of ventral duct of Wirsung
  - Type 3: Incomplete divisum with small caliber communication between ducts

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Most patients asymptomatic
- Other signs/symptoms
  - Can be associated with pancreatitis

Demographics

- Age
  - Congenital anomaly that can present at any age
    - If symptomatic, patients may have recurrent abdominal pain & vomiting
    - Mean age of initial presentation in children: 6 years
- Epidemiology
  - Most common congenital variant of pancreatic ductal development
  - Occurs in 5-10% of general population

Treatment

- Most symptomatic patients respond to sphincterotomy of minor papilla

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Short ventral pancreatic duct does not communicate with long dorsal duct

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1. Rustagi T et al: Magnetic resonance cholangiopancreatography in the diagnosis of pancreas divisum: a systematic review and meta-analysis. Pancreas. 43(6):823-8, 2014
2. Wang DB et al: Pancreatitis in patients with pancreas divisum: imaging features at MRI and MRCP. World J Gastroenterol. 19(30):4907-16, 2013
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Pancreaticobiliary Maljunction

KEY FACTS

TERMINOLOGY

- Pancreaticobiliary maljunction (PBM): Congenital malformation in which pancreatic duct & common bile duct (CBD) join outside of duodenal wall
- Long common channel → pancreatic juices reflux into biliary system → high risk of gallbladder or biliary cancer

IMAGING

- Modalities
  - Ultrasound screening of patients with clinical/laboratory manifestations of hepatobiliary &/or pancreatic disease
  - MRCP for detailed ductal evaluation
  - ERCP to confirm diagnosis, particularly in equivocal cases
- Findings
  - Abnormal union of pancreatic duct & CBD outside of duodenal wall
  - ± visualization of fluid-filled long common channel extending from high confluence to duodenum
  - ± type I or type IV choledochal cyst
  - Associated abnormalities: Gallstones, gallbladder wall thickening, sequelae of pancreatitis

TOP DIFFERENTIAL DIAGNOSES

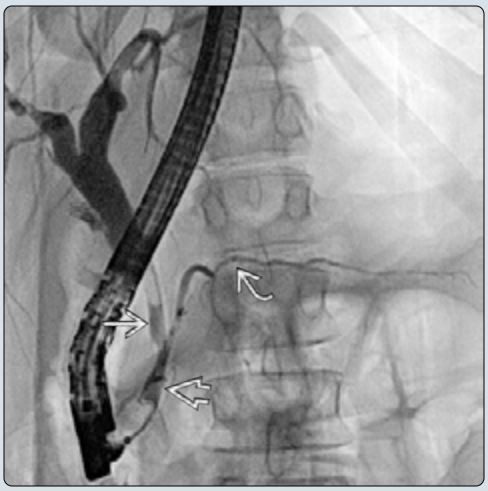
- Pancreas divisum
- Choledochal cyst

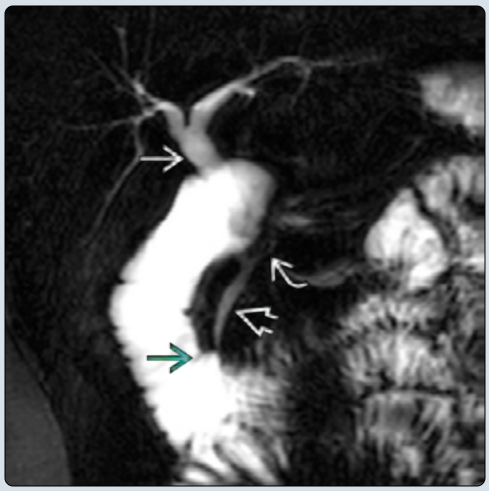
PATHOLOGY

- 200x ↑ risk of biliary cancer (usually in later adulthood)
  - Gallbladder most common site

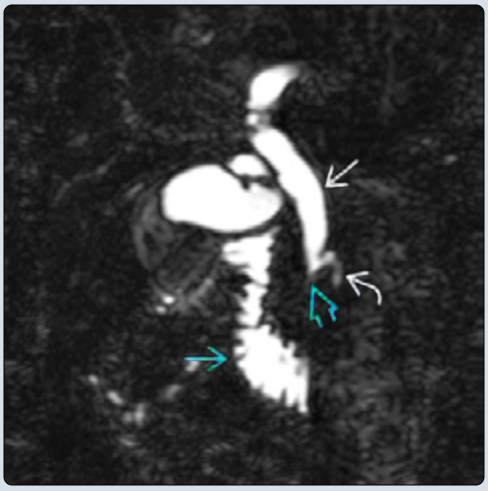
CLINICAL ISSUES

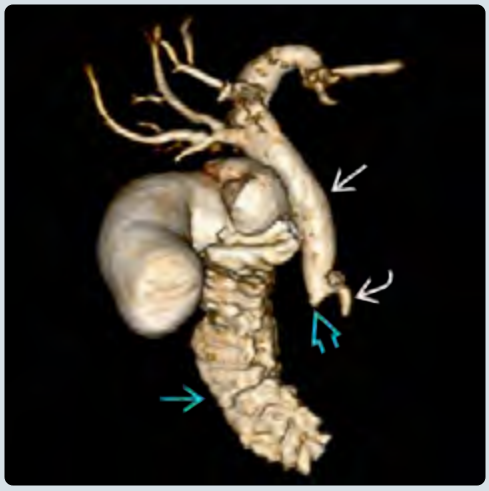
- 2 types of PBM
  - PBM with congenital biliary dilation (choledochal cyst)
    - More likely to present with symptoms in childhood
  - PBM without biliary dilation
    - Often asymptomatic until cancer develops in adulthood
- Treated with cholecystectomy & resection of extrahepatic bile duct due to high risk of malignancy





(Left) *AP ERCP shows partial opacification of a long common channel, which is formed by premature union of the common bile duct (CBD) & the pancreatic duct.* **(Right)** *Coronal MRCP in the same patient shows a long common channel formed by the union of the dilated CBD & the pancreatic duct outside the wall of the duodenum.*





(Left) *Coronal MRCP shows a dilated CBD & a normal caliber pancreatic duct joining in an abnormally high position. The long common channel is not visible on this image but can be inferred based on the location of the confluence & the position of the duodenum.* **(Right)** *3D coronal MRCP in the same patient again shows the abnormal distance between the confluence of the dilated CBD & the distal pancreatic duct relative to the duodenum, implying a long common channel.*

TERMINOLOGY

Synonyms

- Long common channel

Definitions

- Pancreaticobiliary maljunction (PBM): Congenital malformation where pancreatic duct & common bile duct (CBD) join outside of duodenal wall
  - Long common channel allows pancreatic juices to reflux into biliary system
  - Chronic reflux of pancreatic juices leads to high risk of gallbladder or biliary cancer

IMAGING

General Features

- Best diagnostic clue
  - Long common channel extending from abnormally high confluence of pancreatic duct & CBD to duodenum
    - Duct union lies outside of duodenal wall
    - No length of common channel yet agreed upon as diagnostic for PBM

MR Findings

- MRCP
  - Long fluid-filled channel distal to high confluence of pancreatic duct & CBD
  - Type I or type IV choledochal cyst may be present
  - Associated findings: Gallstones, sequelae of pancreatitis

Ultrasonographic Findings

- Transabdominal ultrasound
  - Type I or type IV choledochal cyst may be present
  - May identify nonspecific gallbladder wall thickening, cholelithiasis, dilated CBD
- Endoscopic ultrasound can confirm extraduodenal union of pancreaticobiliary junction

Other Modality Findings

- Endoscopic retrograde cholangiopancreatography (ERCP)
  - Diagnostic modality of choice to confirm ductal anatomy
  - Only method able to evaluate location & effect of sphincter of Oddi

Imaging Recommendations

- Best imaging tool
  - Ultrasound screening of patients with clinical/laboratory manifestations of hepatobiliary &/or pancreatic disease
  - MRCP for detailed ductal evaluation
    - Should be considered in patients with persistent, unexplained gallbladder wall thickening
  - ERCP to confirm diagnosis, particularly in equivocal cases

DIFFERENTIAL DIAGNOSIS

Pancreas Divisum

- Incomplete fusion of dorsal & ventral pancreatic ducts with persistence of 2 separate draining ductal systems

Choledochal Cyst

- Spectrum of malformations of extrahepatic & intrahepatic bile ducts
  - Classified via system developed by Todani
- Focal, multifocal, or extensive ductal dilatation
  - Greater than seen with acute biliary obstruction (which is uncommon in young children)
- May be associated with PBM

PATHOLOGY

Staging, Grading, & Classification

- 2 types of PBM
  - PBM with congenital biliary dilatation (choledochal cyst)
  - PBM without biliary dilatation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - PBM with congenital biliary dilatation
    - Presents in childhood
    - Symptoms: Abdominal pain, vomiting, jaundice, & fever
  - PBM without biliary dilatation
    - Presents in adulthood
    - Most patients asymptomatic until cancer develops
    - Up to 10% of patients present with cholangitis
- Other signs/symptoms
  - Manifestations of pancreatitis or cholelithiasis

Demographics

- More common in patients from Japan

Natural History & Prognosis

- Pancreatic enzymes induce chronic inflammation of biliary tree → proliferation of biliary epithelium → induction of carcinogenesis
  - 200x ↑ risk of biliary cancer
    - Gallbladder cancer most common
  - Cancer more common in PBM patients without biliary dilatation compared to those with dilatation (42.4% vs. 21.6%)
  - Cancers typically occur in 6th or 7th decade of life

Treatment

- Cholecystectomy & resection of extrahepatic bile duct recommended due to high risk of malignancy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- May be found incidentally or with choledochal cyst

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2. Kamisawa T et al: Pancreaticobiliary maljunction and biliary cancer. J Gastroenterol. 50(3):273-9, 2015
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4. Kamisawa T et al: Diagnostic criteria for pancreaticobiliary maljunction 2013. J Hepatobiliary Pancreat Sci. 21(3):159-61, 2014
5. Kamisawa T et al: Recent advances and problems in the management of pancreaticobiliary maljunction: feedback from the guidelines committee. J Hepatobiliary Pancreat Sci. 21(2):87-92, 2014

Annular Pancreas

KEY FACTS

TERMINOLOGY

- Congenital anomaly with ring of pancreatic tissue surrounding 2nd portion of duodenum (D2); results in variable degrees of duodenal obstruction

IMAGING

- Radiograph: ± newborn "double bubble" of dilated stomach & 1st portion of duodenum (D1)
  - Distal bowel gas **is present**
- Upper GI: Circumferential narrowing of D2
- MRCP or ERCP: Split pancreatic duct encircling D2
- MR, CT, or ultrasound: Pancreatic tissue surrounding D2
  - Water useful as oral contrast for MR, CT, & ultrasound
- Diagnosis may be suggested on prenatal imaging

TOP DIFFERENTIAL DIAGNOSES

- Duodenal atresia
- Duodenal web
- Malrotation with midgut volvulus

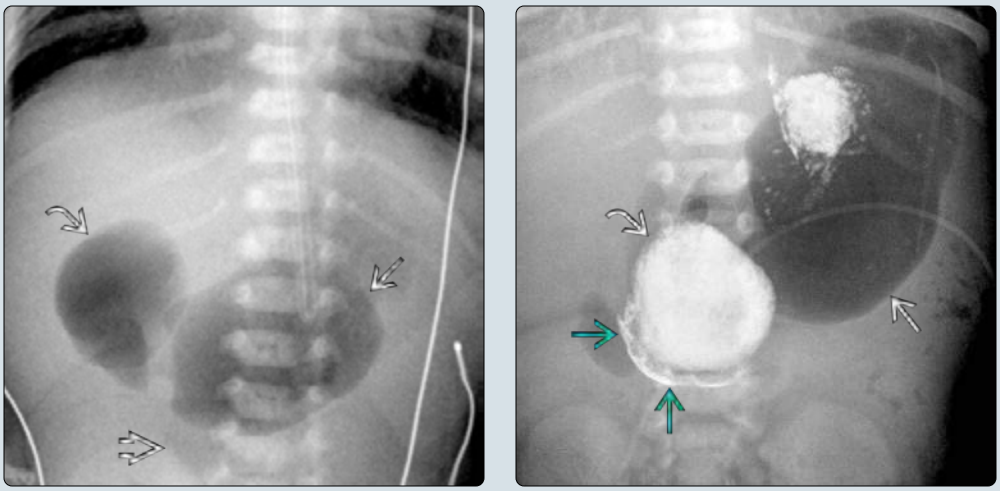
PATHOLOGY

- Associations include
  - Malrotation, duodenal stenosis, tracheoesophageal fistula, cardiac defects, pancreatobiliary anomalies (such as pancreas divisum)
  - Down syndrome

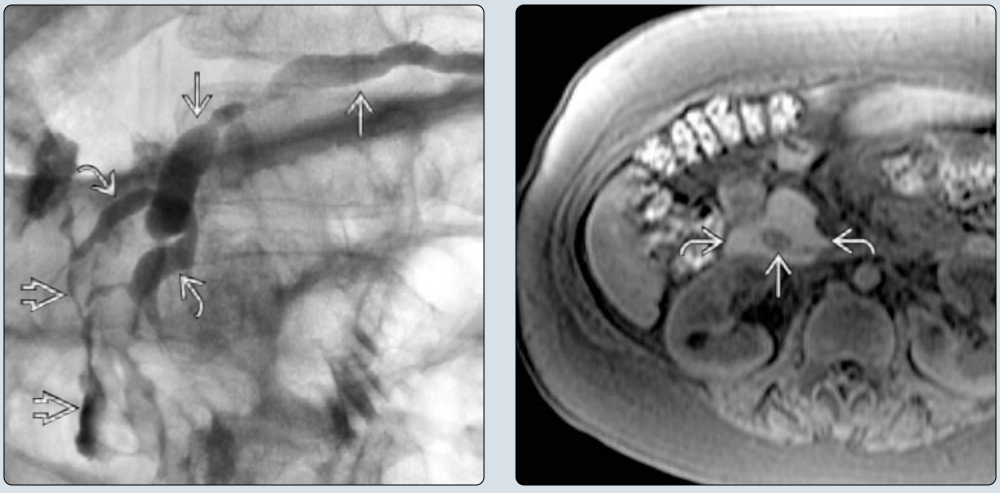
CLINICAL ISSUES

- 3 main presentations
  - Newborns: Signs of proximal duodenal obstruction either on prenatal ultrasound or after birth
    - Postnatal manifestations include vomiting (may be bilious), feeding intolerance, abdominal distension
  - Adults: Abdominal pain, ulcers, pancreatitis
  - Incidental finding: Asymptomatic in 2/3 of patients
- 51.5% diagnosed in childhood (median age: 1 day)
- Treatment of duodenal obstruction: Surgical duodenal bypass

(Left) *AP radiograph of the abdomen in a newborn shows a "double bubble" configuration of the bowel gas with a dilated air-filled stomach & a bulbous proximal duodenum. Small amounts of distal bowel gas are present, suggesting a partial obstruction.* **(Right)** *Frontal image from an upper GI examination in the same patient with annular pancreas shows dilation of the stomach & first portion of the duodenum. Contrast does pass distally into the narrowed second & third portions of the duodenum.*



(Left) *Frontal image from an ERCP in a teenager shows a dilated & irregular main pancreatic duct secondary to recurrent pancreatitis. There are 2 ducts in the pancreatic head encircling the contrast in the second duodenum.* **(Right)** *Axial T1 FS MR in the same patient shows hyperintense pancreatic tissue surrounding the hypointense second duodenum, consistent with annular pancreas.*



TERMINOLOGY

Definitions

- Ring of pancreatic tissue encircling 2nd portion of duodenum (D2)

IMAGING

General Features

- Best diagnostic clue
  - "Double bubble" on prenatal & early postnatal imaging with dilated stomach & 1st portion of duodenum (D1)
    - Duodenum may be bulbous due to longstanding obstruction
  - Cross-sectional imaging shows ring of pancreatic tissue encircling & narrowing D2

Radiographic Findings

- Newborn with partial duodenal obstruction
  - Double bubble of dilated stomach & D1
  - Distal bowel gas **is present** (but may be diminished)

Fluoroscopic Findings

- Upper GI
  - Incomplete duodenal obstruction
    - Double bubble sign with diminished (but not absent) distal bowel gas
    - Delayed emptying of stomach/D1
  - Circumferential narrowing of D2
  - Relatively small, caliber duodenum beyond constriction
- ERCP
  - Split distal pancreatic duct encircling duodenum

CT Findings

- CECT
  - Pancreatic head tissue encircles D2
    - Remainder of pancreas normal in appearance (unless patient has acute or chronic pancreatitis)

MR Findings

- Pancreatic head tissue encircles D2
- MRCP: Split distal pancreatic duct encircles D2

Ultrasonographic Findings

- Can be suggested on prenatal ultrasound
  - Findings of duodenal obstruction including double bubble sign & polyhydramnios
- Postnatal ultrasound
  - Fluid-filled proximal duodenum up to pancreatic head

Imaging Recommendations

- Protocol advice
  - Water useful as oral contrast for MR, CT, & ultrasound
  - T1 FS MR increases conspicuity of pancreatic tissue

DIFFERENTIAL DIAGNOSIS

Duodenal Atresia

- Chronic in utero complete obstruction of D2 presenting in prenatal or newborn periods
- Radiograph (in absence of gastric decompression) shows classic large double bubble **without** distal bowel gas
- Upper GI not necessary

Duodenal Web

- Cause of partial duodenal obstruction
- Symptoms depend on aperture of web
- ± double bubble appearance; distal bowel gas is present
- ± visualization of thin membrane in duodenal lumen
  - May have "windsock" configuration with distal stretching of web beyond origin at proximal duodenal wall

Malrotation With Midgut Volvulus

- Typically presents with bilious emesis in neonates/infants
- Radiograph may show distended stomach & proximal duodenum with normal to ↓ distal bowel gas
  - In acute midgut volvulus, proximal duodenum not as bulbous as with longstanding in utero obstructions
- Upper GI shows abnormal abrupt tapering/"beaking" of distal D2/D3 with corkscrew configuration of contrast beyond transition point
- Ultrasound shows abnormal anterior position of D3 with clockwise "whirlpool" twisting of bowel & mesenteric vessels on color Doppler
- Requires emergent surgery to prevent bowel infarction

PATHOLOGY

Associated Anomalies

- Malrotation, duodenal stenosis, tracheoesophageal fistula, cardiac defects, pancreatobiliary anomalies (such as pancreas divisum)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 3 main presentations
    - Newborn: High-grade duodenal obstruction either on prenatal ultrasound or after birth
      - Signs after birth include emesis (may be bilious), feeding intolerance, abdominal distension
    - Adults: Abdominal pain, pancreatitis, ulcers
    - Incidental: Asymptomatic in 2/3 of patients

Demographics

- Age
  - 51.5% diagnosed in childhood
    - 77% of children diagnosed by 2 days of life
  - 48.5% diagnosed in adults
- Epidemiology
  - Present in 3 of 20,000 autopsies
  - Incidence as high as 1 in 1,000 by imaging
  - 140 cases per 100,000 patients with Down syndrome

Treatment

- Surgical duodenal bypass

SELECTED REFERENCES

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2. Etienne D et al: Annular pancreas: a review of its molecular embryology, genetic basis and clinical considerations. Ann Anat. 194(5):422-8, 2012
3. Zyromski NJ et al: Annular pancreas: dramatic differences between children and adults. J Am Coll Surg. 206(5):1019-25; discussion 1025-7, 2008

KEY FACTS

TERMINOLOGY

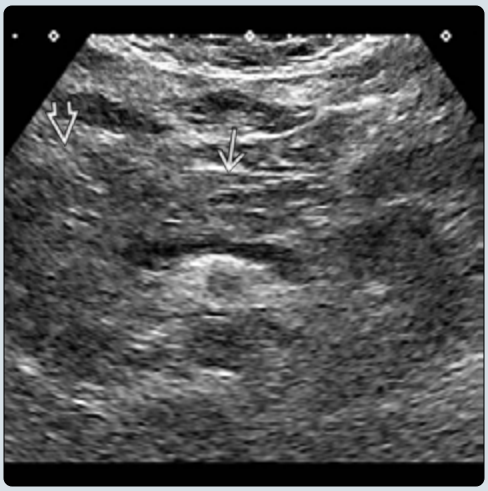
- Acute pancreatitis (AP): Acute inflammation of pancreas with variable involvement of local tissues/remote organs
- 2 of 3 features required for diagnosis: Abdominal pain consistent with disease, 3x rise in serum amylase or lipase, or imaging findings consistent with AP
- 2 types of AP: Interstitial edematous vs. necrotizing
- Fluid collections
  - Acute peripancreatic fluid collection: Fluid collection associated with interstitial edematous pancreatitis during first 4 weeks
  - Pancreatic pseudocyst: Fluid collection associated with interstitial edematous pancreatitis after first 4 weeks
  - Acute necrotic collection: Collection containing fluid & necrotic material during first 4 weeks
  - Walled-off necrosis: Collection containing fluid & necrotic material after first 4 weeks
- Chronic pancreatitis (CP): Relapsing or continuing inflammation with destruction of parenchyma & ducts

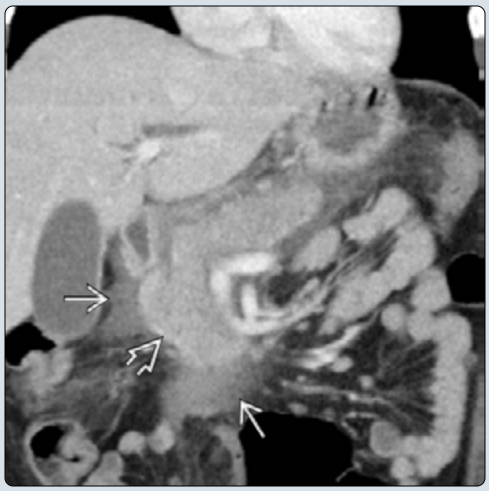
IMAGING

- Interstitial edematous pancreatitis: Gland enlargement with edema & homogeneous (± mildly decreased) enhancement
  - Acute peripancreatic fluid collection: Nonencapsulated collections of variable size & shape
  - Pancreatic pseudocyst: Encapsulated cystic lesion with no internal solid component, > 4 weeks from onset
- Necrotizing pancreatitis: Heterogeneous enhancement (moderately to severely decreased)
  - Acute necrotic collection: Nonencapsulated collection with heterogeneous contents
  - Walled-off necrosis: Well-circumscribed cavity containing necrotic tissue, > 4 weeks from onset
- CP: Atrophic pancreas with dilated duct & pancreatic Ca^{2+}

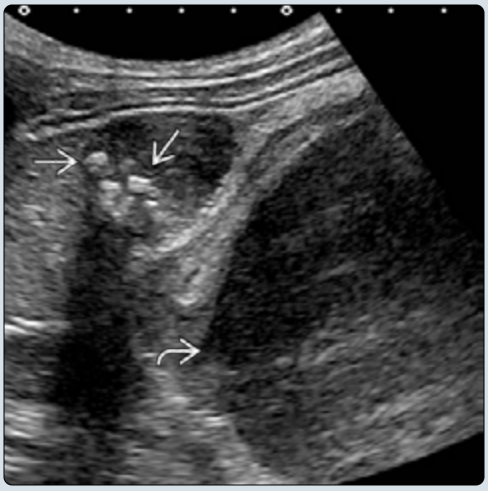
CLINICAL ISSUES

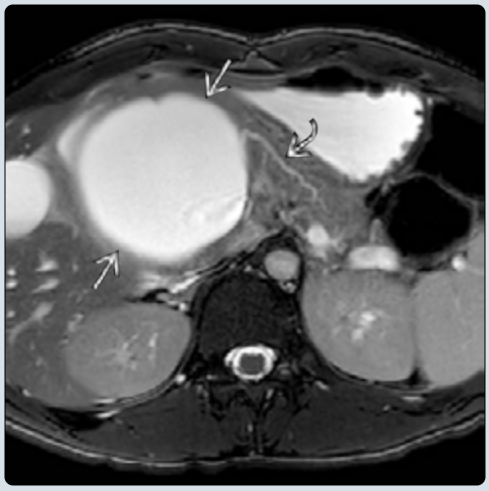
- Epigastric abdominal pain; ↑ amylase & lipase > 3x normal
- Therapy: Fluid resuscitation, early enteral nutrition, pain management





(Left) *Transverse US of the pancreas in an adolescent with abdominal pain & acute interstitial pancreatitis shows a poorly defined hypoechoic pancreas. The head of the pancreas is enlarged to a greater degree than the remainder of the gland, & the border is not distinct.* **(Right)** *Coronal CECT in the same patient shows enlargement of the head of the pancreas with surrounding peripancreatic edema. Interstitial edematous pancreatitis is characterized by homogeneous enhancement of the gland.*





(Left) *Transverse US of the right upper quadrant in an adolescent male with a 2-week history of pancreatitis shows cholelithiasis within the gallbladder lumen. There is a large fluid collection medial & posterior to the gallbladder, consistent with an acute peripancreatic collection.* **(Right)** *Axial 2D SSFP FS MR in the same patient shows the pancreatic duct extending to the large peripancreatic fluid collection. If the collection lasts > 4 weeks after the onset of symptoms, it would be considered a pseudocyst.*

TERMINOLOGY

Definitions

- Acute pancreatitis (AP): Acute inflammation of pancreas with variable involvement of local/regional tissues
  - 2 of 3 features required for diagnosis
    - Abdominal pain consistent with disease
    - Rise in serum amylase or lipase ≥ 3x normal
    - Imaging findings consistent with AP
  - 2 types of AP
    - Interstitial edematous pancreatitis
    - Necrotizing pancreatitis
- Acute recurrent pancreatitis: ≥ 2 distinct episodes of AP with complete resolution of pain for ≥ 1 month or normalization of enzyme levels
- Chronic pancreatitis (CP): Relapsing or continuing inflammatory disease of pancreas leading to irreversible destruction of pancreatic parenchyma & ducts with loss of exocrine function
- Fluid collections
  - Acute peripancreatic fluid collection: Fluid collection associated with interstitial edematous pancreatitis during first 4 weeks
  - Pancreatic pseudocyst: Fluid collection associated with interstitial edematous pancreatitis after first 4 weeks
  - Acute necrotic collection: Collection containing fluid & necrotic material during first 4 weeks
  - Walled-off necrosis: Collection containing fluid & necrotic material after first 4 weeks

IMAGING

General Features

- Best diagnostic clue
  - AP: Edematous pancreas & peripancreatic inflammation
    - Interstitial edematous pancreatitis: Gland enlargement, homogeneous enhancement (which may be mildly decreased diffusely), & edema
      - Acute peripancreatic fluid collection: Nonencapsulated collections of variable size & shape
      - Pancreatic pseudocyst: Encapsulated cystic lesion with no internal solid component > 4 weeks from onset
    - Necrotizing pancreatitis: Gland enlargement, heterogeneous enhancement (with foci of moderately to severely decreased enhancement), & edema
      - Acute necrotic collection: Nonencapsulated collection, may be single or multiple, with heterogeneous contents
      - Walled-off necrosis: Well-circumscribed cavity containing necrotic tissue, often involving pancreatic parenchyma, > 4 weeks from onset
    - Complications: Fluid collections, infection, disconnected duct, pseudoaneurysm of splenic artery, splenic vein thrombosis
  - CP: Pancreatic Ca^{2+} & atrophy
- Location
  - Acute peripancreatic fluid collection/pseudocyst: Adjacent to pancreas in lesser sac or anterior pararenal space
  - Acute necrotic collection/walled-off necrosis: Peripancreatic, pancreatic, or both
- Size
  - AP: Focal or diffuse enlargement of pancreas
  - CP: Atrophy of pancreas, enlargement of pancreatic duct

CT Findings

- Interstitial edematous pancreatitis
  - Focal or diffuse pancreatic enlargement
  - Homogeneous enhancement, often mildly decreased, of edematous pancreatic parenchyma
  - ± surrounding fat inflammation
  - Acute peripancreatic fluid collection: Homogeneous fluid collection without defined capsule
  - Pancreatic pseudocyst: Fluid density cyst with well-defined capsule > 4 weeks from onset
- Necrotizing pancreatitis
  - Can affect pancreas, peripancreatic tissue, or both
  - Area of moderately to severely decreased enhancement within pancreas or peripancreatic mesentery
  - Acute necrotic collection: Irregular hyperdense material within hypodense fluid collection
  - Walled-off necrosis: Fluid density collection in area of prior necrosis > 4 weeks from onset
    - Other findings: Larger size than pseudocyst, paracolic or retrocolic extension, irregular border, fat attenuation debris, thick septations, pancreatic deformity
- CP
  - Atrophic pancreas
  - Dilation of pancreatic duct
  - Coarse Ca^{2+} in pancreas

MR Findings

- Interstitial edematous pancreatitis
  - T1 or T2 FS: Peripancreatic fat stranding
  - T2: Enlarged, hyperintense pancreas
  - Diffuse enhancement
  - Acute peripancreatic fluid collection: Fluid signal on T1 & T2
  - Pancreatic pseudocyst: Fluid signal intensity on T1 or T2 with dependent debris
- Necrotizing pancreatitis
  - Portions of pancreas hypointense on T2
  - Portions of pancreas do not enhance after contrast
  - Acute necrotic collection
    - Nonencapsulated
    - T2 hypointense material layering within hyperintense fluid
  - Walled-off necrosis: Collection with visible capsule in area of prior necrosis, disconnected duct
- MRCP
  - Acute pancreatitis: Look for congenital anomalies
    - Pancreas divisum
    - Pancreatobiliary malfunction
  - Chronic pancreatitis: Look for sequelae of pancreatitis
    - Pancreatic duct strictures
    - Dilated pancreatic duct

Ultrasonographic Findings

- AP: Enlarged, hypoechoic pancreas

- Look for gallstones, biliary dilation
- CP: Atrophic pancreas with echogenic areas of Ca^{2+} & dilated pancreatic duct

Imaging Recommendations

- Best imaging tool
  - MR with MRCP for evaluating pancreas during acute attack
  - US for following known pancreatitis or fluid collection

DIFFERENTIAL DIAGNOSIS

Pancreatic Trauma

- Can cause pancreatitis
- More common in children due to relative size of pancreas in abdomen
- Can be caused by handlebar injury or nonaccidental trauma
- May cause pancreatic transection, duct injury

Pancreatic Lymphoma

- Uncommon site of involvement
- Enlargement of pancreas can be focal or diffuse
- Hypoechoic/hypoenhancing masses + adjacent adenopathy

Shock Pancreas

- Enlarged hyperenhancing pancreas with hypoenhancing septa in setting of hypovolemic shock
- Associated with other findings of shock
- Resolves following fluid resuscitation

Cystic Fibrosis

- Cause of CP
- Usually associated with diffuse fatty infiltration of pancreas
  - ± atrophy with large Ca^{2+}

PATHOLOGY

General Features

- Etiology
  - AP: Caused by acinar cell injury & premature activation of trypsinogen to trypsin in pancreas
    - Anatomic: Pancreas divisum, pancreatobiliary malfunction, choledochal cyst, annular pancreas
    - Biliary obstruction: Gallstones
    - Systemic illness: Sepsis, shock, hemolytic uremic syndrome, systemic lupus erythematosus
    - Drugs: L-asparaginase, valproic acid, azathioprine, mercaptopurine, mesalamine
    - Trauma: Motor vehicle accident, handlebar injury, nonaccidental trauma
    - Idiopathic
    - Metabolic disorders: Cystic fibrosis, hyperlipidemia, hyperparathyroidism
    - Genetic/hereditary
    - Autoimmune
  - CP: Pancreatic injury followed by sustained immune activation where fibrosis dominates
    - Cystic fibrosis: Most common cause of CP
    - Hereditary pancreatitis: Autosomal dominant disorder associated with recurrent bouts of pancreatitis
    - Anatomic causes: Pancreas divisum, annular pancreas, choledochal cyst
- Genetics
  - Multiple genetic mutations associated with CP
    - *SPINK1*, *CFTR*, *PRSS1*, & *PRSS2*

Staging, Grading, & Classification

- Scoring systems used in adults for AP (Ranson & Glasgow) not predictive in children
- Pediatric scoring systems not widespread

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Epigastric abdominal pain
  - Elevated amylase & lipase to level > 3x normal
- Other signs/symptoms
  - Vomiting, anorexia, & nausea
  - Patient appears ill, irritable, & quiet
  - Physical exam findings: Tachycardia, fever, hypotension, abdominal signs

Demographics

- Age
  - Any
- Epidemiology
  - Increasing in frequency over last decade
    - 3.6-13.2 cases per 100,000 individuals per year
  - ~ 50% of patients with hereditary pancreatitis develop CP
  - Mortality rate from AP: 2-10%
    - Decreased mortality rate in children as compared to adults
    - Mortality in children occurs due to multiorgan failure, sepsis, & underlying pathology

Natural History & Prognosis

- Pseudocyst most common complication of AP
- With CP, pain may be transient & not severe
  - End-stage disease leads to endocrine & exocrine dysfunction

Treatment

- Treatment has evolved over past decade
- Current mainstays of therapy: Fluid resuscitation, early enteral nutrition, & pain management

DIAGNOSTIC CHECKLIST

Consider

- AP: Patient with elevated amylase & lipase with enlarged, edematous pancreas
- CP: Atrophic pancreas with dilated duct & pancreatic Ca^{2+}

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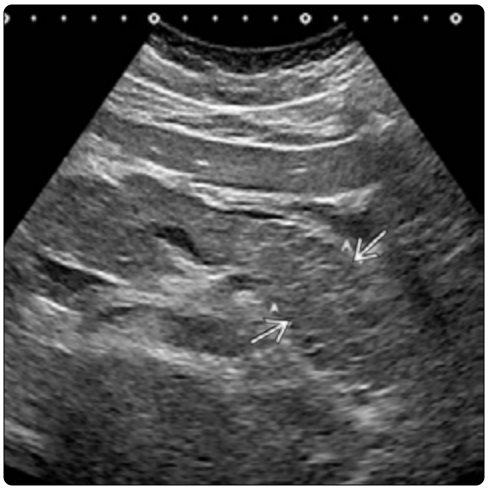
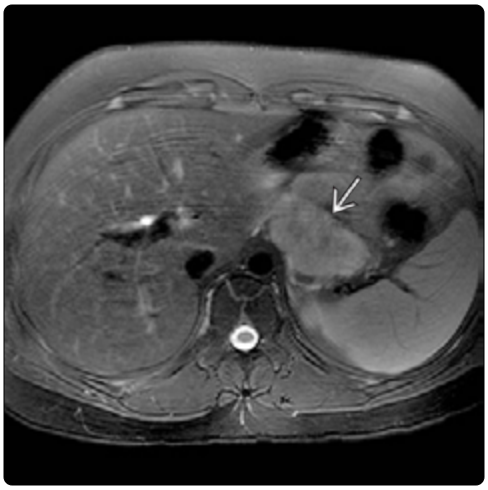
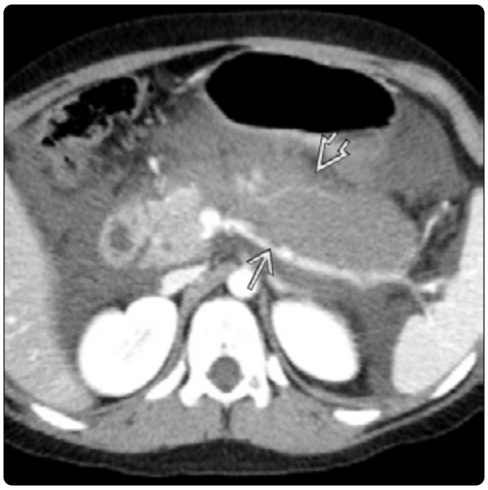
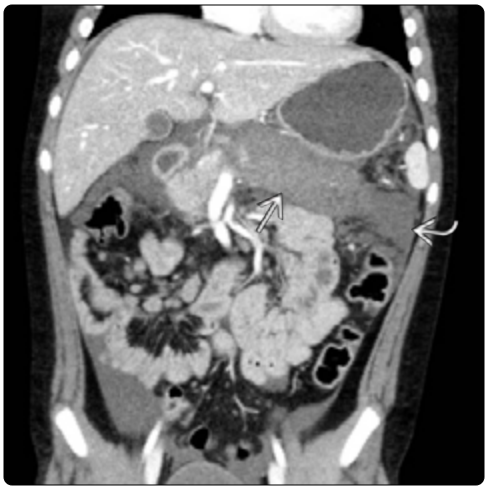


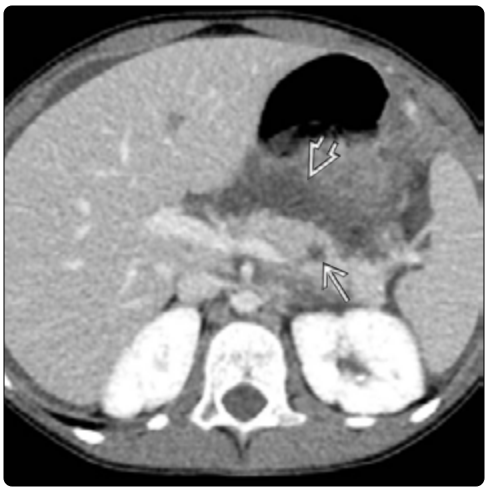


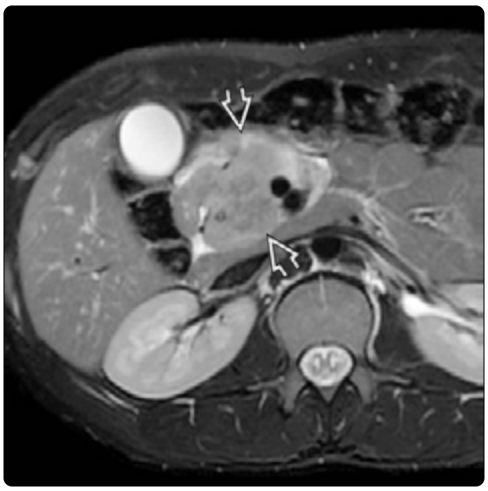
(Left) *Transverse US of the pancreas in an adolescent shows enlargement & mild heterogeneity of the pancreatic body & tail ➡.* **(Right)** *Axial T2 FS MR in the same patient shows enlargement & mildly heterogeneous signal of the pancreatic tail ➡ without substantial surrounding inflammation. Pancreatitis can affect any portion of the pancreas.*





(Left) *Axial CECT in a child after a bicycle handle bar injury shows lack of enhancement ➡ of the pancreatic body & tail. The splenic vein ➡ is irregular, due to partial thrombosis. The lack of enhancement is due to devascularization &/or necrotic pancreatitis.* **(Right)** *Coronal CECT in the same patient shows lack of enhancement ➡ of the pancreatic body & tail with an acute collection ➡ adjacent to the tail . If the collection becomes walled off & lasts > 4 weeks from the injury, it will be termed walled-off necrosis.*





(Left) *Axial CECT in a child with acute onset of pain shows a central area ➡ within the pancreas that does not enhance. In addition, there is fluid in the peripancreatic tissues ➡. These findings are concerning for necrotizing pancreatitis with pancreatic & peripancreatic necrosis.* **(Right)** *Axial T2 FS MR of an adolescent with autoimmune pancreatitis shows enlargement of the pancreatic head ➡ with a mildly heterogeneous appearance. There is a large list of entities that can cause acute pancreatitis.*

Primary Mesenteric Adenitis

KEY FACTS

TERMINOLOGY

- Self-limited benign inflammation of lymph nodes in bowel mesentery; diagnosis of exclusion

IMAGING

- Best diagnostic clue: Cluster of ≥ 3 enlarged lymph nodes, each ≥ 5 mm in size (short axis)
- Ultrasound
  - Enlarged lymph nodes often retain normal nodal architecture with fatty hilum & radiating vessels
  - Pain with compression over nodes
  - ± fat induration; no abscess or phlegmon
  - Normal appendix (< 6 mm & compressible)
- CECT findings
  - Mildly enlarged lymph nodes, most commonly anterior to right psoas muscle
  - Ileal or colonic wall thickening in < 1/3
    - More common < 5 years of age
  - Normal appendix without surrounding inflammation

TOP DIFFERENTIAL DIAGNOSES

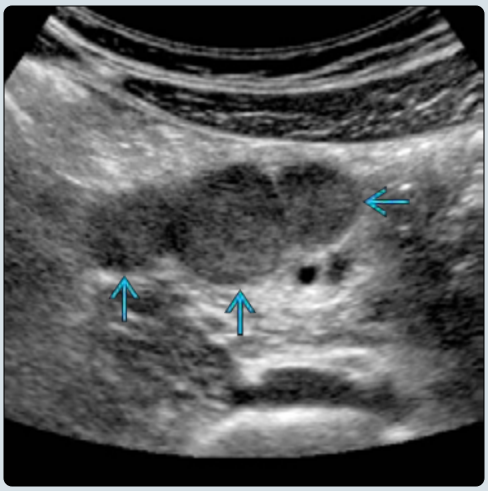
- Appendicitis
- Omental infarction
- Crohn disease
- Burkitt lymphoma

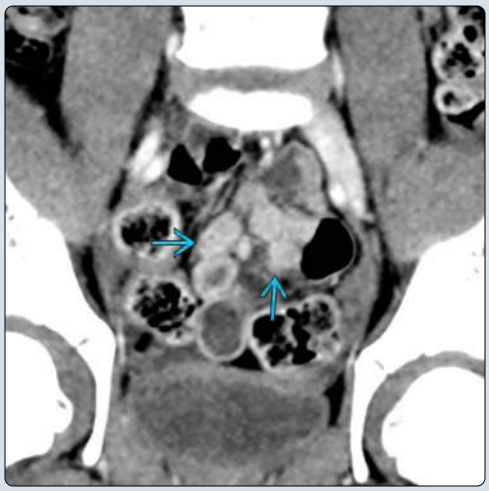
PATHOLOGY

- Majority likely secondary to viral or bacterial infection
- Recent URI in up to 25%

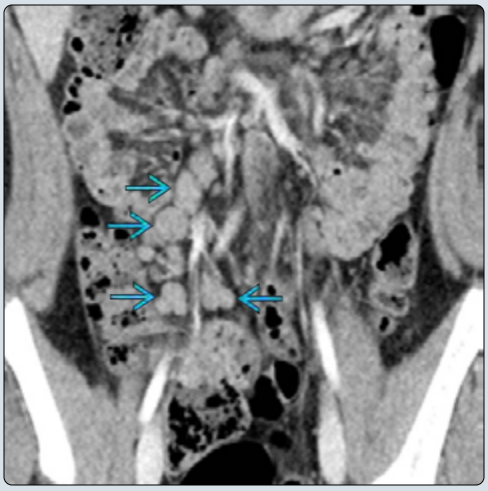
CLINICAL ISSUES

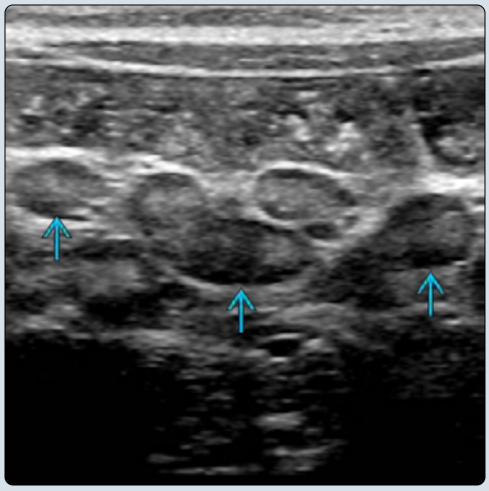
- Most commonly occurs < 15 years of age
- Mimics appendicitis: Similar clinical presentation
  - Most frequent alternative diagnosis to appendicitis
- Treatment conservative for primary mesenteric adenitis
  - Symptoms typically resolve by 2 weeks





(Left) *Transverse ultrasound of the right lower quadrant in a 10 year old with abdominal pain & fever shows a cluster of 3 lymph nodes, each measuring > 5 mm in short axis, at the site of tenderness. Additional enlarged lymph nodes were also present. The appendix was not visualized.* **(Right)** *Coronal CECT in the same patient shows a cluster of 3 lymph nodes, each measuring > 5 mm in size. The appendix was normal (not shown). The patient was diagnosed with mesenteric lymphadenitis & treated conservatively.*





(Left) *Coronal CECT from a 10 year old with abdominal pain shows numerous mildly enlarged lymph nodes in the right abdomen. The appendix was normal (not shown). These findings are consistent with mesenteric adenitis in the absence of other pathology.* **(Right)** *Longitudinal ultrasound in a 2 year old with abdominal pain shows mildly enlarged lymph nodes in the right abdomen. No other pathology was identified. The patient's symptoms resolved with conservative therapy, typical of mesenteric adenitis.*

TERMINOLOGY

Definitions

- Self-limited benign inflammation of lymph nodes in bowel mesentery; diagnosis of exclusion
- Secondary mesenteric adenitis refers to lymphadenopathy associated with detectable intraabdominal inflammatory process, commonly Crohn disease or infectious ileitis
- Most frequent alternative diagnosis to appendicitis

IMAGING

General Features

- Best diagnostic clue
  - ≥ 3 clustered right lower quadrant (RLQ) mesenteric lymph nodes that measure ≥ 5 mm in short-axis diameter in setting of acute or chronic abdominal pain
  - Normal appendix

Ultrasonographic Findings

- ≥ 3 lymph nodes in RLQ or more diffusely in mesentery
  - Size of ≥ 5 mm each in short axis
- Nodes often retain normal sonographic architecture with echogenic fatty hilum & radiating vessels
- May have pain with compression at nodes
- ± ↑ echogenicity of RLQ fat
- ± ileal or colonic wall thickening (especially < 5 years old)
- Normal appendix (< 6 mm & compressible)

CT Findings

- Cluster of ≥ 3 lymph nodes in RLQ mesentery
  - Each node ≥ 5 mm in short-axis diameter (mean: 11 mm)
  - Most commonly anterior to right psoas muscle
  - Often track more proximally along superior mesenteric artery branches
- Normal appendix
- Ileal (up to 33%) or colonic (up to 18%) wall thickening
  - More common < 5 years of age

Imaging Recommendations

- Best imaging tool
  - Graded compression ultrasound
    - No ionizing radiation
    - Confirms tenderness at lymph nodes
    - Normal appendix visualized

DIFFERENTIAL DIAGNOSIS

Appendicitis

- Noncompressible appendix ≥ 6 mm in diameter with surrounding fat induration
- Enlarged lymph nodes in 40-82%, mean of 9 mm
  - Typically less numerous than primary adenitis
- Ileal or colonic wall thickening with appendix perforation

Omental Infarction

- Poorly defined focus of echogenic/hyperattenuating fat immediately deep to anterior right abdominal wall
- Tenderness with compression on ultrasound

Crohn Disease

- Abnormal thickening, hyperenhancement/hyperemia, & diffusion restriction of terminal ileum & cecum
- ± mesenteric fat proliferation, vasa recta engorgement, enlarged lymph nodes, fistula, abscess, small bowel feces sign (due to distal stricture)

Meckel Diverticulum

- Inflamed blind-ending tubular structure arising from ileum
- ± gut signature of wall on ultrasound

Burkitt Lymphoma

- Larger conglomerate nodal masses, potentially inseparable from thickened bowel wall
- Loss of normal nodal architecture
- Rapid doubling time of tumor

Pelvic Inflammatory Disease

- Complex adnexal mass in sexually active girls
- Cervical motion tenderness

PATHOLOGY

General Features

- Majority of primary mesenteric adenitis likely infectious
  - Viral: Coxsackievirus & adenovirus
  - *Yersinia enterocolitica* classic
  - Also *Yersinia pseudotuberculosis*, *Helicobacter jejuni*, *Salmonella*, *Shigella*, *Campylobacter*, *Staphylococcus*
- Recent streptococcal URI in up to 25% (may be reactive)

CLINICAL ISSUES

Presentation

- Diffuse or focal RLQ abdominal pain most common
- ± nausea, vomiting, diarrhea, recent URI, leukocytosis

Demographics

- Most commonly < 15 years old

Natural History & Prognosis

- Symptoms typically resolve by 2 weeks

Treatment

- Primary mesenteric adenitis: Conservative
- Secondary mesenteric adenitis: Treat underlying cause

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Bowel findings uncommon overall in primary mesenteric adenitis & necessitate further investigation
- Appendix must be normal

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Mesenteric Lymphatic Malformation

KEY FACTS

TERMINOLOGY

- Subtype of congenital slow flow vascular malformation due to error of lymphatic vessel formation
- Results in well-defined, cyst-like (macrocystic) &/or infiltrative, solid-appearing (microcystic) mass of abnormal lymphatic channels focally or diffusely within mesentery
  - Individual cyst size: Macrocyst > 1 cm, microcyst < 1 cm

IMAGING

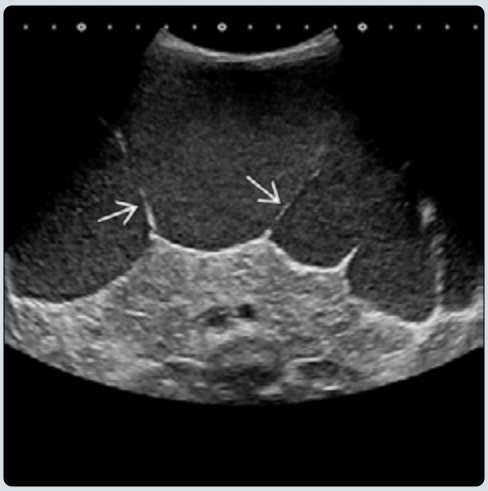
- Macrocystic: Well-defined, lobulated, unilocular or multilocular fluid-filled mass in mesentery
  - Thin septations with minimal enhancement (unless complicated by hemorrhage or infection)
  - Simple vs. complex fluid varies between cystic components
    - ± layering fluid-fluid levels (due to blood products) & fat/chylous fluid; Ca^{2+} uncommon
- Microcystic: More solid appearing, infiltrative
- Best imaging tool: Ultrasound or MR
  - More sensitive than CT for characteristic thin septations & fluid-debris/fluid-fluid levels

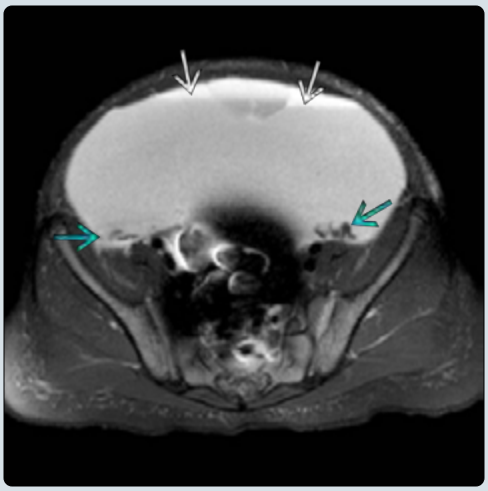
CLINICAL ISSUES

- Generally grow commensurate with patient
  - May rapidly enlarge due to hemorrhage/trauma, infection, or hormonal stimulation (puberty, pregnancy)
- Complications include bowel obstruction, volvulus, infection, hemorrhage
- Most present clinically under age 2 years
  - Abdominal pain, distension, vomiting, palpable mass
  - Commonly asymptomatic
- Treatments include
  - Simple excision, partial bowel resection, sclerotherapy
  - Sirolimus emerging as possible medical therapy

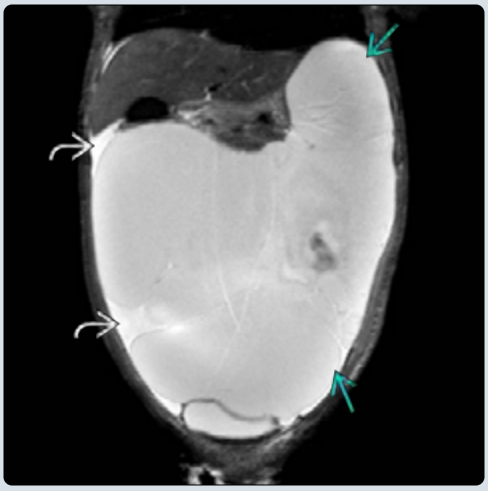
DIAGNOSTIC CHECKLIST

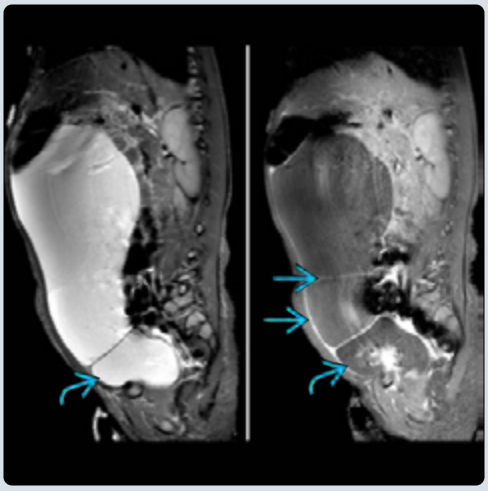
- Verify cystic mass is not actually due to distended hollow organ (such as urinary bladder)





(Left) *Transverse ultrasound through the abdomen in a 2-year-old patient with abdominal distention shows a large hypoechoic mass occupying the anterior peritoneal cavity & displacing bowel posteriorly. Thin septations & swirling granular debris were seen in the mass.* **(Right)** *Axial T2 FS MR in the same patient shows fluid-fluid levels & dependently layering debris in the mass due to prior internal hemorrhage.*





(Left) *Coronal T2 FS MR in the same patient shows that this macrocystic lymphatic malformation occupies the majority of the abdomen & pelvis. Additional free fluid outlines the thin margins of the mass.* **(Right)** *Sagittal STIR (left) & T1 C+ FS (right) MR in the same patient shows only mild rim & septal enhancement of this massive mesenteric lymphatic malformation. Note the distortion of the urinary bladder by the mass.*

TERMINOLOGY

Synonyms

- Mesenteric or omental cyst; lymphangioma (antiquated)

Definitions

- Subtype of congenital slow flow vascular malformation due to error of lymphatic vessel formation
- Results in well-defined cyst-like (macrocystic) &/or infiltrative, solid-appearing (microcystic) mass of abnormal lymphatic channels focally or diffusely within mesentery
  - Individual cyst size: Macrocyst > 1 cm, microcyst < 1 cm
- Mass lacks communication with normal lymphatics

IMAGING

General Features

- Best diagnostic clue
  - Multicystic mass in small bowel mesentery
- Location
  - Lymphatic malformations (LM) overall
    - Vast majority involve neck, axilla, chest wall
    - Mediastinum, omentum, mesentery, retroperitoneum, & extremities less common
      - Mesenteric (< 1% of all LM)
- Morphology
  - Unilocular vs. multilocular
  - Well defined, lobular vs. poorly defined/infiltrative

CT Findings

- Fluid-filled mass ± septations
  - Attenuation depends on composition of cyst (fat/chyle vs. serous fluid vs. hemorrhage); may appear solid

MR Findings

- Unilocular or multiseptated cystic mass
  - Largely of fluid signal intensity
    - Layering fluid-fluid levels often present (due to blood products, typical of slow flow vascular malformations)
  - Components of T1 shortening may be due to hemorrhage or fat
  - Minimal rim/septal enhancement (unless complicated)

Ultrasonographic Findings

- Grayscale ultrasound
  - Mostly anechoic mass with thin septations
  - Can be complex with varying degrees of debris or hemorrhage within different components
    - Debris may be uniform or layer dependently
    - Contents swirl with intermittent compression
- Color Doppler
  - No significant internal vascularity, though LM frequently encase normal vessels

Imaging Recommendations

- Best imaging tool
  - Ultrasound or MR
    - More sensitive than CT for characteristic thin septations & fluid-debris/fluid-fluid levels
- Protocol advice
  - Subtracted (pre- from postcontrast) FS T1 MR images provide clearest assessment of enhancing components

DIFFERENTIAL DIAGNOSIS

Gastrointestinal Duplication Cyst

- Unilocular cyst with sonographic gut signature
  - 5-layer appearance more specific than 3 layer
- ± peristalsis (pathognomonic if seen)

Ovarian Cystic Mass

- Simple cyst (most common abdominal cystic mass of female fetuses) vs. complicated cyst (hemorrhage or infection) vs. cystic neoplasm

Distended/Obstructed Viscus

- Large urinary bladder, chronically obstructed vagina (hydrocolpos), chronic focal dilation of bowel

Other Cysts/Pseudocysts

- Ventriculoperitoneal shunt pseudocyst, hepatic cyst, choledochal cyst, meconium pseudocyst, loculated ascites, peritoneal inclusion cyst, abscess, hematoma

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal pain, distension, vomiting, palpable mass
  - Commonly asymptomatic

Demographics

- Age: Most present clinically under 2 yr

Natural History & Prognosis

- Generally grow commensurate with patient
  - May rapidly enlarge due to hemorrhage/trauma, infection, or hormonal stimulation (puberty, pregnancy)
- Complications
  - Bowel obstruction, volvulus, infection, hemorrhage

Treatment

- Simple excision, partial bowel resection
  - Recurrence up to 100% with incomplete resection
- Sclerotherapy (commonly used in other sites of macrocystic LM) gaining popularity for abdominal LM
  - Limited use previously due to access difficulties & concerns for peritoneal spillage of sclerosing agents
- Sirolimus emerging as possible medical therapy in LM

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Omental Infarction

KEY FACTS

TERMINOLOGY

- Benign self-limited cause of acute abdominal pain due to vascular occlusion → segmental omental infarction

IMAGING

- General features
  - Well-demarcated triangular/wedge-shaped or oval focus of ↑ attenuation/echogenicity within omental fat
  - Right mid to upper abdomen, deep to anterior wall
  - ± thickening of overlying peritoneal membrane
  - ± free intraperitoneal fluid &/or pleural effusion
  - No adjacent inflammatory etiologies (e.g., appendicitis)
- US: Echogenic focus with no internal color Doppler flow; painful to direct sonographic palpation
- CT: Focal fat stranding ± hyperattenuating streaky densities due to fibrous bands or thrombosed vessels
  - ± peripheral enhancement
  - ± whirl or vascular pedicle sign with torsion
- Recommendations
  - Ultrasound: Acceptable 1st-line modality despite lower sensitivity for this diagnosis compared to CECT
    - ↑ sensitivity for surgical diagnoses (e.g., appendicitis)

TOP DIFFERENTIAL DIAGNOSES

- Epiploic appendagitis, appendicitis, slow flow vascular malformation, mesenteric contusion, fat-containing masses

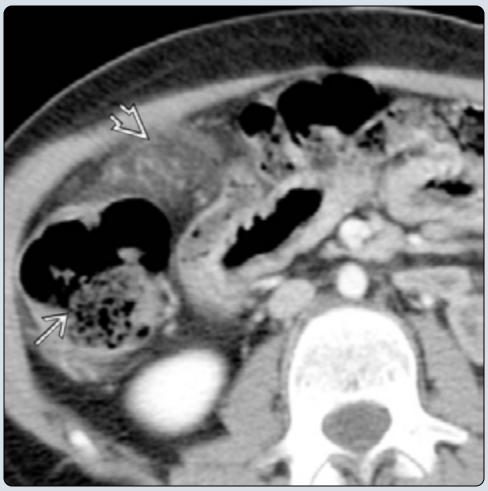
PATHOLOGY

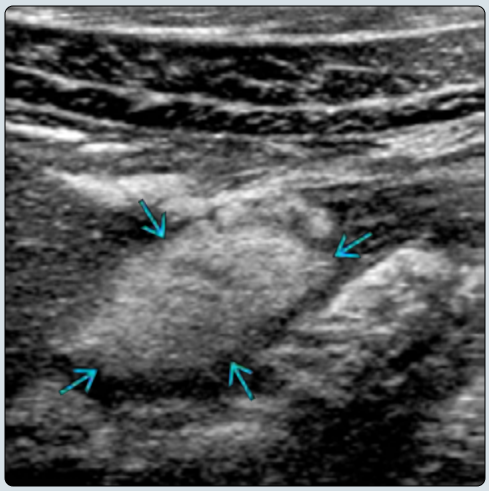
- Etiologies: Anatomic vascular variation, hypercoagulability, torsion, systemic states prone to vascular congestion
- Majority of patients obese

CLINICAL ISSUES

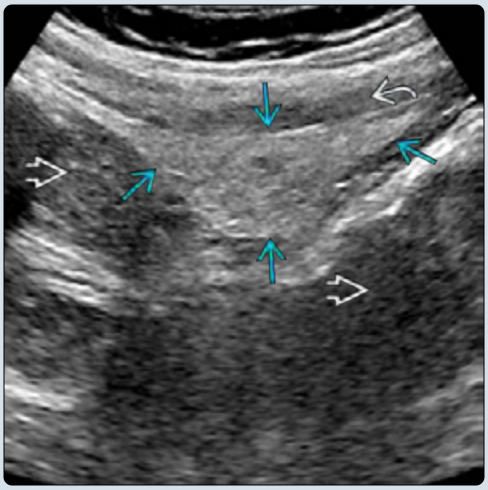
- Abdominal pain & tenderness, nausea, vomiting, fever
- Conservative treatment typically adequate
- Occasional surgical excision if pain intractable, symptoms worsen, or imaging equivocal

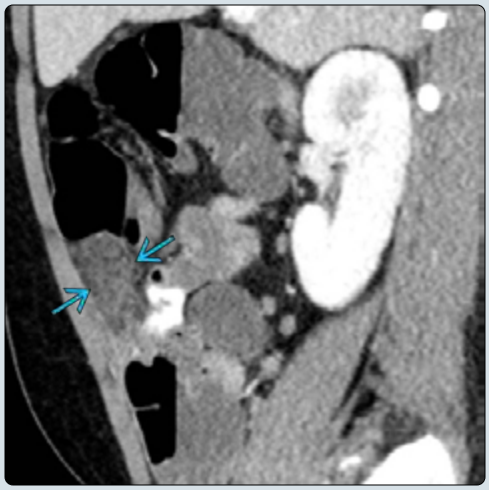
(Left) *Axial CECT in an 11 year old with right abdominal pain depicts a focus of inflammatory stranding ⇨ immediately deep to the abdominal wall & anterior to the ascending colon ⇨, typical of an omental infarct.* **(Right)** *Transverse ultrasound of a 7 year old with abdominal pain shows a well-demarcated, ovoid, hyperechoic focus ⇨ deep to the right anterior abdominal wall. A normal appendix was not visualized. An omental infarct was confirmed at surgery.*





(Left) *Longitudinal ultrasound from a 9 year old with right abdominal pain & fever depicts a hyperechoic focus ⇨ in the omentum between the ascending colon ⇨ & abdominal wall ⇨, consistent with an omental infarct.* **(Right)** *Right sagittal CECT in the same patient shows a corresponding focus of omental hyperattenuation ⇨ just deep to the anterior abdominal wall, consistent with an omental infarction.*





TERMINOLOGY

Definitions

- Benign self-limited cause of acute abdominal pain due to vascular occlusion leading to segmental omental infarction

IMAGING

General Features

- Best diagnostic clue
  - Focal, well-demarcated, triangular/wedge-shaped/oval focus of omental fat stranding (CT)/hyperechogenicity (US) without other sources of inflammation
    - Typically in right upper quadrant (RUQ)

Ultrasonographic Findings

- Relatively hyperechoic, noncompressible, triangular/wedge-shaped or oval focus
- Typically RUQ immediately deep to anterior abdominal wall
- No internal color Doppler flow
- Painful to direct sonographic palpation
- ± free intraperitoneal fluid &/or pleural effusion

CT Findings

- Well-defined focus of hazy ↑ attenuation of omental fat
- Triangular/wedge-shaped or oval
- Typically RUQ between anterior abdominal wall & transverse or ascending colon
- ± streaky/linear densities representing fibrous bands or thrombosed vessels
- ± peripheral enhancement
- ± whirl or vascular pedicle sign: Torsion of omental fat & vessels around engorged vascular pedicle
- ± thickening of overlying peritoneum
- ± free intraperitoneal fluid &/or pleural effusion
- No other source for inflammation (e.g., appendicitis, diverticulitis, colitis, etc.)

Imaging Recommendations

- Ultrasound typically 1st-line modality
  - Low sensitivity (60-80%) for omental infarction but higher for other diagnoses with similar presentations
- CECT has greater sensitivity (90%) for omental infarction

DIFFERENTIAL DIAGNOSIS

Epiploic Appendagitis

- Torsion & resultant inflammation of epiploic appendage along colon, typically in lower quadrants
- Ovoid focus of pericolonic fat inflammation
- Hyperdense central dot sign (thrombosed vessel) classic

Appendicitis

- Enlarged, noncompressible appendix with adjacent inflammatory change

Mesenteric Lymphadenitis

- Cluster of ≥ 3 lymph nodes ≥ 5 mm each (in short axis)
- ± adjacent fat stranding

Mesenteric Contusion

- Typically preceded by clear history of trauma
- ± overlying contusion of subcutaneous fat

Slow Flow Vascular Malformation

- ± poorly defined infiltrative components in venous or lymphatic malformations, typically in conjunction with mass-like abnormal vascular channels/cysts

Meckel Diverticulitis

- Noncompressible blind-ending tubular structure arising from distal ileum with adjacent inflammatory change

Fat-Containing Masses

- Teratoma, lipoblastoma uncommon in anterior abdomen in children; more solid & heterogeneous in appearance

PATHOLOGY

General Features

- Due to anatomic vascular variation, hypercoagulability, torsion (due to scars, hernias, omental cysts), systemic states prone to vascular congestion (right heart failure)

Gross Pathologic & Surgical Features

- Venous stasis &/or thrombosis → venous congestion → omental edema & congestion (early infarct) → hemorrhagic necrosis/infarction → granulomatous infiltrate (6-7 days) → fibrosis & scar

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal pain & tenderness, typically right-sided
- Other signs/symptoms
  - Nausea, vomiting, mild fever, ↑ CRP & WBC

Demographics

- Epidemiology
  - 15% of all cases occur in children
  - M:F = ~ 2-4:1
  - Majority of patients obese

Treatment

- Conservative treatment, including analgesia & antibiotics
  - Complete resolution < 2 weeks
- Occasional surgical excision if pain intractable, symptoms worsen, &/or imaging equivocal

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KEY FACTS

TERMINOLOGY

- Shock bowel

IMAGING

- Diffuse wall thickening & ↑ enhancement of bowel, ± fluid distension
- ↑ enhancement but ↓ caliber of vessels
- Abnormal solid organ appearances may include
  - Adrenal glands: Intense & prolonged enhancement
  - Kidneys: Typically intense & prolonged cortical enhancement; rarely ↓ enhancement < 10 HU (poor prognosis)
  - Pancreas: Diffusely enlarged with reticulated or feathery appearance due to ↑ parenchymal enhancement with edematous intervening septa
  - Liver: Heterogeneous enhancement, periportal edema; gallbladder mucosal enhancement & wall edema
  - Spleen: ↓ enhancement (> 30 HU < liver may be associated with worse prognosis)

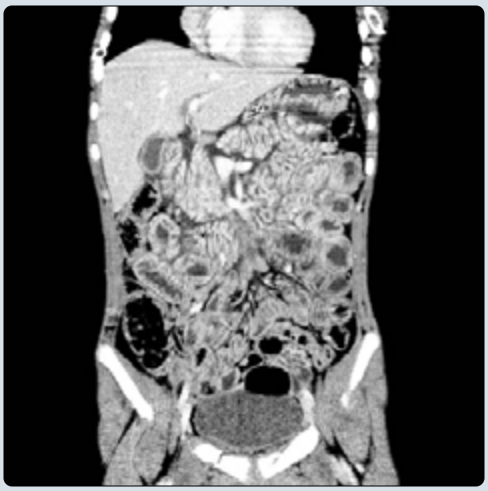
PATHOLOGY

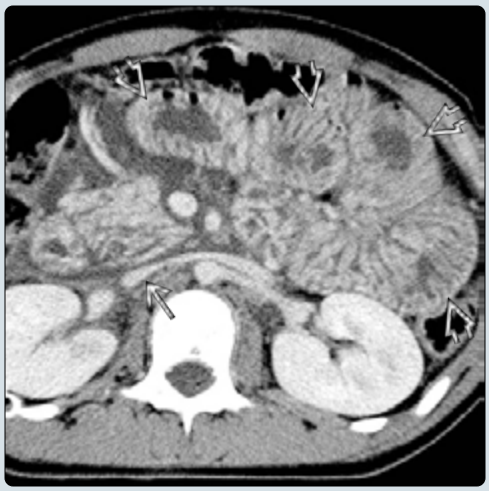
- Shock: Compensated → decompensated → irreversible
- Sympathetic nervous, cardiovascular, & neuroendocrine systems help maintain arterial pressure & cardiac output in compensated state
- Pediatric patients can maintain normal/near normal blood pressure longer than cardiac output → rapid, abrupt progression to decompensated state

CLINICAL ISSUES

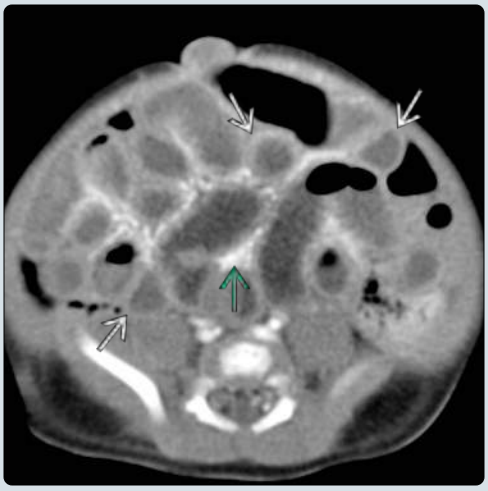
- CT findings often precede clinical recognition of shock
  - In one series, hypotension developed within 10 minutes of CT in 19% of children
  - In same series, 85% mortality rate with presence of hypoperfusion complex (compared with 2% of all children who suffered blunt trauma)
- Requires prompt notification & transfer to clinical service for intense monitoring, volume replacement, supportive measures, & surgery if needed

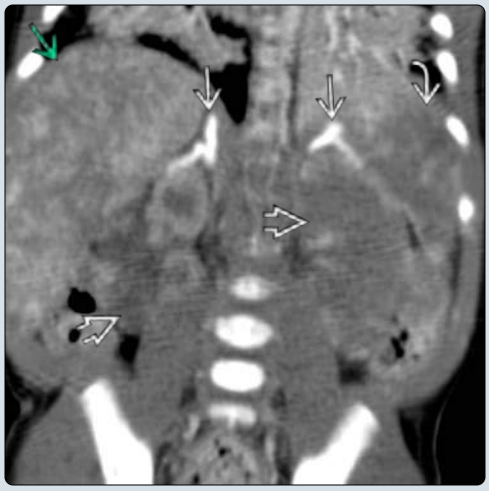
(Left) *Coronal CECT shows findings of shock in a child after an MVA with aortic trauma & a urinary bladder injury (not shown). Note the multiple dilated, fluid-filled loops of small bowel with diffuse wall thickening & increased wall enhancement.* **(Right)** *Axial CECT in the same patient shows multiple loops of fluid-filled small bowel with abnormal feathery wall thickening & increased enhancement. Also note the small, flattened inferior vena cava. These features are typical of the hypoperfusion complex.*





(Left) *Axial CECT of a 2-month-old boy with cardiogenic shock shows diffusely dilated & fluid-filled small bowel with mild wall thickening. There is intense enhancement of the mesenteric vessels.* **(Right)** *Coronal CECT from the same patient demonstrates intensely enhancing adrenal glands, a characteristic finding in hypoperfusion complex. There is patchy, decreased enhancement of the liver, kidneys, & spleen.*





TERMINOLOGY

Abbreviations

- Hypoperfusion complex (HC)

Synonyms

- Shock bowel
- Shock abdomen
- Hypovolemic shock complex
- Hypotension complex

Definitions

- Constellation of CT findings indicative of compensated shock

IMAGING

CT Findings

- Best imaging clues (on CECT)
  - Diffuse small bowel wall thickening with ↑ enhancement
  - Abnormal solid organ enhancement (typically ↑)
  - ↑ enhancement of small caliber vessels
- Bowel (predominantly small intestine)
  - Diffuse mild dilation of fluid-filled loops
  - Diffuse mucosal hyperenhancement (> psoas muscle)
  - Diffuse wall thickening > 3 mm
- Vasculature
  - Flat pancake inferior vena cava (IVC) with circumferential low-attenuation (< 20 HU) fluid halo
    - ± intense enhancement
    - May show normal caliber & shape with aggressive fluid resuscitation
  - ↓ caliber, intensely enhancing aorta, & mesenteric arteries
- Solid viscera
  - Adrenal glands
    - Symmetric, intense, prolonged enhancement
  - Kidneys
    - Typically symmetric, intense, prolonged enhancement of cortex
    - ↓ enhancement (< 10 HU) rarely seen; may represent poor prognosis (black kidney sign)
  - Pancreas
    - Diffusely enlarged, feathery appearance due to edematous intervening septa
  - Spleen
    - ↓ enhancement > 30 HU < liver
    - ↑ enhancement compared to liver also reported in children
  - Liver
    - Heterogeneous with periportal edema
    - Gallbladder mucosal enhancement & wall thickening/surrounding edema
- Generalized mesenteric edema & unexplained ascites
- Thoracic structures
  - ↓ caliber, intensely enhancing aorta & great vessels
  - ↓ caliber superior vena cava & IVC
  - ↓ cardiac chamber volume
  - Thyroidal & perithyroidal edema (shock thyroid)
    - Enlargement with intervening septal edema

Ultrasonographic Findings

- FAST scanning of abdomen for potential injury screening
  - May show peritoneal fluid
  - Diffusely fluid-filled & dilated bowel with wall thickening
  - ↓ caliber of IVC, aorta

Imaging Recommendations

- Best imaging tool
  - CECT

DIFFERENTIAL DIAGNOSIS

Bowel Trauma

- Focal (rather than diffuse) bowel wall thickening, dilation, & enhancement abnormalities (↑ or ↓)
- Adjacent vessel extravasation, mesenteric fluid, extraluminal gas
- Extensive extraintestinal findings favor shock bowel over focal bowel injury (though these entities may coexist)

Transient Small Bowel Intussusception

- Short segment(s) of small bowel showing alternating rings of high & low attenuation (in cross section)
- Typically incidental & self-limited finding
- Seen with higher frequency in trauma patients

Henoch-Schönlein Purpura

- Small vessel vasculitis of unknown etiology
  - Purpuric rash, abdominal pain, arthritis, nephritis, intussusceptions
  - Typical ages of 3-7 yr; boys > girls
- Bowel wall thickening with abnormal attenuation/enhancement due to intramural edema &/or hemorrhage

Graft-vs.-Host Disease

- Bone marrow transplant patient with new skin, liver, & gut abnormalities
- Hyperenhancing but featureless (ribbon-like) small bowel

Other Bowel Inflammatory Processes

- Preexisting bowel conditions may rarely cause hypovolemic shock or may rarely occur coincidental with trauma
  - Pseudomembranous colitis: Pancolitis with severe bowel wall thickening but disproportionately minor pericolonic inflammatory change; related to antibiotic use
  - Crohn disease: Marked thickening & abnormal enhancement of terminal ileum & cecum with surrounding inflammation
  - Infectious enterocolitis: *Shigella*, *E. coli*, etc., show abnormal bowel wall thickening, dilation, & wall enhancement

Bowel Ischemia Due to Vascular Occlusion

- Uncommon in children
  - Iatrogenic (i.e., neonatal vascular catheter), underlying cardiovascular disease, midgut volvulus
- May present with bowel wall thickening, diffuse dilation
- Abnormal enhancement: ↓ in arterial occlusion, ↑ in venous occlusion
- Extensive extraintestinal findings favor shock bowel over primary bowel pathology

PATHOLOGY

General Features

- Shock: Clinical expression of limited oxygen supply/utilization
  - Can result from hypovolemia, head/spine injury, cardiac arrest, septicemia, diabetic ketoacidosis
- Stages of shock
  - Compensated
    - Perfusion to vital organs maintained by response of sympathetic nervous, cardiovascular, & neuroendocrine systems
    - Children may maintain arterial pressure longer than cardiac output, thus appearing stable enough for CT
    - May abruptly transition to decompensated stage
  - Decompensated
    - Response of sympathetic nervous, cardiovascular, & neuroendocrine systems no longer sufficient to maintain perfusion
  - Irreversible
    - Volume replacement unable to reverse depressed cardiac output & hypoperfusion
- Variable organ enhancement results from selective vasoconstriction mediated by ↑ sympathetic tone & renin-angiotensin axis
  - Small caliber aorta & IVC related to vasospasm, hypovolemia
  - Relatively intense & prolonged enhancement of adrenal glands related to central role of adrenal in generating sympathetic response to hypovolemic shock
  - Hyperintense, prolonged enhancement of kidneys related to vasoconstriction of renal efferent arterioles
  - Splenic arterial flow without autoregulatory mechanism; sensitive to sympathetic response, which → vasoconstriction & decreased perfusion
  - Splanchnic vasoconstriction reduces bowel perfusion
    - Altered, ↑ permeability of intestinal mucosa → interstitial leak of fluid & contrast → mucosal enhancement, submucosal edematous wall thickening
    - ↓ fluid reabsorption & ileus may result in fluid-distended lumen
- Associated abnormalities
  - Findings of solid organ injury: Liver, spleen, kidney
    - HC more frequent with ↑ grades of injury, active extravasation
  - Focal bowel injury may also be present
  - In one review, 48% had abdominal organ injury

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pediatric patients in tenuous hemodynamic state
    - Significant hemorrhage after blunt trauma
    - Less common: Neurogenic shock, septic shock, & cardiac arrest
  - Tachycardic
  - Normotensive before decompensation
  - Signs/symptoms of concomitant/causative intracranial & intraabdominal injuries
    - Low GCS: < 6 in 20%

Demographics

- Age
  - CT findings of shock more common & more striking in young children (but can be seen at any age)

Natural History & Prognosis

- HC: Poor prognosis in trauma patient
  - CT findings often precede clinical recognition of shock
    - Pediatric patients able to maintain arterial pressure at or near normal levels longer than cardiac output after severe hemorrhage → stable-appearing patients can rapidly decompensate
      - In one series, progressive hypotension developed within 10 minutes of CT in 19% of children
      - In same series, 85% mortality rate with presence of hypoperfusion complex (compared with 2% of all children who suffered blunt trauma)
- Additional findings associated with worse prognosis
  - Enhancement of kidney <10 HU (black kidney sign)
  - Lower splenic enhancement at initial imaging

Treatment

- Fluid/blood volume replacement
- Intense monitoring
- Surgery when necessary for underlying injuries

DIAGNOSTIC CHECKLIST

Consider

- Diffuse small bowel wall thickening with ↑ enhancement & luminal dilation: Suspect hypoperfusion complex & look for supporting signs
- Requires prompt notification of clinical team & transfer of patient from CT scanner to intensive care unit or operating room

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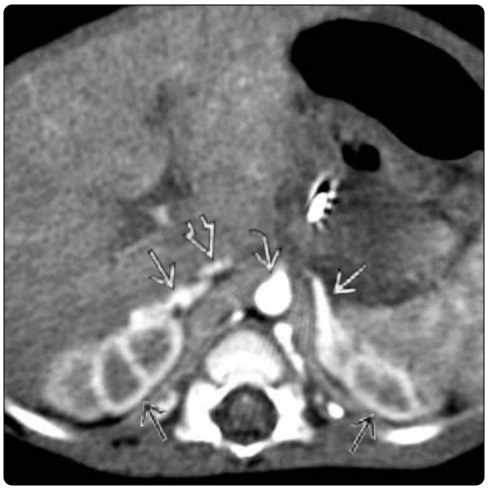
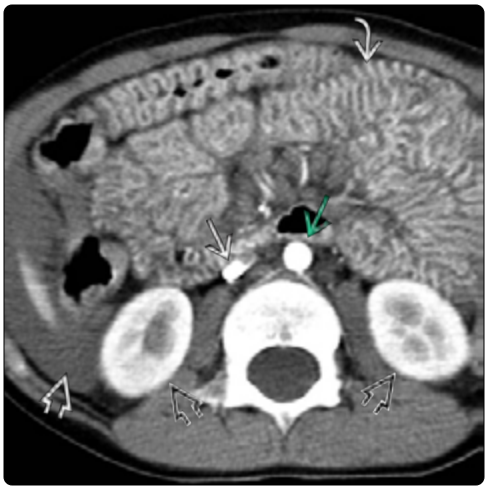
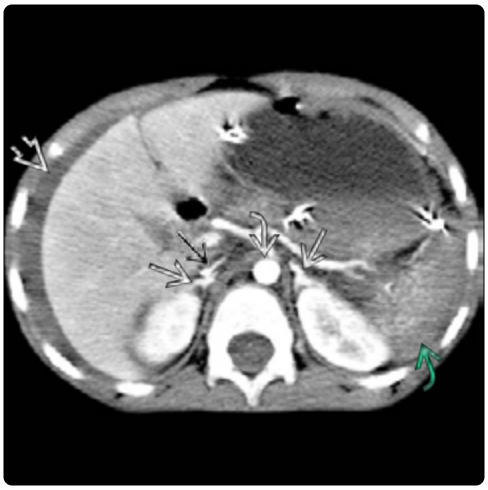
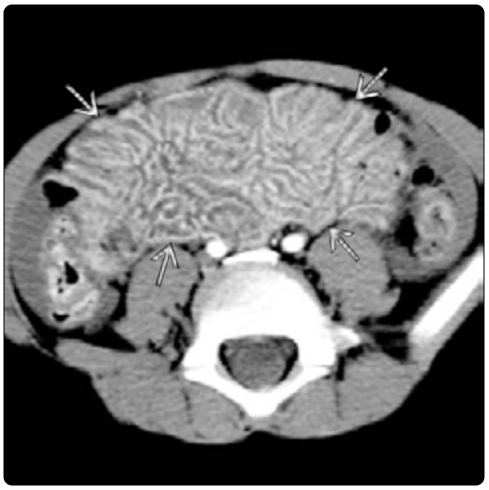


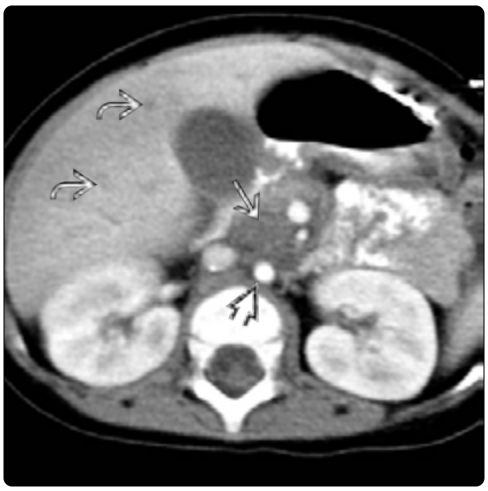


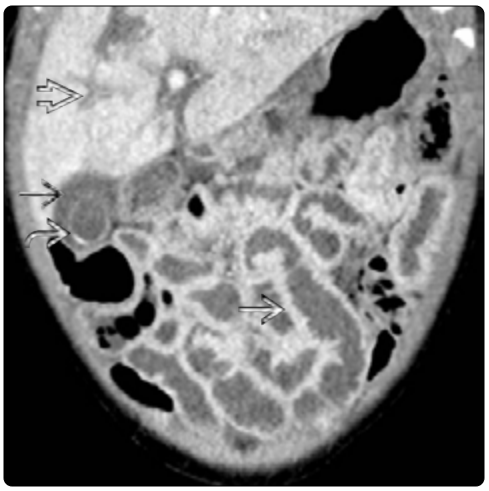
(Left) *Axial CECT shows a small & densely enhancing IVC. There is dense enhancement of the aorta, bilateral adrenal glands, & renal cortex.* **(Right)** *Axial CECT in a hypotensive patient shows diffuse feathery small bowel wall thickening & hyperenhancement, densely enhancing IVC & aorta, dense renal cortical enhancement, & ascites, all typical findings of hypoperfusion complex.*





(Left) *Axial CECT shows hyperenhancement of the adrenal glands & aorta. There is a dense & flattened appearance of the IVC. Note the free fluid in the peritoneal cavity & the poor enhancement of the spleen.* **(Right)** *Axial CECT in the same patient shows diffusely increased small bowel wall enhancement, typical of hypoperfusion complex.*





(Left) *Axial CECT in a 15-month-old girl in septic shock demonstrates edema at the root of the mesentery & hepatic periportal edema. These are nonspecific findings but can be seen in the setting of hypoperfusion complex. The aorta is also intensely enhancing.* **(Right)** *Coronal CECT from a 12-month-old boy nonaccidental trauma victim status post cardiac arrest shows diffuse fluid-distention of small bowel with wall hyperenhancement. Also seen are periportal edema + gallbladder wall enhancement & edema.*

Bowel Injury

KEY FACTS

IMAGING

- Best clue: Bowel wall discontinuity + extraluminal enteric contents + free intraperitoneal air
- Nonspecific signs: Bowel wall thickening, abnormal bowel wall enhancement, mesenteric fluid/stranding
- Sentinel clot sign: Localized mesenteric hematoma adjacent to bowel (> 70 HU)
- In acute blunt trauma setting, oral contrast does not significantly improve sensitivity & may result in inappropriate delay of diagnosis & treatment

TOP DIFFERENTIAL DIAGNOSES

- Hypoperfusion complex (shock bowel)
- Henoch-Schönlein purpura (HSP)
- Inflammatory bowel disease

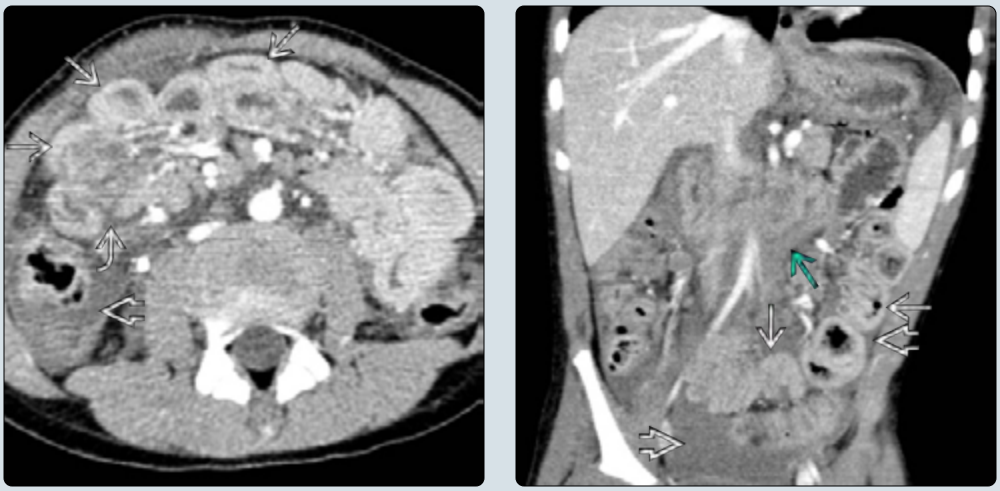
PATHOLOGY

- Blunt trauma 77%: Motor vehicle-related, nonaccidental trauma, & bicycle-related most common
  - Mechanism
    - Sudden ↑ in intraluminal pressure → perforation
    - Crush injury between abdominal wall & spine
    - Shear force between fixed & mobile bowel
  - Associated injuries: Spine (including Chance fracture) (9%), traumatic brain injury (22%), liver (16%), spleen (11%), pancreas (10%), kidney (6%)
- Penetrating trauma 23%
  - With rectal involvement → consider sexual assault

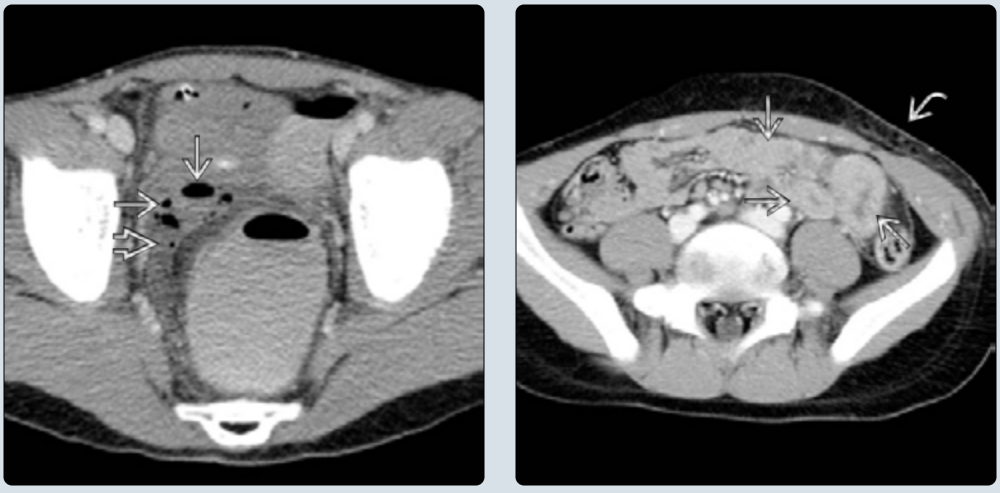
CLINICAL ISSUES

- Signs & symptoms
  - Abdominal pain, peritoneal signs, tachycardia, ↑ WBC
  - Bowel injury in 11% of patients with seat belt sign: Relative risk of 9.4
- Treatment
  - Perforations: Surgical management
  - Contusion/hematoma: Typically nonoperative
    - Clinical deterioration main indicator for surgery

(Left) *Axial CECT in a 7 year old shows diffuse small bowel wall thickening, mesenteric hematoma, & right paracolic gutter fluid of increased density. Small bowel devascularization & perforation of the distal ileum were found at surgery.* **(Right)** *Coronal CECT in a 2-year-old victim of nonaccidental trauma shows thickened bowel loops, regions of increased & decreased bowel wall enhancement, & extensive free fluid. Devascularized jejunum was found at surgery.*



(Left) *Axial CECT with rectal contrast in an 11-year-old boy whose rectum was impaled on a metal stake shows gas & fat stranding in the perirectal tissues. Extraperitoneal perforation of the rectum was demonstrated at surgery.* **(Right)** *Axial CECT in a 12-year-old motor vehicle accident victim shows thick-walled small bowel in the left abdomen. There is edema of the overlying subcutaneous fat with skin thickening, typical of a seat belt sign. Jejunal perforations were found at surgery.*



TERMINOLOGY

Synonyms

- Contusion, hematoma; perforation, laceration, tear, enterotomy, transection

IMAGING

General Features

- Best diagnostic clue
  - CECT: Focal bowel wall thickening with discontinuous bowel wall enhancement, extraluminal gas, & surrounding mesenteric fluid/stranding in trauma setting
- Location
  - Jejunum/ileum 45%, duodenum 27%, colon 37%, stomach 10%

Radiographic Findings

- Dilated bowel loops, focal or generalized
  - Inflammation & ischemia may lead to ileus or obstruction
- Pneumoperitoneum (sensitivity < 30% in cases of bowel perforation due to blunt trauma)
  - Air under diaphragm: Best visualized on upright or left lateral decubitus views
  - ↑ volume of air required for visualization on supine views
    - Rigler sign: Air outlining both sides of bowel wall
    - Falciform ligament sign: Vertically oriented linear density in medial right upper quadrant outlined by air
    - Football sign: Ovoid lucency filling abdominal cavity
    - Inverted V sign: Air outlining medial umbilical folds
- Retroperitoneal air
  - Perforation of retroperitoneal bowel segments (duodenum; ascending or descending colon)
  - Outlines psoas, kidney, &/or hemidiaphragm crus
- Flank stripe sign: Peritoneal fluid separating ascending or descending colon from peritoneal reflection or properitoneal fat
- Dog ear sign: Pelvic fluid separating bowel from bladder
- Splinting: Scoliosis curvature concave toward injury

CT Findings

- CECT
  - Peritoneal fluid
    - Nonspecific, but must consider bowel injury with > trace volume of fluid in absence of solid organ injury or pelvic fracture
  - Bowel wall thickening of 1 or several loops
    - ≥ 3 mm or disproportionate to normal bowel wall
    - Seen in 75% of full-thickness lacerations
    - Intramural hematoma may show focal, mass-like eccentric thickening
  - Abnormal bowel wall enhancement
    - Psoas muscle used as reference
    - ↑ enhancement
      - Patchy & irregular with full-thickness laceration
      - Seen paradoxically with hypoperfusion due to damaged vascular endothelium
    - ↓ enhancement
      - Focal contusion/hematoma, traumatic ischemia, &/or mesenteric vascular injury
      - Discontinuity of enhancing bowel wall from laceration/perforation (specificity 100%, sensitivity 7%)
  - Extraluminal enteric contents
    - Specificity for bowel perforation ~ 100%, sensitivity 12%
    - Oral contrast does not ↑ sensitivity significantly in blunt trauma; may result in unnecessary delay
    - Oral & rectal contrast may help in stable patients with penetrating injury
  - Pneumoperitoneum
    - Angular/curvilinear gas in antidependent abdomen between anterior abdominal wall & liver surface
    - Small round foci of gas may be confined within mesenteric sheets adjacent to disrupted bowel wall
    - Lung windows helpful for detection
  - Mesenteric fluid, hematoma, &/or stranding
    - Often at mesenteric root; polygonal or V-shaped
    - Sentinel clot (> 70 HU) suggests adjacent bowel injury
  - Active vascular extravasation
    - Irregular, nonanatomic focus of ↑ attenuation (isodense to aorta) near vessel
  - Retroperitoneal gas
    - Perforation of duodenum, ascending/descending colon
  - Extraperitoneal gas
    - Rectal perforation

Ultrasonographic Findings

- FAST scan for free fluid in Morison pouch, splenorenal recess, pouch of Douglas, & paracolic gutters
  - Poor sensitivity of 35-44%; not specific for bowel injury
- Bowel wall thickening
- Intramural hematoma: Focal bowel wall thickening of varying echogenicity depending on age
- Pneumoperitoneum: Linear echogenic foci with "dirty" shadowing immediately deep to peritoneal lining

Imaging Recommendations

- Best imaging tool
  - CECT with multiplanar reformats

DIFFERENTIAL DIAGNOSIS

Hypoperfusion Complex (Shock Bowel)

- Diffusely fluid-distended bowel with wall thickening & ↑ enhancement
- ↑ enhancement of adrenals, pancreas, mesenteric vessels
- ↓ size of aorta & inferior vena cava

Henoch-Schönlein Purpura

- Focal or multifocal bowel wall thickening from hemorrhage
- Purpuric rash on legs & upper extremity extensor surfaces

Inflammatory Bowel Disease

- Colicky abdominal pain, recurrent diarrhea ± blood, weight loss, perianal disease, malabsorption
- Terminal ileal & cecal involvement most common
- Wall thickening, enhancement ± adjacent mesenteric stranding, vascular engorgement, fat proliferation

Coagulopathy

- Bleeding into bowel wall

Mimics of Free Intraperitoneal Air From Perforation

- Iatrogenic introduction of air: Diagnostic peritoneal lavage, peritoneal dialysis catheter, recent surgery
- Extension of air from other sites: Pneumomediastinum, pneumothorax, female genital injury, intraperitoneal bladder perforation
- Pseudopneumoperitoneum: Air between abdominal wall & parietal peritoneal layer

PATHOLOGY

General Features

- Etiology
  - Blunt trauma (77%)
    - Motor vehicle-related (46%), nonaccidental trauma (19%), bicycle-related (13%)
    - Intestinal injury in 1-15% of blunt trauma cases
  - Penetrating trauma (23%)
    - Gun shot (52%), stabbing (17%), impalement (30%)
    - Consider sexual assault with rectal trauma
  - Incidence of iatrogenic bowel injury
    - 0.1-7.0% of esophagogastroduodenoscopies, 0.09-3.0% of colonoscopies
- Associated abnormalities
  - Spine injury (including Chance fracture) (9%)
  - Traumatic brain injury (22%)
  - Liver (16%), spleen (11%), pancreas (10%), kidney (6%)
- Injury associated with rectal impalement
  - Anterior: Bladder, uterus, bowel
  - Posterior: Retroperitoneal & vascular structures
  - Perianal: Anus, vagina, urethra

Staging, Grading, & Classification

- American Association for the Surgery of Trauma injury scale
- Grades for stomach injury
  - I: Contusion/hematoma, partial-thickness laceration
  - II: Laceration < 2 cm in GE junction/pylorus, < 5 cm proximal 1/3 stomach, < 10 cm distal 2/3 stomach
  - III: Laceration > 2 cm in GE junction/pylorus, > 5 cm proximal 1/3 stomach, > 10 cm distal 2/3 stomach
  - IV: Tissue loss/devascularization < 2/3 stomach
  - V: Tissue loss/devascularization > 2/3 stomach
- Grades for jejunum, ileum, colon, & rectum injury
  - I: Contusion or hematoma without devascularization, partial-thickness laceration without perforation
  - II: Laceration < 50% circumference
  - III: Laceration ≥ 50% circumference without transection
  - IV: Transection (small bowel/colon), full-thickness laceration with extension into perineum (rectum)
  - V: Transection + segmental tissue loss, devascularization

Gross Pathologic & Surgical Features

- Compression of intestinal loops at impact → sudden ↑ in intraluminal pressure → perforation
- Crush injury between anterior abdominal wall & spine
- Rapid deceleration → shear force between fixed & mobile segments of bowel
- Poor fit of seat belt contributes to injury mechanisms
  - "Submarining" under lap belt or lap belt too high on abdomen ± "jackknifing" (hyperflexion) → compression + crush injuries
  - Higher center of gravity in children → shearing force

CLINICAL ISSUES

Presentation

- Abdominal pain, guarding, rigidity, absent bowel sounds, hypotension, tachycardia, elevated WBC
- Seat belt sign in 11%
- Delayed presentation of symptoms > 24 hours 6.5%

Natural History & Prognosis

- Overall mortality 5%, usually related to associated injuries
- In contrast to adults, delay in diagnosis & intervention up to 24 hours does not significantly affect prognosis
- Can be complicated by sepsis, peritonitis, abscess, stenosis/obstruction
- Risk of postoperative stenosis/obstruction, hernia, leak

Treatment

- Observation + serial examination if diagnosis in question
  - Clinical deterioration main indicator for intervention
- Contusion, hematoma typically nonoperative management
- Perforations managed surgically
  - Gastric: Primary repair (grade II, III); pyloroplasty/partial gastrectomy if not amenable to primary repair (grades IV, V)
  - Small bowel (jejunum, ileum): Primary repair ± debridement (grades I, II, some III); resection & anastomosis (grade IV, V, some III)
  - Colon: Primary repair ± colostomy
  - Rectum: Extraperitoneal → nonoperative; intraperitoneal → primary repair + diverting colostomy

DIAGNOSTIC CHECKLIST

Consider

- Unexplained free fluid with focally thickened bowel & surrounding mesenteric edema/hemorrhage: Bowel injury until proven otherwise
- Relative risk of bowel injury of 9.4 with lap-belt ecchymosis
- Closely analyze bowel in Chance fracture patients

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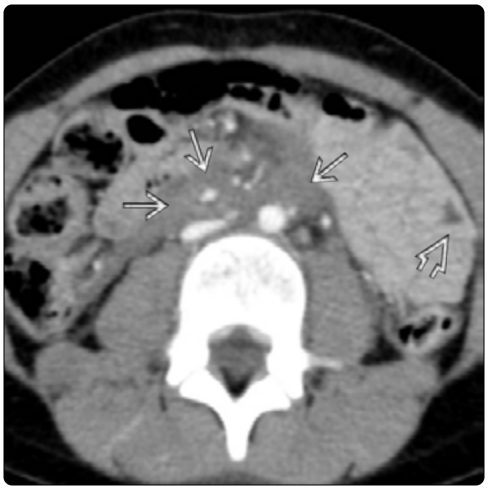
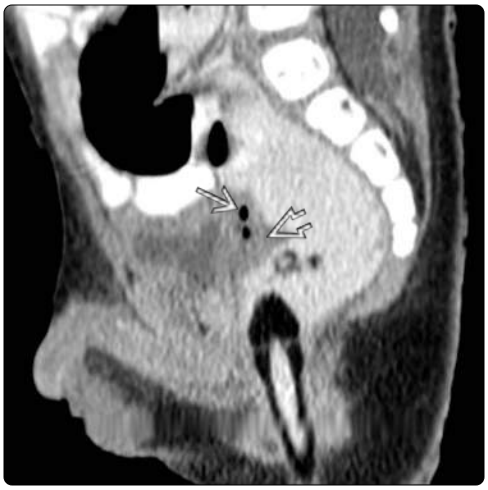
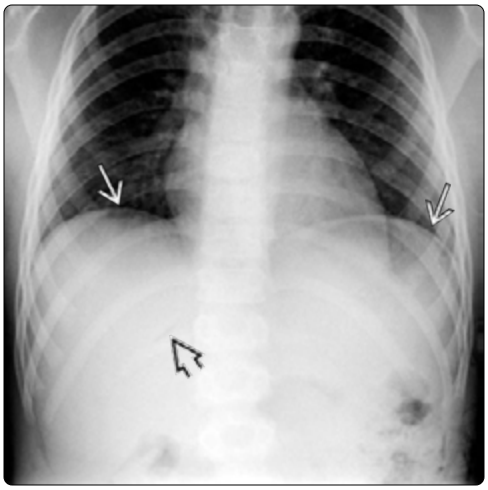
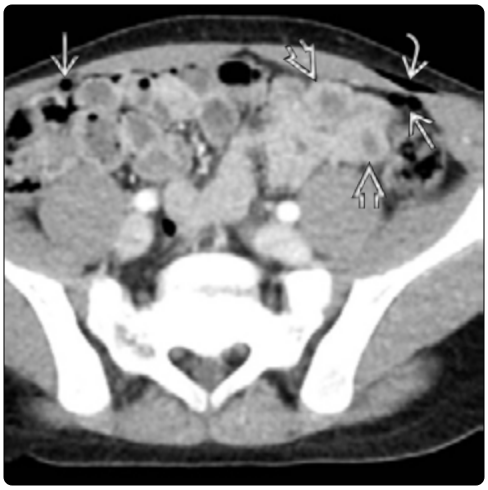


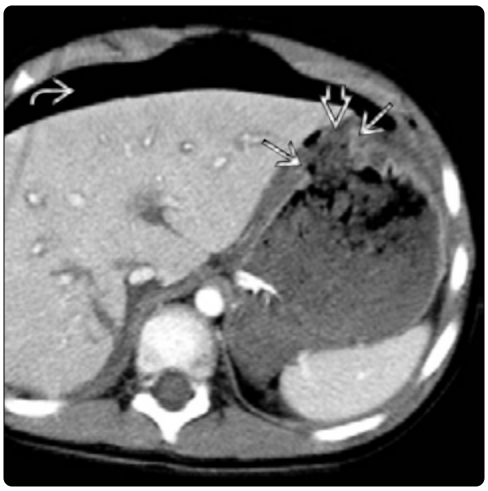


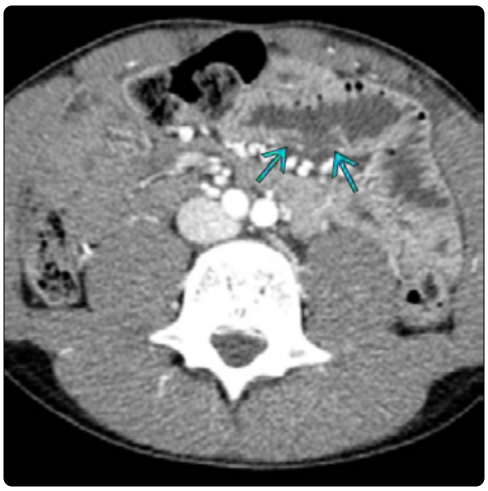
(Left) *Axial CECT from a 7-year-old with a seat belt sign shows high-attenuation fluid at the mesenteric root with foci of small bowel wall thickening. A mesenteric root hematoma & jejunal contusions were found at surgery.* **(Right)** *Sagittal CECT (with rectal contrast) from a 2 year old who was rectally impaled by a pencil shows extraluminal gas & high-attenuation fluid anterior to the rectum in the rectovesical pouch. An intraperitoneal rectal perforation was found at surgery.*





(Left) *Portable upright AP view of the chest in a 10-year-old boy shot with a pellet gun shows lucency under both hemidiaphragms & in Morrison pouch, consistent with free intraperitoneal air.* **(Right)** *Axial CECT from the same patient demonstrates foci of free intraperitoneal air & small bowel wall thickening. Subcutaneous gas was also seen in the abdominal wall near the site of penetration. Multiple jejunal perforations were discovered at surgery.*





(Left) *Axial CECT of a 4-year-old boy after a motor vehicle crash demonstrates anterior gastric wall disruption with extrusion of contents. There is a large volume of pneumoperitoneum.* **(Right)** *Axial CECT in a 12 year old with a bicycle handlebar injury shows decreased & discontinuous enhancement of the posterior wall of a fluid-filled jejunal loop with mild adjacent mesenteric fluid. A jejunal perforation was found intraoperatively.*

KEY FACTS

IMAGING

- Gold standard exam for blunt abdominal trauma: CECT
- Parenchymal laceration
  - Irregular, linear, branching foci of fluid attenuation
  - Posterior right hepatic lobe most commonly involved
  - Extension to bare area/posterior-superior region (segment VII) associated with retroperitoneal hematoma
  - Risk of bile duct injury with extension to porta hepatis
- Hematoma may be intraperitoneal, retroperitoneal, subcapsular, or intraparenchymal
  - Fluid attenuation if hematoma fresh & unclotted
  - ↑ attenuation may represent clot, implicate site of injury
  - ± hematocrit level of settling peritoneal blood in pelvis
    - ↑ density of dependent-most fluid component due to layering cellular blood elements
- Active hemorrhage or extravasation
  - Irregular, nonanatomic focus of high attenuation near vessel; accumulates on delayed images
    - Collection isodense to aorta with arterial injury
  - ± ↑ risk of delayed hemorrhage & nonoperative management failure
- Generalized periportal edema
  - Frequent finding of resuscitation due to distended intrahepatic lymphatics from vigorous hydration

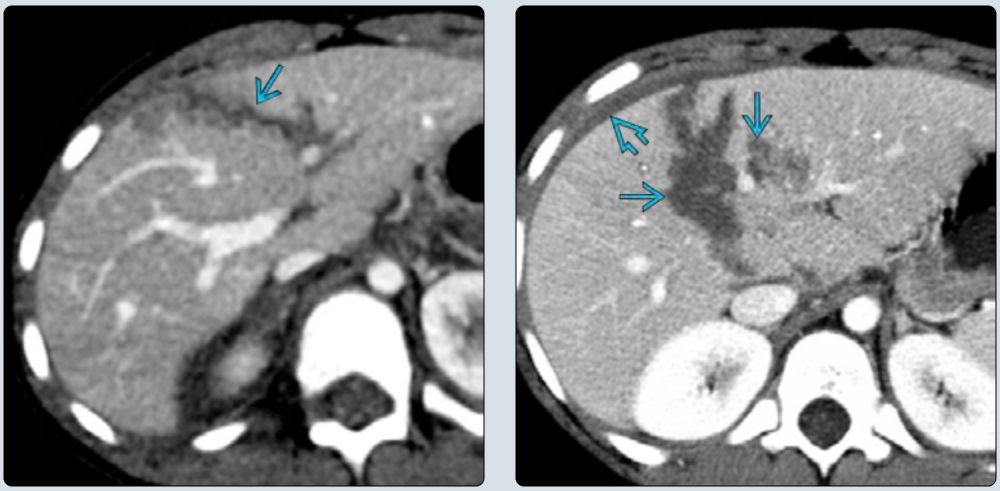
TOP DIFFERENTIAL DIAGNOSES

- Normal fissures
- Artifacts: Beam hardening or motion
- Hepatic abscess

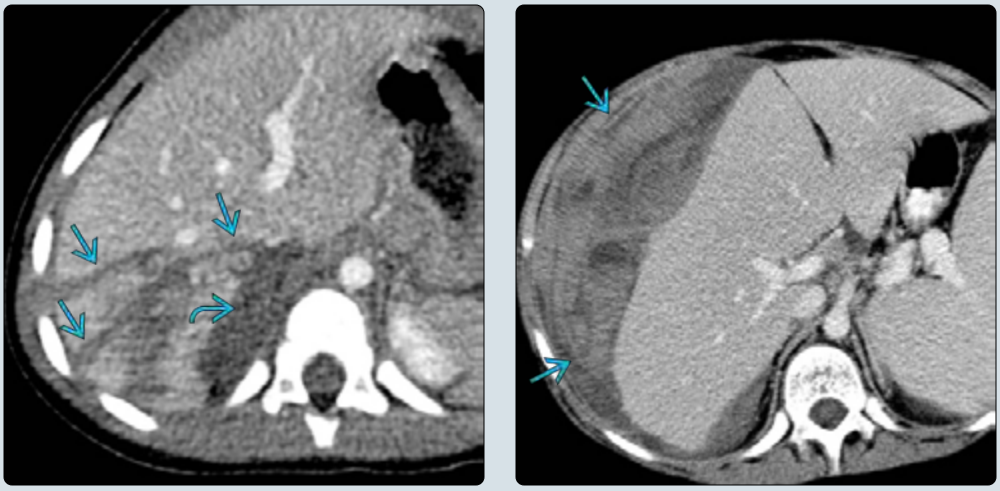
CLINICAL ISSUES

- American Association for the Surgery of Trauma (AAST) grading scale not predictive of need for surgery or outcome
- Nonoperative management of hemodynamically stable patients: > 90% successful
- Complications: Biliary injury, pseudoaneurysm, arteriovenous fistula, delayed hemorrhage, abscess

(Left) *Axial CECT image of a 6-year-old girl after a motor vehicle accident depicts a laceration ⇨ of the medial left hepatic lobe > 4 cm in depth, consistent with a grade III injury.* **(Right)** *Axial CECT in an 11 year old who sustained a handlebar injury to the upper abdomen demonstrates a hypodense laceration ⇨ > 3 cm in depth, consistent with a grade III injury. The laceration extends into both the right and left hepatic lobes. A small subcapsular hematoma is also present ⇨.*



(Left) *Axial CECT in a 2-year-old boy hit by a car shows branching hypodense lacerations ⇨ extending from the superior posterior aspect of the right hepatic lobe (bare area). This disrupted > 25% of the right lobe, consistent with a grade IV injury. There is associated retroperitoneal hemorrhage ⇨.* **(Right)** *Axial CECT in a 12-year-old boy 1 day after liver biopsy shows a mixed attenuation, > 50% surface area subcapsular hematoma ⇨, consistent with a grade III injury. Higher density in the hematoma represents clot.*



TERMINOLOGY

Synonyms

- Liver or hepatic laceration, fracture, or injury

IMAGING

CT Findings

- NECT
  - Intraperitoneal, retroperitoneal, subcapsular, or parenchymal hematoma
    - May be hyperattenuating to unenhanced liver
- CECT
  - Laceration
    - Irregular, linear, branching foci of nonenhancement in liver parenchyma
    - May parallel hepatic or portal veins
  - Hematoma
    - Round, crescentic, or poorly defined fluid collection
    - ↑ attenuation (40-70 HU) may represent clot & implicate site of injury
    - Subcapsular
      - Lentiform or crescentic peripheral collection
      - Compresses convex margin of parenchyma
    - Intraparenchymal
      - Round or irregular
    - Intraperitoneal
      - Poorly defined, tracking dependently in abdomen/pelvis vs. focal collections
      - ± hematocrit level of dependently settling peritoneal blood components in pelvis
    - Retroperitoneal
      - Surrounds inferior vena cava, right adrenal gland
      - Often with lacerations of bare area (segment VII)
  - Active hemorrhage or extravasation
    - Irregular, nonanatomic collection of high attenuation near vessel; accumulates on delayed images
      - Isodense to aorta if arterial injury
  - Pseudoaneurysm
    - Round focus of high attenuation (isodense to aorta)
    - May have diminished attenuation on delayed images
  - Arteriovenous fistula (AVF)
    - Early enhancement of involved hepatic/portal vein, simultaneous with artery
  - Biliary injury (biloma, leak): Delayed, simple-appearing fluid accumulations without clinical hematocrit drop
    - Often with lacerations extending to porta hepatis
  - Periportal edema: Generalized ↓ attenuation surrounding portal veins
    - Most commonly due to lymphatic engorgement from aggressive hydration rather than periportal blood

Ultrasonographic Findings

- Grayscale ultrasound
  - Limited utility for detecting hepatic injury in acute trauma setting
    - Abdominal US: 51% sensitivity, 95% specificity
    - Abdominal CEUS: 84% sensitivity, 99% specificity
    - FAST exam: 55% sensitivity, 83% specificity for intraabdominal injury
  - Echogenicity of hematoma/laceration varies with age
    - Anechoic (initially) → hyperechoic (24 hours: Fibrin & clot) → hypoechoic (2-3 days: Liquefaction of blood)
  - Pseudoaneurysm
    - Anechoic, cyst-like structure near vessel
    - Yin-yang sign on color Doppler
  - Arteriovenous fistula
    - Dilated hepatic artery
    - Arterialized portal/hepatic venous waveform
    - Surrounding tissue vibration
  - Biliary injury (biloma, leak)
    - Delayed appearance of intrahepatic/intraperitoneal anechoic collection ± septation
    - Continued accumulation without hematocrit drop

MR Findings

- T1WI, T2WI signal intensity varies with hematoma age
- MRCP can be useful in evaluation of biliary tree

Nuclear Medicine Findings

- Hepatobiliary scintigraphy
  - Bile leak: Accumulating extrabiliary collection of radiotracer around liver in peritoneal cavity
  - Biloma: Focal, contained extrabiliary radiotracer

Imaging Recommendations

- Best imaging tool: CECT

DIFFERENTIAL DIAGNOSIS

Normal Fissures

- Isolated thin, smooth, linear hypodense foci in characteristic locations for ligamentum venosum, falciform ligament

Artifacts

- Beam hardening: ↓ density streaks extending from ribs
- Excessive patient motion: Random throughout whole image ± adjacent images
- Both artifacts extend beyond liver

Hepatic Abscess

- Round or irregular discrete lesion(s) in ill patient
- Central fluid attenuation with thick, irregular rim & septal enhancement ± surrounding edema

Primary Tumor or Metastatic Disease

- Neoplasm may rupture or bleed, ± trauma

Liver Infarction

- Peripheral, wedge-shaped focus of nonenhancement
- Uncommon in children, occasionally results from trauma

PATHOLOGY

General Features

- Etiology
  - Blunt vs. penetrating trauma: 90% vs. 10%
    - Blunt trauma causes deceleration or shearing injury
      - Motor vehicle-related injuries most common: 69%
      - Falls & recreational accidents: 30%
      - Nonaccidental trauma (NAT): 1%
    - Hepatic injury seen in 25% of blunt trauma cases & 3% of NAT cases overall
- Associated abnormalities

- Concomitant splenic injury: 45%
- Pancreatic injury most common if liver injury ≥ grade IV

Staging, Grading, & Classification

- American Association for the Surgery of Trauma (AAST) grading scale for liver injury
  - Grade I
    - Hematoma: Subcapsular, < 10% surface area
    - Laceration: Capsular tear, < 1-cm parenchymal depth
  - Grade II
    - Hematoma: Subcapsular, 10-50% surface area; intraparenchymal, < 10 cm in diameter
    - Laceration: Capsular tear, 1- to 3-cm parenchymal depth, < 10 cm in length
  - Grade III
    - Hematoma: Subcapsular, > 50% surface area, expanding or ruptured subcapsular, or parenchymal hematoma; intraparenchymal, > 10 cm or expanding
    - Laceration: > 3-cm parenchymal depth
  - Grade IV
    - Laceration: Parenchymal disruption involving 25-75% of hepatic lobe or 1-3 Couinaud segments
  - Grade V
    - Laceration: Parenchymal disruption involving > 75% of hepatic lobe or > 3 Couinaud segments in 1 lobe
    - Vascular: Juxtahepatic venous injuries, i.e., retrohepatic vena cava/central major hepatic veins
  - Grade VI
    - Vascular: Hepatic avulsion

Gross Pathologic & Surgical Features

- Pattern of injury
  - Posterior segment of right lobe most common
    - Greatest volume of liver & surrounded by ribs, spine
    - Fixation by coronary ligaments enhance effect of acceleration-deceleration injury
  - Left lobe most commonly injured in NAT

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - RUQ pain, guarding, rebound tenderness, hypotension, lap-belt ecchymosis (seat-belt sign)

Natural History & Prognosis

- AAST grade not predictive of outcome or need for surgery
- Hemodynamic stability primary determinant of nonoperative success
  - > 90% successfully managed nonoperatively
- Active bleeding by imaging
  - May indicate more severe injury
  - Associated with greater transfusion requirement
  - ± ↑ risk for failure of nonoperative management
- Complications
  - Typically occur with higher grade injuries (> grade III)
  - Biliary: Biloma (most common), bile leak, hemobilia
  - Vascular: Pseudoaneurysm, AVF, delayed hemorrhage
    - Delayed hemorrhage (as late as 2 weeks after injury)
      - 1-3% of patients
      - Association with "blush" on initial CT debated
  - Abscess

Treatment

- Nonoperative management if hemodynamically stable
- American Pediatric Surgical Association (APSA) guidelines for hemodynamically stable isolated liver injury
  - Grade I: Hospital stay 2 days, activity restriction 3 weeks
  - Grade II: Hospital stay 3 days, activity restriction 4 weeks
  - Grade III: Hospital stay 4 days, activity restriction 5 weeks
  - Grade IV: ICU stay 1 day, hospital stay 5 days, activity restriction 6 weeks
  - No routine pre-/postdischarge imaging for grades I-IV
  - Return to full-contact sports at discretion of surgeon
  - APSA does not provide guidelines for grade V, VI injuries
- Laparotomy & surgical hemostasis in unstable patients
- Options for consideration as adjuncts to, or in lieu of, surgery
  - Angiographic embolization for active bleeding
    - Postembolization syndrome common: 90%
  - ERCP with sphincterotomy &/or stent for bile leak
    - Risk of postprocedure infection, pancreatitis
- Utility of routine follow-up imaging in asymptomatic patients debated

DIAGNOSTIC CHECKLIST

Consider

- In infants, CECT warranted if ↑ liver enzymes + clinical/radiological signs of NAT

Image Interpretation Pearls

- Accurate description of hepatic injuries vital, but vigilance & diligence required to seek out additional injuries that may be less obvious (yet of greater clinical significance)

Reporting Tips

- AAST grade guides management

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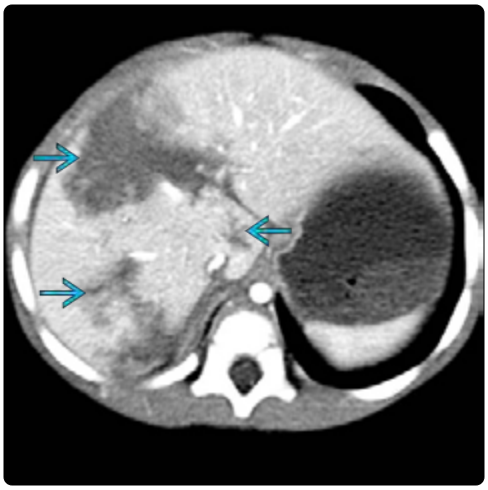
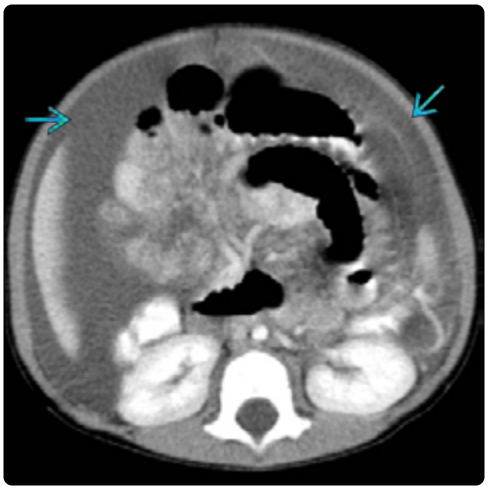
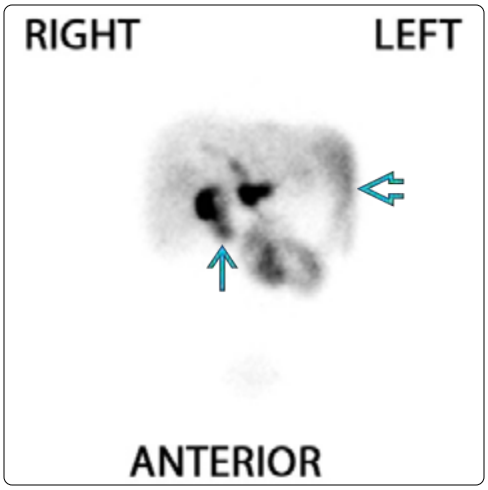
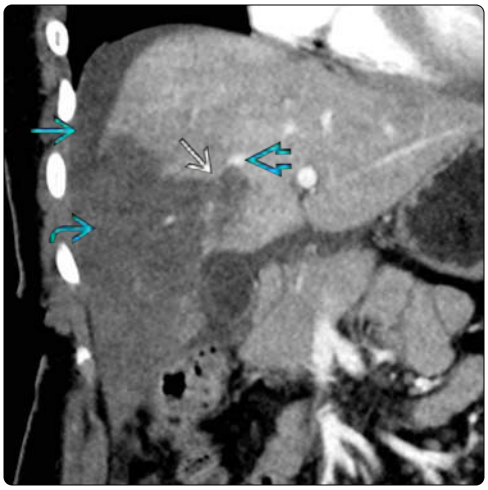


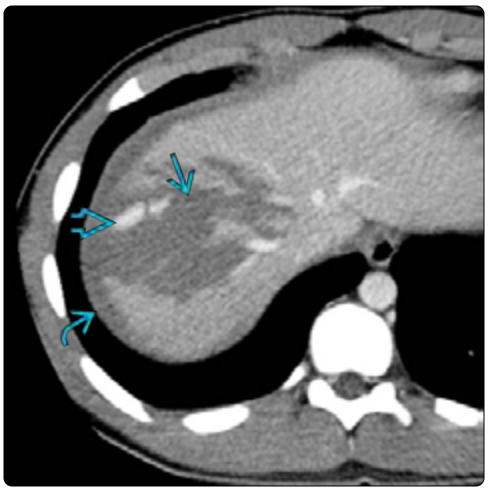


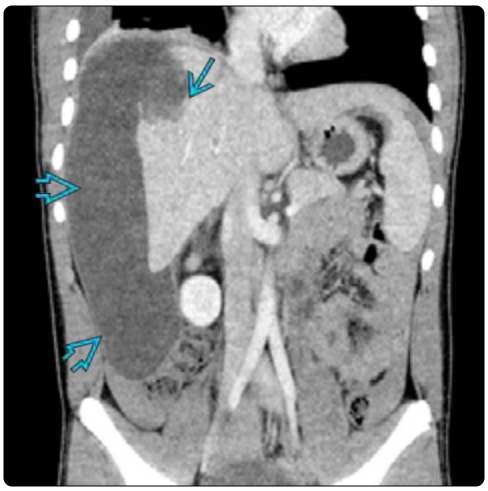
(Left) *Axial CECT in a 14 month old who sustained blunt abdominal trauma shows multiple parenchymal lacerations. The lacerations significantly disrupted > 3 Couinaud segments of the right lobe, consistent with a grade V liver injury.* **(Right)** *Axial CECT in the same child 9 days later shows a large amount of ascites. The amount of free fluid had moderately increased since the prior exam. Clinically, there was concern for a biliary leak in this patient.*





(Left) *Anterior Tc-99m HIDA scan in the same child shows accumulations of radiotracer inferior to the gallbladder and in the left lateral peritoneum, consistent with a biliary leak.* **(Right)** *Coronal CECT of a 14-year-old girl after an all-terrain vehicle accident shows a laceration extending to the right hepatic vein with devascularization of the inferior right hepatic lobe. Injuries to the retrohepatic inferior vena cava or central major hepatic veins are grade V injuries. Hemoperitoneum is also shown.*





(Left) *Axial CECT of a 14-year-old boy status post fall demonstrates a grade IV injury with laceration involving segments 7 and 8, disrupting 25-75% of the right hepatic lobe. There is also active extravasation and perihepatic hemorrhage.* **(Right)** *Coronal CECT of the same patient performed for worsening pain 2 weeks after the initial injury shows that the laceration is contiguous with a large localized fluid collection. Bilious fluid was noted upon drainage, consistent with biloma.*

KEY FACTS

IMAGING

- CECT: Gold standard for blunt abdominal trauma
- Laceration: Irregular, linear, branching foci of fluid density
- Hematoma: Intraperitoneal, retroperitoneal, subcapsular, &/or intraparenchymal collection(s)
  - Fluid attenuation (0-30 HU) if fresh, unclotted blood; ↑ attenuation (40-80 HU) suggests clotted blood, may implicate site of injury by proximity (sentinel clot)
- Active extravasation: Irregular, nonanatomic focus of ↑ density (isodense to adjacent artery); accumulates on delayed images
- Pseudoaneurysm: ↑ density outpouching of arterial lumen (isodense to adjacent artery); washes out with delay
- Complications: Pseudocyst, pseudoaneurysm, arteriovenous fistula, delayed rupture (hemorrhage > 48 hours after trauma), venous thrombosis, abscess

TOP DIFFERENTIAL DIAGNOSES

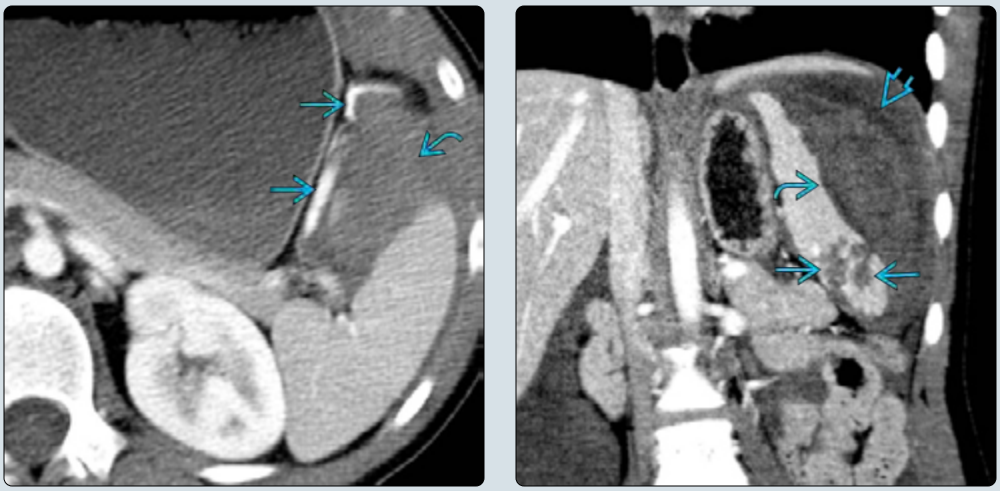
- Splenic cleft
- Artifacts: Beam hardening, early bolus, patient motion
- Hypoperfusion complex
- Splenic infarct
- Splenic abscess

PATHOLOGY

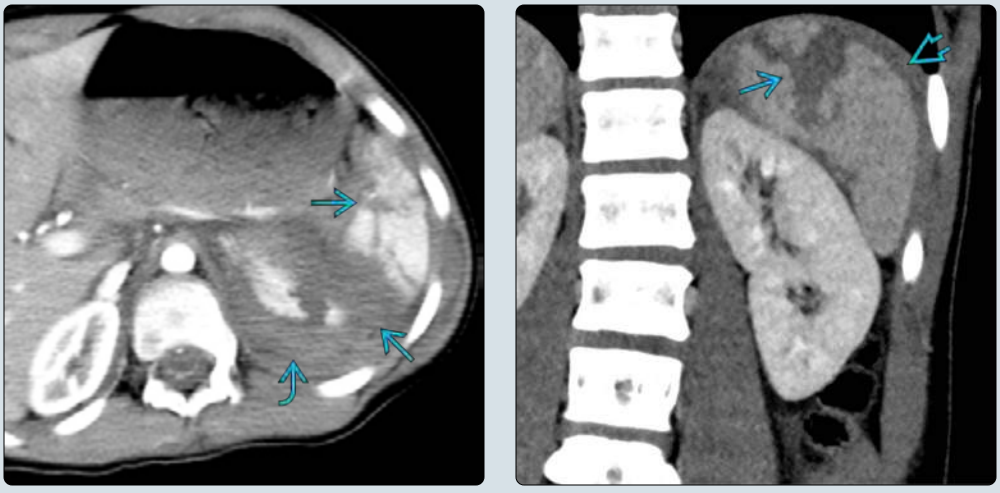
- Typical etiologies: Blunt trauma, motor vehicle collision
- ± additional injuries: Left lower rib fractures, other viscera

CLINICAL ISSUES

- Presentations: LUQ pain & tenderness, hypotension
- Treatment
  - Nonoperative management of isolated blunt splenic injury in stable patients: > 95% success rate
  - American Association for Surgery of Trauma grading scale not predictive of outcomes in pediatrics
    - Injury grade assists with management decisions
  - Unlike adults, active bleeding or pseudoaneurysm alone not clearly predictive of nonoperative treatment failure



(Left) *Axial CECT in a patient with hemophilia and abdominal pain status post fall shows an irregular, nonanatomic collection of dense contrast tracking medial to a perisplenic hematoma, consistent with active hemorrhage.* **(Right)** *Coronal CECT in a patient injured while playing basketball shows a grade III splenic injury with lacerations > 3 cm in depth. A subcapsular hematoma > 50% of the splenic surface area scallops the splenic margin.*



(Left) *Axial CECT shows a fractured spleen with a perisplenic hematoma.* **(Right)** *Coronal CECT in a 14-year-old boy injured while skiing shows a grade III splenic injury. Branching linear hypodensities > 3 cm in depth that extend to the splenic capsule are consistent with lacerations. A small perisplenic hematoma is also seen.*

TERMINOLOGY

Synonyms

- Splenic laceration, fracture, or injury; blunt injury to spleen

Definitions

- Parenchymal injury to spleen ± capsular disruption

IMAGING

General Features

- Best diagnostic clue
  - Jagged, linear fluid density foci in splenic parenchyma ± intraparenchymal or subcapsular fluid collection
  - Perisplenic hematoma > 40 Hounsfield units (HU)

Radiographic Findings

- Radiography
  - Triad: Elevated left hemidiaphragm, left lower lobe atelectasis/collapse, left pleural effusion
  - Left lower rib fractures most common (44% of patients)
  - Medial displacement of gastric bubble
  - Inferior displacement of splenic flexure, bowel gas
  - Hemorrhage may obscure contours of intraperitoneal/retroperitoneal structures: Psoas margin, flank stripe, renal contour, splenic margin

CT Findings

- CECT
  - Accuracy 98%, sensitivity 95%
  - Laceration: Irregular, fluid attenuation, linear/branching foci within parenchyma; may extend to capsule
  - Fracture: Laceration extending from 1 capsular surface to another
  - Rupture: Multiple fractures with discontinuous splenic fragments
  - Hematoma: Fluid attenuation (0-30 HU) collection; ↑ attenuation (40-80 HU) may represent clotted blood
    - Parenchymal: Round or irregular collection in spleen
    - Subcapsular: Peripheral crescentic or lenticular collection compressing parenchymal margin
    - Perisplenic: Collection in adjacent peritoneal spaces
      - Sentinel dense clot adjacent to spleen: Sensitive predictor for splenic injury
    - Hemoperitoneum: Layering dense hematocrit level in dependent pelvis (may be subtle)
    - Retroperitoneal: Typically if splenic hilum injured
      - Involves splenorenal ligament, anterior pararenal space, pancreas
  - Active arterial hemorrhage (extravasation)
    - Irregular, nonanatomic contrast collection; isodense to aorta; accumulates on delayed images
  - Pseudoaneurysm
    - ↑ attenuation outpouching of arterial lumen; isodense to aorta
    - ↓ attenuation on delayed images without ↑ size
    - Almost 75% not seen on initial CECT
  - Venous thrombosis: Focal or diffuse lack of splenic vein enhancement despite visualization of other veins

Ultrasonographic Findings

- Grayscale ultrasound
  - Linear or branching hypoechoic foci
  - Subcapsular or perisplenic collection with variable echogenicity
  - Free intraperitoneal fluid (absent in 25% of splenic injury)
- Color Doppler
  - Splaying or compression of vessels by hematoma
  - Yin-yang appearance of pseudoaneurysm
  - Lack of splenic vein flow with venous thrombus
  - Lack of parenchymal flow with devascularizing injury
- FAST exam: 37-85% sensitivity, 99-100% specificity
- Abdominal CEUS: 93% sensitivity, 99% specificity

Angiographic Findings

- Blush from active arterial extravasation of contrast
- Distortion of vessels, enhancing parenchyma by hematoma
- Pseudoaneurysm: Arterial outpouching
- Venous thrombosis: Filling defect

Imaging Recommendations

- Best imaging tool
  - CECT

DIFFERENTIAL DIAGNOSIS

Splenic Cleft

- Congenital variant in contour of spleen
- Smooth, curvilinear ↓ attenuation focus without other evidence of hemorrhage

Artifacts

- Beam-hardening: Linear hypodense streaks from ribs/arms
- Motion: Blurring or discontinuity of structures across image
- Early bolus: Specific pattern of enhancement
  - Parallel, archiform, "corrugated" waves of ↓ splenic enhancement on early arterial phase CECT
    - 95% resolve by 70 seconds
    - Due to enhancement differences of red & white pulp

Hypoperfusion Complex

- ↓ splenic enhancement
- Fluid-filled bowel with ↑ wall enhancement
- ± flattened IVC, intensely enhancing small caliber aorta

Splenic Infarct

- Wedge-shaped focus of ↓ density with apex towards hilum
- Associated with splenomegaly & systemic disorders

Splenic Abscess

- Round, irregular, singular, or multifocal ↓ attenuation lesion(s)
- Clinical history/findings of infection, immunodeficiency, trauma, emboli, travel, or cat scratch

PATHOLOGY

General Features

- Etiology
  - Blunt trauma
    - Motor vehicle collision most common (40%)
    - Bicycle-related (17%), falls (19%), sports-related injuries (13%)
    - Iatrogenic injury rare (thoracentesis, biopsy, intraoperative)

- Nonaccidental trauma (NAT) rare (1%)
  - Seen in up to 26% of NAT patients
- Preexisting splenic enlargement (EBV, hematologic disorders) predisposes to injury

Staging, Grading, & Classification

- American Association for the Surgery of Trauma (AAST)
  - Grade I
    - Hematoma: Subcapsular, < 10% surface area
    - Laceration: Capsular tear, < 1-cm parenchymal depth
  - Grade II
    - Hematoma: Subcapsular, 10-50% surface area; intraparenchymal, < 5 cm in diameter
    - Laceration: Capsular tear, 1-3-cm parenchymal depth; does not involve trabecular vessels
  - Grade III
    - Hematoma: Subcapsular, > 50% surface area or expanding; ruptured subcapsular or parenchymal hematoma; intraparenchymal hematoma ≥ 5 cm or expanding
    - Laceration: > 3-cm parenchymal depth or involving trabecular vessels
  - Grade IV
    - Laceration: Involving segmental or hilar vessels producing major devascularization (> 25% of spleen)
  - Grade V
    - Laceration: Completely shattered
    - Vascular: Hilar injury that devascularizes spleen
  - Advance 1 grade for multiple injuries up to Grade III

Gross Pathologic & Surgical Features

- Forceful flexion of spleen along axis
- Laceration typically intersegmental, extending towards hilum

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Left upper quadrant tenderness, pain
  - Hypotension (25-30%)
  - Lap-belt ecchymosis, seatbelt sign
- Other signs/symptoms
  - Rib pain: Left lower posterior rib fractures
  - Kehr sign: Pain radiating to left shoulder, phrenic nerve
  - Abdominal rigidity, shock in neonates
  - Not clinically apparent (10-20%)

Natural History & Prognosis

- Excellent prognosis with early diagnosis & intervention
- AAST grading not predictive of outcome in pediatrics
- Hemodynamic stability important for nonoperative success
- Active bleeding, pseudoaneurysm alone do not predict nonoperative treatment failure, but is controversial
  - Active bleeding/blush may be associated with more severe injury, can result in delayed hemorrhage
  - Pseudoaneurysm may result in delayed hemorrhage; many spontaneously resolve
- Complications: Pseudocyst, pseudoaneurysm, arteriovenous fistula, delayed rupture (hemorrhage > 48 hours after trauma), venous thrombosis, abscess

Treatment

- Nonoperative management of isolated blunt splenic injuries in stable patients: > 95% success rate
- Splenectomy or splenorrhaphy when surgery required
  - ↑ hospital stay, ↑ transfusions, ↑ infections
- Embolization of pseudoaneurysm or active hemorrhage may be useful surgical adjunct or alternative
  - No clear guidelines for use
- Routine follow-up CECT does not change outcome or management of asymptomatic patients
  - Utility of follow-up imaging for pseudoaneurysm detection in asymptomatic patients debated
- American Pediatric Surgical Association (APSA) guidelines for hemodynamically stable patient with isolated splenic injury
  - Grade I: Hospital stay 2 days, activity restriction 3 weeks
  - Grade II: Hospital stay 3 days, activity restriction 4 weeks
  - Grade III: Hospital stay 4 days, activity restriction 5 weeks
  - Grade IV: ICU stay 1 day, hospital stay 5 days, activity restriction 6 weeks
  - No pre/postdischarge imaging for Grades I-IV
  - APSA does not provide guidelines for grade V injuries
  - Return to full-contact sports at discretion of surgeon
- Vaccination (pneumococcal, *Haemophilus influenzae* type b, meningococcal, influenza) for patients without functional spleen
- Daily prophylactic antibiotics in patients < 5 years old without functional spleen

DIAGNOSTIC CHECKLIST

Reporting Tips

- Injury grade assists with management decisions

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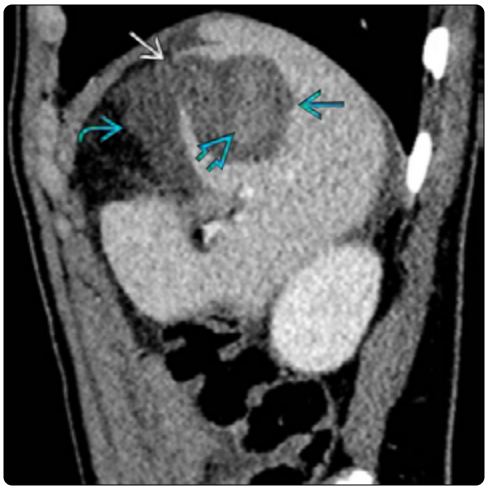
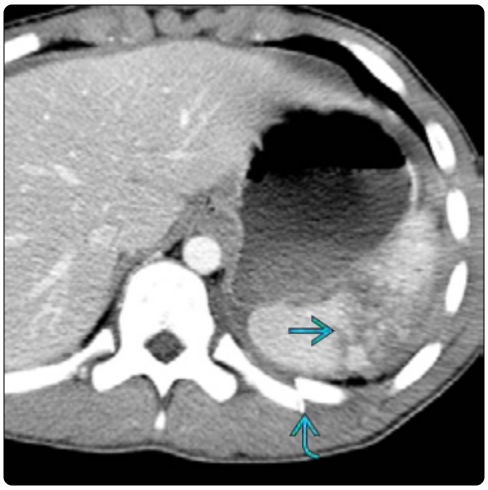
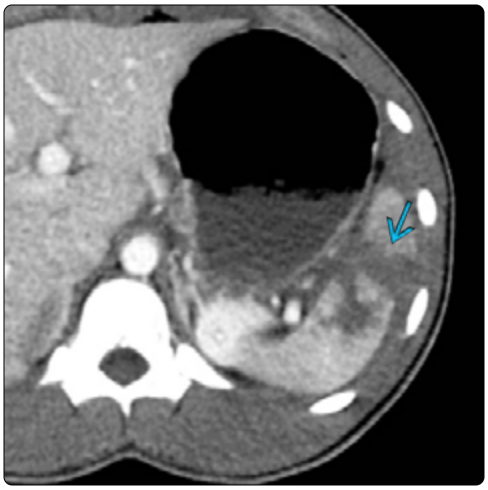
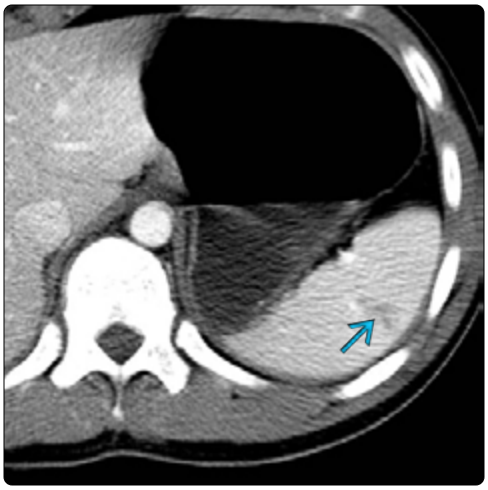


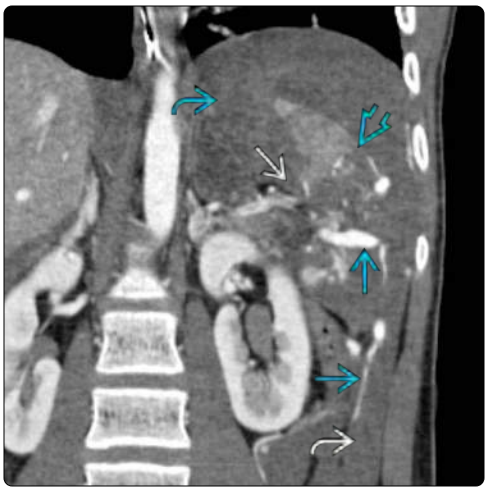


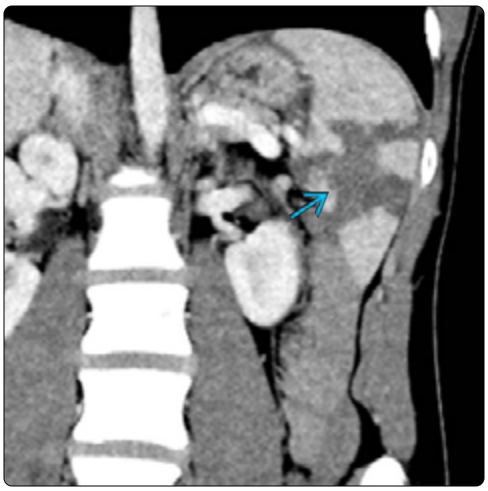
(Left) *Sagittal CECT shows a grade III splenic injury in a 15-year-old boy who fell from a ladder. There is an intraparenchymal hematoma, which has ruptured into a perisplenic collection. High-attenuation material in the hematoma is due to clotted blood.* **(Right)** *Axial CECT in a 14 year old who fell 20-30 feet through a skylight shows a displaced posterior rib fracture and multiple lacerations through the spleen. The lacerations involved segmental and hilar vessels, consistent with a grade IV splenic injury.*





(Left) *Axial CECT in a patient who was assaulted shows a fractured spleen with low density branching lacerations > 3 cm in length that extend to 2 capsular surfaces, consistent with a grade III injury.* **(Right)** *Axial CECT shows a small, irregular, linear hypodense band within the spleen. This was categorized as a grade II splenic injury as it was between 1-3 cm in length.*





(Left) *Coronal CECT in a patient hit by a train shows a grade V injury with a hilar avulsion of a devascularized, shattered spleen. A nonanatomic focus of increased attenuation is due to contrast extravasation from active arterial hemorrhage. There is a large perisplenic hematoma with hemoperitoneum.* **(Right)** *Coronal CECT in a 15-year-old trauma patient depicts a jagged, stellate hypodensity representing a branching laceration, consistent with a grade III splenic injury.*

Duodenal Trauma

KEY FACTS

IMAGING

- CECT best imaging tool in acute trauma setting
- Injuries generally classified as hematoma vs. laceration
  - Hematoma: Nonenhancing intraluminal, intramural, &/or paraduodenal collection of ↑ attenuation
    - Intraluminal: Expansile ovoid or tubular filling defect
      - Coiled-spring appearance on upper GI series
    - Intramural: Mass-like focal wall thickening ± intramural gas; eccentric narrowing of displaced lumen
  - Duodenal laceration/perforation
    - Interruption of bowel wall, extravasation of intraluminal contents, & free retroperitoneal air: Specific but not sensitive
    - Duodenal wall thickening > 3 mm & ↑ attenuation fluid + stranding in retroperitoneum: Sensitive but not specific
- Associated pancreatic trauma in up to 42% of cases

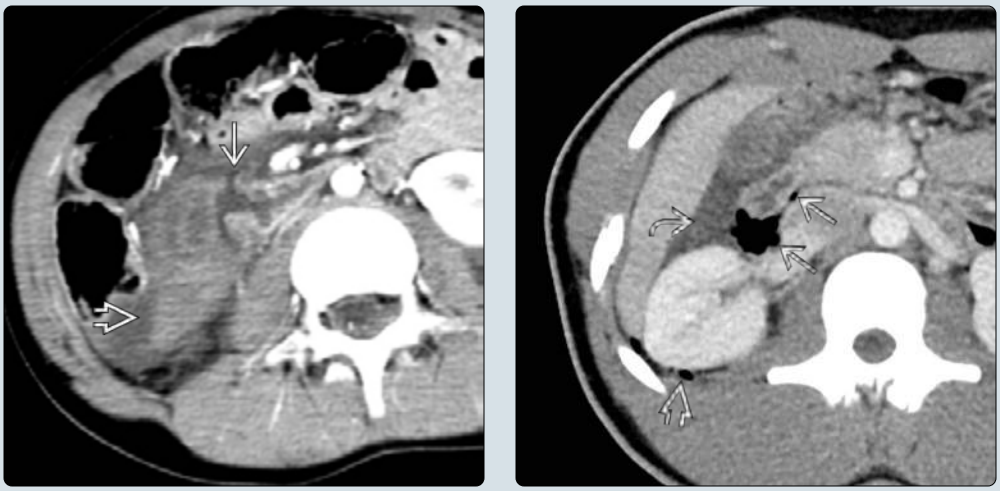
PATHOLOGY

- Blunt trauma > 70%, penetrating trauma 20%
  - Motor vehicle collision, fall, bicycle handlebar injury, assault
  - Consider nonaccidental trauma in young patients (particularly < 2 years old with delayed presentation)
- Also consider with bleeding disorders, anticoagulation, Henoch-Schönlein purpura, recent endoscopic procedures

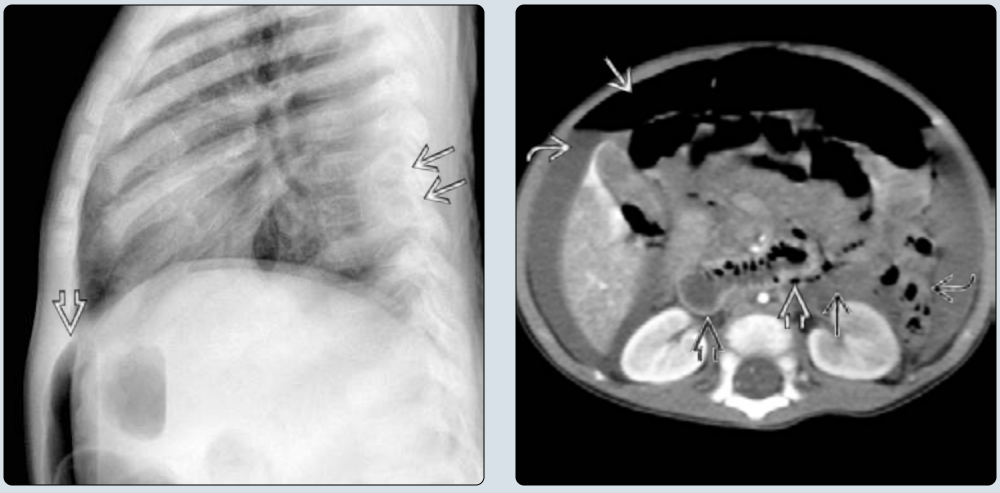
CLINICAL ISSUES

- Common symptoms: Abdominal pain, nausea, vomiting
- Delay in diagnosis & treatment: ↑ morbidity & mortality
- Treatment
  - Intramural/intraluminal hematoma: Initial management nonoperative (bowel rest, total parenteral nutrition, nasogastric decompression); drainage in refractory cases
  - Perforation: Primary surgical repair & drainage vs. more complex procedures in severe cases

(Left) *Axial CECT shows focal disruption of the enhancing duodenal wall with adjacent hematoma. This is a specific but rarely seen finding of traumatic duodenal perforation.* **(Right)** *Axial CECT from a 17-year-old boy with abdominal pain, vomiting, & leukocytosis after being kicked shows retroperitoneal air posterior to the 2nd & 3rd portions of the duodenum (D2 & D3) & posterior to the right kidney. Adjacent fluid is also noted. A duodenal laceration was found at surgery.*



(Left) *Cross-table lateral view of the chest in a 9-month-old male victim of nonaccidental trauma shows free intraperitoneal air. Healing posterior rib fractures are also present.* **(Right)** *Axial CECT from the same patient shows free intra- & retroperitoneal air as well as intra- & retroperitoneal fluid. Mucosal hyperenhancement & small bowel wall thickening are also noted. Perforations of the duodenum & jejunum were repaired surgically.*



TERMINOLOGY

Abbreviations

- D[x] = segment of duodenum (e.g., D1 = 1st segment of duodenum)

IMAGING

General Features

- Location
  - Most commonly involves D2 (46%) or D3 (63%)

Radiographic Findings

- Mass effect of hematoma may lead to
  - Dilated stomach &/or duodenal bulb
  - Displaced bowel gas in right upper quadrant
- Laceration may lead to
  - Retroperitoneal air (D2-D4)
    - Outlines kidneys, psoas muscles
  - Intraperitoneal air (D1)
    - Upright or left-side down decubitus view: Air under diaphragm overlying liver
    - Supine
      - Rigler sign: Air on both sides of bowel wall
      - Falciform ligament sign: Vertical linear density in RUQ outlined by air lucency
      - Football sign: Ovoid lucency of whole abdomen

Fluoroscopic Findings

- Upper GI
  - Typically not performed in acute trauma setting
  - Intramural hematoma
    - Displaced lumen with eccentric or circumferential narrowing/obstruction
    - Wall/fold thickening
  - Intraluminal hematoma
    - Ovoid clot expands duodenal lumen
    - Coiled spring appearance due to contrast tracking between mucosal folds & intraluminal clot

CT Findings

- CECT
  - Hematoma: Well-circumscribed, ovoid, heterogeneous, or homogeneous mass with ↑ attenuation of 40-70 HU
    - Occurs in lumen, bowel wall, or paraduodenal tissues
      - Dense sentinel clot may localize injury site
  - Contusion: Focal duodenal wall thickening/edema
    - > 3 mm or disproportionate to normal bowel wall
  - Vascular insult vs. response to wall injury: Abnormal bowel wall enhancement
    - Judged relative to psoas muscle
    - ↑ enhancement: Paradoxically found with hypoperfusion due to damaged vascular endothelium
    - ↓ enhancement: Suggests traumatic ischemia, vascular compromise
  - Specific signs of duodenal laceration/perforation
    - Interruption of enhancing bowel wall
    - Extraluminal contrast &/or air
      - Retroperitoneal (D2-D4) or intraperitoneal (if D1)
      - Sensitivity for free air with traumatic bowel injury only 30-60%
      - Lung windows ↑ sensitivity for free air

MR Findings

- Not generally performed in acute trauma setting
- T1WI, T2WI: Hematoma signal varies with age
- T1WI C+: Nonenhancing mass; may have enhancing rim

Ultrasonographic Findings

- Grayscale ultrasound
  - Hematoma
    - Focal intramural/intraluminal/paraduodenal mass, wall thickening, or collection
    - Variable echogenicity based on age; ↓ with time
  - Retroperitoneal air: May see linear hyperechogenicity with dirty posterior acoustic shadowing
- Color Doppler
  - No vascularity within hematoma

Imaging Recommendations

- Best imaging tool
  - CECT
    - In blunt trauma setting, oral contrast does not significantly improve sensitivity for duodenal injury → delay in imaging & treatment
  - In cases with persistent clinical symptoms, consider follow-up with ultrasound or UGI every 7 days

DIFFERENTIAL DIAGNOSIS

Enteric Duplication Cyst

- 12% occur in gastroduodenal location
- Well-circumscribed cystic mass
- US: Cyst wall frequently has trilaminar gut signature

Duodenal Ulcer/Duodenitis

- Uncommon in young patients
- Poorly defined proximal duodenal wall edema with interruption of enhancing mucosa
- May perforate → extraluminal gas
- May cause duodenal stricture & gastric outlet obstruction

Crohn Disease

- Involves duodenum in 5-20% of patients
- Wall thickening, hyperenhancement, or stricture
- May form fistula with adjacent bowel

Small Bowel Neoplasm

- Rare in children; Burkitt lymphoma most common
- Typically enhances, unlike hematoma
- May cause duodenal luminal narrowing, obstruction

Pancreatitis

- Enlarged, edematous pancreas with peripancreatic stranding & fluid
- Duodenum may have secondary inflammatory change

Pancreatic Trauma

- Concomitant pancreatic & duodenal injuries common
- Enlarged, edematous pancreas with peripancreatic stranding & fluid
  - Fluid may show ↑ attenuation
- Linear transversely oriented focus of ↓ attenuation due to parenchymal laceration

Annular Pancreas

- Commonly presents in neonatal period
- Ring of pancreatic tissue surrounding D2 results in duodenal stenosis, obstruction

PATHOLOGY

General Features

- Etiology
  - Duodenal injury in 2-10% of children with blunt abdominal trauma
  - Blunt trauma accounts for > 70% of duodenal injuries
    - Motor vehicle-related 50%, falls 17%, bicycle-related 8%, nonaccidental trauma (NAT) 8%, other blunt trauma 17%
      - Consider NAT in younger patients (particularly < 2 yr of age) with delayed presentation to emergency department
  - Penetrating trauma > 20%
  - Iatrogenic
    - Upper endoscopy with biopsy: Hematoma in 0.1-7.0%
      - ↑ risk in bone marrow transplant patients
    - ERCP: Hematoma in 1.3%, perforation in 0.1-0.6%
  - Spontaneous intramural hematomas with bleeding disorders, anticoagulation therapy, vasculitis, Henoch-Schönlein purpura
- Associated abnormalities
  - Abdominal injuries: Pancreas 42%, liver 29%, spleen 17%

Staging, Grading, & Classification

- Grading by American Association for Surgery of Trauma Duodenal Injury Scale
  - I: Hematoma involving single portion of duodenum; partial thickness laceration without perforation
  - II: Hematoma involving >1 portion of duodenum; laceration disrupting < 50% of circumference
  - III: Laceration disrupting 50-75% of circumference of D2 or 50-100% D1/D3/D4
  - IV: Laceration disrupting > 75% of circumference of D2 or involving ampulla or distal common bile duct
  - V: Laceration with massive disruption of duodenopancreatic complex; duodenum devascularized
  - Advance 1 grade for multiple injuries up to grade III

Gross Pathologic & Surgical Features

- 3 mechanisms act in isolation or in combination
  - Compression of intestinal loops at impact
    - Sudden ↑ in intraluminal pressure → perforation
  - Direct force crush-injury between anterior abdominal wall & spine
  - Rapid deceleration → tear at junction of mobile intraperitoneal & fixed retroperitoneal segments

Microscopic Features

- Duodenal hematoma typically develops in submucosal &/or subserosal layers

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Nausea, vomiting, abdominal pain

Natural History & Prognosis

- Overall morbidity 48%, mortality 19%
- Delay in diagnosis & treatment → ↑ length of hospitalization, ICU time, need for total parenteral nutrition (TPN), complication rates
- Mortality usually secondary to associated injuries, most commonly traumatic brain injuries
- Stricture may rarely occur as long-term complication
- Postoperative complications: Postoperative ileus 19%, wound infection 9%, traumatic pancreatitis 5%, abscess 3%, pancreatic fistula 2%, enterocutaneous fistula 2%

Treatment

- Hematoma
  - Nonoperative: Bowel rest, nasogastric decompression, TPN
    - Grade II injury associated with longer duration of TPN compared to grade I
  - Surgical or percutaneous drainage if no improvement
- Laceration/perforation
  - Primary repair + decompression & paraduodenal drain
  - Pyloric exclusion with gastrojejunostomy for complex injuries & wounds involving medial wall of D2
- With associated pancreatic injury
  - Duodenal primary repair/resection & pancreas drainage
  - If concern for biliary injury → ERCP
  - If repair/resection precluded, debridement & Roux-en-Y
  - Pancreaticoduodenectomy if extensive destruction to pancreatic head, common bile duct, & duodenum

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Duodenal injury suggested by wall thickening &/or intraluminal mass + air, fluid, or stranding in retroperitoneum
  - Bowel wall interruption & extravasation rarely seen
- Evaluate pancreas closely for concomitant injury
- Consider NAT in younger patients (especially patients < 2 years old with delayed presentation)

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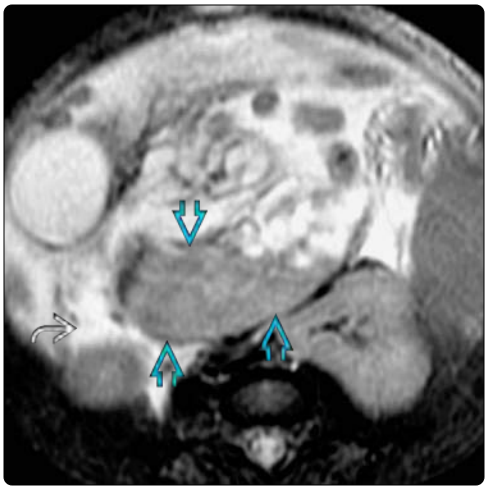
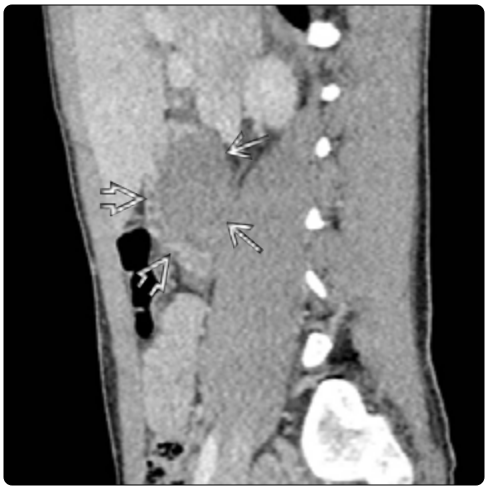
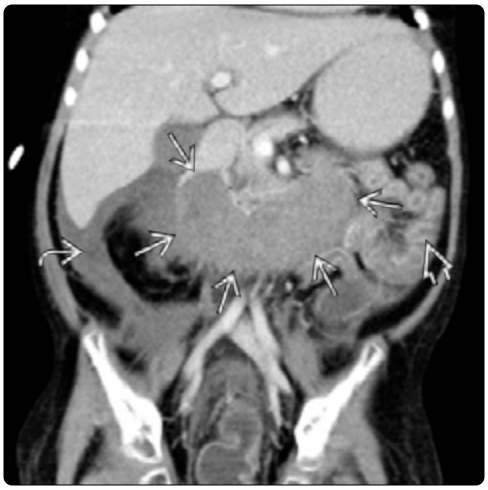
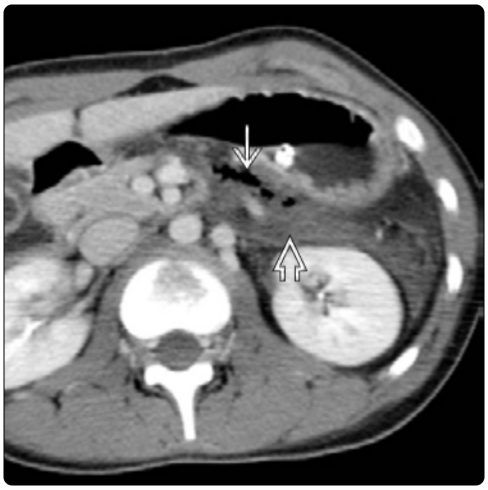


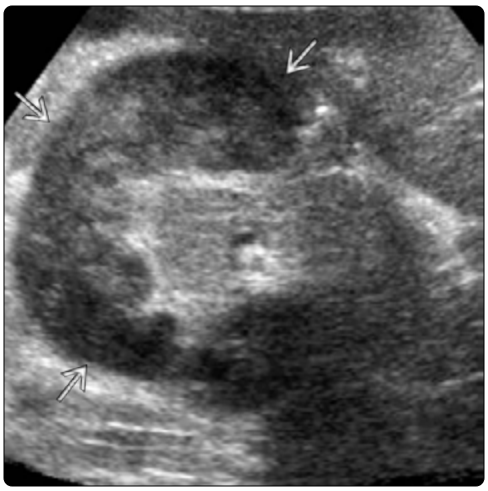


(Left) *Axial T2 FS MR shows an intermediate signal intensity hematoma filling D3 in this patient with liver disease & a preceding endoscopic biopsy. Note the adjacent ascites.* **(Right)** *Sagittal CECT from a 14-year-old boy with a history of abdominal trauma shows a mildly heterogeneous ovoid mass displacing D2 anteriorly, consistent with hematoma.*





(Left) *Coronal CECT from a 5-year-old patient after endoscopic biopsy shows an elongated mass involving D2-D4, consistent with a hematoma. Intraperitoneal hemorrhage is also noted. The mucosal hyperenhancement of bowel loops is due to graft-vs.-host disease.* **(Right)** *Axial CECT from a 16-year-old girl after a motor vehicle accident shows free air & fluid in the anterior pararenal space near D3 & D4. A perforation was found at this site during surgery.*



(Left) *Transverse sonographic image from a 2-year-old girl with pancytopenia & hematemesis after endoscopy with biopsy shows a bulky C-shaped heterogeneous mass of D1-D3, consistent with an intramural or intraluminal duodenal hematoma.* **(Right)** *Lateral view from an upper gastrointestinal series in the same patient demonstrates a large obstructive filling defect expanding D1, consistent with an intraluminal duodenal hematoma.*

Pancreatic Trauma

KEY FACTS

IMAGING

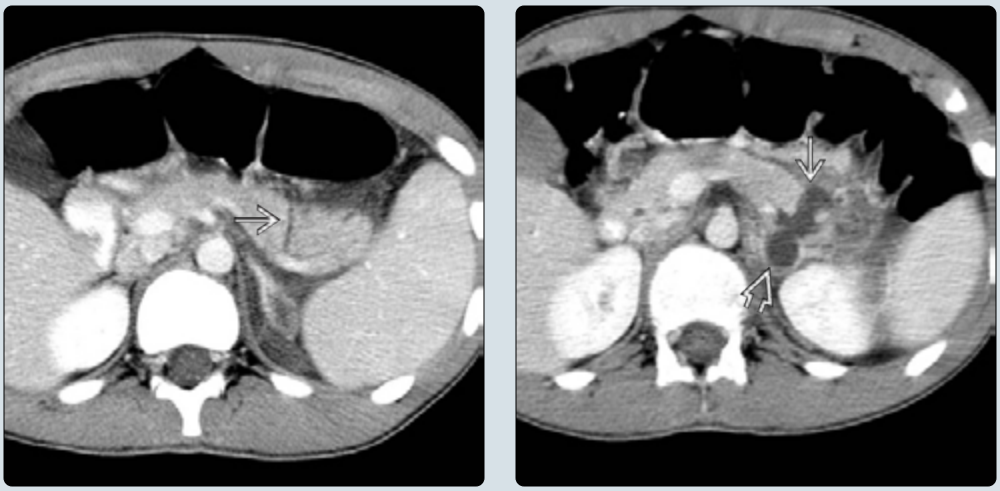
- Modality of choice: CECT
  - Caveat: 20-40% false-negative rate in first 12 hours
- Classic imaging findings
  - Contusion/inflammation: Focal or diffuse ↓ enhancement & ↑ size relative to normal parenchyma
  - Laceration: Linear fluid-attenuation parenchymal defect
    - Usually of short axis; most common at junctions of pancreatic head-neck or body-tail
    - Depth > 50% suggests pancreatic duct disruption
  - Hematoma: Irregular or round fluid collection in/around pancreas; ↑ attenuation (40-80 HU) if clotted
  - Varying degrees of peripancreatic fluid tracking along/within fascial planes & adjacent compartments
- MRCP or ERCP to assess main pancreatic duct injury

PATHOLOGY

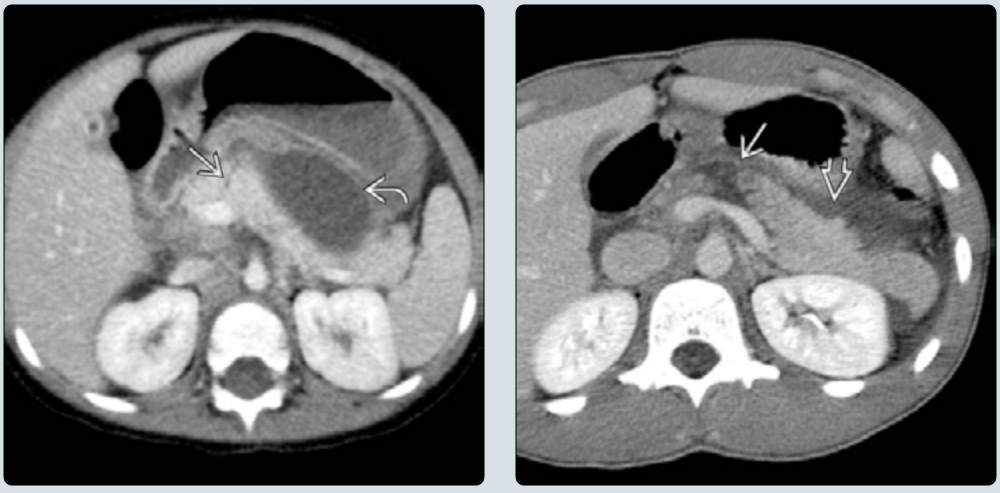
- Mechanisms: Blunt trauma: 93%; penetrating trauma: 7%
- Pancreatic injury in 17% of nonaccidental trauma patients

CLINICAL ISSUES

- Epigastric pain out of proportion to physical findings
- ↑ serum amylase (may be normal initially)
- Nonoperative management: Total parenteral nutrition ± octreotide infusion, percutaneous/endoscopic collection drainage, endoscopic duct stenting
  - Failure more likely with pancreatic duct injury
- Operative management: Surgical drainage, partial/distal pancreatectomy
- Management of American Association for Surgery of Trauma grades
  - I & II: Typically nonoperative
  - III-V: No consensus on management strategy
    - Nonoperative: 43% failure rate; ↑ risk of pseudocysts, ERCP failure, & post-ERCP pancreatitis
    - Operative: Shorter time to resolution; ↑ rates of fistula formation & leak



(Left) *Axial CECT in a 14-year-old who sustained an elbow injury to the abdomen shows a linear, fluid-density laceration through the pancreatic body near the tail. A left adrenal injury was also noted on contiguous images.* **(Right)** *Axial CECT in the same teenager 6 days later shows a pancreatic tail fluid collection extending into the anterior pararenal space. The patient was treated conservatively. A follow-up US 1 month after the initial injury demonstrated resolution.*



(Left) *Axial CECT in a 2-year-old nonaccidental trauma victim with elevated serum amylase shows a laceration at the pancreatic neck-body junction. The > 50% depth of the laceration is concerning for duct injury. A large pancreatic bed fluid collection is seen anteriorly.* **(Right)** *Axial CECT in a 15-year-old boy with a football injury is shown. There is a grade III laceration involving the neck & body of the pancreas. Poorly defined fluid is seen in the peripancreatic tissues.*

IMAGING

General Features

- Best diagnostic clue
  - Linear, fluid-attenuation cleft through short axis of enhancing pancreas + peripancreatic fluid

CT Findings

- CECT
  - Contusion/inflammation
    - Focal or diffuse ↓ attenuation relative to normally enhancing parenchyma
    - Focal or diffuse enlargement of pancreas
  - Laceration
    - Linear fluid attenuation cleft; ↑ attenuation with clotted blood (40-80 HU)
    - Usually involves short axis of organ
      - Most common at junctions of pancreatic head-neck or body-tail
    - Depth > 50% suggestive of pancreatic duct disruption
  - Hematoma
    - Irregular or round parenchymal/peripancreatic fluid attenuation collection; ↑ attenuation with clotted blood (40-80 HU)
  - Fluid collections adjacent to & tracking away from pancreas
    - Separating splenic vein & pancreas
    - Surrounding superior mesenteric & portal veins
    - Separating pancreas & duodenum
    - Intraperitoneal: Lesser sac, paracolic gutters
    - Retroperitoneal: Anterior & posterior pararenal spaces
      - Thickening of fascial planes
  - Pseudocyst
    - Well-circumscribed peripancreatic fluid collection with enhancing wall
      - Usually develops after 4 weeks (but may develop earlier)
    - Components essentially all liquefied

MR Findings

- T2WI
  - ↑ signal intensity in & around pancreas: Edema, hemorrhage, pancreatic enzymes, inflammation
  - Focal or diffuse pancreatic enlargement
  - Linear discontinuity of parenchyma in short axis: ↑ intensity fluid within defect or separating 2 fragments of fractured parenchyma
- T1WI C+
  - Heterogeneous enhancement of pancreas
  - Nonenhancing fluid collections
  - Pseudocyst
    - Not acute finding (requires time for organization)
    - Nonenhancing, low signal intensity fluid collection; hyperintense signal may indicate hemorrhage
    - May have enhancing capsule
- MRCP
  - Diagnostic evaluation of pancreatic duct
  - Disruption of duct continuity ± dilation
    - May be difficult to distinguish disruption vs. compression by extrinsic collection
  - May demonstrate continuity of pseudocyst with pancreatic duct

Ultrasonographic Findings

- Grayscale ultrasound
  - Hematoma
    - Poorly marginated fluid collection in/around pancreas; variable echogenicity
  - Laceration
    - Linear hypoechoic defect in parenchyma; hyperechoic with clot
  - Contusion
    - Focal/diffuse enlargement of hypoechoic gland ± peripancreatic fluid
  - Pseudocyst (on follow-up exam)
    - Anechoic mass ± septations, thick rim
    - Internal echoes may indicate infection or hemorrhage
- For acute pancreatic injury, abdominal US sensitivity 44-71%, specificity 100%

Fluoroscopic Findings

- ERCP
  - Definitive pancreatic duct evaluation
    - Duct may be normal, deviated, or narrowed with contusion or laceration
    - Duct obstruction: Disruption vs. compression
    - Duct laceration confirmed with irregular pooling of contrast adjacent to main/branch duct (extravasation)
      - ± continuity of duct with fluid collection, pseudocyst
      - ± extension of contrast into retroperitoneal spaces
  - At risk for post-ERCP pancreatitis, infection, stricture
  - Not always readily available

Imaging Recommendations

- Best imaging tool
  - CECT
    - Caveat: 20-40% false-negative rate in first 12 hours
  - MRCP or ERCP if main pancreatic duct injury suspected

DIFFERENTIAL DIAGNOSIS

"Shock" Pancreas

- Diffusely enlarged & heterogeneous pancreas
  - Low-attenuation septa throughout enhancing parenchyma
- Typical constellation of findings in hypoperfusion complex
  - Fluid distention of small bowel with thickened, hyperenhancing walls diffusely
  - Intense enhancement of adrenal glands
  - ↓ caliber of hyperenhancing aorta; collapsed inferior vena cava
  - Free fluid & mesenteric stranding

Duodenal Trauma

- Hematoma/contusion
  - Wall thickening, ↓ enhancement, &/or ovoid intramural/intraluminal mass
- Duodenal perforation
  - Retroperitoneal contrast, extraluminal gas, discontinuity of enhancing duodenal wall
- Luminal narrowing, edema/fluid in retroperitoneum

- High association with pancreatic injuries

Pancreatitis

- Focal/diffuse pancreatic enlargement with mildly ↓ enhancement
- Peripancreatic fluid & fat-stranding
- Fluid in lesser sac &/or anterior pararenal space
- Areas of nonenhancement may represent necrosis

PATHOLOGY

General Features

- Etiology
  - Blunt trauma: 93% of pancreatic injuries
    - Motor vehicle related: 36%; bicycle injury: 24%; nonaccidental trauma: 10%; other assault: 8%; fall: 7%; other blunt injury: 15%
  - Penetrating trauma: 7% of pancreatic injuries
    - Gunshot wound: 88% of penetrating injuries
- Pancreatic injury found in
  - 3-12% of blunt abdominal trauma patients
  - 17% of patients admitted for nonaccidental trauma
- Pancreatic duct injury rare: 0.1-0.7% of blunt abdominal trauma
- Mechanism of injury
  - Anterior-posterior force compresses pancreas against spine
  - Children at greater risk than adults

Staging, Grading, & Classification

- American Association for Surgery of Trauma
  - Grade I: Minor contusion, no duct injury; superficial laceration, no duct injury
  - Grade II: Major contusion, no duct injury or tissue loss; major laceration, no duct injury or tissue loss
  - Grade III: Distal transection or parenchymal injury with duct injury
  - Grade IV: Proximal (right of superior mesenteric vein) transection or parenchymal injury involving ampulla
  - Grade V: Massive disruption of pancreatic head
  - Advance 1 grade for multiple injuries up to grade III

CLINICAL ISSUES

Presentation

- Clinical triad
  - Epigastric pain out of proportion to physical findings
  - Leukocytosis
  - ↑ serum amylase
    - May be normal initially
    - Following trend useful to identify injury or complications
    - Magnitude not predictive of injury severity
    - Not specific to pancreatic injury
- Isolated injuries rare (< 30%); average of 3-4 coexisting injuries per patient
  - Hepatic: 46.8%; major vascular: 41.3%; splenic: 28%; renal: 23.4%; duodenal: 19.3%
- Subtle imaging findings may lead to delay in diagnosis & treatment with ↑ morbidity & mortality

Natural History & Prognosis

- Pancreatic duct injury: Critical prognostic factor
  - ↑ morbidity, more likely to fail nonoperative management
- Complications
  - Pancreatitis: 3-26%; pseudocyst: 15-35%; fistula: 2-15%; abscess 10-25%

Treatment

- Grades I & II
  - Nonoperative management if hemodynamically stable
  - Total parental nutrition ± percutaneous or endoscopic fluid drainage
  - ± octreotide infusion: Reduces pancreatic secretions & enhances fistula closure in some populations (cancer & chronic pancreatitis patients)
- Grades III-V
  - No consensus regarding nonoperative versus early operative management
  - Nonoperative management
    - 43% failure rate for injuries ≥ grade III
    - ↑ rates of pseudocyst formation & repeat interventional radiology or endoscopic drainage procedures
    - Moderate failure rate for endoscopic duct stenting resulting in delayed operative intervention
    - At risk for post-ERCP pancreatitis, infection, stricture
  - Operative management
    - ↓ length of hospitalization, time to oral feeding, & time to complete resolution
    - ↑ rate of fistula formation; at risk for postoperative leak
    - Options include: Operative drainage (open cystogastrostomy, Roux-en Y), spleen-sparing partial/distal pancreatectomy
- With penetrating trauma, operative exploration often necessary to manage associated injuries
- Overall, 76% of pancreatic injuries managed nonoperatively

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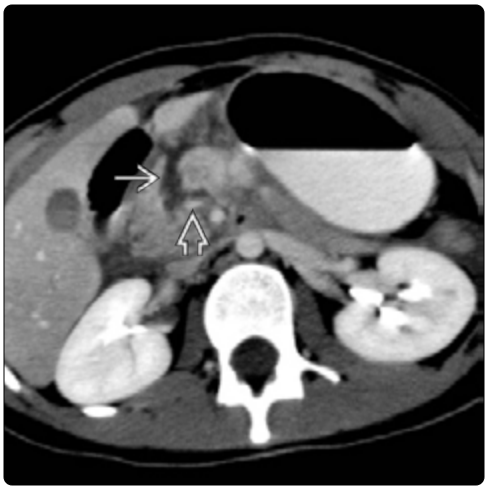
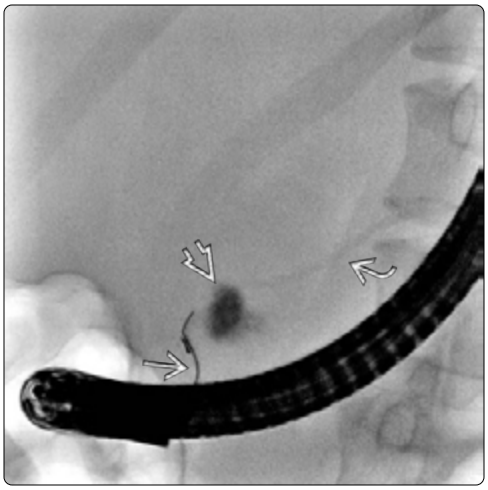
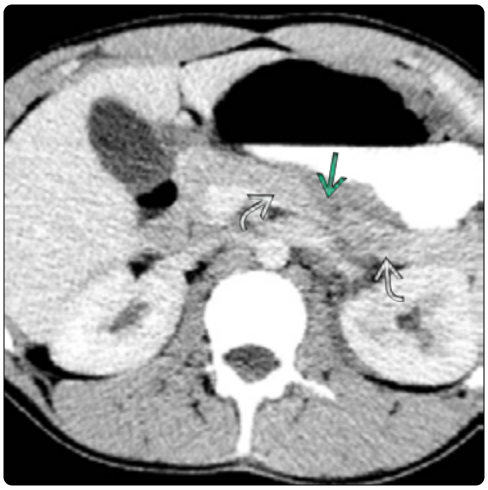
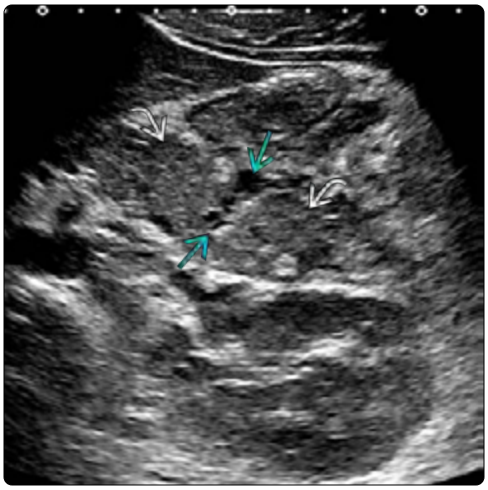


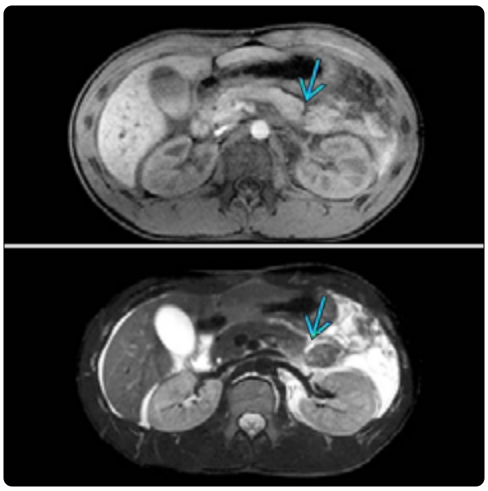


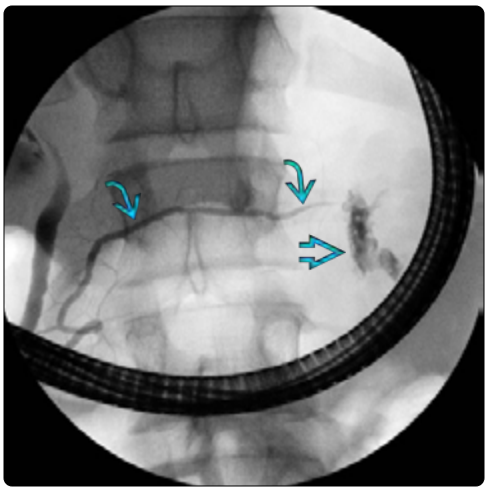
(Left) *Axial CECT from a 14-year-old girl with a soccer injury shows a grade IV proximal transection of the pancreas to the right of the superior mesenteric vein, raising concern for an associated main duct disruption.* **(Right)** *Oblique frontal view from an ERCP in the same patient shows cannulation of the major papilla. There is irregular pooling of contrast at the site of pancreatic neck injury, consistent with main duct disruption. The distal duct is opacified & intact. Stent was subsequently placed.*





(Left) *Axial CECT in a 15-year-old patient after blunt abdominal trauma during a baseball game shows a fluid-attenuation focus interrupting the pancreatic parenchyma, consistent with laceration.* **(Right)** *Axial oblique US through the pancreatic body & tail in the same patient shows the fluid-filled cleft separating the pancreatic parenchyma.*





(Left) *Axial FS 3D T1 GRE (top) & T2 FS (bottom) MRs from the same patient show the site of parenchymal transection with surrounding intraperitoneal & retroperitoneal fluid.* **(Right)** *ERCP in the same patient shows retrograde opacification of the main pancreatic duct with a contrast leak occurring at the site of parenchymal transection, consistent with ductal disruption. This patient ultimately required a spleen-preserving distal pancreatectomy.*

Posttransplant Lymphoproliferative Disease

KEY FACTS

TERMINOLOGY

- Posttransplant lymphoproliferative disorder (PTLD): Spectrum of abnormal lymphoid proliferation in transplant patients

IMAGING

- Any organ system can be affected
- Abdominal region most commonly involved by PTLD, including
  - Gastrointestinal: Bowel wall thickening & dilation, eccentric mass, luminal ulceration, mesenteric stranding, & intussusception
  - Liver: Low-attenuation nodules, periportal infiltration, heterogeneous porta hepatis mass
  - Spleen: Splenomegaly, multiple low-attenuation lesions
  - Kidney: Nephromegaly, multifocal parenchymal masses, heterogeneous renal or pararenal mass
  - Chest: Pulmonary mass, parenchymal nodules, pleural effusion, adenopathy
- Imaging: Lesions often detected by US, CT, or MR
  - DWI MR ↑ conspicuity of nodes against soft tissues
  - PET/CT useful for staging & therapy response

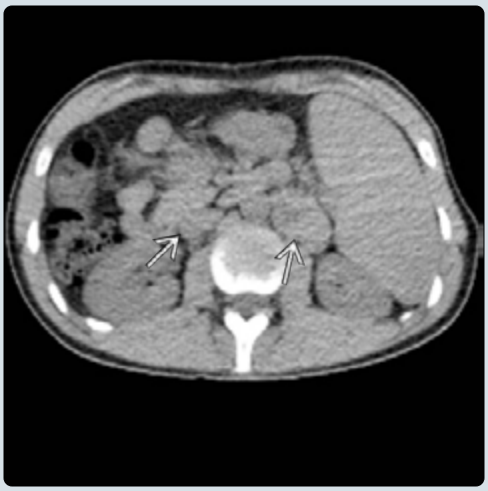
PATHOLOGY

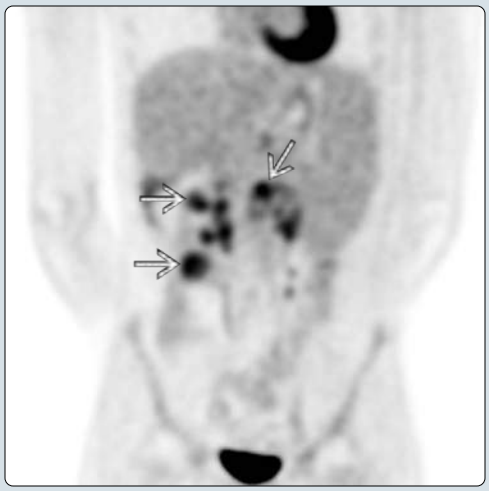
- Risk factors for PTLD: EBV status, young age, type of transplanted organ (& degree of immunosuppression)
  - Greatest risk: Donor EBV positive, recipient EBV negative
  - 50-80% of PTLD biopsies positive for EBV in tumors cells
  - Small bowel transplant at highest risk (20%)
  - Kidney transplant at lowest risk (2%)
- WHO classification: Early lesions, polymorphic, monomorphic, classic Hodgkin lymphoma-like

CLINICAL ISSUES

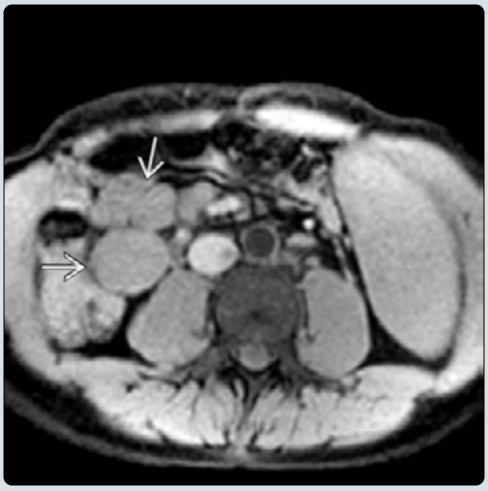
- Most common malignancy in pediatric transplant patients
- Universally fatal if not treated
- Treatment: Reduce immunosuppression, add chemotherapy for lymphoma

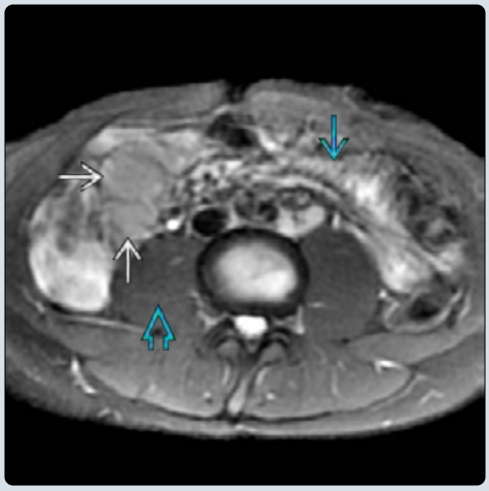
(Left) *Axial NECT in a young adult status post liver transplant shows multiple enlarged retroperitoneal lymph nodes. New nodal masses in patients with a history of transplant should be considered posttransplant lymphoproliferative disease (PTLD) until proven otherwise.* **(Right)** *Coronal FDG PET from a PET/CT in the same liver transplant patient shows multiple nodal foci of FDG uptake. PET/CT is a useful modality to determine the extent of disease in PTLD.*





(Left) *Axial T1 FS MR in the same liver transplant patient shows nodal masses in the right abdomen. PTLD is most common in patients with intestinal transplant & least common in patients with renal transplant.* **(Right)** *Axial T2 FS MR in the same patient shows that the nodal masses are hyperintense to muscle & nearly isointense to bowel. Nodal masses of this size & smaller can be difficult to separate from adjacent bowel loops without DWI or PET imaging.*





TERMINOLOGY

Abbreviations

- Posttransplant lymphoproliferative disorder (PTLD)
- Ebstein-Barr virus (EBV)

Definitions

- Spectrum of abnormal lymphoid proliferation in transplant patients

IMAGING

General Features

- Best diagnostic clue
  - Lymphadenopathy or solid mass occurring virtually anywhere in organ transplant patient
- Location
  - Any organ system can be affected
  - Abdomen most commonly involved by PTLD
    - Transplanted organ, gastrointestinal (GI) tract, & liver most commonly affected
  - Other sites: Tonsils, lungs, kidneys, brain, muscle

Radiographic Findings

- Airway: Enlargement of adenoids & palatine tonsils
- Chest: Pulmonary nodule(s), adenopathy, consolidation, pleural effusion

CT Findings

- CECT
  - Abdomen
    - GI tract
      - Focal or uniform bowel wall thickening, bowel dilation, eccentric bowel wall mass, luminal ulceration, mesenteric stranding, & intussusception
      - More common in distal small bowel & proximal colon
    - Liver
      - Hepatomegaly, low-attenuation nodules, infiltrative pattern along portal vessels, heterogeneous porta hepatis mass
    - Spleen
      - Splenomegaly, multiple low-attenuation lesions
    - Kidney
      - Nephromegaly, multifocal parenchymal masses, heterogeneous renal or pararenal mass
  - Lymph nodes
    - Discrete, homogeneous enlarged lymph node or conglomeration of enlarged lymph nodes
  - Chest
    - Pulmonary mass, parenchymal nodules, pleural effusion, adenopathy
    - Parenchymal nodules usually have homogeneous density, can have halo of ground-glass opacity
  - Neck
    - Nonspecific adenopathy & enlarged tonsils
  - Brain
    - High-attenuation, ring-enhancing parenchymal lesions surrounded by vasogenic edema

MR Findings

- Abdomen
  - Liver
    - Multiple lesions hypointense on T1WI & slightly hyperintense on T2WI with mild peripheral enhancement
  - Spleen
    - Splenomegaly, multiple lesions hypointense to spleen on T1WI & iso- to hypointense on T2WI
  - Kidney
    - Discrete renal mass or diffuse infiltrative lesion causing renal enlargement
      - T1WI: Isointense to kidney
      - T2WI: Slightly hypointense
    - Unilateral or bilateral
    - Hydronephrosis if mass obstructive
  - Discrete or conglomerate nodal masses, often isointense to bowel wall
    - Hyperintense on DWI → stand out against bowel
- Brain
  - Solitary or multiple periventricular mass(es) with vasogenic edema
    - T1WI: Heterogeneously hypo- to isointense with foci of hyperintense hemorrhage
    - T2WI: Mostly hyperintense
    - T1WI C+: Peripheral enhancement

Ultrasonographic Findings

- Solid masses with variable echogenicity & internal vascularity

Nuclear Medicine Findings

- PET
  - ↑ FDG uptake in lesions
  - PET/CT useful for staging & to determine response to therapy

Imaging Recommendations

- Best imaging tool
  - Body: Disease often detected on US, CT, or MR; PET/CT to stage & follow-up disease
  - Brain & spine: MR ± contrast
- Protocol advice
  - DWI helpful in identifying body lesions

DIFFERENTIAL DIAGNOSIS

Graft-vs.-Host Disease

- Allogenic bone marrow transplant patients
- Lymphocytes from donor attack recipient tissues
- Often involves bowel, liver, & skin
  - May have similar symptoms, including abdominal pain & diarrhea
  - CECT shows featureless ribbon-like hyperenhancing bowel
- Treatment: ↑ immunosuppression

Colitis

- Types: Inflammatory, pseudomembranous, neutropenic, infectious
- Symptoms: Fever, diarrhea, abdominal pain
- CECT: Circumferential colonic wall thickening (accordion sign)
- Abdominal radiograph: "Thumbprinting" of colonic wall

Fungal Infection

- *Candida* most common type to infect GI tract, liver, & spleen
- Microabscess in liver, spleen, or kidneys
  - US: Multiple small hypoechoic lesions, some with hyperechoic centers → target or bull's-eye appearance
  - CT: Numerous low-attenuation lesions
  - MR: Hypointense on T1WI & hyperintense on T2WI

Abscess/Septic Emboli

- Visceral &/or soft tissue lesion(s) with irregular thick rim enhancement, surrounding edema

PATHOLOGY

General Features

- Risk factors for PTLD
  - EBV status at time of transplant
    - Greatest risk: Donor EBV positive, recipient EBV negative
    - 50-80% of PTLD biopsies positive for EBV in tumors cells
      - In immunocompromised host, EBV can lead to uncontrolled B-cell expansion
  - Young age
    - Patients more likely to be EBV negative
    - Patients will have longer exposure to immunosuppression
  - Type of transplanted organ
    - Small bowel transplant at highest risk (~ 20%)
    - Kidney transplant at lowest risk (2%)

Staging, Grading, & Classification

- WHO classification
  - Early lesions
    - Similar to infectious mononucleosis
    - Reactive plasmacytic hyperplasia without destruction of normal lymph node or tissue architecture
  - Polymorphic
    - Mixture of monoclonal B-cell lymphocytes & polyclonal T-cell lymphocytes
    - B-lymphocytes EBV positive
  - Monomorphic
    - Exclusively monoclonal
    - Divided into diffuse large B-cell lymphoma, Burkitt lymphoma, & plasma cell myeloma
    - Usually EBV positive
  - Classic Hodgkin lymphoma-like
    - Rare in childhood

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Lymphadenopathy, fever, weight loss, tonsillitis, transplant dysfunction
- Other signs/symptoms
  - Depends on location of mass
    - Airway: Noisy breathing, snoring, changes in voice
    - Abdomen: Abdominal pain, distention, bloody stool, intussusception, multisystem organ failure
    - Chest: May be asymptomatic with incidental opacity on chest radiograph, fever, ↓ pulmonary function tests
    - Head & neck: Mononucleosis-like symptoms including fever, malaise, adenopathy, pharyngitis
    - Neurologic: Seizure, focal neurological deficit

Demographics

- Age
  - Children > adults
- Epidemiology
  - Most common malignancy in pediatric transplant patients (52%)
  - Incidence of PTLD in children much higher than adults
  - Incidence ↑ with ↑ immunosuppression
  - Incidence ↑ in transplants that require more immunosuppression
    - Highest in small bowel transplant & heart-lung transplant, lowest in kidney & liver transplants

Natural History & Prognosis

- Early-onset more likely to be due to EBV infection
- Universally fatal if not treated
- Prognosis depends on degree of immunosuppression
  - More immunosuppressed = poorer prognosis

Treatment

- Reduced immunosuppression
  - Upon EBV seroconversion or ↑ EBV viral load
  - Works best in early lesions & polymorphic PTLD
  - Must balance with risk of transplant rejection
- Chemotherapy
  - For overt malignancy (Burkitt lymphoma)
  - If inadequate response to reduced immunosuppression

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Adenopathy or mass in transplant patient = PTLD until proven otherwise

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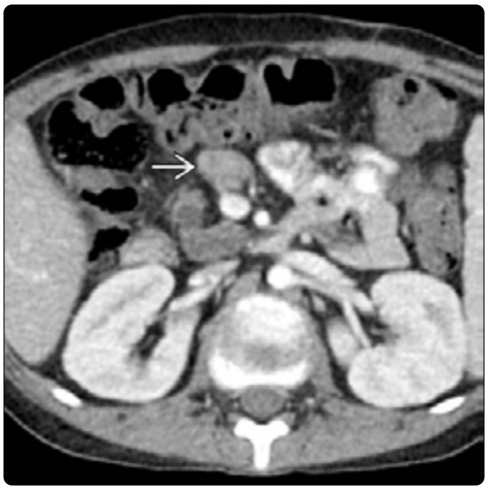
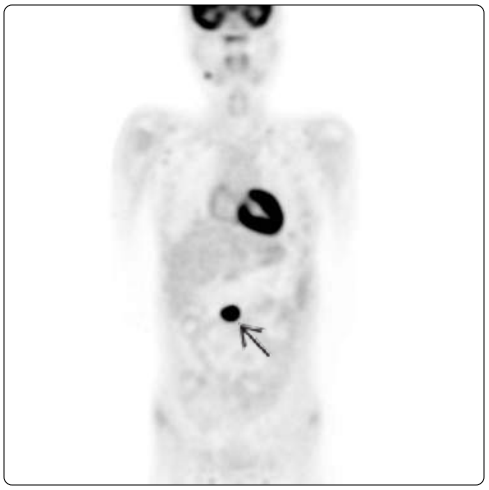
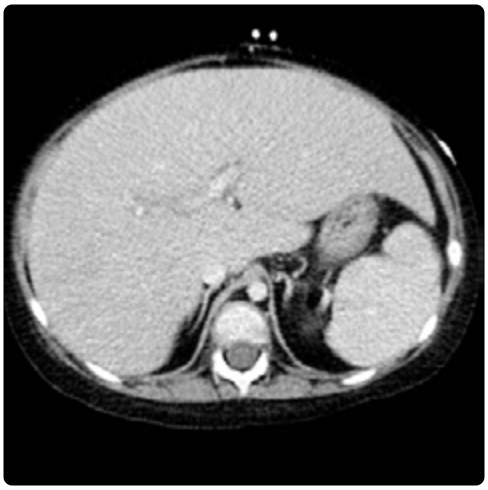
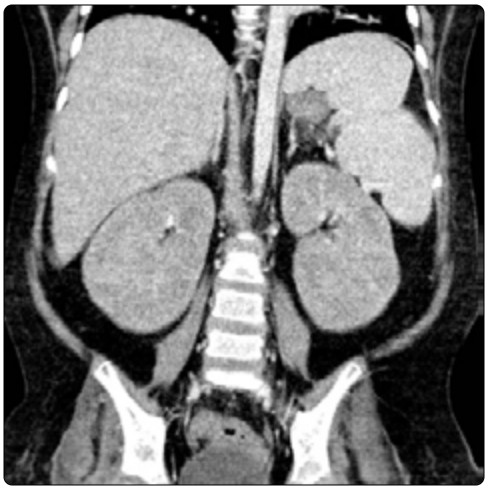


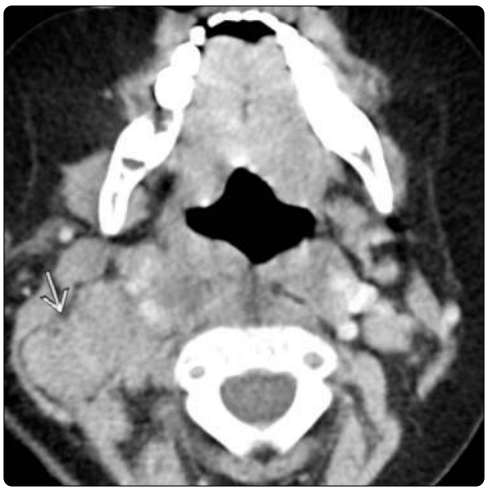


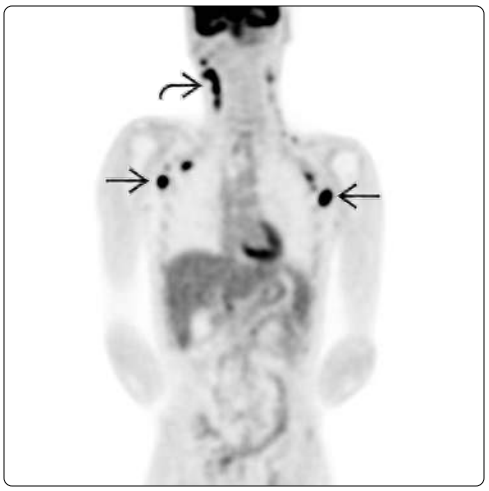
(Left) *Axial CECT in a child with Burkitt lymphoma-type PTLD (after a lung transplant) shows a mildly prominent enhancing soft tissue mass in the mesentery. Burkitt lymphoma-type PTLD represents a monomorphic PTLD.* **(Right)** *Coronal PET/CT in the same patient shows the nodal mass to be FDG avid. There are 4 types of PTLD: Early lesions, polymorphic, monomorphic, & Hodgkin lymphoma-like.*





(Left) *Axial CECT of the abdomen in a patient with PTLD after bone marrow transplant shows hepatosplenomegaly.* **(Right)** *Coronal CECT in the same patient with hepatosplenomegaly shows diffuse enlargement & poor enhancement of the kidneys. PTLD can affect any organ & can appear as a solid mass, organomegaly, or infiltrative tumor.*





(Left) *Axial CECT in the same patient shows cervical adenopathy. Cervical lymph nodes are a common site of PTLD involvement. Other common sites of disease include the liver, GI tract, & tonsils.* **(Right)** *Coronal image from a PET/CT in a patient with monomorphic PTLD after a bone marrow transplant shows bilateral axillary & right cervical adenopathy with increased FDG uptake. Monomorphic PTLD can be divided into diffuse large B-cell lymphoma, Burkitt lymphoma, & plasma cell myeloma.*

Pseudomembranous Colitis

KEY FACTS

TERMINOLOGY

- Colonic inflammation due to *Clostridium difficile* & its toxins A & B → epithelial necrosis & pseudomembranes

IMAGING

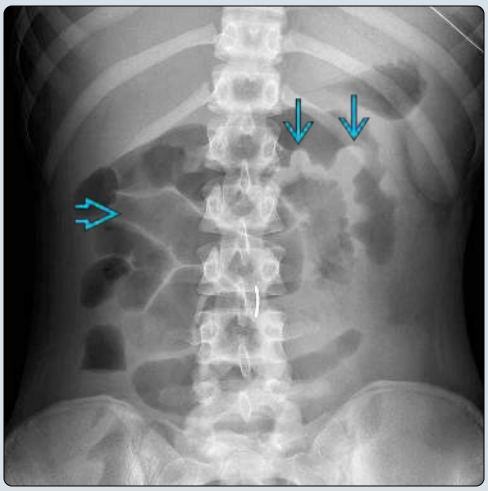
- Classic appearance: Pancolitis with marked wall thickening
- **Radiographs**
  - Thumbprinting ± colonic or small bowel ileus
- **CECT**
  - Accordion sign: Alternating bands of ↑ & ↓ attenuation of colon due to oral contrast interdigitating between edematous haustra
    - Lumen appears stellate in cross section
  - Target sign: Rings of hyperenhancing mucosa & lower attenuation submucosal/muscular edema
  - Pericolonic stranding usually very mild, ± ascites
- **US**
  - US accordion sign: Echogenic bowel contents between hypoechoic thickened wall
  - Pseudomembranes: Linear hyperechogenicity
  - ± ascites
- Paucity of colonic gas with luminal narrowing

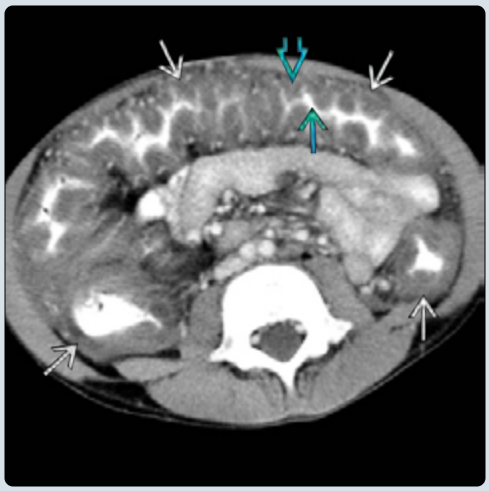
PATHOLOGY

- *C. difficile* overgrowth (usually due to antibiotic therapy) → toxin A & B release
- Inflamed colon with discrete or confluent, raised, yellow-white plaque pseudomembranes on endoscopy

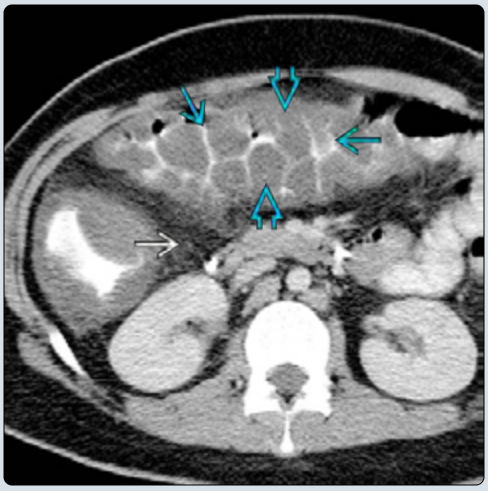
CLINICAL ISSUES

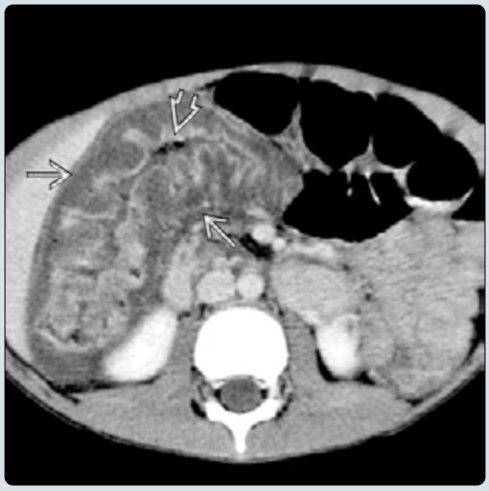
- Most common symptoms: Watery or bloody diarrhea, dehydration, abdominal pain, leukocytosis, fever
- Primary treatments include: Cessation of inciting antibiotic with addition of metronidazole or vancomycin
- If treated early, full recovery expected
  - 20% recurrence rate despite appropriate therapy
- 3% progress to toxic megacolon





(Left) *Supine abdominal radiograph in a 15 year old with known pseudomembranous colitis (PMC) demonstrates thumbprinting along the colon due to haustral thickening. A small bowel ileus is also present.* **(Right)** *Axial CECT in a 10-year-old child with fever, abdominal pain, & diarrhea demonstrates findings of PMC with pancolonic marked wall thickening. The alternating bands of hyperdense contrast between the thickened fluid-density haustra create the accordion sign.*





(Left) *Axial CECT shows marked pancolitis with enteric contrast trapped between thickened haustral folds (the accordion sign). Note the minimal pericolonic fat infiltration, which is typical with PMC.* **(Right)** *Axial CECT demonstrates asymmetric pancolitis with the greatest colonic wall thickening & mucosal enhancement seen in the right & transverse colon. Involvement limited to the proximal colon can be seen in 10-20% of PMC cases.*

TERMINOLOGY

Definitions

- Inflammation of colon by *Clostridium difficile* toxins A & B

IMAGING

General Features

- Location
  - Pancolitis typical
  - Rectum & sigmoid colon: Up to 90% of cases
  - Proximal colon only: 10-20%

Radiographic Findings

- Low sensitivity: Up to 68% can be normal
- Paucity of colonic gas with luminal narrowing
  - Small amounts of gas in lumen outline thickened haustra (thumbprinting)
    - En face view of affected loop may show stellate lucency of gas in narrowed lumen outlining haustra
  - Haustral thickening may be nodular
- ± colonic & small bowel ileus
- ± separation & centralization of bowel loops by ascites
- Toxic megacolon in fulminant cases
  - Colonic dilation, right > left, usually > 6 cm
  - Loss of haustral pattern
  - Pneumoperitoneum if perforation occurs

CT Findings

- CECT
  - Colonic wall thickening
    - Usually marked; average > 10 mm
    - Smooth, plaque-like, nodular or polypoid
  - Accordion sign: Small amounts of oral contrast (or hyperenhancing mucosa) interdigitate between very thick/edematous haustra → alternating bands of ↓ & ↑ attenuation when viewed in long axis
    - Narrowed lumen may appear stellate in cross section
  - Target sign: In cross section/short axis, thickened colonic wall shows rings of hyperenhancing mucosa, lower attenuation submucosal/mural edema, & possible outer layer of serosal enhancement
  - Pericolonic stranding usually mild due to mucosal & submucosal nature of disease; ± ascites
  - ± pneumatosis &/or pneumoperitoneum if severe

Ultrasonographic Findings

- Marked colonic wall thickening effacing lumen
  - Hypoechoic mural edema most pronounced
  - ± loss of wall stratification (gut signature)
  - US accordion sign: Echogenic bowel contents between less echogenic, but thickened, wall
- Pseudomembranes may appear as linear hyperechogenicity
- ± ascites

Imaging Recommendations

- CECT with IV & oral contrast

DIFFERENTIAL DIAGNOSIS

Infectious Colitis

- May be indistinguishable from pseudomembranous colitis

Crohn Disease

- Skip lesions; ileocolic involvement typical
- Strictures, fistulas, abscesses, "creeping fat"

Ulcerative Colitis

- Pancolitis with symmetric wall thickening < 10 mm

Typhlitis (Neutropenic Colitis)

- Usually isolated to right colon

Benign Pneumatosis in Older Children

- History of steroids or bone marrow transplant typical
- Often lacks wall thickening, free fluid

PATHOLOGY

General Features

- *C. difficile* colonization, overgrowth, & toxin production
  - Overgrowth usually due to antibiotic therapy but can be due to chemotherapy, abdominal surgery, obstruction, uremia, hypotension, bowel hypoperfusion
  - Toxins A & B (enterotoxin & cytotoxin) mediate disease

Gross Pathologic & Surgical Features

- Inflamed colon with discrete or confluent, raised, yellow-white plaques (pseudomembranes) on endoscopy

CLINICAL ISSUES

Presentation

- Most common symptoms: Watery or bloody diarrhea, dehydration, abdominal pain, leukocytosis, fever

Natural History & Prognosis

- If treated early, full recovery expected
- Overall mortality up to 3.5%
- 3% progress to toxic megacolon
  - High risk for perforation
  - Mortality very high, reported in 38-80%
- 20% recurrence rate despite appropriate therapy

Treatment

- Cessation of inciting antibiotic, start replacement if needed
- 1st-line therapy with metronidazole or vancomycin
- Fecal microbiota transplantation may be considered for refractory/recurrent cases
- Surgery for severe disease resulting from toxic megacolon, perforation, or if rapidly progressing or refractory

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Neutropenic Colitis

KEY FACTS

TERMINOLOGY

- Necrotizing inflammatory process of bowel most commonly affecting right colon of neutropenic patients (with absolute neutrophil count < 500 cells/μL)

IMAGING

- Right colon most commonly affected but distal colon, appendix, & small bowel can be involved
- Bowel wall thickening: Circumferential & symmetric, typically low attenuation on CECT
- Mucosal hyperenhancement less common than in graft-vs.-host disease (GVHD)
- Pericolic fluid & fat stranding/hyperechogenicity
- Pneumatosis more common compared to other colitides & GVHD
- ± small bowel ileus; ± pneumoperitoneum with perforation
- Modality of choice: US vs. CECT
  - CECT may be more sensitive for early changes & complications (necrosis, perforation, abscess)
  - US more accurate for measurement of bowel wall thickness & uses no ionizing radiation

TOP DIFFERENTIAL DIAGNOSES

- Appendicitis
- Other colitides: Infectious, pseudomembranous, ischemic
- Acute graft-vs.-host disease
- Benign pneumatosis of older children

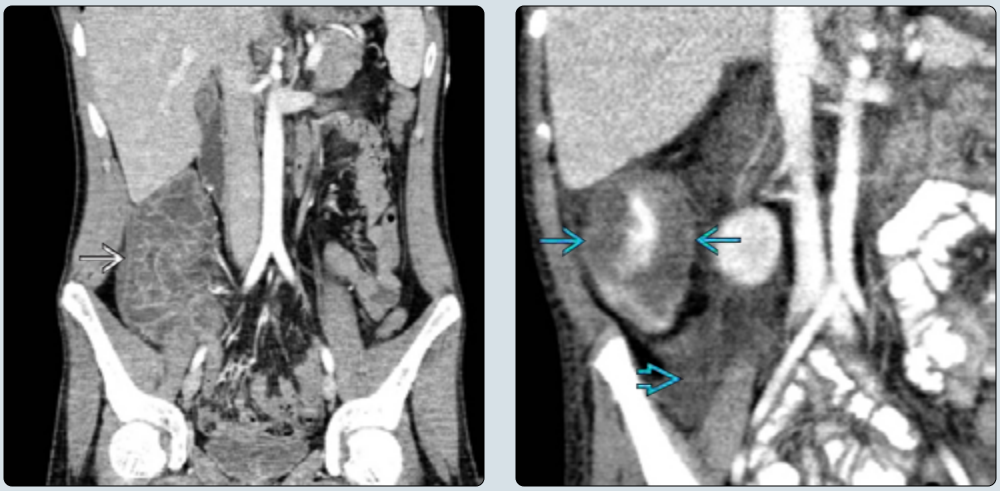
PATHOLOGY

- Multifactorial mucosal injury → bacterial/fungal bowel wall invasion → inflammation, hemorrhage, edema → ulceration, transmural necrosis & perforation

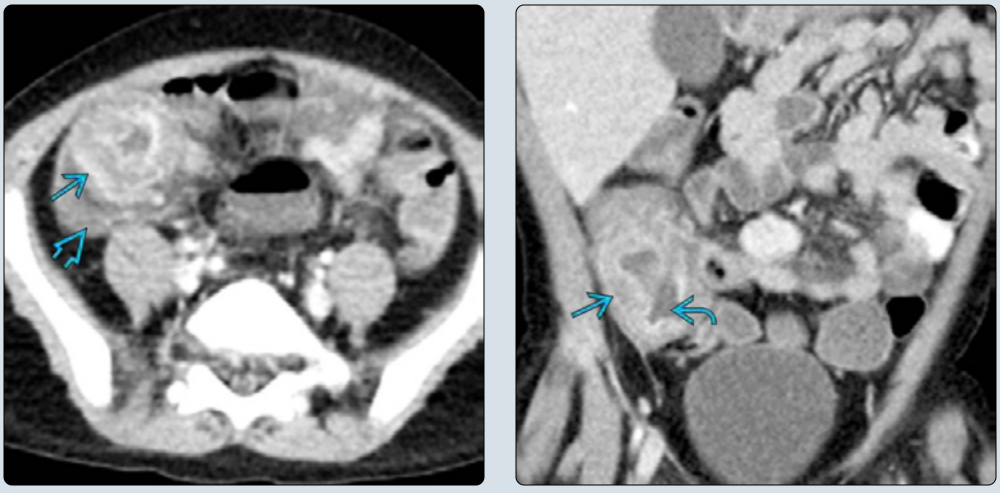
CLINICAL ISSUES

- Clinical triad: Neutropenia, abdominal pain, fever
- Uncomplicated cases: Bowel rest, broad spectrum IV antibiotics (including antifungal), ± granulocyte colony-stimulating factor
- Surgery for perforation, peritonitis, GI bleeding, obstruction

(Left) *Coronal CECT shows marked wall thickening of the cecum & ascending colon in a patient with a history of osteosarcoma status post chemotherapy. The patient presented with neutropenia, vomiting, nausea, & abdominal pain.* **(Right)** *Coronal CECT of a 5-year-old patient with a history of acute myelogenous leukemia presenting with new abdominal pain & neutropenia demonstrates marked edema of the ascending colon as well as free fluid. The findings are consistent with neutropenic colitis.*



(Left) *Axial CECT of a 3-year-old patient with acute lymphocytic leukemia presenting with fever, abdominal pain, & neutropenia shows findings typical of neutropenic colitis, including wall thickening in the cecum & ascending colon & mild associated pericolonic stranding.* **(Right)** *Coronal CECT from the same patient with neutropenic colitis shows bowel wall thickening of the right colon with a fluid-filled lumen. The remaining loops of bowel were normal.*



TERMINOLOGY

Definitions

- Necrotizing inflammatory process of bowel most commonly affecting right colon of neutropenic patients (with absolute neutrophil count < 500 cells/μL)

IMAGING

General Features

- Best diagnostic clue
  - Wall thickening of right colon + pericolic stranding in neutropenic patient with abdominal pain &/or fever without other etiology identified
- Location
  - Cecum & ascending colon most common
  - ± other colonic segments, appendix, & small bowel

Radiographic Findings

- Paucity of bowel gas in right lower quadrant
- "Soft tissue mass" in right lower quadrant
- "Thumbprinting" (thickened haustra)
- ± pneumatosis
- ± small bowel ileus

CT Findings

- Circumferential & symmetric bowel wall thickening > 3 mm, predominantly of ↓ attenuation due to muscularis edema
- ± mucosal & serosal hyperenhancement
- Cecal luminal distention or narrowing
- Pericolic fluid & fat stranding
- ± pneumatosis
- ± dilated adjacent small bowel loops (ileus)
- Pneumoperitoneum if perforation occurs

Ultrasonographic Findings

- Bowel wall thickening > 3 mm
  - Hypoechoic muscularis layer & echogenic mucosal layer
- Pericolic fluid & echogenic fat
- ± echogenic pneumatosis tracking circumferentially in wall

DIFFERENTIAL DIAGNOSIS

Crohn Disease

- Mouth to anus may be affected but ileocolic most common
- Skip lesions (up to 90%) with transmural inflammation
- String sign, strictures, sinus tracts, fistulas, perianal disease

Appendicitis

- Dilated noncompressible appendix + surrounding inflammation
- Bowel typically normal unless appendix perforated

Infectious Colitis

- Nonspecific imaging findings; many possible organisms

Pseudomembranous Colitis

- Typically pancolitis; isolated to right colon in 10-20%
- Marked wall thickening (mean > 10 mm) & nodularity

Acute Graft-vs.-Host Disease

- Prior bone marrow transplant
- Mucosal hyperenhancement & dilated bowel common
- Mild degree of bowel wall thickening
- Small bowel classically featureless & ribbon-like

Benign Pneumatosis of Older Children

- Most commonly occurs in patients on steroids or after bone marrow transplant
- Extensive colonic involvement common
- Bowel wall thickening & free fluid uncommon

Ischemic Colitis

- Uncommon in children but must be considered in ill, complex patients with pneumatosis
- Bowel wall thickening with ↓ enhancement + free fluid

PATHOLOGY

General Features

- Multifactorial etiology: Immunocompromise, therapy toxicity, bowel ischemia, intramural hemorrhage, neoplastic cell infiltration of bowel wall
- Mechanism: Mucositis & mucosal injury → bacterial/fungal invasion of bowel wall → inflammation, hemorrhage, edema → ulceration, transmural necrosis, & perforation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms: Neutropenia, abdominal pain, fever, diarrhea
- Risk factors: Leukemia, hematologic & solid malignancies, solid organ & bone marrow transplant, immunocompromised state, recent chemotherapy
- Incidence: 0.4-6% in pediatric oncology patients

Natural History & Prognosis

- Early diagnosis & treatment essential to reduce risk of transmural necrosis & perforation
- Mortality ↓ over last 3 decades, recently 0-2.5%

Treatment

- Medical management for uncomplicated cases
  - Bowel rest & decompression
  - Broad spectrum IV antibiotics (including antifungal)
  - ± granulocyte colony-stimulating factor
- Surgery for perforation, peritonitis, active GI bleeding, obstruction
  - Commonly hemicolectomy + defunctioning ileostomy

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KEY FACTS

TERMINOLOGY

- Disease of allogenic bone marrow transplant (BMT) or stem cell transplant (SCT) recipients
- Donor T lymphocytes cause damage to epithelial cells lining recipient target organs

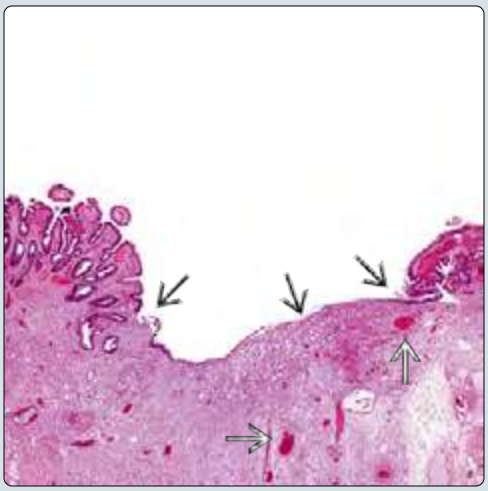
IMAGING

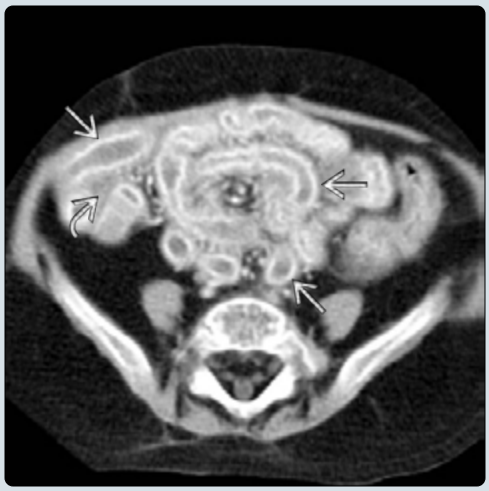
- Best imaging clue: Diffuse bowel wall thickening with smooth central hyperenhancement
  - Corresponds to thin layer of vascular granulation tissue replacing destroyed mucosa
- Radiography
  - Mildly dilated bowel with air-fluid levels
  - ± evidence of bowel wall thickening, pneumatosis
- SBFT: Shows wall thickening as luminal narrowing & separation of small bowel loops
  - Smooth, featureless appearance of intestinal lumen diffusely (ribbon-like)
- CECT
  - Diffusely abnormal bowel with wall thickening & smooth central hyperenhancement
  - Mesenteric inflammatory changes
  - ± hepatomegaly, ascites

CLINICAL ISSUES

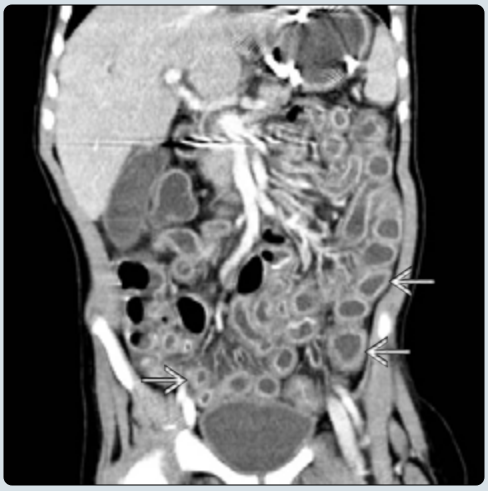
- Affects 30-70% of patients with allogenic BMTs or SCTs
  - Up to 40%, even with complete HLA matching
- Older children have ↑ likelihood of graft-vs.-host disease (GVHD)
- Acute GVHD (< 100 days post transplant)
  - Most commonly involves skin
  - GI tract & liver also commonly involved
- Chronic GVHD (> 100 days post transplant)
  - Features of autoimmune disease
  - Can also affect muscles/joints, lungs, eyes, kidneys
- Treatment: ↑ immunosuppression

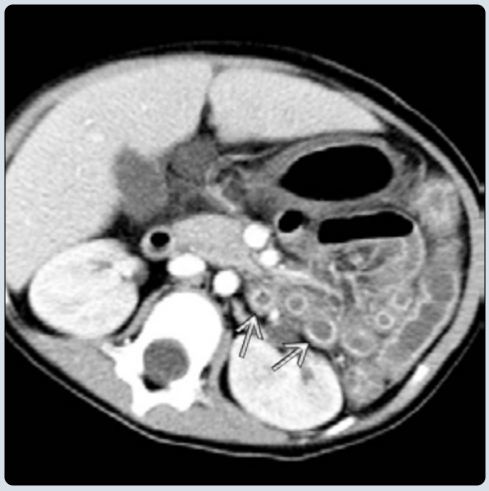
(Left) *Low-power H&E stain of the intestine shows an ulcerated mucosal surface ⇨. Note the underlying blood vessels ➡, which may predispose to bleeding.* **(Right)** *Axial CECT shows fluid-filled, thickened loops of bowel with central hyperenhancement ➡ & mesenteric inflammation ➡. The mucosal surface is relatively smooth & featureless without typical folds (as this layer actually represents vascular granulation tissue replacing destroyed mucosa), a classic appearance of graft-vs.-host disease (GVHD).*





(Left) *Coronal CECT shows multiple fluid-filled loops of abnormal small bowel throughout the abdomen. Note the mildly thickened bowel walls ➡ with smooth central hyperenhancement that is typical of GVHD.* **(Right)** *Axial CECT in the same child shows multiple loops of fluid-filled small bowel with mild wall thickening ➡ & smooth central hyperenhancement, which is due to granulation tissue replacing destroyed mucosa.*





TERMINOLOGY

Abbreviations

- Graft-vs.-host disease (GVHD)

Definitions

- Disease of allogenic bone marrow transplant (BMT) or stem cell transplant (SCT) recipients
- Donor T lymphocytes cause damage to epithelial cells lining recipient target organs (gut, liver, skin)
- Donor bone marrow or stem cells = graft
- Recipient = host

IMAGING

General Features

- Best diagnostic clue
  - Diffuse bowel wall thickening with central, smooth (featureless) hyperenhancement
    - Enhancing layer represents vascular granulation tissue replacing destroyed mucosa
- Location
  - Skin, GI tract, liver
- Size
  - Diffuse intestinal involvement from duodenum to rectum

Radiographic Findings

- May resemble adynamic ileus
  - Multiple mildly dilated small & large bowel loops
  - Air-fluid levels on decubitus/upright images
- ± evidence of bowel wall thickening, pneumatosis

Fluoroscopic Findings

- SBFT may show small bowel wall thickening as luminal narrowing & separation of loops
- Classically shows ribbon-like appearance of smooth, featureless intestinal lumen

CT Findings

- CECT
  - Diffuse bowel abnormality from duodenum to rectum
    - Fluid-filled & mildly dilated
    - Bowel wall thickening/edema
    - Ring-like (in cross section), smooth, featureless central enhancement of mucosa/granulation tissue
      - Hyperenhancing, smooth central lining due to vascular granulation tissue replacing destroyed mucosa
  - Mesenteric inflammatory changes
  - Hepatomegaly
  - Ascites

MR Findings

- Findings similar to CECT
- Techniques described with enterography or 3 Tesla
  - ~ 80% sensitivity

Ultrasonographic Findings

- Grayscale ultrasound
  - Fluid-filled loops of thick-walled bowel
  - Ascites
- Color Doppler
  - Abnormal bowel may have ↑ blood flow due to inflammation

Nuclear Medicine Findings

- PET/CT
  - ~ 80% sensitive, 90% specific

Imaging Recommendations

- Best imaging tool
  - CT with IV contrast
    - Often performed to help differentiate GVHD from other causes of abdominal sepsis in immunocompromised BMT/SCT recipients
  - May show potential complications
    - Perforation & abscess formation
- Protocol advice
  - CECT abdomen & pelvis
    - Positive oral contrast may obscure enhancement pattern
      - Consider negative oral contrast

DIFFERENTIAL DIAGNOSIS

Pseudomembranous Colitis

- Usually pancolitis with marked wall thickening
- Overgrowth of *Clostridium difficile* → enterotoxin production
  - Commonly results from elimination of normal flora by specific antibiotics

Typhlitis

- Cecal inflammation & necrosis in immunocompromised patient

Cytomegalovirus Colitis

- Nonspecific bowel wall thickening
- Adjacent inflammatory changes

Radiation Enteritis

- Thickened bowel wall & adherence of adjacent loops
- Infiltration of adjacent mesenteric or retroperitoneal fat
- Strictures → bowel obstruction

Posttransplant Lymphoproliferative Disorder

- Lymphoid proliferation secondary to chronic immunosuppression
- Associated with Epstein-Barr virus infection or reactivation
- Circumferential bowel wall thickening ± aneurysmal dilation, low-attenuation liver lesions, lymph node enlargement

Shock Bowel (Hypoperfusion Complex)

- Diffuse bowel wall thickening + mucosal hyperenhancement
- Folds accentuated (not featureless like GVHD)
- Additional findings: Hyperenhancing adrenal glands, small caliber aorta, collapsed inferior vena cava
- Relevant clinical history typically clear

PATHOLOGY

General Features

- Complication of allogenic transplants

- Histocompatibility is major predictor of incidence & severity of GVHD
- Development of GVHD requires
  - Immunocompetent cells (T cells) within graft
  - Recipient (host) tissue antigens
    - Donor T cells react to mismatched host antigens & attack host tissues
      - Even with HLA-matched donor recipient, incidence of GVHD still up to 40% due to minor mismatches
    - Recipient incapable of immune response to stop donor T cells from damaging tissues
    - Host inflammatory response also plays role in tissue damage
- May occur as acute or chronic form
  - Distinction based on time from transplant (< or > 100 days)

Staging, Grading, & Classification

- Grade I-IV (mild-severe)
  - Based on involvement of skin, GI tract, & liver

Gross Pathologic & Surgical Features

- Biopsy helpful in confirming diagnosis

Microscopic Features

- Extensive crypt cell necrosis
- In severe cases, diffuse large & small bowel ulceration & mucosal destruction
- Mucosal replacement with thin layer of highly vascular granulation tissue
  - Accounts for central enhancement of bowel wall

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Skin most commonly affected, usually 1st site of involvement in acute GVHD
    - Typical maculopapular rash, ± pain, pruritus
  - GI tract
    - Diarrhea, anorexia, vomiting, pain
    - Rarely GI bleeding in severe cases
      - Due to mucosal ulceration
  - Liver
    - Cholestatic hyperbilirubinemia: Can be clinically indistinguishable from other transplant complications
      - Hepatic venoocclusive disease
      - Drug toxicity
      - Viral infection
    - Hepatomegaly
- Other signs/symptoms
  - Chronic GVHD: More diverse manifestations
    - Often affects skin, mucosal surfaces
    - Can cause oral, ocular, neuromuscular, pulmonary, & renal abnormalities

Demographics

- Age
  - Age at BMT directly related to GVHD
    - Older children have ↑ likelihood of GVHD
- Gender
  - M = F
  - Donor from opposite sex: ↑ incidence
- Epidemiology
  - Affects 30-70% of patients with allogenic transplant of immunocompetent lymphocytes

Natural History & Prognosis

- Prompt diagnosis important
  - Further treatment: ↑ immunosuppression
  - Differentiate from infection (also common after BMT)
- Overall prognosis good: 80-85% survival
- Mortality related to secondary causes
  - Overwhelming infection, hepatic failure, & hemorrhage
- Mortality directly related to severity of disease
  - Stage IV disease = only 5% overall survival
- Use of selectively depleted alloreactive T lymphocytes & umbilical cord blood ↓ incidence of GVHD

Treatment

- Steroids
- ↑ immunosuppression: Cyclosporine, tacrolimus, sirolimus
- Novel therapies: Anticytokine antibodies, monoclonal antibodies

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Diffuse wall-thickening with central featureless hyperenhancement characteristic for GVHD

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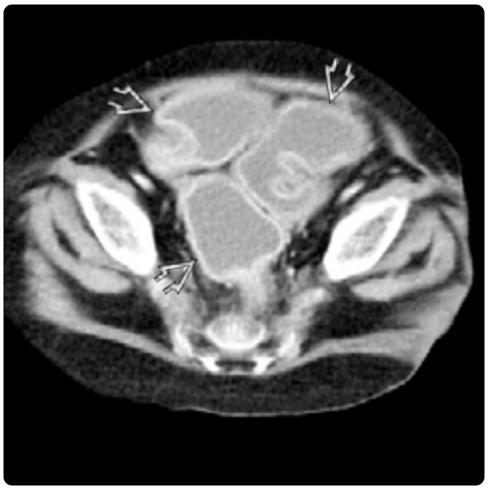
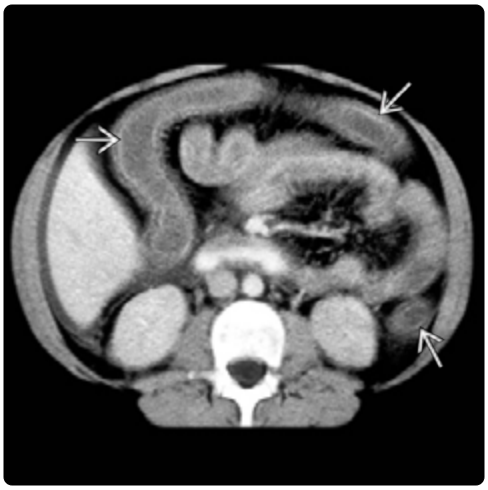
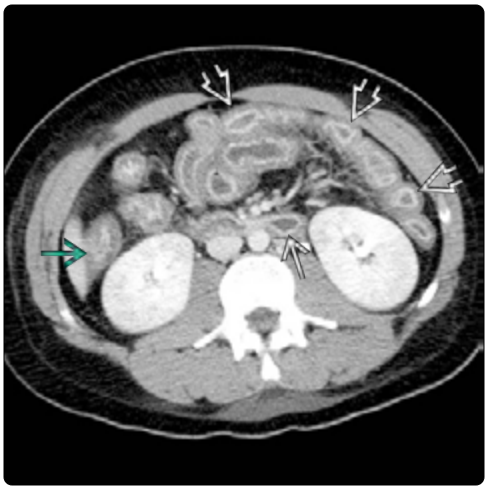
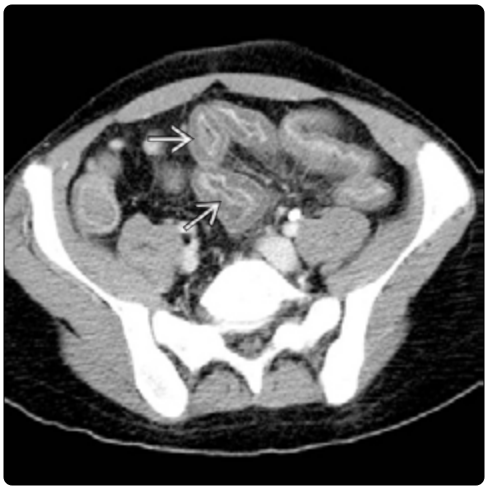


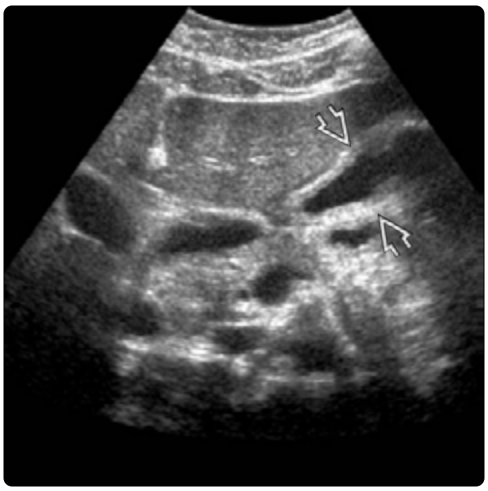


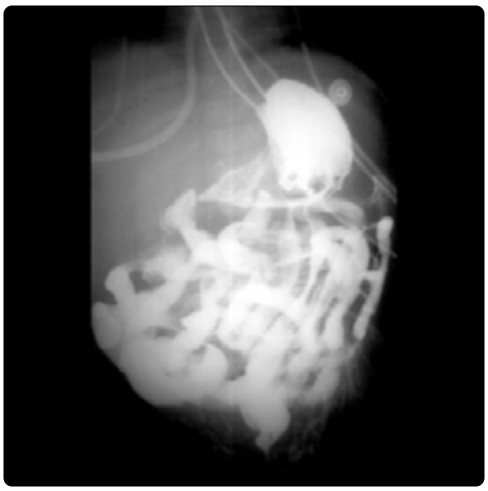
(Left) *Axial CECT shows moderate dilation of multiple fluid-filled loops of large bowel with thin, smooth central hyperenhancement.* **(Right)** *Axial CECT shows multiple loops of dilated, featureless, thickened bowel with a characteristic pattern of central hyperenhancement.*





(Left) *Axial CECT shows multiple loops of fluid-filled bowel with wall thickening & smooth, central enhancement. Note the involvement of the duodenum & colon.* **(Right)** *Axial CECT, more inferiorly located in the same patient, shows bowel loops with moderate to marked wall thickening but only a thin central layer of hyperenhancement. Note the inflammatory stranding in the mesentery.*





(Left) *Transverse US in the same patient demonstrates a fluid-filled bowel loop with wall thickening.* **(Right)** *Frontal SBFT shows diffuse loss of mucosal folds in all visualized bowel loops. Mild bowel wall thickening is present, evidenced by loop separation. This appearance has been termed "ribbon bowel" & is typical of GVHD.*

Chronic Granulomatous Disease in Childhood

KEY FACTS

TERMINOLOGY

- Primary immunodeficiency characterized by recurrent infections at epithelial surfaces & organs with large numbers of reticuloendothelial cells

IMAGING

- Recurrent infections with catalase-positive organisms such as *Aspergillus* or *Staphylococcus aureus*
- Chronic granulomatous disease (CGD) can affect any organ system
- Pneumonia: Consolidation, nodules, reticulonodular opacities, & scarring
  - Complicated by abscess or empyema in 20%
- Lymph nodes
  - Lymphadenopathy present in ~ 100% of patients
  - Suppurative adenitis most common in neck
  - Calcified lymph nodes as sequelae of prior infection
- Osteomyelitis
  - Common locations: Lower & upper extremities
  - Chest wall & vertebral body involvement in up to 1/3 from contiguous spread of *Aspergillus* pneumonia
- Hepatic abscess(es)
- Gastrointestinal
  - CGD involvement mimics Crohn disease

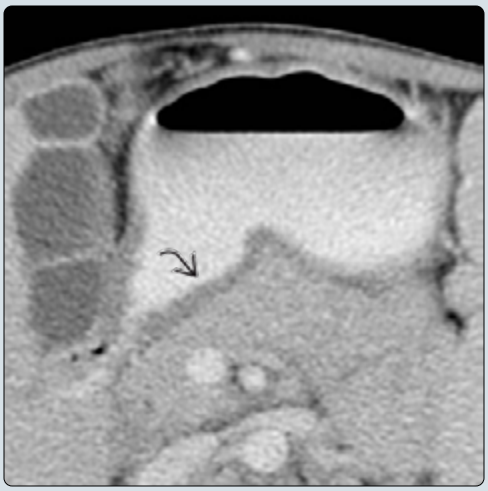
PATHOLOGY

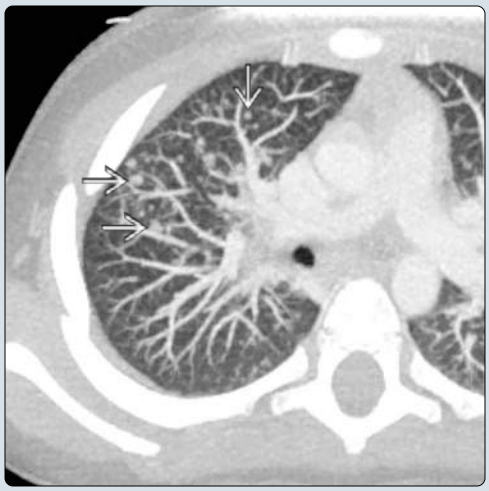
- Defect in 1 of 5 genes encoding for NADPH-oxidase
  - Defect prevents phagocytic cells from producing respiratory burst used to kill pathogens
- 2 types: X-linked > autosomal recessive

CLINICAL ISSUES

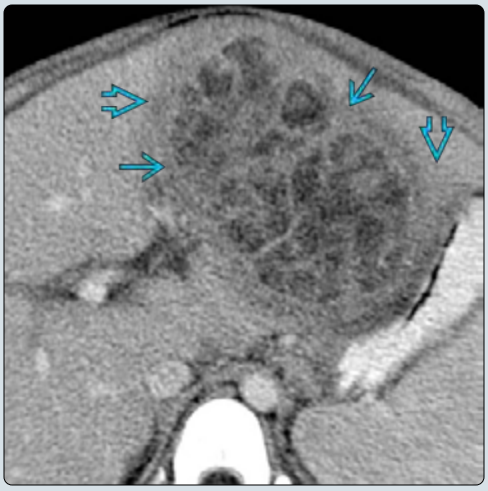
- 76% of patients diagnosed before 5 years of age
- 85% of patients with CGD male
- Pneumonia most common infection in CGD (80%)
- Suppurative adenitis 2nd most common infection (60%)
- Hepatic abscesses occur in 1/3 of patients

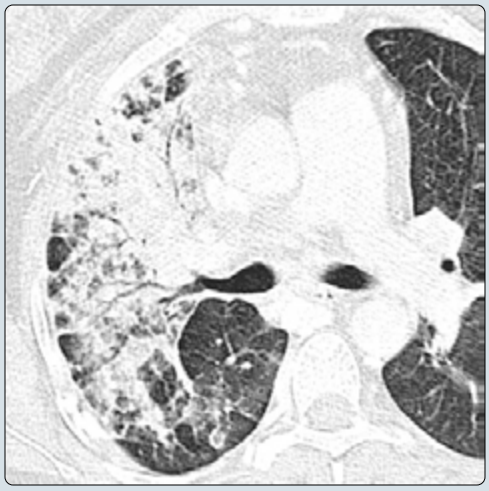
(Left) *Axial CECT of the stomach shows thickening of the gastric antrum, a typical location of inflammation in chronic granulomatous disease (CGD).* **(Right)** *Axial CECT MIP of the chest shows multiple small nodules in the right lung that suggest a pulmonary fungal infection in this patient with CGD.*





(Left) *Axial CECT of the abdomen shows a multiseptated abscess in the left lobe of the liver. Note the surrounding parenchymal edema. Up to 33% of patients with CGD develop a hepatic abscess; 60% of these patients will have multiple abscesses at diagnosis.* **(Right)** *Axial CECT of the chest in a patient with CGD shows pneumonia in the right lung. Pneumonia is the most common type of infection & the most common cause of death in patients with CGD.*





TERMINOLOGY

Abbreviations

- Chronic granulomatous disease (CGD)

Definitions

- Primary immunodeficiency characterized by recurrent infections at epithelial surfaces & organs with large numbers of reticuloendothelial cells

IMAGING

General Features

- Best diagnostic clue
  - Recurrent infections with catalase-positive organisms such as *Aspergillus* or *Staphylococcus aureus*
- Location
  - CGD can affect every organ system

Radiographic Findings

- Chest
  - Consolidation, nodules, reticulonodular opacities, & scarring
- Skeletal
  - Osteomyelitis
    - Common locations: Extremities, chest wall
      - Chest wall & vertebral involvement in up to 1/3 from contiguous spread of *Aspergillus* pneumonia

Fluoroscopic Findings

- Upper GI
  - Gastrointestinal
    - Esophagus: Dysmotility & strictures
    - Stomach: Narrowing of gastric antrum
      - Gastric outlet obstruction, delayed gastric emptying, thickened rugae

CT Findings

- NECT
  - Chest
    - Pneumonia
      - Consolidation, ground-glass opacity, tree-in-bud opacity, centrilobular or random nodules, bronchiectasis, air-trapping, septal thickening, or scarring
      - ± cavitation, mycetoma
      - Abscess or empyema in 20%
    - Prolonged course may lead to chronic findings
      - Mediastinal or hilar adenopathy, pulmonary fibrosis, honeycomb lung, pulmonary arterial hypertension, & pleural thickening
- CECT
  - Lymph nodes
    - Lymphadenopathy in nearly all patients with CGD
    - Suppurative adenitis occurs most commonly in neck
      - Enlarged, enhancing lymph node with central hypodensity ± thick, enhancing septations
    - Calcified lymph nodes as sequelae of prior infection
  - Liver
    - Hepatic abscess occurs in 1/3
      - Multiple in 60% of patients
      - Recur in 40% of patients
      - Abscesses < 1 cm in size enhance homogeneously
      - Abscesses between 1-3 cm have incomplete central enhancement
      - Abscesses > 3 cm have heterogeneous enhancement with thick irregular loculations
      - Ca^{2+} can occur at site of prior infection
    - Hepatomegaly
  - Spleen
    - Splenic abscesses occur in up to 1/3 of patients with hepatic abscess
      - Splenic Ca^{2+} can occur after abscess has healed
    - Splenomegaly
  - Gastrointestinal tract
    - CGD involvement mimics Crohn disease
      - Can affect mouth to anus
      - Stomach: Narrowing of gastric antrum with wall thickening
      - Bowel: Wall thickening, mucosal hyperenhancement, skip lesions, luminal narrowing, fistulas, & perirectal abscess
  - Kidneys
    - Pyelonephritis ± renal abscess
      - Ca^{2+} can occur after renal abscess
  - Sinuses
    - Sinusitis has similar appearance to that in immunocompetent patients (i.e., mucosal thickening, air-fluid levels)
      - Fungal sinusitis can appear hyperdense
      - ± bone destruction

MR Findings

- Liver
  - Abscess
    - T1WI: Hypointense
    - T2WI: Heterogeneously hyperintense with irregular hypointense septations ± surrounding edema
    - T1WI C+: Enhancing septations
  - Hepatosplenomegaly
- Gastrointestinal
  - Stomach: Wall thickening
  - Bowel: Wall thickening, mucosal hyperenhancement, skip lesions, luminal narrowing, fistulas, & perirectal abscess
- Brain
  - Encephalitis
  - Brain abscess
    - Ring-enhancing lesions
    - Typically occur at gray-white matter junctions
    - Surrounding vasogenic edema
  - Meningitis: Thickened, enhancing meninges
- Sinuses
  - Sinusitis
    - Similar appearance to sinusitis in immunocompetent patients
    - Fungal sinusitis: Hypointense on T1/T2WI

Ultrasonographic Findings

- Grayscale ultrasound
  - Lymph nodes

- Suppurative adenitis: Hypoechoic internal contents ± swirling internal debris, thickened septa; absent or only septal central vascularity, ↑ peripheral vascularity
- Late: Hyperechoic foci with posterior shadowing (Ca^{2+}) after prior infection
- Liver
  - Hepatic abscess: Solid or complex-appearing hypo- to isoechoic mass
  - Absent or only septal central vascularity, ↑ peripheral vascularity
- Bladder
  - Urinary tract infection with wall thickening
  - Inflammatory pseudotumors: Focal bladder wall thickening
    - Can mimic rhabdomyosarcoma

Imaging Recommendations

- Best imaging tool
  - Depends on site of infection: US or MR preferred modalities due to frequent imaging in affected patients
    - Patients often receive multiple CECTs over course of life due to rapid scan time & ease of scheduling
  - PET/CT (or potentially whole body MR + DWIBS) can be used if site of infection cannot be identified

DIFFERENTIAL DIAGNOSIS

Recurrent Bacterial Pneumonia

- May be indistinguishable from pneumonia in CGD
- Look for underlying congenital lung lesion

Severe Combined Immunodeficiency

- Primary immunodeficiency with severe T- & B-cell dysfunction
- Infants present with infections due to lack of T cells

X-Linked Agammaglobulinemia

- Immunodeficiency characterized by absence of B cells
- Presents with bacterial infections after maternal antibodies cleared

Common Variable Immunodeficiency

- Immunodeficiency characterized by low levels of immunoglobulins & B cells
- Presents with recurrent bacterial infections

PATHOLOGY

General Features

- Etiology
  - Defect in 1 of 5 genes encoding for NADPH-oxidase
    - Defect prevents phagocytic cells from producing respiratory burst used to kill pathogens
    - Neutrophils able to phagocytose catalase-positive organisms but not kill them
    - Organism lives inside neutrophil causing chronic inflammatory response & granuloma formation
- Genetics
  - Can be transmitted in X-linked or autosomal recessive patterns
    - X-linked transmission in 65-70% of cases

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pneumonia most common infection in CGD, occurring in 80% of patients
    - Most commonly caused by *Aspergillus*
  - Suppurative adenitis 2nd most common infection in CGD, occurring in 60% of patients
    - Most commonly caused by *S. aureus*
  - Hepatic abscesses occur in 1/3 of patients with CGD
    - Most commonly caused by *S. aureus*
  - Osteomyelitis occurs in up to 25% of patients with CGD
    - Most commonly caused by *Serratia marcescens*

Demographics

- Age
  - 76% diagnosed before 5 years
  - 15% diagnosed in 2nd decade
- Gender
  - 85% of patients with CGD male
- Ethnicity
  - 83% of patients with CGD Caucasian
- Epidemiology
  - Occurs in 1 in 200,000-250,000 live births

Natural History & Prognosis

- X-linked disease typically presents at younger age with more severe manifestations
- Recurrent infections typical
- Historically high mortality rate: In 1967, only 21% of patients survived beyond 5 years of age
  - Currently 90% survive into adulthood
  - Median age of death in 2012: 28 years
  - Pneumonia or sepsis most common cause of death

Treatment

- 2 tenets of treatment
  - Prophylaxis
    - Antibiotic: Co-trimoxazole
    - Antifungal: Itraconazole
    - Stimulate superoxide release: Interferon gamma
  - Treatment of acute infection
    - Treated empirically with intravenous antibiotics or antifungals
- Stem cell transplant only known cure for CGD

DIAGNOSTIC CHECKLIST

Consider

- Consider CGD in patients with hepatic abscess, recurrent pneumonia, or other unusual infections

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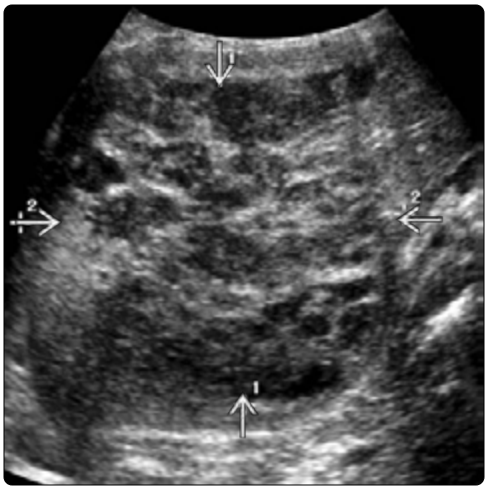
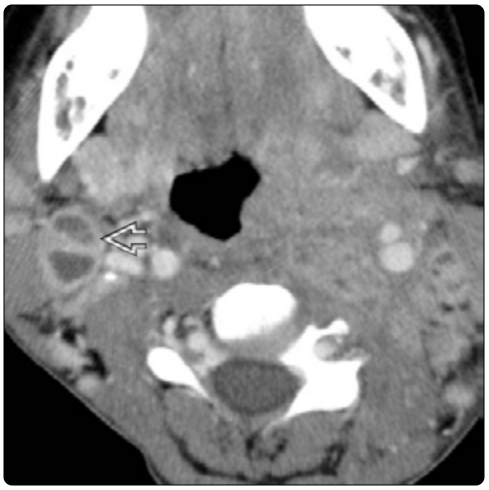
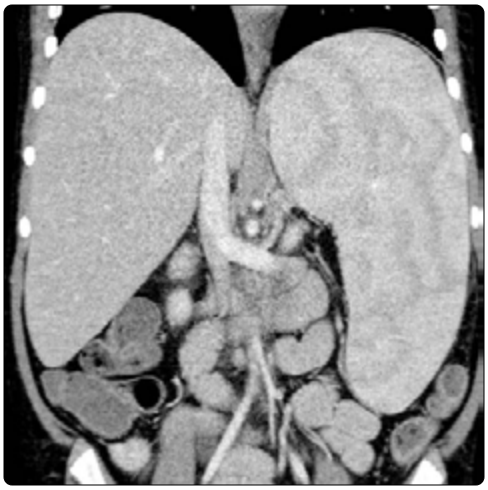
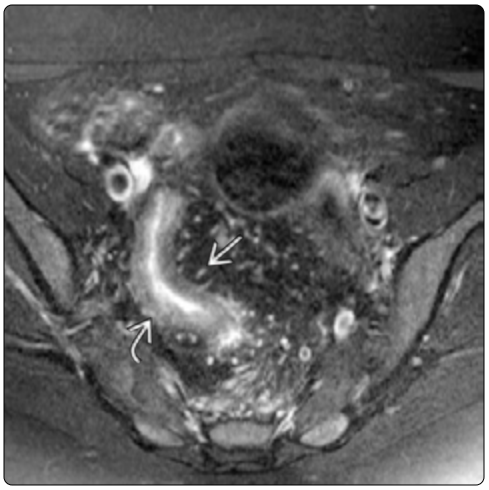


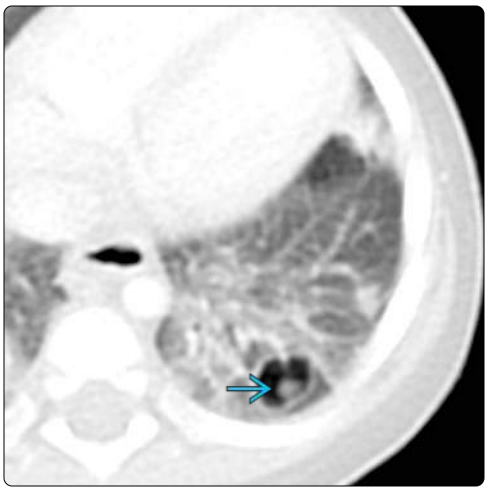


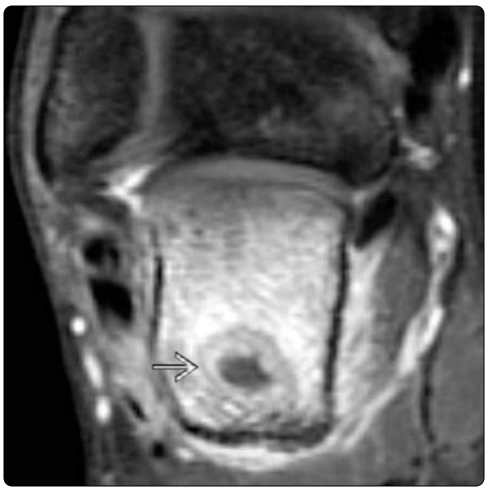
(Left) *Transverse ultrasound of the liver shows a heterogeneous multiseptated abscess. No significant internal vascularity was identified on color Doppler imaging (not shown) to suggest a solid mass.* **(Right)** *Axial CECT of the neck shows a suppurative lymph node in the right cervical chain. All patients with CGD have lymphadenopathy; 60% of patients develop suppurative adenitis.*





(Left) *Coronal CECT of the abdomen shows hepatosplenomegaly without discrete parenchymal lesions.* **(Right)** *Axial T2 FS MR of the pelvis shows diffuse wall thickening of the sigmoid colon with associated engorgement of the vasa recta. Inflammatory bowel disease in patients with CGD can mimic Crohn disease.*





(Left) *Axial CECT shows a cavitary pneumonia with mycetoma (fungus ball) in the left lower lobe. Aspergillus is the most common fungal pneumonia in patients with CGD.* **(Right)** *Coronal T1 C+ FS MR shows a targetoid focus of abnormal enhancement of the calcaneus. The center of this lesion does not enhance, typical of pus. There is a moderately thick rim of enhancing granulation tissue plus marked surrounding marrow edema.*

Pneumatosis in Older Children

KEY FACTS

TERMINOLOGY

- Presence of gas within walls of gastrointestinal tract
- In older children, more frequently benign & associated with variety of underlying conditions & medications

IMAGING

- Radiography 1st-line test
  - CT more sensitive but not often needed
- Curvilinear or round gas collections within bowel wall
  - Colon > small bowel >> stomach
  - Benign pneumatosis often quite extensive
- Look for gas in posterior portions of bowel wall (deep to luminal contents) to differentiate from intraluminal gas
- ± free intraperitoneal air
  - Not necessarily due to intestinal perforation
- CT findings concerning for clinically significant causes: Bowel wall thickening or ↓ enhancement, inflammatory stranding, free fluid

TOP DIFFERENTIAL DIAGNOSES

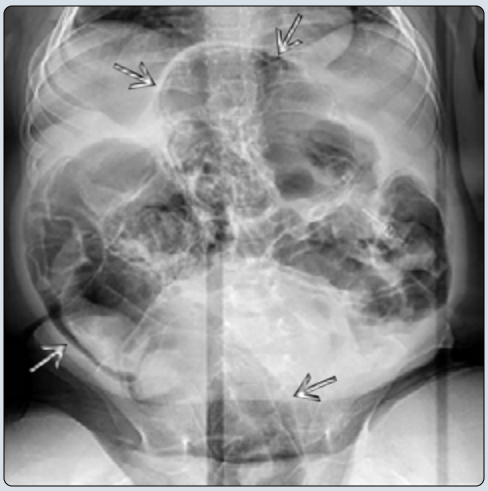
- Benign causes: Medications (corticosteroids), systemic diseases, pulmonary disease, solid organ transplant, bone marrow transplant
- Clinically worrisome causes: Ischemia, bowel obstruction, severe infection, trauma

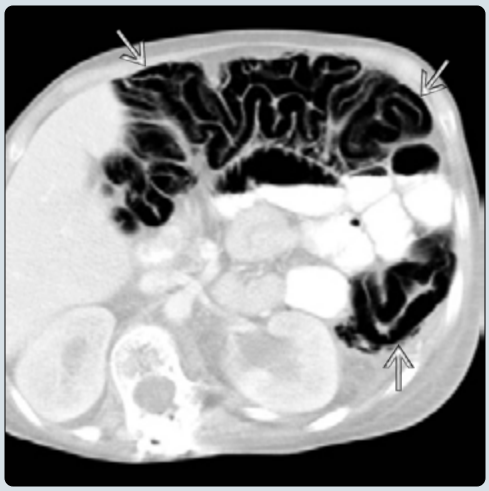
PATHOLOGY

- Multiple proposed mechanisms for gas dissecting into bowel wall; likely multifactorial
  - Mucosal damage &/or ↑ intraluminal pressure allows gas to dissect into wall

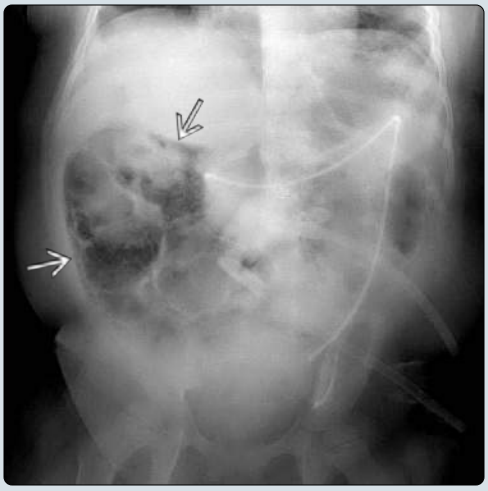
CLINICAL ISSUES

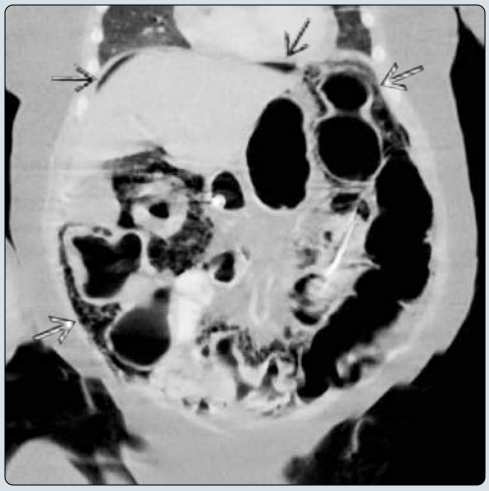
- Managed conservatively unless worrisome clinical features
  - Bowel rest, removal of offending medications, treatment of underlying conditions
- If worrisome clinical/laboratory features → surgery





(Left) *AP radiograph in a 3-year-old boy with multiple medical problems shows extensive curvilinear lucencies separating layers of the colonic wall. This pneumatosis extends all the way to the rectum. The patient was clinically stable, & the pneumatosis was thought to be benign & due to corticosteroid use.* **(Right)** *Axial CECT in a different child shows diffuse pneumatosis throughout the visualized colonic wall.*





(Left) *AP radiograph in a 2-year-old boy with a seizure disorder & multiple medical problems shows colonic pneumatosis, mostly in the ascending colon & proximal transverse colon.* **(Right)** *Coronal CECT in the same 2-year-old boy shows extensive pneumatosis throughout the colon. There is also a small amount of free intraperitoneal gas. Despite the presence of free intraperitoneal gas, the patient was treated conservatively due to his clinical status & lack of other imaging features to suggest ischemia or obstruction.*

TERMINOLOGY

Synonyms

- Pneumatosis intestinalis; pneumatosis coli (confined to colon); pneumatosis cystoides intestinalis

Definitions

- Presence of gas within walls of GI tract
  - Associated with necrotizing enterocolitis in
    - Premature infants
    - Term newborns with
      - Congenital heart disease
      - Hirschsprung disease
      - Maternal use of vasoconstrictive agents (e.g., cocaine)
  - In older children, more frequently benign & associated with variety of underlying conditions & medications

IMAGING

General Features

- Best diagnostic clue
  - Curvilinear or rounded gas collections within bowel wall
    - Colon > small bowel >> stomach
  - Extensive pneumatosis more frequently due to benign causes
    - May involve entire colon

Radiographic Findings

- Curvilinear or rounded foci of gas outlining bowel wall
- ± dilated bowel or evidence of bowel wall thickening
- ± free intraperitoneal air

CT Findings

- CECT
  - Gas collections within wall of bowel
    - Foci of gas in posterior bowel wall may help differentiate from intraluminal gas or gas-containing fecal debris
  - CT findings helpful in differentiating benign from clinically significant causes
    - Extensive pneumatosis: More commonly benign
    - Bowel wall thickening: More commonly significant
    - ↓ bowel wall enhancement: Typically significant
    - Inflammatory stranding: Typically significant
    - Free fluid: Typically significant
  - Some CT findings **not** helpful in differentiating benign from clinically significant causes
    - Linear vs. cystic pattern
    - Distribution in small vs. large bowel
    - Portal venous gas
    - Free intraperitoneal air

Ultrasonographic Findings

- Helpful but can be confusing due to intraluminal gas
- Look for echogenic gas tracking within bowel wall, particularly lateral & posterior aspects
  - Dirty posterior acoustic shadowing & ring-down artifact
  - ± twinkle artifact with color Doppler

DIFFERENTIAL DIAGNOSIS

Benign Causes of Pneumatosis Intestinalis

- Medications
  - Corticosteroids, certain chemotherapy agents
- Transplant patients
  - Bone marrow/stem cell transplants
    - May be associated with graft-vs.-host disease (though characteristic clinical features expected)
  - Solid organ transplants
- Pulmonary disease
  - Mechanical barotrauma
- Autoimmune/rheumatologic diseases
  - ↑ in conditions associated with vasculitis

Clinically Worrisome Causes of Pneumatosis Intestinalis

- Intestinal ischemia
- Severe enterocolitis
- Bowel obstruction
- Trauma

PATHOLOGY

General Features

- Etiology
  - Multiple proposed mechanisms; may be multifactorial
    - Mucosal injury ± limited ability to repair mucosa due to medications (e.g., corticosteroids) or vascular abnormalities (e.g., vasculitis)
    - Abnormal bowel wall lymphatics allowing gas to dissect into wall
    - ↑ intraluminal pressure pushing gas or gas-forming bacteria into bowel wall
    - Dissection of gas from mediastinum → mesentery → bowel wall

Gross Pathologic & Surgical Features

- Gas within submucosal & subserosal spaces
  - May represent gas within dilated intramural lymphatics

CLINICAL ISSUES

Treatment

- Managed conservatively if clinically stable
  - Bowel rest, removal of offending medications, treatment of underlying conditions
- If worrisome by clinical or laboratory features → surgery

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Crohn Disease

KEY FACTS

TERMINOLOGY

- Chronic, recurrent, segmental, granulomatous inflammatory bowel disease; etiology unknown

IMAGING

- Small bowel follow-through: Separated/isolated segment of narrowed bowel with irregular mucosa
- US: Thickened bowel wall with loss of gut signature, ↑ perienteric fat echogenicity, ± bowel wall & perienteric hyperemia
- CT/MR enterography
  - Bowel findings
    - Mucosal hyperenhancement & bowel wall thickening: Most sensitive findings
    - Mural stratification of bowel wall enhancement
    - Strictures showing upstream effects (dilation & small bowel feces sign)
    - Ulcers/fistulas/sinus tracts
  - Mesenteric findings
    - Engorged vasa recta (comb sign & dot sign)
    - Fat stranding, fibrofatty proliferation
    - Phlegmon/abscesses
    - ↑ size & number of lymph nodes
  - Perianal disease
    - Tract/collection extending from anus/skin

PATHOLOGY

- Skip lesions of transmural inflammation occur anywhere from mouth to anus: Ileocolic most common

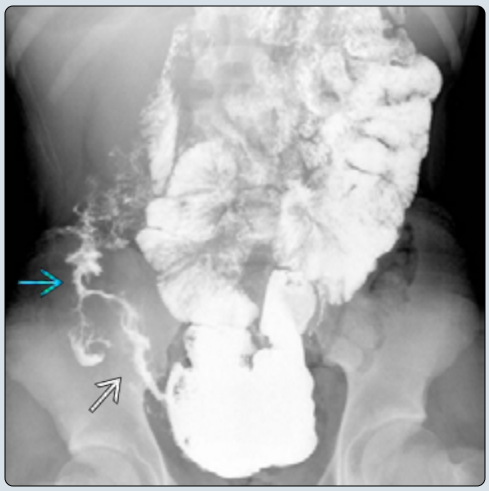
CLINICAL ISSUES

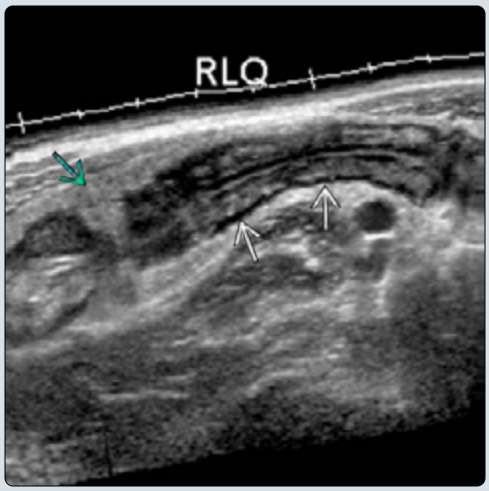
- Annual incidence: 3-15/100,000
- 25% of patients diagnosed in first 2 decades of life

DIAGNOSTIC CHECKLIST

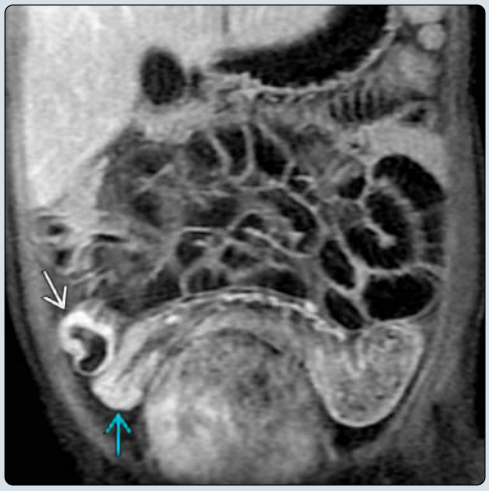
- Extraintestinal manifestations: Cholelithiasis, sclerosing cholangitis, arthritis, & urolithiasis
- Look for penetrating &/or stricturing disease: Alters management

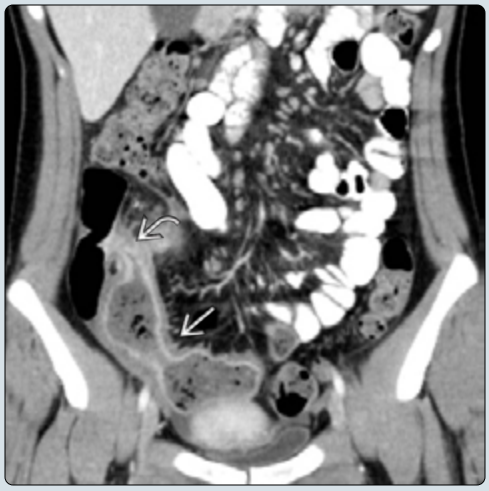
(Left) *Delayed image from a small bowel follow-through in a patient with Crohn disease shows isolation, narrowing, & irregularity of the terminal ileum & cecum. We infer that the separated loops are caused by bowel wall thickening & "creeping fat."* **(Right)** *Longitudinal US of the right lower quadrant (RLQ) shows an abnormal segment of bowel with wall thickening & surrounding echogenic mesenteric fat. Ultrasound may be the 1st modality to suggest Crohn disease in patients being worked up for appendicitis.*





(Left) *Coronal T1 C+ FS MR enterography in an adolescent with Crohn disease shows mucosal hyperenhancement & wall thickening of the ileum & cecum.* **(Right)** *Coronal CECT of the abdomen in a patient with Crohn disease shows an abnormal appearance of the terminal ileum with mucosal hyperenhancement, mild mural stratification, & sacculations. There is a stricture of the distal-most portion of the terminal ileum with upstream dilation & small bowel feces.*





TERMINOLOGY

Abbreviations

- Terminal Ileum (TI)

Definitions

- Chronic, recurrent, segmental, granulomatous inflammatory bowel disease (IBD)

IMAGING

General Features

- Best diagnostic clue
  - Small bowel follow-through (SBFT)
    - Affected bowel loop "isolated" (separated from other loops) with luminal narrowing & mucosal irregularity
  - CT/MR enterography
    - Bowel wall thickening & mucosal hyperenhancement with associated perienteric inflammatory changes
- Location
  - Anywhere from mouth to anus
  - Disease distribution in children (differing from adults)
    - Most common location: Ileocolonic region
    - Isolated TI disease uncommon, occurring in < 10%
    - Small bowel involvement proximal to TI in > 50%
    - Upper GI involvement occurs in 1/3-1/2 of patients
    - Appendix may be involved, even in isolation

Fluoroscopic Findings

- Upper GI with SBFT
  - Separation of bowel loops due to wall thickening & "creeping" proliferative fat
  - Irregular appearance of bowel mucosa
    - Cobblestone pattern: Longitudinal + transverse ulcers
    - Thickened mucosal folds & featureless mucosa (moulage sign)
    - Lymphoid hyperplasia: 1- to 3-mm mucosal elevations, no ring shadow
    - Aphthoid ulcerations: Target or bull's-eye appearance (up to 5 mm)
  - String sign of narrowed bowel lumen
    - Up to 20%, most common in TI
  - Skip lesions (90%)
  - Sacculations at antimesenteric border (due to ↑ luminal pressure)

CT Findings

- CT enterography
  - Technique
    - Low-density oral contrast material to maximize bowel luminal distention
    - IV contrast with imaging in enteric phase (~ 45 s) or portal venous phase (~ 70 s)
  - Bowel findings
    - Mucosal hyperenhancement
    - Bowel wall thickening (lower attenuation edema)
    - Mural stratification: Layers of bowel wall show differential enhancement
    - Luminal narrowing
  - Mesenteric findings
    - Engorged vasa recta (comb sign if tangential or dot sign in cross section)
    - Fat stranding ± fibrofatty proliferation
    - Prominent lymph nodes (↑ in size & number, often not larger than 1 cm in short axis)
  - Penetrating disease
    - Ulcer/fistula/sinus tract
    - Phlegmon/abscess
    - Perianal disease
  - Stricturing disease
    - Luminal narrowing with proximal dilation ± proximal small bowel feces sign or stuck pill/enterolith

MR Findings

- MR enterography
  - Technique
    - Biphasic enteric contrast agent to maximize bowel luminal distension
      - Bright on T2, dark on T1
    - Image in prone position
    - Glucagon may be used to slow/stop bowel peristalsis
    - SSFSE T2 ± FS
    - DWI (particularly helpful if IV contrast not possible)
    - 3D GRE T1 C+ FS sequences
    - Targeted axial & coronal oblique images of anus with suspicion of perianal disease
  - Findings similar to those seen with CT enterography
  - Improved visualization of penetrating/fistulizing disease, particularly in perianal region

Ultrasonographic Findings

- Grayscale ultrasound
  - High-resolution, high-frequency linear transducer with graded compression
  - Thickened bowel wall with loss of normal gut signature
  - ↓ or absent peristalsis
  - ↑ perienteric fat echogenicity & volume
  - Lymphadenopathy
  - Fluid collections
- Color Doppler
  - Bowel wall & perienteric hyperemia

Other Modality Findings

- Direct visualization with upper & lower endoscopy
  - Allows contemporaneous biopsy
  - Cannot view jejunum & ileum
  - Cannot visualize extraintestinal manifestations
- Capsule endoscopy
  - 11- x 27-mm capsule camera
  - Direct visualization of mucosal lesions
  - Risk of capsule not passing beyond area of stricture
  - Retained capsule camera not MR compatible

Imaging Recommendations

- Protocol advice
  - CT & MR enterography: High accuracy for detection of active IBD & associated complications
    - MR enterography lacks ionizing radiation exposure
    - CT enterography may be more sensitive in detecting early/mucosal disease & has less interrater variation
  - Ultrasound less sensitive but may be useful in initially suggesting Crohn disease or following diseased segment

DIFFERENTIAL DIAGNOSIS

Appendicitis

- Dilated, thick-walled, noncompressible appendix with induration of surrounding fat
  - ± hyperemia, appendicolith, abscess
- Adjacent bowel typically normal in absence of appendiceal perforation

Infectious Enterocolitis

- Nonspecific imaging findings of ileocecal infection, resolves with appropriate treatment

Ulcerative Colitis

- Contiguous colitis extending proximally from anus
  - No skip lesions
- ± backwash ileitis
- Not transmural: No strictures, sinuses, or fistulas
- Pseudopolyps, ↑ risk of colon cancer

Pseudomembranous Colitis

- Pancolitis, typically due to *Clostridium difficile* toxins, in setting of antibiotics

Neutropenic Colitis

- Immunocompromised patient; proximal colon involved

PATHOLOGY

General Features

- Etiology
  - Idiopathic IBD with prolonged & unpredictable course
  - Genetic, environmental, infectious, immunologic, & psychologic factors
    - Recent studies have identified number of genes associated with early onset of Crohn disease

Gross Pathologic & Surgical Features

- Skip lesions, edema, inflammation, fibrosis, & strictures

Microscopic Features

- Transmural inflammation, lymphoid aggregates, & noncaseating granulomas (not well developed in 40%)
- Dilation & sclerosis of lymphatic channels

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Children: Pain, diarrhea, & weight loss
    - Weight loss: Presenting feature in 85% of children with Crohn disease
- Other signs/symptoms
  - Malabsorption
    - Interrupted enterohepatic circuit with diminished absorption of bile salts in TI
  - Erythema nodosum & pyoderma gangrenosum
- Laboratory findings
  - Anemia, leukocytosis, ↑ C-reactive protein, & ↑ erythrocyte sedimentation rate
  - ↑ fecal calprotectin
  - p-ANCA(-), ASCA(+): Favors Crohn over ulcerative colitis [ulcerative colitis suggested when (+) p-ANCA & (-) ASCA]

Demographics

- Age
  - Bimodal age distribution with main peak at 18-25 years, smaller peak at 60-80 years
  - 25% of patients diagnosed in first 2 decades of life
- Gender
  - 1.5:1 male predominance in prepubertal patients diagnosed with Crohn disease
- Epidemiology
  - ↑ incidence in developing world
  - ~ 3-15 cases/100,000 annually

Natural History & Prognosis

- Recurrence: 30-53% after resection; only 10-20% lead symptom-free lives
- Complications
  - Stricturing/penetrating
    - Fissures, sinus tracts, fistulas, & abscesses
    - Obstruction (20%), perforation (1-2%)
  - Extraintestinal manifestations in up to 30%
    - Stones: Cholelithiasis, urolithiasis (oxalate)
    - Arthropathy: Enthesis-related arthritis
    - Hepatobiliary: Autoimmune hepatitis, primary sclerosing cholangitis
    - Skin: Erythema nodosum, pyoderma gangrenosum
    - Other: Thromboembolism, pancreatitis, episcleritis, uveitis
  - Malignancy
    - Adenocarcinoma: Latency: 25-30 years
    - Lymphoma

Treatment

- Medical
  - Bowel rest, steroids, azathioprine, mesalamine, metronidazole
  - Biologic agents: Infliximab
- Surgical
  - Resection of diseased bowel, strictureplasty, & primary fistulotomy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Look for penetrating/stricturing disease: Alters clinical management

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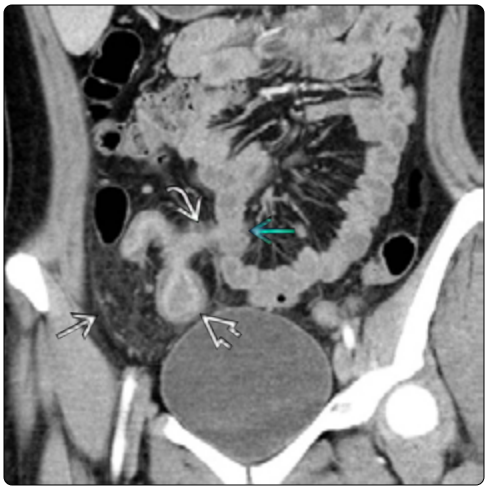
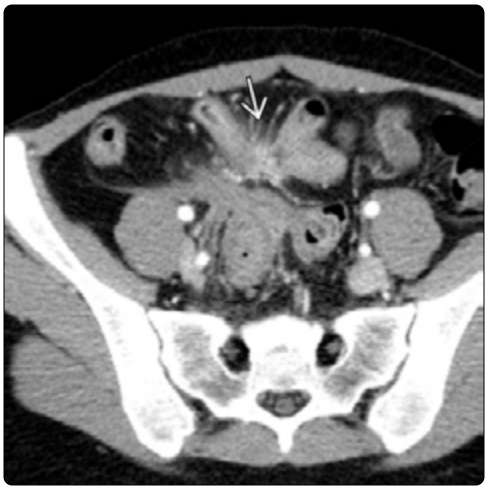
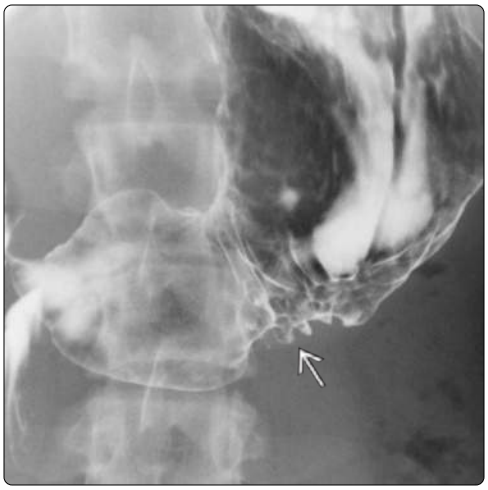
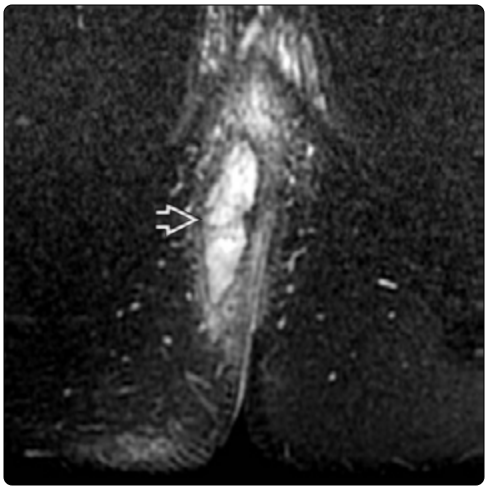


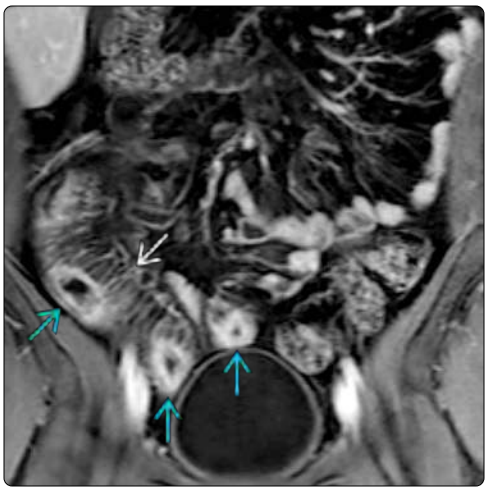


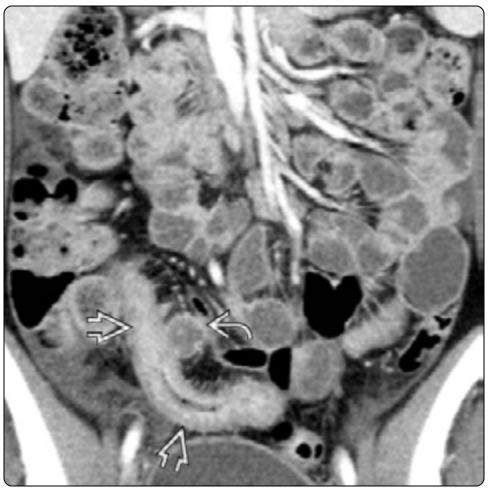
(Left) *Coronal CECT shows multiple findings of Crohn disease. There is focal mucosal hyperenhancement & small bowel wall thickening in the RLQ. A fistula tethers this loop to a nearby ileal loop. There is proliferation & stranding of nearby mesenteric fat.* **(Right)** *Axial CECT in a patient with Crohn disease shows multiple bowel loops tethered centrally, characteristic of a fistula.*





(Left) *Supine frontal image of the stomach from an upper GI in a patient with Crohn disease shows intraluminal protrusion of mucosal irregularity along the greater curvature of the stomach. The area of involvement has "heaped-up" margins with 3 areas of ulceration.* **(Right)** *Axial T2 FS MR of the gluteal region in a child with Crohn disease shows an abscess of the right buttock adjacent to the gluteal crease. This finding is secondary to a perianal fistula (not shown). MR is more sensitive than CECT for detecting perianal fistulas.*





(Left) *Coronal T1 C+ FS MR in a patient with Crohn disease shows an abnormal segment of ileum with mucosal hyperenhancement & bowel wall thickening. The vasa recta supplying this segment of bowel are engorged, demonstrating the comb sign.* **(Right)** *Coronal CECT enterography in a teenager with known Crohn disease shows signs of active inflammation of the terminal ileum with a penetrating ulcer/small abscess along the mesenteric border.*

Ulcerative Colitis

KEY FACTS

TERMINOLOGY

- Chronic, idiopathic, diffuse inflammatory disease that primarily involves colorectal mucosa & submucosa
  - Extends retrograde from rectum
  - No skip lesions or transmural involvement

IMAGING

- CT/MR enterography findings
  - Continuous colonic inflammation extending proximally from rectum for variable distance
  - Mucosal hyperenhancement & wall thickening
  - Thickened, edematous haustra (thumbprinting)
  - Luminal narrowing
  - Mural stratification: More common in ulcerative colitis than Crohn disease
  - DWI MR can aid in detecting inflammation, particularly if IV contrast not available
  - In later disease, dilation or narrowing of colon with loss of haustra

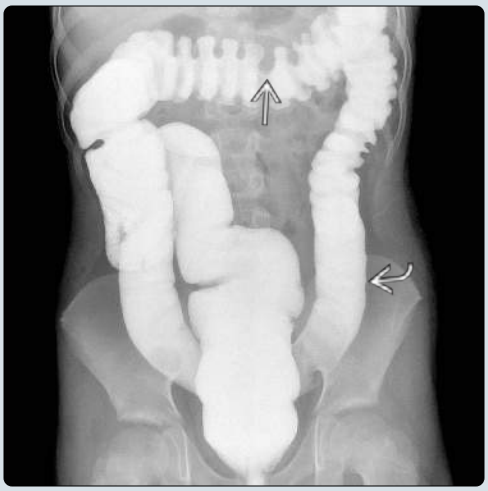
TOP DIFFERENTIAL DIAGNOSES

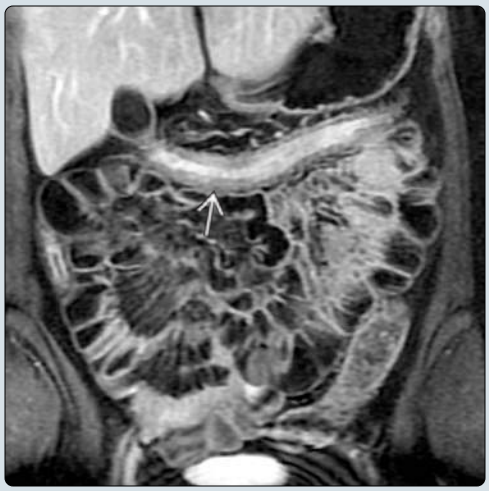
- Crohn disease
- Pseudomembranous colitis
- Infectious colitis

CLINICAL ISSUES

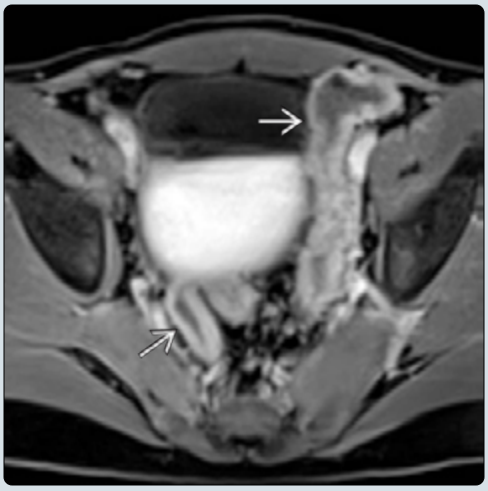
- Majority of patients diagnosed in 4th-5th decades
- Annual incidence: 5-10 cases/100,000 population
  - More common than Crohn disease in preschool age
- Most common presenting symptom: Bloody diarrhea
- Complications
  - Toxic megacolon: 5-10%
  - Stricture: 10%
  - Colorectal cancer risk: ↑ 30x
- Common extraintestinal manifestations: Arthralgias, primary sclerosing cholangitis, uveitis

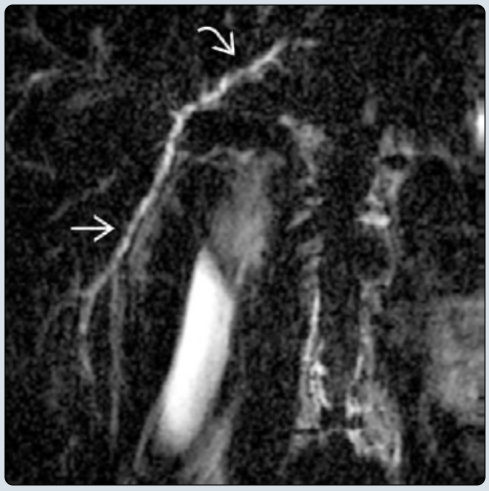
(Left) *Supine radiograph from a contrast enema in an adolescent with ulcerative colitis (UC) shows thickening of haustral markings (thumbprinting) in the transverse colon but loss of haustral markings in the descending colon.* **(Right)** *Coronal T1 C+ FS MR enterography in an adolescent with UC shows diffuse wall thickening & mucosal hyperenhancement of the transverse colon. The transverse colonic lumen is narrowed without visible haustral markings.*





(Left) *Axial T1 C+ FS MR of the pelvis in an adolescent with UC shows diffuse wall thickening & enhancement of the sigmoid colon.* **(Right)** *Coronal MRCP of an adolescent with UC shows diffuse irregularity of the right & left hepatic ducts. The findings are typical of primary sclerosing cholangitis (PSC). Patients with UC & PSC usually have more extensive colonic disease.*





TERMINOLOGY

Abbreviations

- Ulcerative colitis (UC)

Definitions

- Chronic, idiopathic diffuse inflammatory disease that primarily involves colorectal mucosa & submucosa

IMAGING

General Features

- Best diagnostic clue
  - CT/MR enterography: Mucosal hyperenhancement & wall thickening of colon/rectum
- Location
  - Continuous colonic inflammation extending proximally from rectum for variable distance
  - Pancolonic disease present in majority of children at diagnosis

Radiographic Findings

- Thickened, edematous haustra (thumbprinting)
- In later disease, dilation of colon with loss of haustra

Fluoroscopic Findings

- Contrast enema uncommon in current practice due to ileocolonoscopy & CT/MR enterography
- Classic findings on contrast enema: Mucosal irregularity, ulceration, thickened haustra, luminal narrowing
  - Chronic phase: Featureless, foreshortened colon ("lead pipe" colon)

CT Findings

- CECT
  - Diffuse, continuous, symmetric wall thickening of colon
  - Luminal narrowing
  - Mucosal islands or inflammatory pseudopolyps
  - Mural stratification: More common in UC than Crohn disease
  - Inflammatory pericolonic stranding
  - No penetrating disease (fistula, abscess) due to lack of transmural inflammation

MR Findings

- Findings similar to those seen with CT
- DWI can aid in detecting inflammation

Other Modality Findings

- Ileocolonoscopy
  - Allows contemporaneous biopsy
  - Cannot visualize extraintestinal manifestations

Imaging Recommendations

- Sensitivity for detecting colonic inflammation lower with enterography due to colonic nondistention

DIFFERENTIAL DIAGNOSIS

Crohn Disease

- Anywhere from mouth to anus may be involved
- Noncontiguous skip lesions characteristic

Pseudomembranous Colitis

- Pancolitis related to antibiotic use & overgrowth of *Clostridium difficile*

Infectious Colitis

- Nonspecific wall thickening & hyperenhancement
- Distribution varies depending on organism

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Bloody diarrhea
- Less common presenting symptoms: Anemia, poor growth, perianal symptoms
- Complications
  - Toxic megacolon: 5-10%
  - Stricture: 10%
  - Colorectal cancer risk: ↑ 30x
- Extraintestinal manifestations
  - Common: Arthralgias, primary sclerosing cholangitis, uveitis
  - Occur in 16-30% of patients
  - ↑ likelihood of colectomy
- Laboratory finding
  - p-ANCA(+) & ASCA(-) favors UC over Crohn disease

Demographics

- Age
  - Majority of patients diagnosed in 4th-5th decades
  - More common than Crohn disease in preschool age
- Epidemiology
  - Annual incidence: 5-10 cases/100,000 population

Treatment

- Medical: Sulfasalazine, steroids, azathioprine, methotrexate, LTB4 inhibitors
- Surgical: Total colectomy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Continuous involvement of colon extending proximally from rectum for variable length
- Shortened & rigid "lead pipe" colon in long term
- Consider UC in any patient with sclerosing cholangitis

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KEY FACTS

TERMINOLOGY

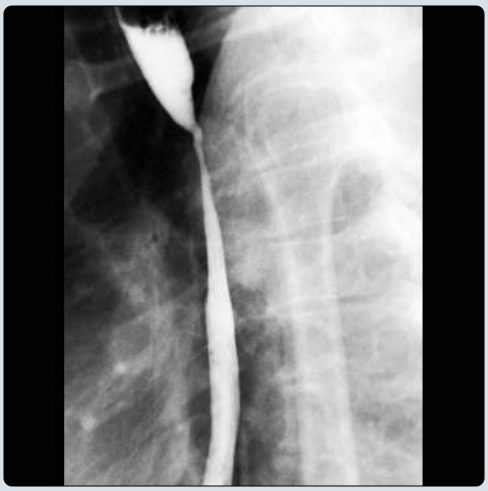
- Acquired narrowing of esophagus, caused by variety of entities; congenital stenosis considered here as well

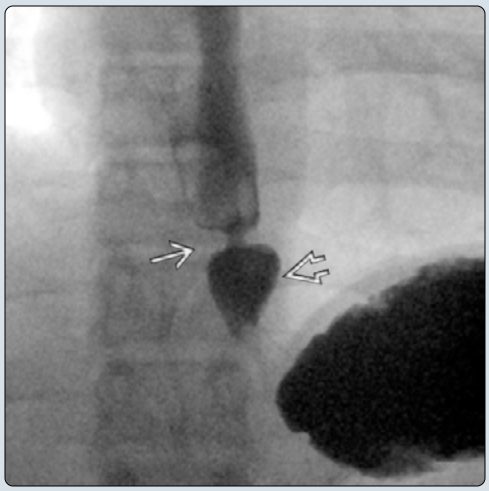
IMAGING

- Focal or diffuse narrowing of esophagus, associated with proximal dilation & altered peristalsis
- Particular segment affected as well as length & degree of narrowing depend on etiology & severity of initial esophageal injury
- Different causes of stricture have different imaging characteristics
- Esophagram findings
  - Esophageal atresia repair: Focal stricture at junction of upper & middle 1/3
  - Peptic stricture: Stricture in distal esophagus
  - Eosinophilic esophagitis: Most commonly normal
  - Caustic strictures: Most common in proximal & mid esophagus
    - Multiple strictures can occur
  - Epidermolysis bullosa: Stricture most common in cervical esophagus
  - Infective esophagitis: May progress to strictures
  - Congenital stenosis
    - Membranous web: Upper or middle 1/3
    - Fibromuscular thickening: Middle or distal 1/3
    - Tracheobronchial cartilage remnant/foregut malformation: Distal 1/3

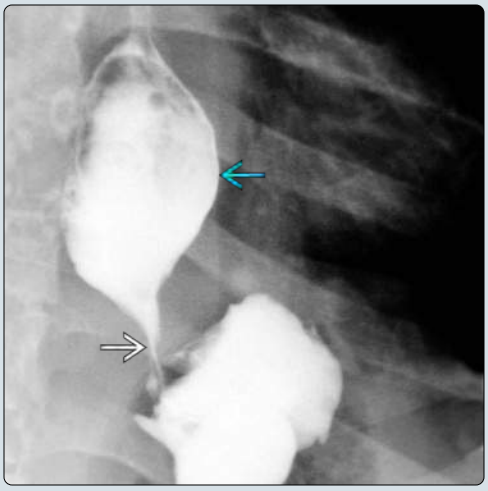
CLINICAL ISSUES

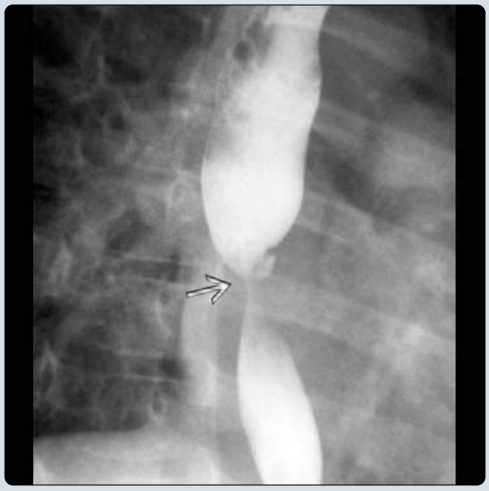
- Infants present with feeding difficulties; food bolus impaction in older children with drooling, vomiting, pain, sensation of food getting stuck
  - ± airway symptoms, such as wheezing, recurrent coughing
- Balloon dilation used for many conditions; stricture resection or esophageal replacement for refractory cases





(Left) *Lateral esophagram shows distention of the proximal esophagus with tapering to a long segment of narrowing secondary to epidermolysis bullosa in an adolescent child.* **(Right)** *Frontal upper GI in an adolescent shows a small hiatal hernia with circumferential narrowing at the gastroesophageal (GE) junction. Rings at the GE junction are termed Schatzki or B rings & are fixed areas of narrowing.*





(Left) *Oblique esophagram shows dilation of the distal esophagus with narrowing at the surgical site of a Nissen fundoplication. This Nissen was too tight & required balloon dilation.* **(Right)** *Oblique image from an upper GI series in an adolescent with eosinophilic esophagitis shows a Schatzki ring in the distal esophagus. Schatzki rings are rare in children & are often associated with eosinophilic esophagitis.*

TERMINOLOGY

Definitions

- Acquired narrowing of esophagus from variety of entities
- Congenital causes of stenosis considered here as well

IMAGING

General Features

- Best diagnostic clue
  - Narrowing of esophagus, associated with proximal dilation & altered peristalsis
  - Particular segment affected as well as length & degree of narrowing depend on etiology & severity of esophageal injury

Radiographic Findings

- Search for radiopaque foreign body
- Search for acute or chronic complications of foreign body or caustic ingestion
  - Perforation of esophagus with pneumomediastinum or pleural effusion
  - Chronically dilated esophagus with air-fluid level on upright view

Fluoroscopic Findings

- Esophagram
  - Focal or diffuse narrowing of esophagus; dysmotility common
  - Different causes of stricture have different imaging characteristics
    - Congenital stenosis
      - Membranous web: Upper or middle 1/3
      - Fibromuscular thickening: Middle or distal 1/3
      - Tracheobronchial cartilage remnant/foregut malformation: Distal 1/3
    - Prior esophageal atresia repair
      - Focal stricture at site of prior anastomosis between dilated proximal esophageal pouch & remaining mid to distal esophagus
    - Peptic stricture
      - Stricture in distal esophagus from chronic gastroesophageal reflux (GER)
    - Caustic strictures
      - Esophagram may not be helpful in acute phase as it delays endoscopy & may not reveal mucosal injury
      - Subsequent strictures can be focal, multifocal, or long segment, depending on ingested substance
      - Most common in proximal & mid esophagus
    - Eosinophilic esophagitis
      - Most common finding: Normal esophagus
      - ± GER, irregular contractions, esophageal strictures or rings, generally small caliber esophagus, & food bolus impaction
    - Epidermolysis bullosa
      - Stricture most common in cervical esophagus
    - Infective esophagitis
      - Severe esophageal infections may progress to strictures
  - Useful to identify complications (e.g., food impaction, leak after dilation or repair)

CT Findings

- CECT may be used in concert with esophagram to identify complications
  - Esophageal injury ± perforation → mediastinitis, abscess
    - Look for extraluminal oral contrast or gas
- Esophageal thickening & periesophageal stranding during acute inflammation

Ultrasonographic Findings

- Endoscopic US may visualize congenital cartilage rings

Imaging Recommendations

- Best imaging tool
  - Esophagram
- Protocol advice
  - Barium used for chronic strictures
  - Water-soluble contrast used for initial imaging if leak suspected
    - Low osmolality with concerns for aspiration
  - Double contrast technique not commonly used in children

DIFFERENTIAL DIAGNOSIS

Achalasia

- Failure of relaxation of lower esophageal sphincter
- Neuromuscular abnormality with thickening of circular & longitudinal muscles
- Esophagus dilated + air-fluid levels in upright position

Vascular Ring/Sling

- Smooth, round impression on esophagus
  - Pulmonary sling: Impression on anterior wall
  - Double aortic arch: Impression on posterior wall with reverse S appearance of esophagus on AP view
  - Right arch with aberrant left subclavian artery: Oblique impression upon posterior esophagus

Extrinsic Compression by Mass

- Neoplastic: Neuroblastoma, plexiform neurofibromas
- Congenital: Foregut duplication cyst, thymic cyst

PATHOLOGY

General Features

- Etiology
  - Postoperative esophageal atresia anastomotic stricture
    - Probably most frequent cause in children; occurs in 9-80% of esophageal atresia repairs
    - May present with retained foreign bodies in proximal pouch
  - Esophageal foreign body
    - Button batteries cause caustic burn injuries in esophagus within hours of ingestion, may lead to perforation or subsequent stricture
    - Chronic foreign bodies in esophagus can cause esophageal edema
  - Peptic strictures
    - Caused by acidic injury due to GER
  - Eosinophilic esophagitis
    - Allergic response to food
  - Postoperative Nissen fundoplication

- Complication of procedure occurs when wrap too tight
- Proximal esophagus dilates secondary to stricture or tight wrap

- Caustic esophagitis & strictures
  - Ingestion of household cleaners, lye ingestion, acid cleaning substance
    - Ingestion usually accidental in younger children, purposeful in teens
  - Type of ingested material can affect appearance of stricture
    - Acid ingestion typically causes short-segment stricture
    - Alkali ingestion typically causes long-segment stricture
  - Strictures can occur as early as 3 weeks after ingestion
- Infective esophagitis
  - Higher risk in immunocompromised
  - *Candida albicans* most common
- Epidermolysis bullosa
  - Hereditary disorder with > 100 distinct genotypes
    - Mutations in genes expressed at dermal-epidermal junction
  - Numerous bullous lesions, sloughing, then healing
  - Patients develop blisters & erosions after minor mechanical trauma
    - Repeated blistering → scar formation

- Associated abnormalities
  - Peptic strictures are more common in patients with associated comorbidity
    - 25% of patients have neurologic impairment

Microscopic Features

- Eosinophilic esophagitis characterized by esophageal mucosal biopsy with > 15 eosinophils per HPF
- Specific diagnosis of infectious esophagitis made by biopsy or culture

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Infants present with feeding difficulties
  - Difficulty swallowing, pain, drooling, vomiting, or sensation of food stuck with bolus impaction
    - Common symptoms in eosinophilic esophagitis, esophageal atresia repair, & tight Nissen fundoplication
  - Airway symptoms, such as wheezing, recurrent coughing episodes
    - Postoperative esophageal atresia patients have tracheomalacia
    - As esophagus dilates, it imprints posterior wall of trachea
  - Systemic diseases (such as scleroderma, dermatomyositis, epidermolysis bullosa) have multiorgan involvement
    - Many systemic diseases have skin findings

Demographics

- Age
  - Depends on disease process
- Gender
  - Eosinophilic esophagitis 4x more common in males
- Ethnicity
  - Eosinophilic esophagitis more common in Caucasians

Natural History & Prognosis

- Strictures may occur at site of prior injury or surgery
- Can progress to obliteration of esophageal lumen
- Recurrent strictures may occur in similar site
- Patients with caustic injection at risk of developing esophageal squamous cell carcinoma
- Patients with GER & Barrett esophagitis have higher risk of developing esophageal adenocarcinoma

Treatment

- Balloon dilation used for many conditions
  - Preferred over dilation using bougie as balloons dilate strictures in radial direction rather than longitudinal direction
  - Children often require multiple dilations depending on symptoms
  - Complications of balloon dilation
    - Perforation, mediastinitis
  - Endoscopic US can be used to triage congenital strictures
- Stricture resection or esophageal replacement reserved for cases where dilation fails
  - Tracheobronchial cartilage remnant classically fails dilation

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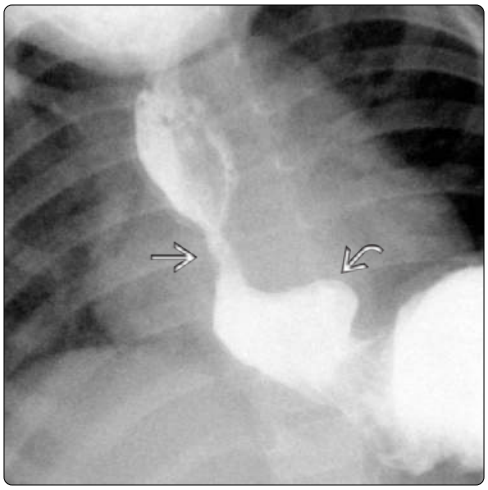
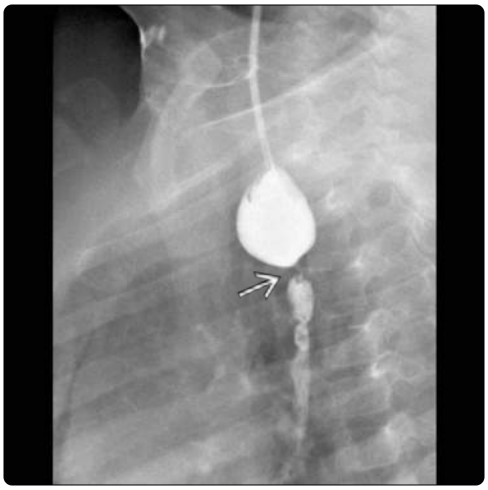
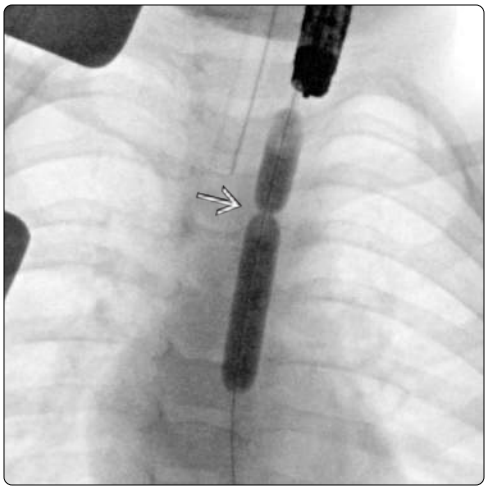
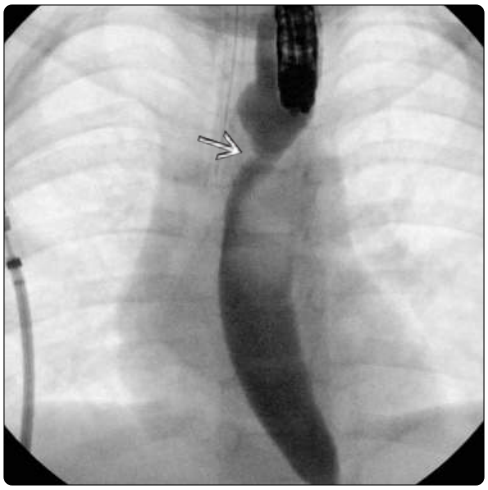


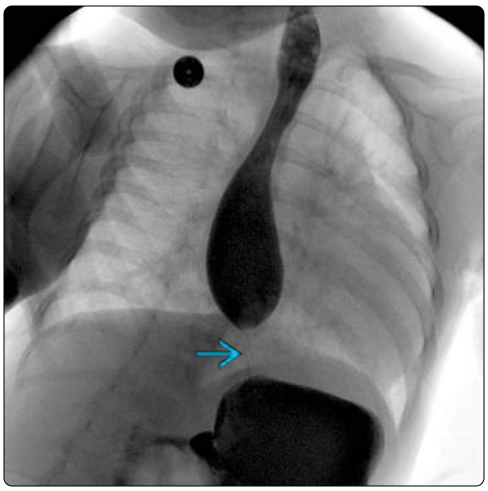


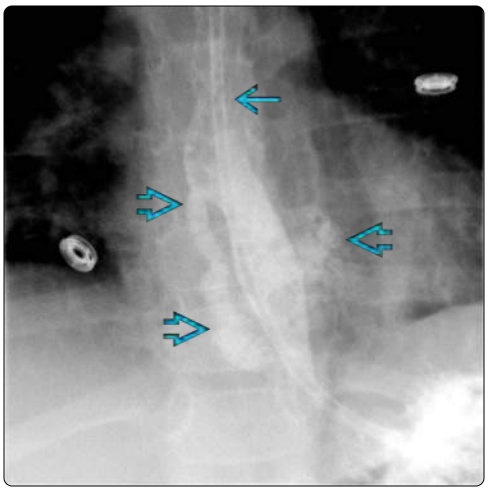
(Left) *Frontal image from an upper GI series in a patient with reflux shows a peptic stricture ➡ of the distal esophagus. The esophagus proximal to the stricture is dilated. In addition to the stricture, a large hiatal hernia ➡ is present.* **(Right)** *Lateral image from an upper GI series shows a tight, short-segment anastomotic stricture ➡ of the upper esophagus in an infant with a history of esophageal atresia repair.*





(Left) *Frontal image during esophageal dilation in the same patient (with a history of esophageal atresia repair) shows a focal waist of the dilating balloon ➡ at the site of the stricture.* **(Right)** *Frontal esophagram image in the same patient immediately following the balloon dilation of the stricture shows a mild residual narrowing ➡ at the site of esophageal anastomosis. While a mild stricture remains, the clinical function of the esophagus was markedly improved after the procedure.*





(Left) *Frontal esophagram in a 1 year old with chronic feeding difficulties shows a severe narrowing of the esophagus ⇨ at the GE junction. The bulbous distal esophagus tapers smoothly up to this level. Resection confirmed a foregut malformation with complete cartilage rings.* **(Right)** *Frontal esophagram in a child with a remote history of a caustic ingestion now status post balloon dilation shows irregularity & narrowing of the mid esophagus ⇨ with pooling of extraluminal contrast ⇨ due to a leak.*

Bezoars

KEY FACTS

TERMINOLOGY

- Specific ingested materials accumulate to form indigestible & potentially obstructive mass in GI tract
- Classified based on ingested material, including
  - Trichobezoar: Hair
  - Phytobezoar: Indigestible food components
  - Lactobezoar: Concretion of milk & mucous proteins

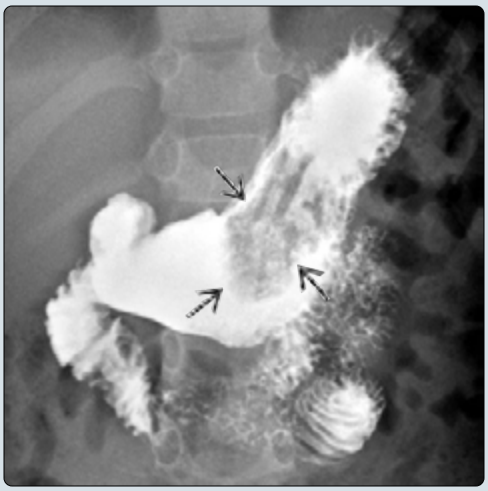
IMAGING

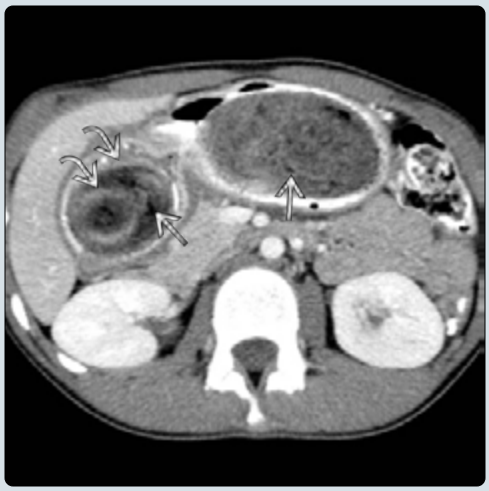
- Radiographs: Round/ovoid/tubular mass with mottled appearance due to air in interstices
  - Distention of affected lumen ± frank obstruction
- Fluoroscopy: Filling defect with contrast in interstices
- US: Echogenic arc-like surface & posterior shadowing
- CT: Concentric architecture & air/contrast in mass
  - ↑ sensitivity for gastric bezoars with window level setting of -100 HU
  - Small bowel bezoars located at transition point in cases presenting with obstruction
  - Floating fat density debris sign & short length help distinguish from small bowel feces sign

CLINICAL ISSUES

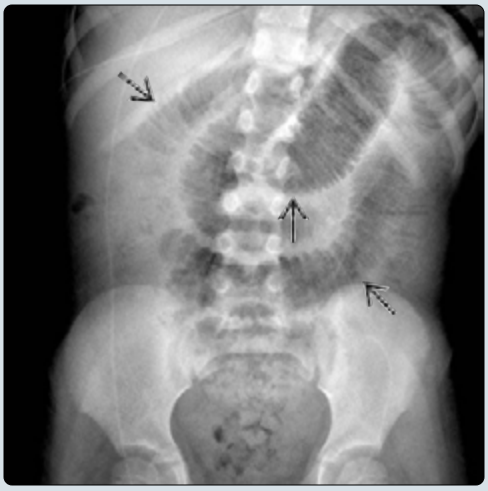
- Presentation: Palpable abdominal mass, abdominal pain, vomiting, distention, dysphagia, bowel obstruction
- Trichobezoar
  - Most common in young females with alopecia, trichotillomania, trichophagia
  - Treatment: Surgical removal by laparotomy
- Lactobezoar
  - Most common in premature neonates
  - Treatment: NPO, IV fluids, ↓ caloric intake ± gastric lavage & saline dissolution; surgery if necessary
- Phytobezoar
  - Most common in adults
  - Treatment: Endoscopic fragmentation ± suction; gastric lavage with enzymatic dissolution; surgery if necessary

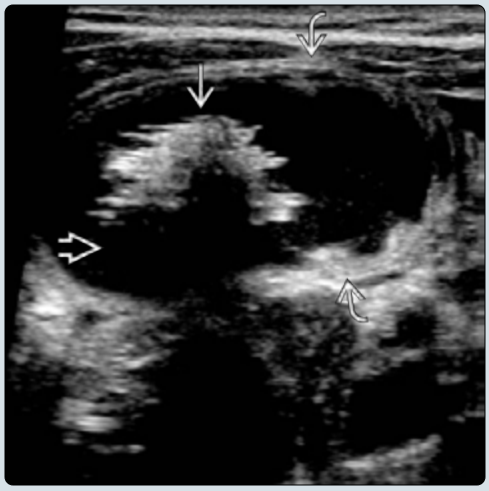
(Left) *Frontal image from an upper GI series in a 6-year-old girl with trichotillomania complaining of pain & emesis shows a mobile ovoid filling defect ⇨ with air & barium in its interstices, consistent with a trichobezoar.* **(Right)** *Axial CECT from a 13-year-old girl with abdominal pain & a palpable mass shows concentric rings ⇨ & trapped air ⇨ in the interstices of the mass. The mass conforms to the stomach contour & is surrounded by contrast. A trichobezoar was removed surgically.*





(Left) *Frontal supine abdominal radiograph of a 7-year-old girl with pain, emesis, & alopecia shows multiple loops of dilated small bowel ⇨ concerning for a small bowel obstruction.* **(Right)** *Longitudinal grayscale ultrasound from the same patient shows a mass impacted in the distal ileum. The surrounding bowel is dilated & fluid filled ⇨. The mass has a broad, echogenic, arc-like surface ⇨ with dense posterior acoustic shadowing ⇨. A trichobezoar was removed at surgery.*





TERMINOLOGY

Definitions

- Specific ingested materials accumulate to form indigestible & potentially obstructive mass in gastrointestinal (GI) tract
- Classified based on ingested material, including
  - Trichobezoar: Hair
  - Phytobezoar: Indigestible food components, typically vegetable or fruit fibers
  - Lactobezoar: Concretion of coagulated milk & mucous proteins
- Rapunzel syndrome: Gastric trichobezoar that extends distally through pylorus

IMAGING

Radiographic Findings

- Radiography
  - Mottled appearance of solid matter in (typically proximal) GI tract due to lucencies of gas in interstices of accumulated ingested material
    - Nonspecific: Abscess, stool, or recently ingested food can have similar appearance
  - Bezoars rarely identified on radiographs alone
  - ± evidence of small bowel obstruction: Dilated proximal bowel with air-fluid levels

Fluoroscopic Findings

- Upper GI
  - Round, ovoid, or tubular filling defects
  - May have mottled appearance due to contrast within interstices of accumulated ingested material
  - Gastric bezoars
    - Majority freely mobile
    - Gastric dilation with ↓ or absent peristalsis
  - Small bowel bezoars commonly present with findings of obstruction
    - Impacted filling defect
    - Dilated, air/fluid-filled loops of proximal bowel

Ultrasonographic Findings

- Intraluminal mass with broad, hyperechoic, arc-like surface & posterior acoustic shadowing
  - "Dirty" shadowing due to intermixed air
  - Lactobezoar lacks posterior acoustic shadowing
- Dilated, fluid-filled bowel in cases of obstruction
  - Compression of bowel loops may show fluid shifting around intraluminal mass
- May see color Doppler twinkle artifact at hyperechoic surface
  - No true internal vascularity in mass
- Low sensitivity to detect multiple bezoars & gastric bezoars
  - Only 2/5 cases with multiple intestinal bezoars identified as such in 1 study
  - Same study identified only 2/8 gastric bezoars associated with intestinal bezoars

CT Findings

- Circumscribed intraluminal mass with interstitial air
- Mass may have concentric ring architecture
- Seeds from persimmon fruit may appear as hyperdense foci in mass
- Gastric bezoars
  - May float or fill lumen
  - May not be identified on soft tissue windows: Window setting of -100 HU may improve sensitivity
- Small bowel bezoars located at bowel transition point in cases presenting with obstruction
  - Appearance strongly mimics small bowel feces sign
    - Bezoars may extend distally into nondilated bowel
    - Floating fat densities in dilated bowel loops proximal to obstruction may help distinguish from small bowel feces
      - Floating fat density debris sign reported using window level of -50 HU & width of 500 HU
    - Mean attenuation < -11.75 HU & length < 9.5 cm have also been reported to help differentiate from small bowel feces
- Up to 97% sensitivity for gastrointestinal bezoars

Imaging Recommendations

- Abdominal radiographs typically obtained due to presenting symptoms
- CECT
  - Advantages
    - High sensitivity
    - Can identify multiple bezoars
    - Allows evaluation of entire abdomen
  - No consensus on use of oral contrast
  - Often ordered in setting of suspected small bowel obstruction
- Upper GI
  - Dynamic study allows evaluation of intraluminal mass mobility
  - Can identify mucosal injuries to gastric & bowel wall
  - Ideally performed after fasting

DIFFERENTIAL DIAGNOSIS

Small Bowel Feces

- Classically seen in dilated bowel just proximal to transition point of obstruction
- Longer length, greater density, & lack of encapsulated appearance more likely than bezoars

Ingested Material

- Recently ingested food in stomach can simulate bezoar on radiographs
- Food & gas in stomach show "dirty" shadowing on US
  - Gastric food should be minimal with adequate NPO ("nothing by mouth") status for exam

Neoplasm

- Intraluminal & adjacent extraluminal masses: Lymphoma, neuroblastoma, juvenile polyp, etc.
- Solid mass with visible internal vascularity by color Doppler
- Calcified masses may simulate US features of bezoars
  - True calcification not typical of bezoars
- Intraluminal neoplasm has connection to gastric/bowel wall

Gallstone Ileus

- Gallstone may have similar appearance to bezoar on US
- Pneumobilia classic feature in gallstone ileus
- Extremely uncommon in children

PATHOLOGY

General Features

- Trichobezoar
  - Hair resistant to digestion & peristalsis
  - Forms concretion with mucus + food
- Lactobezoar
  - GI tract capacity overwhelmed
    - Caloric & protein intake > age-related dietary reference (erroneous preparation of formula, low birth-weight formula)
    - Medication-induced antagonization of gastric secretion & motility
    - Likely multifactorial as also seen with breast milk & cow's milk
  - GI tract capacity overload → vomiting & diarrhea → dehydration → excessive water reabsorption → lactobezoar
- Phytobezoar
  - Indigestible fruit & vegetable fibers: Cellulose, tannin, lignin
    - Persimmon common culprit
  - Gastroparesis/↓ gastric peristalsis: Major risk factor; may be due to
    - Gastric surgery
    - Medical conditions: Hypothyroidism, diabetes, cystic fibrosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Palpable abdominal mass, abdominal pain, vomiting, distention, dysphagia, small bowel obstruction
- Other signs/symptoms
  - Trichobezoar: Alopecia, psychiatric conditions (anxiety, trichotillomania, trichophagia, depression)
  - Lactobezoar: Dehydration, diarrhea, respiratory distress, weight loss/failure to thrive, feeding intolerance
  - Phytobezoar: Small bowel obstruction most common presentation
  - Lamerton sign: Mobile, palpable, & indentable mass in upper abdomen

Demographics

- Trichobezoar
  - Most common bezoar in pediatric population
  - Almost all documented cases female
- Lactobezoar
  - > 70% of documented cases: Patient ≤ 30 days old
  - > 75% of documented cases: Patient born prematurely
- Phytobezoar
  - Most common in adult population
  - Prevalence varies among ethnic groups & geographic locations relating to food preferences

Natural History & Prognosis

- Complications
  - Obstruction, mucosal erosion/ulceration, GI bleeding, perforation
  - Less common: Intussusception, jaundice, protein-losing enteropathy, pancreatitis

Treatment

- Trichobezoar
  - Laparotomy typically required due to bezoar size; allows evaluation of GI tract
  - Laparoscopy reported
  - Endoscopy valuable for diagnosis
    - Poor therapeutic option: No success in series of 40 reported cases due to obstruction
      - Large size of bezoars
      - Bezoars resistant to fragmentation
      - With fragmentation, components may impact distally
  - Psychiatric referral critical to prevent recurrence
- Lactobezoar
  - Conservative management: NPO, IV fluids, ↓ caloric density
  - Gastric lavage with saline + N-acetylcysteine if conservative management fails
  - Surgery for perforation or failure of conservative management & gastric lavage
- Gastric phytobezoar
  - Endoscopic fragmentation ± suction retrieval
  - Gastric lavage with enzymatic or biochemical dissolution
    - N-acetylcysteine, papain, metoclopramide, cellulase, Coca-Cola

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Adjust CT window level & width: ↑ sensitivity to bezoars

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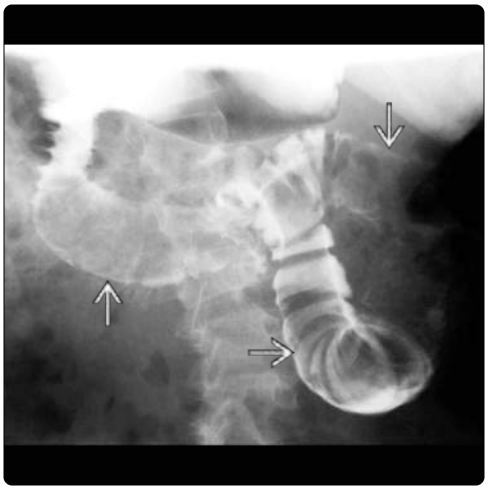
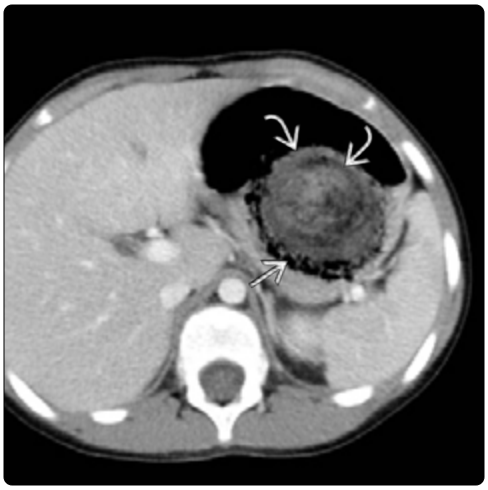
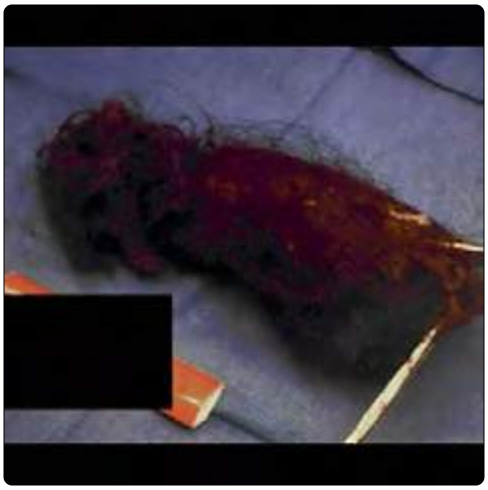
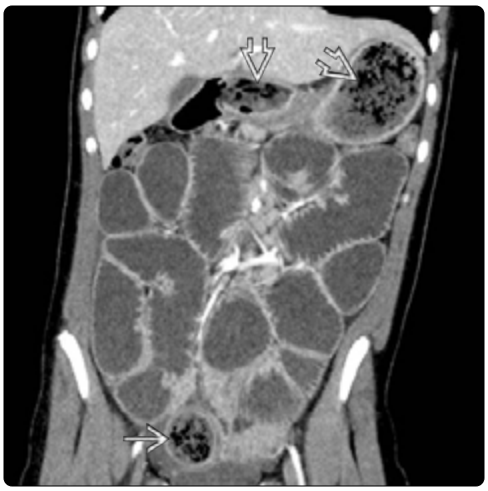


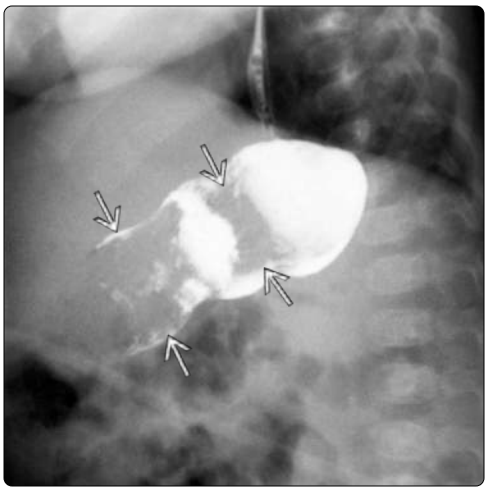
(Left) *Frontal image from an upper GI series demonstrates an elongated filling defect in the duodenum & proximal jejunum that is surrounded by contrast & conforms to the bowel contour* ➡. **(Right)** *Surgical photograph of the specimen removed from the proximal small bowel in the same patient shows an elongated trichobezoar.*





(Left) *Axial CECT from an 8-year-old girl with a history of trichotillomania, trichophagia, & reports epigastric pain shows an intraluminal mass with concentric architecture* ➡. *Air is trapped within the peripheral interstices* ➡ *of the mass, consistent with a bezoar.* **(Right)** *Specimen image from the same patient demonstrates the large trichobezoar that was removed from the stomach during laparotomy.*





(Left) *Coronal CECT from a 14-year-old girl with abdominal pain & emesis shows diffuse dilation of fluid-filled small bowel with a mass at the transition point* ➡. *The appearance of the mass mimics small bowel feces but was confirmed to be an impacted trichobezoar. A large gastric bezoar* ➡ *was also confirmed.* **(Right)** *Lateral view from an upper GI series in a 3-month-old boy with failure to thrive & emesis shows a large fragmented filling defect in the stomach* ➡. *A lactobezoar was confirmed at endoscopy.*

Gastrointestinal Duplication Cysts

KEY FACTS

TERMINOLOGY

- GI duplication cysts have 3 defining characteristics
  - Well-developed coat of smooth muscle
  - Epithelial lining representing some part of GI tract
  - Attachment to (± communication with) GI tract
    - Rarely: Isolated cyst with no persistent attachment

IMAGING

- Cystic (80-90%) vs. tubular (10-20%) lesions with well-defined wall
  - Can occur anywhere along GI tract; most frequently associated with jejunum/ileum (53%) & esophagus (18%)
- Contiguous (but not necessarily communicating) with adjacent GI tract
- Ultrasound best modality to visualize key features, though findings with highest specificity have lowest sensitivity
  - Gut signature: Trilaminar appearance of wall less specific than 5-layered wall
  - Y sign: Hypoechoic muscularis propria divided at point of attachment between cyst & adjacent bowel
  - Peristalsis of cyst wall pathognomonic (when visualized)

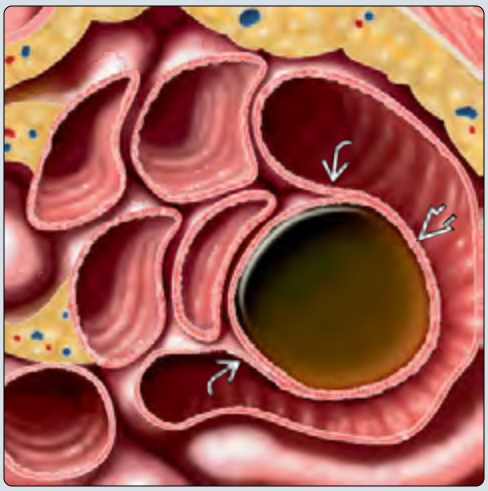
TOP DIFFERENTIAL DIAGNOSES

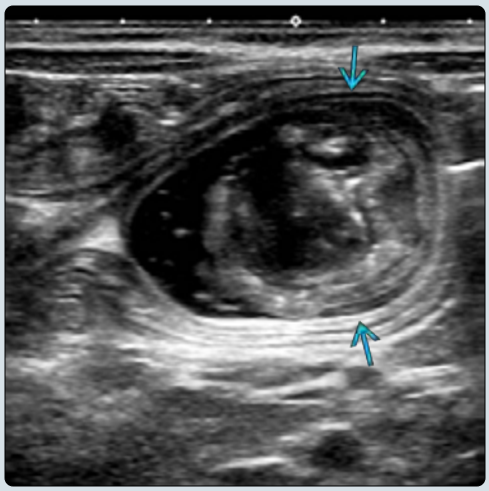
- Mesenteric lymphatic malformation
- Ovarian cyst
- Meconium pseudocyst
- Urachal cyst
- Hydrometrocolpos
- Choledochal cyst
- Aneurysm

CLINICAL ISSUES

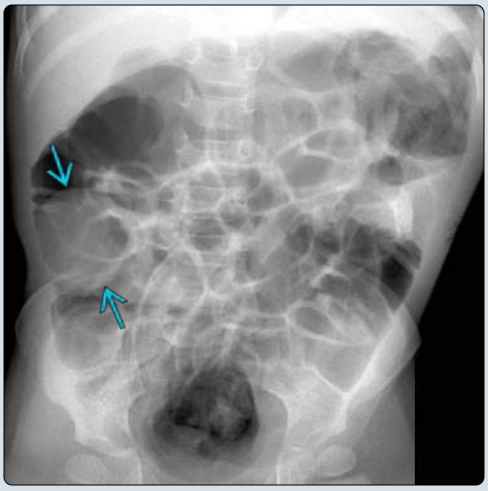
- Congenital lesions, most often presenting < 2 years of age
  - Pain, mass, rectal bleeding; may be incidental
  - Small bowel obstruction or intussusception most frequent presentation for ileal duplications
- Complete surgical resection ideal

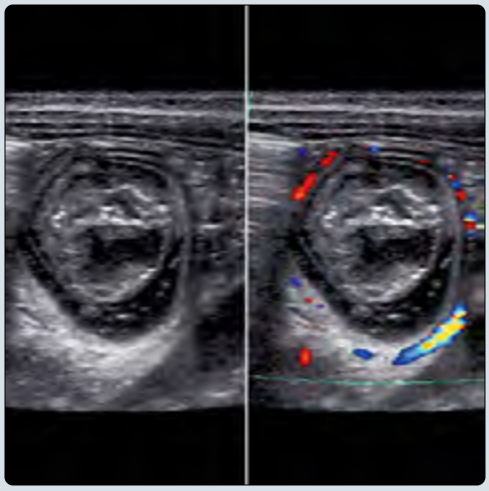
(Left) *Axial graphic shows an enteric duplication cyst between sectioned bowel loops. Note that the muscular layer of the bowel is continuous with that of the duplication cyst, creating a Y as the muscle splits.* **(Right)** *Transverse ultrasound of the upper abdomen in a 15 month old with intermittent pain & vomiting shows an ileocolic intussusception extending to the transverse colon & terminating in a complex cystic mass with a trilaminar wall. The mass was initially mistaken for fluid trapped around the intussusceptum.*





(Left) *Frontal radiograph in the same patient after successful air-enema reduction (confirmed by free reflux of air into the small bowel) shows a persistent round mass outlined by air in the mid right abdomen, concerning for a lead point.* **(Right)** *Transverse grayscale (left) and color Doppler (right) ultrasounds in the same patient after the air-enema reduction show the residual complex cystic mass with no internal vascularity. This mass was found to be an ileocecal duplication cyst with internal debris upon resection.*





TERMINOLOGY

Synonyms

- Foregut/midgut/hindgut malformation or duplication, enteric cyst, enterocyst, enterocystoma, enterogenous cyst, dorsal enteric cyst, alimentary tract duplication

Definitions

- GI duplication cysts have 3 defining characteristics
  - Well-developed coat of smooth muscle
  - Epithelial lining representing some part of GI tract
  - Attachment to (± communication with) GI tract
    - Ultra rare isolated cysts: No longer attached
  - Neurenteric cysts often have associated vertebral & intraspinal anomalies
    - Most frequent with esophageal duplications

IMAGING

General Features

- Best diagnostic clue
  - Cystic lesion with well-defined wall; contiguous (but not necessarily communicating) with adjacent GI tract
  - Cyst wall has similar appearance to bowel (gut signature) on ultrasound
  - Peristalsis pathognomonic (but not always visualized)
- Location
  - Can occur anywhere along alimentary tract
    - 75% abdominal, 20% thoracic
  - Most frequently associated with jejunum/ileum (53%) & esophagus (18%)
  - Less common: Colon (13%), stomach (7%), duodenum (6%), floor of mouth (1%), retrorectal (4%), retroperitoneal (< 1%), intrapancreatic (< 1%)
  - Esophageal duplications
    - Right > left; inferior > superior
  - Gastric duplications
    - Typically occur at greater curvature or posterior wall
  - Duodenal duplications
    - 2nd or 3rd portions most common
  - Tubular duplications
    - Can be short or very long
      - Rarely, complete duplication of GI tract segment
    - Tubular form more likely to extend above & below diaphragm
  - Multiple cysts: 1-15%
    - Single GI segment affected more frequently than different segments
  - Isolated cyst (separated from GI tract): Ultra rare
- Morphology
  - Cyst (80-90% of duplications)
    - Spherical, ovoid, or dumbbell in shape
    - Sharing bowel wall > > isolated cyst in peritoneum with mesenteric stalk
  - Tubular duplication (10-20%)
    - Perpendicular branch of bowel with solitary communication vs. parallel tube (double-barrel configuration) communicating at both ends

Radiographic Findings

- Abdomen
  - Displacement of bowel gas (depending on size of cyst)
  - ± bowel obstruction
- Chest: Middle or posterior mediastinal/paraspinal mass

Fluoroscopic Findings

- Contrast by mouth or rectum may demonstrate continuity of duplication with normal GI tract

CT Findings

- CECT
  - Ovoid, round, or tubular lesion
  - ± thick, enhancing wall, especially if inflamed
  - ± fluid-debris levels
  - Duplication may be difficult to separate from adjacent normal bowel, especially if no oral contrast given

MR Findings

- Best for intraspinal extension of neurenteric cysts
- MR enterography may have increasing role

Ultrasonographic Findings

- Grayscale ultrasound
  - Gut signature of cyst wall
    - Trilaminar appearance classically described but not specific
      - Deep → superficial layers: Hyperechoic mucosa, hypoechoic muscle, hyperechoic serosa
    - 5-layered wall difficult to visualize but more specific
      - Deep → superficial layers: Hyperechoic mucosa, hypoechoic muscularis mucosa, hyperechoic submucosa, hypoechoic muscularis propria, hyperechoic serosa
    - Wall signature may be disrupted by inflammation, ulceration, or perforation
  - Y sign (specific): Split hypoechoic muscularis propria at point of attachment between cyst & adjacent bowel
  - Peristalsis uncommon but specific
  - Variable cystic contents
  - Wall calcification rare
  - May shift locations slightly in abdomen between exams
- Color Doppler
  - ± vascularity in wall

Nuclear Medicine Findings

- Tc-99m pertechnetate uptake in duplications containing ectopic gastric mucosa

Imaging Recommendations

- Best imaging tool
  - Ultrasound for pediatric abdominal cyst
  - CT or MR for intrathoracic lesions

DIFFERENTIAL DIAGNOSIS

Mesenteric Lymphatic Malformation

- Cystic, often with fluid-fluid levels from hemorrhage
- Unilocular vs. multiseptated
- Round, well-defined vs. irregular, infiltrating
- Microcystic components may appear solid

Ovarian Cyst

- Simple vs. hemorrhagic vs. mixed cystic & solid
- Follicles visible in distorted ovary

Meconium Pseudocyst

- Localized complex collection (debris, Ca^{2+}) secondary to in utero bowel perforation
- Bowel obstruction usually present

Urachal Cyst

- Lies along midline tract from bladder dome to umbilicus

Hydrometrocolpos

- Moderate to marked vaginal distention in neonate or teenager
- Relatively small uterus projects off superior aspect of obstructed vagina

Choledochal Cyst

- Right upper quadrant cyst, often elongated, communicating with biliary system

Aneurysm

- Uncommon in children but may appear as cystic mass
- Fills with color flow on Doppler

PATHOLOGY

General Features

- Etiology
  - Pathogenesis remains unclear; theories include
    - Split notochord theory
    - Possible early embryologic error in branching/diverticularization
    - Error in epithelial recanalization
    - Fusion of longitudinal folds along bowel
    - Result of vascular compromise

Gross Pathologic & Surgical Features

- Attachment ± communication with parent bowel much more common than complete isolation

Microscopic Features

- GI duplication cysts lined with either stratified squamous or columnar epithelium
- Supported by muscular & serosal layers
- 50-60% contain gastric mucosa or pancreatic tissue
- Duplications secreting alimentary tract substances (gastric acid, pancreatic enzymes, mucus) predisposed to rupture & hemorrhage

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, mass, rectal bleeding
  - Small bowel obstruction or intussusception most frequent presentation for ileal duplications
  - May be incidental
- Other signs/symptoms
  - Ulceration, perforation, hemorrhage
  - Vascular compromise, urinary retention reported from mass effect
  - Respiratory symptoms in thoracic & oral cavity lesions
  - May be detected on prenatal sonography

Demographics

- Age
  - Congenital lesions, most often presenting < 2 years of age
  - 30% present in adulthood
- Epidemiology
  - F:M = 2:1
  - Prevalence of 1 per 4,500 fetal/neonatal autopsies

Natural History & Prognosis

- Very good prognosis overall
- Complications include ulceration, perforation, hemorrhage, volvulus, intussusception
- Pancreatitis, acute or chronic, if cyst near pancreas
- Rare reports of malignant transformation
  - Most common with colonic duplications

Treatment

- Complete surgical resection ideal
- Partial excision with removal of lining or marsupialization may be required in difficult regions

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Look for sonographic gut signature of cyst wall (5-layered wall more specific than trilaminar appearance)
  - Trilaminar wall may also be seen with
    - Fluid-distended hollow organs (bowel, vagina, urinary bladder) or Meckel diverticulum
    - Alternate histologies occurring in layers (pseudogut signature)
    - Artifact

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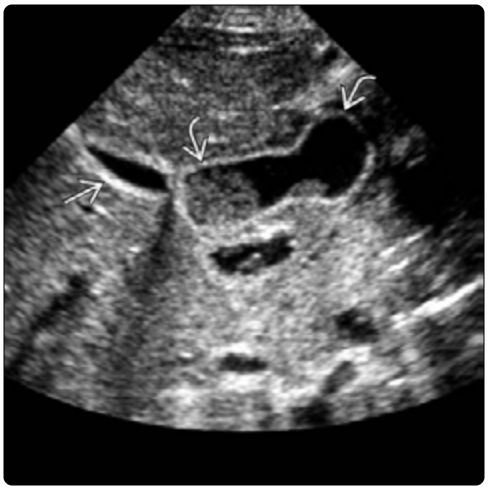
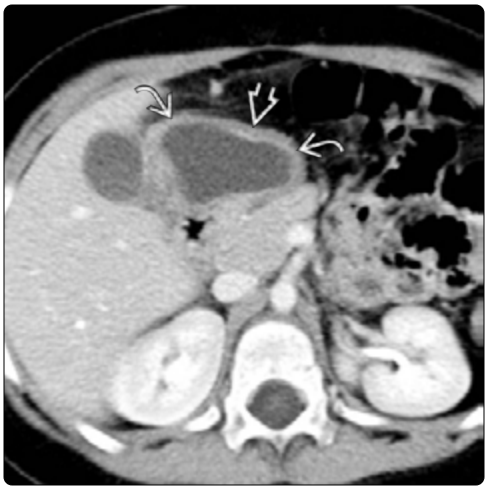
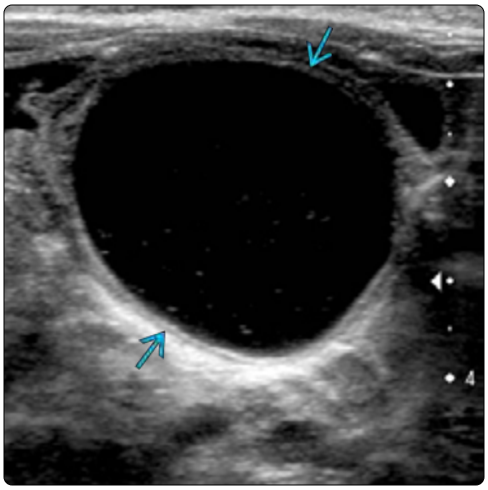
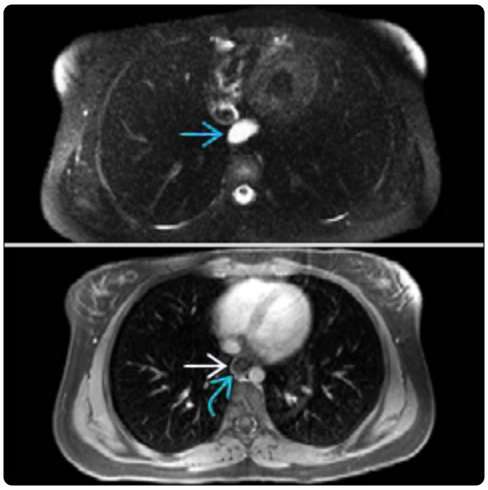


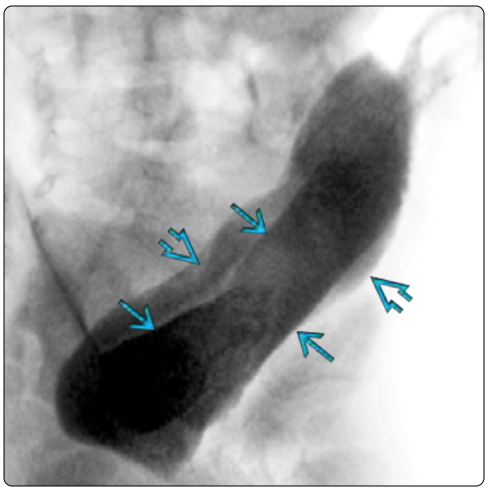


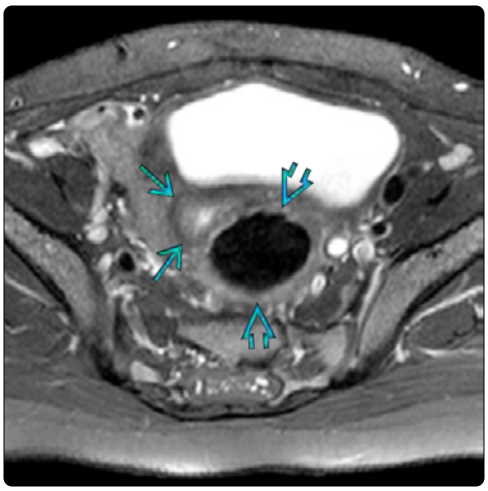
(Left) *Transverse ultrasound shows echogenic debris in a cystic mass adjacent to the liver margin in a newborn. The mass, which abuts the gallbladder, was seen on prenatal ultrasound & favored to represent an enteric duplication cyst. The baby was asymptomatic and was temporarily lost to follow-up.* **(Right)** *Axial CECT in the same patient, now 4 years old & complaining of postprandial pain, shows a cystic lesion with a thickened trilaminar wall. Resection confirmed a duodenal duplication cyst.*





(Left) *Transverse ultrasound of the abdomen in a neonate shows a well-circumscribed round mass with minimal internal debris, posterior acoustic enhancement, & trilaminar wall. An ileal duplication cyst was confirmed at surgery.* **(Right)** *Axial T2 FS (top) & T1 C+ FS (bottom) MRs in a patient with chest pain show a well-circumscribed, ovoid T2 hyperintense mass with mild rim enhancement along the anterior margin of the esophagus. An esophageal duplication cyst was confirmed upon resection.*





(Left) *Oblique high-pressure distal colostogram in an anorectal malformation patient shows overlapping opacification (from sequential ostomy injections) of parallel functional & nonfunctional limbs of a rectosigmoid tubular duplication.* **(Right)** *Axial T2 FS MR of the pelvis in the same patient shows the collapsed nonfunctional limb of the tubular duplication adjacent to the gas-distended functional rectosigmoid colon.*

Small Bowel Intussusception

KEY FACTS

TERMINOLOGY

- Telescoping of proximal small bowel segment (intussusceptum) into contiguous distal small bowel segment (intussuscipiens)

IMAGING

- Bowel-within-bowel appearance: Alternating layers of bowel wall & mesenteric fat
- Target appearance in cross section on CT & US
- Continuity of central fat with mesenteric fat
- Mesenteric vessels extending into or out of mass
- Crescent of gas or fluid between intussusceptum & intussuscipiens
- Small bowel intussusception (SBI) features that differentiate it from ileocolic intussusception (ICI)
  - Smaller diameter (mean of 1.5 vs. 2.6 cm)
  - Less hyperechoic mesenteric fat centrally
  - Entrapped lymph nodes less common
  - More common in periumbilical or left abdomen

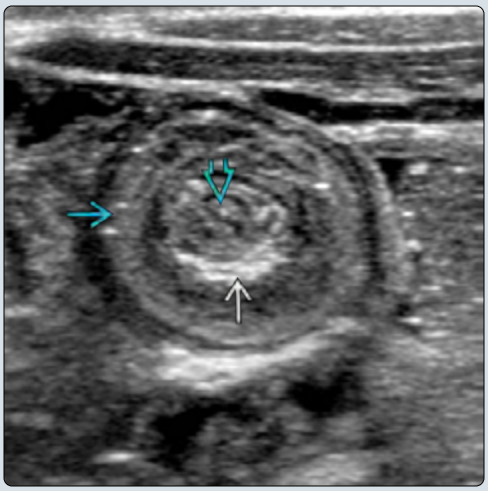
PATHOLOGY

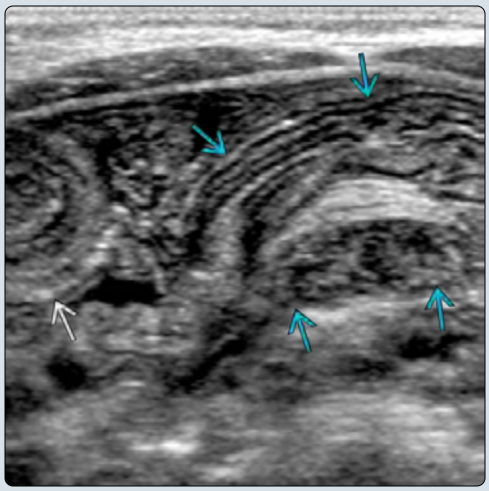
- Lead points (uncommon): Lymphoid hyperplasia, Meckel diverticulum, duplication cyst, adhesions, polyps, intramural hematoma, foreign body, enteric tubes
- Associated pathology: Henoch-Schönlein purpura, malabsorption syndromes (celiac), cystic fibrosis
- May occur postoperatively

CLINICAL ISSUES

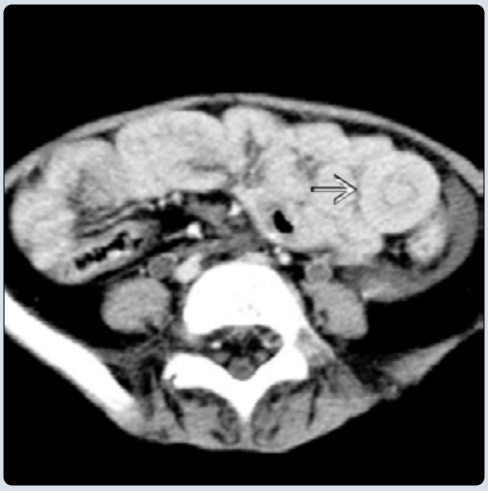
- Majority self-limited & spontaneously reduce
- Sonographic findings that may indicate need for surgery
  - Length > 3.5 cm
  - Findings of small bowel obstruction (SBO) & ascites
  - Pathologic lead point (but US has low sensitivity)
- Postoperative SBI typically surgically reduced
- Complications more likely with delayed presentation & diagnosis: SBO, ischemia, necrosis

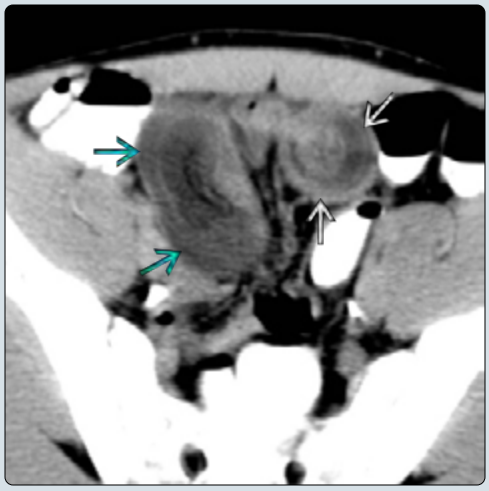
(Left) *Transverse periumbilical US from a 14 month old with pain & vomiting shows a target appearance of the bowel that is consistent with an intussusception. A crescent of hyperechoic mesenteric fat is interposed between the intussuscipiens & intussusceptum. The location & diameter are typical of a small bowel intussusception (SBI).* **(Right)** *US from the same patient demonstrates a SBI > 3 cm in length in both the longitudinal & axial planes.*





(Left) *Axial CECT in a child with pancreatitis shows an incidental left lower quadrant SBI. Most SBIs are asymptomatic & resolve without intervention.* **(Right)** *Axial CECT shows a long (> 3.5 cm length) SBI in both axial & longitudinal planes. An underlying large Meckel diverticulum was found at surgery.*





TERMINOLOGY

Definitions

- Telescoping of proximal small bowel segment (intussusceptum) into contiguous distal small bowel segment (intussuscipiens)

IMAGING

General Features

- Best diagnostic clue
  - Bowel-within-bowel appearance with alternating layers of bowel wall & mesenteric fat
  - Layering target appearance in cross section
  - Continuity of fat in mass with mesenteric fat
  - Mesenteric vessels extending into or out of mass
  - Crescent of gas or fluid between intussusceptum & intussuscipiens
  - ± proximal bowel dilatation
  - ± pathologic lead point

Ultrasonographic Findings

- Layered alternating bands of hypoechoic bowel wall & hyperechoic mesenteric fat
  - Target appearance in cross section
  - ± pseudokidney appearance in long axis: Hyperechoic mesenteric fat centrally mimics renal sinus fat; hypoechoic bowel wall mimics renal cortex
- Small bowel intussusception (SBI) features that differentiate it from ileocolic intussusception (ICI)
  - Smaller diameter
    - Mean diameter of 1.5 cm (range 1.1 cm to 2.5 cm)
      - Size may ↑ with age, edema, hemorrhage or intramural mass
  - Less hyperechoic mesenteric fat centrally
    - Thin crescent or no visible fat
    - Ratio of central fat core diameter to outer wall thickness > 1.0 in all ICI & < 1.0 in all SBI
  - SBI more common in periumbilical or left abdomen
  - Entrapped lymph nodes less common in SBI
- Majority transient & spontaneously reduce during exam
- Sonographic findings that may indicate need for surgery
  - Length of SBI > 3.5 cm
    - Sensitive (93%) & specific (100%) independent predictor of need for surgery
  - Findings of small bowel obstruction (SBO) & ascites
  - Pathologic lead point (but US has low sensitivity)

CT Findings

- Layered alternating bands of ↑ (enhancing bowel wall) & ↓ (mesenteric fat ± fluid) attenuation
- Target appearance in transverse plane

Imaging Recommendations

- US for initial imaging evaluation
  - Should be able to clearly trace small bowel into & out of intussusception (further differentiates SBI vs. ICI)
- CECT occasionally used to further investigate if
  - Symptomatic patient
  - Findings present that may indicate need for surgery

DIFFERENTIAL DIAGNOSIS

Ileocolic Intussusception

- ICI extends from right lower quadrant into right upper quadrant
- ICI mean diameter of 2.6 cm in cross section
- Entrapped lymph nodes more likely with ICI
- Vomiting, bloody stool, leukocytosis more likely with ICI

Meckel Diverticulum

- Tubular mass (± gut signature on US)
- Complications (20%): Obstruction, bleeding, perforation, & intussusception

Henoch-Schönlein Purpura

- Hypersensitivity reaction with small vessel vasculitis
- Purpuric rash on legs or extensor surface of arms
- Mural bleed predisposes to intussusceptions

PATHOLOGY

General Features

- Etiology
  - Abnormal peristalsis → invagination of proximal bowel & mesenteric fat into contiguous bowel segment
  - Lead points (uncommon): Lymphoid hyperplasia, Meckel diverticulum, duplication cyst, polyps, intramural hematoma, foreign body, enteric tubes
  - Associated pathology: Henoch-Schönlein purpura, malabsorption syndromes (celiac), cystic fibrosis
  - Prior abdominal or nonabdominal procedures
    - > 85% of postoperative intussusceptions SBI

CLINICAL ISSUES

Presentation

- Abdominal pain, distension, vomiting, blood in stool
- 65% asymptomatic in one study

Natural History & Prognosis

- Majority self-limited & spontaneously reduce

Treatment

- Asymptomatic patient without concerning imaging features: Conservative management (often spontaneously reduces during study)
- Symptomatic patient or concerning imaging features: Surgical consult ± CECT/follow-up US

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Henoch-Schönlein Purpura

KEY FACTS

TERMINOLOGY

- Immune complex-mediated small vessel vasculitis commonly affecting skin, GI tract, urologic system, joints

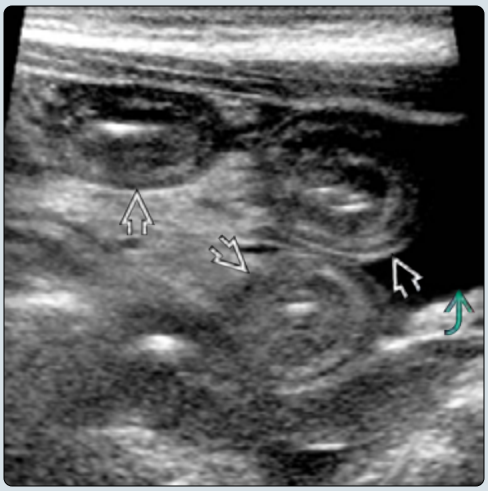
IMAGING

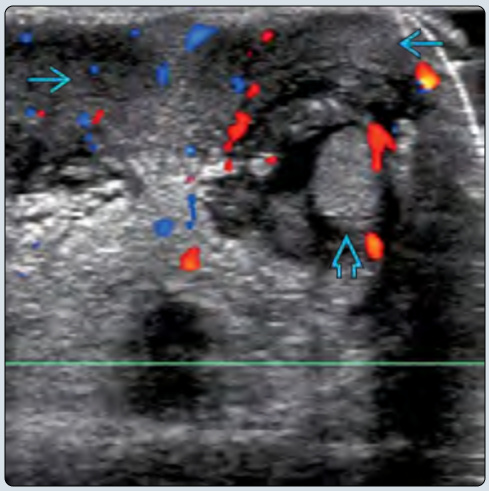
- GI features (up to 75%)
  - Circumferential bowel wall thickening of discontinuous segments of variable lengths
    - "Thumbprinting" on radiographs
    - Due to intramural hemorrhage &/or edema
  - Intussusception, commonly ileoileal
- Urologic features (up to 60%)
  - Henoch-Schönlein purpura (HSP) nephritis
    - Normal or bilaterally enlarged & echogenic kidneys
  - Stenosing ureteritis
    - Hydroureteronephrosis & thick urothelium
  - Scrotal wall thickening, edema, & hyperemia
- Arthritis/arthralgias (up to 82%)
- Neurologic features (up to 2%)
  - Cerebral edema, intracranial hemorrhage, cerebral vein thrombosis, posterior reversible encephalopathy syndrome
- Pulmonary features (up to 5%)
  - Diffuse alveolar hemorrhage (DAH)
    - Alveolar infiltrates & ground-glass opacities
    - Pleural effusions, often large & requiring chest tube

CLINICAL ISSUES

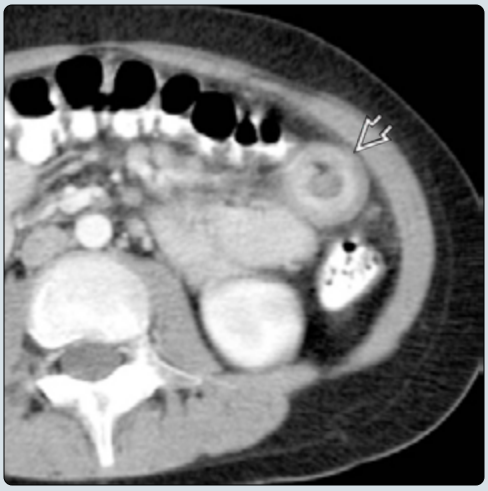
- Most common primary pediatric vasculitis at 49%
- Purpura or petechiae ultimately in 100%
- Peak age: 7 years
- Typically self-limited, resolves in 3-4 weeks
  - Treatment largely conservative & directed to specific systems involved
  - Recurrence in up to 1/3 of cases
  - HSP nephritis: 20% develop nephritic/nephrotic syndrome
  - DAH: Up to 28% mortality

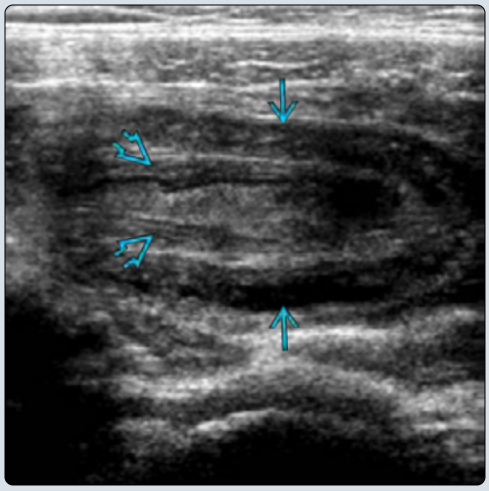
(Left) *Transverse US through the right lower quadrant in an 8 year old with Henoch-Schönlein purpura (HSP) demonstrates multiple thick-walled loops of small bowel & a small amount of free fluid.* **(Right)** *Transverse color Doppler US of the scrotum shows marked thickening, edema, & hyperemia of the scrotal wall in a 6-year-old patient with HSP. A normal left testicle is also seen.*





(Left) *Axial CECT in a 9 year old with HSP, vomiting, & abdominal pain demonstrates a thick-walled loop of the small bowel with mucosal hyperenhancement & mild surrounding mesenteric inflammation.* **(Right)** *Longitudinal US from a 4 year old with HSP & abdominal pain shows a small bowel intussusception with well-delineated intussuscipiens & intussusceptum. This intussusception was > 5 cm long & required surgical reduction.*





TERMINOLOGY

Abbreviations

- Henoch-Schönlein purpura (HSP)

Definitions

- Systemic nonthrombocytopenic immune complex-mediated small vessel vasculitis

IMAGING

General Features

- Multisystem diffuse vasculitis most commonly affecting gastrointestinal (GI) tract, urologic system, joints, & (to lesser extent) nervous system & lungs
- Imaging findings nonspecific & must be considered in context of clinical & laboratory findings to make diagnosis

Gastrointestinal Findings

- Bowel wall edema with submucosal & intramural hemorrhage
  - Circumferential bowel wall thickening involving discontinuous segments of variable lengths
  - "Thumbprinting" from fold thickening on radiographs
  - Hypo- or hyperattenuating bowel wall on CT
  - Hypomotility of involved bowel loops
- Mesenteric vascular engorgement
- ↑ attenuation or echogenicity of mesenteric fat
- Ileus or obstruction
- Intussusception occurs in up to 14% of HSP cases
  - Ileoileal most common, ~ 50%
- Rarely: Ischemia, pneumatosis, & spontaneous bowel perforation
- Nonspecific lymphadenopathy
- Hydropic gallbladder

Urologic Findings

- HSP nephritis in 30-50% of HSP cases
  - Normal or bilaterally enlarged kidneys
  - Diffusely ↑ echogenicity of renal cortex
- Stenosing ureteritis
  - Secondary to ureteral/periureteral vasculitis
  - Hydroureteronephrosis, urothelial thickening
- Bladder & ureteral wall hematomas
- Acute scrotum
  - Scrotal wall edema & hyperemia ± hydrocele
  - Normal or slightly hyperemic testicles & epididymis
  - ± spermatic vein thrombosis
- Penile HSP
  - Diffuse edema of shaft & foreskin
  - Penile purpura may precede extremity rash
    - Hypoechoic avascular lesion
  - Thrombosis & priapism

Joint Findings

- Arthritis with synovitis & effusion, soft tissue swelling

Neurologic Findings

- Cerebral edema
- Intracranial hemorrhage
- Cerebral vein thrombosis
- Posterior reversible encephalopathy syndrome
  - Patchy cortical & subcortical white matter lesions most pronounced in posterior circulation distribution (parietal, occipital, cerebellar)
    - Hypodense on CT; ↓ T1, ↑ T2 & FLAIR on MR, most without diffusion restriction
  - Due to hypertension or cerebral vasculitis

Pulmonary Findings

- Edema related to kidney failure
- Diffuse alveolar hemorrhage (DAH) in 0.8-5%
  - Alveolar infiltrates & ground-glass opacities
  - Reticulonodular opacities less common
  - Pleural effusions, often large & requiring chest tube

DIFFERENTIAL DIAGNOSIS

Idiopathic Thrombocytopenic Purpura

- Life-threatening multisystem disorder with disseminated microvascular thrombi
- ↓ platelets (whereas HSP may have ↑ platelets)
- Anemia, petechiae, microscopic hematuria

Hemolytic Uremic Syndrome

- Microvascular lesions with platelet aggregation
- Caused by enterohemorrhagic strains of *Escherichia coli*, especially *E. coli* 0157:H7
- Transient, severe, crampy abdominal pain with bloody diarrhea; precedes renal failure by 3-14 days

Infectious or Inflammatory Colitis

- Abdominal pain & bloody diarrhea
- Pseudomembranous colitis characterized by fever, diarrhea, & diffuse colonic mucositis, typically while on antibiotics
  - Toxin produced by *Clostridium difficile* most important cause of antibiotic-associated colitis
- Crohn disease most commonly with ileocecal involvement

Child Abuse

- Bruising on extremities may simulate HSP rash
- Bowel hematomas can be seen in both HSP & child abuse

Juvenile Idiopathic Arthritis

- Arthralgias involving 1 or more joints: Knee (90%), ankle (70%), wrist (70%)
  - Joint effusion, synovial thickening/hyperemia, bone marrow changes ± erosions
- ± rash

Leukemia

- Numerous possible symptoms & signs, including petechiae, pain, swelling, adenopathy

Testicular Torsion

- Doppler demonstrates ↓ or absent blood flow of acutely painful testicle

Scrotal Cellulitis

- Lacks other features of HSP
- May have history of insect bite

PATHOLOGY

General Features

- Etiology
  - Inflammatory disorder of unknown cause; characterized by specific immune complexes in venules, capillaries, & arterioles

Gross Pathologic & Surgical Features

- HSP nephropathy demonstrates mesangial hypercellularity, endocapillary proliferation, necrosis, cellular crescents, leukocyte infiltration
- HSP has no specific diagnostic laboratory markers
  - Serum IgA levels ↑; platelets ± normal or high

Microscopic Features

- Leukocytoclastic angiitis initiated by deposition of immune complexes
- Renal biopsy indistinguishable from IgA nephropathy
  - Changes can be minimal mesangial proliferation to severe necrotizing glomerulonephritis
  - Glomerular crescents associated with poor renal outcome & development of end-stage renal disease
- Pulmonary involvement
  - DAH predominate finding

CLINICAL ISSUES

Presentation

- Consensus criteria for HSP
  - Purpura or petechiae with lower limb predominance & at least 1 of following
    - Abdominal pain
    - Arthritis or arthralgia
    - Renal involvement
    - IgA deposition on biopsy
- Purpura or petechiae, 100%
  - Lower limb predominance
  - May develop bullous lesions
  - May not be initial manifestation
- Arthralgia & arthritis, up to 82%
  - Typically oligoarthritis of lower limbs
  - May precede rash in 25% of cases
- GI disturbances, 50-75%
  - Abdominal pain, vomiting, GI bleeding (which may be massive), intussusception, pancreatitis, hydropic gallbladder, protein-losing enteropathy
  - GI symptoms can precede rash in up to 43% of cases
- Renal, 20-60%
  - Hematuria, hypertension, proteinuria, acute renal failure
  - HSP nephritis may develop up to 6 months after acute presentation
- Urogenital, up to 27%
  - Scrotal swelling, priapism, ureteral stenosis
  - 3% of all acute scrotal cases
- Neurological, 2%
  - Seizure, intracranial hemorrhage, headache, focal deficit, polyradiculopathy
- Pulmonary, 0.8-5.0%
  - Hemoptysis, dyspnea, chest pain
- Conditions associated with or preceding HSP include
  - Upper respiratory infection
  - Vaccinations with measles, yellow fever, typhoid
  - Environmental exposures, such as drugs, cold exposure, insect bites

Demographics

- Most common primary pediatric vasculitis, 49%
- Majority occur between 3-15 years of age
  - Peak age: 7 years
- Annual incidence 20 per 100,000
- Seasonal variation, most cases occur in winter

Natural History & Prognosis

- Typically self-limited, resolves in 3-4 weeks
- Recurrence in up to 1/3 of cases
- 20% of HSP nephritis cases develop nephritic or nephrotic syndrome
  - Long-term renal impairment in 20-44% of patients that develop nephritic or nephrotic syndrome
  - End-stage renal disease < 1%
- DAH often severe: 50% require mechanical ventilation, 28% mortality

Treatment

- Conservative management of symptoms
  - Steroids for severe skin lesions
  - Oral prednisolone for GI disease controversial
- Reduction of ileocolic intussusceptions
- Hemodynamic monitoring with possible endoscopy for GI bleeding
- HSP nephritis
  - No evidence-based treatment
  - Current therapeutic approach includes
    - Angiotensin-converting enzyme inhibitors
    - IV or oral corticosteroids
    - Cyclophosphamide if severe
    - Early-onset plasmapheresis

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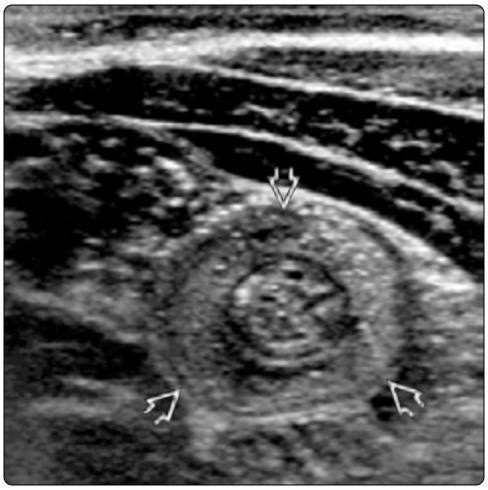
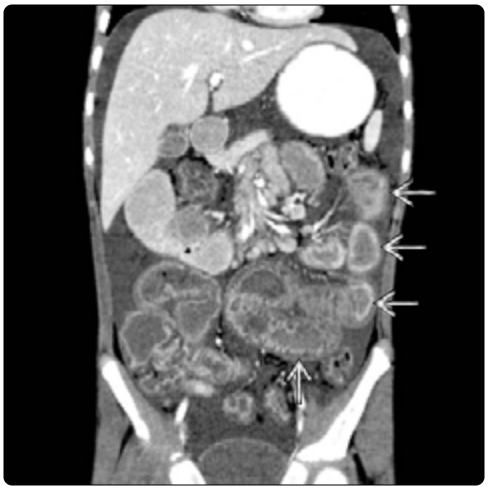
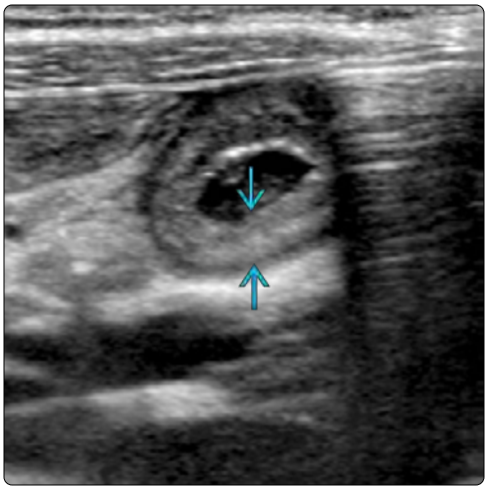
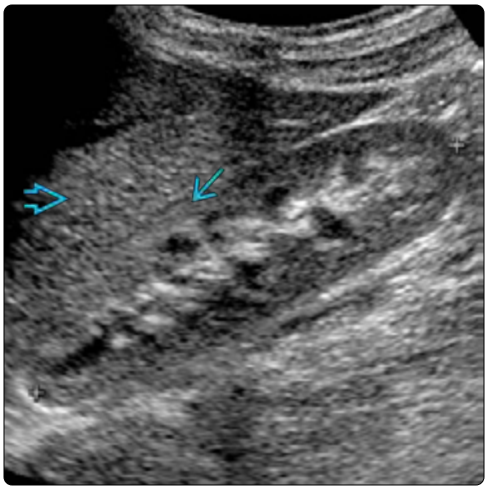


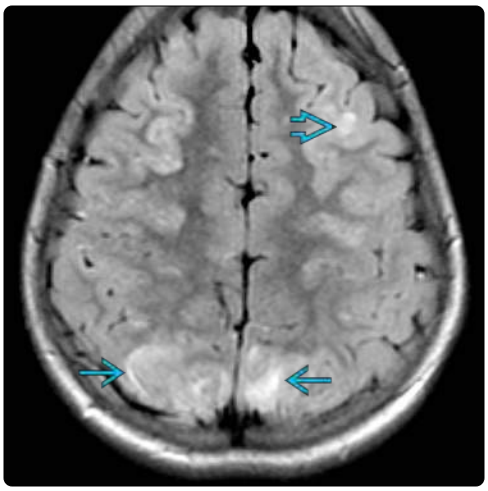


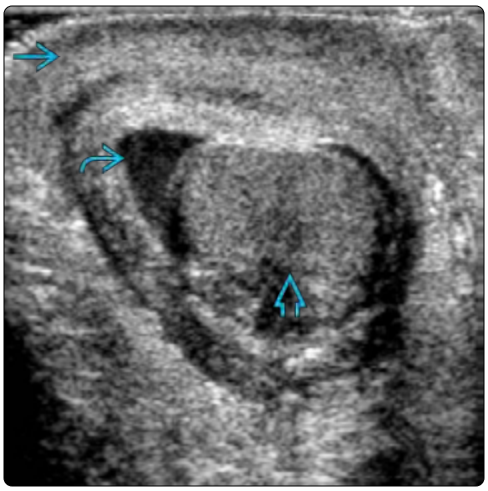
(Left) *Transverse US in a 5 year old with HSP & abdominal pain demonstrates a small bowel intussusception in cross section with a target appearance ➡. Multiple small bowel intussusceptions were encountered during the exam (not shown).* **(Right)** *Coronal CECT in an 8 year old with rash, proteinuria, & abdominal pain shows multiple areas of bowel wall thickening & mucosal hyperenhancement ➡, bowel dilation, & diffuse ascites. A skin biopsy showed IgA vasculitis consistent with HSP.*





(Left) *Transverse US in an 11 year old with known HSP demonstrates small bowel wall thickening ⇨, which can be seen with intramural edema &/or hemorrhage.* **(Right)** *Longitudinal US in the same patient demonstrates increased echogenicity of the renal cortex ⇨ that is equivalent to or slightly greater than the adjacent liver ⇨. The patient presented with worsening hypertension.*





(Left) *Axial FLAIR MR of the brain after the same patient with HSP & hypertension developed seizures shows superficial foci of increased signal intensity in the parietal ⇨ > frontal ⇨ lobes. Occipital lobe involvement was also present (not shown). The findings are consistent with posterior reversible encephalopathy syndrome.* **(Right)** *Transverse US demonstrates marked scrotal wall thickening ⇨, a small hydrocele ⇨, & a normal testicle ⇨ in a 21 month old with HSP.*

KEY FACTS

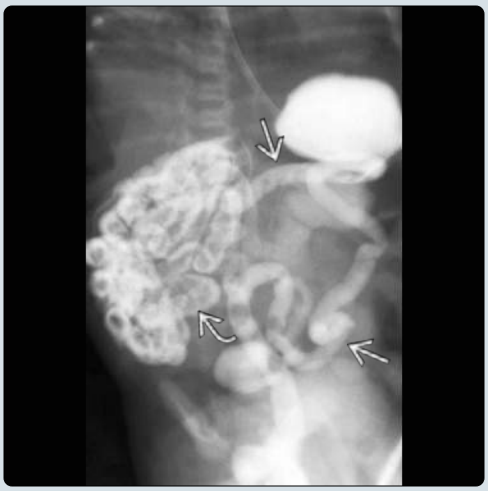
TERMINOLOGY

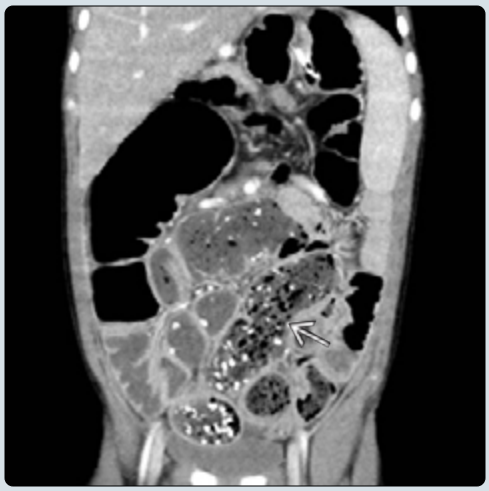
- Cystic fibrosis (CF): Autosomal recessive multisystem disorder caused by dysfunctional chloride ion transport across epithelial surfaces

IMAGING

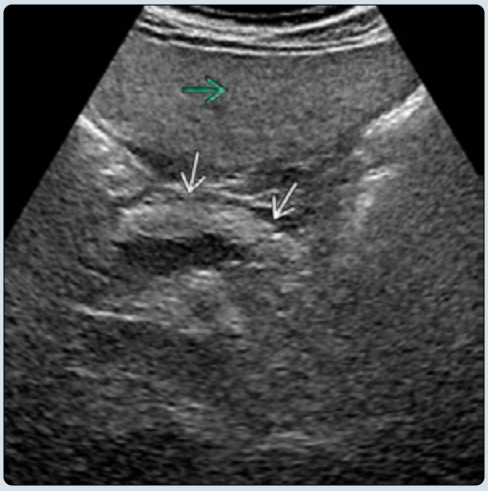
- Pancreas: Abdominal organ most commonly involved in CF
  - Pancreatic insufficiency: Present in > 70% of patients with CF at initial diagnosis
  - Pancreatitis: Can be acute or chronic
    - Acute: More common in patients with pancreatic sufficiency
    - Chronic: May appear as lipomatous hypertrophy, Ca^{2+}, &/or gland atrophy
  - Pancreatic cystosis: Rare sequela caused by occlusion of small ductules
- Intestinal
  - Gastroesophageal reflux disease: Prevalence 6-8x higher than general population
  - Meconium ileus: Congenital bowel obstruction caused by tenacious meconium occluding terminal ileum
  - Distal intestinal obstruction syndrome (DIOS): Meconium ileus equivalent in older children
  - Constipation: Excess stool throughout colon
  - Intussusception: 10x higher than general population
  - Appendix: Enlarged without inflammation
    - Appendicitis rates lower in CF patients
  - Fibrosing colonopathy: Fibrotic colonic disease in older patients associated with high doses of replacement pancreatic enzymes
- Hepatobiliary: Most common cause of CF mortality after pulmonary complications
  - Hepatic fibrosis/cirrhosis: Occurs in 5-15% of children/adolescents with CF
  - Focal biliary cirrhosis: Most common cause of liver pathology in patients with CF
  - Microgallbladder: Caused by atresia/stenosis of cystic duct

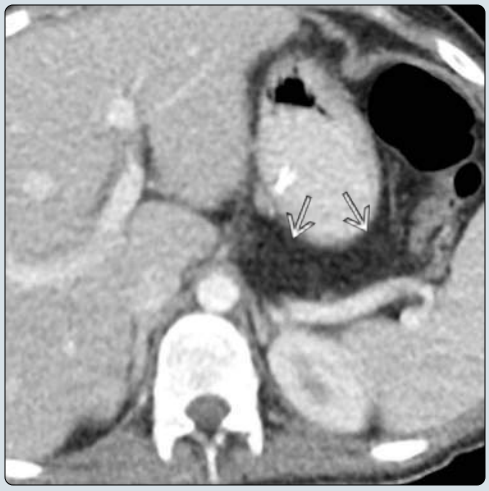
(Left) *Neonatal contrast enema shows a microcolon, typical of a congenital distal bowel obstruction. Contrast refluxes into the terminal ileum, outlining filling defects of tenacious meconium. Meconium ileus is almost always due to cystic fibrosis (CF).* **(Right)** *Coronal CECT in the same patient 9 years later shows stool in the ileum. The proximal small bowel was dilated & fluid filled (not shown). Distal intestinal obstruction syndrome (DIOS) occurs more commonly in CF patients that have a history of meconium ileus in infancy.*





(Left) *Ultrasound of the abdomen shows the pancreas to be hyperechoic & atrophied, typical of fatty replacement in this CF patient. There is also loss of the normal echogenic portal triads in the liver, suggesting hepatic steatosis.* **(Right)** *Axial CECT in a child with CF shows fatty replacement of the pancreas. CF is the most common cause of pancreatic lipomatosis. Patients with CF can also get acute or chronic pancreatitis, though acute pancreatitis is more common in patients with normal pancreatic exocrine function.*





TERMINOLOGY

Definitions

- Cystic fibrosis (CF): Autosomal recessive multisystem disorder caused by dysfunctional chloride ion transport across epithelial surfaces
  - Presenting symptoms of CF in infants & young children most commonly GI related

IMAGING

General Features

- Associated conditions in CF patients
  - Pancreas: Abdominal organ most commonly involved
    - Pancreatic insufficiency: Present in > 70% of patients at initial detection
    - Pancreatitis: Can be acute or chronic
      - Acute: Occurs in 10% of patients with pancreatic sufficiency
      - Chronic: May appear as lipomatous hypertrophy, Ca^{2+}, &/or gland atrophy
    - Pancreatic cystosis: Rare sequela caused by occlusion of small ductules
  - Intestinal
    - Gastroesophageal reflux disease (GERD): Prevalence 6-8x higher than in general population
    - Meconium ileus: Congenital bowel obstruction (not ileus) caused by tenacious meconium occluding terminal ileum
    - Distal intestinal obstruction syndrome (DIOS): Meconium ileus equivalent occurring in all ages
    - Constipation: Excess stool throughout colon, may be particularly firm
    - Appendicitis: Occurs less commonly than general population
      - However, uninflamed appendix typically enlarged in CF patients: Mean diameter of 8.3 mm
    - Intussusception: 10x more common than general population
    - Fibrosing colonopathy: Fibrotic colonic disease associated with high doses of replacement pancreatic enzymes
    - Malignancy: Intestinal malignancies occur 23x more commonly in CF patients (typically adults)
  - Hepatobiliary: Most common cause of mortality after pulmonary complications
    - Steatosis: Occurs in up to 2/3 of children & adolescents
    - Hepatic fibrosis/cirrhosis: Occurs in 5-15% of children/adolescents
    - Focal biliary cirrhosis: Most common cause of liver pathology
    - Microgallbladder: Caused by atresia/stenosis of cystic duct

Radiographic Findings

- Radiography
  - Chronic pancreatitis: Occasionally see scattered Ca^{2+}
  - Meconium ileus: Distal obstruction bowel gas pattern occurring in newborn
  - DIOS: Dilated small bowel loops with stool mass in right lower quadrant of older child
  - Constipation: Large amount of stool throughout colon

Fluoroscopic Findings

- Upper GI
  - GERD: Retrograde propulsion of contrast from stomach to esophagus
  - Duodenal fold thickening
- Contrast enema
  - Meconium ileus
    - Microcolon with filling defects of abnormal meconium in terminal ileum
    - Complicated by ischemia, atresia, volvulus, or perforation in up to 50% of cases
    - Enema can be therapeutic in uncomplicated cases
  - DIOS
    - Normal colon with filling defects in terminal ileum
    - Enema can be therapeutic
  - Fibrosing colonopathy: Colonic stricture, loss of haustra, & colonic shortening
- Air enema
  - Used to treat ileocolic intussusception
    - Intussusception reduced when air refluxes into small bowel

CT Findings

- Acute pancreatitis: Appears as pancreatitis in patients without CF
- Chronic pancreatitis: Varying amounts of fatty replacement, pancreatic atrophy, Ca^{2+}
- Pancreatic cystosis: Multiple cysts of varying sizes in pancreas
- DIOS: Mass of stool in terminal ileum
- Appendix: Larger than normal patients & often filled with inspissated secretions despite lack of inflammation
- Steatosis: Diffuse or patchy areas of low attenuation in liver
- Hepatic fibrosis/cirrhosis: Small liver with nodular contour; may have findings of portal hypertension
- Microgallbladder: Gallbladder much smaller than expected

MR Findings

- Pancreatitis: Symptomatic patients may have pancreatic ductal anomalies such as pancreas divisum on MRCP
- Steatosis: Loss of hepatic signal on opposed-phase images

Ultrasonographic Findings

- Pancreatic lipomatosis: Echogenic pancreas of varying size
- Pancreatic cystosis: Multiple cysts of varying size in pancreas
- Appendix: Larger than normal patients & often filled with inspissated secretions without inflammation
- Intussusception: Target sign or pseudokidney sign
- Steatosis: Hyperechoic liver (compared to right kidney) with poor visualization of portal triads & right hemidiaphragm

Imaging Recommendations

- Best imaging tool
  - Depends on patient symptoms
    - Neonate with vomiting or failure to pass meconium
      - Radiograph with distal obstruction pattern → contrast enema
    - Abdominal pain in older child
      - DIOS, constipation: Radiograph

- Pancreatitis: Ultrasound followed by MR/MRCP
- Intussusception: Radiograph followed by ultrasound followed by air-reduction enema

– Liver disease
- US or MR can assess hepatic fat content, fibrosis, & findings of portal hypertension

DIFFERENTIAL DIAGNOSIS

Neonate With Failure to Pass Meconium

- Hirschsprung disease
  - Functional obstruction of bowel due to lack of distal enteric (typically distal-most colonic) ganglion cells
  - Abnormal rectosigmoid ratio on contrast enema
  - Microcolon rare
- Meconium plug/small left colon
  - Transient functional immaturity of distal colon causes obstruction
  - Transition at splenic flexure
- Ileal atresia
  - Microcolon with "cut-off" to retrograde contrast flow at site of atresia in ileum
    - Contrast does not reflux into dilated loops
  - Minority of cases associated with other bowel atresias
- Anorectal malformation
  - Multiple variations, none with clinically normal anus

PATHOLOGY

General Features

- Genetics
  - Autosomal recessive
  - CF transmembrane conductance regulator (*CFTR*) gene mutation (chromosome 7q31.2)
    - > 1,500 genetic defects can result in CF; ΔF508 most common
      - Genotypes with less severe lung disease may have more severe extrapulmonary disease
- Etiology
  - *CFTR* gene mutation → lack of chloride ion secretion → ↑ sodium retention & fluid absorption → ↑ viscosity of luminal secretions → obstruction of ducts of solid organs & hollow viscera
  - CFTR protein located in epithelial cells of airway, GI tract, sweat glands, & GU system
  - Most GI manifestations due to combination of disordered motility, thick intestinal secretions, pancreatic insufficiency, & medications used to control pulmonary or pancreatic symptoms
- Associated abnormalities
  - Pulmonary disease

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Meconium ileus: Failure to pass meconium, emesis in newborn
  - Intussusception: Colicky abdominal pain, intermittent fussiness
  - Pancreatic insufficiency: Greasy stools, flatulence, bloating, failure to thrive
  - GERD: Heartburn, dysphagia, dyspepsia
  - DIOS: Crampy abdominal pain, distended abdomen, palpable right lower quadrant mass, anorexia, weight loss, flatulence
  - Constipation: Frequent or chronic abdominal pain, palpable mass of stool
  - Fibrosing colonopathy: Abdominal pain, distension, vomiting, & constipation in older patients

Demographics

- Age
  - Most patients diagnosed with CF in 1st year of life during neonatal screening
  - Meconium ileus occurs in neonates
  - Other findings can be found throughout life; patients may not manifest until later in childhood
- Epidemiology
  - CF most common lethal genetic defect in Caucasians
    - 1 in 2,500 Caucasian live births
      - 1 in 20 Caucasians = CF carriers
    - 1 in 17,000 African American live births

Natural History & Prognosis

- Most common cause of death: Progressive lung disease
- Median survival: 41.1 years
  - Survival increased > 10 years since 2002

Treatment

- CFTR modulators: New class of therapy designed to help CFTR protein function
  - Improves pulmonary function
  - Uncertain effect on extrapulmonary manifestations
- Meconium ileus
  - Uncomplicated: Dilute gastrografin enema + patient hydration
  - Complicated: Surgery
- DIOS: Dilute gastrografin enema + patient hydration
- Intussusception: Air-contrast enema
- Pancreatic insufficiency: Pancreatic enzyme replacement

DIAGNOSTIC CHECKLIST

Consider

- CF in neonate with distal bowel obstruction
- Abdominal manifestations of CF cause significant morbidity & can contribute to pulmonary complications

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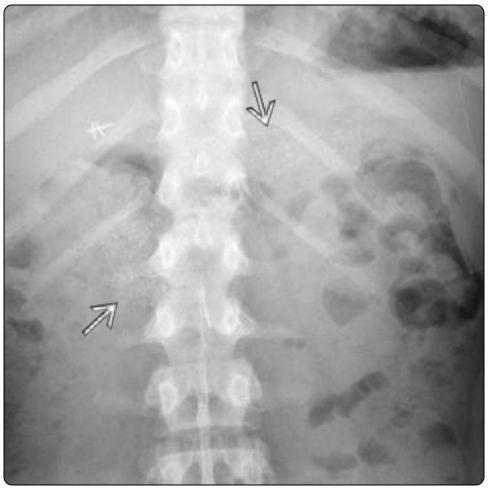
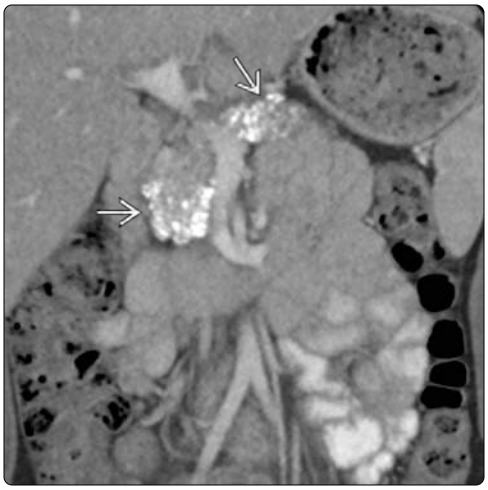
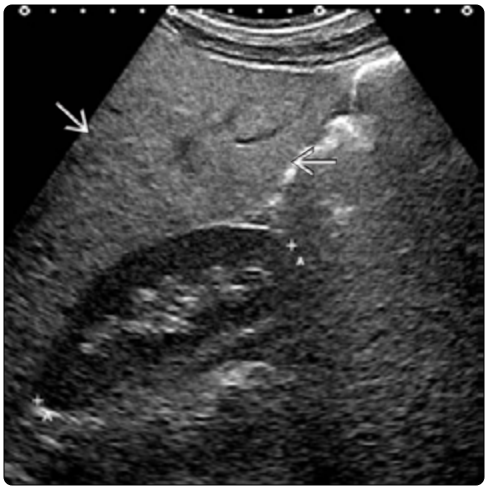
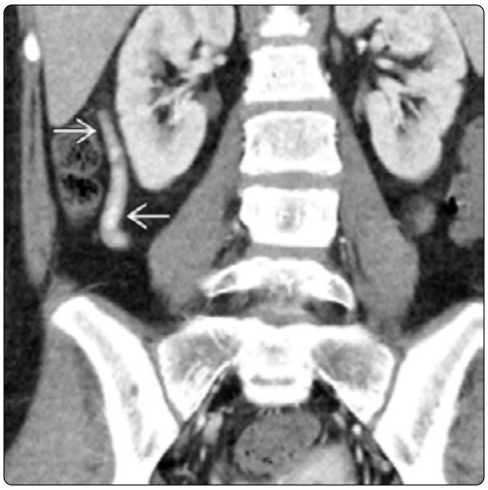


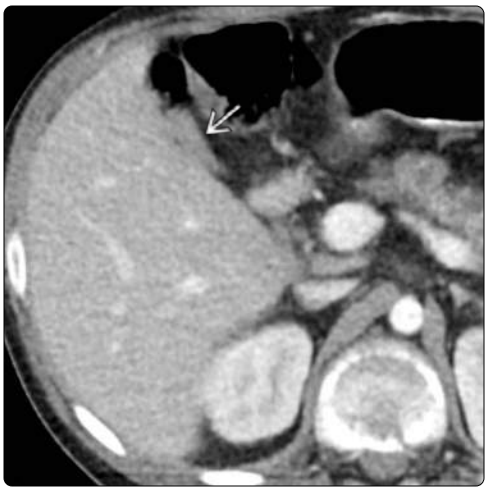


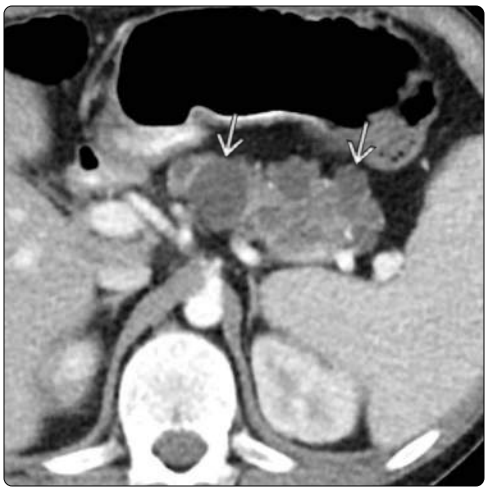
(Left) *Abdominal radiograph shows speckled Ca^{2+} throughout the course of the pancreas. Findings of chronic pancreatitis include Ca^{2+} & gland atrophy.* **(Right)** *Coronal CECT in the same patient shows the diffuse speckled pancreatic Ca^{2+}. Pancreatic Ca^{2+} are more commonly scattered & fewer in number than in this case. Complete fatty replacement is the most common imaging finding of the pancreas in patients with CF.*





(Left) *Longitudinal ultrasound in an adolescent with CF shows diffusely increased echogenicity of the liver when compared to the right kidney. Additionally, there is poor visualization of the echogenic portal triads of the liver, consistent with steatosis.* **(Right)** *Coronal CECT in an adolescent with CF shows an enlarged (9 mm), mucous-filled appendix. Patients with CF often have an enlarged but uninflamed appendix. The incidence of appendicitis is 1% in CF patients as compared to 7% in the general population.*





(Left) *Axial CECT in a child with CF shows a microgallbladder, a classic finding that is caused by stenosis or obstruction of the cystic duct.* **(Right)** *Axial CECT shows multiple cysts of varying sizes in the pancreatic tail. Pancreatic cystosis is a rare entity caused by obstruction of multiple pancreatic ductules.*

Burkitt Lymphoma

KEY FACTS

TERMINOLOGY

- Aggressive B-cell non-Hodgkin lymphoma (NHL)
- Most common form of NHL in children ≤ 15 years of age

IMAGING

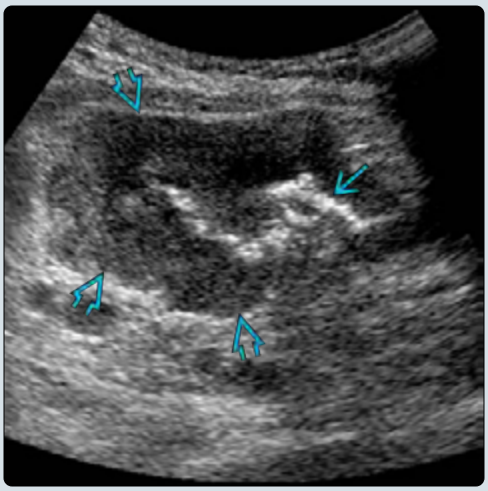
- Most common imaging appearance depends on type
  - Sporadic: Abdominal soft tissue mass &/or bowel wall thickening; may involve solid organs
    - Discrete or conglomerate masses &/or infiltrating, encasing, poorly defined lesions
  - Endemic: Head & neck soft tissue mass
- Ultrasound
  - Homogeneous, hypoechoic soft tissue mass(es)
  - Irregular bowel wall thickening; ± intussusception
- CT/MR
  - Moderately enhancing homogeneous mass(es) &/or thickened bowel wall (ileocecal region)
  - Enlarged lymph nodes
  - ± peritoneal thickening or nodularity

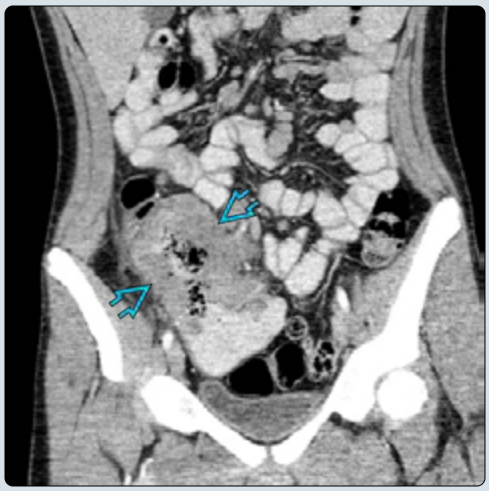
PATHOLOGY

- 3 distinct subtypes
  - Endemic
    - Commonly involves head & neck (mandible)
    - Almost always associated with Epstein-Barr virus (EBV)
    - More common in Africa & other sites along equator
  - Sporadic
    - Most commonly involves abdomen (60-80%)
      - Bowel, mesentery, solid organs, gonads
      - May involve head & neck, chest, superficial sites
    - Less commonly associated with EBV (15%)
    - More common in North America, Europe
  - Immunodeficiency related

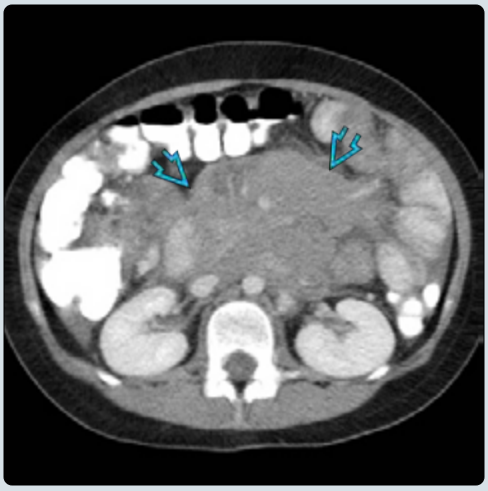
CLINICAL ISSUES

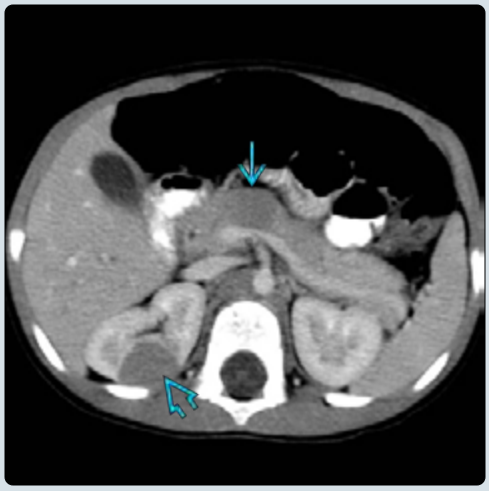
- Mean age: 8 years
- Short doubling time of neoplasm (~ 24 hours) may lead to rapid growth, acute presentations
- With appropriate treatment, survival ~ 90%





(Left) *Transverse abdominal ultrasound in a 12-year-old boy with Burkitt lymphoma shows marked hypoechoic & somewhat irregular bowel wall thickening in the right lower quadrant. The central echogenic foci represent gas within the compressed bowel lumen.* **(Right)** *Coronal CECT in the same 12-year-old boy with Burkitt lymphoma shows marked nodular bowel wall thickening.*





(Left) *Axial CECT in a 9-year-old girl found to have Burkitt lymphoma shows a large, conglomerate lymph node mass engulfing bowel loops & multiple vessels.* **(Right)** *Axial CECT in a 5-year-old boy with Burkitt lymphoma shows a homogeneous, low-attenuation mass in the right kidney. A similar focus of tumor is visualized in the pancreatic neck. Additional lesions were also present in the left kidney (not shown).*

TERMINOLOGY

Synonyms

- Small, noncleaved lymphoma

Definitions

- Aggressive B-cell non-Hodgkin lymphoma (NHL)
- Most common form of NHL in children ≤ 15 years of age
  - 40% of childhood NHL
  - 3-4% of all childhood malignancies

IMAGING

General Features

- Best diagnostic clue
  - Homogeneous abdominal soft tissue mass or focal, marked bowel wall thickening involving ileocecal region
- Location
  - Sporadic
    - Abdominal involvement most common (60-80%)
      - Bowel wall, most commonly ileocecal region (including appendix)
      - Mesentery & peritoneal lining
      - Can involve solid organs including kidneys & gonads
    - Head & neck
    - Others: Mediastinum, bone marrow, CNS
  - Endemic
    - Head & neck most common
      - Classically involves mandible

CT Findings

- CECT
  - Homogeneous, solid soft tissue mass
  - Moderate to marked irregular bowel wall thickening involving ileocecal region
    - May cause intussusception
  - Conglomerate mesenteric lymph node masses
  - Nodular or mass-like peritoneal thickening

MR Findings

- T2WI FS
  - Intermediate-signal soft tissue mass
  - Bowel wall thickening
  - Enlarged lymph nodes
- DWI
  - Positive diffusion restriction
- T1WI C+ FS
  - Moderate enhancement of mass or thickened bowel wall, enlarged lymph nodes

Ultrasonographic Findings

- Grayscale ultrasound
  - Irregular bowel wall thickening
    - Thickened bowel wall usually hypoechoic
    - May cause ileocolic intussusception
  - Solid, hypoechoic mass or enlarged lymph nodes
- Color Doppler
  - Internal blood flow suggesting solid tumor &/or vessel encasement
  - Thickened bowel wall causes less hyperemia than infectious or inflammatory causes

Nuclear Medicine Findings

- ↑ uptake on FDG PET

DIFFERENTIAL DIAGNOSIS

Other Potentially Extensive Abdominal Neoplasms

- Hodgkin lymphoma
- Rhabdomyosarcoma
- Ovarian neoplasm
- Desmoplastic small round cell tumor

Other Causes of Bowel Wall Thickening

- Inflammatory bowel disease
- Infectious enterocolitis

PATHOLOGY

General Features

- Translocation t(8:14)(q24:q32) in 70-80% of patients
  - Linked to deregulated expression of *MYC* oncogene

Staging, Grading, & Classification

- Classified into 3 distinct subtypes
  - Endemic
    - Africa, Papua New Guinea, & other locations clustered around equator
      - Linked to malaria
    - Almost always associated with Epstein-Barr virus (EBV)
  - Sporadic
    - North America, Europe
    - Less commonly associated with EBV (15%)
  - Immunodeficiency related
    - Associated with HIV infection

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Sporadic: Abdominal pain, distention, obstruction
  - Endemic: Head & neck mass, usually mandible
- Other signs/symptoms
  - Recurrent ileocolic intussusception
- Short doubling time of neoplasm (~ 24 hours) may lead to rapid growth, acute presentations

Demographics

- Mean age: 8 years; most common between 5-9 years
- Boys > > girls

Natural History & Prognosis

- With appropriate treatment, survival ~ 90%

Treatment

- Chemotherapy ± surgery, radiation therapy

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Castleman Disease

KEY FACTS

TERMINOLOGY

- Benign lymphoproliferative disorder
  - Also termed angiofollicular lymph node hyperplasia
- Can be unicentric (most common) or multicentric/systemic
- Most common location overall: Mediastinum
  - Can occur in abdomen & other nodal sites

IMAGING

- Radiographs: Soft tissue mass ± punctate Ca^{2+}
- Ultrasound: Hypoechoic, sometimes hypervascular mass
- NECT/CECT
  - Well-circumscribed, avidly enhancing mass
    - ± Ca^{2+} in branching (arborizing) pattern
- MR
  - Low to intermediate T1; intermediate to high T2, DWI
    - ± central ↓ T2 signal due to Ca^{2+} or fibrosis
  - Avid arterial phase enhancement characteristic
- PET: FDG avidity typically less than lymphoma

TOP DIFFERENTIAL DIAGNOSES

- Lymphoma
- Granulomatous infection
- Neurogenic tumor
- Inflammatory myofibroblastic tumor

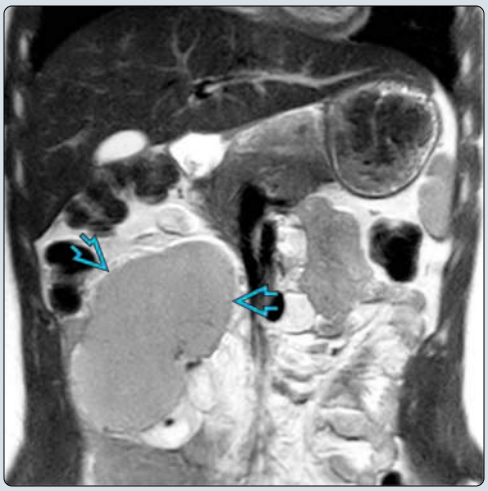
PATHOLOGY

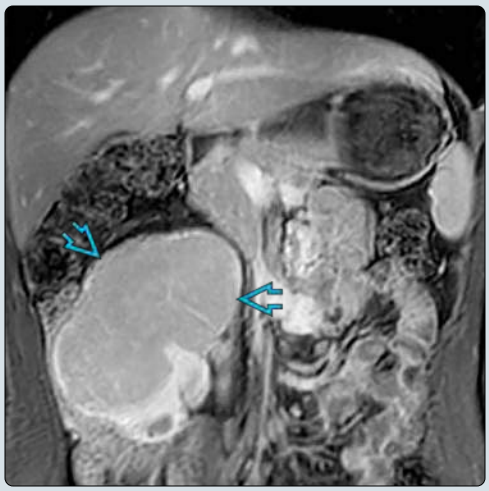
- Hyaline-vascular type
  - Most common, usually unicentric asymptomatic mass
  - Typically in adolescents & young adults
- Plasma cell type
  - Rare in children, can be multicentric
  - More commonly with systemic symptoms (fever, anorexia, weight loss) & laboratory abnormalities
- Mixed type: Features of both

CLINICAL ISSUES

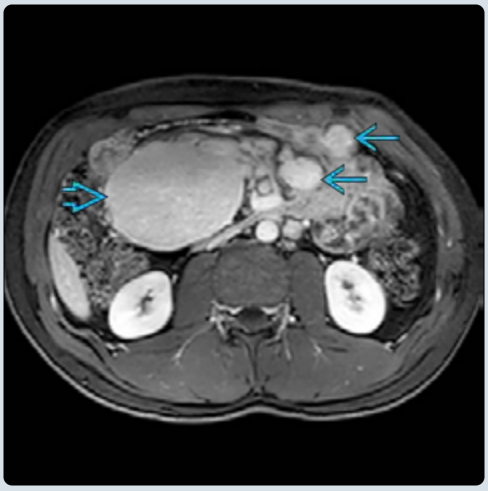
- Unicentric form: Excellent prognosis with surgical excision
- Multicentric form: Systemic treatment; worse prognosis

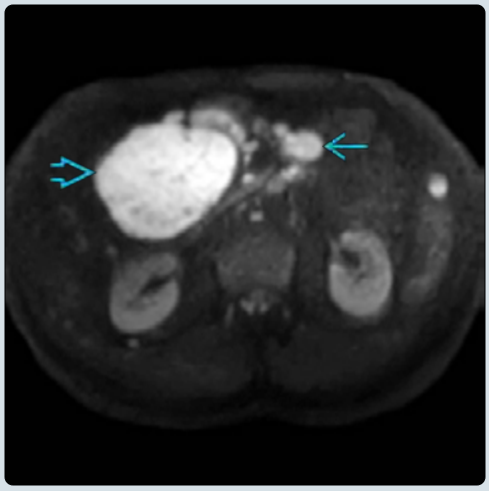
(Left) *Coronal T2 SSFSE MR in a 16 year old with abdominal pain shows a moderate-sized abdominal mass of homogeneous intermediate signal intensity centered in the mesentery.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows moderate homogeneous enhancement of the right mesenteric mass.*





(Left) *Axial T1 C+ FS MR in the same patient again shows moderate homogeneous enhancement of the right mesenteric mass as well as several enlarged mesenteric lymph nodes.* **(Right)** *Axial DWI MR in the same patient shows marked hyperintense signal throughout the mass & adjacent lymph nodes. The corresponding ADC map (not shown) confirmed restricted diffusion. Biopsy revealed Castleman disease.*





KEY FACTS

TERMINOLOGY

- Spindle cell neoplasm with low malignant potential in children & young adults
- Many antiquated terms, including inflammatory pseudotumor & plasma cell granuloma

IMAGING

- Locations: Lung, abdominal solid & hollow organs, mesentery
  - Most common primary lung neoplasm in children
- Typically well-defined, round or lobulated mass
  - May be discrete & bulky or infiltrative and ill-defined in mesentery & bowel
- Heterogeneously enhancing on CT/MR; ± delayed enhancement, central necrosis, or central Ca^{2+}
- May be T2 hypointense on MR with ↑ fibrous stroma

TOP DIFFERENTIAL DIAGNOSES

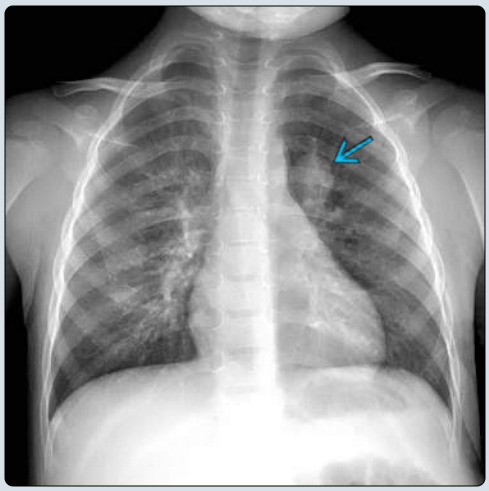
- Other fibrous lesions
- Soft tissue sarcomas
- Lymphoma
- Soft tissue abscess
- Castleman disease
- Desmoplastic small round cell tumor

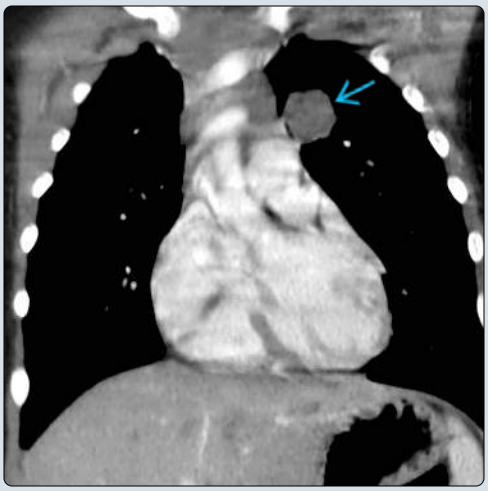
PATHOLOGY

- Spindle cell proliferation + inflammatory cells
- *ALK* oncogene mutation in ~ 60%

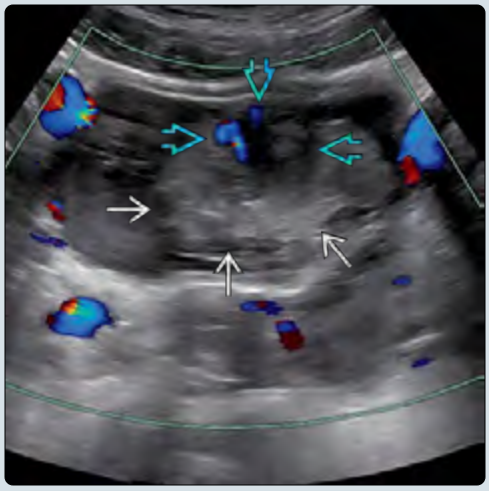
CLINICAL ISSUES

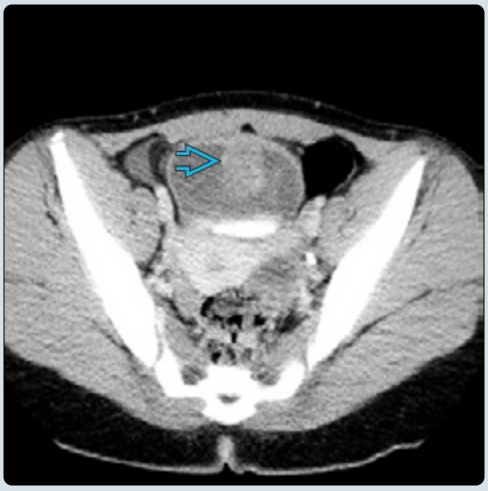
- Symptoms due to local mass effect ± nonspecific constitutional symptoms
- Low risk of distant metastases (~ 5%) with local/regional recurrence more frequent
  - Most morbidity/mortality from local invasion/mass effect
- Excellent prognosis with complete surgical resection
  - If not feasible → chemotherapy ± steroids & radiation





(Left) *Frontal chest radiograph in a 3-year-old boy with a cough who was ultimately found to have an inflammatory myofibroblastic tumor (IMT) is shown. A well-defined perihilar left upper lobe mass is present ⇨.* **(Right)** *Coronal CECT in the same 3-year-old boy with an IMT demonstrates a well-defined, nearly homogeneous, relatively hypoenhancing mass situated in the left upper lobe ⇨. IMT should be remembered as the most common pediatric lung neoplasm.*





(Left) *Color Doppler US of the urinary bladder in a 13-year-old girl with hematuria shows echogenic avascular debris ➡ in the bladder, consistent with clotted blood. However, there is also a solid, hypoechoic mass anteriorly ⇨ with central internal blood flow.* **(Right)** *Axial CECT through the pelvis in the same 13-year-old girl shows a mildly enhancing intraluminal soft tissue mass anteriorly within the bladder ⇨. An IMT was confirmed upon resection.*

KEY FACTS

TERMINOLOGY

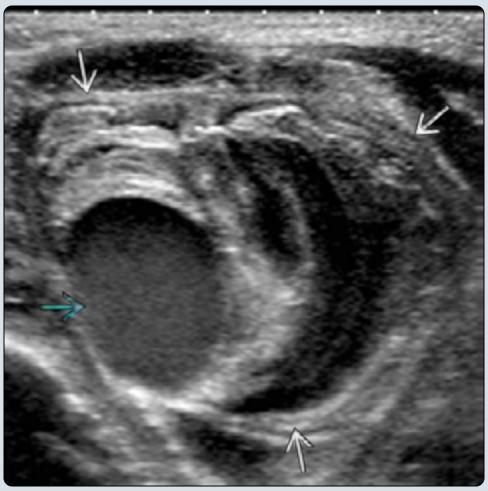
- Dilation of abdominal aorta &/or major arterial branches
- Beyond idiopathic, etiologies include
  - Syndromic: Tuberous sclerosis, neurofibromatosis type 1, connective tissue disorders
  - Inflammatory arteritides: Takayasu, Kawasaki, Behçet
  - Infectious: Bacterial, mycobacterial, fungal
  - Traumatic: Iatrogenic (umbilical arterial catheter), blunt trauma

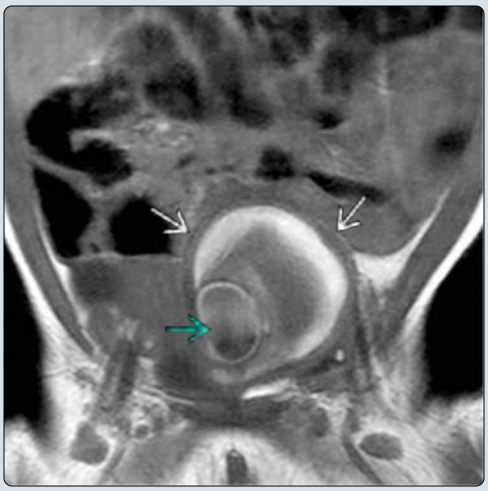
IMAGING

- Any portion of aorta & branches may be affected
  - ± dilation &/or stenosis of visceral branches
  - Normal aortic diameter varies with patient age & size
- Ultrasound excellent tool for initial investigation of pediatric abdominal mass (± pulsation)
  - Layers of clot may mimic heterogeneous neoplasm
  - Color Doppler essential to exclude aneurysm upon visualization of any "cystic" lesion
- Best imaging tool to define extent & branch involvement: CTA or MR angiography
  - Fusiform or saccular dilation of aorta &/or branches
  - Vessel wall enhancement in inflammatory etiologies
- Consider multistation/whole-body MRA with blood pool contrast agent to look for distant aneurysms in settings of systemic disease or idiopathic aneurysm

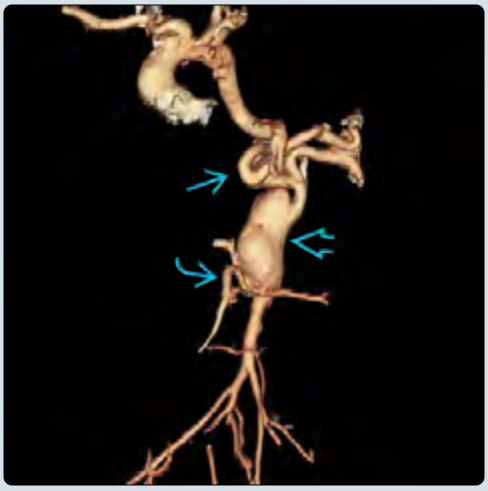
CLINICAL ISSUES

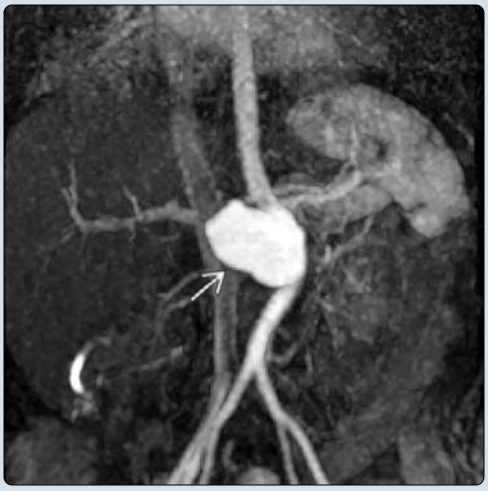
- Presentations include
  - Asymptomatic ± pulsatile abdominal mass
  - Stenosis &/or thromboembolism of branch vessels
    - End organ hypoperfusion; resultant renin-mediated hypertension if renal arteries involved
  - Rupture (rarely) with abdominal pain & shock
  - Signs/symptoms of underlying systemic disease
- Treatment: Surgical repair vs. endovascular stent grafting
  - Medical therapy to ↓ inflammation, control blood pressure; IV antibiotics for infection





(Left) *Transverse ultrasound of the lower abdomen in a 4-month-old infant shows a large heterogeneous mass ➔ along the expected course of the left common iliac artery. The ovoid hypoechoic center ⇨ completely filled in with color flow on Doppler imaging (not shown).* **(Right)** *Coronal T1 MR in the same patient shows layers of alternating signal intensity in the mural thrombus ➔ around the aneurysm lumen ⇨ due to different ages of clotted blood. Multiple aneurysms were found in this patient with polyarteritis nodosa.*





(Left) *CTA 3D reconstruction in a 3 month old with congenital anomalies & hypertension shows an aneurysm of the abdominal aorta ⇨ with involvement of the celiac trunk & superior mesenteric artery ⇨. The patient also had occlusion of the lower thoracic aorta with resultant formation of multiple collateral vessels ⇨.* **(Right)** *Coronal MIP MR angiogram in a 19 day old with severe hypertension shows a saccular aneurysm of the infrarenal abdominal aorta ➔ causing narrowing of the renal arteries.*

TERMINOLOGY

Definitions

- Dilation of abdominal aorta &/or its major branches
  - True aneurysm: All layers of arterial wall expanded
  - Pseudoaneurysm: Adventitia contains focal rupture

IMAGING

General Features

- Best diagnostic clue
  - Focally dilated aorta &/or visceral branches
    - Normal aortic dimensions vary with patient age & size
      - > 2.5 cm diameter enlarged for any age
- Location
  - Can affect any portion of aorta & branches
    - Infrarenal aneurysms most common
  - Aortic aneurysm associated with dilation &/or stenosis of adjacent visceral branches
  - Distant aneurysms often found with systemic diseases & idiopathic aneurysms
- Morphology
  - Aneurysm can be fusiform or saccular

CT Findings

- CECT
  - ↑ luminal diameter of aorta
    - Look for associated aneurysms in visceral branches
    - Also associated with vascular stenoses
  - May see vessel wall thickening or enhancement if due to inflammatory causes (e.g., Takayasu arteritis)

MR Findings

- T1WI C+ FS
  - ± vessel wall enhancement with inflammatory causes
- MRA
  - Dilated aorta or visceral branches ± associated stenoses

Ultrasonographic Findings

- Grayscale ultrasound
  - Dilation of aorta/artery: Well-defined, pulsatile, anechoic, round or tubular mass (cystic appearing)
  - Heterogeneous mural thrombus due to layers of clot ± calcification
    - May compose majority of aneurysm
- Color Doppler
  - Confirms vascular nature of round or tubular anechoic structure (rather than cyst or fluid-filled tube)
  - ± ↑ velocities at stenosis; turbulent flow with parvus et tardus waveforms, spectral broadening beyond stenosis

Imaging Recommendations

- Best imaging tool
  - CT angiography or MR angiography
  - Catheter-based digital subtraction angiography (DSA) may be employed in therapy
- Protocol advice
  - Multistation/whole-body MR angiography with albumin-binding blood pool contrast agent
    - Prolonged arterial enhancement allows for multistation screening for other aneurysm sites (in setting of systemic disease or idiopathic aneurysm)

DIFFERENTIAL DIAGNOSIS

Abdominal Cyst

- Well-defined cysts such as ovarian cyst, gastrointestinal duplication cyst, mesenteric lymphatic malformation
- Color Doppler will show no significant internal flow

Solid Abdominal Mass

- Large, heterogeneous, partially thrombosed aneurysm may mimic neoplasm such as Wilms or ovarian tumor

PATHOLOGY

General Features

- Etiology
  - Syndromes: Connective tissue disorders (Ehlers-Danlos, Marfan), tuberous sclerosis, neurofibromatosis type 1
  - Traumatic: Iatrogenic (umbilical artery catheter), blunt abdominal trauma (uncommon)
  - Inflammatory arteritides: Takayasu (most common), Kawasaki, polyarteritis nodosa, Behçet disease
  - Infectious: Bacterial, mycobacterial, fungal
  - Idiopathic
- Associated abnormalities
  - Various features of underlying syndromes
  - Multifocal abdominal &/or distant aneurysms not uncommon, even in idiopathic setting

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic vs. pulsatile abdominal mass
  - High blood pressure due to renal artery stenosis/compression → renin-mediated (renovascular) hypertension
- Other signs/symptoms
  - Abdominal pain & shock due to rupture (rare)
  - Signs/symptoms of associated syndromes or septic emboli in infectious setting

Natural History & Prognosis

- Rupture can lead to life-threatening hemorrhage
- Visceral organ hypoperfusion due to associated arterial stenoses or thromboses

Treatment

- Surgery vs. endovascular stent grafting
  - May require operative reimplantation of branches
- Medical therapy to ↓ inflammation, treat infection, control blood pressure

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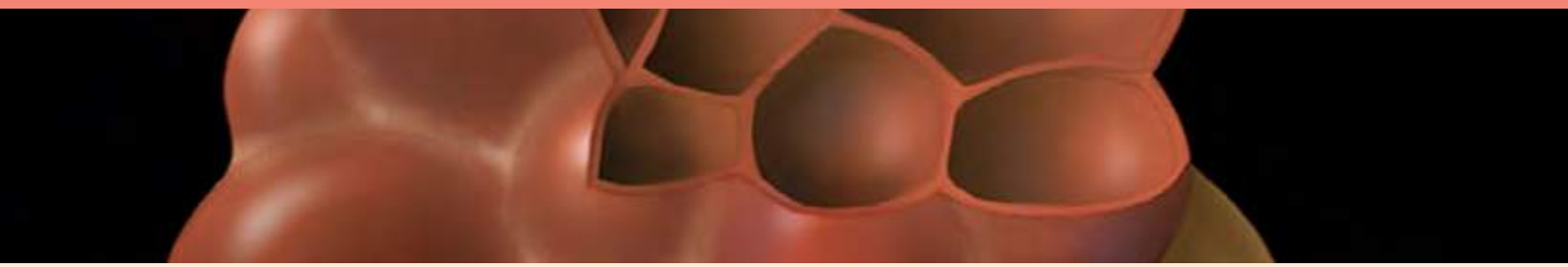
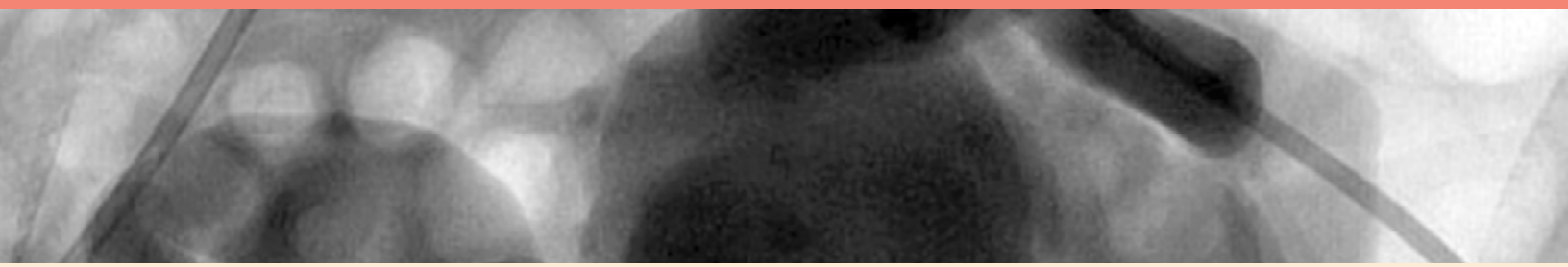
SECTION 5

Genitourinary





Miscellaneous Renal Conditions

Bladder Abnormalities

Adrenal Abnormalities

Uterine and Ovarian Abnormalities

Scrotal/Testicular Abnormalities

Pediatric Genitourinary Tract Disorders

Congenital Abnormalities

Congenital anomalies of the genitourinary tract are commonly discovered during the evaluation of pediatric patients. Many of these abnormalities are found incidentally, while others become symptomatic, causing infections, stone disease, pain, hematuria, renal failure, or, in severe bilateral cases, respiratory distress (due to pulmonary hypoplasia at birth).

The most commonly seen anomalies are ureteropelvic duplications, vesicoureteral reflux, ureteropelvic junction obstruction, & renal ectopia & fusion anomalies.

Renal Cystic Disease in Pediatrics

Pediatric renal cystic diseases range from the often lethal autosomal recessive polycystic kidney disease (ARPKD) to simple cortical cysts. A myriad of other diagnoses fall in between these 2 extremes with variable degrees of symptoms & clinical impact. Recently, most of the renal cystic diseases have been reclassified as ciliopathies, helping explain in many cases their multisystem organ involvement.

Multicystic dysplastic kidneys (MCDKs) are often diagnosed prenatally & confirmed with imaging in the newborn period. The lack of renal function in an MCDK on nuclear studies confirms the diagnosis.

ARPKD is a rare disorder in which the renal parenchyma is virtually replaced by microscopic cysts & occasional macrocysts. In severe cases, the kidneys are massively enlarged, function only minimally, & cause oligohydramnios in utero. Some complications of oligohydramnios, including lung hypoplasia, contribute to the mortality of this lethal condition. There is a variable degree of hepatic involvement in this disorder.

Autosomal dominant polycystic kidney disease (ADPKD) is a more common disorder characterized by macroscopic cysts that typically increase in both size & number over years. Patients are often aware of the family history of cystic renal disease in relatives with hypertension or renal insufficiency. Cysts can be seen in very young children, but ADPKD typically presents in the teenage years, either incidentally or with hematuria.

Other renal cysts found in the pediatric population include simple cortical cysts, calyceal diverticula, cystic renal dysplasia (secondary to chronic obstruction), syndromic cysts (such as tuberous sclerosis, von Hippel-Lindau, Meckel-Gruber, & others), & cystic neoplasms (such as multilocular cystic nephroma).

Renal "Masses"

Hydronephrosis is by far the most common cause of an enlarged kidney in pediatric patients & can present as a palpable mass on physical exam, prompting imaging.

Wilms tumor is the most common pediatric renal malignancy with a peak incidence at 3.6 years of age. These tumors are heterogeneous on imaging, but their origin from the kidney provides an important clue to their diagnosis. Renal vein thrombus & vascular invasion are common.

Nephroblastomatosis is a rare entity often associated with syndromes & chromosomal abnormalities. It is defined as the persistence of primitive nephrogenic rests in the kidney after birth. The rests fail to differentiate into functional renal tissue & do not enhance like normal parenchyma on imaging. Its importance lies in its frequent degeneration into Wilms tumor.

Mesoblastic nephroma is the most common renal tumor in neonates. It has a peak age of 3 months as opposed to the 3-year range for Wilms. Mesoblastic nephroma is classically solid, but it can be cystic or have mixed solid & cystic components.

Multilocular cystic nephroma is an unusual tumor with a bimodal age distribution, affecting young boys & adult women. Tumors are cystic with many septations, occasionally herniating into the central renal pelvis. An important association with the *DICER1* mutation (resulting in predisposition to various other tumors) has recently been described.

Angiomyolipoma is a benign tumor of mixed cellularity, typically with prominent fatty components & vascularity that can result in hemorrhage. The fatty component provides the key to the imaging diagnosis. In children, these lesions are most often seen in association with tuberous sclerosis.

Renal medullary carcinoma is a very rare & highly aggressive tumor associated with sickle cell trait.

Uncommon renal masses in pediatrics include renal lymphoma (typically multifocal), clear cell sarcoma, ossifying renal tumor of infancy, renal cell carcinoma, & rhabdoid tumor.

Uterine & Ovarian Abnormalities

These abnormalities often present either in the newborn period (due to the influence of maternal hormones) or in adolescence at menarche. Imaging should always include a search for associated renal anomalies, which are frequently coincident.

Adrenal Abnormalities

The most important adrenal abnormality in children is neuroblastoma. This tumor typically is seen in children < 2 years of age. Imaging commonly shows a calcified mass encasing the adjacent aorta & inferior vena cava; the mass may also extend into the spinal canal. Venous tumor thrombus is rare, unlike in Wilms tumors. Bony metastases are more common in neuroblastoma vs. lung metastases in Wilms.

Testicular Abnormalities

Torsion, infection, & inflammation are the most common reasons for testicular/scrotal imaging in pediatric patients. Congenital anomalies may predispose to inflammatory conditions or spermatic cord twisting that leads to ischemia. Tumors are less frequent & typically present without pain.

Other Renal Processes

Pyelonephritis is imaged very frequently in pediatrics with the goal of identifying predisposing anatomic variants & complications of infection that require intervention.

Renal vein thrombosis may be seen in newborn infants & other ill children, particularly in the setting of dehydration &/or systemic disorders.

Renal stones are increasingly diagnosed by ultrasound.

Bladder Abnormalities

Neurogenic bladder is the most common genitourinary entity imaged. Children typically show an evolution from a high-compliance, large-capacity bladder in new neurogenic bladders to a poorly distensible, low-capacity bladder in chronic neurogenic bladders. Bulking agents, such as Deflux, may be used to treat vesicoureteral reflux & will be visible on follow-up imaging. Cloaca & other anorectal malformations

are rarely imaged (except at dedicated centers). Bladder rhabdomyosarcoma is also uncommon but should be considered when a solid or multicystic mass is visualized in the bladder.

Imaging the Pediatric Genitourinary Tract

Ultrasound

Ultrasound is the mainstay of imaging in the pediatric genitourinary tract. Infants & children typically have excellent sonogenic body habitus. The advantages of ultrasound include that it is painless, noninvasive, portable, relatively inexpensive, & does not involve ionizing radiation. Although ultrasound is user dependent, assessments of renal lengths & hydronephrosis should be reproducible from exam to exam. With the approval of ultrasound contrast agents, ultrasound may be utilized increasingly for vesicoureteral reflux evaluation.

Fluoroscopic Voiding Cystourethrogram

Fluoroscopic VCUGs are most often performed to exclude vesicoureteral reflux but can also evaluate voiding dysfunction, enuresis, urethral abnormalities, & predisposing causes of infections. The study requires bladder catheterization by experienced healthcare providers, education of parents & children, optimal environmental distractions, & age-appropriate communication.

The VCUG is performed with the patient awake after aseptic bladder catheterization (with urine specimen collection optional). Standard views include a scout image of the pelvis & kidney regions, an early filling image of the bladder to exclude intraluminal abnormality, oblique views of the fully distended bladder to detect ureterovesical junction abnormalities, voiding images of the urethra, post void images of the pelvis & kidney regions, & spot views of any reflux or other abnormality.

Tips to aid with patient cooperation include the following: Providing distractions during the exam (such as videos, music, & toys; a child life specialist can be invaluable for both the physician & patient), pouring warm water over the perineum or toes if the patient has difficulty voiding, running water in the sink to provide an audible cue, or continuing to fill the bladder to 2x the estimated capacity, causing overflow incontinence (though this is not recommended in high spinal cord injury patients or postsurgical bladders).

Nuclear Medicine

Nuclear imaging is 2nd only to ultrasound in terms of utility in working up pediatric genitourinary abnormalities. Nuclear imaging studies examine the physiologic function of the urinary tract, not just the anatomy. All nuclear exams require an injection or catheterization, making patient & parental acceptance less than that for ultrasound.

Diuretic renal scans are widely used to monitor obstructive progression of hydronephrosis or surgical success.

Nuclear cystograms are widely used to follow vesicoureteral reflux, though historically lower radiation exposures (compared to VCUG) are now less substantial due to current fluoroscopic techniques.

DMSA renal cortical scans are still more sensitive & specific than ultrasound for documenting pyelonephritis & renal scarring.

Glomerular filtration studies are indispensable in patients on chemotherapy & with chronic renal insufficiency.

Magnetic Resonance Imaging

MR provides exquisite detail of genitourinary anatomy, & MR urography is able to quantify renal uptake & excretion rates. In fact, MR urography has the capacity for complete anatomic & physiologic assessment during a single exam, which may be useful in complex disorders. MR studies are motion sensitive, often requiring sedation, & are more costly compared to the other modalities. Although contrast-enhanced studies or sedation require intravenous access, MR does not utilize ionizing radiation.

Computed Tomography

Due to the risks of ionizing radiation, CT use is limited in pediatrics, most often for trauma & stone disease.

Intravenous Pyelogram

IVPs are rarely utilized in the pediatric population.

Embryology

A basic understanding of developmental anatomy will help explain the spectrum of congenital anomalies seen in the pediatric genitourinary tract.

During the 5th week of gestation, the mesonephric duct develops a budding tube (the ureteric bud) near its attachment to the cloaca. As this ureteric bud elongates, it reaches the metanephric blastema & undergoes a series of branchings to form the major & minor calyces, the renal pelvis, & the ureter. Signals from the ureteric bud induce changes in the metanephric blastema to form renal tubules, collecting tubules, & glomeruli of individual renal lobules. By 16- to 18-weeks gestation, the fetal kidneys are the major source of amniotic fluid for the fetus. The induction of mature renal lobules is complete by 32- to 36-weeks gestation.

While the kidney is forming, it migrates cephalad & rotates so that the renal pelves, which are initially directed anteriorly, are ultimately directed medially. At week 6, the kidneys lie in front of the primitive sacrum in the pelvis. By weeks 10-12, the kidneys have relatively ascended to their expected location along the upper lumbar spine. As the kidneys migrate, they pick up & discard nearby vascular attachments to both the aorta & inferior vena cava. This accounts for accessory vessels in normally located kidneys & for multiple vessels seen with ectopic & crossed fused kidneys.

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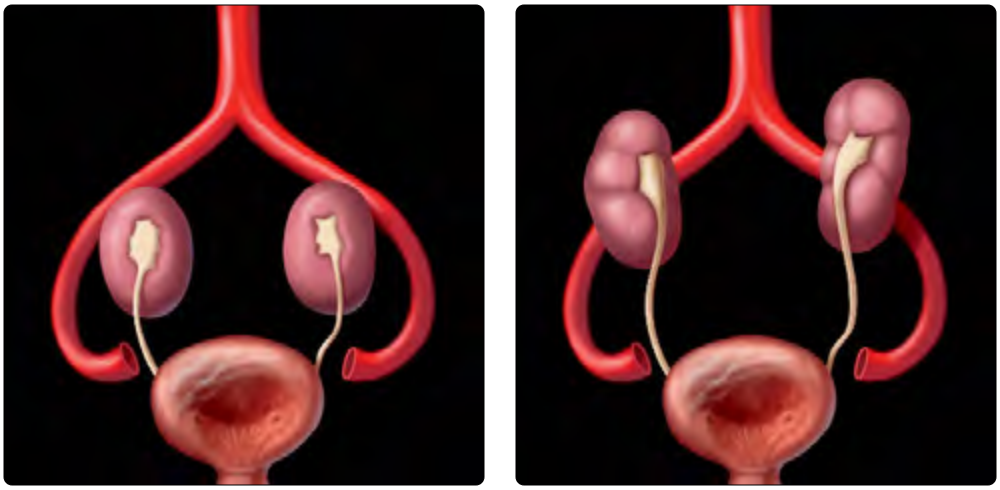
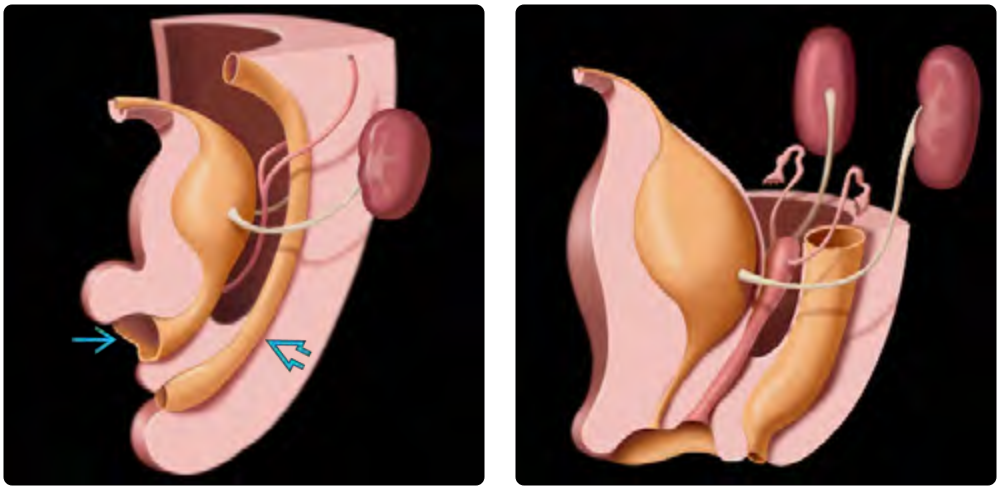
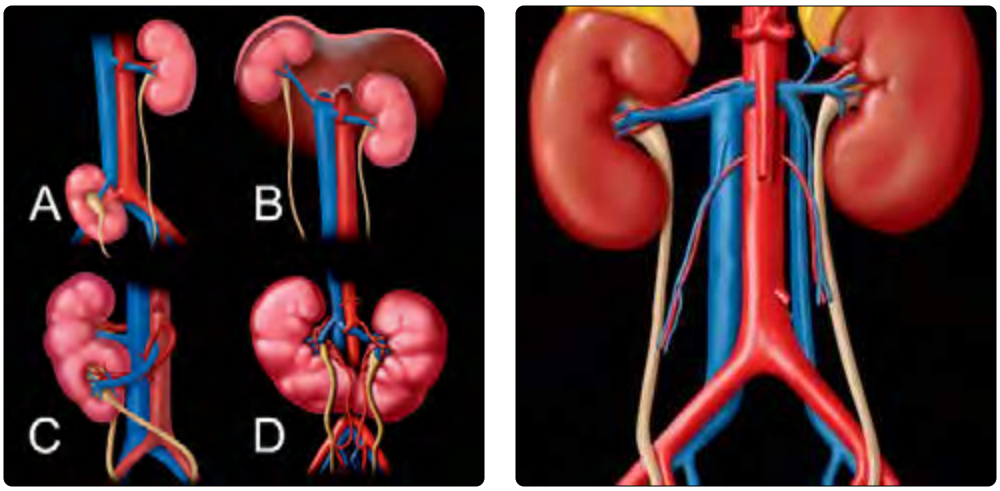
(Left) *Graphic shows early kidney formation as the ureteric bud extends from the primitive bladder/cloaca to the metanephric blastema & induces the formation of renal lobules. Note the low-starting position of the kidneys, in front of the sacrum, & the anterior location of the renal pelves.* **(Right)** *Graphic shows early ascent of the kidneys up from the fetal pelvis with rotation of the renal pelves medially. The renal axis is still vertical, but this will become oblique along the psoas muscle with further ascent.*

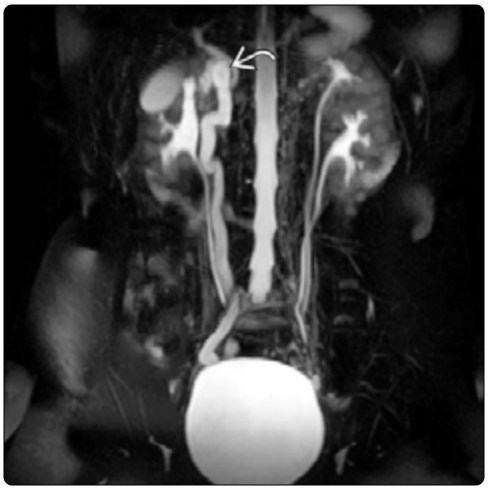


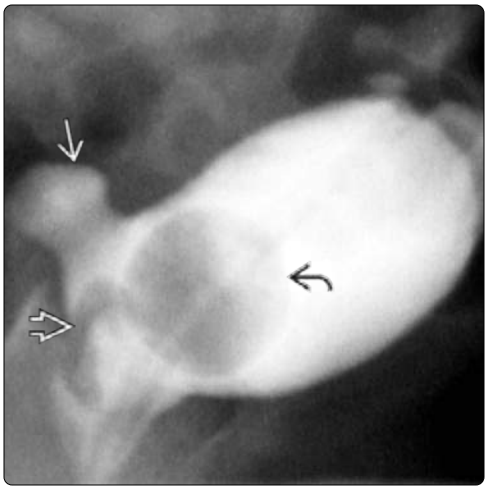
(Left) *Graphic from a lateral perspective of an early fetus shows the interrelated development of the genital & urinary organs. The mesonephric duct has differentiated into müllerian ducts budding from a common urogenital sinus ⇨ in this female fetus. The hindgut lies posterior ⇨.* **(Right)** *Graphic depicts a later developmental stage with separate urinary & genital tracts, ascent & rotation of the kidneys, & development of the uterus & fallopian tubes. The colon is seen posteriorly.*



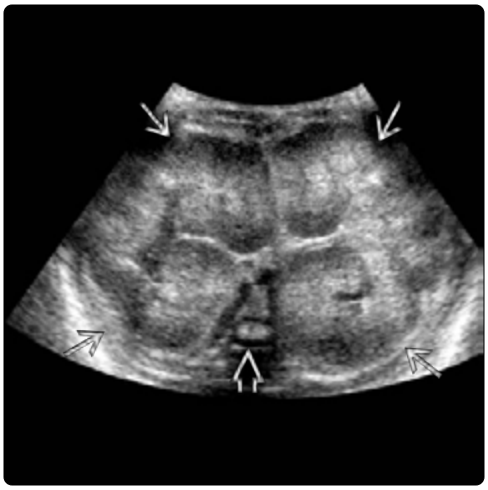
(Left) *Graphic illustrates the spectrum of renal ascent & fusion anomalies: (A) pelvic kidney, (B) thoracic kidney, (C) crossed fused ectopia, & (D) horseshoe kidney. Note the aberrant blood supply to the ectopically located kidneys.* **(Right)** *Graphic shows typical normal anatomy with kidneys in the upper lumbar region & renal pelves directed medially with single ureters & single renal arteries.*

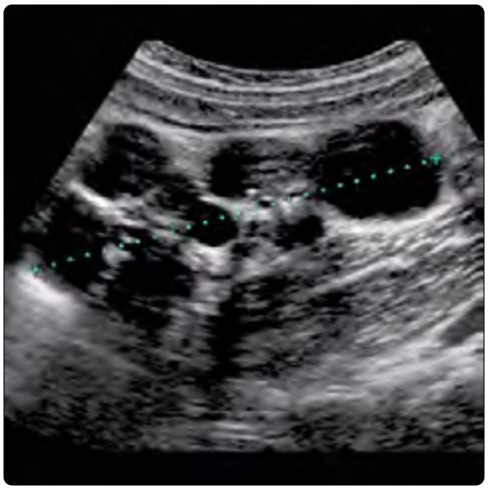




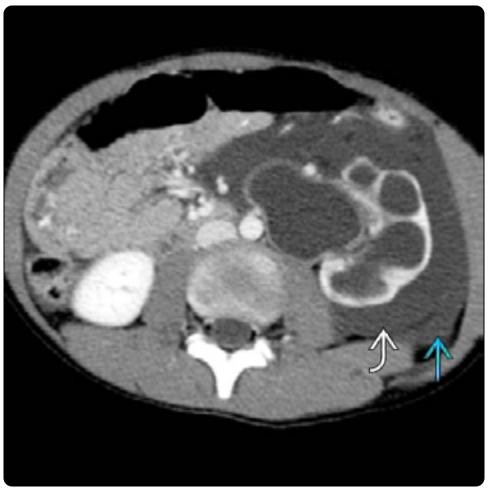


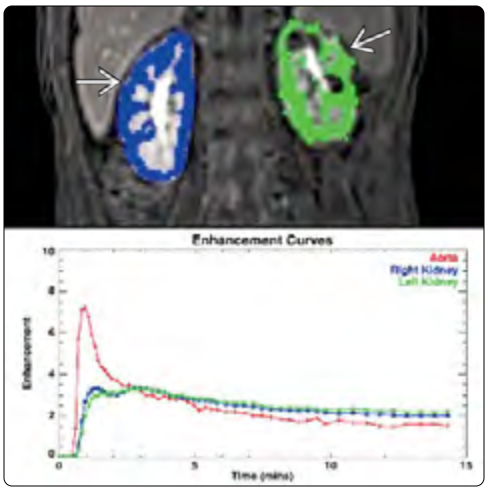
(Left) *Coronal MR urogram shows bilateral duplicated collecting systems & ureters in a teenager with flank pain & "giggle incontinence," likely related to the dilated right upper pole moiety ➡ inserting ectopically in the vagina (not shown).* **(Right)** *Voiding cystourethrogram shows an unusual case of a prolapsing ureterocele ➡ below the Foley catheter balloon ⇨. The ureterocele was causing intermittent bladder outlet obstruction. This patient also has a periureteral diverticulum ➡.*





(Left) *Transverse anterior abdominal US in a newborn shows massively enlarged echogenic kidneys ➡ in a patient with autosomal recessive polycystic kidney disease. The spine ➡ is visible in the midline posteriorly.* **(Right)** *Longitudinal US shows cysts completely replacing normal renal parenchyma in a case of multicystic dysplastic kidney.*





(Left) *Axial CECT shows marked hydronephrosis of the left kidney with a perinephric collection in a teenager following blunt abdominal trauma. The perinephric ➡ & paracolic gutter ⇨ fluid are due to rupture of a newly diagnosed ureteropelvic junction obstruction.* **(Right)** *Coronal contrast-enhanced MR urography illustrates the color-coded regions of interest for each kidney ➡ & the corresponding enhancement curves compared to the aorta in this normal study.*

Normal Neonatal Kidney

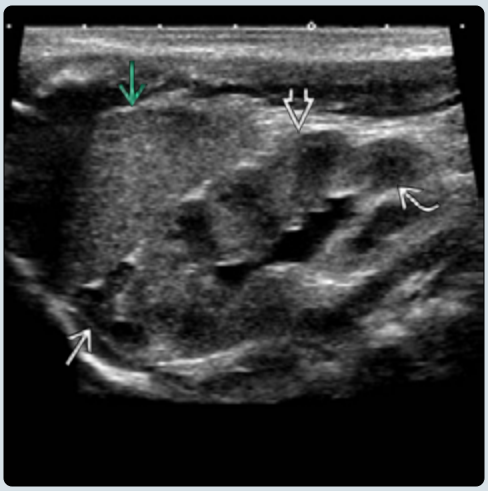
KEY FACTS

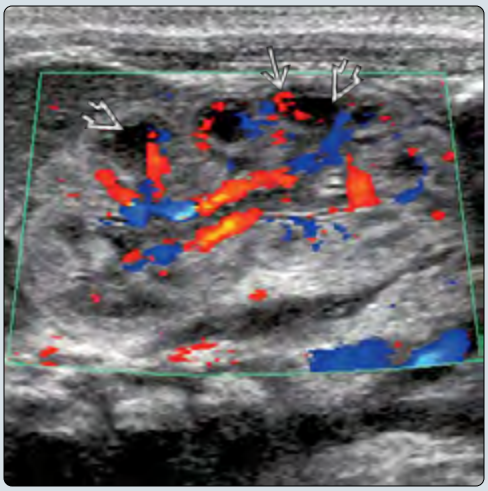
IMAGING

- Renal cortical echogenicity of normal term infant often same as adjacent normal liver
  - Cortical echogenicity ↑ in very premature infants compared to liver/spleen
- Medullary pyramids large & hypoechoic
  - Renal cortex thin relative to medullary pyramids
- Corticomedullary differentiation of infants & children > adults
  - Partially due to differences in cellular composition
  - Partially due to ↑ resolution of higher frequency transducers penetrating less overlying tissue
- Central echo complex less hyperechoic than adults
  - Less peripelvic fat in infants than adults
- Transient ↑ in echogenicity of pyramidal tips commonly seen in neonates; etiology uncertain
  - Self-limited: Usually resolves within few days
- Fetal lobulation ("lobation") of kidneys persists into newborn period & sometimes into childhood/adulthood
  - Not to be mistaken for scarring
- Arterial resistive indices ↓ over 1st year of life

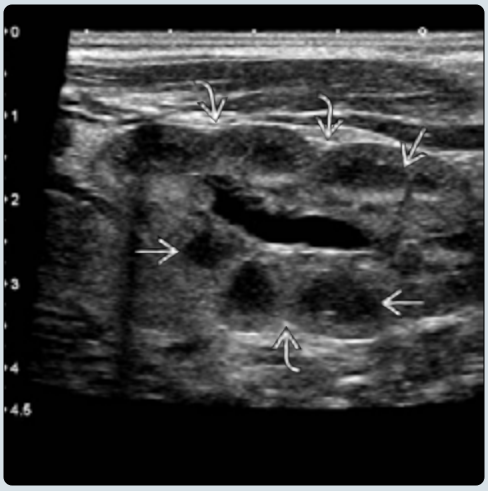
TOP DIFFERENTIAL DIAGNOSES

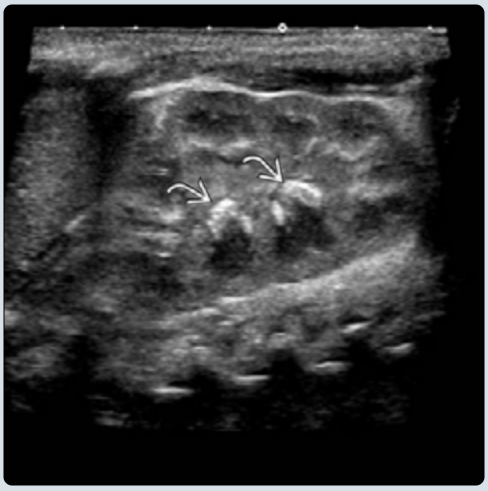
- Urinary tract obstruction
  - Pyramids do not interconnect as dilated calyces would
- Cystic renal disease
  - Normal pyramids line up around central echo complex
- Medical renal disease
  - Normal hyperechogenicity of neonatal renal cortex can mimic medical renal disease
  - Corticomedullary differentiation persists in normal kidneys of young children
- Renal scar
  - Fetal lobulation: Indentations between lobes/pyramids
  - Scars: Indentations within lobes/pyramids with dilated subtending calyces





(Left) *Longitudinal US of a newborn left kidney, adrenal gland, & spleen shows semicircular echogenic cortex overlying each hypoechoic pyramid. The renal cortical echogenicity is similar to the adjacent spleen.* **(Right)** *Longitudinal color Doppler US of a normal kidney in a 35-gestational-week newborn shows normal branching vessels extending to the corticomedullary junctions, confirming that the hypoechoic foci are renal pyramids, not cysts or dilated calyces.*





(Left) *Longitudinal prone US of the left kidney with a high-frequency linear transducer shows relatively echogenic cortex & underlying hypoechoic pyramids. Cortical indentations are seen between each pyramid ("fetal lobation").* **(Right)** *Prone longitudinal US of a newborn kidney shows increased echoes at the tips of the medullary pyramids, postulated to be related to Tamm-Horsfall proteinuria or other solute precipitation. This is a transient, self-limited finding in infancy.*

IMAGING

Ultrasonographic Findings

- Echogenicity of renal cortex in healthy term infant often same as adjacent normal liver
  - Renal cortex less echogenic than liver in older children & adults
- Renal cortex echogenicity typically ↑ in very premature infants compared to liver/spleen
  - May see striations in cortex & medulla
    - Due to vasculature in cortex & collecting ducts in medulla
- Corticomedullary differentiation of infants & children > adults
  - Partially due to differences in cellular composition of renal parenchyma
    - Greater density of glomeruli in renal cortex in neonates
  - Partially due to better resolution from higher frequency transducers with less tissue overlying kidney
  - If renal cortical echogenicity much > adjacent liver, suspect underlying renal parenchymal disease
- Renal cortex thin relative to medulla
- Medullary pyramids large & hypoechoic
  - ↑ echogenicity at tips of pyramids common in neonates, typically transient
    - Zone of maximal echogenicity at apex of pyramids with ↓ in echogenicity towards base of pyramids
      - Base of pyramids typically spared
    - Usually affects multiple pyramids but can range from 1 to all
    - Self-limited: Usually resolves within days as glomerular filtration rate ↑
      - May require 10 or more days, especially in premature infants
    - Historically thought to be due to transient precipitation of Tamm-Horsfall proteins
      - Not definitively proven
      - Exacerbated by hypernatremic dehydration (similar to reversible appearance of hypernatremic dehydration with oliguria in older patients)
- Central echo complex less hyperechoic than in adults
  - Less peripelvic fat in infants than in adults
- Neonatal kidneys often more spherical than kidneys in older children & adults
- Fetal lobulation ("lobation") of kidneys persists into newborn period (& sometimes into childhood/adulthood)
  - Not to be mistaken for scarring
- Sulcation: Interrenicular junction line, Odonno sulcus, junctional parenchymal defect
  - Commonly occurring hyperechoic sulcus in upper pole; controversial etiology
  - Not to be mistaken for scarring
- Rapid growth of kidney in 1st few months
  - Mainly due to disproportionate growth of cortical renal tubules
- Doppler shows ↓ intraparenchymal renal arterial resistive indices (RI) during early life
  - RI in preterm neonate: Up to 0.9 normal
  - RI < 1 year of age: 0.6-0.8 normal
  - RI > 1 year of age: 0.5-0.7 normal

Imaging Recommendations

- Best imaging tool
  - Ultrasound with high-frequency linear transducer
    - 14-6 MHz
  - Posterior or lateral decubitus approach

DIFFERENTIAL DIAGNOSIS

Urinary Tract Obstruction

- Normal hypoechoic pyramids in neonatal kidney can mimic dilated calyces (resulting in misinterpretation as hydronephrosis)
  - Pyramids especially prominent due to hyperechoic cortex
- Normal pyramids line up around central echo complex
  - Pyramids do not interconnect as dilated calyces would
- Position of arcuate artery at corticomedullary junction can help identify hypoechoic structure as pyramid

Cystic Renal Disease

- Normal hypoechoic pyramids in neonatal kidney can mimic cysts
- Normal pyramids line up around central echo complex
- Position of arcuate artery at corticomedullary junction can help identify hypoechoic structure as pyramid

Medical Renal Disease

- Normal hyperechogenicity of neonatal renal cortex can mimic medical renal disease
  - Cortical echogenicity in newborn same as or slightly > liver or spleen
- If echogenicity much > liver/spleen, consider other causes
  - Look for loss of corticomedullary differentiation

Renal Scar

- Normal fetal lobulation/lobation of neonatal kidney with relatively thin cortex can mimic scarring
- Fetal lobulation: Indentations between lobes/pyramids
- Scars: Indentations within lobes/pyramids, often with dilated subtending calyces

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Normal Neonatal Adrenal Gland

KEY FACTS

IMAGING

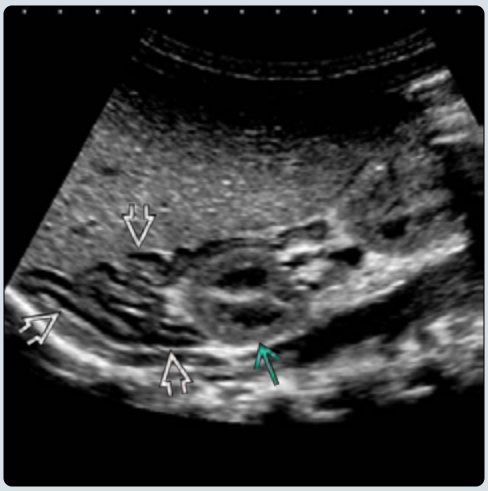
- US nicely shows newborn adrenal gland
  - Doppler useful in setting of neonatal adrenal lesions
- Normal outer hypoechoic fetal adrenal cortex & inner echogenic adrenal medulla have layered arrangement with mildly undulating margins
- Shape of newborn adrenal varies: Described as capital letters A, Y, V, X, or Z, or λ
- Appearance evolves during infancy
  - Newborn adrenal gland quite large (5 g), almost 2x weight of adult adrenal gland
  - Mainly due to prominent fetal cortex, which functions in utero & grows until term
    - Adrenal cortex initially much thicker than medulla; overall contour convex outward
  - Fetal cortex starts to involute after delivery; gradually ↓ until almost inapparent by 6 months of age
    - After 2-3 months, cortex & medulla equivalent in thickness; contour starts to flatten

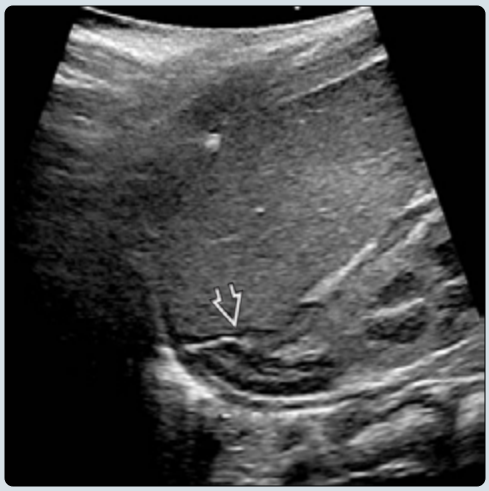
TOP DIFFERENTIAL DIAGNOSES

- Congenital adrenal hyperplasia
- Neonatal adrenal hemorrhage
- Neuroblastoma
- Extralobar bronchopulmonary sequestration
- Adrenal cyst
- Adrenal insufficiency
- Wolman disease

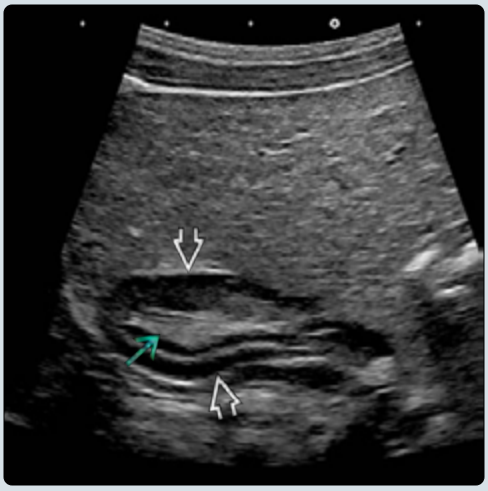
CLINICAL ISSUES

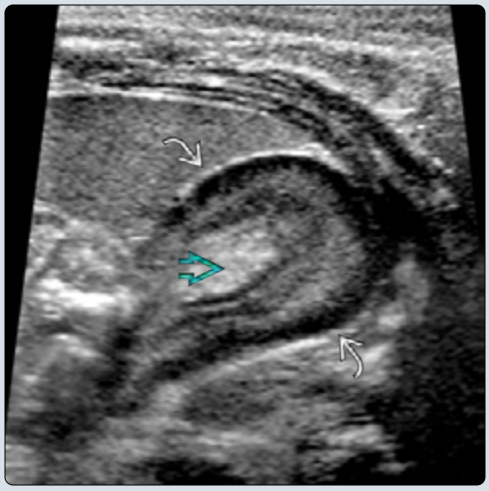
- Prominent newborn adrenal gland typically noted incidentally on spinal or renal US
- Awareness of normal newborn adrenal appearance avoids inappropriate work-up





(Left) *Longitudinal ultrasound of the right upper quadrant shows the folded, alternating layers of a normal newborn adrenal gland draped over the right kidney. The outer fetal adrenal cortex is hypoechoic compared with the inner echogenic medulla.* **(Right)** *Longitudinal view of the left adrenal gland in this newborn infant has a lambda shape.*





(Left) *Transverse ultrasound of the right suprarenal region shows echogenic fat between the limbs of the normal newborn adrenal gland.* **(Right)** *Transverse ultrasound of another normal adrenal gland shows a nearly "doughnut" configuration of the left adrenal gland with echogenic perirenal fat centrally. Despite its various shapes, the normal adrenal gland should not have any focal mass or distortion of the normal corticomedullary layers.*

TERMINOLOGY

Synonyms

- Suprarenal gland

Definitions

- Part of hypothalamic-pituitary axis that regulates many body functions & responds to stress
- **Adrenal cortex**: Largest part of adrenal gland; 3 layers produce different hormones (described below from superficial to deep)
  - **Zona glomerulosa**: Aldosterone (mineralocorticoid)
    - Retains sodium & wastes potassium; regulates fluid & electrolyte balance, helps maintain BP
  - **Zona fasciculata**: Cortisol (glucocorticoid)
    - Regulates metabolism of glucose, protein, & fat; responds to stress by ↑ blood glucose levels & cardiac output
  - **Zona reticularis**: Dehydroepiandrosterone
    - Sex hormone that works much like testosterone
- **Adrenal medulla**: Makes & stores epinephrine, norepinephrine

IMAGING

General Features

- Best diagnostic clue
  - Layered arrangement of outer hypoechoic fetal cortex & central echogenic medulla
  - Shape described as capital letters A, Y, V, X, or Z, or λ
- Location
  - Cephalad to kidney
- Size
  - 0.9-3.6 cm length, 0.2-0.3 cm thick
- Morphology
  - Newborn adrenal cortex much thicker than medulla; overall contour convex outward
    - Margins mildly undulating throughout
    - Small (< 5 mm) round focus of accessory adrenal tissue (with similar layered echogenicity) described in < 6% of neonates; typically not visible on follow-up
  - After 2-3 mo, cortex & medulla equivalent in thickness; contour starts to flatten
  - Around 6 mo, corticomedullary differentiation lost on imaging; contours flatten
  - After 1 yr, resembles adult gland with thin limbs & flat or concave margins

Imaging Recommendations

- Best imaging tool
  - US nicely shows newborn adrenal gland
- Protocol advice
  - Doppler helpful in evaluating adrenal lesions

DIFFERENTIAL DIAGNOSIS

Congenital Adrenal Hyperplasia

- Enlarged bilateral adrenal glands with redundant folds of cortex & medulla resembling sulci & gyri of brain (cerebriform appearance)
- Length > 2 cm & width > 4 mm suggests diagnosis (gland weight may reach 15 g)
- Due to deficiency in 1 of 5 enzymes necessary to produce hormones cortisol & aldosterone, leading to overproduction of androgen
  - Females manifest with ambiguous genitalia; males manifest with salt-wasting crisis

Neonatal Adrenal Hemorrhage

- Perinatal bleeding into normal gland, leading to avascular mass distorting adrenal tissue; gradually resolves
- Associated with perinatal stress: Asphyxia, sepsis, labile BP, birth trauma, coagulopathy
- Hemorrhage bilateral in 10% → ↑ risk of adrenal insufficiency

Neuroblastoma

- Most common malignancy in infancy
- Congenital/neonatal neuroblastoma tends to have good prognosis, even with disseminated disease (stage 4S/MS)
  - May spontaneously involute
- Neonatal neuroblastomas can be more cystic than tumors in older children

Extralobar Bronchopulmonary Sequestration

- Solid or mixed cystic & solid mass; can be in or below diaphragm above kidney

Adrenal Cyst

- Sequelae of prior hemorrhage or in association with Beckwith-Wiedemann syndrome

Adrenal Insufficiency

- Primary adrenal insufficiency rare in newborn; often due to adrenal hypoplasia congenita, syndromes of achalasia-addisonianism-alacrima syndrome or intrauterine growth restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, & genital anomalies
- Congenital aplasia very rare; found in 10% with unilateral renal agenesis

Wolman Disease

- Rare autosomal recessive lipid storage disorder
- Deficiency of lysosomal acid lipase → triglycerides & cholesterol esters build up in liver, spleen, adrenal glands
- Markedly enlarged adrenals with dystrophic Ca^{2+}

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prominent newborn adrenal gland usually noted incidentally on renal or spine US
  - Awareness of normal newborn adrenal appearance avoids inappropriate work-up

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Normal Prepubertal Uterus and Ovaries

KEY FACTS

TERMINOLOGY

- Thelarche: Onset & progress of breast development
- Adrenarche: Onset & progress of pubic/axillary hair development
- Menarche: 1st episode of vaginal bleeding

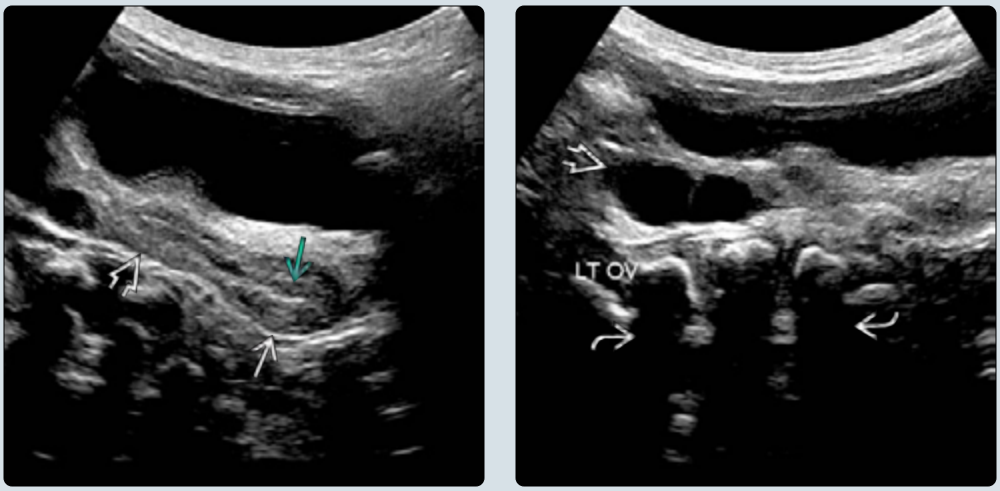
IMAGING

- Female genital organs best imaged with ultrasound or MR
- Uterus & cervix
  - Neonatal
    - Cervix larger than uterus, ~ 2:1 ratio
  - Prepubertal
    - Cervix & uterus equivalent
  - Pubertal
    - Uterus enlarges > cervix, 2:1 or 3:1 ratio
- Ovaries
  - Volumes
    - Neonatal ovarian volume generally < 1 mL
    - Prepubertal up to 3 mL (age 1-6 years)
    - Pubertal 3-20 mL
  - Morphology
    - Cysts can be seen in any age
    - If 3-5 cm, consider repeat scan in 6-8 weeks
    - Cysts can predispose to ovarian torsion
- **Best imaging tool**
  - Ultrasound screening, MR in complex cases

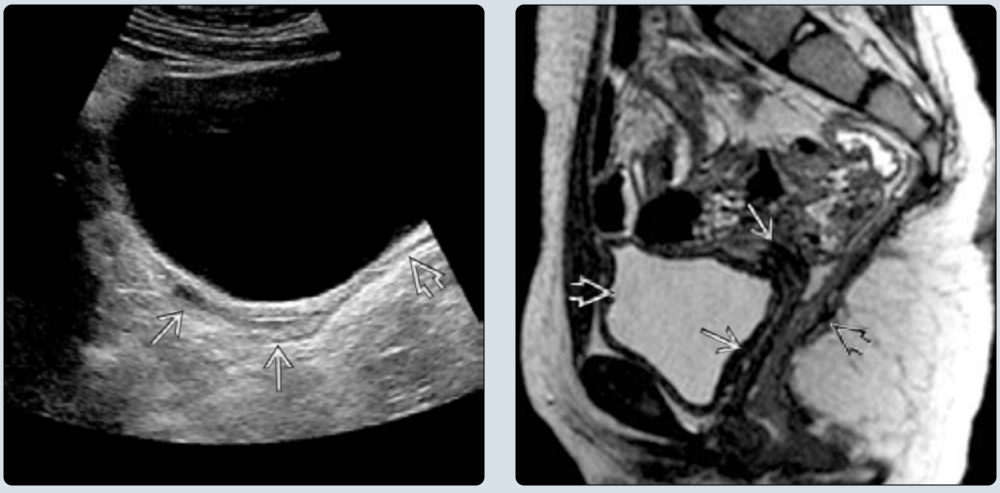
TOP DIFFERENTIAL DIAGNOSES

- Ambiguous genitalia
- Amenorrhea
  - Turner syndrome
  - Müllerian anomalies
  - Hypothalamic pituitary abnormalities
  - Constitutional/familial/other
- Prepubertal vaginal bleeding
  - Foreign body/abuse
- Ectopic ovary

(Left) *Midline longitudinal pelvic ultrasound in a newborn girl shows a relatively large cervix compared to the uterus due to maternal hormones. A thin ellipse of fluid is present in the lower uterine segment/upper cervical os.* **(Right)** *Longitudinal ultrasound just left of midline in the same newborn shows 2 small follicles in the left ovary. Physiologic cysts are common in the 1st few months of life. Note the posterior acoustic shadowing from the sacral vertebral segments posteriorly.*



(Left) *Midline longitudinal ultrasound through a very distended urinary bladder in a 7-year-old girl shows a thin, tubular cervix & uterus above a collapsed vaginal vault. Note the lack of fundal broadening in this prepubertal girl.* **(Right)** *Midline sagittal T2 MR in a 6-year-old girl being followed for a sacrococcygeal tumor shows a typical tubular appearance of the vaginal vault, cervix, & uterus (all blending together) between the bladder & collapsed rectosigmoid colon.*



TERMINOLOGY

Definitions

- Varied appearance of uterus & ovaries with age
  - Prominence in newborns due to maternal hormones
  - Small, quiescent stage in ages 1-8 years
  - Growth with hormonal surge during puberty
- Thelarche: Onset & progress of breast development
- Adrenarche: Onset & progress of pubic & axillary hair development
- Menarche: 1st episode of vaginal bleeding

IMAGING

General Features

- Best diagnostic clue
  - Female genital organs best imaged with ultrasound or MR
- Size
  - Uterus & cervix
    - Neonatal
      - Cervix larger than uterus, ~ 2:1 ratio
      - Uterus ~ 3.5 cm long x 1.5 cm thick
    - Prepubertal
      - Cervix & uterus equivalent
      - Uterus ~ 2.5-4 cm long x 1 cm thick
    - Pubertal
      - Uterus enlarges > cervix, 2:1 or 3:1 ratio
      - ~ 5-8 cm long x 1.5 cm thick x 3 cm transverse
  - Ovaries
    - Neonatal volume generally < 1 mL
    - Prepubertal up to 3 mL (age 1-6 years)
    - Pubertal 3-20 mL
- Morphology
  - Uterus
    - Neonatal: Endometrial echoes visible, fluid possible
    - Prepubertal: Very thin endometrial line
    - Pubertal: Measurable endometrial stripe
  - Ovaries
    - Ovarian cysts can be seen at any age
    - If 3-5 cm, consider 4-6 week follow-up
    - Cysts can predispose to torsion

Ultrasonographic Findings

- Primary diagnostic modality for uterus & ovaries

MR Findings

- Useful in complex anatomy or when ultrasound indeterminate
- Multiplanar & 3D images possible

CT Findings

- Genital findings seen incidentally in trauma/pain evaluation
- May be used in staging & follow-up of tumors

Imaging Recommendations

- Best imaging tool
  - Ultrasound; MR for complex cases
- Protocol advice
  - Endovaginal scanning avoided until adolescents sexually active or using tampons (postpubertal patients)
  - Transabdominal
    - Adequate hydration preceding exam as well-distended urinary bladder needed for sonographic window
      - Use gentle transducer pressure on bladder to avoid inducing micturition
    - Alternatively, use Foley catheter to fill bladder
  - Limited need for Doppler

DIFFERENTIAL DIAGNOSIS

Ambiguous Genitalia

- Pelvic ultrasound in neonatal period searching for gonads: Include scrotum/labia
  - Female pseudohermaphroditism due to congenital adrenal hyperplasia
  - True hermaphroditism

Amenorrhea

- Turner syndrome
  - Typical prepubertal uterus & streak ovaries
  - 5-15% normal pubertal onset: Mosaicism
- Müllerian anomalies
  - Müllerian agenesis
  - Obstructive müllerian anomalies
  - Nonobstructive müllerian anomalies
- Hypothalamic pituitary abnormalities
- Constitutional/familial/other

Prepubertal Vaginal Bleeding

- Foreign body/abuse
- Vaginal rhabdomyosarcoma
- Precocious puberty

Ectopic Ovary

- Canal of Nuck hernia containing ovary

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Ureteropelvic Junction Obstruction

KEY FACTS

TERMINOLOGY

- Ureteropelvic junction (UPJ) obstruction most common form of urinary tract obstruction in children

IMAGING

- Marked pelvocaliectasis that ends abruptly at UPJ with normal caliber ureter downstream
- Dilated calyces relatively uniform in size & distribution; all connect centrally to disproportionately dilated pelvis
- Thinned but otherwise intact renal parenchyma
- Doppler US shows ↑ renal resistive indices with obstruction
- Severity of delayed nephrogram, excretion, & collecting system drainage (on IVP, CECT, MRU, or nuclear renal scan) depends on degree of obstruction
- Contrast entering dilated collecting system (by excretion, retrograde injection, or vesicoureteral reflux during VCUG) may be very dilute due to mixing with retained urine
- ± crossing vessel at obstruction site on CECT, MRU, or US
- Nuclear medicine renal scan well-established for initial assessment & follow-up of renal function & obstruction
- MR urography may provide optimal combination of anatomic & physiologic assessment

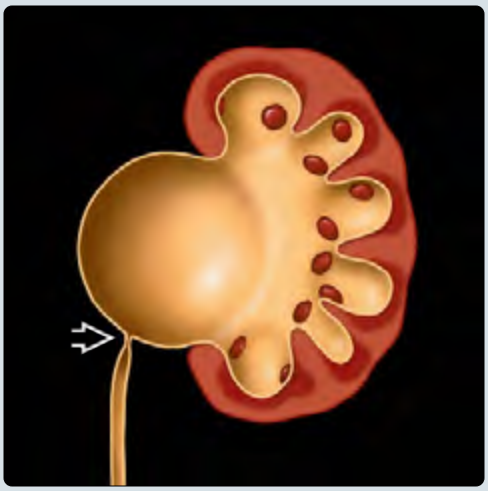
PATHOLOGY

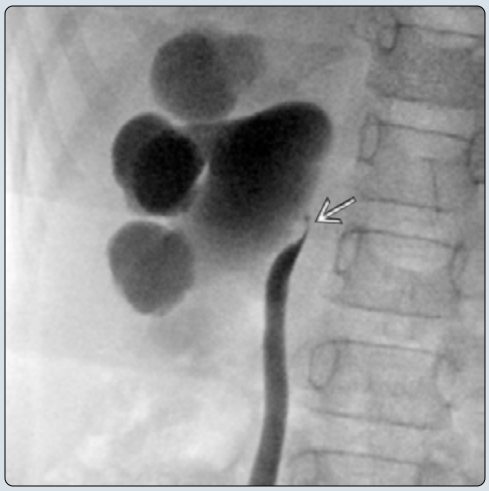
- Theoretical etiology of obstruction at UPJ
  - Abnormal smooth muscle arrangement impairs distensibility
  - Abnormal innervation of proximal ureter (Hirschsprung equivalent)
  - Crossing vessel or fibrous scar at UPJ
- UPJ obstruction associated with contralateral MCDK

CLINICAL ISSUES

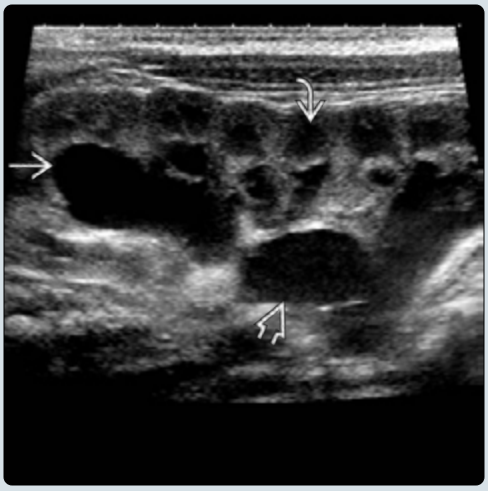
- May be diagnosed antenatally or in infancy/childhood with urinary tract infection, intermittent abdominal/flank pain, or hematuria
- Treatment: Pyeloplasty, ureteroureterostomy, or endoureteral balloon plasty/stenting (infants)

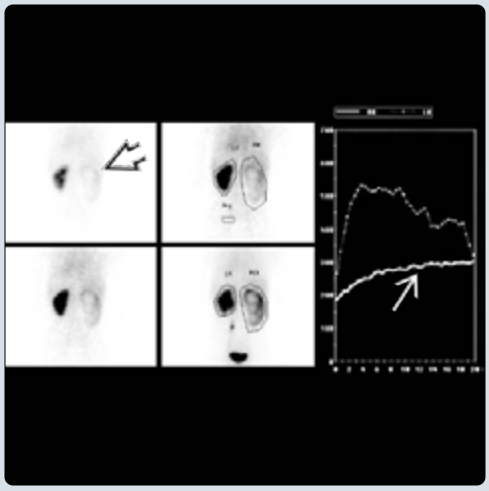
(Left) *Graphic shows a ureteropelvic junction (UPJ) obstruction with massive dilation of the renal pelvis, disproportionate to the dilated calyces. Note the focal narrowing at the UPJ ⇨ & the nondilated proximal ureter.* **(Right)** *Frontal fluoroscopic image in the OR during cystoscopy & retrograde ureterography shows a dilated right renal collecting system & abrupt caliber change at the UPJ ⇨. Intraoperative imaging may be performed to exclude an intraluminal polyp or stone & determine the best surgical approach.*





(Left) *Longitudinal US of an infant with prenatal hydronephrosis shows dilated calyces ⇨ & a large renal pelvis ⇨, subsequently diagnosed as a UPJ obstruction. Hypoechoic renal pyramids ⇨ are normal in infants.* **(Right)** *Posterior images of a Tc-99m MAG3 diuretic renal scintigraphy show minimal uptake in a dilated right kidney ⇨. The time activity curve for the right kidney shows progressive accumulation of counts ⇨ with no washout during the exam, typical of a UPJ obstruction.*





TERMINOLOGY

Abbreviations

- Ureteropelvic junction (UPJ) obstruction

Synonyms

- Pelviureteric obstruction

Definitions

- Variable degree of blockage to urine flow at level of UPJ
- Most common urinary tract obstruction in pediatrics
- May be diagnosed antenatally with sonography or present in infancy or later childhood with UTI, intermittent abdominal pain, vomiting, hematuria
  - Occasionally found incidentally during trauma work-up

IMAGING

General Features

- Best diagnostic clue
  - Marked pelvocaliectasis that ends abruptly at UPJ with normal caliber ureter
  - Disproportionate enlargement of renal pelvis
- Morphology
  - UPJ obstruction has been likened to Hirschsprung disease with focal transition zone & aperistaltic segment
  - Obstruction may be extrinsic rather than intrinsic
    - Crossing vessel at UPJ in 25% infants
- Enhancement/uptake
  - Severity of delayed nephrogram, excretion, & collecting system drainage depends on degree of obstruction

Radiographic Findings

- Radiography
  - May see mass effect from enlarged hydronephrotic kidney; ± scoliosis/splinting with pain
- IVP
  - Delayed nephrogram
  - Delayed contrast excretion into dilated collecting system
    - Contrast gradually opacifies distended renal pelvis, which tapers abruptly
    - Contrast may be very dilute due to mixing with retained urine
  - Delayed ureteral visualization

Ultrasonographic Findings

- Grayscale ultrasound
  - Moderate to severe pelvocaliectasis without hydroureter
  - Dilated calyces relatively uniform in size & distribution; all connect centrally to disproportionately dilated pelvis
  - Abrupt tapering of pelvis at UPJ
  - Thinned but otherwise intact renal parenchyma
- Pulsed Doppler
  - ↑ resistive indices (RIs) with obstruction
    - May be able to correlate RIs with degree of obstruction (when contralateral kidney normal & can serve as internal standard)
    - Because RIs change with age, strict cutoff value for obstruction not applicable in pediatrics
- Color Doppler
  - Search for crossing aberrant vessel at site of obstruction
  - Ureteral jets (in urinary bladder) useful in excluding complete obstruction
  - Qualitative assessment for dampened, infrequent, or abnormally angulated jets (in setting of partial obstruction) is difficult

Fluoroscopic Findings

- Voiding cystourethrogram
  - With ≥ grade 2 vesicoureteral reflux (VUR), contrast may be seen entering disproportionately dilated renal pelvis (relative to ureter)
    - Chronically dilated ureter may kink at UPJ, creating additional etiology for pelvocaliectasis
    - Look for delayed drainage of high grade VUR
  - May see contralateral VUR
- Intraoperative retrograde ureterogram variably used to confirm focal narrowing, identify crossing vessels (by negative impression), search for intraluminal polyp or stone
  - Shows abrupt transition of normal caliber ureter to dilated renal pelvis
  - Contrast entering renal pelvis becomes very dilute by retained volume of urine

CT Findings

- CECT
  - Delayed nephrogram in enlarged kidney
  - Marked renal pelvic > calyceal dilation with normal or nonvisualized ureter
  - May see crossing vessel at UPJ
  - Delayed contrast excretion into collecting system

MR Findings

- Hydronephrosis without hydroureter
- MR urography ± contrast particularly helpful in visualizing crossing vessels
- Drainage curves & differential function can be used to guide timing of surgery

Nuclear Medicine Findings

- Renal scans show delayed uptake, excretion, & drainage of radiotracer
  - Early central photopenia of affected kidney
  - Gradual & progressive accumulation of radiotracer replacing central photopenia
  - Little to no ureteral activity; variable change with diuretic administration
- Time activity curve provides quantified assessment of radiotracer distribution throughout scan
  - Time to half of peak activity (T½) can measure of severity of obstruction
    - T ½ < 10 min is normal
    - T ½ > 20 min is obstructed
    - T ½ between 10-20 minutes is indeterminate
- Nuclear studies used for initial assessment & follow-up of function & obstruction
- Study should be performed in standardized fashion with adequate hydration, bladder drainage, & diuretic administration

Other Modality Findings

- Whitaker test (pressure monitoring during direct fluid infusion into collecting system): Historically important, now rarely performed

Imaging Recommendations

- Best imaging tool
  - Sonography usually performed 1st
  - Nuclear renal scan then used to grade degree of obstruction & determine if surgical intervention or percutaneous drainage required
  - MR urography may be viable single test alternative, optimizing anatomic & physiologic assessment
- Protocol advice
  - Obstruction often partial, can improve or worsen over time
  - Affected children typically undergo serial exams every 6-12 months (if they remain asymptomatic) to determine when to intervene

DIFFERENTIAL DIAGNOSIS

Multicystic Dysplastic Kidney

- No discernible normal renal parenchyma
- Cysts do not interconnect
- Largest cyst not usually located centrally

Ureteral Fibroepithelial Polyp

- Found in 5% of UPJ obstructions
- Increasing awareness with pediatric ureteroscopy

Megacalycosis or Congenital Megacalyces

- Idiopathic dilation & ↑ number of calyces without enlarged renal pelvis
- Drainage in megacalycosis normal or minimally delayed
- Often presents with hematuria after minor trauma

Hydronephrosis of Other Etiologies

- VUR
- Renal stone
- Ureterovesical junction obstruction
- Ureterocele

Megaureter

- Dilated ureter with narrowed distal aperistaltic segment
- Obstructed or nonobstructed, refluxing or nonrefluxing

PATHOLOGY

General Features

- Etiology
  - At surgical resection, massively dilated pelvis often too distorted to confirm any single theory
  - Theoretical etiology of obstruction at UPJ
    - Abnormal smooth muscle impairs distensibility
    - Abnormal innervation of proximal ureter (Hirschsprung equivalent)
    - Crossing vessel or fibrous scar at UPJ
- Associated abnormalities
  - Contralateral UPJ obstruction associated with multicystic dysplastic kidney (MCDK)
    - Requires prompt intervention since MCDK nonfunctional & UPJ may compromise remaining renal function

Staging, Grading, & Classification

- Anteroposterior pelvic diameter > 10 mm in 3rd-trimester fetus or newborn suggests obstruction

Microscopic Features

- Obstructive nephropathy leads to tubulointerstitial fibrosis & loss of renal function
- Nerve fibers depleted in muscular layer in ureteric walls
- Denervation results in dysfunction/atrophy of muscle fibers & ↑ collagen fibers within muscle layers
- Deficient Cajal cells (pacemaker cells in smooth muscles), which enhance ureteral peristalsis
- Abnormal accumulations of intercellular & interstitial collagen also seen pathologically

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Antenatally detected on fetal sonogram or MR
- Other signs/symptoms
  - Infants & children: Urinary tract infection, intermittent abdominal pain, flank pain, or hematuria
  - In older children who present with symptomatic UPJ obstruction, crossing vessel causative in ~ 50%

Natural History & Prognosis

- May improve or deteriorate spontaneously
- Prognosis excellent if renal function has not been compromised by longstanding, high-grade obstruction
- Following successful surgery, pelvocaliectasis persists for years on sonography
- Appropriate renal growth & adequate drainage on nuclear scans help measure surgical success

Treatment

- Pyeloplasty (open or laparoscopic surgery)
  - Tapered, dismembered pyeloplasty has been classic surgery for UPJ obstruction
  - Narrowed segment resected, or crossing vessel rerouted
  - Ureteral stents often left in place (crossing surgical anastomosis) for several weeks postop
  - Open or laparoscopic procedures preferable when crossing vessels or aberrant vessels recognized
- Endoscopic incision (a.k.a. endopyelotomy)
- Endopyeloplasty (horizontal percutaneous suturing of conventional longitudinal endopyelotomy incision)
- Ureterocalicostomy, which is reconstructive option in rare patient with surgically failed UPJ or difficult anatomy due to fibrosis or other concurrent problems
- Endoureteral balloon plasty/stenting
- Percutaneous drainage, temporizing, if infected

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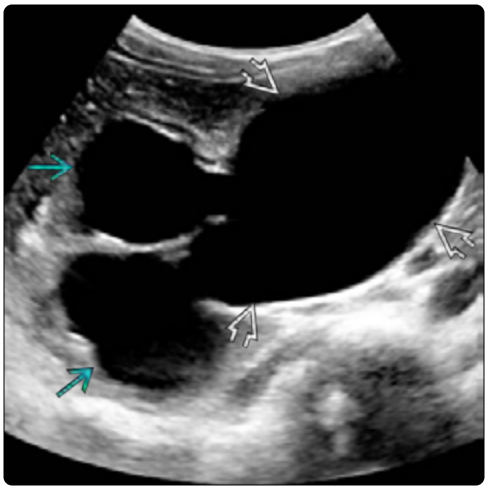
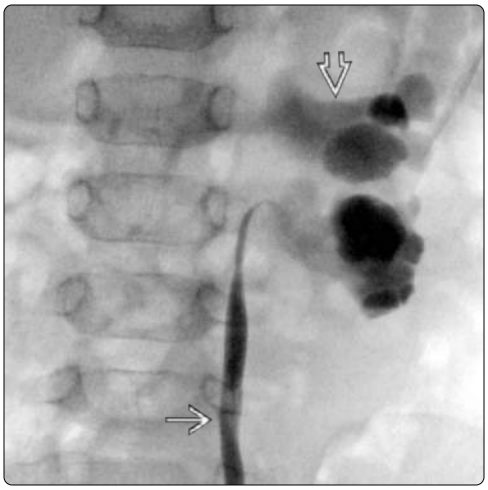
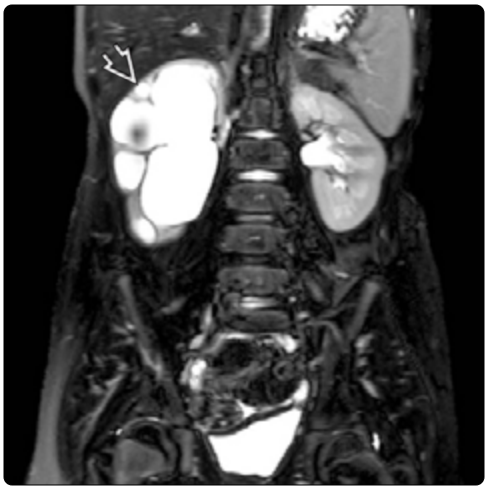
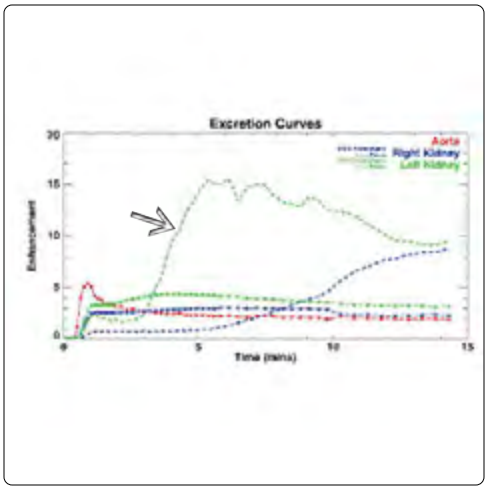


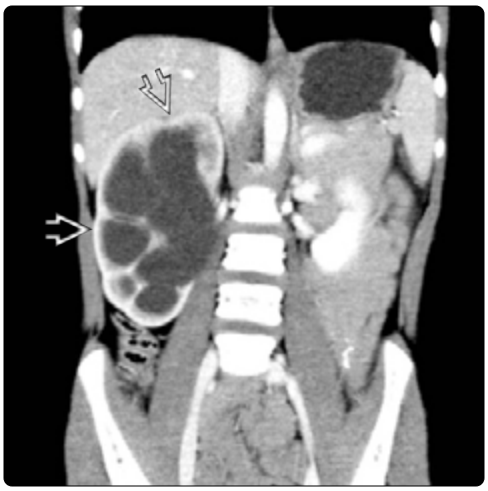


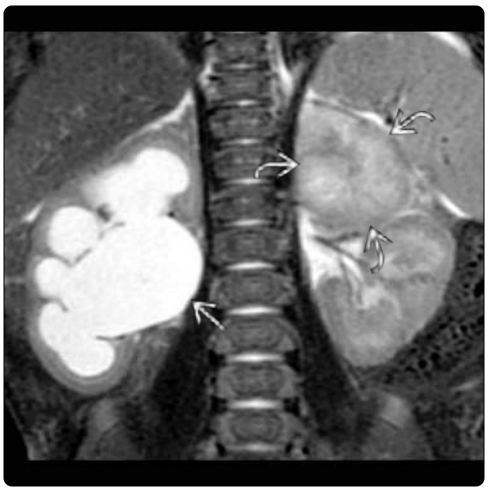
(Left) *Transverse US of the left kidney shows dramatically dilated calyces converging on the centrally located & very dilated renal pelvis. No ureter was identified draining the dilated system, suggesting a UPJ obstruction.* **(Right)** *Retrograde ureteral injection shows a normal caliber ureter with dilution of contrast in the massively dilated left renal pelvis & collecting system in this case of a left UPJ obstruction.*





(Left) *Coronal MR urography shows marked right pelvocalicectasis in this teenager with intermittent flank pain caused by a UPJ obstruction. A crossing vessel was not identified.* **(Right)** *MR urography excretion curves (in a different patient) are similar to nuclear time activity curves. In this case, the left kidney shows delayed uptake, excretion, & drainage, typical of a UPJ obstruction.*





(Left) *Coronal CECT shows a markedly hydronephrotic right kidney. On adjacent images (not shown), the renal pelvis appeared dilated, but no ureter was visualized, consistent with UPJ obstruction.* **(Right)** *Coronal T2 FS MR shows an incidentally discovered right UPJ obstruction in an infant being evaluated for a left adrenal hemorrhage vs. mass. The adrenal mass was resected & found to be a neuroblastoma. The right UPJ obstruction was also repaired.*

KEY FACTS

TERMINOLOGY

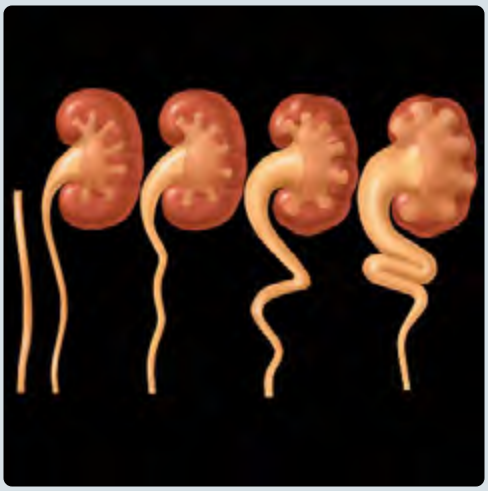
- Retrograde flow of urine from bladder toward 1 or both kidneys

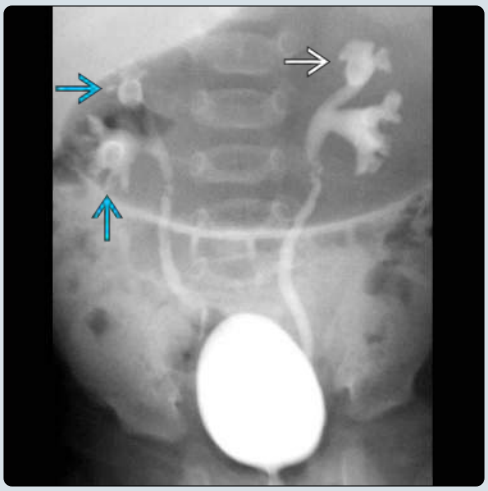
IMAGING

- International Reflux Study Committee grading system of vesicoureteral reflux (VUR)
  - I: Reflux into ureter but not reaching renal pelvis
  - II: Reflux reaching pelvis without blunting of calyces
  - III: Mild calyceal blunting
  - IV: Progressive calyceal & ureteral dilation
  - V: Very dilated & tortuous collecting system
  - ± intrarenal reflux as modifier to grade II+
- Voiding cystourethrogram preferred whenever anatomic detail of upper tracts & urethra needed
- Nuclear cystogram preferred when anatomy is known (e.g., renal ultrasound normal) &/or for follow-up studies
- Use of renal US alone for screening controversial: Variable sensitivity & specificity for scar as compared to DMSA

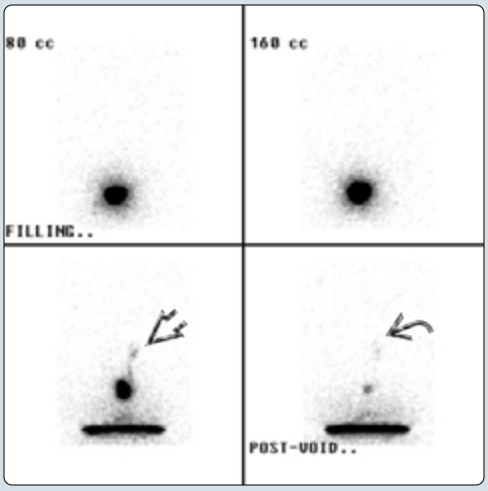
CLINICAL ISSUES

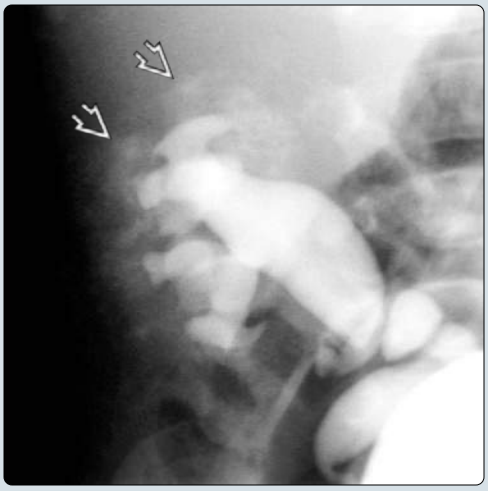
- Up to 2% of general population
- VUR seen in 25-40% of children with acute pyelonephritis
- VUR seen in 5-50% of asymptomatic siblings of children with documented reflux
- 80% outgrow VUR before puberty
- ↑ grade or longer standing VUR, more numerous UTIs, & subsequent renal scarring: ↑ incidence of renal insufficiency, hypertension, & end-stage renal disease
- Treatment options
  - Prophylactic antibiotic therapy (medical management)
  - Ureteral reimplantation surgery (surgical management)
  - Endoscopic periureteral injections (minimally invasive endoscopic management)
    - Treatment-induced hydroureteronephrosis following endoscopic procedures uncommon & usually self-limited





(Left) *Graphic depiction of the International Reflux Study Committee grading system is shown. Note the progressive level of reflux, dilation, calyceal blunting, & ureteral tortuosity from grade I on the left to grade V on the right.* **(Right)** *Voiding cystourethrogram (VCUG) in an infant shows bilateral vesicoureteral reflux (VUR). The calyces are sharp ⇨ on the right (grade II). On the left, the calyces are slightly blunted ➡ & the ureter is mildly dilated & tortuous, making this grade III VUR.*





(Left) *Posterior nuclear cystogram images show radiotracer extending into the right ureter & reaching the intrarenal collecting system ⇨, likely corresponding to fluoroscopic grade II VUR. Note that reflux occurs only during voiding & drains well on the postvoid image ⇨.* **(Right)** *Frontal VCUG shows high-grade VUR into the right kidney with a dilated tortuous ureter, blunted calyces, & intrarenal reflux into the tubules ➡. These findings constitute grade V VUR with intrarenal reflux.*

TERMINOLOGY

Abbreviations

- Vesicoureteral reflux (VUR)

Definitions

- Retrograde flow of urine from bladder toward 1 or both kidneys

IMAGING

General Features

- Best diagnostic clue
  - Contrast instilled into urinary bladder opacifies at least 1 ureter
  - Contrast may reach intrarenal collecting system
  - Findings often seen only transiently

Fluoroscopic Findings

- Voiding cystourethrogram
  - Requires bladder catheterization for contrast installation
  - Preliminary scout film useful, especially when there has been prior surgery or urolithiasis
  - Early filling image of bladder best to show intraluminal abnormalities: Ureterocele, polyp, mass
  - Contrast seen in ureter &/or renal collecting system confirms VUR
  - Oblique views of distended bladder helpful to show periureteral diverticula & ureteric insertion site in cases of VUR
  - Ectopic ureters that insert below bladder neck only reflux during voiding (if they do drain to bladder)
    - May not visualize ectopic ureters unless they are inadvertently catheterized
    - In such cases, separate catheter should also be passed into urinary bladder to study other ureters for VUR
  - Voiding images of urethra performed to exclude distal pathology, which may contribute to back pressure
    - Obtain voiding images without catheter in place
  - In cases of high-grade reflux, delayed upright image used to assess drainage from upper tract & exclude concomitant ureteropelvic junction or ureterovesical junction obstruction
    - Note that contrast refluxed into obstructed, dilated system may become very diluted

Ultrasonographic Findings

- Varying degrees of renal collecting system &/or ureteral dilation ± urothelial thickening
  - Normal US without dilation does not exclude significant VUR
- Look for signs of scarring: Globally small kidney or polar foci of cortical thinning subtended by dilated calyces
- US contrast agent can be instilled into bladder for sonographic cystogram
  - Bladder catheterization still required; no ionizing radiation used
  - Urethral evaluation possible with ultrasound probe on perineum
- Doppler optimized for low velocity (SMI) has been used to show retrograde flow in ureter without contrast

Nuclear Medicine Findings

- Nuclear cystogram
  - Study performed with posterior gamma camera
  - Tc-99m pertechnetate instilled into bladder via catheter
  - Imaging performed throughout bladder filling & voiding
  - With VUR, radiotracer activity extends cephalad from bladder in varying amounts
  - Imaging continuous
  - Grade I VUR harder to see on nuclear cystogram due to bladder activity
  - Nuclear cystogram gives no information about urethral abnormalities
- Renal cortical scan
  - Study performed with posterior pinhole imaging
  - Radiotracer: Tc-99m DMSA or MAG3
  - Photopenic renal cortical foci due to acute pyelonephritis or chronic scarring

Imaging Recommendations

- Best imaging tool
  - Controversial
  - Guidelines from American Academy of Pediatrics & National Institute for Health & Care Excellence in United Kingdom both advocate less imaging than historically performed
  - Traditionally
    - Voiding cystourethrogram (VCUG) preferred whenever anatomic detail of upper tracts or urethra needed
    - Nuclear cystogram preferred when anatomy is known &/or for follow-up studies
    - Cystosonography alternative at sites with ultrasound contrast agents
  - Historic "bottom-up" approach to UTI work-up: VCUG + renal US; DMSA if 1st-line studies abnormal or with febrile UTI
  - Current "top-down" approach to UTI work-up: Renal US ± DMSA; VCUG if 1st-line studies abnormal
  - **Use of renal US alone for screening controversial due to variable sensitivity (37-100%) & specificity (65-99%) for scar as compared to DMSA**
  - DMSA therefore advocated by some as better test for scarring, which serves as surrogate for high-grade VUR requiring treatment (with scarring in 50% of grades IV-V but < 10% of grades I-III VUR)
    - Note that RIVUR trial showed no difference in progressive scarring on antibiotic prophylaxis vs. placebo
    - ↑ scars in older patients, higher grade VUR, & those with recurrent febrile UTI
  - Though anatomic detail < VCUG, continuous imaging of nuclear medicine ↑ detection of transient VUR so that nuclear cystogram is more sensitive for VUR

DIFFERENTIAL DIAGNOSIS

Fluoroscopic Mimics of VUR

- Normal bowel wall contrasted by intraluminal air, contrast or stool **or** bony iliopectineal line can mimic contrast in ureter
  - Clarify with oblique views & comparison to scout image

- Ventriculoperitoneal tubing & other intraabdominal catheters can resemble contrast-filled ureter

Urolithiasis, Especially Staghorn Calculus

- Density may simulate contrast in renal pelvis
- Check scout image, watch for postvoid drainage

Sonographic Mimics of Vesicoureteral Reflux

- Normally peristalsing ureter or renal pelvis
- Distended distal ureter in patients with very full bladders
- Pathologies causing obstruction of ureter or renal pelvis

PATHOLOGY

General Features

- Etiology
  - Shortened or abnormally angulated insertion of ureter into bladder theorized to result in primary VUR
  - Vast majority (80%) of pediatric patients outgrow primary VUR, presumably due to changes at level of ureterovesical junction
  - VUR may also be secondary to periureteral (Hutch) diverticulum, ureterocele, bladder outlet obstruction, voiding dysfunction, or neurogenic bladder
  - Probable association of sterile reflux with renal scarring
    - Antibiotic prophylaxis after 1st UTI reduces febrile UTI recurrences, not scarring
  - Risk factors: Family history, sex, age at presentation, duplication, & other voiding dysfunctions
- Associated abnormalities
  - Multicystic dysplastic kidney
  - Ectopic kidney
    - Note that reflux most commonly involves contralateral orthotopic kidney
  - VUR present in 1/3 of patients with acute pyelonephritis

Staging, Grading, & Classification

- **International Reflux Study Committee grading system of VUR**
  - I: Reflux into ureter but not reaching renal pelvis
  - II: Reflux reaching pelvis without blunting of calyces
  - III: Mild calyceal blunting
  - IV: Progressive calyceal & ureteral dilation
  - V: Very dilated & tortuous collecting system
  - ± intrarenal reflux as modifier to grade II+

Gross Pathologic & Surgical Features

- Deficiency or immaturity of longitudinal muscle in submucosal ureter
- Abnormal angle of ureteral insertion through bladder wall, which tends to correct as ureter grows & elongates
- Distortion of ureteral insertion by adjacent bladder anomaly

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually discovered during work-up of febrile UTI

Demographics

- Age
  - VUR most common in children < 2 years old
  - 0.5x as likely in those 3-6 years old
  - 0.3x as likely in those 7-11 years old
  - 0.15x as likely in those 12-21 years old
- Gender
  - F:M = 2:1
- Epidemiology
  - Up to 2% of general population
  - VUR in 25-40% of children with acute pyelonephritis
  - VUR in 5-50% of asymptomatic siblings of children with documented reflux

Natural History & Prognosis

- 80% outgrow VUR before puberty
- With higher grade or longer standing VUR, more numerous UTIs, & subsequent renal scarring: Incidence of renal insufficiency, hypertension, & end-stage renal disease increases

Treatment

- Prophylactic antibiotic therapy (medical management)
  - Reduces febrile UTI recurrences, not scarring
- Ureteral reimplantation surgery (surgical management)
- Endoscopic periureteral injections (minimally invasive endoscopic management) utilizing inert material to alter shape of abnormal/refluxing ureterovesical junction
  - Treatment-induced hydroureteronephrosis following endoscopic procedures uncommon & usually self-limited

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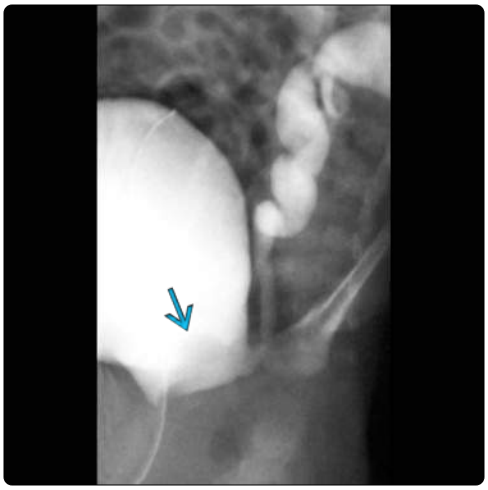
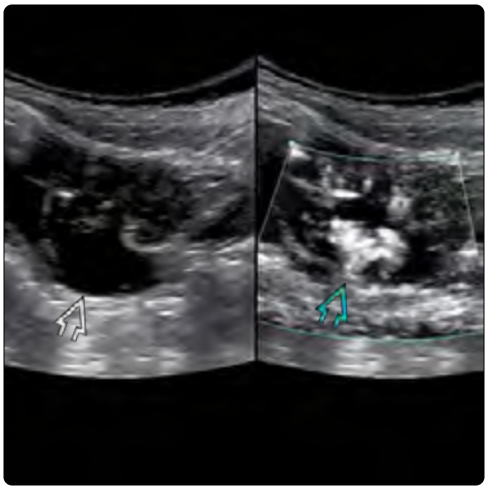
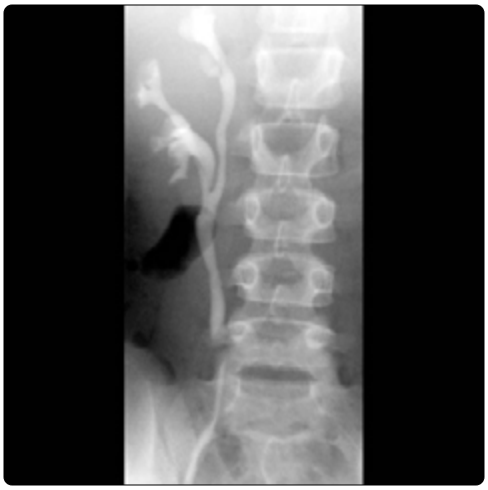
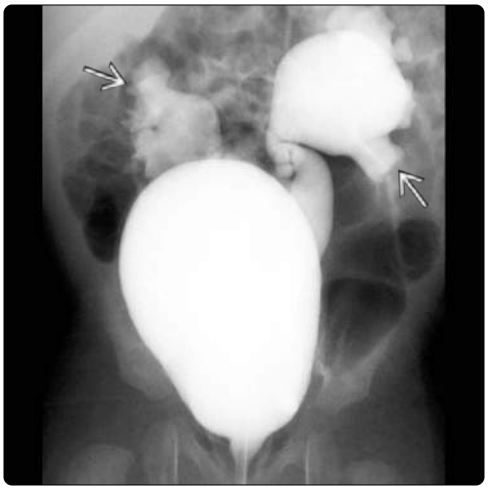


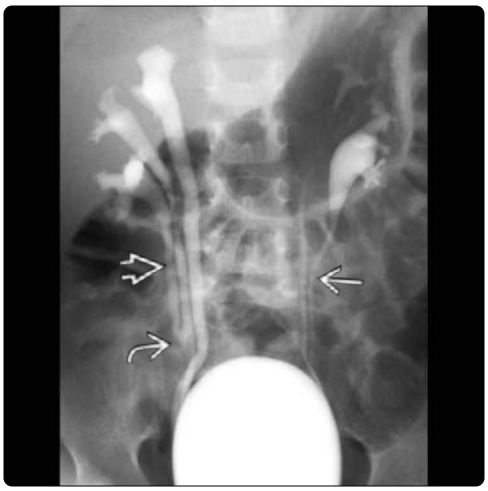


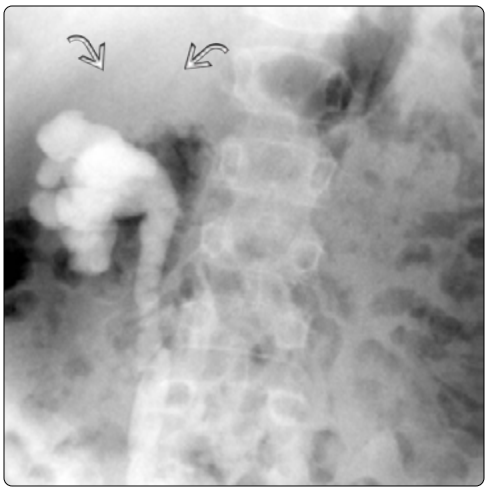
(Left) *VCUG shows high-grade VUR on the left side associated with a lobulated filling defect in the bladder base, corresponding to a refluxing ureterocele.* **(Right)** *Prone posterior US in twin view shows the anechoic renal pelvis on the left image & swirling echogenic urine refluxing into the pelvis on the right image, which used Doppler SMI to detect slow velocity flow. A similar appearance would be expected on a contrast-enhanced cystosonography.*





(Left) *VCUG shows VUR into a partially duplicated collecting system, which resembles the letter "y". VUR can sometimes be seen fluoroscopically entering one limb of the "y" 1st, receding, & then filling the other limb, the so-called "yo-yo" reflux.* **(Right)** *VCUG shows large-volume VUR into both kidneys, which have very dilated collecting systems. Grade V reflux was diagnosed in this infant with prenatal hydronephrosis.*





(Left) *Frontal VCUG shows high-grade VUR into a triplicated ureter on the right plus lower grade reflux into a duplicated ureter on the left. Two of the 3 right ureters join inferiorly.* **(Right)** *VCUG shows grade IV VUR into the lower pole of a duplicated right kidney. Note that no upper pole calyces are seen, & there are 10 calyces visualized. Also, the long axis points abnormally to the ipsilateral shoulder. US can confirm a duplication as the cause for this "drooping lily" rather than a cyst or mass.*

Ureteropelvic Duplications

KEY FACTS

TERMINOLOGY

- Presence of 2 separate pelvicalyceal collecting systems in 1 kidney; 2 draining ureters may
  - Join above bladder: Partial duplication (most common, often of no consequence)
  - Insert into bladder separately: Complete duplication

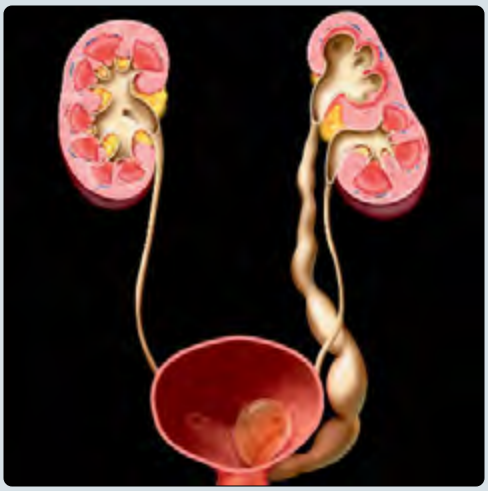
IMAGING

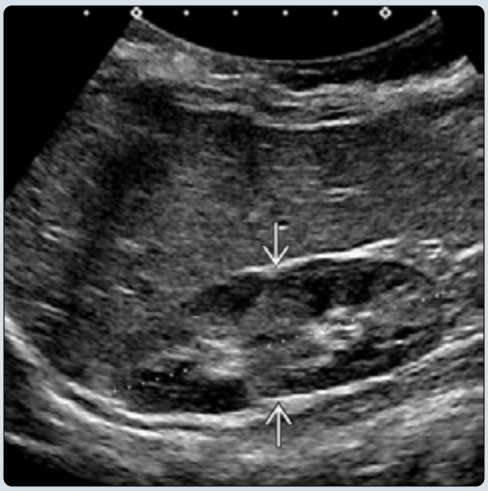
- Central renal sinus fat separated by bar of cortical tissue
  - Renal parenchyma otherwise normal unless complications of VUR &/or obstruction present
- Separate renal pelves often visible on either side of this tissue, ± separate proximal ureters
- Duplicated kidneys tend to be larger than nonduplex kidneys, even without hydronephrosis
- With complete duplication, ureter draining upper pole of kidney inserts in bladder inferior & medial to ureter draining lower pole of kidney (Weigert-Meyer rule)
  - Lower pole ureter inserts orthotopically in trigone
  - Upper pole ureteral orifice ectopic in location & often associated with ureterocele
- Corollary to Weigert-Meyer rule
  - Upper pole tends to obstruct
  - Lower pole tends to have vesicoureteral reflux (VUR)
    - Lack of dilation of ureter & collecting system in no way excludes VUR
- Drooping lily sign: Classic appearance of opacified lower pole collecting system on IVP or VCUG, displaced by mass-like upper pole hydronephrosis
  - Correlation with US critical to confirm upper pole obstruction rather than other mass lesion

CLINICAL ISSUES

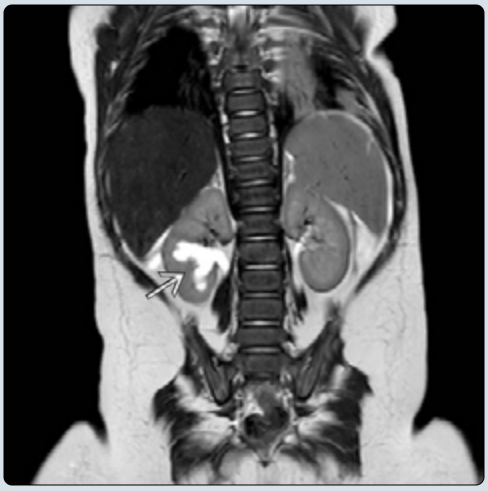
- Incidence of 12-15% in general population
- Treatment depends on extent of anomalies & complications

(Left) *Coronal graphic shows a normal right kidney & completely duplicated left kidney with a poorly draining upper pole ectopic ureterocele seen in the bladder medial & inferior to the lower pole ureteral orifice. The lower pole ureter on the left inserts into bladder orthotopically at the trigone.* **(Right)** *Longitudinal US in a 1 year old being evaluated for fever shows an uncomplicated duplicated right kidney with a band of cortex ⇒ obliquely crossing the central sinus structures.*





(Left) *Frontal VCUG image in a patient with a duplicated right kidney shows grade 4 vesicoureteral reflux into the lower pole moiety ⇒. Upper pole moieties tend to obstruct & lower poles tend to reflux.* **(Right)** *Coronal T2 MR in a 4 month old shows a duplicated right kidney with moderate pelvocaliectasis affecting only the lower pole ⇒. The left kidney is not duplicated.*



TERMINOLOGY

Synonyms

- Duplicated kidney, duplex collecting system, partial/incomplete/complete duplication, bifid pelvis

Definitions

- Presence of 2 separate pelvicalyceal collecting systems in 1 kidney
  - 2 draining ureters may
    - Join above bladder (partial duplication)
    - Insert into bladder separately (complete duplication)

IMAGING

General Features

- Best diagnostic clue
  - Identification of 2 renal pelves or proximal ureters on any imaging modality
- Location
  - Duplication can involve any length of urinary tract (from renal parenchyma to urethra)
  - Left > right
- Size
  - Duplicated kidneys tend to be larger than nonduplex, even without hydronephrosis
- Morphology
  - Central renal sinus fat completely separated by bar of renal cortex; renal parenchyma often otherwise normal
    - Unless vesicoureteral reflux (VUR) has led to scarring or obstruction has led to thinning/dysplasia
  - Duplex system has > 10 calyces, 2 renal pelves, & 1 or 2 ureters
  - > 1 renal artery & vein very common
  - If duplication complete, ureter draining upper portion of kidney inserts in bladder inferior & medial to ureter draining lower segment of kidney (Weigert-Meyer rule)
  - Corollary to Weigert-Meyer rule
    - Upper pole tends to obstruct
    - Lower pole tends to have VUR
  - Upper pole ureteral orifice is, by definition, ectopic in location & may be associated with ureterocele

Fluoroscopic Findings

- Voiding cystourethrogram
  - Look for VUR (lower pole > > upper pole)
  - Saddle VUR or yo-yo VUR unique to partial duplications, which have single distal ureter
    - VUR contrast first enters 1 component of upper tract collecting system, drains, & then refluxes into 2nd component
  - Ectopic ureter may only be visualized when inadvertently catheterized (e.g., intersphincteric ureter)
    - If this occurs, contrast opacification can help delineate anatomy
    - However, catheterization of urinary bladder must also occur to look for VUR into other ureters

Ultrasonographic Findings

- Grayscale ultrasound
  - Band of renal cortex splits echogenic sinus fat
  - Separate renal pelves often visible, ± separate proximal ureters
  - Distal ureters more difficult to identify due to bowel gas near bladder
    - While dilated distal ureter may be suggestive of VUR, remember that
      - Lack of dilation of ureter & collecting system in no way excludes VUR
      - Severe VUR may not show dilation at US
  - Look for thin-walled ureterocele protruding into urinary bladder, assess for presence & location of ureteral jets
  - Remember to survey for concomitant genital anomalies, especially of uterus
- Color Doppler
  - May show elevated resistive indices (RIs) in obstructed moiety
  - Useful to define arterial anatomy
  - Used to confirm lack of blood flow in renal pelves & tubular ureters
  - Can show foci of hypoperfusion from superimposed infection (pyelonephritis)

CT Findings

- CECT can demonstrate course of ureters (particularly with delay) & locate renal arteries
- Presence of supernumerary calyces & bifid pelvis can be more challenging to detect on axial imaging; reformats helpful

MR Findings

- MR urography useful to show fluid within collecting systems
- Often best test when ectopic ureter cannot be demonstrated with other imaging
- Coronal thin or thick slab series may be most useful

Nuclear Medicine Findings

- Occasionally, nuclear study can be 1st to suggest renal duplication
- Renal scans used to show differential function, drainage, & scarring
- Results help guide surgical intervention

Other Modality Findings

- Obstructed hydronephrotic upper pole moiety of duplicated system exerts mass effect on lower pole moiety, displacing/rotating it inferiorly
  - Drooping lily sign: Classic appearance of inferiorly displaced/rotated opacified lower pole collecting system on IVP or VCUG
  - Can be seen on coronal CT, MR, & US
  - Beware that true suprarenal masses can also create this fluoroscopic appearance, necessitating cross-sectional investigation (typically US)

Imaging Recommendations

- Best imaging tool
  - US usually 1st-line study for suspected GU abnormality
  - VCUG & nuclear studies generally complete imaging work-up if duplicated system causing problems
  - MR useful in complex cases; can provide complete anatomic & physiologic work-up
- Protocol advice

- When searching for ectopic ureter from poorly functioning &/or chronically obstructed upper pole, consider MR urography

DIFFERENTIAL DIAGNOSIS

Column of Bertin

- Normal variant of junctional parenchyma, typically in mid kidney; looks like focally thickened cortex

Segmental Multicystic Dysplastic Kidney

- Upper pole multicystic dysplastic kidney of duplicated system can mimic obstructed, hydronephrotic upper pole moiety

Adrenal Mass

- Can mimic drooping lily sign by displacing collecting system inferiorly

PATHOLOGY

General Features

- Etiology
  - Early branching of ureteric bud or 2 ureteral buds arising from wolffian duct
  - Each bud induces formation of its own nephrons when it meets metanephric blastema
  - Abnormal branching of ureteric bud can also give rise to supernumerary kidney or triplicate collecting system (both very rare)
- Associated abnormalities
  - Duplications of bladder, urethra, & genital structures associated with renal duplication
    - Genital anomalies in 1/2 of affected females
  - Ureteropelvic junction obstruction more common in duplicated kidneys

Gross Pathologic & Surgical Features

- Almost always has 2 renal arteries & veins, often with separate renal artery orifice from aorta
- In absence of complications, renal parenchyma & collecting system tissue normal

Microscopic Features

- Depends on complications: Scarring, hydronephrosis, fibrosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Most often discovered antenatally or incidentally on imaging studies performed for other reasons
- Other signs/symptoms
  - Symptomatic duplications may lead to infection, obstruction, calculi, scarring, hematuria, abdominal or flank pain, voiding dysfunction, urinary retention

Demographics

- Age
  - Congenital; usually discovered early in life
- Gender
  - Complete duplications much more common in women
  - Partial duplications have no gender predilection
- Epidemiology
  - Incidence of 12-15% in general population, but only 1% found de novo in cadaveric renal donors

Natural History & Prognosis

- Prognosis varies with type of duplication & severity of complications
- Chronic obstruction or VUR, infection, &/or scarring may lead to secondary hypertension & renal insufficiency

Treatment

- Depends on extent of anomalies & complications
  - Associated ureteroceles incised, unroofed, resected, or reimplanted
  - Ectopic ureter may be reimplanted if kidney it subtends retains good function; otherwise resected
  - Hydronephrosis treated surgically to improve drainage
    - Cortical thinning & relative function used to determine surgical course: Salvage vs. resection
  - Infections treated with antibiotics & evaluated for urine stasis & VUR
  - VUR may be managed conservatively (medically) or treated surgically, depending on grade & associated anomalies
  - Calculi removed, fragmented, or monitored
  - Voiding dysfunction assessed to exclude ectopic ureter/ureterocele, prolapsing cecoureterocele, & bladder dyskinesia

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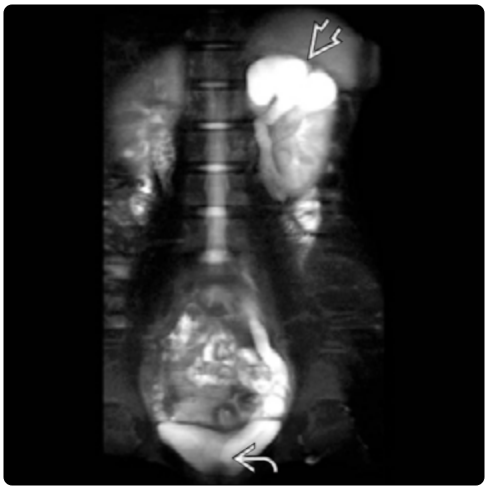
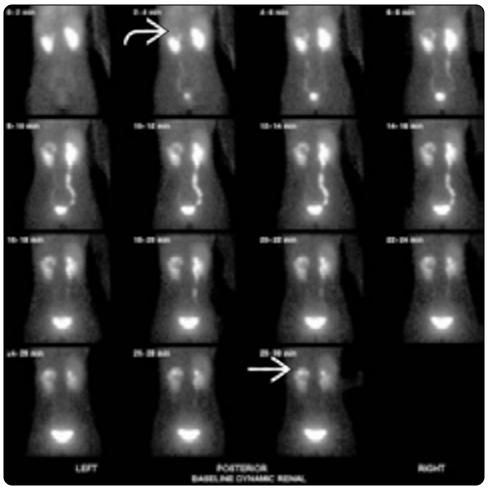
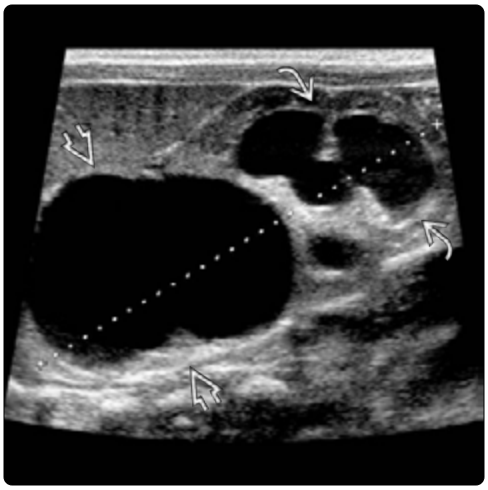
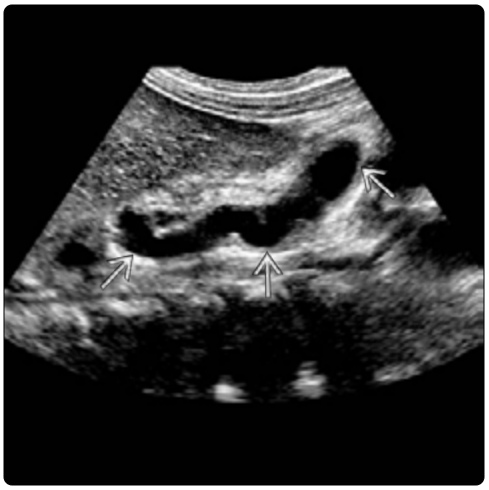


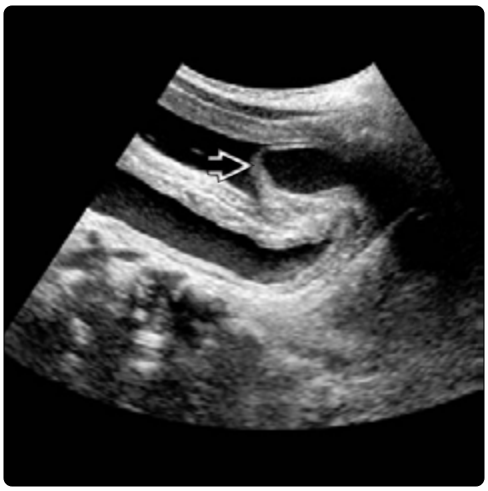


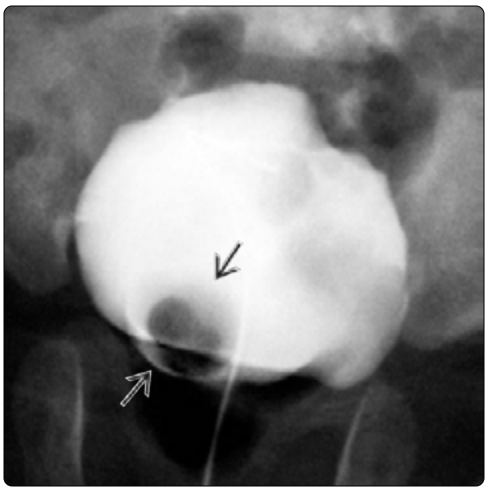
(Left) *Coronal MIP MR urogram in a teenager shows a duplicated left kidney & obstructed upper pole moiety, which drains ectopically into the vagina. Note the cortical thinning of the left upper pole from chronic injury.* **(Right)** *Posterior images from a Tc-99m MAG3 renogram in the same patient show delayed uptake by the upper pole of the left kidney with poor function & excretion. The postdiuretic washout curve for the left upper pole was flat (not shown).*





(Left) *Longitudinal oblique US in a patient with a duplicated right kidney shows severe dilation of the upper pole moiety with marked parenchymal thinning. There is also mild to moderate dilation of the lower pole moiety. Cursors mark the length of the kidney.* **(Right)** *Longitudinal oblique US in the same patient shows the tortuous course of the dilated right upper pole ureter.*





(Left) *Longitudinal oblique US in the same patient follows the right upper pole dilated ureter into the bladder base, where the ureter ends in an ectopic ureterocele. This patient has a completely duplicated right kidney with an obstructed upper pole moiety.* **(Right)** *Frontal voiding cystourethrogram (VCUG) shows a round filling defect in the bladder base from an ectopic ureterocele. Ureteroceles can be very large & occasionally prolapse during voiding, causing bladder outlet obstruction.*

Ureterocele

KEY FACTS

TERMINOLOGY

- Congenital cystic dilation of distal submucosal portion of 1 or both ureters within urinary bladder
- Categorized according to ureterocele position
  - Intravesical: Completely contained within bladder
  - Extravesical: Partially extends to bladder neck, urethra, or perineum
- Categorized according to ureterocele insertion
  - Orthotopic (simple): Orifice located in normal anatomic position in bladder trigone
  - Ectopic: Orifice located anywhere else
- Categorized according to type of kidney drained
  - Single system vs. duplicated system with 2 ureters
  - 1 ureter of duplicated system must be ectopic
- Weigert-Meyer rule: Ureter from upper pole (UP) moiety of duplicated kidney inserts inferior & medial to normal lower pole (LP) moiety insertion site at trigone
- Any ureterocele may obstruct & cause hydronephrosis

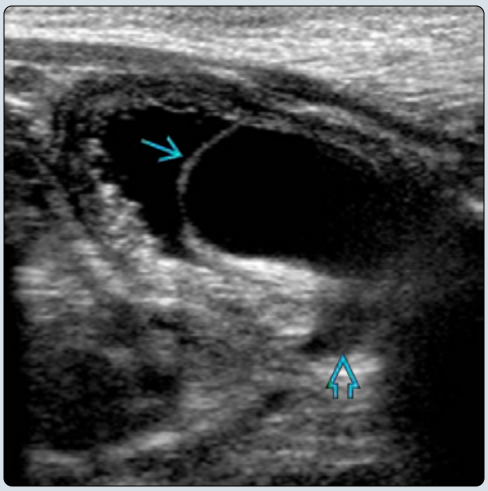
IMAGING

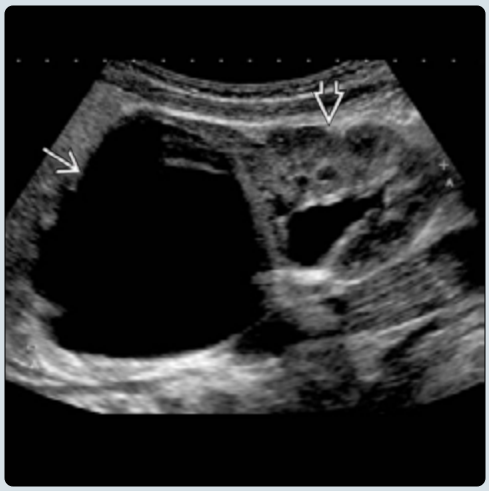
- Round/ovoid filling defect in urinary bladder
  - Thin walled & cystic-appearing on US, MRU
    - Wall appears thicker, collapsed after surgery
- ± visualization of associated dilated distal ureter
- In duplicated system, UP moiety (associated with ureterocele) typically obstructs & LP moiety typically refluxes
  - Varying degrees of hydronephrosis & parenchymal dysplasia of UP moiety
  - Vesicoureteral reflux into LP moiety classically shows drooping lily sign
    - Due to rotation of LP system by obstructed UP

CLINICAL ISSUES

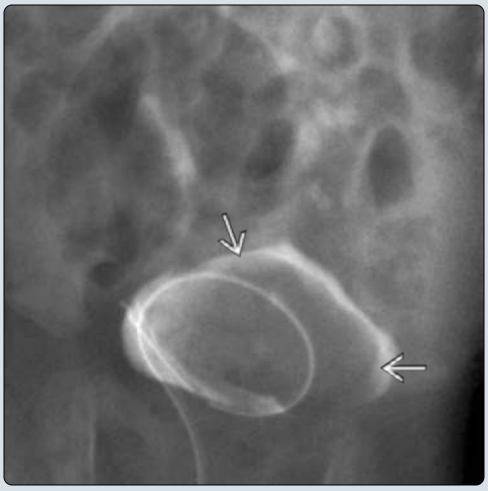
- Ectopic, extravesical variety > orthotopic, simple, intravesical variety by 3:1 ratio
- Typical treatment: Endoscopic incision of ureterocele, especially if infected or obstructed in neonate

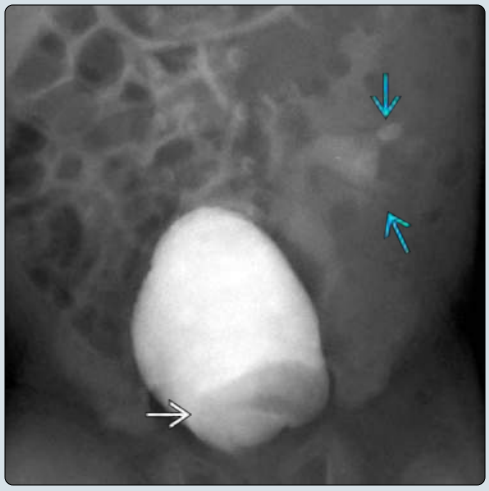
(Left) *Longitudinal ultrasound of the urinary bladder in an 18 day old with prenatally detected hydronephrosis shows a large thin-walled cyst ⇨ filling much of the bladder lumen, typical of a ureterocele. The cyst connects to a dilated distal left ureter ⇨.* **(Right)** *Longitudinal ultrasound of the duplicated left kidney in the same patient shows severe pelvocaliectasis & parenchymal thinning of the left upper pole (UP) moiety ➡. The lower pole (LP) moiety shows mild pelvocaliectasis with more normal-appearing renal parenchyma ➡.*





(Left) *Frontal early filling image from a VCUG in the same patient shows the ureterocele as a large ovoid filling defect ➡ partially outlined by contrast in the urinary bladder.* **(Right)** *Postvoid VCUG in the same patient shows grade 2-3 vesicoureteral reflux into the LP collecting system ⇨. The drooping lily configuration of contrast within the LP strongly suggests obstruction of an UP moiety (as confirmed on the ultrasound). The UP ureterocele ➡ remains visible in the contrast-filled bladder.*





TERMINOLOGY

Definitions

- Congenital cystic dilation of distal submucosal portion of 1 or both ureters within urinary bladder
- Categorized according to ureterocele position
  - Intravesical: Completely contained within urinary bladder
  - Extravesical: Partially extends into bladder neck, urethra, or perineum
- Categorized according to ureterocele insertion
  - Orthotopic (simple): Orifice located in normal anatomic position in bladder trigone
  - Ectopic: Orifice located anywhere else
    - Ectopic ureter may drain to other genitourinary locations without ending as ureterocele
- Categorized according to type of kidney drained
  - Single system ureterocele: Kidney gives rise to single collecting system with solitary ureter
    - Typically simple, intravesical ureterocele
  - Duplex system ureterocele: Kidney has completely duplicated collecting system with 2 complete ureters
    - 1 ureteral insertion ectopic by definition (often extravesical ureterocele)
- Cecoureterocele: Unique subtype elongated beyond ureteral orifice by tunneling under trigone & urethra
- Any type of ureterocele may become obstructed & cause hydronephrosis

IMAGING

General Features

- Best diagnostic clue
  - Round or ovoid cystic filling defect in bladder
- Location
  - Left > right; bilateral in 10%
  - Ectopic, extravesical variety > orthotopic, simple, intravesical variety by 3:1 ratio
  - Weigert-Meyer rule: Ureter from upper pole (UP) moiety of duplicated kidney inserts inferior & medial to normal insertion site of lower pole (LP) moiety in bladder trigone
    - UP moiety typically obstructs, LP typically refluxes
  - Ureterocele may prolapse in & out of bladder

Radiographic Findings

- IVP
  - Elongated cobra head filling defect in contrast-filled bladder
  - Possible delayed/diminished function of UP moiety

Fluoroscopic Findings

- Voiding cystourethrogram (VCUG)
  - Best seen on early bladder-filling image before contrast too dense & intravesical pressure compresses ureterocele
  - Vesicoureteral reflux (VUR) frequently seen in LP moiety
    - May create drooping lily sign where nonopacified obstructed UP moiety distorts opacified refluxing LP moiety
      - Long axis of opacified LP moiety collecting system points toward ipsilateral shoulder
  - If ureterocele everts, creates appearance of diverticulum & may cause VUR

Ultrasonographic Findings

- Grayscale ultrasound
  - Anechoic, thin-walled cyst inside urinary bladder
  - Ureteral jet & connection to dilated distal ureter may be demonstrated
  - ± duplicated kidney with hydroureteronephrosis
    - Single system kidney typically shows uniform degree of pelvocaliectasis
    - Obstructed UP moiety of duplex kidney variable in morphology
      - Often shows worse pelvocaliectasis than LP
      - Variable degrees of UP parenchymal thinning ± cystic dysplasia
      - Variable size of UP moiety: Enlarged with severe pelvocaliectasis vs. small with dysplastic parenchyma in setting of large ureterocele (ureterocele disproportion)
  - After incision (to relieve obstruction), residual partially or totally collapsed ureterocele often has thick undulating wall
- Color Doppler
  - Ureteral jet useful to exclude complete obstruction of ureterocele

MR Findings

- T2WI
  - MR urogram (MRU) sequences useful to detect poorly functioning UP moiety & draining ureter
  - Gynecologic anomalies in 50% of females with duplication

Nuclear Medicine Findings

- Nuclear cystogram
  - Ureterocele difficult to see on nuclear cystogram unless very large or prolapsing
- Nuclear renal scan
  - Used to assess function of obstructed or poorly functioning UP moiety
  - Poor function on nuclear scan predictive of severe histologic changes in renal parenchyma; helps justify heminephrectomy

Imaging Recommendations

- Best imaging tool
  - VCUG & ultrasound best 1st-line studies
  - Nuclear renal scan most established technique for evaluating differential function in duplicated kidney
  - MRU & IVP reserved for difficult or complex cases
    - Increasing use of MRU as comprehensive study of anatomy & function
- Protocol advice
  - VCUG best test to assess dynamic nature of ureterocele: Everting, refluxing, prolapsing, &/or causing bladder outlet obstruction

DIFFERENTIAL DIAGNOSIS

Bladder Mass

- Lacks communication with distal ureter; typically solid
  - Rhabdomyosarcoma, neurofibroma, polyp: Nonmobile with variable internal vascularity
  - Focal cystitis: Variety of infectious & noninfectious causes

- Fungus ball, hematoma: Often mobile; no internal vascularity

Mass Effect From Sigmoid Colon

- Can appear as filling defect on VCUG: Get oblique views

Bladder Hutch Diverticulum

- Periureteral diverticulum; separate from distal ureter
- May mimic everted ureterocele; does not usually prolapse into bladder to cause filling defect

Ovarian Cyst

- May deform but does not typically bulge into bladder

PATHOLOGY

General Features

- Etiology
  - Embryology/anatomy
    - Thought to result from delayed canalization of Chwalla membrane during embryogenesis → obstruction of ureteral orifice
      - Chwalla membrane: Primitive separation of ureteral bud from developing urogenital sinus
- Associated abnormalities
  - VUR into lower pole of duplicated system: 50%
  - VUR in contralateral kidney: 25%

Microscopic Features

- In heminephroureterectomy specimens, histologic changes include chronic interstitial inflammation, fibrosis, tubular atrophy, glomerulosclerosis, & dysplasia
- Normal urothelium in dilated intramucosal or submucosal segment

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Prenatal detection of hydronephrosis typical
  - Febrile UTI most common postnatal presentation
- Other signs/symptoms
  - Simple ureterocele may not be diagnosed until adulthood
  - Ectopic ureterocele usually diagnosed in infancy or shortly after toilet training: Presents with hematuria, UTI, chronic enuresis, or hydronephrosis
  - Rarely presents with prolapse & acute bladder outlet obstruction
  - Other rare presentations: Failure to thrive, cyclic abdominal pain, ureteral calculus

Demographics

- Age
  - Most often detected antenatally
- Gender
  - Ectopic ureterocele F:M = 4-7:1
- Epidemiology
  - ~ 1/4,000 children; less frequently diagnosed in adults

Natural History & Prognosis

- Prognosis excellent if nonobstructing & nonrefluxing
- Prognosis variable if prolonged obstruction or high-grade VUR has compromised renal function
- Antenatal diagnosis reported to improve overall course: Fewer infections, fewer surgical procedures
- Ureteroceles diagnosed antenatally should be treated surgically within 1st weeks of life since rate of UTI exceeds 50% despite prophylactic antibiotics

Treatment

- Endoscopic incision (puncture or unroofing) of ureterocele, especially if infected or obstructed in neonate
  - Following incision, ureterocele wall appears thickened, irregular, even mass-like
  - Endoscopic incision may convert obstructed ureterocele into refluxing ureterocele
- Ureteral reimplantation surgery: Extravesical reimplantation, ureteroureterostomy, ureteropyelostomy
  - Patients undergoing bilateral ectopic ureterocele repair at ↑ risk for postoperative voiding dysfunction
    - Unclear if risk present preoperatively or due to trigonal surgery
- Heminephroureterectomy if UP moiety very poorly functioning
- Antenatal laser ablation of ureteroceles has been performed in fetuses with severe bilateral hydronephrosis, bladder outlet obstruction, & oligohydramnios
- Success after single procedure ↑ in single ureters; many require > 1 surgical procedure
- Best surgical management remains controversial

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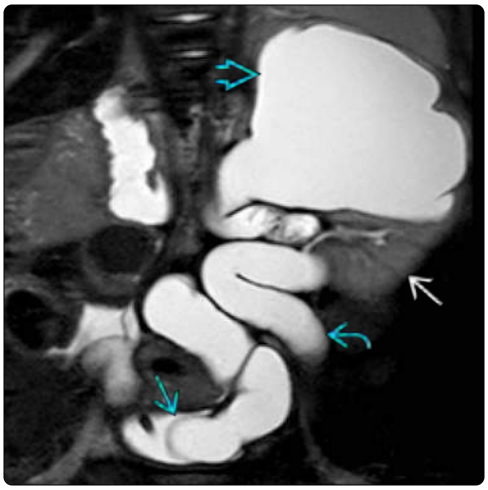
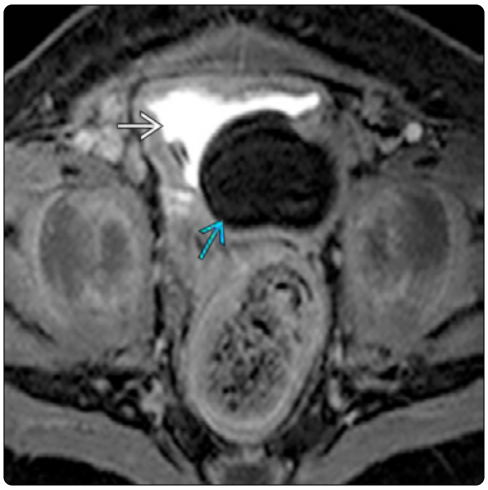
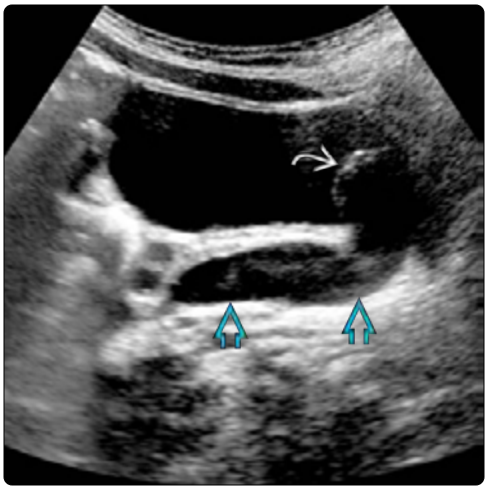
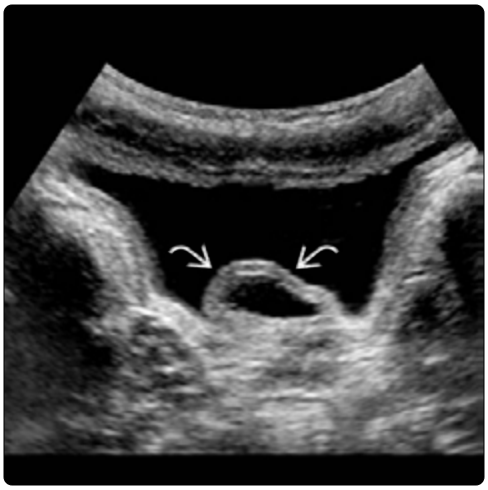


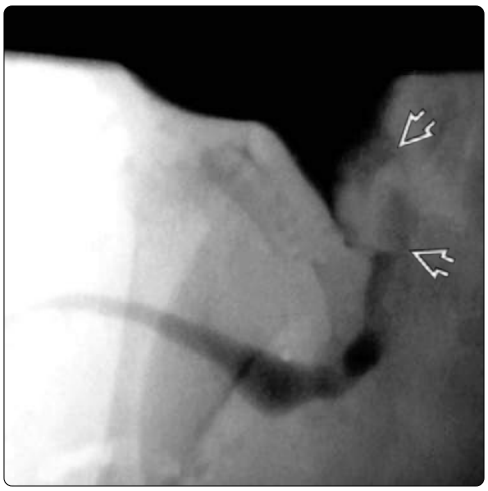


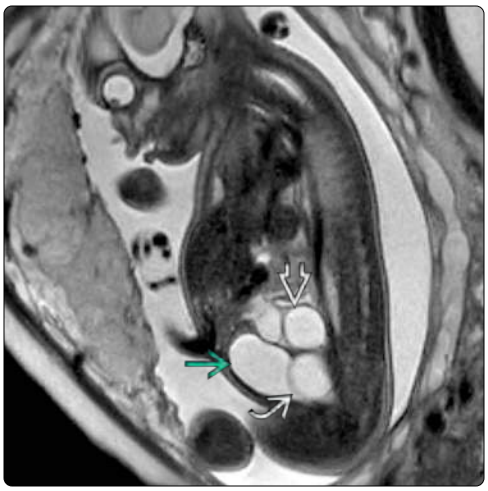
(Left) *Coronal T2 FS MR in an infant shows an ectopic ureterocele partially filling the urinary bladder & connecting to a markedly dilated UP ureter. There is severe UP hydronephrosis due to obstruction. Note the normal LP moiety.* **(Right)** *Delayed axial T1 C+ FS MR in the same patient shows no contrast in the ureterocele due to the long-standing UP obstruction with decreased function. Contrast has filled the surrounding bladder by excretion from the remaining urinary tract. (Courtesy J.R. Dillman, MD.)*





(Left) *Longitudinal ultrasound shows a dilated distal ureter entering the bladder & forming a thin-walled bulbous end, typical of a ureterocele. This ureterocele is inserting low in the bladder base & is likely ectopic.* **(Right)** *Transverse ultrasound through the bladder base in a patient after a ureterocele incision shows a partially collapsed, relatively thick-walled ureterocele. After endoscopic incision, ureteroceles tend to decompress & may show wall thickening with an undulating contour, as in this case.*





(Left) *Oblique voiding cystourethrogram shows prolapse of a ureterocele into the posterior urethra as voiding starts & the catheter is expelled. Voiding stopped seconds later due to urethral obstruction.* **(Right)** *Sagittal T2 SSFSE MR from a fetal MR shows a thin-walled ureterocele within the urinary bladder with a tortuous & dilated ureter superiorly.*

Primary Megaureter

KEY FACTS

TERMINOLOGY

- Megaureter: General term for ureteral dilation
  - Can be due to vesicoureteral reflux (VUR), obstruction, both, or neither
- Primary obstructive megaureter: Functional obstruction at juxtavesical segment of ureter due to absent peristalsis

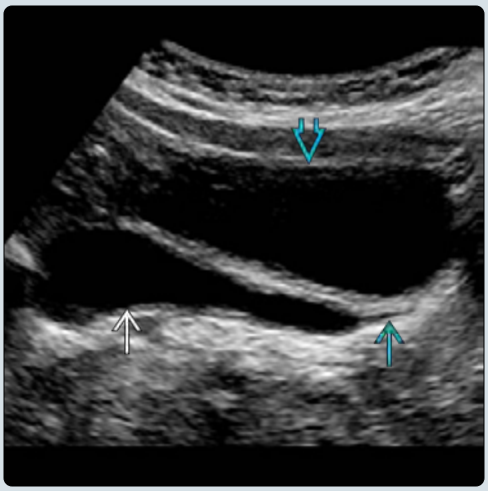
IMAGING

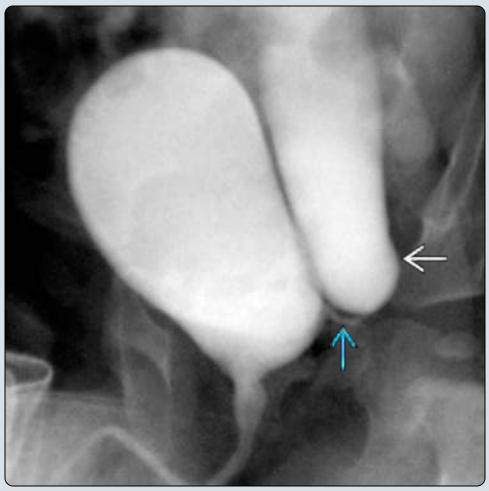
- Best imaging clues
  - Variable degree of hydroureteronephrosis with transition to nondilated distal ureter
    - Nondilated aperistaltic segment involves terminal 0.5-4 cm of ureter
    - Most commonly unilateral: 67% on left
  - VUR: Ipsilateral in 5%, contralateral in < 10%
  - ± debris, calculi in dilated ureter
  - Long-term obstruction can lead to parenchymal thinning & lack of renal growth
- Best imaging modalities
  - Ultrasound best screening exam for urinary tract anomalies (pre- or postnatally)
  - Diagnostic & treatment considerations refined with
    - Fluoroscopic VCUG (to assess for VUR &/or bladder outlet obstruction)
    - Nuclear medicine diuretic renography to assess for renal function & delayed ureteral drainage
    - MR urography can assess anatomy & physiology

CLINICAL ISSUES

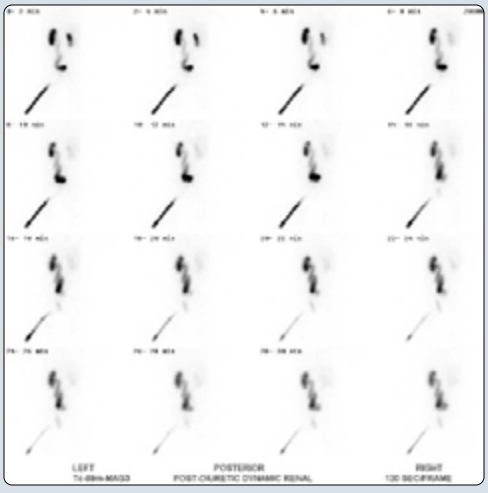
- M:F = 4:1
- Diagnosed prenatally in nearly 50%; remaining cases present over wide range of ages
- May be asymptomatic but can present with UTI, abdominal/flank pain, hematuria ± calculi, renal failure
- 70% regress spontaneously by 7 years of age
- Surgery considered for worsening renal function or recurrent UTIs

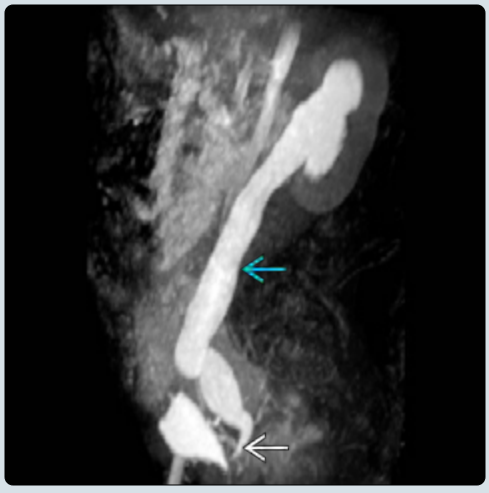
(Left) *Longitudinal ultrasound at the left ureterovesical junction (UVJ) of a male infant shows a dilated left ureter ➡ tapering distally to a normal caliber aperistaltic juxtavesical segment ⇨ that shows urothelial thickening. The urinary bladder ⇨ is seen anteriorly.* **(Right)** *Oblique fluoroscopic image from a voiding cystourethrogram in the same patient shows vesicoureteral reflux into the normal caliber juxtavesical segment ⇨ with transition to the proximally dilated ureter ➡.*





(Left) *Dynamic posterior view postdiuretic images from a Tc-99m MAG3 renogram in the same patient show qualitatively slow drainage of the left dilated ureter. The T1/2 for drainage of the left ureter was > 20 minutes on the quantitative evaluation, consistent with obstruction.* **(Right)** *Oblique MIP from an MR urogram in a patient with a primary obstructive megaureter shows a narrow juxtavesical segment of the left ureter ➡ with proximal hydroureteronephrosis ⇨.*





TERMINOLOGY

Definitions

- Megaureter: Ureteral diameter ≥ 7 mm
- 4 types classified by presence of vesicoureteral reflux (VUR) &/or obstruction
  - Nonrefluxing obstructed (NR, O)
  - Refluxing obstructed (R, O)
  - Refluxing nonobstructed (R, NO)
  - Nonrefluxing nonobstructed (NR, NO)
- Primary obstructive megaureter: Congenital functional obstruction at juxtavesical segment of ureter
  - Presumably due to absent peristalsis in normal caliber distal segment, though exact etiology unclear

IMAGING

General Features

- Best diagnostic clue
  - Hydroureteronephrosis (HUN) with short segment of normal caliber distal ureter
- Location
  - Transition from normal caliber distal ureter to proximal dilation 0.5-4 cm above ureterovesical junction
  - 60% unilateral (of which 67% involve left ureter)

Ultrasonographic Findings

- Grayscale ultrasound
  - Unilateral or bilateral HUN
  - ± renal parenchymal thinning & lack of renal growth
  - Hyperperistalsis of dilated ureter up to distal aperistaltic normal caliber segment
  - ± intraluminal debris (proteinaceous or infectious)
  - ± calculus formation

Fluoroscopic Findings

- Voiding cystourethrogram
  - If no VUR, study may appear normal
  - Ipsilateral VUR in 5%, contralateral in < 10%
  - When VUR present, assess for obstruction
    - Delayed drainage of dilated ureter
    - Short segment of normal caliber distal ureter
- Retrograde ureterogram (by urology service)
  - More reliably demonstrates distal adynamic ureter

Nuclear Medicine Findings

- Diuretic renography (Tc-99m MAG3 or DTPA)
  - Delayed clearance of radiotracer from dilated ureter & collecting system after furosemide administration
    - Prolonged ureteral ± renal T-1/2 (> 20 minutes)

MR Findings

- MR urography
  - Comprehensive anatomic & functional evaluation
    - Can assess renal parenchymal function & urinary clearance, similar to nuclear medicine renal scan

DIFFERENTIAL DIAGNOSIS

Refluxing Nonobstructive Megaureter

- Usually high-grade VUR; drains into bladder at end of VCUG
- Much more common than primary obstructive form
- May be idiopathic or seen with neurogenic bladder, posterior urethral valves, prune-belly syndrome, megacystis-megaureter, duplicated ureters, incised ureterocele

Nonrefluxing Nonobstructive Megaureter

- Idiopathic aperistaltic distal ureter may occur without quantitative obstruction
- Polyuria (diabetes insipidus) may cause general ureteral dilation

Mechanical Obstruction

- Unilateral (ureteral obstruction)
  - Stricture, ureterocele, urolithiasis, retrocaval ureter, extrinsic mass, ureteral valve (rare)
- Bilateral (bladder outlet or urethral obstruction)
  - Bladder outlet: Posterior urethral valves, cloaca with hydrocolpos, extrinsic mass
  - Urethral: Stricture, polyp, anterior valve, atresia, primary megalourethra
  - Unclear: Prune-belly syndrome

PATHOLOGY

General Features

- Etiology unclear: Possibilities include ↑ collagen deposition, muscle hypertrophy, ↓ number of interstitial cells of Cajal

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic vs. febrile UTIs, abdominal/flank pain, hematuria ± calculi, gradual renal failure

Natural History & Prognosis

- 70% resolve spontaneously by 7 years of age
  - Initial ureteral diameter < 8.5 mm: Likely to resolve
  - Initial ureteral diameter > 15 mm: Likely to persist
- May develop renal dysfunction or recurrent UTIs

Treatment

- Conservative management
  - Prophylactic antibiotics; imaging every 6-12 months
- Surgical management: Temporary double J stent, various ureteral reimplantation options
  - Consider with worsening renal function or recurrent UTIs

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Megaureter-Megacystis

KEY FACTS

TERMINOLOGY

- Megaureter-megacystis syndrome (MMS): Marked vesicoureteral reflux (VUR) leads to large urinary bladder due to repetitive recycling of urine
  - Bladder dysfunction develops gradually

IMAGING

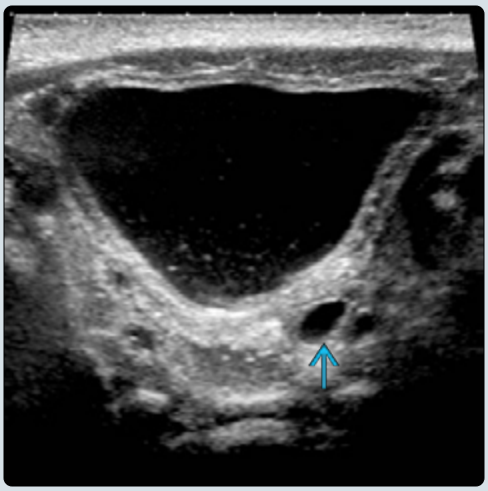
- On voiding cystourethrogram (VCUG)
  - Large urinary bladder with smooth, thin wall
    - > 2x estimated normal bladder capacity
  - Unilateral or bilateral high-grade VUR
    - VUR may occur (during filling &/or voiding) into single system or lower pole of duplex system
  - Voiding demonstrates complete bladder emptying
  - However, bladder rapidly refills (almost to capacity) by drainage of retained refluxed contrast from dilated ureters & collecting systems
    - Termed aberrant micturition, though bladder neck & urethra normal

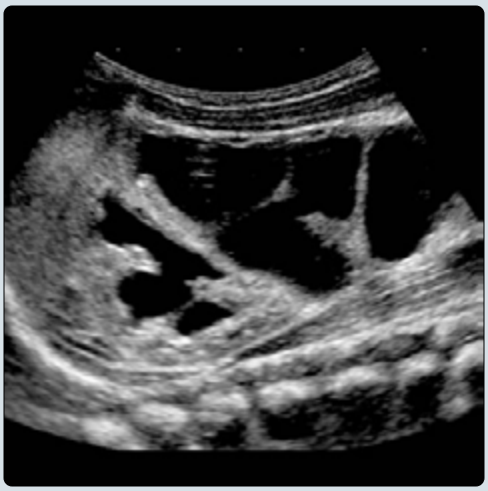
TOP DIFFERENTIAL DIAGNOSES

- Mechanical bladder outlet obstruction
  - Posterior urethral valves
  - Extrinsic/intrinsic mass
  - Obstructing ureterocele
- Functional bladder outlet obstruction
  - Neurogenic bladder
- Megacystis of unclear etiology
  - Prune-belly (Eagle-Barrett) syndrome
  - Megacystis-microcolon-hypoperistalsis syndrome
  - Megalourethra

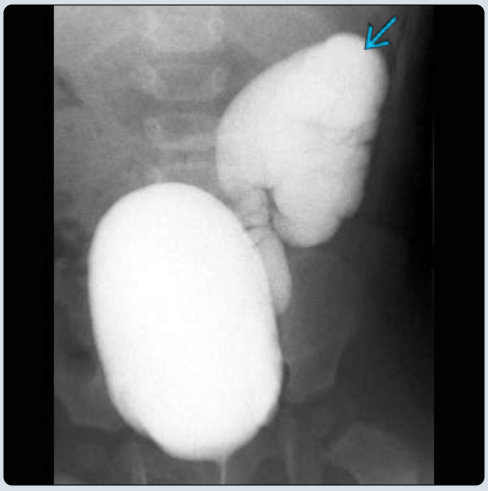
CLINICAL ISSUES

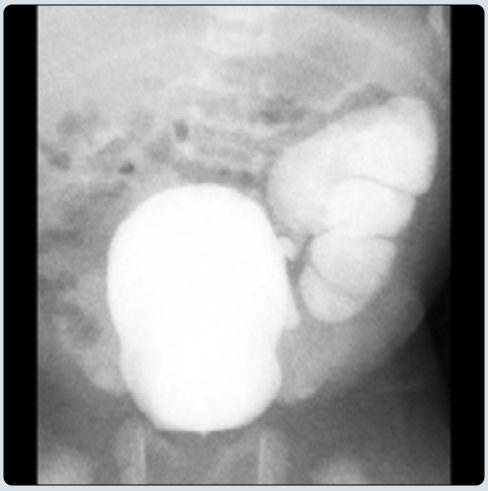
- Most commonly presents with urinary tract infections or abnormal prenatal ultrasound
- Bladder function may deteriorate if VUR not improving
- With spontaneous resolution or surgical cessation of VUR, bladder function usually improves





(Left) *Transverse ultrasound in a 3-day-old boy with a history of prenatal hydronephrosis shows a large thin-walled bladder & left hydroureter ⇨ with urothelial thickening.* **(Right)** *Longitudinal ultrasound of the left kidney in the same patient shows asymmetric upper pole (UP) vs. lower pole (LP) pelvocaliectasis & cortical thinning, suggesting collecting system duplication. As no ureterocele was seen in the bladder, this finding suggests an obstructed ectopic UP ureter & high-grade LP vesicoureteral reflux (VUR).*





(Left) *Frontal VCUG in the same patient shows grade V left lower pole VUR ⇨ with a large bladder. (The orientation of the visualized collecting system long axis towards the ipsilateral shoulder suggests duplication.) The bladder capacity was > 2x the estimated normal capacity.* **(Right)** *Frontal VCUG image hold in the same patient after complete voiding shows immediate bladder refilling by the drainage of refluxed contrast, typical of megaureter-megacystis syndrome (MMS).*

KEY FACTS

TERMINOLOGY

- Megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS): Rare disease of smooth muscle dysfunction resulting in poor motility of GI & GU tracts
- Synonyms: Berdon syndrome (described by Berdon et al in 1976) & familial visceral myopathy

IMAGING

- Variably dilated, featureless small bowel
  - Poor/absent peristalsis
- Colon **not** dilated: Unused microcolon, no haustrations
- Abnormal bowel fixation: Malrotation
- Variable pelvocaliectasis & hydroureter
- Bladder very dilated with poor or no emptying
  - Nonobstructive urinary tract dilation
    - Polyhydramnios in utero with megacystis suggestive
  - Bladder aspiration improves urinary tract dilation
- Severe abdominal distention → pulmonary compromise

TOP DIFFERENTIAL DIAGNOSES

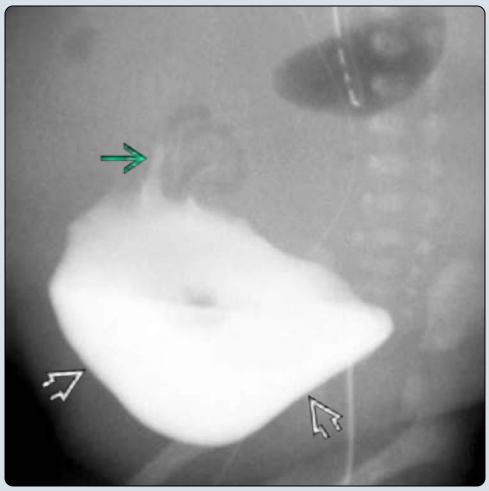
- Posterior urethral valves
- Prune-belly syndrome
- Congenital megalourethra
- Cloacal malformation
- Hirschsprung disease

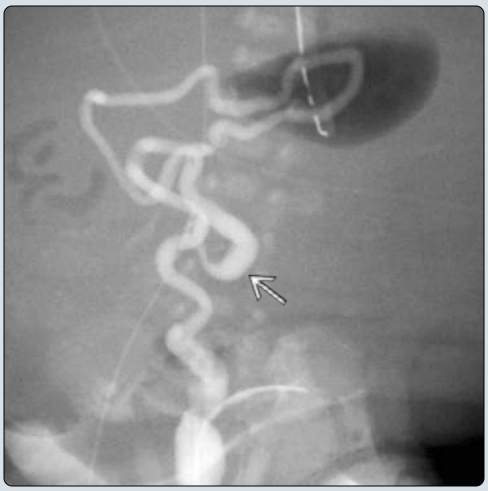
PATHOLOGY

- Mutation in *ACTG2* gene → abnormal smooth muscle actin
- Autosomal dominant, often sporadic

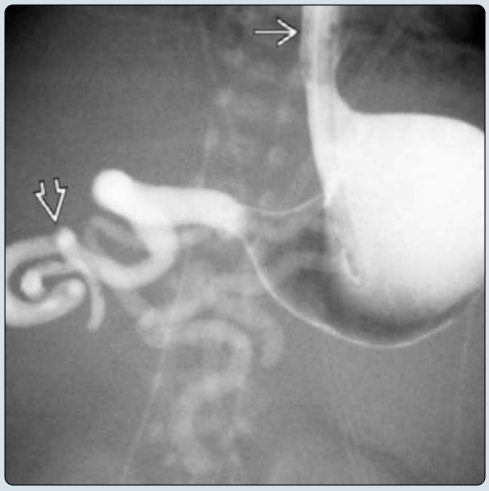
CLINICAL ISSUES

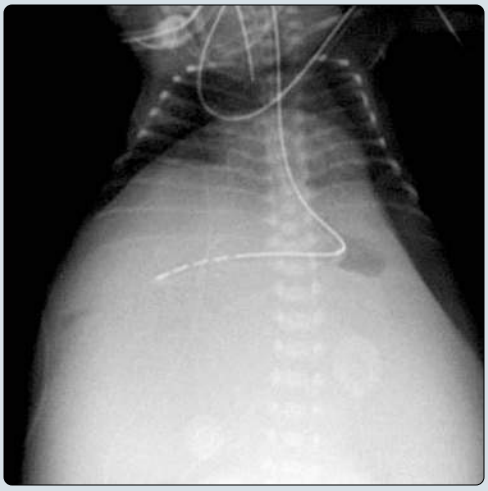
- F > > M
- Clinical triad: Abdominal distention, bilious emesis, failure to pass meconium or urine
- Previously lethal within 1 year
- Survival improved with TPN & multivisceral transplant





(Left) *Anterior oblique view of the urinary bladder during a VCUG shows an enlarged, smooth-walled bladder in this newborn with MMIHS. Contrast in a tubular projection from the bladder dome likely represents a urachal remnant.* **(Right)** *Frontal contrast enema in a newborn with MMIHS shows a very small caliber, unused microcolon without haustrations positioned in the midline abdomen. The cecum is overlying the spine. Marked hydronephrosis is displacing the bowel.*





(Left) *Frontal upper GI image through a nasogastric tube shows full-column gastroesophageal reflux into the mildly dilated esophagus with distal passage of contrast into the featureless tubular small bowel malpositioned in the right upper abdomen, consistent with malrotation.* **(Right)** *Frontal view of the chest in a newborn with MMIHS shows massive abdominal distention impressing on the lung bases. Pulmonary function & development are compromised by mass effect both in utero & postnatally in MMIHS.*

Prune-Belly Syndrome

KEY FACTS

TERMINOLOGY

- Prune-belly syndrome (PBS): Congenital triad of
  - Urinary tract dilation
  - Cryptorchidism
  - Abdominal wall muscle deficiency/laxity (with thin wrinkled skin resembling prune)

IMAGING

- Radiographs: Enlarged abdomen with laterally bulging flanks
  - ± undulation or "wrinkling" of redundant skin
  - ± centralization of bowel gas due to markedly dilated ureters &/or renal collecting systems
  - Small, bell-shaped thorax ± pneumothorax
- VCUG: Marked bilateral vesicoureteral reflux
  - Dilated posterior urethra, but true mechanical obstruction uncommon
- US: Dilated bladder & ureters; ± caliectasis, cystic renal dysplasia, urachal anomalies
  - Empty scrotum (cryptorchidism)
- MR urography can help delineate complex anatomy & assess renal drainage & dysplasia

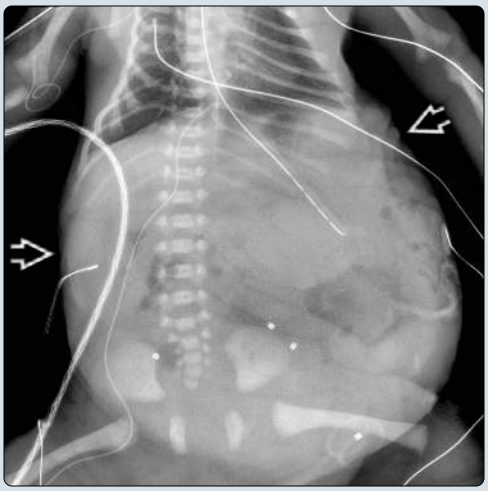
TOP DIFFERENTIAL DIAGNOSES

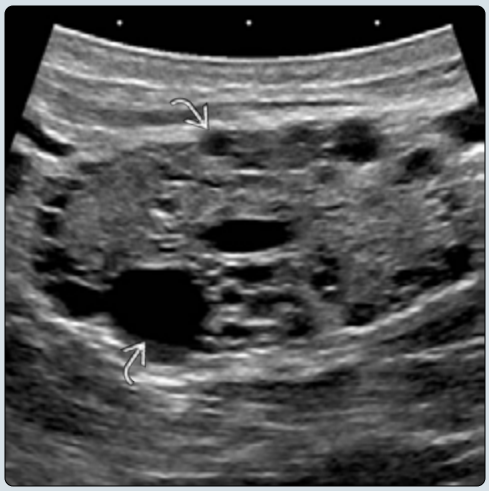
- Posterior urethral valves
- Severe vesicoureteral reflux
- Primary megaureter
- Cloacal anomaly

CLINICAL ISSUES

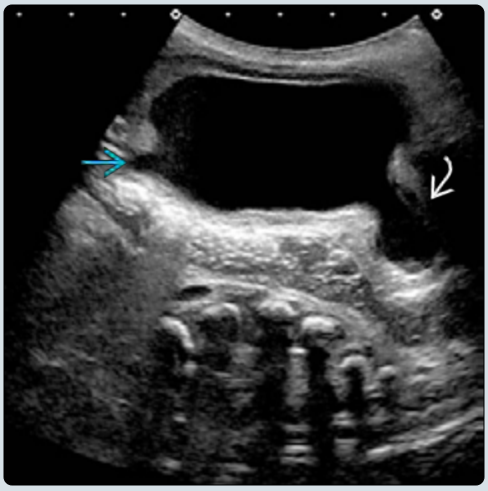
- Large flaccid abdomen with redundant skin, cryptorchidism, ± Potter facies
- 1 in 30,000 to 40,000 live births; 95-99% male
- Variable renal impairment & pulmonary hypoplasia determine prognosis
- Many survivors but with chronic health issues
  - Dialysis or renal transplant in 10-20%

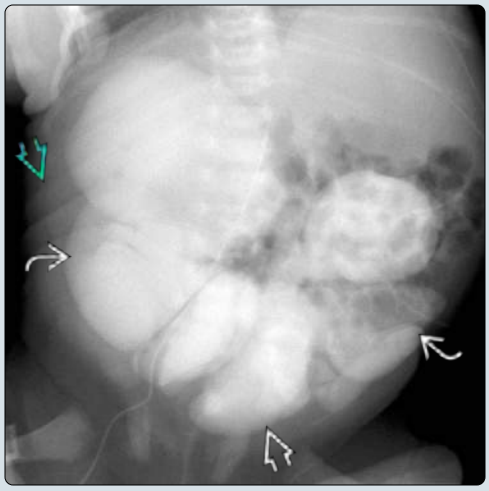
(Left) *Frontal abdominal radiograph in a newborn shows an enlarged abdomen with a flaccid, undulating abdominal wall & a small thorax, typical of prune-belly (or Eagle-Barrett) syndrome (PBS or EBS).* **(Right)** *Longitudinal US in the same newborn with PBS shows numerous cysts scattered through the echogenic renal parenchyma with poor corticomedullary differentiation. The findings are typical of cystic dysplasia of the kidneys, likely due to long standing bladder outlet obstruction.*





(Left) *Longitudinal US of the pelvis in newborn boy with PBS shows a keyhole appearance of the bladder with posterior urethral dilation, bladder wall thickening, & a bladder diverticulum. No obstructing urethral anomaly was seen on VCUG.* **(Right)** *Frontal VCUG in a patient with PBS shows infused contrast filling the bladder & rapidly refluxing into markedly dilated, tortuous ureters bilaterally. The ureters hold several times the volume of the bladder. Note the wrinkled abdominal wall.*





TERMINOLOGY

Synonyms

- Eagle-Barrett syndrome, Obrinsky syndrome, abdominal muscle deficiency, triad syndrome

Definitions

- Prune-belly syndrome (PBS): Congenital triad of
  - Urinary tract dilation
  - Cryptorchidism
  - Abdominal wall muscle deficiency/laxity (with thin wrinkled skin resembling prune)

IMAGING

General Features

- Best diagnostic clue
  - Urinary tract dilation with abdominal distention & laxity

Radiographic Findings

- Enlarged abdomen with laterally bulging flanks
  - ± undulation or "wrinkling" of redundant skin
- ± centralization of bowel gas due to markedly dilated ureters &/or renal collecting systems
- Small, bell-shaped thorax ± pneumothorax
  - Longstanding oligohydramnios → pulmonary hypoplasia

Fluoroscopic Findings

- Dilated bladder with lobulation, elongation, wall-thickening, diverticula, ± urachal anomaly
- Marked bilateral vesicoureteral reflux
- Dilated posterior urethra; true mechanical obstruction uncommon

MR Findings

- MR urography can help sort through complex anatomy, assessing renal function & drainage

Ultrasonographic Findings

- Dilated bladder & ureters, ± caliectasis & cystic renal dysplasia
- Urachal anomalies common
- Empty scrotum (cryptorchidism)
- Deficient abdominal wall musculature

Nuclear Medicine Findings

- Delayed drainage of ureters common

DIFFERENTIAL DIAGNOSIS

Posterior Urethral Valves

- "Keyhole" bladder with high-grade obstruction of posterior urethra by valve tissue in males

Severe Vesicoureteral Reflux

- May be associated with megacystis but typically isolated

Primary Megaureter

- Adynamic distal ureter with proximal dilation

Cloacal Anomaly

- Failure of separation of distal genitourinary & gastrointestinal tracts → single perineal orifice
- Hydrocolpos can cause bladder outlet obstruction

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome

- Prognosis worse in megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS)
- MMIHS more common in females

Congenital Megalourethra

- Findings of bladder outlet obstruction but entire urethra dilated

Urethral Atresia

- Very rare; anhydramnios → pulmonary hypoplasia

PATHOLOGY

General Features

- Etiology
  - Controversial: Mesodermal defect vs. early obstruction of bladder outlet
  - Debated whether PBS truly unique entity vs. phenotype of various bladder outlet pathologies

CLINICAL ISSUES

Presentation

- Large, flaccid abdomen with wrinkled, redundant skin
- Cryptorchidism
- Potter facies if oligohydramnios

Demographics

- 95-99% male
- 1 in 30,000 to 40,000 live births

Natural History & Prognosis

- Variable renal impairment & pulmonary hypoplasia determine prognosis
- Many survivors but with chronic health issues
  - Genitourinary: 100%
  - Cardiovascular: 10%
  - Orthopedic: 20%
  - Gastrointestinal

Treatment

- Bladder outlet: Vesicostomy, cystoplasty, Mitrofanoff
- Dialysis or renal transplant in 10-20%
- Orchiopexy
- Abdominoplasty, rectus femoris grafts

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Posterior Urethral Valves

KEY FACTS

TERMINOLOGY

- Varying degrees of chronic urethral obstruction due to fusion &/or prominence of plicae colliculi (which are normal concentric folds within posterior urethra)
- Occurs exclusively in males

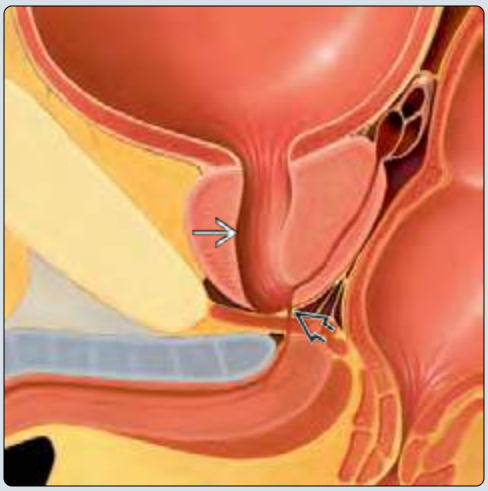
IMAGING

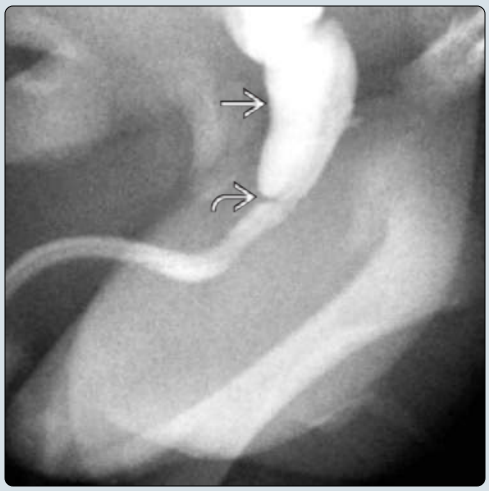
- Voiding cystourethrogram (VCUG)
  - Abrupt transition from dilated posterior urethra to small bulbar urethra at level of valvular tissue; actual valve tissue may not be visible
  - Bladder dilation, wall trabeculation, muscular hypertrophy, diverticula, ± patent urachus
  - Vesicoureteral reflux (50-70%)
- Ultrasound
  - Bilateral hydroureteronephrosis
  - Echogenic, dysplastic kidneys with poor corticomedullary differentiation ± cortical cysts, urinomas, ascites
  - Lobular bladder with thickened, irregular wall ± diverticula
- Diagnosis made on VCUG, cystoscopy, or cystosonography

CLINICAL ISSUES

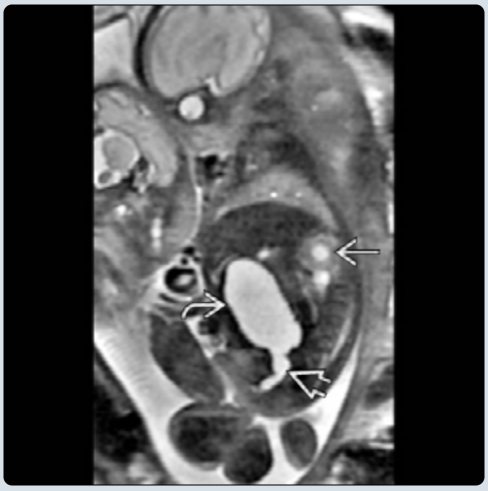
- Severity & duration of obstruction determines age of presentation & clinical symptoms, which include
  - Perinatally: Oligohydramnios, hydronephrosis, anuria, urinary ascites, urinoma, pulmonary hypoplasia
  - In infancy: Urinary tract infection, sepsis, urinary retention, poor urinary stream, failure to thrive
  - In childhood: Abnormal voiding patterns, hesitancy, straining, poor stream, large postvoid residual, renal insufficiency/failure
- Catheterization at birth to relieve obstruction, followed by urgent endoscopic valve ablation
- 30-40% will eventually develop end-stage renal disease
- 75% have long-term urinary bladder dysfunction
- Fertility issues common in long term survivors

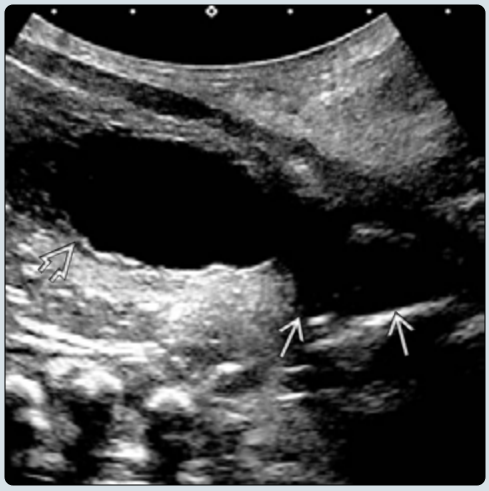
(Left) *Sagittal graphic shows an enlarged posterior urethra extending through the prostate with an abrupt change in urethral caliber just distal to the verumontanum at a typical level of valve tissue.* **(Right)** *Lateral oblique voiding cystourethrogram (VCUG) in a newborn with posterior urethral valves (PUV) shows the same anatomy from the previous graphic. There is a dilated posterior urethra with an abrupt change in urethral caliber just distal to the valve tissue.*





(Left) *Sagittal SSFSE T2 MR in a 3rd-trimester twin gestation shows a fetus with an elongated mildly thick-walled urinary bladder, hydronephrosis, & a dilated posterior urethra typical of PUV.* **(Right)** *Longitudinal US in a newborn with PUV shows a dilated posterior urethra inferior to the bladder base. Note the irregular bladder wall thickening.*





TERMINOLOGY

Abbreviations

- Posterior urethral valves (PUV)

Definitions

- PUV: Varying degrees of chronic urethral obstruction due to fusion &/or prominence of plicae colliculi (which are normal concentric folds within posterior urethra)
- Valve bladder: Varying degrees of urinary bladder dysfunction secondary to chronic bladder obstruction

IMAGING

General Features

- Best diagnostic clue
  - Dilated posterior male urethra with distinct caliber change at level of valves
    - Actual valve tissue very thin, may not be visible
  - Diagnosis made on voiding cystourethrogram (VCUG), cystoscopy, or cystosonography
- Location
  - PUV lie just distal to prostatic urethra

Radiographic Findings

- Dilated urinary bladder creates pseudomass in pelvis, displacing bowel loops superiorly
- Perinephric urinoma, dilated ureters, & dilated renal collecting systems may cause additional mass effect
- ± small bell-shaped thorax of pulmonary hypoplasia, ± pneumothorax

Fluoroscopic Findings

- Voiding cystourethrogram
  - Hallmark: Abrupt caliber change in posterior urethra
    - Dilated posterior urethra gives rise to small caliber bulbar & penile urethra
    - Actual valve tissue need not be seen on imaging to make diagnosis
    - Note: Urethral catheter left in place during voiding may "stent" valve tissue, obscuring caliber change & pushing valve tissue against urethral wall
  - Associated findings
    - Bladder dilation, wall trabeculation, muscular hypertrophy, diverticula
    - Vesicoureteral reflux (VUR) (50-70%)
    - Urinary ascites
    - Urinoma/perinephric urine collection
    - Reflux into utricle or other ducts
    - Patent urachus

Ultrasonographic Findings

- Grayscale ultrasound
  - Bilateral or unilateral hydroureteronephrosis
  - Echogenic, dysplastic kidneys with poor corticomedullary differentiation ± cortical cysts, urinomas, ascites
  - Lobular bladder with thickened, irregular wall ± diverticula
  - Angling transducer toward bladder neck may reveal dilated posterior urethra
  - Cystosonography with US contrast agents used in Europe > USA
    - Eliminates radiation of fluoroscopic VCUG
    - Exam performed from transperineal approach
    - Bladder catheterization & voiding during imaging still required

MR Findings

- MR urogram
  - Anatomic findings mirror US, though dilated ureteral course more discernible by MR
  - Physiologic study using dynamic contrast evaluation of renal function & drainage
    - Caution required with low glomerular filtration rate
    - Patients with severe renal failure receiving gadolinium-based contrast agents at risk for nephrogenic systemic fibrosis

Nuclear Medicine Findings

- Cortical imaging for scarring & differential renal function
- Diuretic renogram to evaluate additional obstructing lesion of ureter or renal pelvis

Other Modality Findings

- At direct cystoscopy, valve tissue translucent, may be pushed back against outer walls of urethra by inflowing irrigation fluid
- Similar drawbacks exist for fluoroscopic retrograde urethrogram, which may not show dynamic change in urethral caliber

Imaging Recommendations

- Best imaging tool
  - Fluoroscopic VCUG
  - Cystosonography used in countries where contrast agents available
- Protocol advice
  - VCUG: Removal of catheter while patient voiding will best demonstrate urethral configuration consistent with PUV; however, catheter removal should 1st be discussed with clinical team prior to procedure

DIFFERENTIAL DIAGNOSIS

Prune-Belly Syndrome (Eagle-Barrett)

- Hydroureteronephrosis
  - Posterior urethra dilated but nonobstructed
- Absence/hypotonia of abdominal wall musculature
- Cryptorchidism

Anterior Urethral Valves

- Very rare entity where prominent semilunar fold obstructs anterior urethra

Voiding Dysfunction

- Detrusor external sphincter dyssynergia can resemble PUV transiently (due to opening of bladder neck with bulging posterior urethra & contracted external sphincter)
- Appearance not persistent

Ureterocele

- Prolapse into posterior urethra can cause obstruction

Megalourethra

- Rare entity where entire length of urethra enlarged due to absence of corpus spongiosum

Urethral Stricture

- Postsurgical, posttraumatic, or postinfectious
- History key to making this diagnosis

Megacystis Microcolon Intestinal Hypoperistalsis Syndrome

- M:F = 1:4
- Dilated hypoperistaltic bowel with malrotation
- Nonobstructive megacystis

Megacystis-Megaureter Association

- Large volume VUR cycling between dilated upper tracts & bladder

PATHOLOGY

General Features

- Associated abnormalities
  - Longstanding oligohydramnios leads to pulmonary hypoplasia, which may not be survivable

Staging, Grading, & Classification

- Type I (most common): Anterior fusion of plicae colliculi
- Type II (rarest): Longitudinal folds from verumontanum to bladder neck
- Type III (rare): Disc, ring, or windsock-type tissue distal to verumontanum

Gross Pathologic & Surgical Features

- Valve tissue typically very thin but functions like sail, causing near complete obstruction to antegrade flow of urine

Microscopic Features

- Valve tissue thin, normal urothelium
- Bladder wall will show muscular hypertrophy & fibrosis
- Kidneys may show tubulointerstitial fibrosis & dysplasia

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Severity & duration of obstruction determine age of presentation & clinical symptoms, which include
    - Perinatally: Oligohydramnios, hydronephrosis ± renal dysplasia & renal failure, anuria, urinary ascites, urinoma, pulmonary hypoplasia with respiratory distress
    - In infancy: Urinary tract infection, sepsis, urinary retention, poor urinary stream, failure to thrive
    - In childhood: Abnormal voiding patterns, hesitancy, straining, poor stream, large postvoid residual, renal insufficiency/failure
  - Historically 1/3 present at each age: Perinatal, infancy, childhood
    - Up to 1/2 of all cases now diagnosed in utero
    - Fewer cases now diagnosed in infancy & childhood

Demographics

- Gender
  - Males only
- Epidemiology
  - Incidence: 1 in 5,000-8,000 births

Natural History & Prognosis

- Varies with degree of renal dysplasia related to chronic obstruction & VUR
- Unilateral reflux & urinary ascites protective for contralateral kidney (relieve pressure)
- Renal insufficiency has bimodal peak, developing in 1st months of life vs. adolescence after initial improvement
  - Lowest creatinine in 1st year of life (nadir Cr) has some prognostic value
- 30-40% will eventually develop end-stage renal disease
- 75% with long-term urinary bladder dysfunction
  - Poor bladder compliance, overactivity, & myogenic failure (incomplete bladder emptying, overflow incontinence)

Treatment

- Catheterization at birth to relieve obstruction
- Urgent endoscopic valve ablation
  - VUR ↓ by 30% after valve ablation
  - Improved renal function with early treatment
- Secondary bladder surgeries often needed: Bladder augmentation or continent diversion (Mitrofanoff)
  - ↑ risk of malignancy decades after augmentation using GI tract
- Long-term follow-up necessary to monitor renal function & bladder compliance
- Dialysis, renal transplant for end-stage renal disease
  - 10-15% of pediatric renal transplants due to PUV
  - Comparable transplant survival rates in PUV patients vs. patients with nonobstructive etiologies
- Fetal intervention performed in cases of severe oligohydramnios; conflicting data on long-term renal function benefit, though procedures may ↑ "pulmonary survivors" needing renal replacement therapy
  - Serial amnioinfusions to restore normal amniotic fluid volume (to promote lung development)
  - Serial bladder taps vs. vesicoamniotic shunt
  - In utero cystoscopy with valve fulguration

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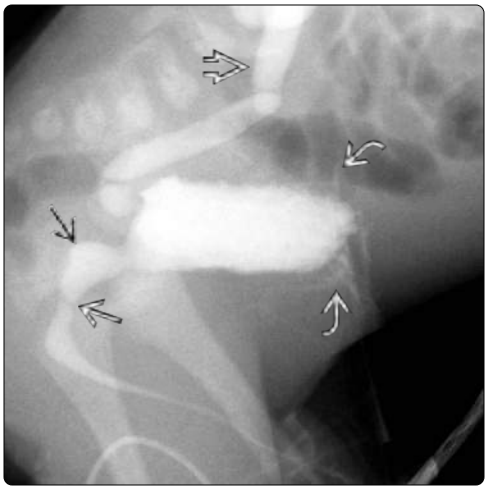
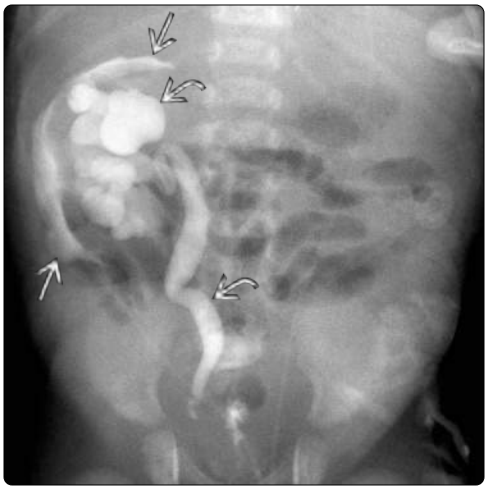
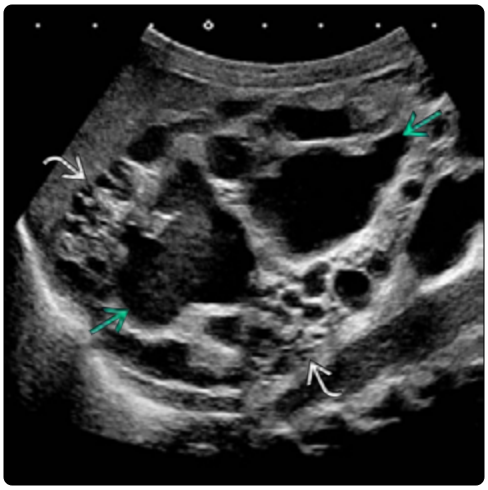
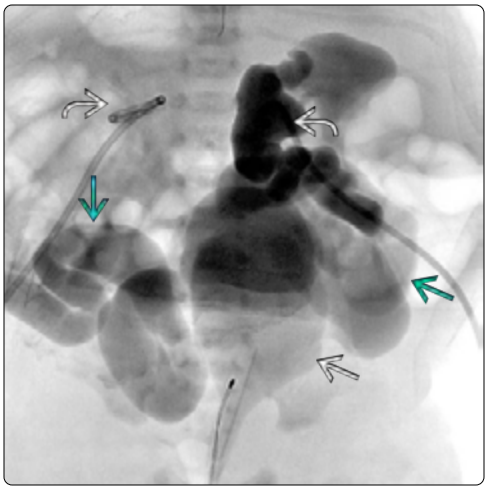


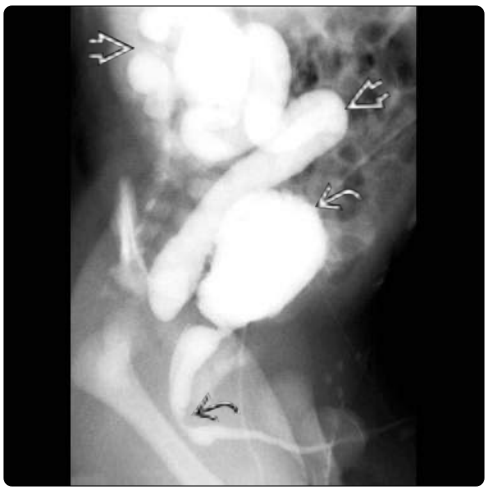


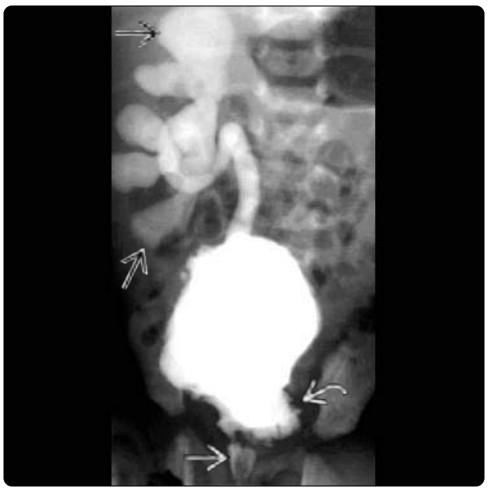
(Left) *Lateral VCUG in a newborn with PUV shows the thin valvular tissue ➡ at the inferior margin of the dilated posterior urethra ⇨. The urinary bladder wall is markedly thickened with contrast extending between the trabeculations ⇶. High-grade unilateral vesicoureteral reflux (VUR) ➡ is noted.* **(Right)** *Frontal VCUG in the same patient after voiding shows contrast pooling around the kidney ➡. This urinoma is due to high- pressure VUR ⇶ causing a forniceal rupture in the setting of a chronic bladder outlet obstruction.*





(Left) *Longitudinal US of the left kidney in a newborn with PUV shows marked pelvocaliectasis ⇨ & cystic dysplastic changes of the renal cortex ⇶ due to chronic obstruction in utero.* **(Right)** *Frontal prone radiograph during bilateral percutaneous nephrostomy tube placements ⇶ shows contrast filling severely dilated ureters ⇨ & a lobulated urinary bladder ➡.*





(Left) *Oblique VCUG in a newborn shows PUV ⇶, massive unilateral VUR ➡, & thickening of the bladder wall ⇶.* **(Right)** *Frontal VCUG in the same patient shows high-grade VUR into the right kidney ⇨ with irregular bladder wall thickening ⇶ & a dilated posterior urethra ➡. High-grade unilateral VUR is protective of the contralateral renal function.*

Urachal Abnormalities

KEY FACTS

TERMINOLOGY

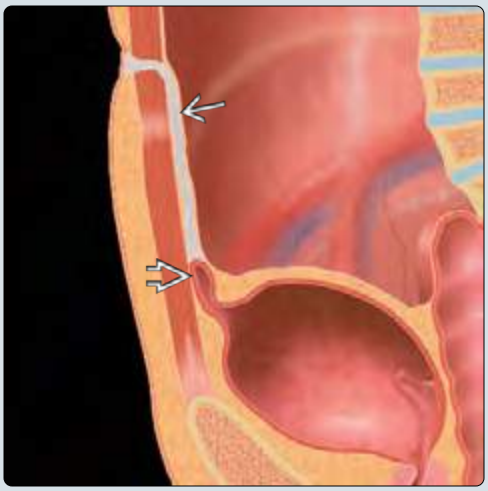
- Persistence of all or portion of connection between bladder dome & umbilicus; remnant of fetal allantoic stalk

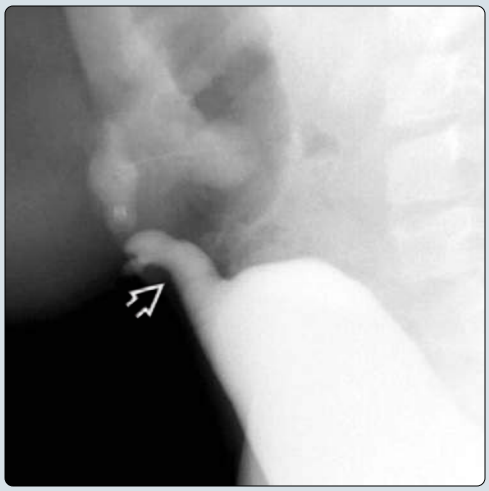
IMAGING

- Patent urachus or urachal fistula
  - Open channel from bladder to umbilicus through which urine can leak
- Urachal sinus
  - Persistence of superficial segment of channel opening onto skin surface
- Urachal diverticulum
  - Persistence of deep segment of tract, creating point or diverticulum off of anterior-superior bladder wall
- Urachal cyst
  - Persistence of intermediary segment with fibrous attachments to bladder & umbilicus
- Size & shape depend on type of remnant, location, & presence of inflammation
- Helpful hints for US scanning
  - Bladder should be fairly full
  - Patient should be relaxed as Valsalva maneuver can squeeze fluid out of patent tract
  - Begin scanning at inferior bladder level & sweep transducer upward toward umbilicus
  - Gentle pressure on bladder dome can push fluid into patent tract to aid visualization

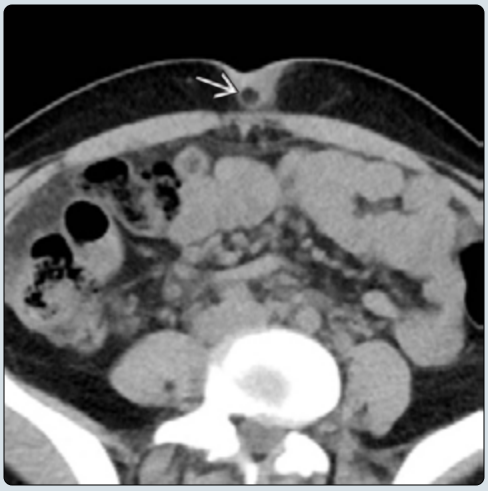
CLINICAL ISSUES

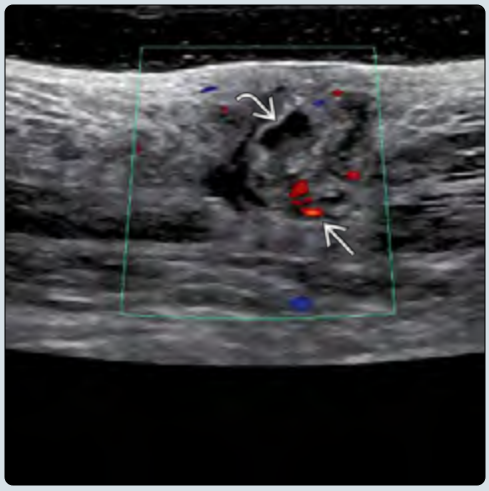
- Incidence of patent urachus: 1 in 40,000
  - Other urachal anomalies more common, though reliable statistics not available
- Generally, prognosis excellent
  - Urachal tract resected; no further follow-up needed
    - Open surgery previously, now laparoscopic
- Risk of malignancy in adults if not resected
  - Urachal malignancies < 1% of all bladder cancers





(Left) *Sagittal graphic shows a urachal diverticulum ⇨ & fibrotic tract ⇨ to the umbilicus. When the entire tract remains open, it is called a patent urachus. The urachal sinus & urachal cyst are additional variations along this spectrum.* **(Right)** *Lateral voiding cystourethrogram image in an infant with a history of umbilical drainage shows a tubular contrast-filled connection ⇨ between the bladder dome & the umbilicus, consistent with a patent urachus.*





(Left) *Axial NECT in a teenager shows a low-density cyst ⇨ at the base of the umbilicus with thickening of the periumbilical tissues.* **(Right)** *Longitudinal US of the same patient shows mild hyperemia ⇨ & heterogeneity of the periumbilical tissues with a small fluid collection centrally ⇨. A small urachal cyst was resected. Additional considerations for this appearance could include an evolving hematoma, abscess, inflamed dermoid, or other lesions.*

TERMINOLOGY

Synonyms

- Patent urachus, urachal fistula, urachal remnant, urachal cyst, urachal sinus, urachal diverticulum

Definitions

- Partial or complete persistence of connection between bladder dome & umbilicus; remnant of fetal allantoic stalk

IMAGING

General Features

- Best diagnostic clue
  - Fluid or cyst along tract between bladder dome & umbilicus in patient with umbilical drainage
- Location
  - Midline between dome of bladder & umbilicus
- Size
  - Variable
- Morphology
  - Patent urachus or urachal fistula
    - Open channel from bladder to umbilicus through which urine can leak
  - Urachal sinus
    - Persistence of superficial segment of channel opening onto skin surface
  - Urachal diverticulum
    - Persistence of deep segment of channel creating point or diverticulum off of anterior-superior bladder wall
  - Urachal cyst
    - Localized, fluid-containing persistence of intermediary segment with fibrous attachments to bladder & umbilicus
    - Case reports of intravesical urachal cysts along anterior superior bladder wall

Radiographic Findings

- Cross-table lateral view in neonate may be 1st test ordered to exclude hernia
- In general, radiography not useful

Fluoroscopic Findings

- Voiding cystourethrogram (VCUG)
  - Best test to document patency of urachus
  - Tends to underestimate length or extent of remnant
  - Inflammation along tract can intermittently block lumen
  - True lateral views necessary (with full bladder)
  - Radiopaque marker on umbilicus can be helpful

Ultrasonographic Findings

- Grayscale ultrasound
  - Size & shape depend on type of remnant, location, & presence of inflammation
    - Diverticulum of fully patent urachus contiguous with anterior superior aspect of bladder
  - Often has thick, well-defined wall (especially if inflamed)
  - Wall may have layered appearance mimicking gut signature
  - Remnant may or may not contain fluid
  - Fluid-debris levels may be present in urachal cysts
- Color Doppler
  - Doppler useful in
    - Assessing degree of hyperemia when infected
    - Excluding vascular anomaly
- Helpful hints when scanning for urachal remnants
  - Bladder should be fairly full
  - Patient should be relaxed as Valsalva maneuver can squeeze fluid out of patent tract
  - Begin scanning at bladder level & sweep transducer upward toward umbilicus
  - Gentle pressure on bladder dome can push fluid into patent tract to aid visualization

CT Findings

- Occasionally recognized incidentally on CT scans performed for other reasons
- Infected urachal cyst may present with "rule out abscess" manifestations
- Look for round, triangular, or tubular fluid-filled structure extending along (superficial to) anterior aspect of peritoneal cavity between urinary bladder & umbilicus
- Localized cyst may be seen when proximal & distal segments of tract fibrotic
- Surrounding inflammation common

MR Findings

- Similar findings as CT
- Often incidental finding on MR scan

Nuclear Medicine Findings

- Flow of renally-excreted radiotracers into tract from urinary bladder may mimic other pathology
  - FDG-PET could falsely suggest malignancy

Imaging Recommendations

- Best imaging tool
  - Ultrasound defines static anatomy well
  - VCUG useful to show flow dynamics & confirm patency

DIFFERENTIAL DIAGNOSIS

Granulation Tissue of Umbilical Stump

- Most often confused with urachal remnants, especially urachal sinus
- No deeper discrete fluid-filled tract identified

Omphalitis

- Poorly defined, heterogeneous inflammation of anterior abdominal wall just deep to umbilicus
- Can be present in conjunction with urachal remnant
- In severe cases, may need to treat with antibiotics & reevaluate

Umbilical Hernia

- Usually readily discerned by physical exam & imaging
- Hernia may contain peristalsing bowel or omentum
- Helpful to scan patient upright & with Valsalva

Hemangioma of Umbilical Cord

- Doppler ultrasound will show high volume of low-resistance arterial blood flow in lobulated mass rather than stagnant fluid

PATHOLOGY

General Features

- Etiology
  - In embryos, allantois
    - Forms from caudal end of yolk sac
    - Functions as primitive bladder as well as blood-forming organ
    - Normally involutes by 2nd month of gestation
    - Obliteration forms median umbilical ligament
    - May have segments persist as urachal remnants
  - Urachus lies in space of Retzius
    - Between transversalis fascia anteriorly & peritoneum posteriorly
- Associated abnormalities
  - Periumbilical associations
    - Omphalocele
    - Omphalomesenteric remnant
  - Bladder outlet associations
    - Posterior urethral valves
    - Urethral atresia
    - Cloacal anomalies
    - Urogenital sinus malformation
  - Miscellaneous associations
    - Myelomeningocele (perhaps because of neurogenic bladder)
    - Unilateral kidney & other renal anomalies
    - Vaginal atresia

Gross Pathologic & Surgical Features

- Well-defined stalk with mucosal lining & varying degrees of fibrosis/lumen obliteration

Microscopic Features

- Cellular histology varies, not simple urothelium
  - Transitional cell epithelium
  - Columnar epithelium
  - Glandular epithelium
  - Squamous epithelium
- Varied cell types explain variety of malignant cell lines found in adults with urachal tumors
- Fibrostromal histology has low risk of malignant transformation, while epithelial histology has higher risk
  - Imaging & symptoms do not differentiate between histologic subtypes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Varies with type of urachal remnant
    - Patent urachus presents with drainage from umbilicus, urinary tract infection, & relapsing periumbilical inflammation
    - Occasionally, urachus remains patent in response to bladder outlet obstruction (posterior urethral valves, pelvic mass, etc.) & will close when outlet repaired
    - Urachal sinus presents with periumbilical tenderness, wet umbilicus, or nonhealing granulation of umbilicus
    - Urachal cyst presents in childhood or adolescence with suprapubic mass, fever, pain, & irritative voiding symptoms
    - Urachal diverticula are often asymptomatic & discovered incidentally; rarely they enlarge, fail to drain during urination, & become predisposed to infection or stone formation

Demographics

- Age
  - Patent urachus seen primarily in newborns
  - Urachal sinus also typically diagnosed within 1st few months of life
  - Urachal cysts may go undetected until childhood or adulthood
  - Urachal diverticulum often undetected (autopsy finding)
- Gender
  - M > F
  - 2:1 ratio
- Epidemiology
  - Incidence of patent urachus: 1 in 40,000
  - Other urachal anomalies more common, though reliable statistics not available

Natural History & Prognosis

- Generally excellent prognosis
  - Urachal tract resected; no further follow-up needed
- Risk of malignancy if not resected: Adenocarcinoma, mucinous cystadenocarcinoma, villous adenoma
  - Urachal malignancies < 1% of all bladder cancers
  - Typically occur in patients 40-70 years of age
  - Majority of cancers occur in men (~ 75%)
  - Present with pain, hematuria or mucinous micturition
  - Local tumor invasion common at time of urachal cancer diagnosis
- Reports of using urachal remnant instead of appendix for Mitrofanoff

Treatment

- Resection of entire tract
  - Open surgery previously, now laparoscopic
- Often performed in staged fashion
  - Inflammation & infection treated, allowed to heal
  - Definitive surgery delayed
- When accompanied by bladder outlet obstruction
  - Surgery to fix outlet obstruction must be performed 1st
    - Patent urachus serves as pop-off valve in these cases
  - Following correction of bladder outlet problems, allow ~ 1 year for patent urachus to close independently
  - If drainage persists, reassess bladder outlet; if functioning well, resect tract

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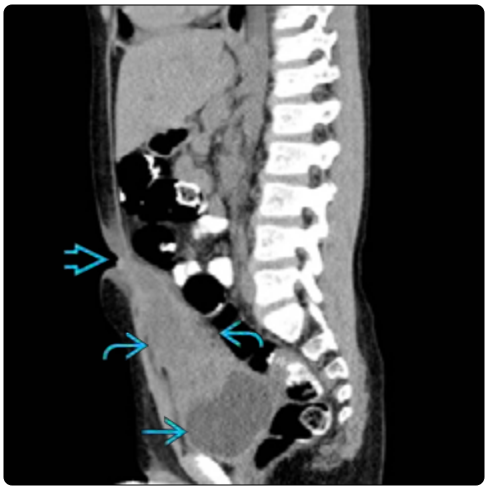
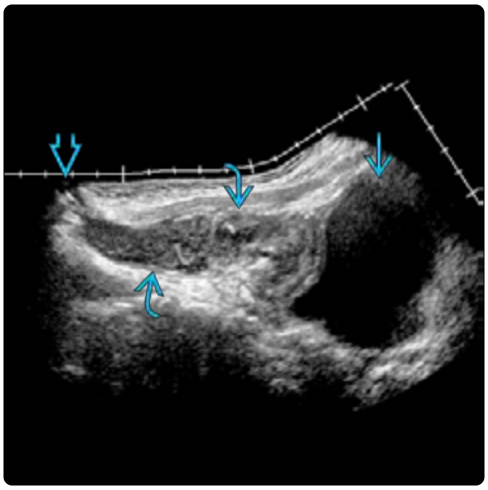
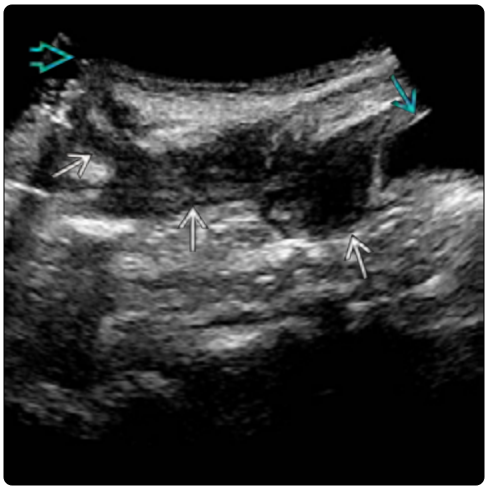
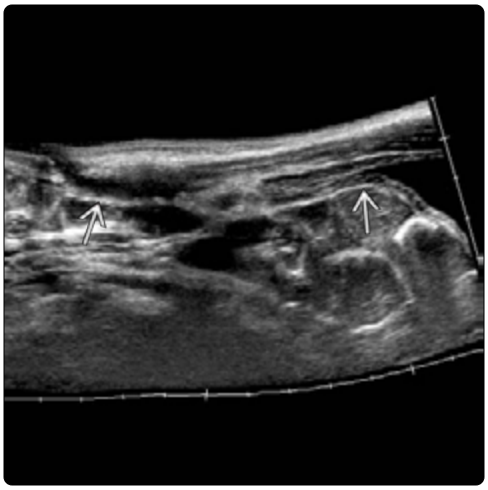


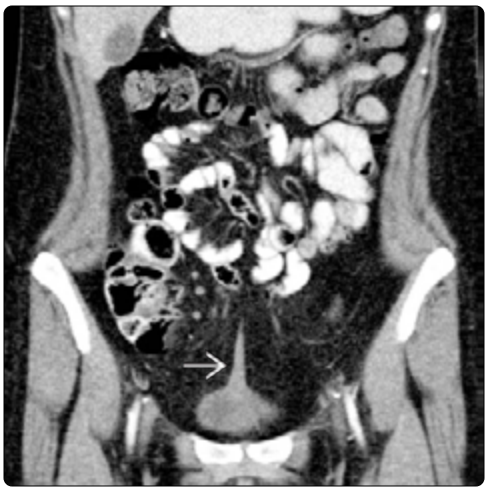


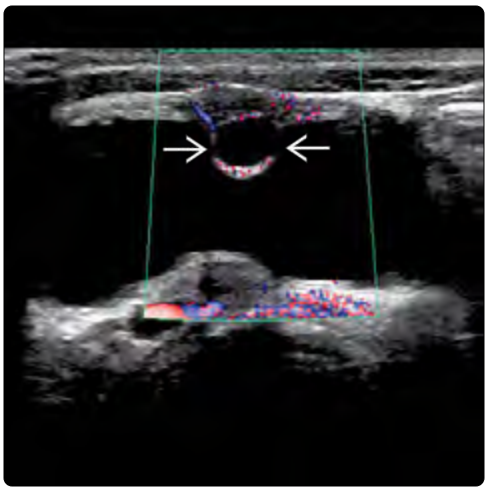
(Left) *Sagittal NECT in a 7-year-old boy with abdominal pain & fever shows mildly heterogeneous soft tissue density between the bladder & umbilicus. The appendix was normal (not shown), & US was ordered for further evaluation.* **(Right)** *Sagittal US in the same patient shows a complex collection extending from the bladder to the umbilicus. No flow of internal mobile contents to the bowel, bladder, or skin was demonstrated. At surgery, an infected urachal remnant was resected.*





(Left) *Longitudinal US in a 6-year-old boy with fever & periumbilical pain shows an irregularly shaped hypoechoic tract between the bladder & umbilicus. Foul smelling fluid/urine was expressed from the tract during the scan, consistent with an infected patent urachus.* **(Right)** *Longitudinal US in the same patient after a course of antibiotics shows a decreased caliber of the patent urachus, which was subsequently surgically excised. Note the pointed shape of the bladder dome (which is usually round).*





(Left) *Coronal CECT performed for suspected appendicitis in a patient with pain shows a thin, triangular soft tissue lesion extending from the dome of the bladder toward the umbilicus, either a urachal remnant or bladder diverticulum.* **(Right)** *Transverse color Doppler US shows blood flow in the rim of a cystic lesion along the anterior-superior margin of the bladder. At surgery, an unusual invaginating urachal remnant was resected.*

Cloaca

KEY FACTS

TERMINOLOGY

- Anorectal malformation of female patients in which rectum, urethra, & vagina converge to form common channel draining to single perineal orifice (which allows clinical diagnosis)
- Imaging performed for operative planning & to evaluate associated malformations

IMAGING

- Radiographs: Multiple dilated bowel loops, ± bowel Ca^{2+}
- US: Renal, pelvic, & spine
  - Cystic pelvic mass: Most commonly hydrocolpos (> 30% of all cloaca patients)
    - Rarely dilated bladder, meconium pseudocyst
  - ± hemivaginas/hemiuteri (40%)
  - ± hydroureteronephrosis
  - ± echogenic calcified meconium in dilated bowel or bladder
  - ± spinal anomalies
- Postcolostomy distal colostogram/cloacagram prior to definitive repair

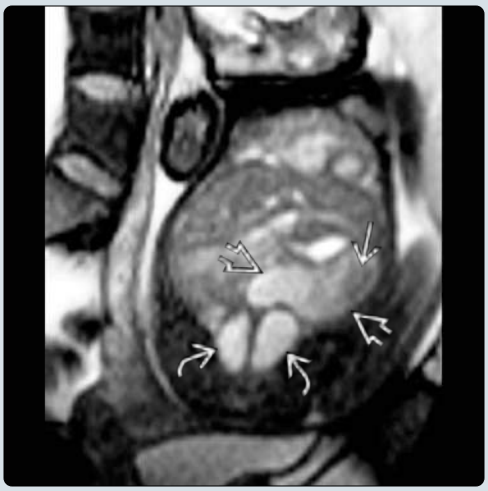
TOP DIFFERENTIAL DIAGNOSES

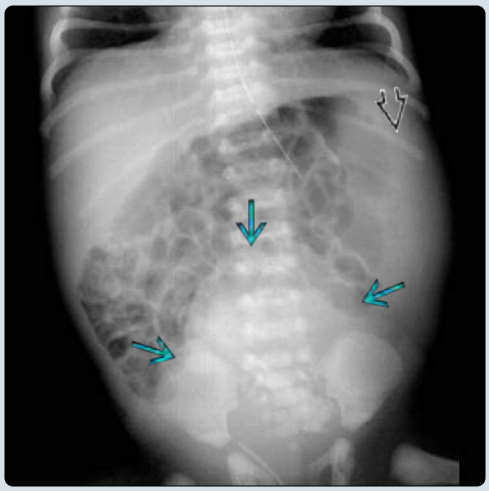
- Anorectal malformation
- Other low/distal neonatal bowel obstructions
- Isolated hydrocolpos
- Megacystis-microcolon-intestinal hypoperistalsis syndrome

CLINICAL ISSUES

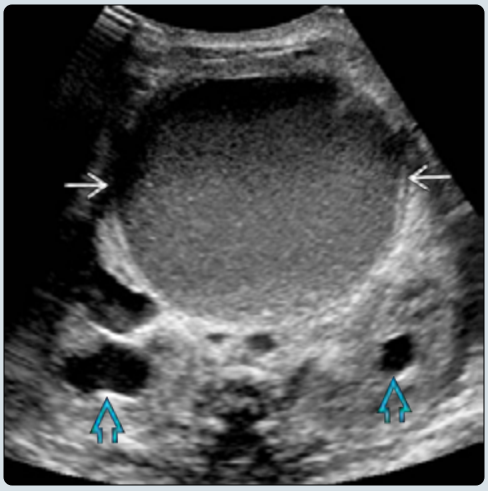
- Hydrocolpos → ↓ renal function without urgent drainage
- Prognosis based on common channel length
  - < 3 cm: Most common type (56%), less complex repair, better functional prognosis
  - > 3 cm: Less common type (44%), more complex repair, worse functional prognosis

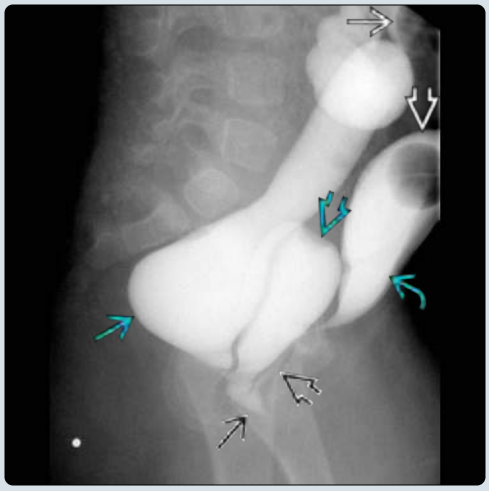
(Left) *Coronal SSFP MR in a 31-week gestation female fetus with a cloaca shows 2 dilated hemivaginas (HVs) & a dilated but tapering distal colon. The heterogeneous, low-signal material within the dilated colon suggests meconium ± Ca^{2+}.* **(Right)** *AP abdominal radiograph in a 1-day-old girl with a cloaca shows a large soft tissue mass arising from the pelvis or displacing bowel superiorly & laterally, likely representing hydrocolpos. The left flank soft tissue mass is likely due to a meconium-filled colon.*





(Left) *Transverse US in the same patient shows a markedly distended vagina filled with echogenic fluid. There is moderate hydronephrosis of both kidneys. The urinary bladder (not shown) was compressed by the hydrocolpos.* **(Right)** *Injection of a mucous fistula & vesicostomy shows filling of the rectum, vagina (with cervical impression), & bladder plus urethra. All of these structures drain into a common channel (or cloaca).*





TERMINOLOGY

Synonyms

- Cloacal malformation

Definitions

- Anorectal malformation in female patients in which rectum, urethra, & vagina converge to form common channel that drains to single perineal orifice (which allows clinical diagnosis)

IMAGING

General Features

- Best diagnostic clue
  - Dilated bowel + hydrocolpos in female fetus (or neonate with single perineal orifice)

Radiographic Findings

- Neonatal intestinal obstruction
  - Multiple dilated bowel loops with variable degree of rectal distention
  - Displacement of gas-filled bowel by dilated structures
    - Hydrocolpos (> 30% of all patients)
    - ± meconium filled colon
  - ± bowel or bladder Ca^{2+} from mixing of urine & stool
  - ± curvilinear peritoneal Ca^{2} (if bowel perforation)
  - Additional findings
    - ± spinal anomalies
    - ± radial ray anomalies
    - ± cardiomegaly, pulmonary edema → congenital heart disease
    - ± proximal esophageal pouch → esophageal atresia

Ultrasonographic Findings

- Neonatal cystic pelvic mass: Commonly hydrocolpos
  - Dilated vagina(s) mimics bladder
- Hemivaginas/hemiuteri (40%)
- ± hydroureteronephrosis (HUN); other GU anomalies (renal agenesis, cross-fused ectopia, horseshoe kidney)
- ± echogenic calcified meconium in dilated bowel or bladder
- ± ascites
- ± spinal cord anomalies

Fluoroscopic Findings

- Voiding cystourethrogram
  - Not often helpful precolostomy; difficult bladder catheterization
  - Usually postrepair to check for vesicoureteral reflux, neurogenic bladder
- Fistulagram
  - Postcolostomy: Inject mucous fistula; opacify distal colon ± bladder, single/hemivagina(s), common channel
    - Length of common channel important to operative planning, prognosis
  - ± cloacagram by rotational fluoroscopy or MR

MR Findings

- Fetal MR: Dilated bowel containing T1 bright meconium; absence of T1 bright rectal meconium beyond 20-weeks gestational age; hydrocolpos of single or hemivagina(s)
  - ± bilateral HUN

DIFFERENTIAL DIAGNOSIS

Anorectal Malformation

- Other types in female patients: Imperforate anus + rectovestibular fistula, rectoperineal fistula, or no fistula
  - True rectovaginal fistula rare
  - > 1 perineal orifice present clinically

Other Low/Distal Neonatal Bowel Obstructions

- Anus present on physical exam
- Contrast enema to differentiate
  - Meconium ileus
  - Ileal atresia
  - Hirschsprung disease
  - Small left colon/meconium plug syndrome
  - Colonic atresia

Isolated Hydrometrocolpos

- Vaginal septum or imperforate hymen; anus present on physical exam

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome

- Large bladder may mimic hydrocolpos; anus present on physical exam

PATHOLOGY

General Features

- Etiology
  - Failed division of embryologic cloaca by urorectal septum before 6th gestational week

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Single perineal orifice; abdominal distention

Natural History & Prognosis

- Hydrocolpos → ↓ renal function in neonate unless urgent drainage
- Prognosis
  - < 3 cm: Most common type (56%), less complex repair, better functional prognosis
  - > 3 cm: Less common type (44%), more complex repair, worse functional prognosis

Treatment

- Surgical repair depends on length of common channel
  - < 3 cm: Posterior sagittal anorectoplasty (PSARP) + urogenital mobilization
  - > 3 cm: PSARP ± laparotomy(oscopy) + complex surgical reconstruction

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Bladder Exstrophy

KEY FACTS

TERMINOLOGY

- Classic bladder exstrophy (CBE): Low midline abdominal wall defect with exposure of bladder plate & urethra + low-set umbilicus
  - Split lower abdominal skin & rectus abdominis + pubic symphysis diastasis
  - Bifid clitoris in females, epispadias in males
  - Deficient genitalia & pelvic floor muscles
- Epispadias: Abnormal dorsal urethral opening
  - With CBE, entire dorsal urethra open with abnormal bladder sphincter

IMAGING

- Absent urinary bladder by prenatal sonography with normal amniotic fluid
- Pubic symphysis diastasis
- Lobular soft tissue at lower abdominal wall
- High rates of vesicoureteral reflux & dysfunctional bladder prior to ureteral reimplants & bladder neck reconstruction

TOP DIFFERENTIAL DIAGNOSES

- Cloacal exstrophy/OEIS complex
- Omphalocele
- Gastroschisis
- Impaired urine production (absent bladder)

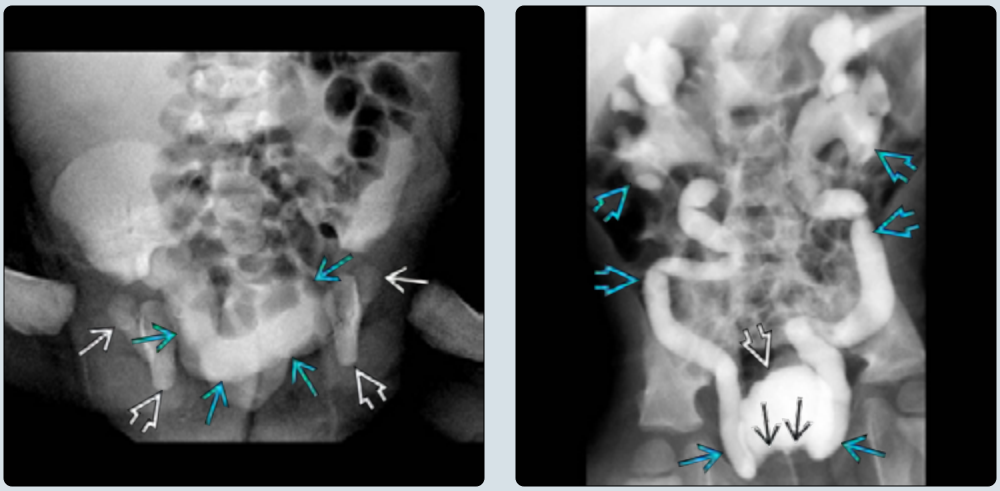
PATHOLOGY

- Cloacal membrane rupture due to failed mesodermal ingrowth

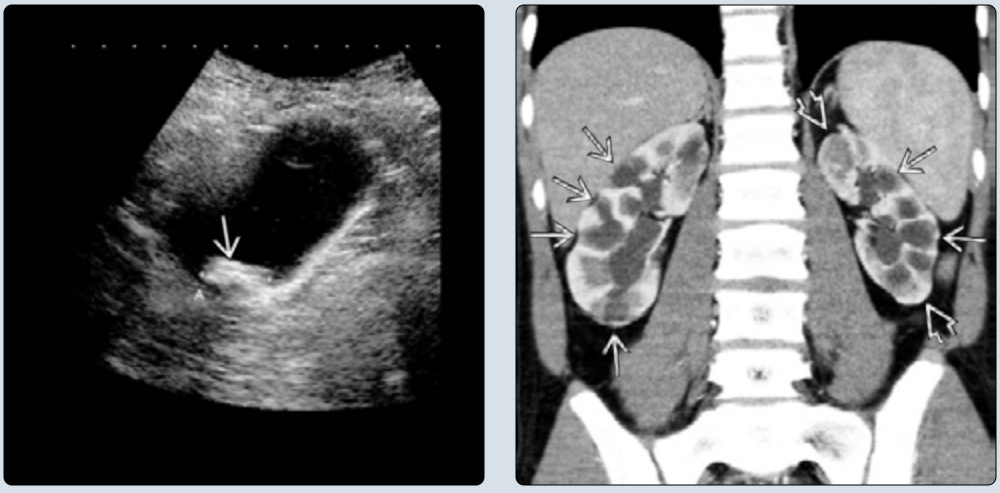
CLINICAL ISSUES

- Incidence of 1:10,000-1:50,000 live births
- 2.3-6:1 = M:F
- Modern staged repair vs. complete primary repair
  - Continence achieved in up to 81%
  - Most perform clean intermittent catheterization

(Left) *AP pelvic radiograph in a neonate with bladder exstrophy shows lobular soft tissues of the open bladder plate with splayed pubic & ischial bones. Note the normal sacrum. This can help differentiate this entity from cloacal exstrophy in which the sacrum is dysplastic or hypoplastic.* **(Right)** *Frontal VCUG in a 3-year-old girl after bladder exstrophy repair shows a small lobulated bladder with elevated bladder base, J- shaped ureterovesical junctions, & bilateral high-grade vesicoureteral reflux (VUR).*



(Left) *Transverse US of the bladder in an adolescent with a history of bladder exstrophy shows a large echogenic stone with posterior shadowing at the dependent portion of the bladder. These may be treated by extracorporeal shock wave lithotripsy. Note the irregular bladder wall.* **(Right)** *Coronal CECT in a 17-year-old boy with a history of repaired bladder exstrophy shows multifocal renal scars bilaterally with a globally scarred small left kidney secondary to VUR of infected urine after bladder closure.*



TERMINOLOGY

Synonyms

- Exstrophy-epispadias complex

Definitions

- Classic bladder exstrophy (CBE): Low midline abdominal wall defect (AWD) with exposure of bladder plate & urethra + low-set umbilicus
  - Split lower abdominal skin & rectus abdominis + pubic symphysis diastasis
  - Bifid clitoris in girls, epispadias in boys
- Epispadias: Abnormal dorsal urethral opening
  - In CBE, entire urethra open with abnormal bladder sphincter

IMAGING

General Features

- Best diagnostic clue
  - Absent urinary bladder on prenatal US but normal amniotic fluid
  - Pubic symphysis diastasis on postnatal radiographs

Radiographic Findings

- Pubic symphysis > 1 cm
- Lobular lower abdominal wall soft tissue from everted bladder
- ± inguinal hernias (with gas-filled bowel within hernia)

Fluoroscopic Findings

- Voiding cystourethrogram
  - Performed after bladder closure
  - High rate of vesicoureteral reflux (VUR)
  - Variable bladder capacity, shape
  - Postop findings: Bladder rupture, urethral stricture/fistula

CT Findings

- External rotation of pubic & iliac bones, pubic bone shortening, acetabular retroversion

Ultrasonographic Findings

- Prenatal
  - Nonvisualized urinary bladder but normal amniotic fluid
  - Low position of umbilicus
  - Lobular soft tissue at infraumbilical abdominal wall
  - Diminutive genitalia
  - Splayed echogenic pubic & iliac bones
- Postnatal
  - Variable bladder size, shape, & wall thickness post repair
  - VUR → pelvocaliectasis ± pyelonephritis & scarring

Nuclear Medicine Findings

- DMSA to evaluate split function & scarring

MR Findings

- Assessment of pelvic floor musculature deficiencies

Imaging Recommendations

- Best imaging tool
  - Prenatal ultrasound/MR, postnatal clinical exam

DIFFERENTIAL DIAGNOSIS

Cloacal Exstrophy/OEIS Complex

- Low midline AWD
- Absent bladder with normal amniotic fluid
- Exposed bladder halves divided by everted cecum
  - ± elephant trunk appearance of prolapsed ileum
- Omphalocele, imperforate anus, spine anomalies

Omphalocele

- Central AWD
- Bowel & other viscera herniate into umbilical cord base
  - Contents covered by sac
- Lower abdominal wall & urinary bladder intact

Gastroschisis

- Off-midline AWD: Bowel herniated right of umbilicus
  - No covering membrane
- Lower abdominal wall & bladder intact

Impaired Urine Production (Absent Bladder)

- Bilateral renal abnormalities (same or combination) → oligo-/anhydramnios → pulmonary hypoplasia
  - Multicystic dysplastic kidney
  - Ureteropelvic junction obstruction
  - Polycystic renal disease, recessive
  - Renal agenesis

PATHOLOGY

General Features

- Cloacal membrane rupture due to failed mesodermal ingrowth

CLINICAL ISSUES

Demographics

- Gender
  - M:F = 2.3-6:1
- Epidemiology
  - 1:10,000-1:50,000 live births

Natural History & Prognosis

- Continence in up to 81%: Most clean (± intermittent catheterization)

Treatment

- Modern staged repair
  - Stage I, newborn: Close AWD, bladder, & posterior urethra + iliac osteotomy
  - Stage II, age 6-12 months: Epispadias repair
  - Stage III, age 4-5 years: Reconstruct bladder neck + ureteral reimplants
- Complete primary repair ± ureteral reimplant

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Renal Ectopia and Fusion

KEY FACTS

TERMINOLOGY

- Includes horseshoe or pancake kidney; crossed fused ectopia; pelvic, iliac/pelvic (ptotic/mobile), & thoracic kidneys
- Results from abnormal ascent & rotation of fetal kidney
- Found anywhere from presacral to intrathoracic; may be bilateral or unilateral; may cross midline
- Malpositioned kidneys more susceptible to trauma, iatrogenic injury, obstruction, infection, & stones

IMAGING

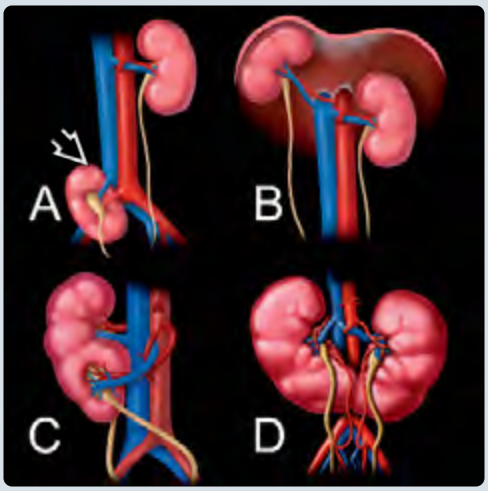
- US typically sufficient to document location & gross morphology of ectopic & fused kidneys
  - Accurate lengths difficult to obtain due to more globular, less reniform shape & poor definition of margin with contralateral kidney
- Other modalities used to answer specific questions (as needed): Drainage, stones, vascular supply, ureteral course
- Isthmus of horseshoe kidney may contain functioning renal tissue or fibrotic nonfunctional tissue
- Ureteral insertion site in bladder provides clue to site where kidney initially formed (i.e., lower pole ureter of crossed fused ectopia inserts into trigone on contralateral side)
- Colon typically occupies empty renal fossa

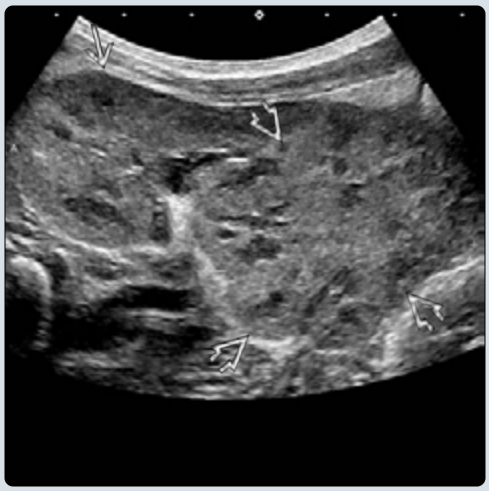
PATHOLOGY

- Horseshoe kidneys associated with genital anomalies, VACTERL, Turner, & other syndromes
- Adrenal ectopia reported in association with renal ectopia
- Vesicoureteral reflux (20-30%), contralateral renal dysplasia (4%), cryptorchidism (5%), hypospadias (5%)

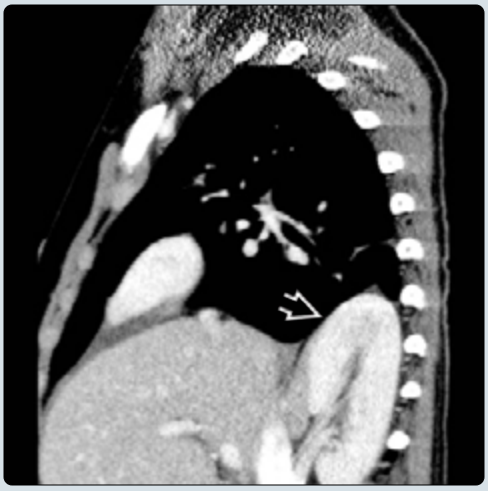
CLINICAL ISSUES

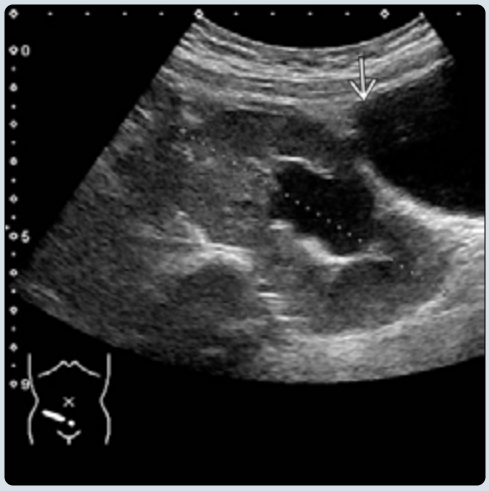
- Horseshoe kidney most common: 1 in 400 births
- All types of ectopia more common in boys than girls
- Primary concern: Avoidance of iatrogenic injury to renal parenchyma & supplying vessels during routine surgery
- Treat complications of obstruction, reflux, & stones





(Left) *Graphic shows variations of renal ectopia & fusion: (A) Pelvic kidney ⇨, (B) subdiaphragmatic/thoracic kidney, (C) crossed fused renal ectopia, & (D) horseshoe kidney.* **(Right)** *Longitudinal US shows a crossed fused renal ectopia in the left abdomen with a relatively normal upper moiety ⇨ & a malrotated, globular lower moiety ⇨. Note that the long axis of each moiety is different, which helps distinguish this entity from a duplication. Also, no renal tissue will be seen in the contralateral renal fossa in this setting.*





(Left) *Sagittal CECT shows a right kidney ⇨ bulging into the posterior right lung base in this patient with a subdiaphragmatic or thoracic kidney.* **(Right)** *Longitudinal US shows a pelvic kidney (between cursors) abutting the bladder dome ⇨ in the right lower quadrant. It is easy to imagine this kidney being injured during a laparoscopic appendectomy.*

TERMINOLOGY

Synonyms

- Includes horseshoe or pancake kidney; crossed fused ectopia; pelvic, iliac/pelvic (ptotic/mobile), & thoracic kidneys

Definitions

- Normal renal tissue in abnormal location
- Results from abnormal ascent & rotation of fetal kidney
- Kidneys form at sacral level & ascend to L1 by term; renal pelvis initially directed anteriorly but rotates 90° medially as it ascends
- Malpositioned kidneys more susceptible to trauma, iatrogenic injury, obstruction, infection, & stones
- High incidence of multiple renal arteries & veins

IMAGING

General Features

- Best diagnostic clue
  - Normal renal parenchyma with abnormal location, axis of orientation, or position of renal pelvis
- Location
  - Anywhere from presacral to intrathoracic, bilateral or unilateral; may cross midline
- Size
  - Overall renal parenchyma volume similar to orthotopic kidneys; longitudinal measurements vary based on ectopic kidney shape
- Morphology
  - Normal cortex, pyramids, & collecting system; however, pelvocaliectasis commonly associated
  - Isthmus or midline junctional zone of horseshoe kidney may contain functioning renal tissue or fibrotic nonfunctional tissue

Radiographic Findings

- Radiography
  - Renal shadows altered from expected positions
  - May simulate midline or pelvic mass
  - Bowel gas may be
    - Displaced by ectopic kidney
    - Occupying renal fossa (with repositioning of splenic & hepatic flexures of colon most commonly)
- IVP
  - Functional renal parenchyma in atypical location; pelvocaliectasis commonly associated
  - Ectopic kidneys, especially thoracic & horseshoe varieties, may appear mass-like on initial scout view
    - Expect bowel to occupy empty renal fossa
  - Early nephrogram may be missed on tightly coned films; not included in field of view
  - Abnormalities of vasculature & ureter also common
    - Ureteral insertion site in bladder provides clue to where kidney initially formed (i.e., lower pole ureter of crossed fused ectopic kidney inserts into trigone on contralateral side)
  - Oblique views often helpful to profile abnormally rotated collecting system
  - May require fluoroscopic spot views to capture ureteral course
  - Pelvic compression devices should be avoided

Fluoroscopic Findings

- Renal ectopia may be noted incidentally during other fluoroscopic procedures: Gastrointestinal tract studies, voiding cystourethrogram (VCUG), or genitograms

Ultrasonographic Findings

- Grayscale ultrasound
  - Normal echotexture of abnormally positioned renal parenchyma
    - Hydronephrosis & scarring may alter echotexture & architecture
  - Accurate length measurements difficult to obtain due to more globular, less reniform shape & poor definition of margin with contralateral kidney
    - Volumes can be helpful
  - May be difficult to see pelvic kidneys due to adjacent bowel gas
  - Colon typically occupies empty renal fossa
- Color Doppler
  - Useful in detecting aberrant vessels & localizing urinary bladder ureteral jets
- Power Doppler
  - Useful in cases of pyelonephritis where parenchymal perfusion ↓ in infected segments

CT Findings

- NECT
  - May initially be seen on renal stone CT
  - Soft tissue "mass" identified along with absence of renal tissue in renal fossa
  - Associated hydronephrosis or stone disease frequent clue to aberrant renal position
    - Look for aberrant ureteral course during stone assessment
- CECT
  - Look for abnormal location, rotation, axis, & ureteral course
  - Expect normal renal enhancement & excretion of contrast, except in obstruction cases
  - Delayed imaging may be useful, depending on study indication
    - To assess parenchyma & collecting system for laceration in trauma setting
    - To localize distal ureters
- CTA
  - Occasionally used for mapping vessels preoperatively

MR Findings

- Similar to other modalities: Abnormal location, axis, & rotation

Angiographic Findings

- Reserved for renal donors or patients undergoing surgery who have renal ascent & rotation abnormalities not clearly defined by other imaging modalities

Nuclear Medicine Findings

- May be found incidentally on renal imaging studies or whole-body exams performed for unrelated reasons

- Nuclear renal studies sometimes requested specifically to document presence of pelvic kidney that could not be appreciated on US due to intervening bowel gas
- Ectopic kidneys show normal uptake of radiopharmaceutical with variable degrees of pelvocaliectasis

Imaging Recommendations

- Best imaging tool
  - US typically sufficient to document location & gross morphology of ectopic & fused kidneys
  - Other modalities used to answer specific questions (as needed): Drainage, stone disease, vascular supply

DIFFERENTIAL DIAGNOSIS

Mass in Typical Ectopic Location

- Renal tissue still visible in normal locations
- Thoracic considerations
  - Bronchopulmonary sequestration
  - Neuroblastoma
  - Hernia/eventration
- Abdominal
  - Lymphoma
  - Any large neoplasm or cyst of solid organ, viscus, or mesentery
- Pelvis
  - Ovarian tumors
  - Sacrococcygeal teratoma
  - Pelvic rhabdomyosarcoma

Pseudokidney of Intussusception

- Depending on image orientation, ileocolic intussusception can mimic reniform shape & architecture on US
- Real-time cross-sectional sweep or cine through entire length of intussusception will reveal nature of "mass"

Simulated Ectopia Related to Severe Kyphoscoliosis

- Mimics horseshoe kidney or crossed fused ectopia but lacks connection between right & left kidneys
- Cross-sectional imaging or orthogonal views clarify difference

PATHOLOGY

General Features

- Genetics
  - Horseshoe kidneys associated with genital anomalies, VACTERL (vertebral, anorectal, cardiac, tracheoesophageal, renal, limb abnormalities), Turner, & other syndromes
  - Other abnormalities of ascent & rotation less frequently associated with syndromes
  - Geographic "hot spots" for ectopia suggest either common exposure to teratogenetic factors or hereditary condition with variable penetrance
- Associated abnormalities
  - Urologic abnormalities associated with simple ectopia
    - Vesicoureteral reflux (20-30%)
    - Contralateral renal dysplasia (4%)
    - Cryptorchidism (5%)
    - Hypospadias (5%)
  - Adrenal ectopia reported in association with renal ectopia
  - Cardiac & skeletal anomalies common
- Parenchyma developmentally normal, though secondary changes of obstruction, scarring, & nephrolithiasis not uncommon
- Embryology/anatomy
  - Results from abnormal ascent & rotation of metanephric blastema after induction by ureteric bud
  - Multiple supplying vessels & draining ureters common

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often discovered incidentally
  - May be suspected on antenatal US
  - Can present later in infancy as palpable mass or with UTI or obstruction

Demographics

- Gender
  - All types of ectopia more common in boys than girls
- Epidemiology
  - Horseshoe kidney incidence: 1 in 400 births; most common fusion anomaly
  - Crossed fused ectopic kidney less common
  - Simple ectopia seen in 1 in 900 (autopsy series)

Natural History & Prognosis

- Aside from complications of obstruction, stone formation, UTI, & injury, most ectopic kidneys function normally
- Primary concern: Avoidance of iatrogenic injury to renal parenchyma & supplying vessels during routine surgery, especially laparoscopic surgery
- Slightly ↑ risk of Wilms & carcinoid tumors reported in horseshoe kidneys
- Prognosis generally excellent
  - 1/3 of patients with horseshoe kidney asymptomatic throughout life

Treatment

- Treat complications of obstruction, reflux, & stones

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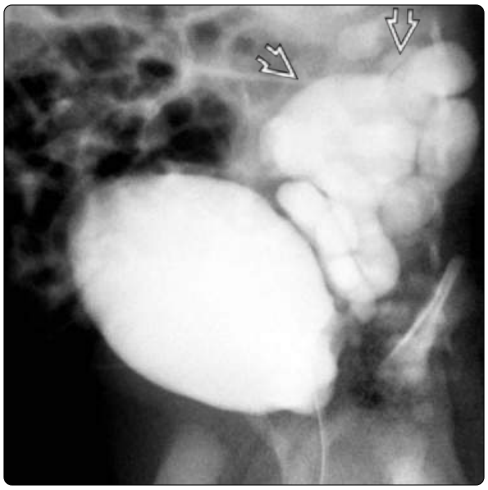
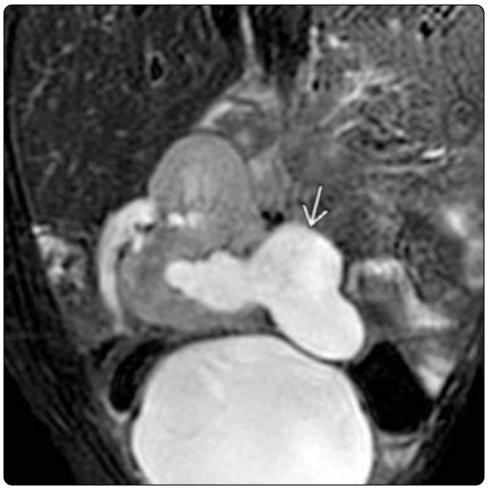
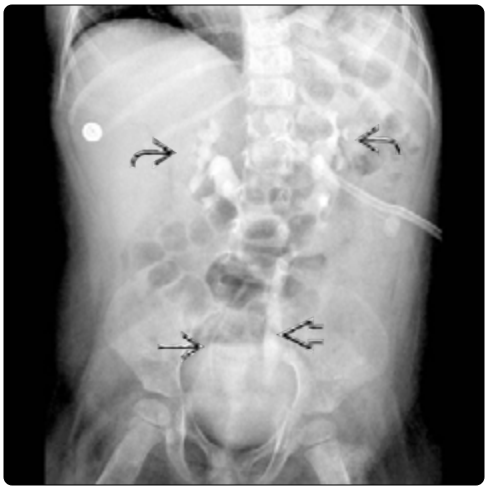
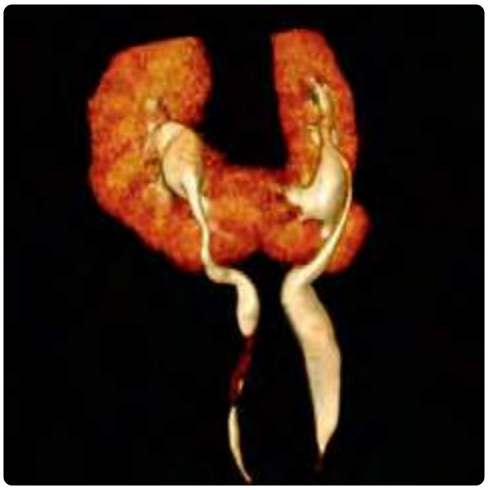


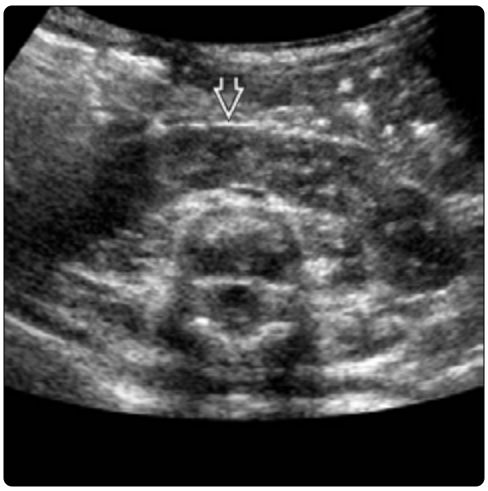


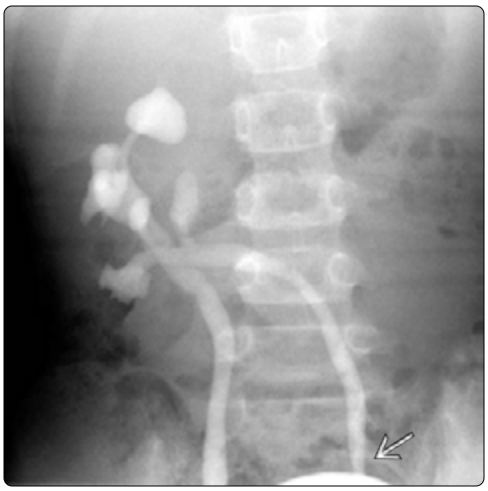
(Left) *Oblique voiding cystourethrogram (VCUG) shows grade 5 vesicoureteral reflux into the left kidney, which is located in the pelvis. Pelvic kidneys are sometimes called "ptotic," as if they fell down from the renal fossa; however, their location is actually due to a failure of ascent during fetal life.* **(Right)** *Coronal T2 FS MR urography shows a dilated collecting system & tortuous ureter in this case of a pelvic kidney.*





(Left) *Frontal IVP shows abnormally rotated & fused renal parenchyma crossing the midline in this case of horseshoe kidney. Horseshoe kidneys may have several ureters & renal vascular pedicles. This patient has 2 ureters, the right being normal in caliber & the left being slightly dilated.* **(Right)** *3D reformation from a CECT scan in a patient with multiple congenital anomalies shows a horseshoe kidney with fused lower poles & anteriorly directed renal pelves.*





(Left) *Transverse US in a patient with a horseshoe kidney shows a band of renal parenchyma crossing over the spine.* **(Right)** *Frontal fluoroscopic image during a VCUG shows a crossed fused kidney with the lower moiety ureter crossing the midline to the left side of the bladder (since this kidney initially formed as a left kidney).*

Renal Agenesis

KEY FACTS

TERMINOLOGY

- Congenital absence of functioning renal tissue
- **C**ongenital **a**bnormality of **k**idney & **u**rinary **t**ract (CAKUT)

IMAGING

- Complete absence of 1 or both kidneys
  - No ectopia or fusion anomaly
- Bowel fills renal fossa
- Linear configuration of ipsilateral adrenal gland
  - "Lying down adrenal" rarely mistaken for abnormal kidney in neonate
- ± compensatory hypertrophy of remaining kidney
- Best modality: Ultrasound (pre- or postnatal)

TOP DIFFERENTIAL DIAGNOSES

- Involuted multicystic dysplastic kidney
- Ectopic kidney
- Crossed fused renal ectopia
- Prior nephrectomy

PATHOLOGY

- Unilateral renal agenesis associated with
  - Müllerian abnormalities
    - **O**bstructed **h**emi**v**agina, **i**psilateral **r**enal **a**genesis (OHVIRA syndrome)
  - Contralateral renal anomalies in 31-48%
  - Extrarenal anomalies in up to 30%
- Bilateral renal agenesis results in Potter sequence: Oligo-/anhydramnios, lung hypoplasia, dysmorphic facies, ± fetal demise

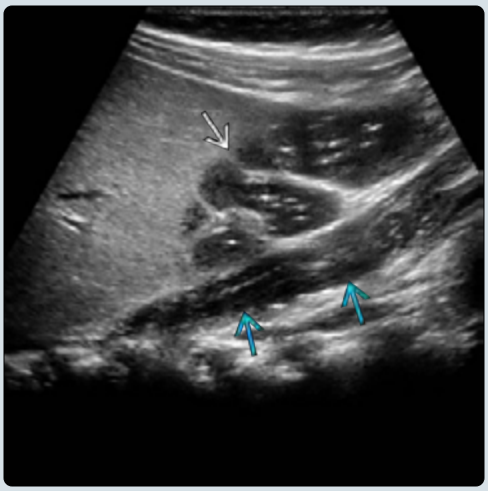
CLINICAL ISSUES

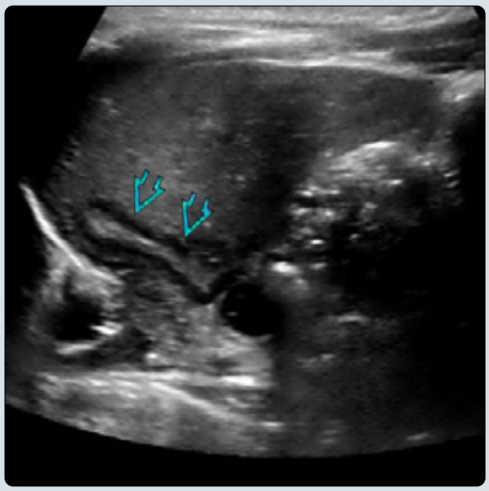
- Unilateral usually asymptomatic; associated with medical renal disease later in life
- Bilateral typically fatal with respiratory distress, pneumothoraces, bell-shaped chest, & anuria at delivery

DIAGNOSTIC CHECKLIST

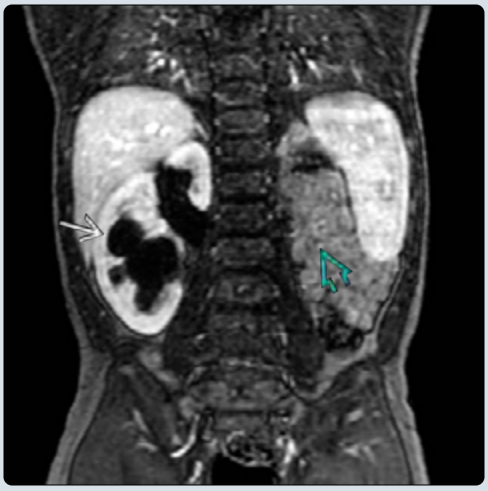
- Voiding cystourethrogram for contralateral reflux

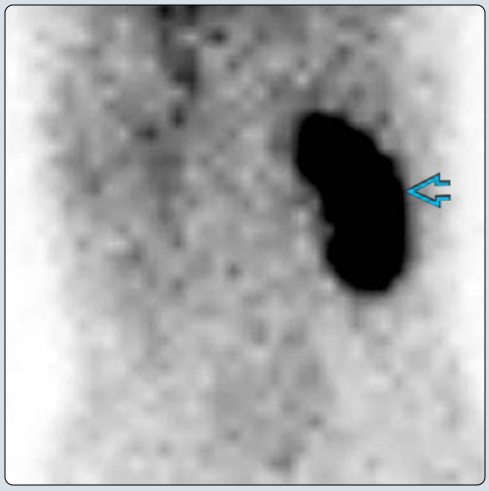
(Left) *Longitudinal US of the right renal fossa in a 1-day-old girl with right renal agenesis shows absence of the kidney in the expected location along the right psoas muscle.* *There are normal, fluid-filled bowel loops in the right renal fossa.* **(Right)** *Oblique US of the right renal fossa in a 1-day-old boy with right renal agenesis shows an elongated, linear adrenal gland (lying down adrenal).*





(Left) *Coronal T1 C+ FS MR in a 3 year old with left renal agenesis shows an absence of renal tissue in the left renal fossa (which is filled with decompressed bowel).* *The right renal collecting system is dilated secondary to a ureteropelvic junction obstruction.* **(Right)** *Posterior coronal image from a Tc-99m DTPA renal scan in a 2 year old with left renal agenesis shows normal radiotracer uptake in the right kidney with no radiotracer uptake on the left.*





TERMINOLOGY

Definitions

- Congenital absence of functioning renal tissue

IMAGING

General Features

- Best diagnostic clue
  - US, CT, MR: Absence of kidney in renal fossa without fusion anomaly or ectopia + associated abnormal linear configuration of ipsilateral adrenal gland
    - Lying down adrenal sign

Radiographic Findings

- Radiography
  - Unilateral: Absence of normal renal shadow
  - Bilateral: Newborn with respiratory distress & abnormal facies shows chest sequelae of pulmonary hypoplasia
    - Small, bell-shaped thorax
    - Pneumothoraces ± pneumomediastinum

Ultrasonographic Findings

- Absent kidney(s) without fusion anomaly or ectopia
- Ipsilateral elongated linear adrenal gland
  - Rarely mistaken for dysplastic kidney in neonate
- Unilateral: ± compensatory hypertrophy of remaining kidney
- Bilateral: Typically detected by prenatal US
  - Oligo-/anhydramnios + lack of urinary bladder distention
  - No identifiable renal tissue in either renal fossa
  - No renal arteries by Doppler
  - No renal ectopia

Nuclear Medicine Findings

- Renal scans show lack of renal uptake & excretion

Imaging Recommendations

- Best imaging tool
  - Ultrasound (prenatal or postnatal)
- Protocol advice
  - Renal function should be screened prior to CT/MR IV contrast administration if renal anomalies known/suspected
  - Consider pelvic imaging in girls due to association with müllerian abnormalities

DIFFERENTIAL DIAGNOSIS

Involuted Multicystic Dysplastic Kidney

- Gradual atrophy of reniform collection of cysts

Ectopic Kidney

- Often located in pelvis

Crossed Fused Renal Ectopia

- Contralateral kidney normally positioned with ectopic kidney lying across midline & fused to lower pole
- Divergent long axes of 2 fused kidneys
- Separate ureters insert normally into urinary bladder

Prior Nephrectomy

- Due to renal tumor, severe trauma, obstructed nonfunctioning kidney

PATHOLOGY

General Features

- Etiology
  - Renal development begins in 5th week of gestation
  - Agenesis results from abnormal interaction between ureteric bud of mesonephric duct & metanephric mesenchyme
- Associated abnormalities
  - Unilateral renal agenesis
    - Müllerian abnormalities in girls
      - **O**bstructed **h**emi**v**agina, **i**psilateral **r**enal **a**genesis (OHVIRA syndrome)
      - May be due to ectopic insertion of ureter with resultant obstruction & renal aplasia
      - Typically with uterine didelphys configuration
    - Contralateral renal abnormalities in 31-48%
      - Vesicoureteral reflux most common
      - Others: Contralateral ureteropelvic junction obstruction, megaureter, collecting system duplication
    - Extrarenal abnormalities present in up to 30%
  - Bilateral agenesis → Potter sequence/syndrome: Oligohydramnios, lung hypoplasia, dysmorphic facies

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Unilateral agenesis usually asymptomatic
  - Bilateral agenesis typically presents in utero with oligohydramnios & pulmonary hypoplasia ± clubfeet, fetal demise
    - At delivery → respiratory distress, abnormal facies, anuria

Demographics

- Epidemiology
  - Overall incidence ~ 1/2,000
  - Incidence based on prenatal diagnosis only ~ 1/8,000
    - Difference may be due to confusion of involuted multicystic dysplastic kidney with true renal agenesis

Natural History & Prognosis

- Unilateral renal agenesis associated with medical renal disease later in life
  - ↑ incidence of hypertension, impaired glomerular filtration rate, & proteinuria
    - May be due to hyperfiltration by remaining kidney
- Bilateral renal agenesis typically incompatible with life
  - Serial amnioinfusions may create "pulmonary survivor" to be sustained by peritoneal dialysis until renal transplant

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Multicystic Dysplastic Kidney

KEY FACTS

TERMINOLOGY

- Congenital nonfunctional kidney replaced by multiple cysts & dysplastic tissue
- Tend to involute with time: Cysts shrink & residual tissue may lose reniform shape

IMAGING

- Reniform-shaped multicystic mass occupying renal fossa
  - ± lobulated outer contour (due to cysts of variable size)
  - Wide range of sizes: Up to 15 cm in length in newborn period; may be only 1-2 cm after years of involution
- Cysts of varying size do not connect
  - Largest cyst typically peripheral, not central
- Poorly defined intervening echogenic parenchyma without normal corticomedullary architecture
- Can be segmental in duplicated kidneys
- Nuclear scintigraphy documents lack of renal function in MCDK

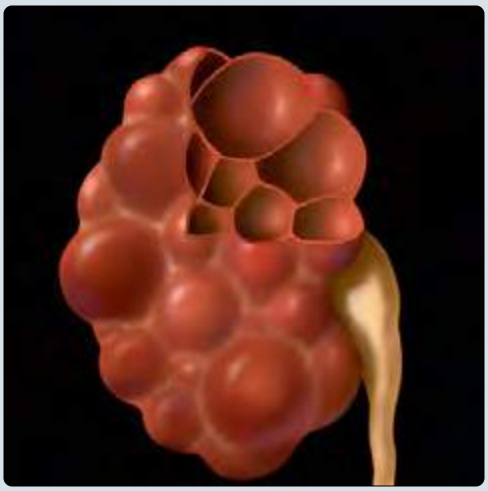
PATHOLOGY

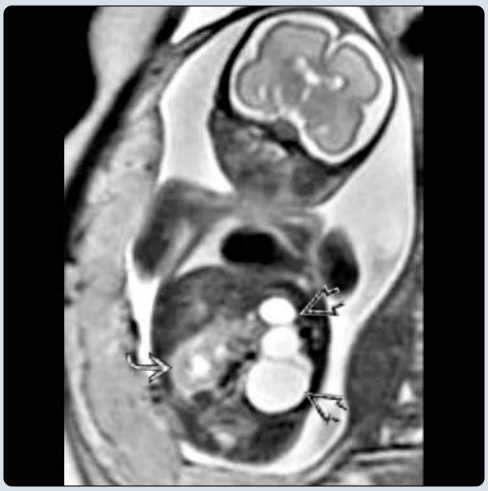
- Probably due to atresia of ureter or ureteropelvic junction (UPJ) during metanephric stage of intrauterine development
- Up to 40% of patients with MCDK have contralateral renal abnormality
  - UPJ obstruction & VUR most common

CLINICAL ISSUES

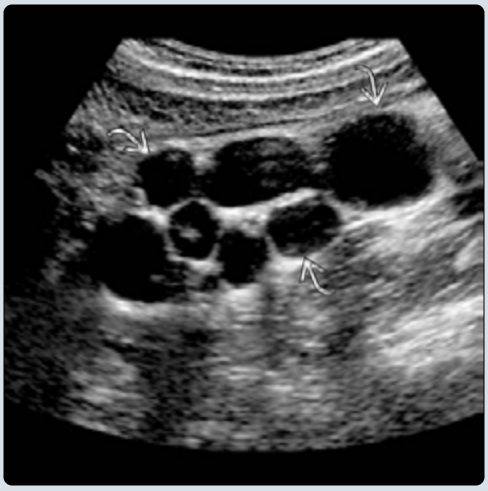
- MCDK 2nd most common abdominal mass in neonate (after hydronephrosis)
- > 50% discovered antenatally or in infancy as palpable mass
- Unilateral MCDK with normal contralateral kidney: Excellent prognosis
  - Vast majority involute with time & remain asymptomatic
  - Rare reports of Wilms tumor developing in MCDK
- Unilateral MCDK with abnormal contralateral kidney: May develop renal insufficiency
- Bilateral MCDK: Incompatible with life

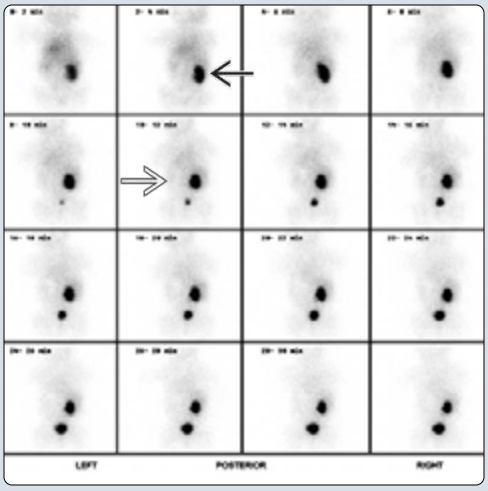
(Left) *Frontal graphic of the right kidney shows multiple cysts of varying size replacing the renal parenchyma. There is minimal intervening dysplastic renal tissue. A ureter may or may not be recognizable at the renal hilum on imaging studies.* **(Right)** *Coronal SSFSE T2 fetal MR shows a multicystic mass in the left renal fossa ⇨ with no discernible normal left renal tissue, most consistent with a left multicystic dysplastic kidney (MCDK). The right kidney ⇨ & volume of amniotic fluid appear normal.*





(Left) *Prone postnatal US of the left flank in the same patient shows multiple cysts ⇨ of varying size replacing the left kidney. A reniform shape is retained, but no normal left renal parenchyma is seen.* **(Right)** *Posterior images of the same infant during a Tc-99m MAG3 renal scan show normal function of the right kidney ⇨ but no function on the left side ⇨, confirming a left MCDK. Early transient activity in MCDK on nuclear scans merely reflects that the tissue is being perfused, but continued images show no function.*





TERMINOLOGY

Abbreviations

- Multicystic dysplastic kidney (MCDK)

Definitions

- Congenital nonfunctional kidney replaced by multiple cysts & dysplastic tissue
- 2 forms of MCDK generally recognized
  - Pelvoinfundibular (more common): Theoretically results from atresia of ureter or renal pelvis; cysts are remnants of dilated calyces
  - Hydronephrotic (occurs less frequently): Results from atretic segment of ureter; cysts replace entire pelvocalyceal system
- Tend to involute with time: Cysts shrink & residual tissue may lose reniform shape

IMAGING

General Features

- Best diagnostic clue
  - Cysts & dysplastic echogenic tissue replace entire kidney
  - Nuclear scintigraphy documents lack of renal function
    - If some delayed excretion present, consider poorly functioning hydronephrosis
- Location
  - Renal fossa most common but can occur ectopically from pelvis to chest
- Size
  - Wide range: Up to 15 cm in length in newborn period; may be only 1-2 cm after years of involution
- Morphology
  - Numerous cysts of varying size with dysplastic intervening parenchyma
  - No recognizable normal corticomedullary architecture
  - Renal shape typically preserved, though cysts may result in lobulated contour
  - Can be segmental in duplicated kidneys

Radiographic Findings

- Radiography
  - Indirect evidence: Space-occupying lesion in flank
- IVP
  - No enhancement of or excretion from MCDK
  - May see transient blush during contrast bolus infusion due retained vascularity

Ultrasonographic Findings

- Grayscale ultrasound
  - Reniform-shaped multicystic mass occupying renal fossa
    - ± lobulated outer contour (due to cysts of variable size)
  - Cysts of varying size do not connect
  - Largest cyst typically peripheral rather than central
  - Poorly defined intervening echogenic parenchyma without normal corticomedullary architecture
- Color Doppler
  - Minimal, if any, vascularity in parenchyma; central hilar vessels tend to be small
- Often followed with annual US scans to
  - Assess growth of contralateral kidney
  - Confirm involution of MCDK
    - Watch for unusual growth of MCDK that has been reported with Wilms tumor arising in these lesions

CT Findings

- NECT
  - Low-attenuation cysts (though some may contain debris) replacing normal renal parenchyma
- CECT
  - No parenchymal contrast enhancement
  - No contrast excretion on delayed images
  - No hydronephrosis

MR Findings

- Cysts replacing normal renal parenchyma
- Usually noted incidentally on MR performed for other indications

Nuclear Medicine Findings

- MAG3 scintigraphy
  - Initial blood flow images may show slight perfusion of MCDK but sequential images show lack radiotracer uptake or excretion
- Tc-99m DMSA renal cortical scan
  - Radiotracer may localize to scattered tubular cells in dysplastic parenchyma

Other Modality Findings

- Retrograde ureterogram will show blind-ending ureter
  - Different from rapid change in caliber of ureter & communication with calyces seen in ureteropelvic junction (UPJ) obstruction or other causes of hydronephrosis

Imaging Recommendations

- Best imaging tool
  - Ultrasound for initial identification
  - Nuclear scan to confirm nonfunction of suspected MCDK & assess drainage of contralateral kidney

DIFFERENTIAL DIAGNOSIS

Hydronephrosis

- "Cysts" (dilated calyces) all relatively uniform in size & connect centrally
- Largest "cyst" (renal pelvis) lies central

Congenital Mesoblastic Nephroma

- Solid or mixed solid & cystic neonatal/infantile renal mass
- "Claw" of renal tissue splayed along tumor margin

Wilms Tumor

- Predominantly large solid heterogeneous round mass with "claw" of renal tissue splayed along tumor margin

Multilocular Cystic Nephroma

- Discrete round renal mass in males 3 months to 2 years of age
- Numerous thin-walled cysts throughout mass with central herniation of cysts
- "Claw" of renal tissue splayed along tumor margin

Tuberous Sclerosis

- Numerous cysts &/or angiomyolipomas bilaterally

Autosomal Recessive Polycystic Kidney Disease

- Enlarged, echogenic bilateral kidneys with striated parenchyma & intermixed small cysts

End-Stage Renal Disease

- Bilateral kidneys tend to be small & echogenic, resembling involuted MCDK
- Cysts may be present from underlying disease or secondary to dialysis

PATHOLOGY

General Features

- Etiology
  - Probably due to atresia of ureter or UPJ during metanephric stage of intrauterine development
- Genetics
  - Generally considered sporadic; reported familial cases of autosomal dominant inheritance with variable expression & penetrance
- Associated abnormalities
  - Genitourinary abnormalities in 25-40%
    - Up to 40% of patients have contralateral abnormality
      - UPJ obstruction & vesicoureteral reflux (VUR) most common
    - Megaureter
    - Cystic dysplasia of testis
    - VUR in 12-26%
  - Nonurologic abnormalities
    - Cardiac & musculoskeletal most common
  - Associated syndromes: Turner syndrome, trisomy 21, chromosome 22 deletions, Waardenburg syndrome, others

Gross Pathologic & Surgical Features

- Walls of cysts vary in thickness; fibrotic dysplastic tissue replaces normal renal stroma; may be quite large & nonreniform in shape

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - > 50% discovered antenatally or in infancy as palpable mass
  - Can have delayed presentation as incidental finding when
    - Symptoms of contralateral UPJ obstruction evaluated
    - Patient evaluated for urinary tract infection
    - Patient imaged for traumatic injury or spine abnormality

Demographics

- Age
  - Congenital abnormality, usually presents in infancy
- Gender
  - M = F
- Incidence
  - 2nd most common abdominal mass in neonate
    - Hydronephrosis most common
  - 0.03% in autopsy series
  - 1 in 4,300 live births

Natural History & Prognosis

- Unilateral MCDK with normal contralateral kidney: Excellent prognosis
  - Vast majority involute with time & remain asymptomatic
    - ~ 50% involute in 1st decade
    - MCDK > 5 cm in newborn less likely to involute
  - Can have complications of infection or mass effect
  - Rare reports of hypertension with unilateral MCDK
    - In patients treated with nephrectomy for hypertension, blood pressure normalized in only 50%, suggesting other sources
  - Rare reports of Wilms tumor developing in MCDK
- Unilateral MCDK with abnormal contralateral kidney
  - If contralateral kidney has delayed diagnosis of UPJ or ureterovesical junction obstruction or VUR, renal insufficiency can be problem
  - VUR into contralateral kidney associated with smaller renal size during 1st year of life; may lead to recurrent pyelonephritis, scarring, hypertension
- Bilateral MCDK: Incompatible with life

Treatment

- Surgical excision when complicated by focal enlargement (potential Wilms tumor), recurrent infections, mass effect, or hypertension
- Otherwise, serial ultrasounds for 3-5 years to monitor

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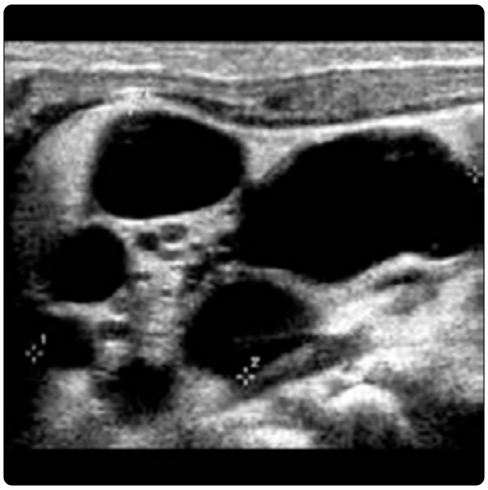
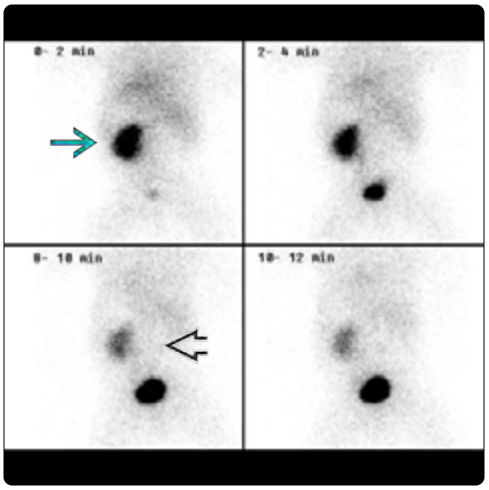
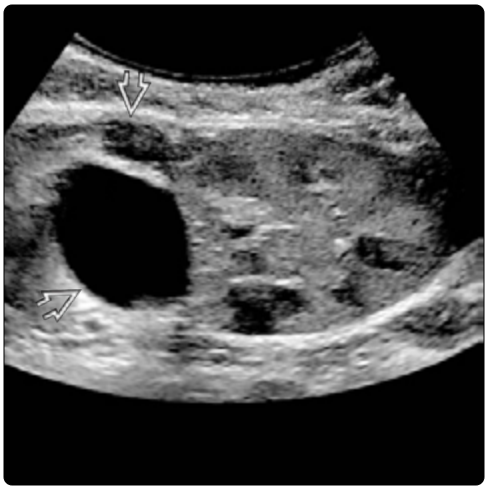
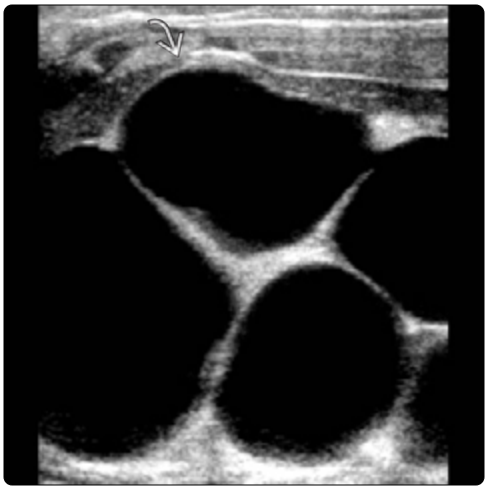


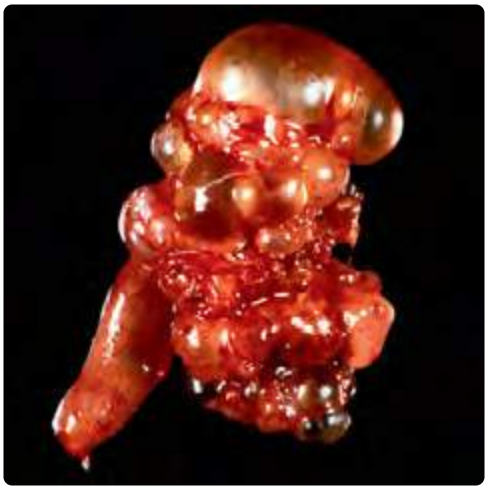


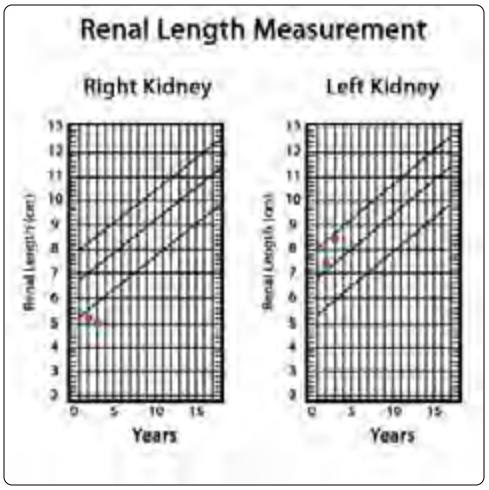
(Left) *Longitudinal US shows cursors roughly marking the borders of a MCDK. Note that the largest cyst is in the lower pole, not in the expected location of the renal pelvis (helping to differentiate MCDK from hydronephrosis). Only echogenic dysplastic tissue is visualized between the cysts.* **(Right)** *Posterior images from a Tc-99m MAG3 renal scan show normal function & excretion of only the left kidney ⇨ in this patient with a right-sided MCDK ⇨. Nuclear renal scans are used to confirm the lack of functioning renal tissue.*





(Left) *Longitudinal US shows an unusual case of a segmental MCDK in an infant noted to have cysts on a prenatal scan. The 2 cysts in the upper pole of the kidney ⇨ do not communicate, & there is echogenic solid parenchyma between the cysts. The lower pole is normal. The contralateral kidney in this infant was congenitally absent.* **(Right)** *Longitudinal US shows near complete replacement of the renal parenchyma ⇨ with cysts in this MCDK. No normal renal tissue is visualized.*





(Left) *Gross pathology shows numerous cysts replacing the renal parenchyma & distorting the contour of this MCDK. Note that the largest cyst in this specimen is in the upper pole, not in the renal hilum.* **(Right)** *This chart measures renal growth over serial exams in children. This particular patient has a shrinking right-sided MCDK & a rapidly growing normal left kidney, which would be expected to show compensatory hypertrophy.*

Polycystic Kidney Disease, Autosomal Recessive

KEY FACTS

TERMINOLOGY

- Single gene ciliopathy with marked bilateral renal enlargement due to dilated distal tubules & collecting ducts
- Synonyms: Autosomal recessive polycystic kidney disease (ARPKD), infantile polycystic kidney disease

IMAGING

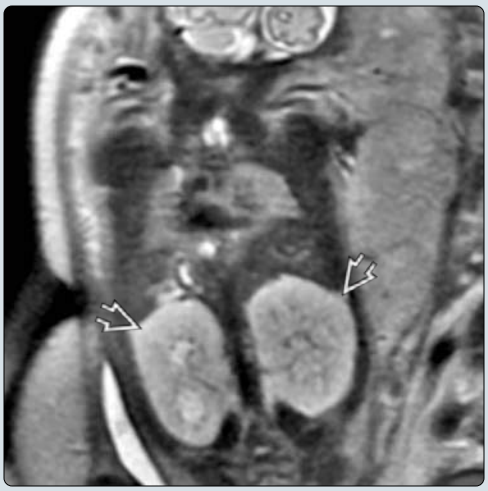
- Radiographs: Bilateral flank "masses" bulging lateral abdominal contours & displacing bowel gas centrally
  - Pulmonary hypoplasia & history of oligohydramnios
    - Bell-shaped thorax ± pneumothorax, pneumomediastinum
- US: Bilaterally enlarged echogenic kidneys in newborn with loss of corticomedullary differentiation
  - 2-6 standard deviations (SD) above mean size for age
  - Dilated, radially arranged tubules on high-resolution linear transducers ± small cysts (< 1 cm)
  - Tiny, punctate hyperechoic foci (likely calcium deposits) develop with time & correlate with renal failure
- MR: Large kidneys of diffusely high signal intensity on T2
  - Intervening low-signal septa
- Variable degrees of liver disease

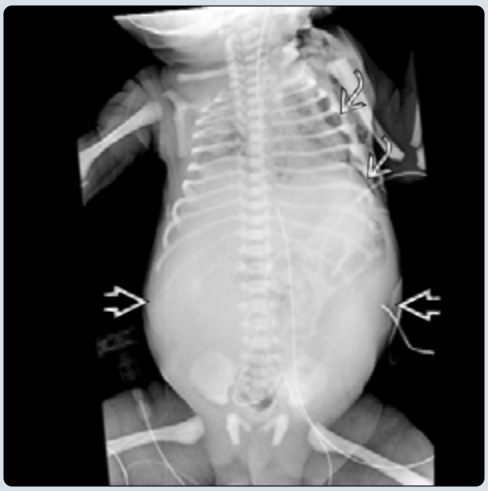
PATHOLOGY

- Autosomal recessive, due to mutations of polycystic kidney & hepatic disease 1 gene (*PKHD1*) on chromosome 6p12.2
  - Risk of recurrence in subsequent pregnancies: 25%

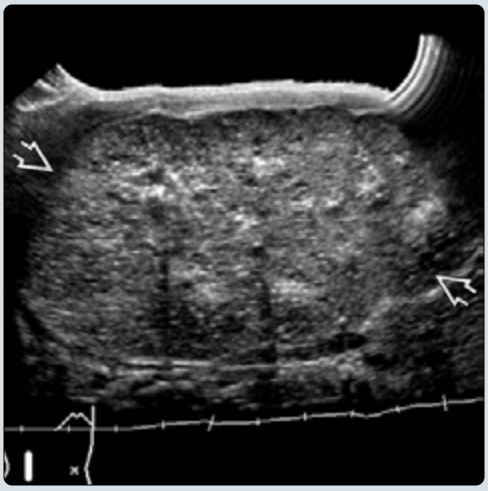
CLINICAL ISSUES

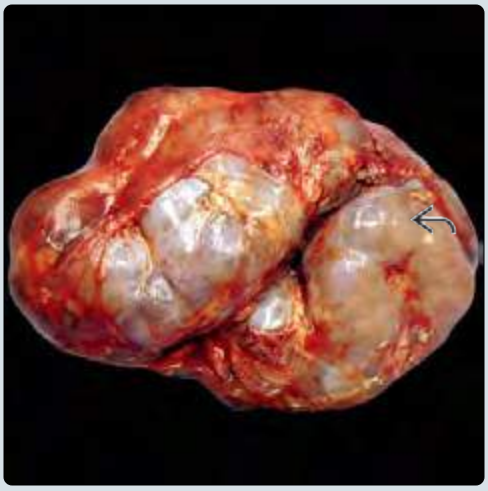
- Perinatal form: More severe renal disease with pulmonary hypoplasia, less hepatic disease
  - Renal replacement therapy (dialysis or transplant)
- Juvenile form: Less renal disease, more hepatic disease
  - Portal hypertension & fibrosis develop in 50%
- Severity & outcomes vary within affected families
- Survival rate for milder forms up to 82% at age 3 years & 79% at 15 years





(Left) *Coronal SSFSE T2 fetal MR shows marked enlargement & increased signal of both kidneys without hydronephrosis. Note the lack of amniotic fluid around this fetus, reflecting poor renal function in this case of autosomal recessive polycystic kidney disease (ARPKD).* **(Right)** *Frontal view of a newborn infant with a history of oligohydramnios shows bowel loops displaced centrally by bilateral flank "masses" (due to massively enlarged kidneys). Note the bell-shaped thorax & left pneumothorax.*





(Left) *Extended field of view US shows a newborn kidney measuring >10 cm in length. There is loss of corticomedullary differentiation with replacement of the renal parenchyma by microscopic cysts & dilated tubules with hyperechoic walls.* **(Right)** *Gross pathology shows a kidney removed from an infant with ARPKD in order to relieve mass effect & permit peritoneal dialysis. Macroscopic cysts are visible along the surface of this kidney, which is > 14 cm long.*

TERMINOLOGY

Synonyms

- Autosomal recessive polycystic kidney disease (ARPKD), infantile polycystic kidney disease

Definitions

- Single gene ciliopathy characterized by dilated distal convoluted tubules & collecting ducts

IMAGING

General Features

- Best diagnostic clue
  - Bilaterally enlarged hyperechoic kidneys in newborn with loss of corticomedullary differentiation
  - Pulmonary hypoplasia & history of oligohydramnios further support diagnosis
- Location
  - Massive kidneys fill flanks & displace adjacent organs
- Size
  - 2-6 standard deviations (SD) above mean size in majority
- Morphology
  - Reniform shape generally maintained
  - Loss of normal intrarenal architecture
- Additional features
  - Variable degrees of liver disease
    - Cystic dilation of intrahepatic bile ducts
      - Central dot sign of linear or punctate portal venous radicle encased by duct
    - Hepatic fibrosis
    - Secondary signs of portal hypertension: Splenomegaly, ascites, abnormal portal vein

Radiographic Findings

- Radiography
  - Bilateral flank "masses" bulging lateral abdominal contours & displacing bowel gas centrally
  - Pulmonary hypoplasia with bell-shaped thorax ± pneumothorax, pneumomediastinum

MR Findings

- Large kidneys of diffusely high signal intensity on T2WI
  - May have striated appearance of dilated tubules ± small, discrete cysts with low-signal septa
- Uniform or grainy intermediate to low signal on T1WI
- On MR urography series, radially arranged, dilated tubules visible throughout kidney
- No urine in bladder

Ultrasonographic Findings

- Enlarged, hyperechoic kidneys
  - Renal size maximal by 1-2 years old
- Poor or absent corticomedullary differentiation
- Dilated, radially arranged tubules seen in 2/3 of affected kidneys by high-resolution linear transducers
- Focal rosettes consisting of clusters of radially oriented, dilated collecting tubules also reported
- Small cysts may be present, generally < 1 cm in diameter; seen in ~ 1/2 of patients
  - Cysts > 1 cm only in minority of cases
- Diffuse microcystic appearance also described
- Tiny, punctate hyperechoic foci develop with time & correlate with renal failure
  - Do not cause posterior acoustic shadowing but may cause ring-down artifact
  - Likely due to calcium deposits
    - Calcium citrate & calcium oxalate crystals found histologically
- Doppler suggests random arrangement of intrarenal vessels rather than typical branching pattern
- Prenatal ultrasound findings
  - Kidneys > 2 SD above mean for gestational age; may be 4-6 SD above
  - Enlargement may not occur until mid 2nd trimester
  - Cysts may be visible but do not predominate
  - Abnormal corticomedullary differentiation in variable patterns
  - Oligohydramnios
  - Fetal bladder not visible
  - Associated musculoskeletal abnormalities due to mechanics: Oligohydramnios limits movement

Nuclear Medicine Findings

- Hepatobiliary scintigraphy
  - Delay in maximal hepatocyte uptake; delayed tracer excretion into biliary tree & gut; cystic foci of hepatic photopenia early with gradual & prolonged tracer accumulation
- Tc-99m DMSA renal cortical scans
  - Loss of kidney outline & internal structure; patchy tracer uptake with focal defects throughout kidneys

Imaging Recommendations

- Best imaging tool
  - Ultrasound
- Protocol advice
  - High-frequency linear US transducer in infants
  - Serial renal measurements in fetuses at risk
  - Renal circumference:abdominal circumference ratio
    - > 2 SDs above mean
  - Amniotic fluid assessment
    - 1st- or 2nd-trimester oligo carries poor prognosis

DIFFERENTIAL DIAGNOSIS

Bilateral Multicystic Dysplastic Kidney

- Macroscopic cysts of varying size with no normal renal parenchyma
- Essentially lethal disorder

Autosomal Dominant Polycystic Kidney Disease

- Renal enlargement less severe than ARPKD, may be asymmetric
- Small cysts may be visible in 3rd trimester, renal echogenicity normal or with echogenic cortex, amniotic fluid normal
  - Check family history & scan kidneys of parents

Cystic Renal Dysplasia

- Small with numerous cysts & echogenic parenchyma
- Due to chronic obstruction, infection, or vascular injury
  - May be unilateral or asymmetric

Meckel-Gruber Syndrome

- Encephalocele, large polycystic kidneys, & polydactyly
  - Microcephaly may be clue if oligohydramnios limits views

Tuberous Sclerosis

- Rhabdomyoma: Echogenic cardiac mass
- Tubers & subependymal nodules in brain: May be difficult to detect in newborn
- Renal cysts: May be seen in utero
- Angiomyolipomas: Not usually seen in newborn

PATHOLOGY

General Features

- Etiology
  - Ectatic distal convoluted tubules & collecting ducts
    - ↑ volume medulla → renal enlargement
    - ↑ in reflective interfaces from dilated tubules creates high echogenicity
- Genetics
  - Autosomal recessive
    - Risk of recurrence in subsequent pregnancies: 25%
  - Gene called polycystic kidney & hepatic disease 1 (*PKHD1*)
    - Chromosomal locus 6p12.2
  - Carrier frequency: 1 in 70 of general population
- Associated abnormalities
  - Musculoskeletal abnormalities related to oligohydramnios
  - Pulmonary compromise as result of oligohydramnios & mass effect from marked renal enlargement (which elevates diaphragm)

Microscopic Features

- Ectatic distal convoluted tubules & collecting ducts
- Tubules described as saccular or cylindrically enlarged

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Bilateral renal enlargement, renal insufficiency ± respiratory distress
    - Pulmonary hypoplasia may be life-limiting
- Other signs/symptoms
  - Fetal presentation: Enlarged kidneys at imaging
    - Majority detected > 24 weeks
    - Diagnosis reported at 16 weeks in at-risk fetus
      - Most kidneys look normal up to 20 weeks
    - May look normal up to late in 2nd trimester
      - Reports of delayed oligohydramnios > 28 weeks
    - If fetal diagnosis, most stillborn or neonatal death
  - Marked nephromegaly may cause dystocia at birth

Demographics

- Age
  - More severe disease typically presents in infancy
  - Milder forms of ARPKD can present in childhood & survive to adulthood
- Gender
  - M = F
- Epidemiology
  - 1:20,000 births

Natural History & Prognosis

- Perinatal form
  - Severe renal disease
  - Pulmonary hypoplasia
  - Minimal hepatic fibrosis
  - Worst prognosis; high mortality in 1st month of life
- Juvenile form
  - Milder renal disease; more severe hepatic fibrosis
  - Liver disease more relevant in survivors
    - Portal hypertension & fibrosis develop in ~ 1/2 of patients
  - Survival rate has ↑ for milder form to 82% at age 3 years & 79% at 15 years
- Perinatal survivors will require renal replacement therapy (dialysis or transplant)
  - If prolonged survival, liver disease becomes relevant
  - Liver transplant currently only available treatment for hepatic component of this disease
  - Long-term survivors have had combined renal & hepatic transplants
- Severity & outcomes vary within affected families
  - Systemic hypertension seen in 75%
  - Chronic ventilatory support in 30-50%

Treatment

- With fetal detection
  - Monitor fetal abdominal circumference for risk of dystocia
  - Counsel on outcomes, interventions (serial amnioinfusions, postnatal pulmonary & renal support) vs. comfort care options
  - Deliver at tertiary center
- With survivors
  - As needed, dialysis (peritoneal in infants) as bridge to renal transplant
    - Dialysis or transplant in 25-60% by 10 years old
  - Nephrectomy when renal size compromises respiratory status or peritoneal dialysis
  - Liver transplant when associated with progressive hepatic fibrosis
    - Combined liver kidney transplant (CLKT)
    - Kidney after liver transplant (KALT)

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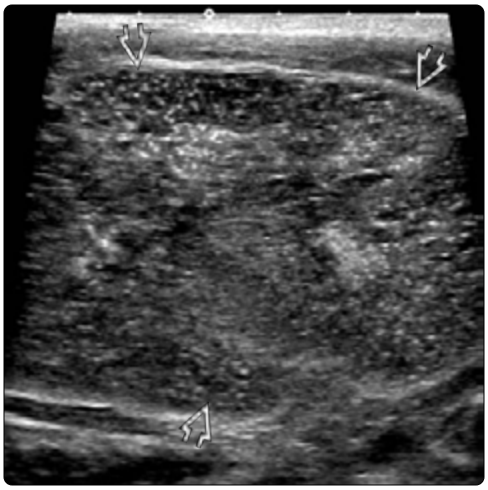
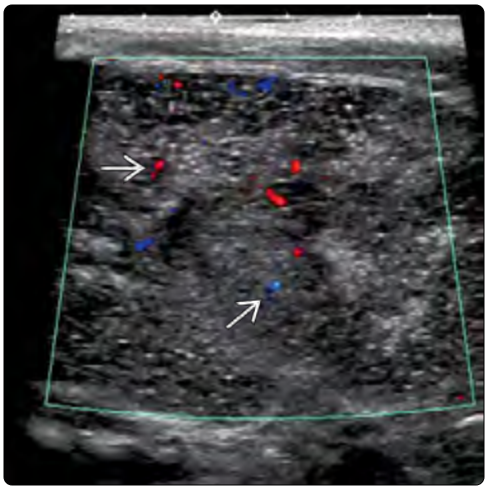
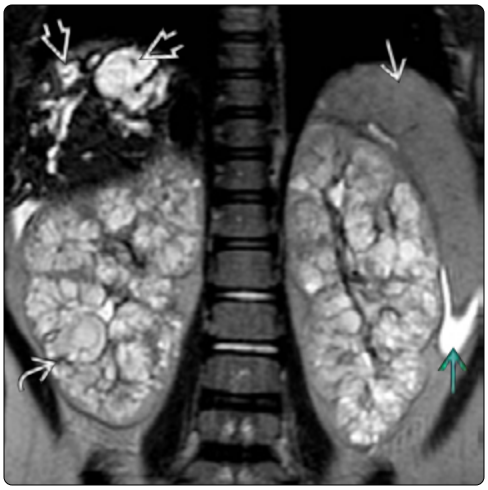
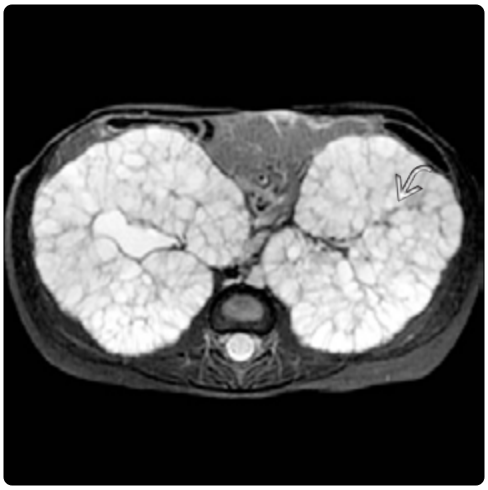


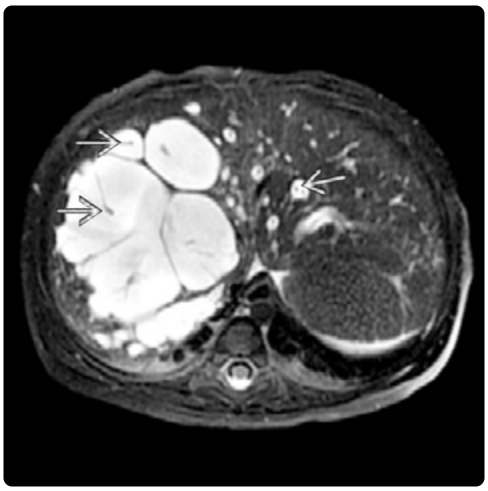


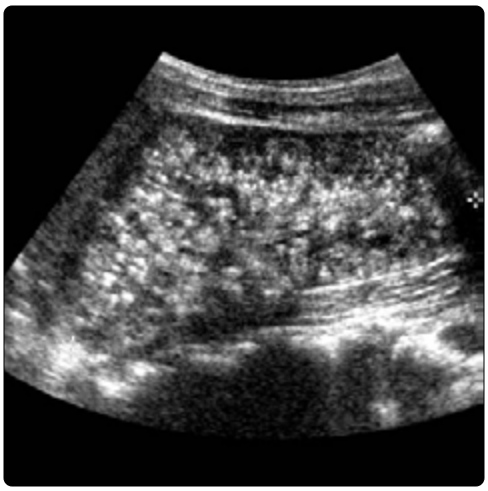
(Left) *Longitudinal US using a high-frequency linear transducer on the back of this infant with ARPKD optimally demonstrates the tiny cysts & dilated tubules replacing the normal renal parenchyma. The hyperechoic dots & dashes represent the highly reflective walls of the cysts & tubules.* **(Right)** *Color Doppler US in the same patient shows small vessels coursing randomly in the replaced renal parenchyma (rather than in the organized pattern of normal interlobular & arcuate branches).*





(Left) *Coronal T2 FS MR in a 4 year old with ARPKD & Caroli disease shows the enlarged kidneys replaced with tiny cysts & lace-like fibrous septa. A central dot pattern of biliary ductal dilation is noted. The progressive hepatic fibrosis has led to splenomegaly & ascites from portal hypertension.* **(Right)** *Axial T2 FS MR in a child with ARPKD & Caroli disease shows complete replacement of the kidneys with cysts & lace-like fibrous septa. The kidneys are markedly enlarged.*





(Left) *Axial T2 FS MR in a patient with ARPKD & Caroli disease shows the dilated intrahepatic biliary tree affecting the right lobe of the liver more than the left. The central dots within these fluid-filled spaces are tiny portal vein branches.* **(Right)** *Longitudinal US in a patient with ARPKD shows nephromegaly with numerous punctate echogenic foci throughout the kidney (an appearance which was bilateral). The echogenic foci likely represent calcium deposits & correlate with worsening renal function.*

Polycystic Kidney Disease, Autosomal Dominant

KEY FACTS

TERMINOLOGY

- Autosomal dominant polycystic kidney disease (ADPKD)
- Hereditary ciliopathy characterized by multiple renal cysts & various other systemic manifestations
- Cystic organ involvement: Kidneys (100%), liver (50%), pancreas (9%), brain/ovaries/testis (1%)
- Cerebral "berry" aneurysms (5-10% in adults)

IMAGING

- Renal size within 2 standard deviations above normal at time of diagnosis in 1/2 of pediatric patients
- Scattered renal cysts, of variable number & size
  - ↑ throughout life: 54% of ADPKD cysts appear in 1st decade; 72% occur within 2nd decade
  - May be complicated by hemorrhage, infection, or rupture
- Renal parenchyma often otherwise normal

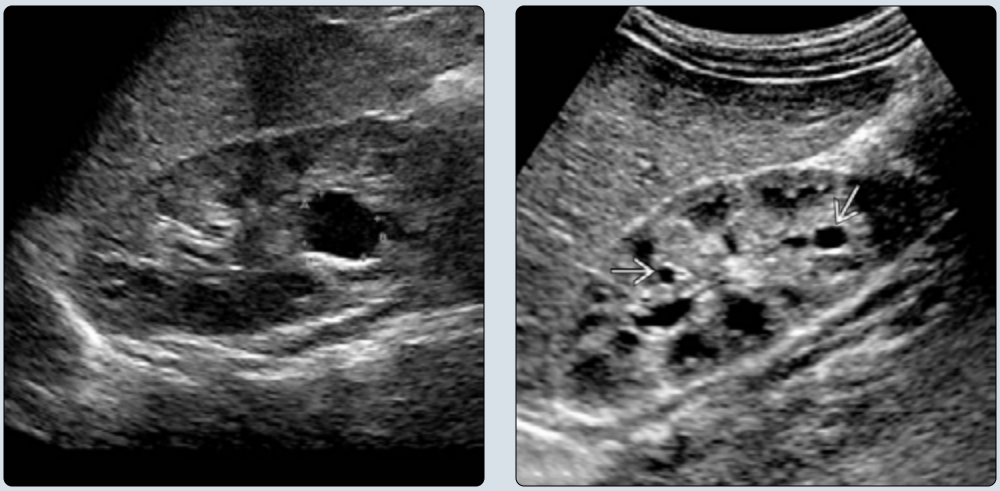
PATHOLOGY

- 90% autosomal dominant; 10% spontaneous mutations
- Types of ADPKD based on gene location
  - *PKD1*: Short arm of chromosome 16 (90%)
  - *PKD2*: Long arm of chromosome 4 (10%)
- Family history lacking in almost 1/2 of patients due to variable expressivity & spontaneous mutations

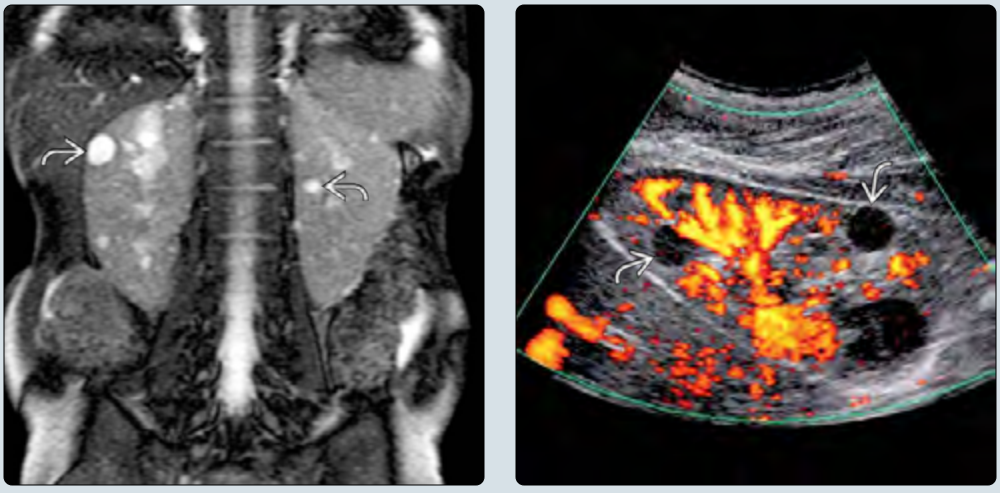
CLINICAL ISSUES

- Typically asymptomatic in childhood
  - Discovered incidentally or when screening children of affected adults
  - Flank pain, hematuria, hypertension, & renal failure also reported in children
- Prognosis excellent in childhood
- Prognosis in adulthood variable: Hemorrhage, infection, rupture; renal failure; hypertension; rarely malignancy
- 4th leading cause of chronic renal failure in world

(Left) *Longitudinal US in a 5 year old being screened for cysts due to a relevant family history shows a solitary central cyst between the cursors, likely due to autosomal dominant polycystic kidney disease (ADPKD).* **(Right)** *Longitudinal oblique US in a 9 month old with a family history of renal failure shows several small cysts ➡ in the medullary portion of the right kidney.*



(Left) *Coronal SSFP MR in the same 9 month old shows numerous cysts ➡ of varying size scattered through both kidneys in this patient with ADPKD.* **(Right)** *Longitudinal power Doppler US shows normal blood flow in renal parenchyma between the cysts ➡. Vascular compression by enlarging cysts is one theory to explain progressive renal insufficiency in ADPKD.*



TERMINOLOGY

Synonyms

- Autosomal dominant polycystic kidney disease (ADPKD), adult polycystic kidney disease

Definitions

- Hereditary ciliopathy characterized by multiple renal cysts & various other systemic manifestations
- Cystic organ involvement
  - Kidneys (100%), liver (50%), pancreas (9%), brain/ovaries/testis (1%)
- Noncystic manifestations
  - Cardiac valve (26%), hernias (25%), colonic diverticula
  - Aneurysms: Cerebral "berry" aneurysms (5-10%); aortic or coronary aneurysms less common

IMAGING

General Features

- Best diagnostic clue
  - Innumerable macrocysts ± enlarged kidneys
- Size
  - Cysts tend to be > 1 cm in diameter
  - Renal size within 2 standard deviations above normal at time of diagnosis in 1/2 of pediatric patients

CT Findings

- CECT
  - No enhancement of uncomplicated cysts
    - Hypodense relative to enhanced normal renal tissue
  - Complicated (hemorrhagic) cysts
    - Hyperdense (60-90 HU)
    - ± curvilinear mural Ca^{2+} or calculi in cysts
  - Complicated (infected) cysts
    - Hypodense; ± gas in infected cyst
    - Thick, irregular wall, thickened renal fascia
    - Variable wall enhancement

MR Findings

- T1WI
  - Uncomplicated & infected cysts: Hypointense
  - Complicated (hemorrhagic cysts)
    - Signal intensity varies with age of hemorrhage
    - Classic appearances suggesting blood products: Hyperintense, fluid-fluid levels
- T2WI
  - Uncomplicated cysts: Hyperintense contents, thin wall
  - Complicated: Variable signal intensity, blood products may be hypointense & layering; mural thickening

Ultrasonographic Findings

- Grayscale ultrasound
  - Multiple well-defined, round, anechoic cysts bilaterally
  - Renal contour typically normal early in life; may become lobulated as more cysts form
  - Renal size & echotexture often normal in young patients, aside from few cysts

Imaging Recommendations

- Best imaging tool
  - Ultrasound (sensitivity, specificity, accuracy 97+%)

DIFFERENTIAL DIAGNOSIS

Autosomal Recessive Polycystic Kidney Disease

- Very enlarged newborn kidneys
- Echogenic parenchyma with dilated tubules & loss of corticomedullary differentiation; few small cysts

Isolated Simple Cysts

- Very few in number, normal renal function

Tuberous Sclerosis

- Echogenic, fat-containing renal angiomyolipomas as well
- Characteristic brain & cutaneous findings

Acquired Cystic Disease of Dialysis

- Early stage: Small kidneys with multiple cysts
- Advanced stage: Indistinguishable from ADPKD

Other Causes of Bilateral Renal Enlargement

- Lymphoma/leukemia
- Nephroblastomatosis
- Glomerulonephritis
- Renal vein thrombosis
- Hemolytic uremic syndrome

PATHOLOGY

General Features

- Genetics
  - 90% autosomal dominant; 10% spontaneous mutations
  - Family history lacking in almost 1/2 of patients due to variable expressivity & spontaneous mutations

CLINICAL ISSUES

Presentation

- Typically asymptomatic in childhood
  - Discovered incidentally or when screening children of affected adults
- Flank pain, hematuria, hypertension, & renal failure also reported in children

Demographics

- Variable age at diagnosis: Childhood to 8th decade
  - Prevalence of cysts ↑ with age
    - 54% appear in 1st decade of life
    - 72% occur within 2nd decade
- Incidence: 1 in 400 to 1,000 persons in USA

Natural History & Prognosis

- Prognosis excellent in childhood
- Prognosis in adulthood variable
  - Hemorrhage, infection, rupture, renal failure, hypertension, rarely malignancy
- 4th leading cause of chronic renal failure in world

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Wilms Tumor

KEY FACTS

TERMINOLOGY

- Malignant tumor of primitive metanephric blastema
- Most common abdominal tumor in children 1-8 years old

IMAGING

- Ultrasound frequently 1st study performed; CT & MR better characterize tumor & local extent
  - Large, heterogeneous but predominantly solid hypoechoic (US)/hypoenhancing (CT/MR) renal mass
  - Careful evaluation of
    - Adjacent soft tissues for tumor rupture, adenopathy
    - Renal vein & inferior vena cava for tumor thrombus
    - Contralateral kidney for synchronous tumor or nephrogenic rests
- Chest radiograph or CT: Lung metastases in 10-20%

TOP DIFFERENTIAL DIAGNOSES

- Neuroblastoma: Extrarenal, more often calcified
- Congenital mesoblastic nephroma: < 3-12 months
- Clear cell sarcoma: Look for skeletal metastases
- Renal cell carcinoma: Equal incidence to Wilms after age 12
- Nephroblastomatosis: Multiple bilateral nephrogenic rests

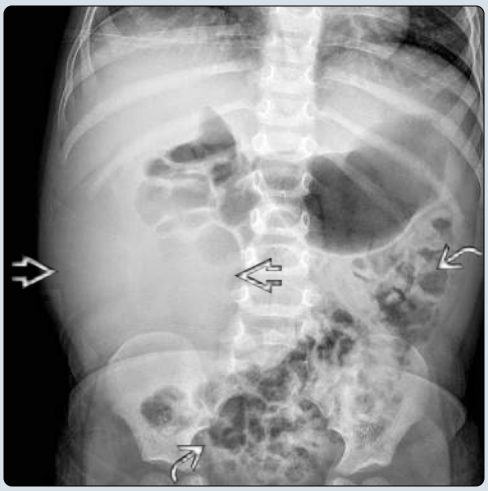
PATHOLOGY

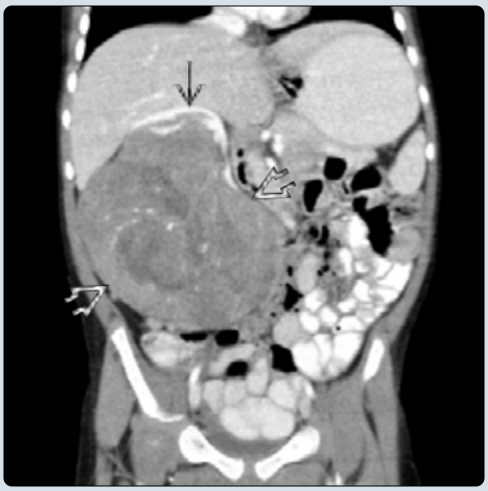
- Arises from primitive metanephric blastemal tissue
  - Persistence after 34 weeks gestation termed "nephroblastomatosis" → 30-40% develop Wilms tumor
- Associated predisposing syndromes in 10%
  - Quarterly screening renal ultrasounds until age 8

CLINICAL ISSUES

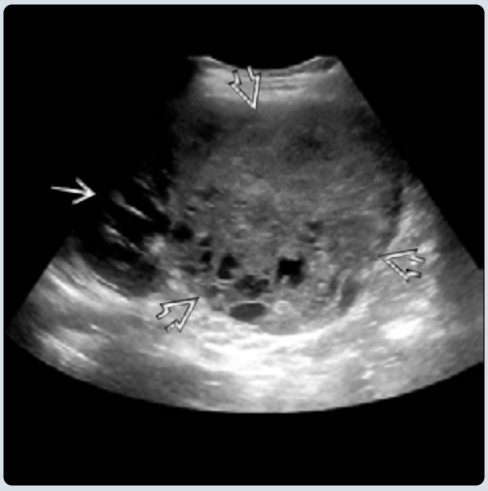
- 80% of cases in children < 5 years old
- Typical presentation: Incidentally discovered palpable mass
- Preferred treatment: Up front complete surgical resection
  - Preoperative chemotherapy for unresectable or bilateral tumors or tumor thrombus above hepatic veins
  - Postoperative chemotherapy ± radiation
- > 90% 5-year-survival for localized abdominal disease

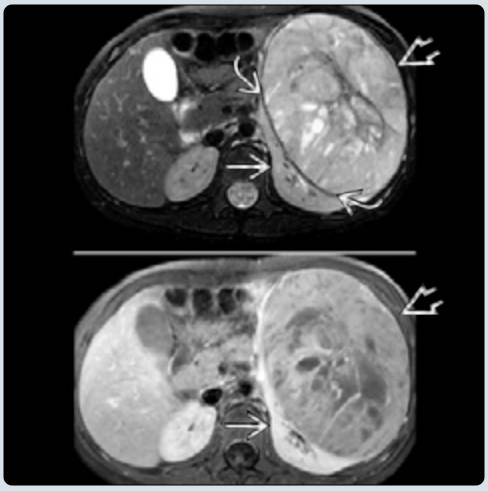
(Left) *AP abdominal radiograph of a 3-year-old child with a firm, palpable mass shows leftward displacement of bowel loops by a right-sided soft tissue mass. CT was performed next in this child due to suspicion for a Wilms tumor.* **(Right)** *Coronal CECT in the same patient confirms a large, heterogeneous, solid mass arising from the right kidney. A residual "claw" of normal renal tissue is splayed along the upper pole of this Wilms tumor. No contralateral lesions or venous invasion were identified.*





(Left) *Longitudinal US in a 5 year old with hypertension & abdominal fullness shows a round heterogeneous mass extending out of the lower pole of the left kidney. Wilms tumor was confirmed upon surgery.* **(Right)** *Axial T2 (top) & T1 C+ (bottom) FS MR images show a large, well-defined, heterogeneous mass arising from a splayed "claw" of the left kidney in a 6 year old with a Wilms tumor. Note the presence of a T2 hypointense pseudocapsule around the tumor.*





TERMINOLOGY

Synonyms

- Nephroblastoma, embryoma of kidney (archaic)

Definitions

- Malignant tumor of primitive metanephric blastema
- Most common abdominal neoplasm ages 1-8 years

IMAGING

General Features

- Best diagnostic clue
  - Large, heterogeneous renal mass extending into renal vein & inferior vena cava (IVC) in young child
- Location
  - > 90% unilateral, 5-10% bilateral
- Size
  - Typically quite large (mean diameter: 5-10 cm)
- Morphology
  - Usually spherical; sometimes lobulated or multicentric
  - Usually smooth contours; may have local extension

Radiographic Findings

- Mass displacing adjacent bowel
- Ca^{2+} visible in 10%

CT Findings

- NECT
  - Ca^{2+} in 15%
  - Lung metastases in 10-20% at time of diagnosis
    - Well-defined solid nodules
- CECT
  - Large, poorly enhancing, heterogeneous mass replacing most of kidney
    - "Claw" of residual renal tissue along tumor margin
  - Often has well-defined margins or pseudocapsule
  - Displaces adjacent organs, especially bowel
  - ± adjacent adenopathy
  - Predilection for invasion of renal vein & IVC
  - May have local extension into perirenal fat or gross tumor rupture with distant ascites

MR Findings

- T1WI: Typically heterogeneous, predominantly intermediate to low signal but may have foci of high T1 signal due to blood products
- T2WI: Typically heterogeneous, predominantly high signal
- T1 C+: Often poor, heterogeneous enhancement
- MRV: Useful in determining vascular invasion

Ultrasonographic Findings

- Grayscale ultrasound
  - Findings similar to CT & MR
    - Large, hypoechoic, heterogeneous mass
    - May see local invasion & adenopathy
  - Often difficult to image entire tumor without extended field of view
- Color Doppler
  - Can be useful to detect tumor thrombus vs. compression of veins by bulky mass

Echocardiographic Findings

- Used to assess intracardiac tumor thrombus
  - Especially when preoperative chemotherapy used to "shrink" tumor thrombus
  - Intracardiac tumor thrombus may change surgical approach, necessitate cardiothoracic surgical input

Nuclear Medicine Findings

- Bone scan
  - Not routinely used as bone metastases occur very late
- PET
  - Increasing use in Wilms (& all pediatric tumors)
  - Primarily has adjunctive, problem-solving role
    - Differentiates scar from active tumor post chemotherapy
    - May predict response to neoadjuvant therapy

Imaging Recommendations

- Best imaging tool
  - Ultrasound often performed initially for palpable mass
  - CT or MR to further characterize tumor, local extent, adenopathy
    - MR better for detection of contralateral lesions
  - Chest radiograph or CT for staging
- Protocol advice
  - Doppler US or contrast-enhanced CT or MR venography for assessment of ipsilateral renal vein & IVC

DIFFERENTIAL DIAGNOSIS

Neuroblastoma

- Suprarenal (adrenal gland) or paraspinal (sympathetic chain)
  - Typically displaces rather than invades kidney
- Much more likely than Wilms to contain Ca^{2+}, cross midline, & engulf or "lift" adjacent vessels

Congenital Mesoblastic Nephroma

- Solid or mixed solid & cystic tumor of infants
  - > 90% diagnosed in 1st year of life
  - Most common renal tumor < 3 months of age

Multilocular Cystic Nephroma

- Entirely cystic mass with numerous thin septations
- Classically herniates into renal pelvis
- Pediatric cases typically boys 2-3 years of age

Renal Cell Carcinoma

- Solid renal mass typically seen in 2nd decade of life
- Incidence equal to Wilms after 12 years of age

Renal Rhabdoid Tumor

- Nonspecific solid renal mass in young child
- May have synchronous intracranial rhabdoid tumor

Clear Cell Sarcoma

- Solid renal mass with skeletal metastases at diagnosis

Angiomyolipoma

- Benign fat-containing, vascular tumors with variable enhancement
- Tuberous sclerosis complex in most pediatric angiomyolipomas (AMLs)
  - AMLs typically multiple & accompanied by simple cysts

Pyelonephritis

- Developing abscess can mimic cystic neoplasm but typically more infiltrative
- Patients often have clinical & laboratory features of upper urinary tract infection

Renal Medullary Carcinoma

- Poorly defined, infiltrating renal mass; can mimic phlegmonous pyelonephritis
- Lymphangitic pulmonary spread common
- Typically in adolescents with sickle cell trait or disease

Nephroblastomatosis

- Multiple nonenhancing foci of residual primitive nephrogenic rests, usually bilateral
- Individual lesions relatively small & homogeneous
- High association with bilateral Wilms tumor, Beckwith-Wiedemann syndrome, hemihypertrophy

Lymphoma

- May have multifocal solid bland renal masses, typically in setting of other confluent adenopathy ± splenic disease

PATHOLOGY

General Features

- Etiology
  - Primitive metanephric blastema differentiates by 34 weeks gestation
  - Persistence of metanephric blastema (nephrogenic rests) termed "nephroblastomatosis"
    - Found in 1% of infant autopsies, 4% of resected multicystic dysplastic kidney
    - Wilms tumor develops in 30-44%
- Genetics
  - Numerous somatic & germ line mutations described
    - 10-20% have 11p13 (*WT1*) gene mutation
  - Only 2% of Wilms tumors familial
- Associated abnormalities
  - Genitourinary anomalies
  - Overgrowth syndromes (Beckwith-Wiedemann, isolated hemihypertrophy)
  - WAGR syndrome: Wilms tumor, aniridia, genitourinary anomalies, mental retardation
  - Sporadic aniridia
  - Denys-Drash syndrome
  - Trisomy 18
  - Sotos syndrome
  - Bloom syndrome

Staging, Grading, & Classification

- Similar systems used by Children's Oncology Group (COG)/National Wilms Tumor Study Group (NWTSG) or Société Internationale d'Oncologie Pediatrique (SIOP)
  - I: Confined to kidney, completely excised
  - II: Local extension, completely resected
  - III: Incomplete resection, no distant metastases
  - IV: Distant metastases to lung, liver, brain, or bone
  - V: Bilateral synchronous tumors

Microscopic Features

- 4-10% have unfavorable (anaplastic) histology

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic flank mass, hematuria, vomiting, failure to thrive
- Other signs/symptoms
  - Hypertension, fever from tumor necrosis, anemia

Demographics

- Age
  - 80% of cases occur in children < 5 years old
  - Peak: 3.6 years
    - Syndromic tumor occurrence typically younger
- Epidemiology
  - 500 new cases each year in United States
  - 1/3 of patients with sporadic aniridia have Wilms; 1% of Wilms tumor patients have aniridia

Natural History & Prognosis

- Prognosis based on stage, tumor size, histology
- 5-year-survival for localized abdominal disease > 90%

Treatment

- Up front complete surgical resection (nephrectomy) preferred
  - Limited uses of renal-sparing resection: Predisposition to bilateral tumors or preexisting solitary kidney
- Preoperative chemotherapy for unresectable or bilateral tumors or tumor thrombus extending above hepatic veins
- Postoperative chemotherapy ± radiation
- Bone marrow transplant usually reserved for relapses

DIAGNOSTIC CHECKLIST

Consider

- Children with predisposing syndromes require ultrasound screening every 3 months until 8 years of age

Image Interpretation Pearls

- Careful assessment of ipsilateral renal vein & IVC, adjacent soft tissues & lymph nodes, contralateral kidney, & lungs
- Look for free fluid that might suggest tumor rupture

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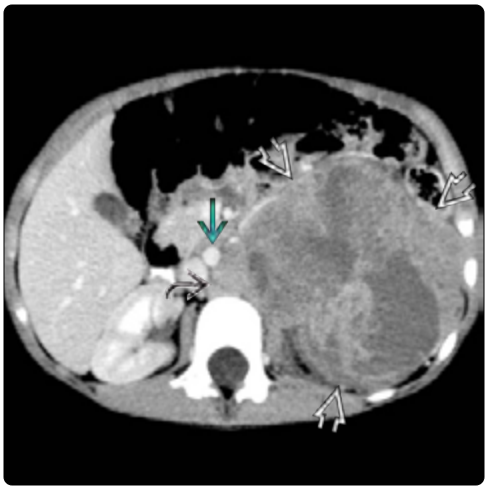
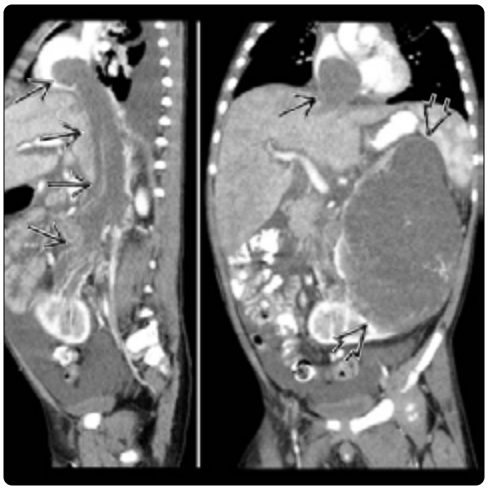
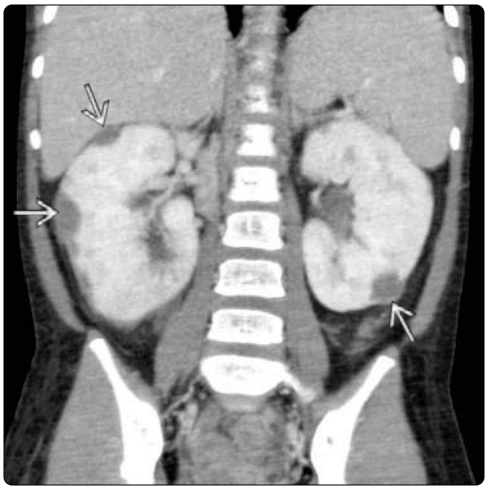
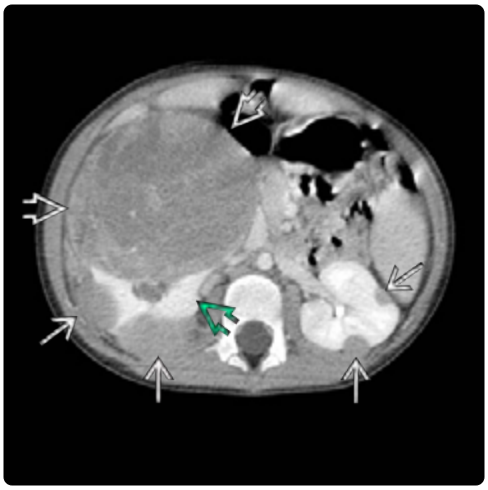


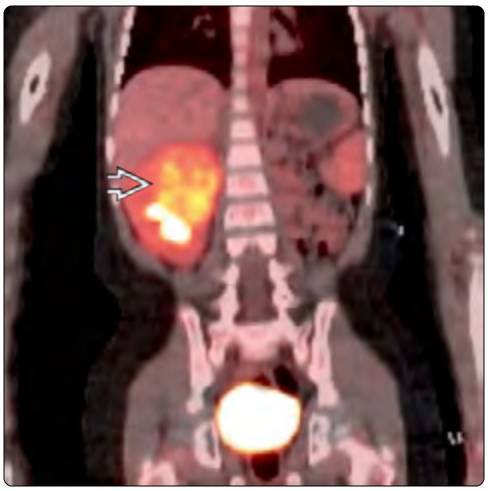


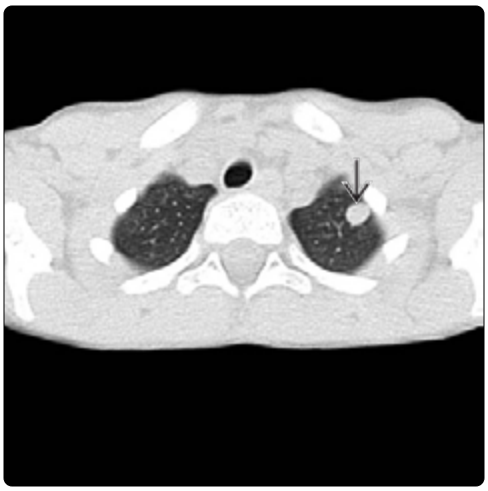
(Left) *Axial CECT shows a large, poorly defined, heterogeneous Wilms tumor arising from the left kidney in a 4-year-old girl. Note the enlarged lymph node displacing the aorta anteriorly & to the right.* **(Right)** *Sagittal (left) & coronal (right) CECTs show tumor thrombus extending through the left renal vein, up the inferior vena cava, & into the right atrium from a Wilms tumor in the left kidney. CECT has high sensitivity & specificity for cavoatrial tumor thrombus in children with Wilms tumor.*





(Left) *Coronal CECT of a 3 year old with a palpable mass shows numerous round nonenhancing lesions in both kidneys. These residual nephrogenic rests (nephroblastomatosis) will give rise to a Wilms tumor in 30-40% of affected patients.* **(Right)** *Axial CECT through the kidneys in the same patient shows a large, well-defined Wilms tumor in the right kidney. The nephrogenic rests are seen as numerous, bland-appearing cortical lesions in the periphery of both kidneys.*





(Left) *Coronal FDG PET/CT in a child with a history of left nephrectomy for Wilms tumor shows a contralateral recurrence with metabolically active tumor in the right upper kidney. FDG PET is useful in assessing chemotherapy response in children with relapsed Wilms tumor, even when the tumor size is unchanged on other modalities.* **(Right)** *Axial NECT through the lung apices in a 3 year old with Wilms tumor demonstrates a well-defined, biopsy-confirmed metastatic lung nodule in the left upper lobe.*

Nephroblastomatosis

KEY FACTS

TERMINOLOGY

- Nephrogenic rests (NR): Persistent metanephric blastema; precursor to Wilms tumor
  - Perilobar NR: Occur at periphery of kidney
  - Intralobar NR: Occur within kidney
- Nephroblastomatosis: Multiple or diffuse NR

IMAGING

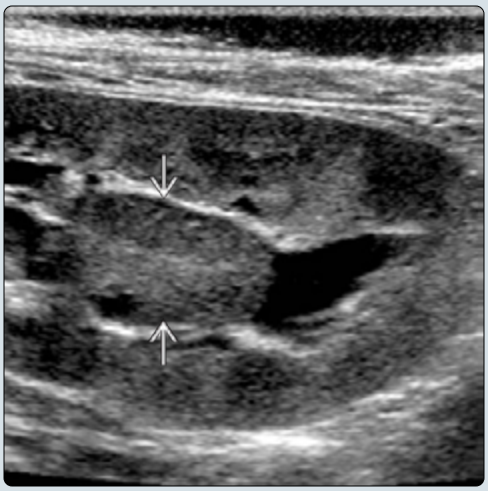
- Homogeneous, multifocal ovoid or subcapsular, rind-like renal masses that enhance less than normal kidney on CT/MR
  - Diffuse disease: Thick, uniform, homogeneous rind of hypoenhancing abnormal tissue
  - Multifocal disease: Scattered nodules resembling normal renal cortex on precontrast imaging
- Differentiating NR from Wilms tumor
  - NR: Diffusely homogeneous appearance; Wilms tumor: Heterogeneous
  - NR: Usually < 2 cm; Wilms tumor: Usually > 3 cm
  - NR: Usually oval or lenticular in shape; Wilms tumor usually spherical
- Imaging findings suggesting conversion of NR to Wilms
  - Rapid ↑ in size
  - Stable or ↑ size while on chemotherapy
  - Nodule within initial lesions
  - New heterogeneous appearance of mass

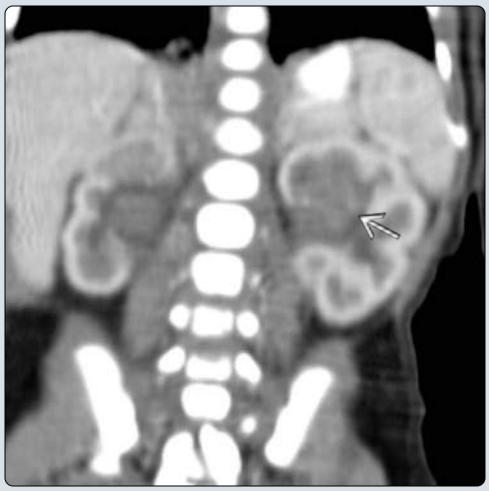
PATHOLOGY

- Biopsy often not helpful as microscopic patterns of NR & Wilms tumor can appear similar

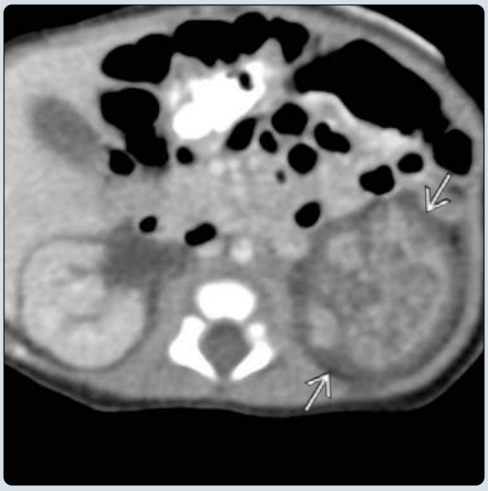
CLINICAL ISSUES

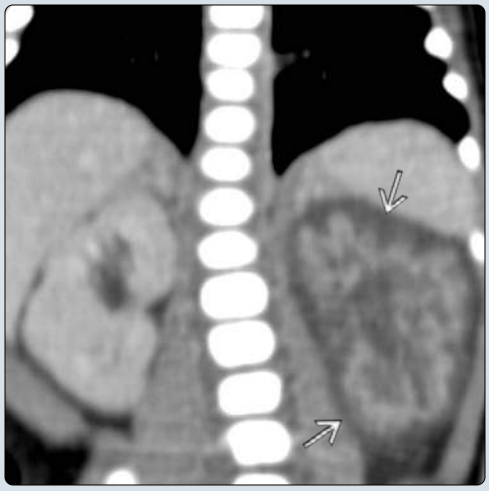
- Most NR spontaneously regress
  - Currently, no specific treatment protocol advocated
- Common associated syndromes: Beckwith-Wiedemann syndrome, hemihypertrophy, sporadic aniridia, & WAGR syndrome
  - Screening renal US at 3-month intervals up to age 8





(Left) *Longitudinal ultrasound of the left kidney in a 4 day old shows a centrally located, ovoid, isoechoic nephrogenic rest. Intralobar nephrogenic rests are less common than the perilobar form but have a higher risk of malignant transformation to Wilms tumor.* **(Right)** *Coronal CECT of the abdomen in the same patient shows a homogeneously hypodense intralobar nephrogenic rest. Intralobar rests are associated with the WT1 gene, sporadic aniridia, & Drash syndrome.*





(Left) *Axial CECT in a neonate shows unilateral diffuse perilobar nephrogenic rests as a thin rind of hypodense tissue surrounding the left kidney.* **(Right)** *Coronal CECT in the same patient shows the unilateral diffuse perilobar nephrogenic rests of the left kidney. Perilobar nephrogenic rests are more common than the intralobar form & are associated with the WT2 gene, Beckwith-Wiedemann syndrome, & hemihypertrophy.*

TERMINOLOGY

Definitions

- Nephrogenic rests (NR): Metanephric blastema that persists after birth; precursor to Wilms tumor
- Nephroblastomatosis: Presence of multiple or diffuse NR
- Intralobar NR: Occur anywhere in renal parenchyma
- Perilobar NR: Occur in peripheral cortex

Associated Syndromes

- Perilobar NR associated with Beckwith-Wiedemann syndrome, hemihypertrophy, Perlman syndrome, trisomy 18
- Intralobar NR associated with Drash syndrome, sporadic aniridia, WAGR syndrome

IMAGING

General Features

- Best diagnostic clue
  - Multifocal ovoid vs. subcapsular, rind-like homogeneous renal masses that enhance much less than normal kidney
    - ↓ enhancement due to hypovascularity
  - 2 main patterns of disease similar on all modalities
    - Diffuse disease: Thick, uniform, homogeneous rind of hypoenhancing abnormal tissue
    - Multifocal disease: Scattered nodules resembling normal renal cortex on precontrast imaging
- Location
  - Intralobar: Occur within kidney
  - Perilobar: Occur at periphery of kidney
- Size
  - Microscopic to several centimeters (usually < 3 cm)
- Morphology
  - Kidneys enlarged in diffuse disease
    - May lose normal corticomedullary differentiation
  - NR may deform renal surface, creating nodular or lobulated appearance
  - In diffuse perilobar NR, interfaces between hypoenhancing NR & centrally located normal parenchyma can be jagged or irregular

Ultrasonographic Findings

- Grayscale ultrasound
  - Homogeneously hypoechoic or isoechoic to renal parenchyma
    - May not be able to distinguish NR from background kidney
  - Enlarged, distorted kidneys in diffuse disease
    - Poor renal corticomedullary differentiation
- Color Doppler
  - Hypovascular lesions

CT Findings

- NECT: Isodense to slightly hyperdense to renal cortex
- CECT: Homogeneous hypodense lesions enhancing < normal renal tissue

MR Findings

- T1WI
  - Homogeneous masses isointense to slightly hypointense to renal parenchyma
- T2WI
  - Homogeneous masses isointense to renal parenchyma
- DWI
  - Can restrict diffusion
- T1WI C+
  - Homogeneous, hypointense masses that enhance < renal parenchyma

Nuclear Medicine Findings

- PET
  - Avid FDG uptake in Wilms tumor
    - Differentiation of Wilms from NR on PET not yet established

Imaging Recommendations

- Best imaging tool
  - MR preferred over CT due to superior contrast resolution & lack of radiation
  - Intravenous contrast administration essential
    - Most sensitive method to identify NR

Differentiation from Wilms tumor

- Best differentiator: NR → diffusely homogeneous; Wilms tumor → heterogeneous
- Weaker differentiators
  - NR usually < 2 cm; Wilms tumor usually > 3 cm
  - NR usually oval or lenticular in shape; Wilms tumor usually spherical
  - Wilms tumor surrounded by fibrous pseudocapsule
  - Larger Wilms tumors compress & distort calyces, invade vessels
- Imaging findings suggesting conversion from NR to Wilms tumor
  - Rapid ↑ in size
  - No change or growth while patient on chemotherapy
  - Nodule within initial lesions
  - Newly heterogeneous appearance of mass

DIFFERENTIAL DIAGNOSIS

Wilms Tumor

- Solid mass with heterogeneous enhancement
- Bilateral in 4-13% of patients
- NR: Precursor to Wilms tumor
  - NR present in 30-40% of unilateral Wilms tumor cases
  - NR present in 94% of patients with metachronous contralateral Wilms tumor
    - 1% of unilateral Wilms tumor patients develop metachronous disease
    - ↑ risk in children < 12 months of age with NR at diagnosis
  - NR present in 99% of patients with synchronous bilateral Wilms tumors
    - Represent 4-7% of all Wilms tumors
    - Present at slightly younger age than unilateral Wilms tumor (2.6 vs. 3.3 years)

Renal Lymphoma

- Multifocal, homogeneous, hypodense lesions on CECT
- Other manifestations of disease typically present
  - Multifocal &/or confluent adenopathy
  - Splenomegaly

- Unusual in infants & young children

Pyelonephritis

- Wedge-shaped foci of renal parenchymal hypoenhancement (CECT) or hypoperfusion (color Doppler US)
- Can cause striated nephrogram

PATHOLOGY

General Features

- Etiology
  - Metanephric blastema that persists beyond 36 weeks gestational age

Staging, Grading, & Classification

- 2 pathologic subtypes
  - Perilobar rests (90%): In renal cortex or at corticomedullary junction
    - Associated with 1-2% risk of developing Wilms tumor
  - Intralobar rests (10%): Deeper in renal parenchyma
    - Associated with 4-5% risk of developing Wilms tumor
- 3 patterns of distribution
  - Unifocal
  - Multifocal
  - Diffuse
- 4 developmental phases
  - Incipient/dormant
    - Contains primitive epithelial cells
    - Rarely progresses to Wilms tumor
  - Regressing/sclerosing
    - Cells show signs of maturation or sclerosis
    - Rarely progresses to Wilms tumor
  - Hyperplastic
    - Adenomatous benign neoplasms that ↑ in size
  - Neoplastic

Gross Pathologic & Surgical Features

- Diffuse nephroblastomatosis
  - White plaques or whorls of tissue replacing renal parenchyma
    - ± peripheral rind
    - ± small cysts
    - ± masses of hyperplastic or neoplastic (Wilms) tissue

Microscopic Features

- Biopsy often not helpful in distinguishing NR from Wilms tumor as microscopic patterns can be similar
  - Imaging more helpful

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic vs. flank mass
  - Incidental finding of ipsilateral or contralateral kidney in setting of Wilms tumor

Demographics

- Age
  - Most common in infancy
    - May not be detectable on initial imaging study
  - Incidence ↓ with age
- Gender
  - M = F
  - Synchronous & metachronous bilateral Wilms tumors more common in females
- Epidemiology
  - NR present in 1% of perinatal autopsies
    - Incidence ↓ significantly in 1st months of life
  - Most sporadic, but ↑ risk with some syndromes

Natural History & Prognosis

- Risk for developing Wilms tumor
  - Only small percentage of patients with NR will develop Wilms tumor
  - NR present in 30-40% with unilateral Wilms tumor
  - Found in up to 99% of bilateral Wilms tumors
  - Risk for developing Wilms tumor highest in younger children; ↓ with age
  - 35% of diffuse hyperplastic perilobar NR develop Wilms tumor (highest risk group)

Treatment

- Most NR spontaneously regresses
- Currently, no specific treatment protocol advocated
  - Can be treated empirically with chemotherapy
- Children with syndromes at risk for Wilms tumor typically screened regularly with imaging for development of nephroblastomatosis/Wilms tumor
  - Screening renal sonography at 3-month intervals up to 8 years of age
  - US more useful to identify Wilms tumor than NR
    - On US, look for new/enlarging spherical mass
- MR C+ or CECT if renal ultrasound shows mass → follow-up with MR > CECT to minimize radiation
- ↑ size of nephroblastomatosis sometimes treated empirically as stage 1 Wilms tumor without biopsy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Nephroblastomatosis appears homogeneous on all imaging modalities (US, CT, MR)
  - Enhances much less than surrounding kidney
- Wilms tumor tends to be heterogeneous

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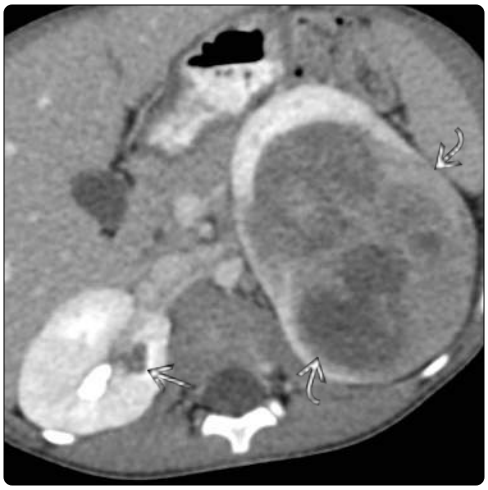
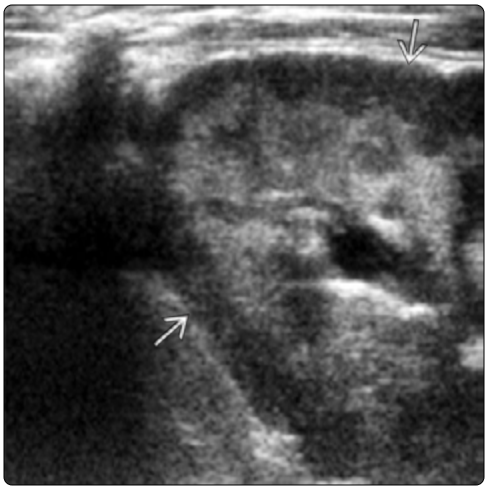
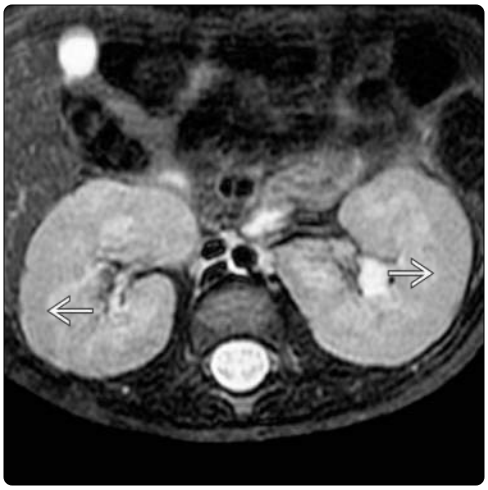
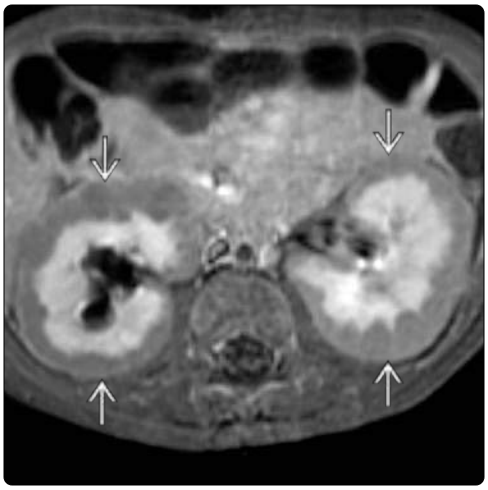


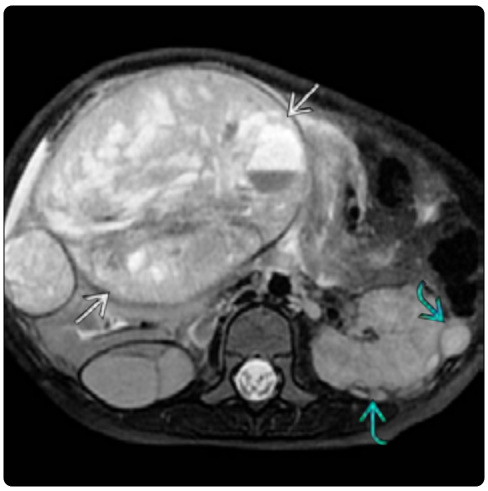


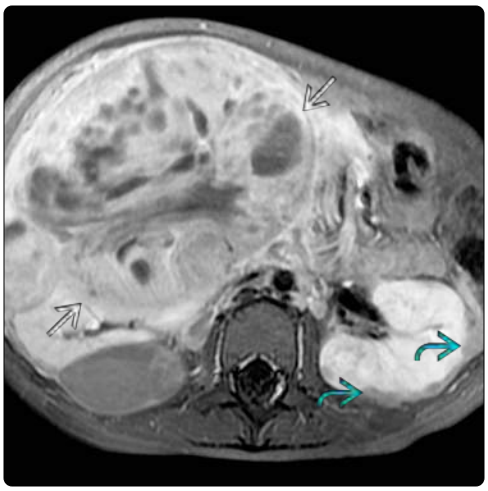
(Left) *Axial CECT in a young child shows a large Wilms tumor arising from the left kidney. There is a solitary intralobar nephrogenic rest in the right kidney. Nephrogenic rests usually have a homogeneous appearance. This is in contrast to a Wilms tumor, which is typically heterogeneous.* **(Right)** *Ultrasound of the kidney in a patient with Beckwith-Wiedemann syndrome shows a diffuse, hypoechoic rim of tissue at the periphery of the kidney, consistent with diffuse perilobar nephrogenic rests.*





(Left) *Axial T2 FS MR of the abdomen in a young patient shows diffuse enlargement of the kidneys, each of which have a subtle rim of hypointense tissue peripherally. Nephrogenic rests are often isointense to slightly hypointense to the normal renal parenchyma on T1 & T2 MR.* **(Right)** *Axial T1 C+ FS MR of the kidneys in the same patient shows diffuse bilateral hypoenhancing perilobar nephrogenic rests.*





(Left) *Axial T2 FS MR of the abdomen shows a large, heterogeneous Wilms tumor of the right kidney arising in a background of numerous smaller bilateral renal lesions.* **(Right)** *Axial T1 C+ FS MR of the abdomen in the same patient shows heterogeneous enhancement of the large Wilms tumor of the right kidney. The nodular foci in the left kidney show hypoenhancement, typical of nephrogenic rests in nephroblastomatosis.*

Multilocular Cystic Nephroma

KEY FACTS

TERMINOLOGY

- Benign mixed mesenchymal & epithelial renal neoplasm

IMAGING

- Large multilocular cystic renal mass in young male or middle-aged female
  - Mean size: 9-10 cm
- Cystic components generally follow imaging characteristics of simple fluid on all modalities
- Septa show variable enhancement
  - No excretion of contrast into cysts
- Mass may herniate into renal hilum
- "Claw" of normal, splayed/compressed renal tissue at periphery of mass

TOP DIFFERENTIAL DIAGNOSES

- Cystic Wilms tumor
- Multicystic dysplastic kidney
- Mesoblastic nephroma

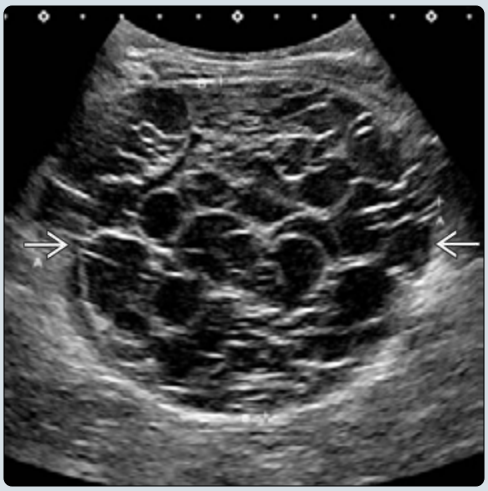
PATHOLOGY

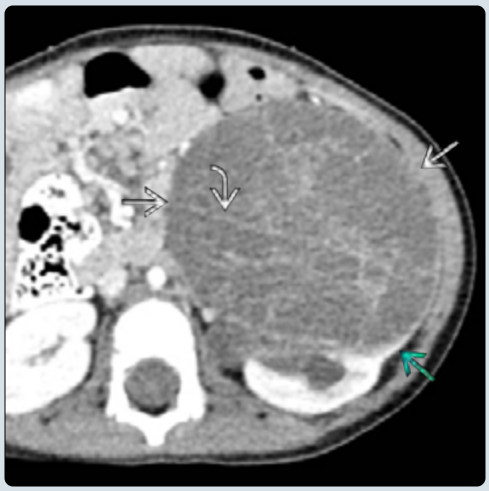
- > 70% of multilocular cystic nephromas (MLCNs) have *DICER1* gene mutation
  - Tumor predisposition syndrome at risk for pleuropulmonary blastoma & other malignancies
  - Anaplastic sarcoma of kidney may arise in *DICER1* associated MLCN

CLINICAL ISSUES

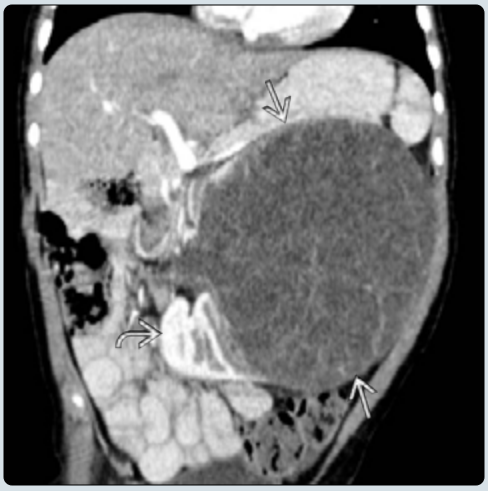
- Bimodal age & sex distribution
  - Children: M:F = 2-3:1; occurs between 3 months & 2 years
  - Adults: F:M = 4-9:1; occurs from 40-60 years
- Presentation
  - Painless abdominal/flank mass
  - Hematuria & urinary tract infection less frequent in children
- Benign mass with excellent prognosis overall
  - Resection usually curative

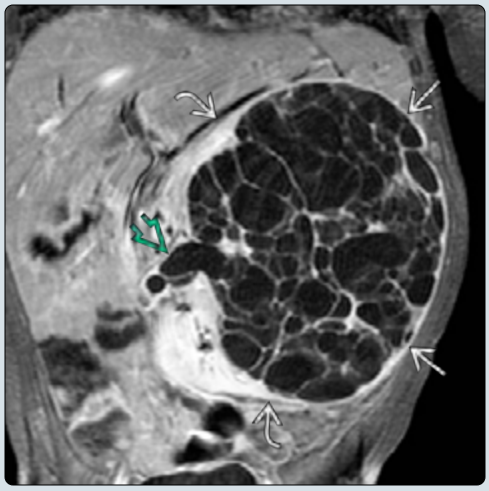
(Left) *Transverse ultrasound of the left kidney in a 2-year-old girl shows a large, multiloculated mass. While multilocular cystic nephroma is more common in young boys, 25-40% occur in girls.* **(Right)** *Axial CECT in the same patient shows a large cystic mass arising from the left kidney. There are multiple fine septations throughout the mass, which demonstrate mild enhancement. Note the "claw" of enhancing renal tissue stretched over the mass, a useful sign in determining the organ of origin for a large mass.*





(Left) *Coronal CECT shows a large cystic mass occupying the upper pole of the left kidney. The normal lower pole of the kidney is displaced inferiorly & medially by the mass.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows only septal enhancement of the large multicystic renal mass, which otherwise followed fluid signal on all sequences. There is a "claw" of renal tissue along the medial border of the mass. A portion of the mass herniates into the renal pelvis, typical of multilocular cystic nephroma.*





TERMINOLOGY

Abbreviations

- Multilocular cystic nephroma (MLCN); cystic partially differentiated nephroblastoma (CPDN)

Definitions

- Benign mixed mesenchymal & epithelial renal neoplasm

IMAGING

General Features

- Best diagnostic clue
  - Large multilocular cystic renal mass in young male or middle-aged female
- Size
  - Mean size: 9-10 cm
- Morphology
  - Well-circumscribed cystic mass with thick fibrous capsule
    - Fluid-filled locules of varying size with thin septa
    - ± herniation of mass into renal pelvis

CT Findings

- Large, well-defined, multiloculated cystic mass
  - Mass may herniate into renal hilum
  - Cystic components have attenuation equal to or slightly greater than water
  - Septa have variable enhancement
  - No excretion of contrast into cystic components of mass

MR Findings

- Multiloculated cystic mass
  - Splayed "claw" of normal, compressed renal tissue stretched along mass periphery
  - Cystic components usually follow fluid signal intensity on all sequences with no contrast excretion into cysts
  - Fibrous septa & capsule hypointense on T1 & T2 with variable enhancement
- Herniation of mass into renal pelvis

Ultrasonographic Findings

- Large, well-defined multiloculated cystic renal mass
  - Innumerable anechoic cysts + hyperechoic septa/capsule
  - Septa have variable thickness

Imaging Recommendations

- Best imaging tool
  - US: Excellent for investigating palpable mass in child
  - CT or MR with contrast: Further characterize mass & define extent

DIFFERENTIAL DIAGNOSIS

Wilms Tumor

- Most common renal neoplasm of childhood
- Predominantly cystic form uncommon

Multicystic Dysplastic Kidney

- Presents in newborn/neonate, unlike MLCN
- No normal renal parenchyma

Mesoblastic Nephroma

- Presents < 1 year of age
  - Most common renal neoplasm of infancy
- Can be mixed cystic & solid

PATHOLOGY

General Features

- Genetics
  - *DICER1* (14q32.13) mutation inactivates tumor suppressor gene → autosomal dominant tumor predisposition syndrome, at risk for
    - Pleuropulmonary blastoma (PPB)
    - MLCN: > 70% of have *DICER1* mutation
      - > 80% of these patients develop PPB
    - Ovarian sex cord-stromal tumor
    - Embryonal rhabdomyosarcoma of urinary bladder or uterine cervix
  - Low penetrance (15%) → no tumors in most carriers

Microscopic Features

- Septa lined by flattened or cuboidal epithelium with areas of eosinophilic cuboidal cells protruding into lumen
- MLCN & CPDN differentiated by septa
  - MLCN: No undifferentiated elements
  - CPDN: Contain blastemal ± other embryonal elements

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless abdominal/flank mass
- Other signs/symptoms
  - Hematuria & urinary tract infection less frequent in children

Demographics

- Overall bimodal age & sex distribution
  - 3 months to 2 years of age: 2-3x more common in males
  - 40-60 years of age: 4-9x more common in females
- *DICER1* associated MLCN: < 4 years of age

Natural History & Prognosis

- MLCN: Benign mass with excellent prognosis in isolation
- Reports of *DICER1* associated anaplastic sarcoma of kidney (ASK) arising in MLCN

Treatment

- Complete or partial nephrectomy usually curative

DIAGNOSTIC CHECKLIST

Consider

- *DICER1* genetic testing, annual chest radiographs (for PPB)

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Mesoblastic Nephroma

KEY FACTS

TERMINOLOGY

- Mesoblastic nephroma (MN): Hamartomatous renal tumor of young infants
- Composed predominantly of elongated spindle cells
- Classic benign vs. more aggressive cellular variants
  - Studies vary on which type more common

IMAGING

- Solitary renal mass in fetus or infant
- Well-defined oval/round mass
  - Classic type usually solid, smaller
  - Cellular type usually larger with cystic/necrotic/hemorrhagic foci

PATHOLOGY

- Classic type similar to infantile myofibroma
- Cellular type similar to infantile fibrosarcoma
  - Local recurrence, metastases

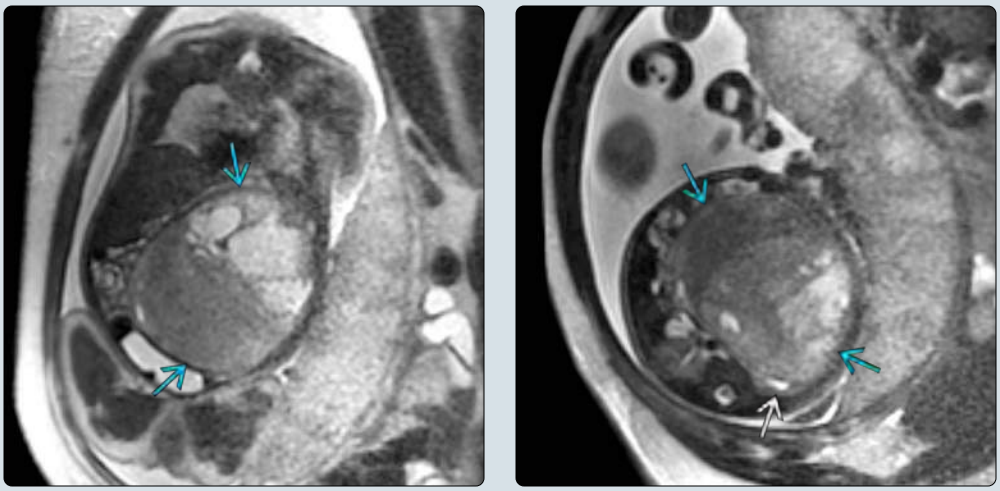
CLINICAL ISSUES

- 3-6% of childhood renal tumors
- Presentations include
  - Palpable abdominal mass in young infant
  - Hypertension, hypercalcemia, hematuria
  - Prenatally detected renal mass with polyhydramnios (70%), preterm labor
- Most MN diagnosed before 3 months of age
  - Classic type more common in this timeframe
- After 3 months
  - Wilms tumor becomes more common
  - Remaining MN more likely to be cellular
- Nephrectomy with wide margins usually curative

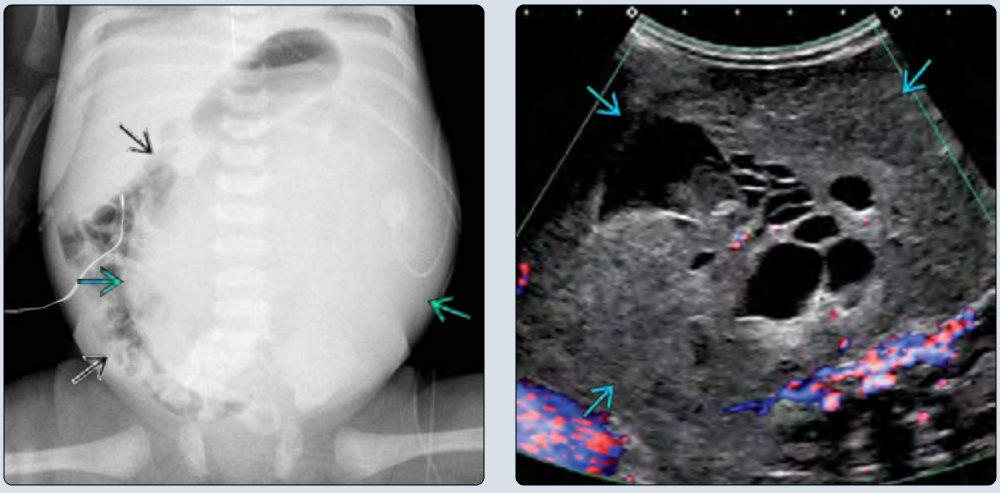
DIAGNOSTIC CHECKLIST

- Preoperative feature for best differentiating solid renal masses in children: Age

(Left) *Coronal T2 SSFSE MR image in a 37-week gestation fetus shows a large mixed cystic & solid mass ⇨ in the left fetal abdomen.* **(Right)** *Axial T2 SSFSE MR in the same fetus shows the relationship of the large mass ⇨ to the residual left kidney ⇨, though it is difficult to tell if the kidney is splayed over the mass or merely compressed by it. Additionally, the amniotic fluid volume is greater than typically seen for this gestational age.*



(Left) *Postnatal AP radiograph of the abdomen in the same patient shows displacement of gas-filled bowel loops ⇨ into the right abdomen by a large round mass ⇨.* **(Right)** *Longitudinal color Doppler ultrasound of the abdomen in the same patient shows the 11-cm mixed cystic & solid mass ⇨ with little detectable internal vascularity. The cellular subtype of mesoblastic nephroma was confirmed upon resection.*



TERMINOLOGY

Synonyms

- Congenital mesoblastic nephroma (MN), fetal renal hamartoma, Bolande tumor
  - Bolande described MN as histologically distinct from Wilms tumor in 1967

Definitions

- Hamartomatous renal tumor of young infants composed predominantly of elongated spindle cells
  - Classic benign MN vs. more aggressive cellular variant

IMAGING

General Features

- Best diagnostic clue
  - Solitary renal mass in fetus or neonate
- Location
  - Intrarenal but may replace entire kidney & cross midline when large
  - Metastases very uncommon
- Size
  - Variable, from < 1 cm diameter to > 15 cm
    - Cellular variant often much larger than classic type
- Morphology
  - Well-defined oval/round mass
    - Usually solid
    - Cystic/necrotic/hemorrhagic foci imply more aggressive cellular variant
  - "Claw" of residual renal parenchyma splayed along tumor margin helps confirm renal origin
  - Hydronephrosis & vascular invasion not typical
  - Irregularity/discontinuity of margins, infiltration of adjacent soft tissues, surrounding fluid &/or free pelvic fluid suggest tumor rupture
    - More common with cellular type

Radiographic Findings

- Mass effect from large round tumor

CT Findings

- NECT
  - Ca^{2+} not typically seen
  - Pockets of high attenuation corresponding to hemorrhage in cellular type
- CECT
  - Heterogeneous enhancement of solid components
  - Cystic/necrotic/hemorrhagic foci of nonenhancement typical of cellular type

MR Findings

- T1WI
  - Intermediate to dark
  - Hemorrhage may be bright
- T2WI
  - Variable for solid components
  - Bright cystic foci ± fluid-fluid levels from hemorrhage
- T1WI C+
  - Heterogeneous enhancement of solid components
  - Rim/septal enhancement of cystic foci

Ultrasonographic Findings

- Grayscale ultrasound
  - Smaller masses leave renal shape largely intact
    - Smooth contours to mass by imaging
  - Larger masses may fill abdomen & cross midline
  - Variable echogenicity
  - Hyper- & hypoechoic ring pattern at periphery, more common in classic type
  - Prenatal findings include polyhydramnios (70%), rarely hydrops
- Color Doppler
  - Vascular invasion not seen

Imaging Recommendations

- Best imaging tool
  - Ultrasound best initial tool for investigating palpable abdominal mass in child
  - MR will better define local extension, adenopathy

DIFFERENTIAL DIAGNOSIS

Wilms Tumor

- Most common pediatric renal tumor
- Imaging appearance may be identical to MN
- Best preoperative discriminator from MN: Age
  - Average presentation of Wilms: 3-4 years of age
  - Rare in utero

Neuroblastoma

- Suprarenal/paraspinal mass displacing (or rarely invading) kidney
- Often contains Ca^{2+}
- Frequently engulfs & displaces vessels, crosses midline

Adrenal Hemorrhage

- Avascular cystic or heterogeneous suprarenal mass in newborn
- Decreasing size over time

Autosomal Recessive Polycystic Kidney Disease

- Markedly enlarged, echogenic & striated kidneys bilaterally

Multicystic Dysplastic Kidney

- Cysts of varying size completely replacing renal parenchyma
  - Not cysts within well-defined round mass

Ossifying Renal Tumor of Infancy

- Extremely rare tumor characterized by ossification/Ca^{2+}

Rhabdoid Tumor

- Aggressive rare solid renal tumor of infancy
- Association with CNS atypical teratoid rhabdoid tumors

Clear Cell Sarcoma of Kidney

- 2nd most common pediatric renal tumor
- Classically causes bone metastases
- Average presentation: 1-4 years of age

Ureteropelvic Junction Obstruction

- Moderate to marked hydronephrosis, often with disproportionate pelvis dilation
- No hydroureter or bladder abnormality

Multilocular Cystic Nephroma

- Large multicystic mass herniating into central collecting system
- Average presentation: 3 months to 2 years of age

PATHOLOGY

General Features

- Genetics
  - Sporadic
  - No recurrence risk in siblings
  - Cellular subtype of MN shares t(12;15)(p13;q25) chromosomal translocation with infantile fibrosarcoma
    - Fusion of genes *ETV6* & *NTRK3*

Staging, Grading, & Classification

- 3 histologic subtypes (reports vary regarding most common type)
  - Classic: Similar features to infantile myofibroma/fibromatosis
  - Cellular or atypical: Similar features to infantile fibrosarcoma
  - Mixed
- Staging by Wilms tumor criteria

Gross Pathologic & Surgical Features

- Whorled appearance, similar to uterine fibroid
- No capsule despite well-defined imaging appearance
- Cut surface usually yellow-tan, solid, rubbery stromal tissue
- Cystic, hemorrhagic foci in cellular type

Microscopic Features

- Classic: Bundled spindle cells, entrapped tubules & glomeruli, few mitoses
- Cellular: Sheets of randomly arranged spindle cells with few bundles, high mitotic rate, nuclear atypia

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Palpable abdominal mass in young infant
  - Prenatally detected renal mass with polyhydramnios, preterm labor
- Other signs/symptoms
  - Hypertension, hypercalcemia, hematuria

Demographics

- Age
  - Usually diagnosed in first 6 months of life
    - Most MN diagnosed before 3 months of age
      - Classic type most likely in this period
    - After 3 months
      - Wilms tumor becomes more common
      - Remaining mesoblastic nephromas more likely to be cellular variants
- Gender
  - 1-2:1 = M:F
- Ethnicity
  - No ethnic predisposition
- Epidemiology
  - 3-6% of childhood renal tumors
  - 93% of renal tumors detected in utero
  - 90% diagnosed before 1 year of age
    - Wilms tumor more common after 3 months of age

Natural History & Prognosis

- Can show rapid growth despite benign histology
- Large abdominal circumference may result in dystocia at delivery
- Excellent prognosis for classic subtype
  - Complete surgical resection curative
- Cellular subtype more aggressive
  - 10% relapse with local recurrence or metastases
    - Most common metastatic site: Lung
    - Liver, heart, brain, & bone metastases also reported

Treatment

- Amnioreduction for polyhydramnios
- Referral to pediatric surgeon/urologist
- Resection in neonatal period
  - Nephrectomy with wide margins usually curative
  - Adjuvant chemotherapy for patients > 3 months of age with stage III (incomplete resection/positive margins) cellular type
- 5-year event-free survival of 94%

DIAGNOSTIC CHECKLIST

Consider

- Best differentiating factor for solid renal masses in children: Age

Image Interpretation Pearls

- Look for residual splayed renal parenchyma
  - Claw sign helps confirm renal origin
  - Inferiorly displaced, distorted kidney suggests extrarenal tumor
- Look for findings related to rupture
  - Cysts predispose to intraoperative rupture
  - Surrounding soft tissue findings may suggest preoperative tumor rupture

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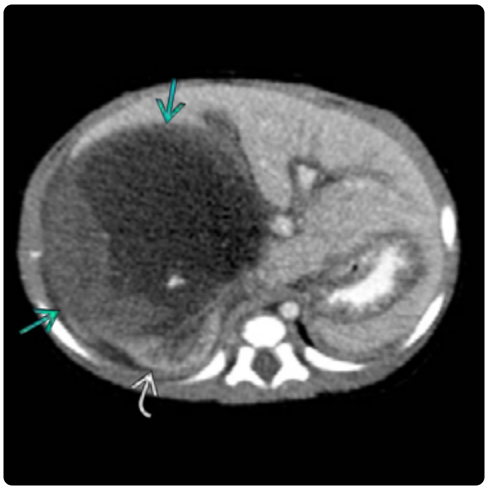
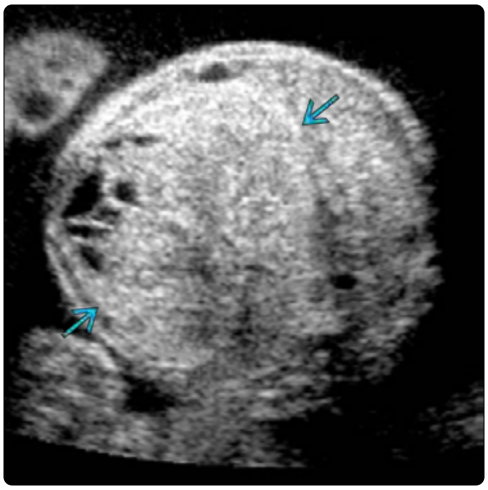
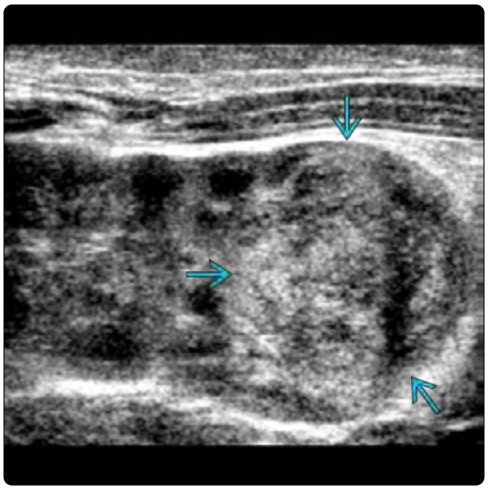
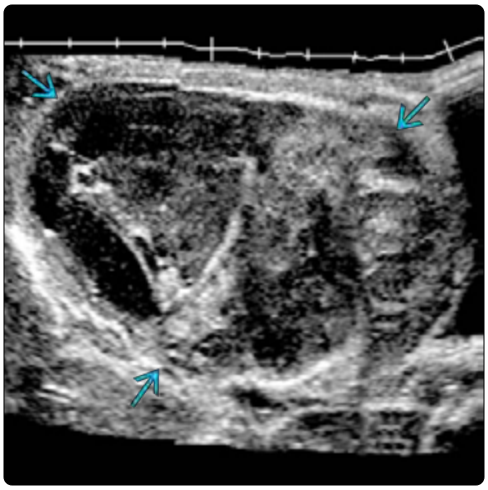


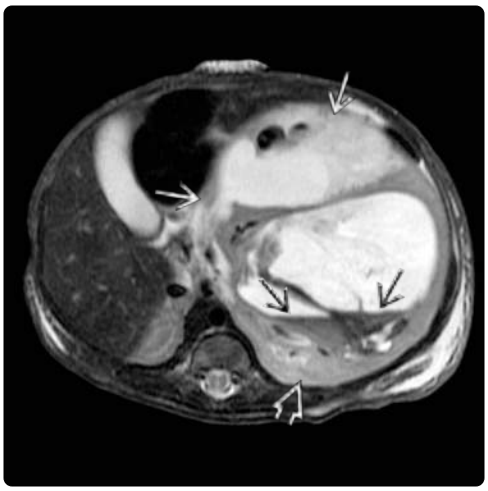


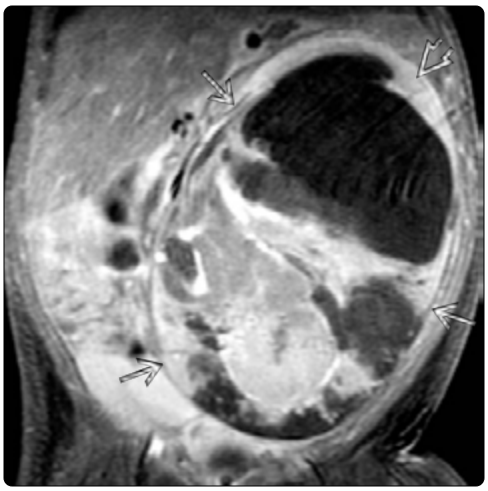
(Left) *Axial CECT in a patient with cellular mesoblastic nephroma shows a crescent of residual right kidney splayed along the posterior margin of the heterogeneous mass. This claw sign helps to identify the kidney as the organ of origin for the tumor.* **(Right)** *Transverse ultrasound in a 2nd trimester fetus with polyhydramnios shows a large predominantly solid mass in the right abdomen. No right kidney could be identified. Mesoblastic nephroma was confirmed postnatally at resection.*





(Left) *Longitudinal ultrasound in a newborn shows a relatively small solid (but heterogeneous) mass involving only the lower pole of this kidney. The patient age, tumor size, & imaging features are typical for the classic type of mesoblastic nephroma.* **(Right)** *Longitudinal panoramic ultrasound in a 5 day old with abdominal fullness shows a large, heterogeneous mass in the left flank replacing most of the left kidney.*





(Left) *Axial T2 FS MR in the same patient shows a heterogeneous mass in the left flank. The layering fluid-fluid levels are typical of hemorrhage within cysts. A "claw" of the splayed residual left kidney is seen posteriorly.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows heterogeneous enhancement of the tumor with large foci of nonenhancement. A "claw" of residual renal parenchyma is seen superiorly. Resection of this cellular mesoblastic nephroma was complicated by intraoperative rupture/spill.*

Rhabdoid Tumor

KEY FACTS

TERMINOLOGY

- Rare, highly aggressive neoplasm in young children
- Commonly arises from kidney
- Extrarenal sites include CNS, soft tissues > liver, lung

IMAGING

- Contrast-enhanced CT or MR most useful
  - Large, heterogeneous renal mass
    - Foci of hemorrhage, necrosis, &/or Ca^{2+} separating tumor lobules
      - Ca^{2+} more common than Wilms
  - Crescentic subcapsular fluid collection common
  - Sinus/hilar, local, & vascular invasion common
  - Metastasizes most frequently to lungs

PATHOLOGY

- ~ 2% of pediatric renal tumors
- Associated with inactivation or deletion of *SMARCB1* (hSNFS/INI1) gene on chromosome 22q11

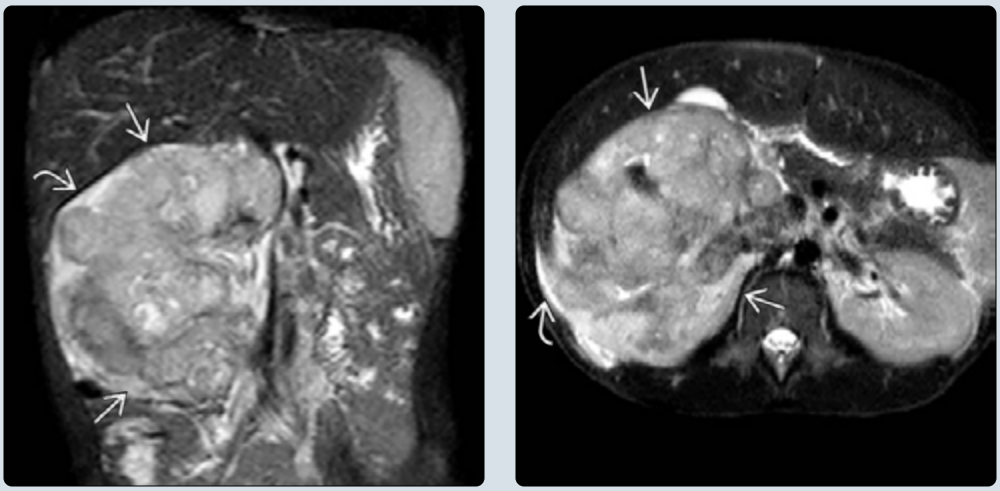
CLINICAL ISSUES

- Mean age of presentation: ~ 11 months
  - Younger than mean age for Wilms tumor
- Signs/symptoms
  - Most common: Palpable abdominal mass, hematuria (gross or microscopic)
  - Others include fever, anemia, hypercalcemia
- Aggressive neoplasm with poor prognosis
  - Patients often present with advanced disease
    - Stages 3-4 in ~ 70% at presentation
  - Overall 5-year survival around 20%
  - Prognosis worse in younger patients (< 6 months of age)

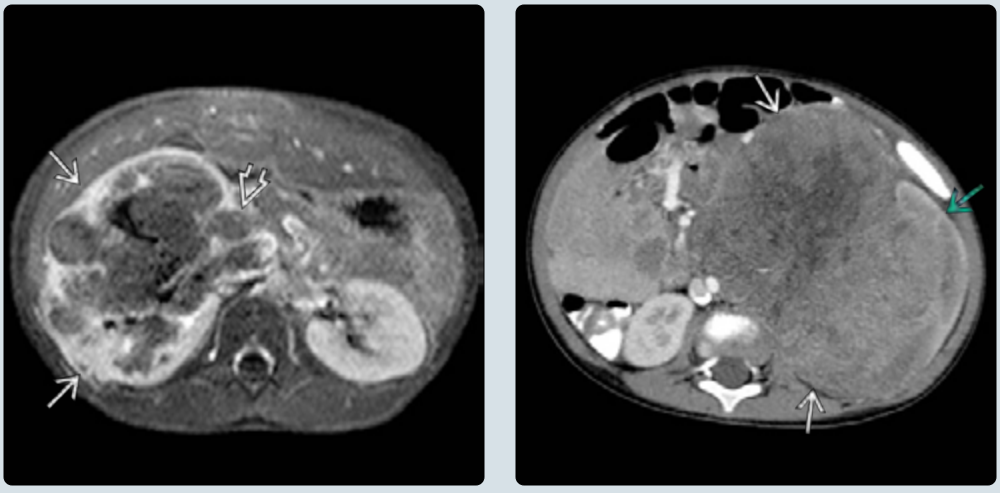
DIAGNOSTIC CHECKLIST

- Consider brain MR to evaluate for synchronous or metachronous CNS neoplasm

(Left) *Coronal T2 FS MR in an 11-month-old boy ultimately diagnosed with a rhabdoid tumor shows a large, heterogeneous, lobulated mass ➡ replacing the right kidney. A subcapsular fluid collection ➡ is noted at the periphery of the mass.* **(Right)** *Axial T2 FS MR in the same patient with a right renal rhabdoid tumor shows a crescentic focus of subcapsular fluid ➡ at the periphery of the heterogeneous tumor ➡.*



(Left) *Postcontrast T1 C+ FS MR in the same patient demonstrates heterogeneous enhancement of the large right renal mass ➡. Note the hilar invasion by the tumor lobules ➡.* **(Right)** *Axial CECT in a 14 month old with a renal rhabdoid tumor demonstrates a very large mass with heterogeneous enhancement ➡ arising from the left kidney ➡.*



KEY FACTS

TERMINOLOGY

- Bone metastasizing tumor of childhood
- Rare malignant renal neoplasm in young children
  - ~ 3% of pediatric renal tumors

IMAGING

- CECT or MR best for characterizing tumor & evaluating local extension
  - Best clue: Large, heterogeneous renal mass + osseous metastases in young child
    - Cystic & necrotic foci common
    - Ca^{2+}: 25%
    - Vascular invasion: 5%
    - Regional lymph node involvement: Up to 30%
    - Delayed recurrence in bones, lungs
- Metastatic work-up: Chest CT & nuclear medicine bone scan

PATHOLOGY

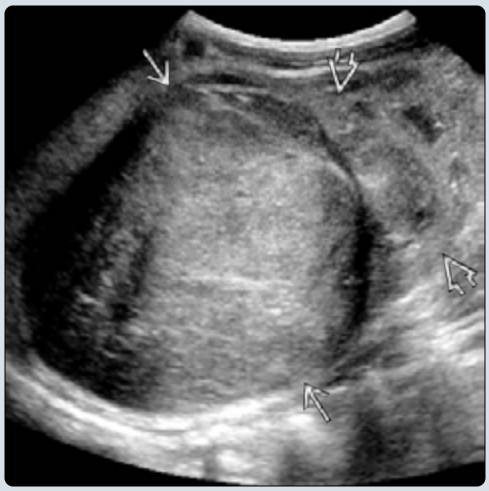
- Histologically heterogeneous: Clear cells may be absent
- Staging similar to Wilms tumor

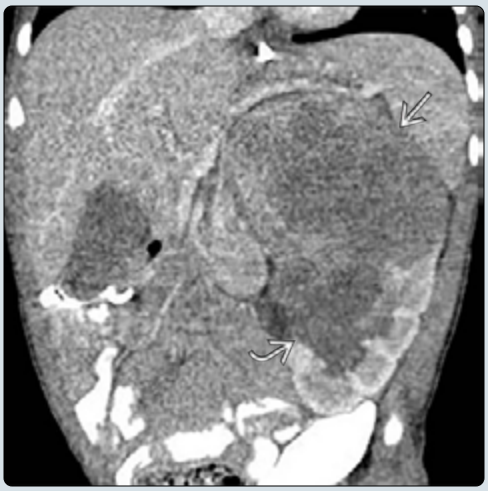
CLINICAL ISSUES

- Most common presentation: Palpable abdominal mass
  - ± hematuria
  - Rarely presents with pain from osseous metastases
- Mean age at presentation: 36 months
- Male predominance of 2:1
- 5-year survival ~ 70%
  - Up to 98% for stage I disease
  - ~ 50% for stage IV disease
- Treatment: Combination chemotherapy (including doxorubicin), surgery, & radiation (depending on stage)

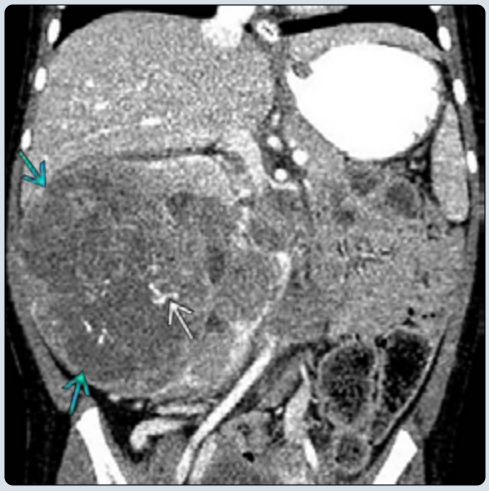
DIAGNOSTIC CHECKLIST

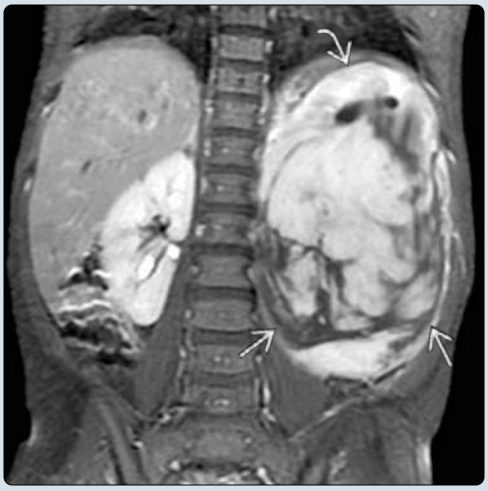
- Pediatric solid renal masses rarely distinguishable by imaging alone
  - Age, clinical history, pattern of spread helpful





(Left) *Longitudinal ultrasound in a 2-year-old boy with a palpable abdominal mass shows a solid mass arising from the upper pole of the left kidney. Note the splayed residual left renal parenchyma forming the claw sign.* **(Right)** *Coronal CECT in the same patient shows a large, hypoenhancing mass arising from the upper pole of the left kidney. The mass is causing obstruction of the lower portion of the left renal collecting system. Clear cell sarcoma was confirmed at resection.*





(Left) *Coronal CECT in a 3-year-old girl with clear cell sarcoma shows a heterogeneous mass arising from the right kidney. Note the Ca^{2+} within the mass.* **(Right)** *Coronal T1 C+ FS MR in an 18-month-old boy with clear cell sarcoma shows a large, heterogeneous mass arising from the left kidney with areas of nonenhancement corresponding to necrosis. Some preserved renal parenchyma is seen in the upper pole.*

Ossifying Renal Tumor of Infancy

KEY FACTS

IMAGING

- Best clue: Small renal pelvic mass with Ca^{2+} in infant
- CECT
  - Polypoid or lobulated mass within collecting system
  - Ca^{2+} typically present
    - Hyperattenuation may be vague with only microCa^{2+}
  - Variable enhancement
- MR: May be low in signal intensity on T2 due to Ca^{2+}
- Ultrasound
  - Small, echogenic mass within collecting system
    - ± collecting system dilation due to partial obstruction
  - Posterior acoustic shadowing due to Ca^{2+}
  - Variable internal vascularity
    - Presence of true internal flow helps separate lesion from nonneoplastic material filling collecting system

TOP DIFFERENTIAL DIAGNOSES

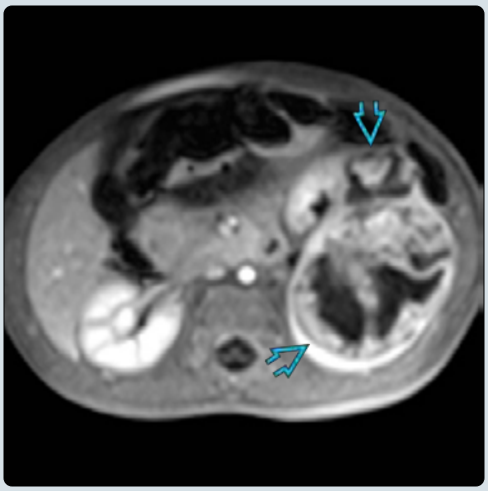
- Staghorn calculus
  - Large Ca^{2+} filling variable degrees of collecting system
  - No detectable pulsed Doppler signal
  - Extremely rare in young children
- Mesoblastic nephroma
  - Most common renal neoplasm < 3 months old
  - Solid or mixed solid & cystic; Ca^{2+} uncommon
- Wilms tumor
  - Most common pediatric renal neoplasm > 3 months old
  - Typically solid; Ca^{2+} uncommon

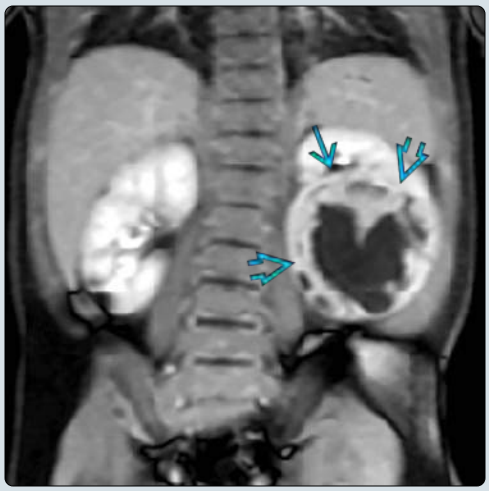
PATHOLOGY

- Originates from medullary pyramid papilla
- 3 histological elements, including osteoid cores, osteoblasts, & spindle cells

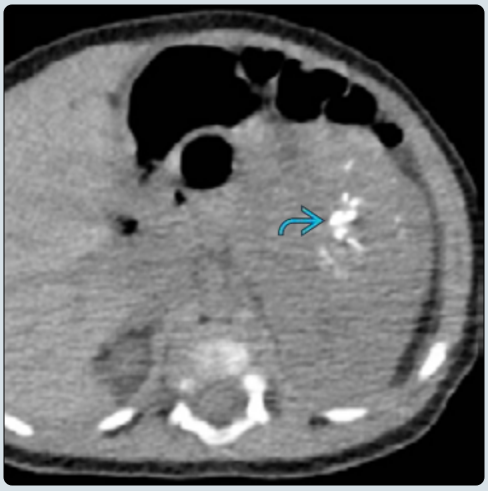
CLINICAL ISSUES

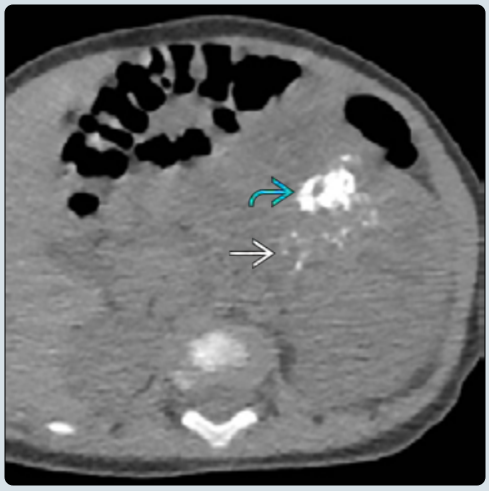
- Most common presentation: Hematuria under 1 year of age
- Benign clinical behavior
  - No reports of metastatic spread or recurrence
- Surgical resection (renal sparing if possible) curative





(Left) *Axial T1 C+ FS MR in a 5-month-old boy with hematuria shows a lobulated, heterogeneously enhancing mass in the left kidney.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows that the heterogeneously enhancing mass is centered in the left renal lower pole but protrudes into the collecting system centrally. The mass is larger than typically seen with an ossifying renal tumor of infancy.*





(Left) *Axial NECT of the upper abdomen (done as part of a metastatic work-up chest CT in the same 5-month-old patient) shows coarse, dense Ca^{2+} within the left renal mass, typical of an ossifying renal tumor of infancy.* **(Right)** *Slightly inferior axial NECT of the upper abdomen in the same patient shows the Ca^{2+} within the left renal mass extending into the region of the central collecting system, typical of an ossifying renal tumor of infancy.*

KEY FACTS

TERMINOLOGY

- Rapidly growing tumor of renal medulla

IMAGING

- Best clue: Infiltrating, poorly defined renal mass in young adult with sickle cell trait
  - Tumor arises in renal medulla with extension towards renal sinus & cortex
  - Heterogeneous tumor enhancement due to necrosis
  - Kidney may maintain reniform shape
  - Central masses may cause caliectasis
  - Extension to perinephric tissues common
- 70% occur in right kidney
- Mean size: 6 cm
- Lung metastases & lymph node involvement common
  - Lung metastases often have lymphangitic pattern with clustered or diffuse poorly defined nodules intermixed with interstitial thickening

TOP DIFFERENTIAL DIAGNOSES

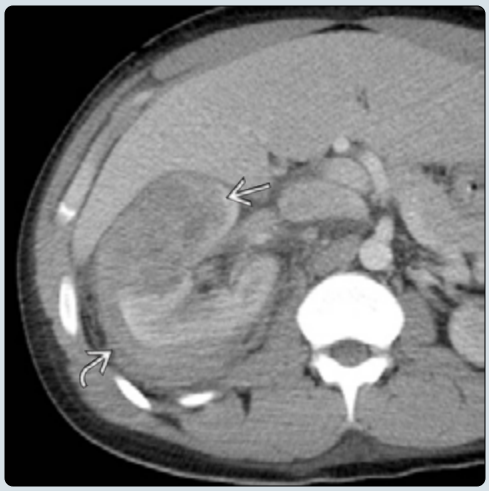
- Pyelonephritis with developing abscess
- Wilms tumor
- Renal cell carcinoma

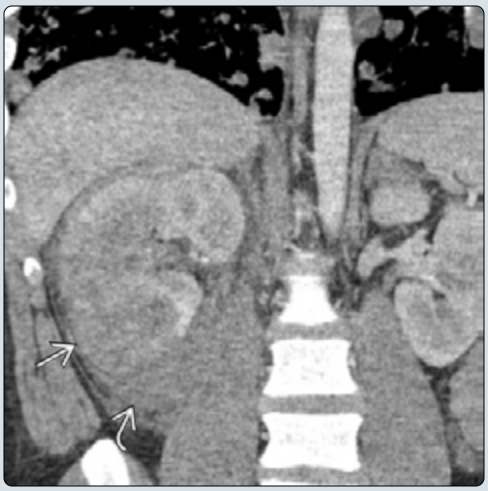
PATHOLOGY

- 88% occur in patients with sickle cell trait
  - May be related to sickling blood & local renal environment

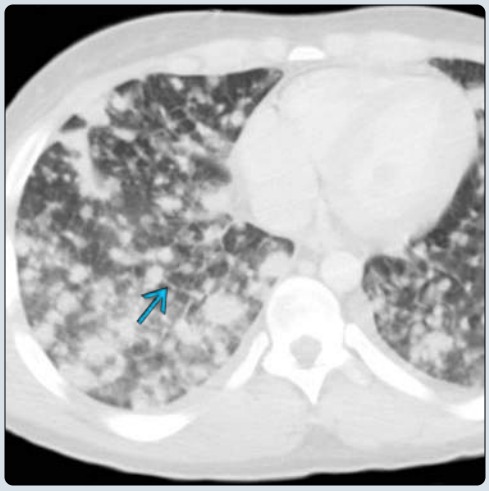
CLINICAL ISSUES

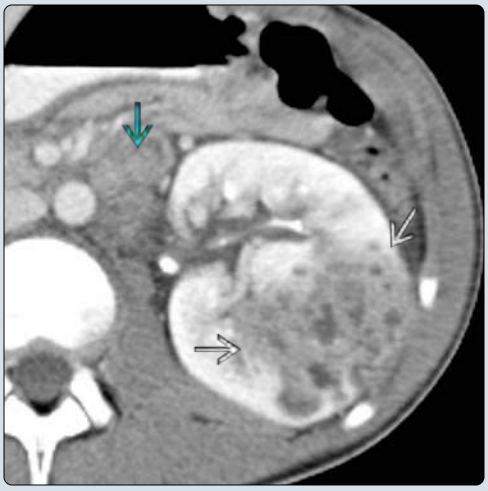
- Most common presenting symptoms: Hematuria & pain
- Other symptoms: Weight loss, respiratory symptoms, nausea, & vomiting
- 2.4x more common in males
- Therapy has not not been effective
  - Typically treated with nephrectomy & chemotherapy
  - Poor prognosis with median overall survival of 5 months





(Left) *Axial CECT in a young adult with sickle cell trait & renal medullary carcinoma (RMC) shows a poorly defined infiltrative mass of the right kidney with perinephric extension. RMC is more common in the right kidney.* **(Right)** *Coronal CECT in the same patient shows the infiltrative mass of the right kidney with associated perinephric extension. Extensive lung metastases are visible. RMC characteristically has an infiltrative appearance, & the kidney may maintain its reniform shape.*





(Left) *Axial CECT in the same patient shows extensive lung metastases with nodular opacities intermixed with interstitial thickening, typical of lymphangitic spread. Metastases are common in RMC with the most common sites being lymph nodes, lungs, & liver.* **(Right)** *Axial CECT in a young adult with flank pain & sickle cell trait shows an infiltrative, heterogeneous mass of the left kidney. This typical heterogeneous enhancement pattern is caused by necrosis & hemorrhage. Note the adjacent adenopathy.*

Angiomyolipoma

KEY FACTS

TERMINOLOGY

- Benign hamartomatous tumor consisting of abnormal blood vessels, smooth muscle, & fat
- Almost always associated with tuberous sclerosis complex (TSC) in children

IMAGING

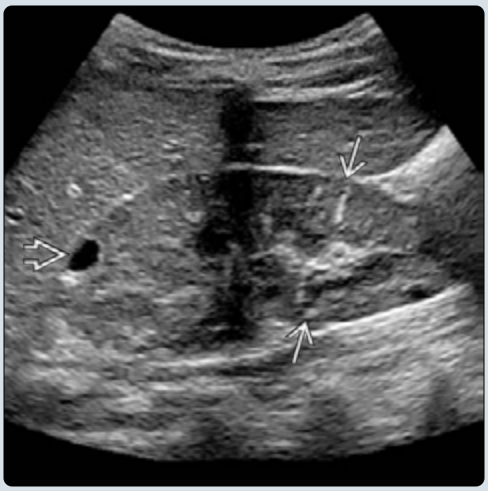
- Renal masses showing variable amounts of fat
  - Fat-containing renal mass diagnostic of angiomyolipoma (AML) in child
    - Signal dropout on opposed-phase MRs
    - Contrast not needed for diagnosis with known TSC
      - Potentially useful for vascular evaluation
- Bilateral AMLs in 95% of TSC patients
- Vast majority of AMLs occur in kidneys; liver, pancreas, other abdominal organs less common
- Benign renal cysts also common in TSC
- Tortuous vessels ± aneurysms in larger AMLs
  - Vessels may be splayed by fatty components or cysts

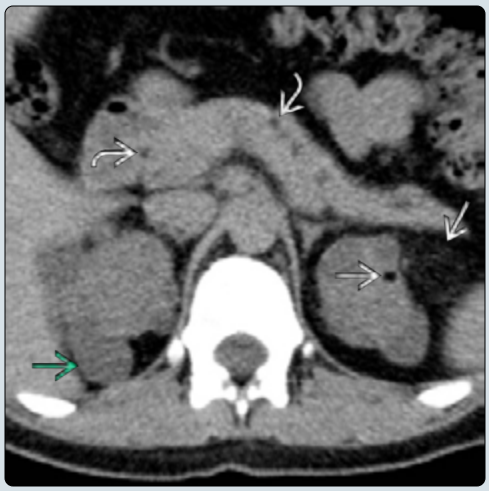
PATHOLOGY

- 2 patterns of disease: Sporadic vs. TSC-associated AMLs
  - Sporadic lesions extremely rare in children
- Underlying mutations of tumor suppressor genes *TSC1* or *TSC2* → dysregulation of mammalian target of rapamycin (mTOR) → ↑ cell growth & division
- Lymphangioleiomyomatosis: Numerous thin-walled pulmonary cysts in TSC patients, likely metastatic AMLs

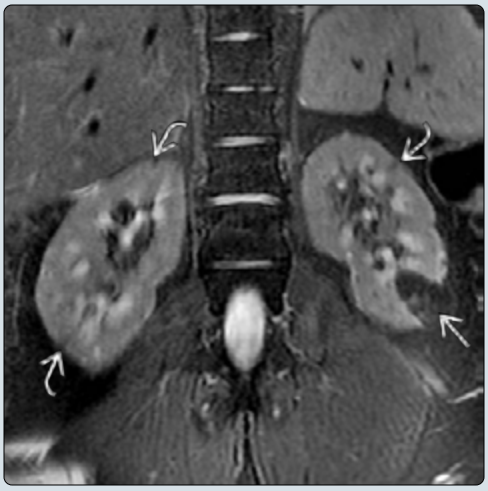
CLINICAL ISSUES

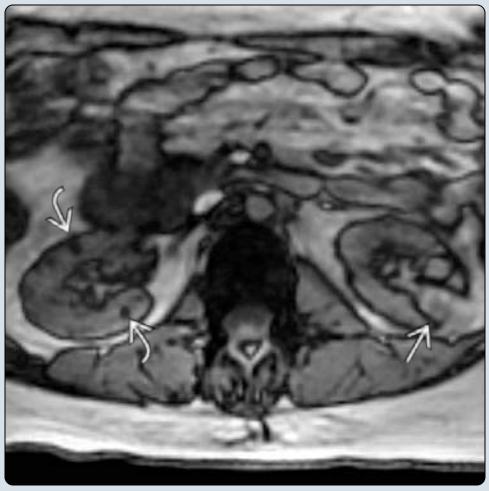
- Pediatric AMLs usually asymptomatic
  - Children typically have clinical (especially cutaneous & neurologic) manifestations of TSC
- Conservative symptomatic management for AMLs in TSC
  - mTOR inhibitors used for lesions > 3 cm in size
- Possible life-threatening spontaneous hemorrhage
  - Hemorrhage rare if AML ≤ 4 cm or aneurysm < 5 mm
    - Risk from sporadic AML data may not apply to TSC
  - Treated with arterial coil embolization





(Left) *Longitudinal US of the right kidney in a patient with tuberous sclerosis complex (TSC) shows multiple small, echogenic foci consistent with small angiomyolipomas (AMLs). A small simple cyst is also present in the upper pole.* **(Right)** *Axial NECT shows multiple lesions of both kidneys. Some lesions are of fat density while others are hypodense compared to the liver & paraspinal muscles. The fat density lesions are AMLs while the other hypodense lesions represent cysts. Small AMLs are also present in the pancreas.*





(Left) *Coronal T2 FS MR of the kidneys shows multiple renal AMLs bilaterally. The largest lesion is in the lower pole of the left kidney & follows fat signal intensity. Multiple tiny, fat-containing lesions are also seen along the renal cortex bilaterally.* **(Right)** *Axial T1 opposed-phase MR shows multiple AMLs of both kidneys. The large lesion in the left kidney has a peripheral rim of signal dropout at its interface with the normal renal parenchyma. The smaller lesions show complete internal signal dropout.*

TERMINOLOGY

Abbreviations

- Angiomyolipoma (AML)
- Tuberous sclerosis complex (TSC)

Definitions

- Benign hamartomatous tumor consisting of abnormal blood vessels (angio), smooth muscle (myo), & fat (lipoma)

Associated Syndromes

- Almost always associated with TSC in children

IMAGING

General Features

- Best diagnostic clue
  - Renal masses containing variable amounts of fat
  - Benign renal cysts also common in patients with TSC
    - Variable signal depending on proteinaceous content
    - Cysts do not contain fat or enhance
  - Cysts in lung bases in TSC patient → Lymphangioleiomyomatosis (LAM)
- Location
  - Bilateral renal AMLs in 95% of TSC patients
  - Vast majority of AMLs occur in kidneys
    - Can occur in liver, pancreas, & other abdominal organs
    - Hepatic AMLs in 13% of TSC patients with renal AMLs
      - Hepatic AMLs typically multiple
      - Renal AMLs typically diffuse in this setting
- Size
  - Variable: Range from tiny (mm) to large (many cm)
- Morphology
  - Variable shape, fat content, & number in kidneys

CT Findings

- NECT
  - Renal mass with intramural fat → diagnostic of AML in children
  - Lipid-poor lesions slightly hyperdense compared to kidney
- CECT
  - Contrast not needed in setting of known TSC
  - Lesions may significantly enhance with contrast (depending on extent of vascular component)
  - Lipid-poor lesions show homogeneous & prolonged enhancement
- CTA
  - ± aneurysmal renal vessels in large AMLs

MR Findings

- T1WI
  - Mass contains variable amounts of high-signal fat
  - Low signal in fatty portion on fat-saturated images
- T2WI
  - Variable signal intensity
  - Low signal on fat-saturated images
- T1WI C+
  - Variable enhancement
    - With high fat content, may show minimal enhancement
    - With high vessel content, may show marked enhancement
  - Contrast not needed for diagnosis in setting of known TSC
    - May be useful for vessel assessment
- In phase/opposed phase
  - Signal dropout on opposed-phase images
    - Small lesions become completely hypointense
    - Large lesions have hypointense rim
  - Opposed phase useful to identify extrarenal AMLs

Ultrasonographic Findings

- Grayscale ultrasound
  - Hyperechoic mass relative to normal kidney & liver
  - Lipid-poor lesions may be isoechoic to slightly hypoechoic compared to kidney
- Color Doppler
  - Can detect aneurysms in larger lesions

Angiographic Findings

- Characteristic tortuous vessels with multiple aneurysms
- Vessels may be splayed by larger fatty lesions or cysts

Imaging Recommendations

- Best imaging tool
  - US to screen children with known TSC
    - Once lesions detected, US less useful due to ↓ reliability of lesion characterization & measurement
      - Particularly in setting of numerous &/or large, heterogeneous lesions
  - TSC patients generally followed with noncontrast MR
    - Minimize CT as these patients will undergo many studies during their lifetime
    - Renal MR performed at same time as brain MR
- Protocol advice
  - MR with in-phase/opposed-phase sequences most sensitive for detecting small lesions
  - T1 & T2 MR performed with & without fat saturation
  - IV contrast not necessary for diagnosis in patients with known TSC

DIFFERENTIAL DIAGNOSIS

Renal Cell Carcinoma

- Rare in children
- Rarely reported to contain fat
  - Lipid-poor AML may mimic renal cell carcinoma (RCC)
- Ca^{2+} in mass highly suggestive of RCC
  - Ca^{2+} extremely rare in AML
- Association of RCC with TSC controversial
  - Even if risk ↑, RCC remains very rare in TSC
    - Lipid-poor AMLs much more common

Renal Cyst

- Commonly seen in patients with TSC
- Hemorrhagic cyst or cyst with high protein content can mimic AML on MR (high T1, T2 signal)
- Fat-saturated MRs help differentiate fat from proteinaceous fluid

Wilms Tumor

- Most common pediatric renal malignancy

- Solid mass that rarely contains fat
- ± invasion of renal vein & inferior vena cava
- Metastases to lymph nodes, liver, lung
- ↑ frequency in certain syndromes
  - Beckwith-Wiedemann
  - Hemihypertrophy
  - Congenital aniridia
  - WAGR syndrome

Retroperitoneal Teratoma

- Heterogeneous, fat-containing mass; ± Ca^{2+}
- Displaces/compresses kidney rather than arising from it

PATHOLOGY

General Features

- Etiology
  - Benign renal tumor with mixed vascular, muscle, & fatty elements
  - 2 main patterns of disease
    - Sporadic lesions
      - ~ 80% of AMLs overall; however, sporadic AMLs extremely rare in children
      - Usually solitary
      - More common in females due to stimulation by estrogen/progesterone
      - Risk of hemorrhage in lesions ≥ 4 cm in size
    - TSC-associated lesions
      - Multiple bilateral tumors
      - Typically larger than sporadic AMLs
      - ± ↓ risk of hemorrhage vs. sporadic lesions
- Genetics
  - 2 tumor suppressor genes associated with TSC
    - *TSC1*, band 9q34, encodes for hamartin
    - *TSC2*, band 16p13.3, encodes for tuberin
  - Hamartin & tuberin help inhibit activation of mammalian target of rapamycin (mTOR)
    - Mutations lead to uncontrolled cell size & division
- Associated abnormalities
  - LAM: Numerous thin-walled cysts throughout lungs
    - Likely represent metastatic AMLs

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Characteristic skin & neurologic stigmata of TSC
  - AMLs usually asymptomatic
  - Possible life-threatening spontaneous hemorrhage
    - Flank/abdominal pain, hematuria
    - Risk factors for spontaneous hemorrhage
      - AML size > 4 cm, aneurysm size > 5 mm
      - Hemorrhage risk based on data from sporadic AMLs; may not apply to TSC
- Other signs/symptoms
  - Renal failure (usually not until adulthood)

Demographics

- Age
  - ± small AMLs in TSC patients in early childhood
    - 45% of have renal cysts or AMLs by 6 years of age
    - Frequency of renal lesions ↑ with age
- Gender
  - TSC-associated AMLs → M:F = 1:1
  - Sporadic AMLs much more common in females
- Epidemiology
  - AMLs in 0.3-3% of population in autopsy series
  - AMLs seen in up to 95% of children with TS

Natural History & Prognosis

- TSC-associated AMLs slowly enlarge into adulthood
- Massive replacement of renal parenchyma with AMLs may result in end-stage renal disease

Treatment

- Conservative, symptomatic management for patients with TSC-associated AMLs
  - Lesions can ↓ in size with mTOR inhibitors; reserved for lesions > 3 cm in size
- Treatment related to risk of hemorrhage in patients with sporadic AMLs
  - ≤ 4 cm: Conservative management with follow-up imaging
  - > 4 cm: Partial nephrectomy or arterial coil embolization

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Classic appearance: Well-circumscribed, fatty renal mass
- 3 renal lesion types seen on MR in patients with TSC
  - Lipid-rich AML: Bright on T1 & T2; ↓ signal with fat saturation
  - Lipid-poor AML: Isointense to muscle on T1 & T2; no change with fat saturation
  - Cyst (typical): Dark on T1, bright on T2; no change with fat saturation
    - Hemorrhage/protein in cyst may alter internal signal

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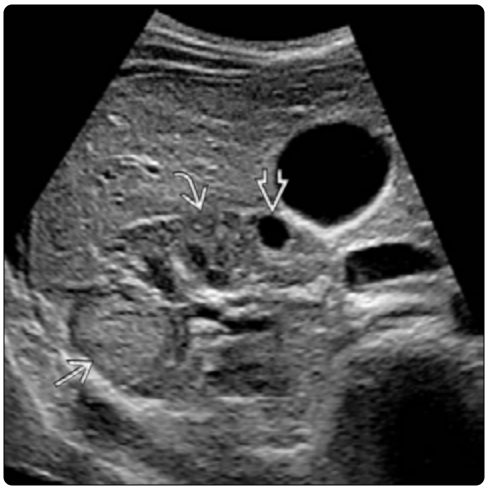
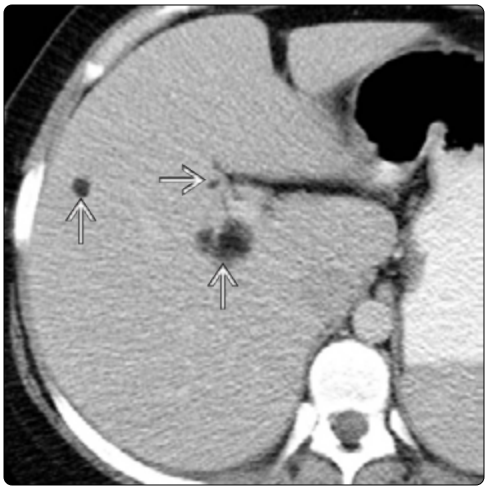
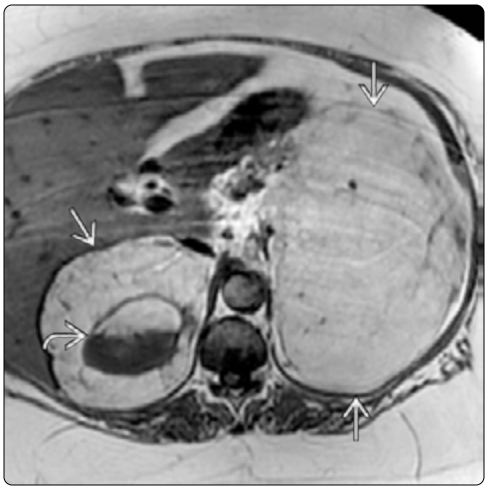
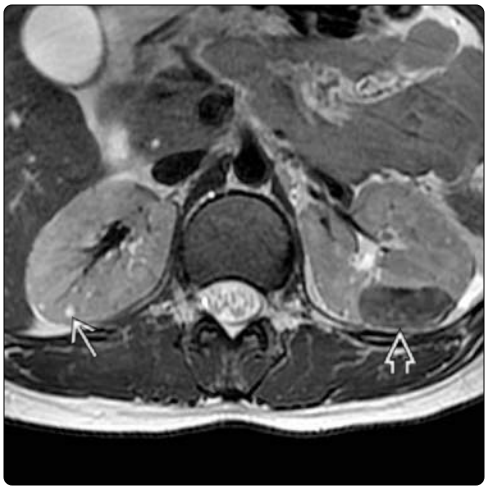


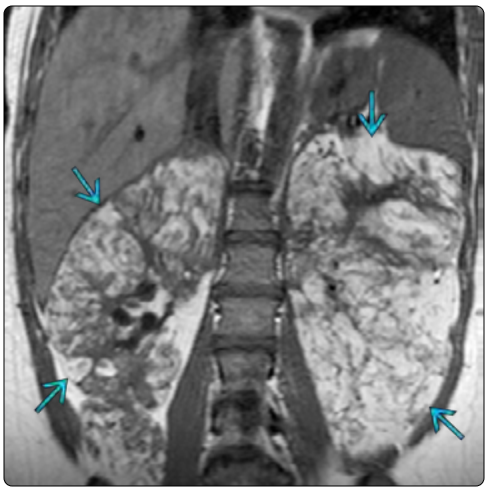


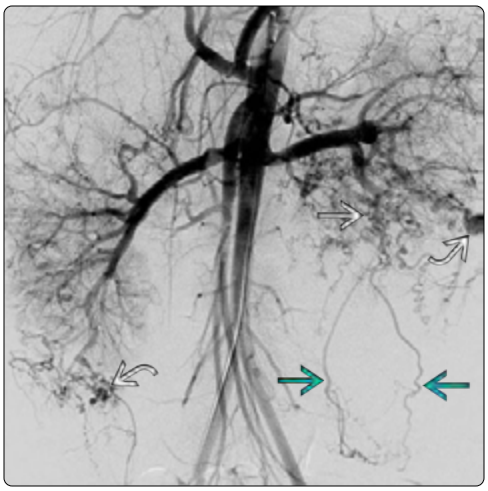
(Left) *Transverse US of the right kidney shows a slightly hyperechoic lipid-poor AML laterally. There is another smaller AML in the kidney as well as a small cortical cyst, typical of TSC.* **(Right)** *Axial CECT shows fat-containing AMLs in the liver. Hepatic AMLs are present in 13% of patients who have bilateral renal AMLs in the setting of TSC.*





(Left) *Axial T1 MR in a TSC patient shows diffuse fatty replacement of the kidneys with AMLs. An AML in the upper pole of the right kidney has hemorrhaged with layering blood seen in this lesion.* **(Right)** *Axial T2 MR in a TSC patient shows multiple lesions of both kidneys. Based on this sequence alone, the hyperintense lesions could represent cysts or lipid-rich AMLs while the larger, hypointense lesion could represent a fat-poor AML or hemorrhagic cyst.*





(Left) *Coronal T1 MR in a TSC patient shows marked enlargement & diffuse replacement of the bilateral renal parenchyma by fat-containing AMLs.* **(Right)** *Frontal image from a DSA with aortic injection of contrast in the same patient shows opacification of the renal arteries with multiple tortuous tumor vessels & aneurysms of both kidneys. Some of the tumor vessels are splayed by the fatty masses. This pattern is typical of multiple AMLs in the setting of TSC.*

Pyelonephritis

KEY FACTS

TERMINOLOGY

- Acute infection of renal parenchyma; often difficult to clinically distinguish from lower urinary tract infection (UTI)
- Classic imaging appearance: Focal swelling & ↓ perfusion of affected parenchyma visible on nuclear scintigraphy, US, CECT, MR, & IVP
  - Imaging work up of UTI controversial
  - See professional society guidelines

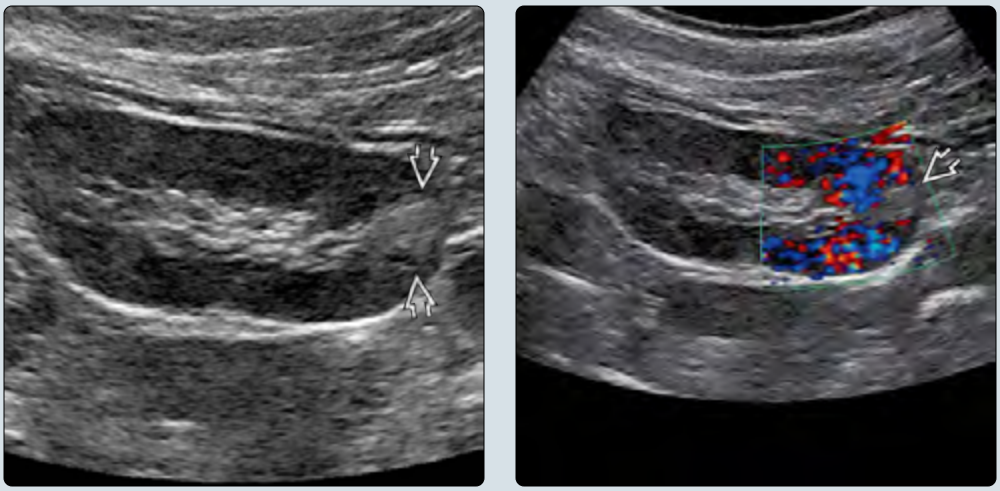
IMAGING

- Marked inflammatory response to infection causes swelling that alters normal tissue properties & effectively ↓ radiologic contrast agent delivery to site, which results in
  - Photopenic focus on nuclear cortical scan
  - ↓ perfusion on Doppler imaging with altered echotexture on grayscale US
  - Striated or wedge-shaped foci of ↓ enhancement on CECT/MR/IVP
- US with Doppler least invasive & readily available but less sensitive than nuclear renal cortical scans, CT, & MR
- US frequently performed to search for associated complications (abscess, stones, scarring), congenital anomalies, & hydronephrosis

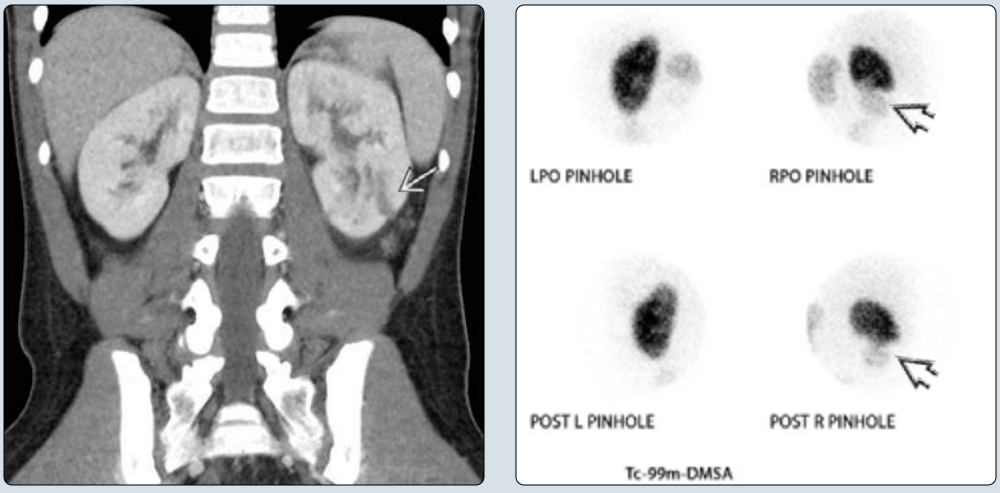
CLINICAL ISSUES

- Symptoms nonspecific: Malaise, irritability, fever, abdominal/flank pain, vomiting, hematuria, dysuria, change in urinary habits/enuresis
- Treatment: 7- to 14-day course of antimicrobial therapy; may be started IV & changed to oral
  - Obtain work-up for VUR & congenital anomalies
- Complications: Perirenal abscess, necrotizing papillitis, pyonephrosis (obstruction), & cortical scarring
- Permanent scarring more likely < 2 years old
- Recurrent infections & subsequent scarring lead to end-stage renal disease in small but significant percentage of pediatric patients

(Left) *Longitudinal US of the left kidney in a 4-year-old girl with fever & abdominal pain shows a focal area of increased echotexture in the lower pole* ⇨. **(Right)** *Color Doppler US of the lower pole shows decreased blood flow* ⇨ *in the echogenic area consistent with pyelonephritis. Ultrasound findings of pyelonephritis can be subtle; color (& especially power) Doppler is useful to localize an abnormality.*



(Left) *Coronal CECT image in a 13 year old scanned for possible appendicitis shows focal decreased enhancement* ⇨ *in the left lower pole with adjacent fat stranding, consistent with pyelonephritis.* **(Right)** *Posterior pinhole images from Tc-99m DMSA renal cortical scintigraphy show absent radiotracer in the lower pole of the right kidney* ⇨, *consistent with acute pyelonephritis. Large, wedge-shaped photopenic areas suggest pyelonephritis, while smaller, crescent-shaped cortical defects suggest scarring.*



TERMINOLOGY

Synonyms

- Acute lobar nephronia, focal bacterial nephritis

Definitions

- Acute infection of renal parenchyma
- Overview
  - Classic imaging appearance: Focal swelling & ↓ perfusion of affected parenchyma visible on nuclear scintigraphy, US, CT, & MR
  - Associated with vesicoureteral reflux (VUR) in ~ 1/3 of cases
  - Permanent scarring more likely in children < 2 years old
  - Patients can have variable presentation: Fever, lethargy, irritability, vomiting, abdominal/flank pain, hematuria, or dysuria
- Imaging work-up of urinary tract infection is controversial
  - See professional society guidelines, which vary internationally

IMAGING

General Features

- Best diagnostic clue
  - Marked inflammatory response to infection causes swelling that alters normal tissue properties & effectively ↓ radiologic contrast agent delivery to site, which results in
    - Photopenic area on nuclear cortical scans
    - ↓ perfusion on Doppler imaging & altered echotexture on grayscale US
    - Striated or wedge-shaped areas of ↓ enhancement on CT or IVP
    - Focal ↓ contrast enhancement on MR
- Morphology
  - Areas of infection tend to be peripheral & wedge-shaped; can be more rounded & mass-like appearance when inflammation more severe

Radiographic Findings

- IVP
  - Striated nephrogram classic, though IVPs rarely performed in pediatric patients

Ultrasonographic Findings

- Grayscale ultrasound
  - Localized or generalized swelling; unilateral renal enlargement may be only clue to pyelonephritis
  - Poor corticomedullary differentiation & focal areas of ↑ or ↓ echogenicity
  - Occasionally, rounded or mass-like areas of altered echotexture noted
- Color Doppler
  - ↓ perfusion noted in areas of pyelonephritis on color or power Doppler imaging
  - Adding power Doppler to US exam improves diagnostic accuracy & sensitivity
  - ↑ resistive indices with parenchymal edema (not specific)

CT Findings

- CECT
  - Wedge-shaped or round areas of poor enhancement
  - May have streaky enhancement
  - Inflammatory changes in perirenal fat
  - Occasionally mass-like; may distort normal renal contour & appear as partially cystic neoplasm during abscess development

MR Findings

- T1WI
  - Loss of corticomedullary differentiation
- T2WI
  - High signal intensity in affected areas of renal parenchyma
  - May also see inflammatory changes in perirenal fat
  - Fat suppression techniques may be more sensitive
- STIR
  - ↑ signal of renal parenchyma & surrounding tissues
  - With contrast administration, ↓ signal on STIR
    - More sensitive technique in correct clinical setting

Nuclear Medicine Findings

- Nuclear scintigraphic findings
  - ↓ accumulation of renal cortical agents, typically in wedge-shaped distribution that points toward renal hilum
    - Tc-99m DMSA or glucoheptonate
    - Pinhole collimation & SPECT imaging improve diagnostic sensitivity & accuracy
  - No volume loss until scarring ensues
  - Findings persist for up to 6 weeks after acute infection

Imaging Recommendations

- Best imaging tool
  - US with Doppler least invasive & readily available but less sensitive than nuclear renal cortical scans, CECT, & MR
  - US frequently performed to search for associated complications (abscess, stones, scarring), congenital anomalies, & hydronephrosis

DIFFERENTIAL DIAGNOSIS

Renal Infarction

- Wedge-shaped pattern of ↓ perfusion
- Retained thin rim of capsular enhancement
- May see Doppler abnormalities of vessels

Renal Scarring

- May be more superficial than pyelonephritis
- Associated cortical volume loss & dilated calyx

Renal Mass

- Heterogeneous well-circumscribed mass, typically round
  - Very uncommon but aggressive renal medullary carcinoma can be very poorly defined & infiltrative, mimicking pyelonephritis/abscess & surrounding inflammatory change; check for history of sickle trait
- Large masses (Wilms, mesoblastic nephroma, etc.) may only show claw of residual splayed renal parenchyma along margin of tumor

Renal Contusion/Laceration

- Focal ↓ enhancement (often jagged or linear) with surrounding fluid in setting of trauma

PATHOLOGY

General Features

- Etiology
  - Infection may occur via ascending route (VUR) or hematogenous spread; may be related to recent instrumentation
  - Vast majority of urine cultures grow gram-negative bacilli, typically normal inhabitants of intestinal tract (*Escherichia coli* most common)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often nonspecific: Malaise, irritability, fever, abdominal/flank pain, vomiting, hematuria, dysuria, change in urinary habits/enuresis
- Other signs/symptoms
  - Strong-smelling urine in any age
- Laboratory studies
  - Urine dipstick for nitrite, leukocyte esterase; both associated with higher likelihood of positive urine culture
  - Urine for Gram stain; *E. coli* causative in > 80% of 1st-time UTIs, *Klebsiella* is 2nd most common
    - Urine specimen for culture: Catheter specimen, clean-catch midstream, or suprapubic aspirate
    - Urine culture considered positive when single organism grows as follows
      - > 1,000 colony-forming units (cfu)/mL for suprapubic aspirate
      - Or > 10,000 cfu/mL for catheter specimen
      - Or > 100,000 cfu/mL for clean-catch midstream specimen
  - Bloodwork: Leukocytosis, occasionally positive blood cultures as well
- Complications
  - Perirenal abscess, necrotizing papillitis, pyonephrosis (obstruction), & cortical scarring
  - Recurrent infections & subsequent scarring lead to end-stage renal disease in small but significant percentage of pediatric patients
  - Recent study found that 1/2 of all patients with acute pyelonephritis went on to develop scarring
  - Some studies have found risk of scarring to be greater in younger patients

Demographics

- Gender
  - At least 2x as common in girls vs. boys
- Ethnicity
  - Less common in African Americans
- Epidemiology
  - Associated with VUR in ~ 25-40%
  - Higher incidence in obstruction, duplicated kidneys, other anomalies

Natural History & Prognosis

- Generally excellent, unless there are complications or recurrent infections
- Potential sequelae of renal scarring, chronic renal failure, hypertension, & pregnancy-related complications

Treatment

- 7- to 14-day course of antimicrobial therapy; may be started IV & changed to oral
- Imaging work-up for VUR & congenital anomalies
- Prophylactic antibiotics for VUR & other predisposing conditions controversial

DIAGNOSTIC CHECKLIST

Consider

- May be difficult clinically to distinguish lower UTI (cystitis) from pyelonephritis

Image Interpretation Pearls

- Partially cystic renal mass could be abscess, particularly if small to moderate in size with surrounding inflammatory changes
  - Pyelonephritis much more common than tumor
  - Abscess does not always cause positive urine testing
  - In correct clinical setting, consider short-term follow-up after antibiotics
- Conversely, very uncommon renal medullary carcinoma (RMC) often poorly defined & infiltrative, mimicking pyelonephritis/abscess
  - Check for history of sickle trait (associated with RMC) if clinical history of pyelonephritis not apparent

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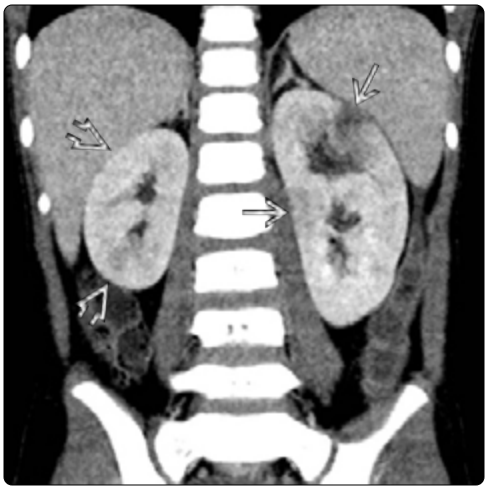
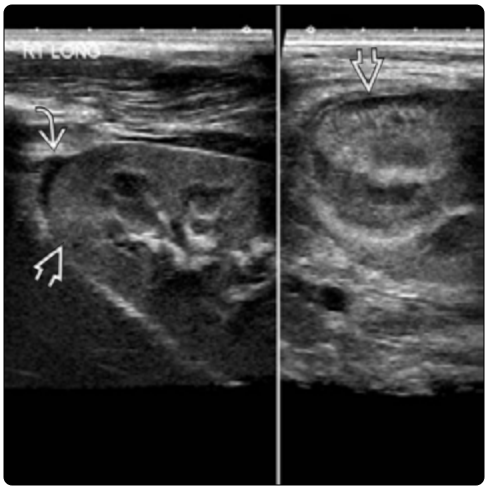
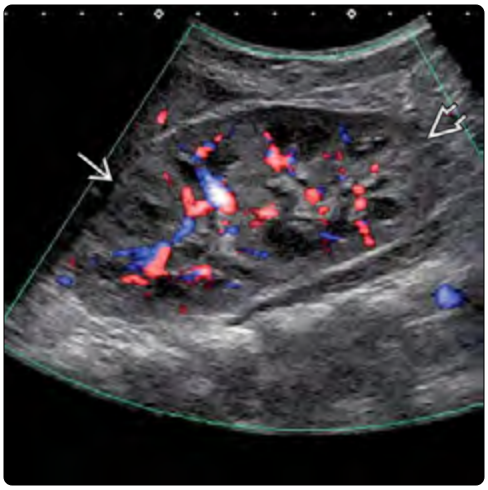
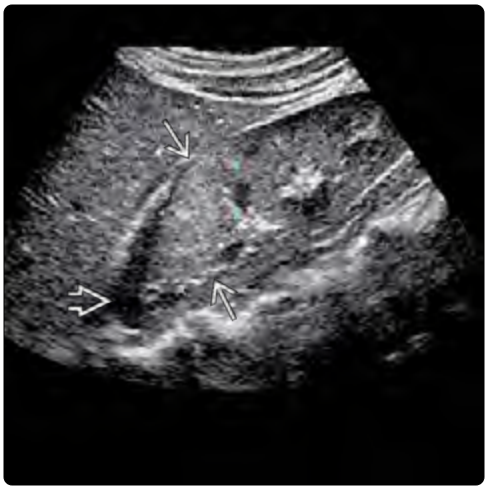


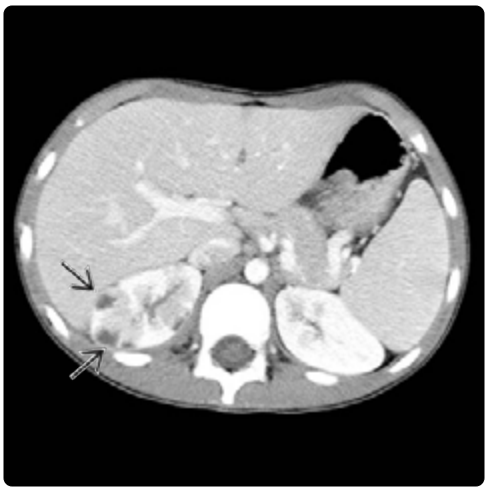


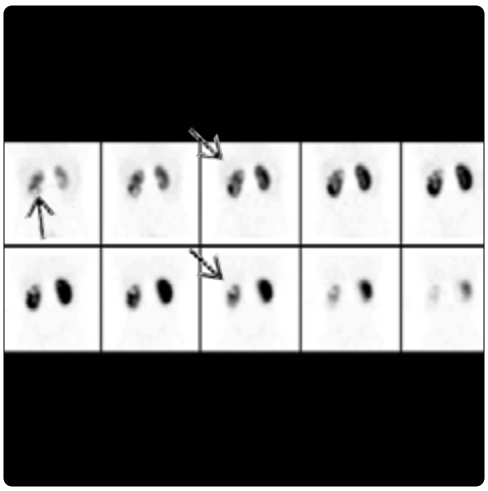
(Left) *Coronal CECT scan performed in a 6 year old with abdominal pain shows multiple foci of diminished cortical enhancement in the left ⇨ & right ⇨ kidneys, consistent with pyelonephritis.* **(Right)** *Longitudinal ultrasounds with a high-frequency linear transducer show bilateral areas of altered cortical echogenicity ⇨ plus a crescent of fluid ⇨ adjacent to the upper pole of the right kidney in a child with bilateral pyelonephritis.*





(Left) *Color Doppler shows poor blood flow to both the upper pole ⇨ & lower pole ⇨ of the kidney in a patient with pyelonephritis related to vesicoureteral reflux (VUR). Pyelonephritis & scarring often involve the renal poles in patients with VUR.* **(Right)** *Longitudinal US performed for refractory urinary tract infection shows an area of increased echoes in the right kidney upper pole ⇨ plus a thin crescent of perinephric fluid ⇨. These findings are consistent with pyelonephritis.*





(Left) *Axial CECT in a patient with immune compromise shows multiple fluid-density foci in the right kidney ⇨, suggesting developing abscesses. With subsequent increased size & confluence, drainage was ultimately required.* **(Right)** *Coronal anterior SPECT images from a Tc-99m DMSA cortical scan in the same patient show multiple defects in the right kidney ⇨, consistent with pyelonephritis & developing abscesses seen on the same patient's CECT scan. The left kidney was normal.*

KEY FACTS

IMAGING

- Protocol
  - CECT in late cortical or early nephrographic phase
  - Obtain delayed images to detect collecting system injury if parenchymal laceration or perinephric fluid present on initial images
- Imaging findings
  - Renal parenchymal contusion, laceration
  - Subcapsular or perinephric/perirenal hematoma
  - Collecting system/ureteropelvic junction (UPJ) laceration
    - Fluid attenuation [0-40 Hounsfield units (HU)]: Unclotted blood &/or urine
    - Intermediate attenuation (40-70 HU): Clotted blood &/or urine
    - High attenuation (> 150-200 HU) on delayed images only: Urinary contrast leak
  - Complete UPJ avulsion: Unopacified ipsilateral ureter distal to injury on delayed images
  - Active hemorrhage: High attenuation (isodense to vessel) nonanatomic collection on initial images; accumulates on delayed images
  - Vascular thrombosis or avulsion: Delayed or persistent renal enhancement vs. subtotal or global nonenhancement (± preservation of thin enhancing rim due to capsular arterial supply)

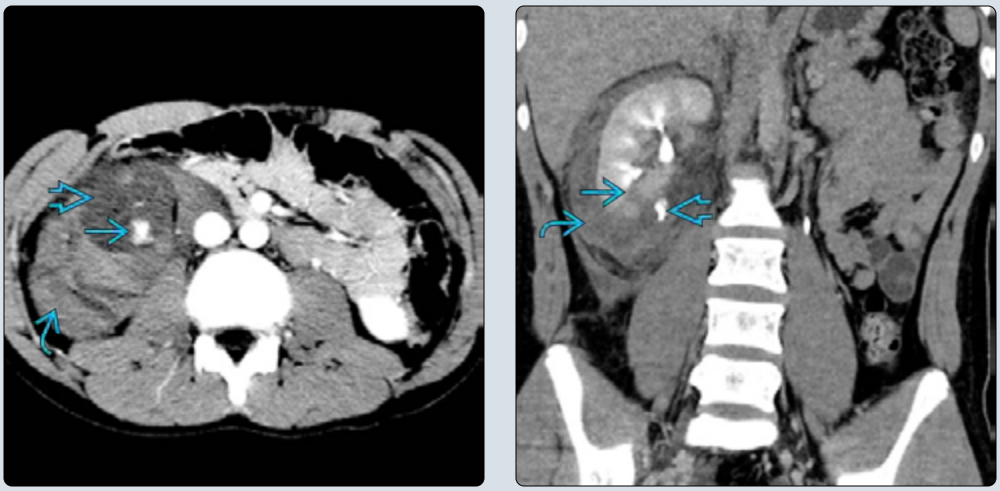
PATHOLOGY

- Etiology: Blunt trauma 90%, penetrating trauma 10%

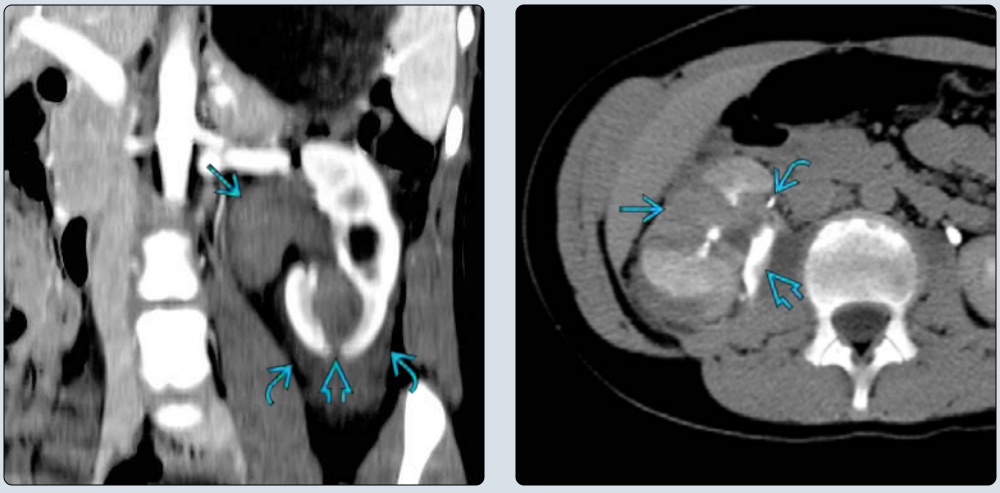
CLINICAL ISSUES

- Most common presentations: Flank pain, hematuria
- Successful nonoperative management in hemodynamically stable patients: 85%
  - Most common complication: Urinoma
- Possible predictors of intervention: Vascular contrast extravasation, collecting system clot, interpolar urinoma, urinoma > 4.3 cm, perirenal hematoma > 3.5 cm, dissociated renal fragments, > 25% devitalized fragments

(Left) *Axial CECT of a 15 year old after a football injury shows active arterial extravasation ⇨ along a devascularized right renal lower pole ⇨. There is a large perinephric hematoma ⇨.* **(Right)** *Delayed coronal CECT on a follow-up exam in a 14-year-old boy after a skiing accident shows a grade IV laceration ⇨ with urinary contrast extravasation ⇨ from the lower pole collecting system. The perinephric urinoma ⇨ had increased in size compared to the initial study, requiring a ureteral stent.*



(Left) *Coronal CECT in a 15 year old after a car accident shows a lower pole renal laceration ⇨ with perinephric blood &/or urine ⇨. A hematoma expands the collecting system, renal pelvis, & proximal ureter ⇨.* **(Right)** *Delayed axial CECT in a 15-year-old boy after a snowmobile accident shows a right renal grade IV laceration ⇨ that extends to the UPJ. Extravasation of contrast-opacified urine is noted ⇨. The ureter distal to the injury is opacified with contrast ⇨, indicating that the UPJ tear is incomplete.*



TERMINOLOGY

Definitions

- Trauma to parenchyma &/or collecting system of kidney

IMAGING

General Features

- Best diagnostic clue
  - Defect of renal parenchymal enhancement + perirenal hematoma on CECT
    - ± extravasation of contrast-opacified urine or blood

CT Findings

- CECT
  - Contusion
    - Region of ↓ parenchymal enhancement with ↑ attenuation on delayed images (persistent nephrogram)
    - May be radiologically occult
  - Hematoma
    - Fluid attenuation collection; ↑ density [40-70 Hounsfield unit (HU)] with clot formation
    - Subcapsular
      - Peripheral crescentic or lentiform collection
      - Flattening/indentation of underlying parenchyma
    - Perinephric/perirenal
      - Between renal parenchyma & Gerota fascia
      - Thickened lateral conal fascia
      - May displace (but not indent) kidney; may have mass effect on colon
  - Parenchymal laceration
    - Irregular, linear fluid attenuation defect (40-70 HU if contains clot)
  - Collecting system/ureteropelvic junction (UPJ) laceration
    - Perinephric collection of fluid attenuation or higher
      - Urine or urine mixed with hematoma
    - Collection attenuation & size ↑ on delayed images
    - Delayed images with UPJ/proximal ureter injury show
      - Opacification of ureter distal to injury: Partial tear
      - Unopacified ureter distal to injury: Complete tear/avulsion
  - Main renal artery thrombosis
    - Partial or complete lack of arterial enhancement
    - Subtotal/global renal nonenhancement: Infarction
      - Cortical rim sign: Rim of enhancing peripheral cortex perfused by collateral flow from renal capsular artery; may be absent in initial 8 hours
    - Retrograde opacification of renal vein from inferior vena cava
  - Segmental vascular infarct
    - Triangular nonenhancing parenchymal focus
    - Injury due to accessory, capsular, or intrarenal segmental arterial branches
  - Pseudoaneurysm
    - Nonanatomic focus of ↑ density (isodense to aorta) adjacent to artery
    - Stable size with contrast washout on delayed images
  - Active hemorrhage
    - Irregular high density collection (isodense to adjacent vessel) on initial images
    - Persists, ↑ in size on delayed images
  - Main renal vein thrombosis
    - Enlarged vein with filling defect of thrombus
    - Nephromegaly with delayed nephrogram & excretion
    - Less common than main renal artery thrombosis

Ultrasonographic Findings

- Grayscale ultrasound
  - Hematoma
    - Parenchymal, subcapsular, or perinephric collection
    - Echogenicity varies with age of blood products
    - Can be difficult to differentiate from urinoma
  - Laceration
    - Hypoechoic linear/branching defect interrupting renal architecture; hyperechoic if contains clot
- Color Doppler
  - Thrombosis: Various aberrant waveforms possible depending on extent & location of thrombus
    - Tardus-parvus waveform distal to proximal nonocclusive arterial thrombus/stenosis
    - Reversal of diastolic flow with thrombosis of renal vein or distal artery
    - Absence of flow in occluded vessel
  - Parenchymal ischemia/infarct: Focally or globally ↓ or absent color Doppler signal
- CEUS
  - Improved detection of segmental & subcapsular infarcts
  - Improved confidence for diagnosing renal artery & vein occlusion, & differentiating complete vs. partial occlusion
  - Allows visualization of hyperechoic active extravasation
- Sensitivity & specificity vary widely across multiple studies
  - FAST US: Sensitivity 23-100%, specificity 98-100%
  - Abdominal US: Sensitivity 36-90%, specificity 98-100%
  - CEUS: Sensitivity 69%, specificity 99%

Nuclear Medicine Findings

- Dimercaptosuccinic acid (DMSA) renal scintigraphy
  - Assesses relative renal function after injury
  - Focal to global absence of DMSA uptake

Imaging Recommendations

- Best imaging tool
  - American Urological Association recommendations
    - CECT for initial evaluation
      - Portal venous phase (late cortical or early nephrographic phase)
      - Delayed (5-15 minute) images to evaluate for urinary contrast leak in stable patient with renal laceration &/or perinephric fluid on initial images
    - Follow-up CT in 48 hours for
      - Grade IV or V injuries (due to ↑ complication rates)
      - Injury grade < IV with clinical signs of complication
  - European Society of Uroradiology recommendation
    - US with color Doppler for minor or moderate trauma with low pretest probability of injury

DIFFERENTIAL DIAGNOSIS

Normal Variant Contour

- Dromedary hump, column of Bertin, fetal lobation

Pyelonephritis

- Enlarged kidney with delayed or striated nephrogram in patient with flank pain, fever, abnormal urinalysis
- Abscess can mimic multicystic mass with irregular rim/septal enhancement

Renal Neoplasm

- Most pediatric renal tumors
  - Present as abdominal distention, palpable mass, &/or hematuria
  - Have overlapping imaging features
    - Differential best determined by patient age & clinical history
  - Can rupture or hemorrhage (causing pain)
- Angiomyolipoma (AML) unique
  - Fat-containing renal masses typically associated with tuberous sclerosis
  - AML > 4 cm, or aneurysm > 5 cm, prone to spontaneous hemorrhage (uncommon in children)

PATHOLOGY

General Features

- Etiology
  - Blunt trauma in 90%, penetrating trauma in 10%
  - Motor vehicle-related 47%, falls 19%, sports-related 13%, abuse & assault 13%, bicycle accidents 8%
  - Seen in up to 19% of abdominal trauma cases due to child abuse
  - Rapid deceleration: ↑ risk of vascular pedicle & UPJ injury
  - Children at ↑ risk compared to adults due to ↑ organ mobility & relative size, ↓ perirenal fat, low renal position, elastic rib cage with ↓ renal coverage, fetal lobations (which may act as cleavage plane)
  - Preexisting renal abnormalities predispose to injury
- Associated abnormalities
  - Concomitant injuries: Hepatic 22%, pulmonary 19%, splenic 16%

Gross Pathologic & Surgical Features

- American Association for the Surgery of Trauma Scale
  - Grade I: Contusion with microscopic/gross hematuria & normal urologic studies; nonexpanding subcapsular hematoma without laceration
  - Grade II: Nonexpanding perirenal hematoma confined to retroperitoneum; laceration < 1-cm depth without urine extravasation
  - Grade III: Laceration > 1-cm depth without collecting system rupture/urine extravasation
  - Grade IV: Laceration through cortex, medulla, & collecting system; main renal artery or vein injury with contained hemorrhage
  - Grade V: Completely shattered kidney; renal hilum avulsion that devascularizes kidney
  - Advance 1 grade for bilateral injuries up to grade III

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Flank pain/tenderness, hematuria, ecchymosis
    - Hematuria may be absent, even in high-grade injury

Natural History & Prognosis

- Majority (79%) of injuries low-grade (I-III)
- Of all renal injuries, 85% managed conservatively
- 50% of grade V injuries & 44% of penetrating injuries managed surgically
- Increased likelihood of intervention associated with
  - Vascular contrast extravasation (particularly medial), collecting system clot, interpolar contrast urinoma, urinoma > 4.3 cm, dissociated renal fragments, > 25% devitalized fragments, perirenal hematoma > 3.5 cm
- Early complications (< 4 weeks)
  - Urinoma: Most common; most reabsorb
  - Delayed bleeding: Due to arteriovenous fistula/pseudoaneurysm, usually 2-3 weeks after injury
  - Perinephric abscess, sepsis, infected urinoma
- Late complications (> 4 weeks)
  - Page kidney (hypertension due to renal parenchymal compression by subcapsular hematoma, which ↓ renal perfusion & activates renin-angiotensin system)
  - Hydronephrosis, calculi, chronic pyelonephritis

Treatment

- Nonoperative in hemodynamically stable patients
- Ureteral stent ± percutaneous urinoma drain &/or percutaneous nephrostomy for complications of ↑ urinoma, ↑ pain, ileus, fistula, infection
- Endoscopic or open surgical intervention for renal pelvis/proximal ureteral avulsion
- Renal artery thrombosis: Thrombolysis, stent
- Angioembolization in hemodynamically unstable patients with uncontrolled bleeding
- Surgery (often nephrectomy) in hemodynamically unstable patients or those failing nonoperative management

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- View CECT in multiple planes (as transverse lacerations may be occult on axial images)

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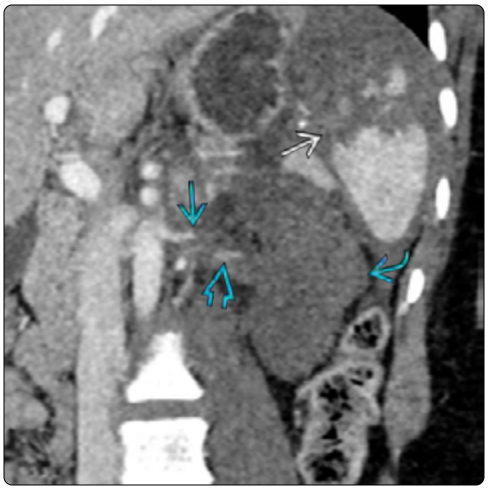
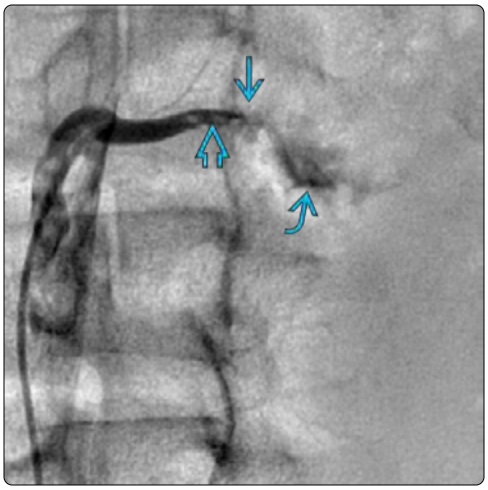
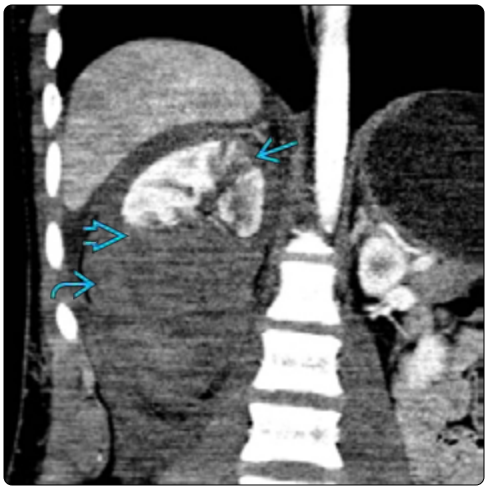
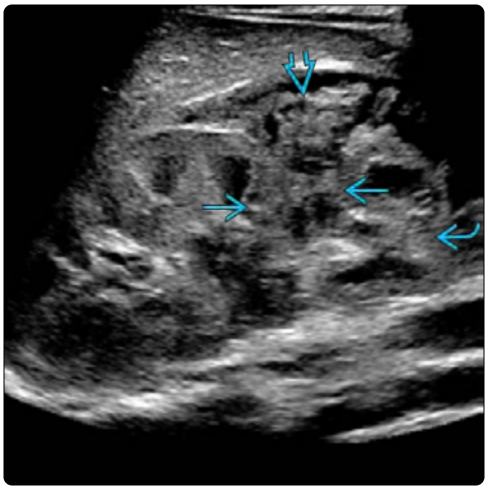


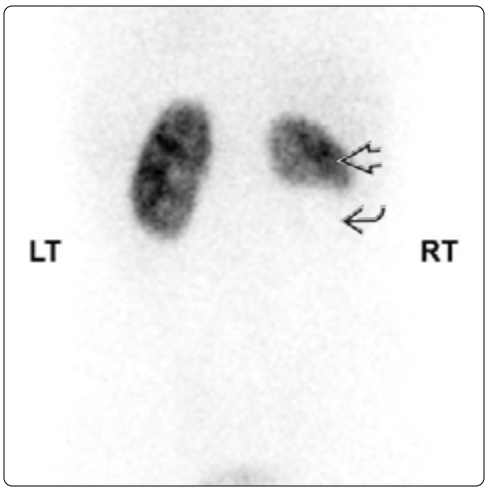


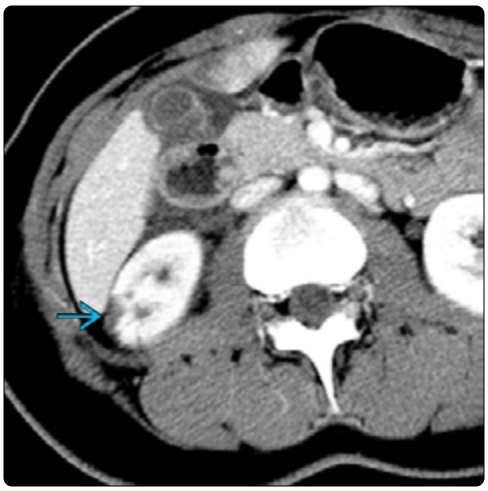
(Left) *Coronal oblique CECT from a 13-year-old girl with a grade V renal injury shows cut-off of the left renal arterial opacification with adjacent contrast extravasation. The left kidney is devascularized. A grade IV splenic injury is also noted.* **(Right)** *Frontal view from catheter angiography of the left renal artery in the same patient shows complete left renal artery avulsion with proximal vessel thrombosis & distal contrast extravasation. A revascularization attempt failed.*





(Left) *Coronal CECT from a 15-year-old boy after a sledding accident shows that the right kidney is shattered (grade V) with no enhancement of lower pole fragments below the fracture plane. There is a large perirenal hematoma as well as a laceration of the upper pole.* **(Right)** *Longitudinal ultrasound in the same patient shows a midkidney fracture with heterogeneous perinephric hematoma. A portion of the fragmented lower pole is seen but lacked color Doppler signal (not shown).*





(Left) *Posterior view from a 2-month follow-up Tc-99m DMSA scan in the same patient shows the right kidney has normal uptake in the upper pole but none in the lower pole. The split renal function was 60% left & 40% right.* **(Right)** *Axial CECT of a 15 year old who sustained a grade IV liver laceration during a motor vehicle collision shows a small hypodense renal contusion consistent with a grade I injury.*

Renal Stones

KEY FACTS

TERMINOLOGY

- Concretion in urinary system

IMAGING

- Most stones seen in kidneys & upper urinary tract
- Range in size from 1-2 mm to > 1 cm
- NECT most sensitive modality to detect stones
  - Calcified density in urinary system
- Ultrasound shows hyperechoic urinary tract focus with posterior acoustic shadowing &/or twinkle artifact
- Important to report signs of obstruction
  - Hydroureteronephrosis
  - Nephromegaly
  - Perinephric/periureteral edema
  - Lack of ureteral jet in urinary bladder on Doppler ultrasound
  - High-resistance renal artery flow on Doppler ultrasound
  - Delayed renal enhancement & excretion after IV contrast administration

TOP DIFFERENTIAL DIAGNOSES

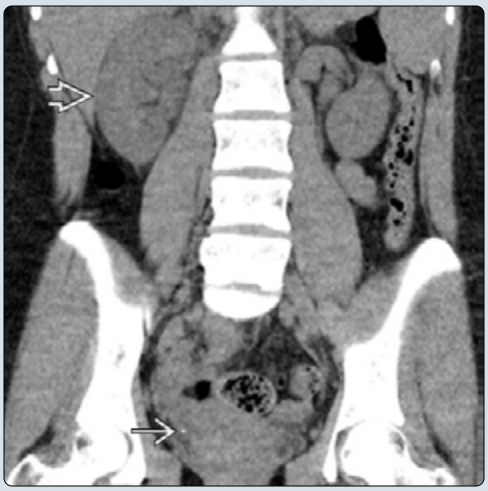
- Fecal material
- Phleboliths
- Nephrocalcinosis
- Ureteropelvic junction obstruction

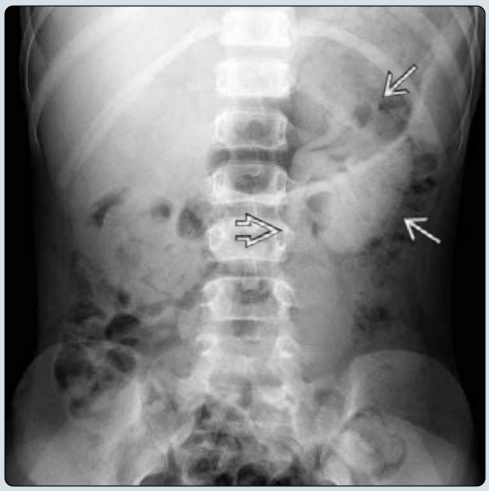
PATHOLOGY

- 75% of children have identifiable metabolic predisposition

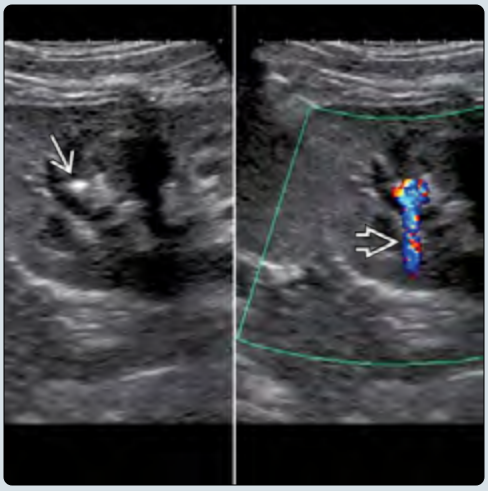
CLINICAL ISSUES

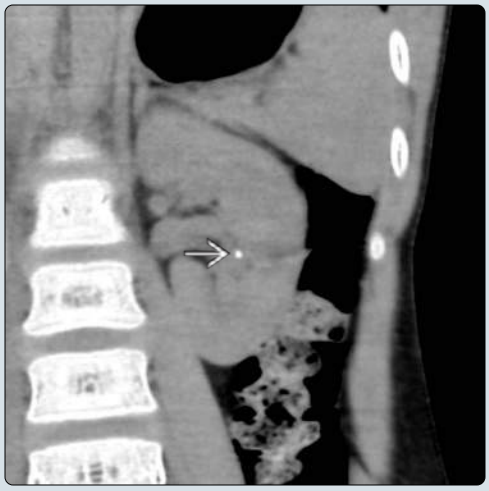
- Presentations
  - 94% of adolescents have colicky flank pain
  - Nonspecific symptoms (abdominal pain, nausea, vomiting, & irritability) in younger children
  - Microscopic hematuria in up to 90% of patients
- Treatment depends on stone size, location, presence of obstruction, & underlying etiology





(Left) *Coronal NECT in a young adult shows a 1- to 2-mm calcific density in the distal right ureter near the ureterovesical junction. The right kidney appears mildly swollen with minimal perinephric stranding.* **(Right)** *IVP in an adolescent shows a delayed nephrogram of the left kidney. Contrast is being excreted & extends only a few centimeters in the proximal left ureter before it abruptly stops. Contrast did not pass beyond this point on more delayed images.*





(Left) *Transverse ultrasound images of the left kidney in an adolescent show an echogenic focus within the renal collecting system. With color Doppler, there is considerable twinkle artifact posterior to the echogenic focus, typical of a calculus.* **(Right)** *Coronal NECT in the same patient shows a small nonobstructing calcified stone in the lower pole of the left kidney. Approximately 75% of pediatric patients with renal stones have an identifiable predisposition to stone formation.*

TERMINOLOGY

Synonyms

- Nephrolithiasis, nephrolith, urolithiasis, urolith, urinary stone, kidney stone, renal calculi

Definitions

- Concretion in urinary system

IMAGING

General Features

- Best diagnostic clue
  - Calcific density in renal collecting system, ureters, or urinary bladder
- Location
  - Usually seen at renal collecting system, ureteropelvic junction (UPJ), or ureterovesical junction (UVJ)
    - Most stones seen in kidneys & upper urinary tract
- Size
  - Range in size from 1-2 mm to > 1 cm

Radiographic Findings

- Radiography
  - Irregular calcified density over region of kidneys, ureters, or bladder
  - If stone seen on radiograph, can be used to follow disease
- IVP
  - Used to identify obstructive calculi in known stone formers
  - Often modified for children to ↓ radiation dose
  - Signs of urinary obstruction
    - Enlarged kidney
    - Hydronephrosis
    - Hydroureter up to stone
    - Filling defect at level of calculus
    - Delayed, prolonged nephrogram with delayed collecting system excretion

CT Findings

- NECT
  - Most sensitive modality to detect urinary stones
  - Stone appears as calcified density in renal collecting system, ureters, or bladder
  - Stones can obstruct urinary system
    - Most stones not obstructive
    - Signs of obstruction
      - Hydronephrosis
      - Hydroureter
      - Perinephric or periureteral stranding
      - Enlarged kidney
      - Loss of slightly hyperattenuating pyramids

Ultrasonographic Findings

- Grayscale ultrasound
  - Hyperechoic focus with posterior acoustic shadowing
    - Shadow artifact may not be present on newer ultrasound machines
  - Stones usually visible in renal collecting system (at UPJ, UVJ, or bladder)
    - Midurethral stones difficult to find due to bowel gas
  - Ultrasound can be used to identify obstruction of urinary system
    - Signs of obstruction
      - Hydronephrosis
      - Hydroureter to level of urinary stone
      - Enlarged kidney
- Color Doppler
  - Twinkle artifact: Rapidly changing random mixture of color in/posterior to stone
    - Thought to be due to rough but strongly reflective surface of stone
  - Signs of obstruction
    - High-resistance renal artery flow
    - Lack of ureteral jet in urinary bladder

Imaging Recommendations

- Best imaging tool
  - NECT most sensitive modality to detect stones
    - Often 1st study in 1st-time stone disease
    - Many perform CT in prone position to better distinguish UVJ vs. bladder stone
    - Beware multiple CTs due to radiation dose
  - In known stone formers, presence or absence of obstruction may be only imaging question
    - US detects urinary obstruction without radiation

DIFFERENTIAL DIAGNOSIS

Fecal Material

- Colonic contents may overlie kidneys, ureters, or bladder on radiographs & mimic nephrolithiasis

Phleboliths

- Round Ca^{2+} with lucent center in abnormal stagnant vein
- Most common in pelvis, near bladder

Nephrocalcinosis

- Ca^{2+} within renal parenchyma
- ± cortical, medullary, or diffuse

Ureteropelvic Junction Obstruction

- Chronically dilated renal collecting system due to intrinsic or extrinsic process; no visible stone

Transient Neonatal Renal Medullary Hyperechogenicity

- Evenly distributed echogenic renal pyramid tips
- Incidental, benign finding lasting up to 3 weeks
- Not clearly due to Tamm-Horsfall proteinuria

PATHOLOGY

General Features

- Etiology
  - 75% of children with stones have identifiable predisposition
    - Metabolic cause of stones in 40-50% of patients
    - Structural urinary tract anomalies in 30% of patients
    - Infection in 4% of patients
- Metabolic causes of pediatric stone disease
  - Calcium stones
    - Hypercalciuria: Most common metabolic abnormality causing pediatric stones

        - Accounts for 34-50% of children with identifiable metabolic cause of stones
      - Hyperuricosuria: Can be caused by excess purine production or ingestion, renal tubular disorders, medications, or juvenile gout
        - Present in 2-20% of children with stones
      - Hypocitruria: Can be caused by renal tubular acidosis
        - 10% of children with stones
      - Hyperoxaluria: Can be caused by primary hyperoxaluria or ↑ intestinal absorption due to bowel disease
        - Found in 10-20% of children with stones
    - Uric acid stones: Seen with excessively acidic urine such as in diarrheal states or diet high in animal protein
      - Radiolucent stones
    - Struvite stones: Caused by infection with urease-splitting bacteria
      - Often appear as staghorn calculi
- Hereditary causes of pediatric stone disease
  - Adenine phosphoribosyltransferase deficiency
    - Autosomal recessive inborn error of adenine metabolism
    - Most common manifestation: Radiolucent stones
    - Pathognomonic round/brown DHA crystals in urine
  - Cystinuria
    - Most common cause of inherited kidney stones
    - Accounts for up to 25% of pediatric stones
    - Defect in proximal tubular resorption of filtered cystine
    - Average age of 1st stone 12-13 years
    - Pathognomonic hexagonal crystals in urine
  - Dent disease
    - Rare X-linked renal tubular disorder
    - Characterized by low molecular weight proteinuria
    - Variable hypercalciuria, nephrocalcinosis, & stones
    - Progresses to chronic kidney disease in 30-80% of affected patients
  - Familial hypomagnesemia with hypercalciuria & nephrocalcinosis
    - Autosomal recessive disorder characterized by renal magnesium wasting, hypercalciuria, nephrocalcinosis, & renal failure
    - Present in childhood with recurrent urinary tract infections, polyuria, polydipsia, hematuria, & pyuria
    - End-stage renal disease by early adulthood
  - Primary hyperoxaluria
    - Metabolic disorder where hepatic enzyme deficiencies lead to overproduction of oxalate
    - Present with nephrolithiasis in 1st decade
    - ± treated with liver/kidney transplant

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depends on age
    - 94% of adolescents present with colicky flank pain
    - Nonspecific symptoms (abdominal pain, nausea, vomiting, & irritability) in younger children
  - Microscopic hematuria in up to 90% of patients
- Other signs/symptoms
  - Gross hematuria in 14-32%
  - Concomitant urinary tract infection in 8-20%

Demographics

- Age
  - Can occur at any age
- Gender
  - 1.4-2.1x more common in male patients
- Ethnicity
  - More common in Caucasians
  - Rare in African Americans & Asians
- Epidemiology
  - Prevalence of stones in kids increasing
    - 18 per 100,000 patients in 1999; 57 per 100,000 in 2008
  - Potential causes for increasing prevalence
    - Increasing obesity & related poor dietary habits
    - ↓ water intake coupled with ↑ sodium intake
    - ↓ dietary calcium (from milk)
    - ↑ use of CT (improved detection)

Natural History & Prognosis

- Higher morbidity of stone disease in children
  - Longer hospital stay
- Stones < 5 mm may pass spontaneously
- Residual stones can act as nidus for more stones

Treatment

- Treatment depends on underlying etiology
- ↑ fluid intake
  - Poor long-term compliance
- Expectant management
  - Up to 60% of stones < 5 mm pass spontaneously
- Medical treatment depends on underlying cause
  - Hypercalciuria: ↑ fluid intake, ↓ dietary sodium, & ↓ dietary calcium to recommended levels
    - Thiazide diuretics can ↓ urinary calcium
  - Hyperoxaluria: Potassium citrate & calcium supplements may ↓ intestinal oxalate absorption
  - Hyperuricosuria: Urinary alkalinization
- Surgical management required in 22%
  - Extracorporeal shock wave lithotripsy (ESWL)
    - Used for treatment of proximal ureteral calculi, lower pole stones < 1 cm, & midupper pole stones < 2 cm
  - Ureteroscopy
    - Improved stone-free rates (88-100%) compared to ESWL for ureteral stones
    - Similar stone-free rates compared to ESWL for intrarenal stones (58-62%)
  - Percutaneous nephrolithotomy
    - Reserved for larger stone burden/treatment failure

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3. Edvardsson VO et al: Hereditary causes of kidney stones and chronic kidney disease. Pediatr Nephrol. 28(10):1923-42, 2013
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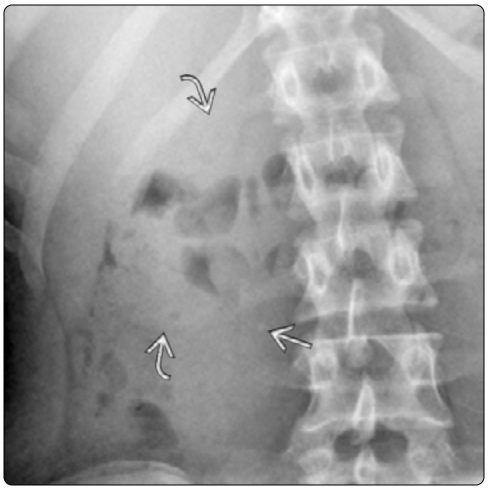
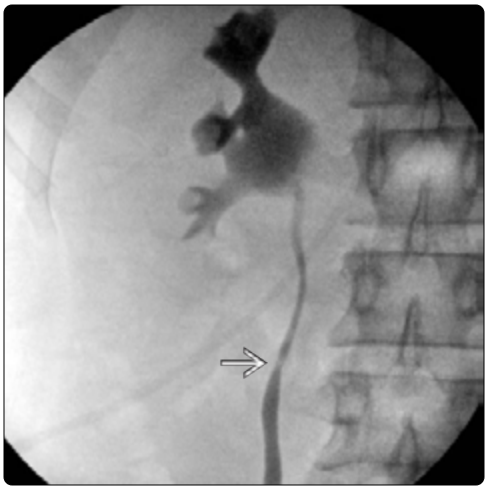
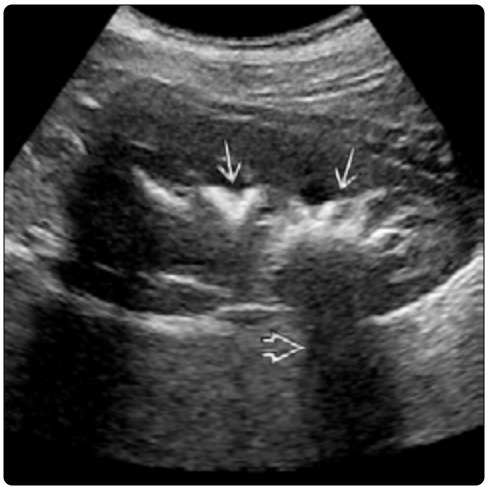
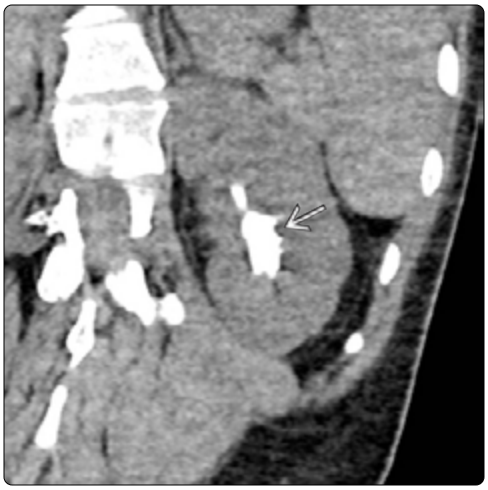


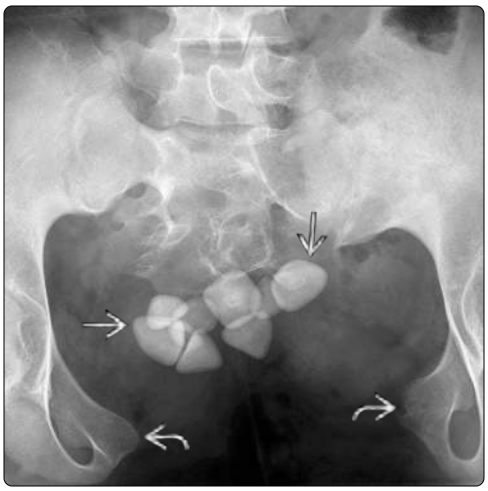


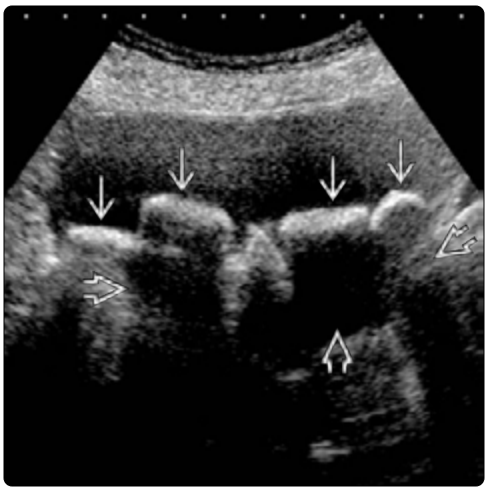
(Left) *AP radiograph of the abdomen in an adolescent shows a small calcified stone ➡ inferior & medial to the right renal shadow ➡, likely in the proximal ureter.* **(Right)** *Retrograde pyelogram in the same patient shows a small filling defect ➡ in the midportion of the right ureter at the level of the nonobstructing stone.*





(Left) *Longitudinal ultrasound of the left kidney in a young adult shows the renal collecting system filled with echogenic material ➡, consistent with a large staghorn calculus. Note the large amount of posterior acoustic shadowing ➡.* **(Right)** *Coronal NECT in the same patient shows the large staghorn calculus of the left kidney ➡. Patients with staghorn calculi may require surgical removal of such large stones.*





(Left) *AP radiograph of the pelvis shows multiple large stones ➡ within the urinary bladder. In addition, there is diastasis of the pubic symphysis ➡ in this patient with a history of bladder exstrophy.* **(Right)** *Transverse ultrasound in the same patient shows multiple echogenic stones ➡ in the urinary bladder. Note the associated posterior acoustic shadowing ➡. In patients with chronic medical issues (such as cerebral palsy & neurogenic bladder), urinary stones can be caused by urine stasis.*

Transient Neonatal Renal Medullary Hyperechogenicity

KEY FACTS

TERMINOLOGY

- Temporary sonographic appearance of hyperechoic renal medullary pyramids in some normal neonates
  - Neonatal renal pyramids usually hypoechoic to renal cortex on US
- Etiology unclear: Not definitively linked to ↑ concentration of normal Tamm-Horsfall glycoproteins in neonatal urine
  - Though appearance previously has been termed Tamm Horsfall proteinuria

IMAGING

- Renal US shows ↑ echogenicity of medullary pyramids in neonate with normal renal function
  - Usually involves tips & central portions of pyramids
  - Bases of pyramids typically remain hypoechoic
  - Diffuse involvement of entire medullary pyramid uncommon
  - May appear as layering gradient of ↑ echogenicity, brightest at tips of pyramids
  - Posterior acoustic shadowing typically absent
  - Bilateral > unilateral renal involvement
  - Segmental involvement (sparing of some medullary pyramids) > diffuse (involvement of all pyramids)
  - Normal renal cortical echogenicity & size
- ↑ echogenicity of renal medullary pyramids usually resolves spontaneously by 10 days

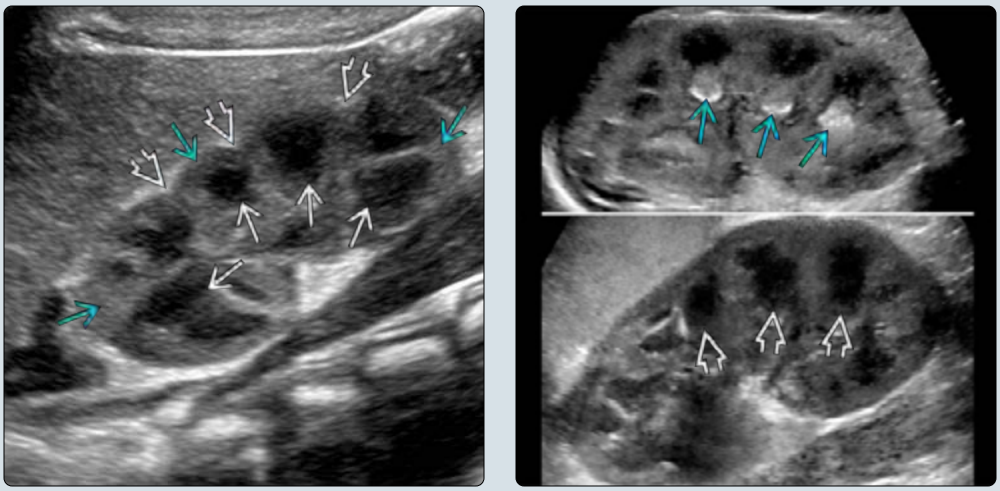
CLINICAL ISSUES

- If renal insufficiency/failure present, consider pathologic causes for ↑ echogenicity

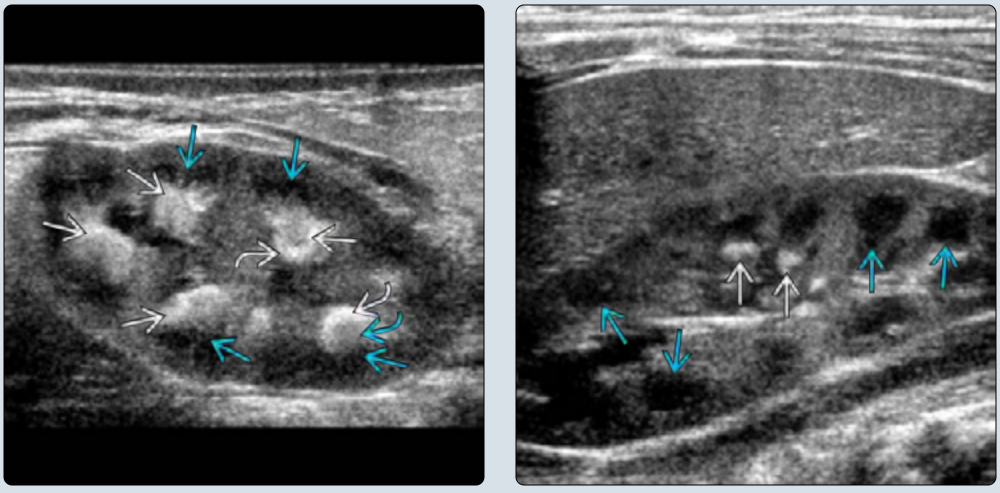
DIAGNOSTIC CHECKLIST

- Normal finding up to 10th day of life; repeat US not indicated
- If hyperechoic medullary pyramids seen beyond 10-14 days or in setting of renal dysfunction, then another diagnosis should be considered

(Left) *Longitudinal ultrasound shows a normal right kidney in a 1-day-old girl. In the neonatal period, the medullary pyramids are normally hypoechoic ➡ compared to the renal cortex ⇨. Also note the normal fetal lobations ➡.* **(Right)** *Longitudinal ultrasound of the left kidney in a neonate with normal renal function on the 1st day of life (top) show that tips & central portions of multiple renal pyramids are hyperechoic ⇨. One month later (bottom), the pyramids ➡ have a normal hypoechoic appearance.*



(Left) *Longitudinal renal ultrasound in a newborn shows ↑ echogenicity ➡ in all renal pyramids. Note the extensive involvement of individual pyramids with a gradient of ↑ echogenicity that is most pronounced at the tips ➡ & less so in the central portions ⇨. Only the bases of the pyramids have a normal hypoechoic appearance ⇨.* **(Right)** *Longitudinal renal ultrasound in a newborn shows discrete foci of ↑ echogenicity at the tips of 2 medullary pyramids ➡. Other pyramids ⇨ have a normal hypoechoic appearance.*



TERMINOLOGY

Synonyms

- Tamm Horsfall proteinuria (THP)

Definitions

- Temporary sonographic appearance of hyperechoic renal medullary pyramids in some normal neonates
- Etiology unclear (not definitively linked to Tamm-Horsfall proteins)

IMAGING

General Features

- Best diagnostic clue
  - ↑ echogenicity of medullary pyramids in neonate with normal renal function
    - Neonatal renal medullary pyramids usually hypoechoic relative to cortex
- Location
  - Bilateral > unilateral
  - Segmental involvement (sparing of some medullary pyramids) > diffuse (involvement of all pyramids)
- Morphology
  - Usually involves tips & central portions of pyramids
  - Typically spares bases of pyramids

Ultrasonographic Findings

- ↑ echogenicity of medullary pyramids
  - Usually involves tips & central portions of pyramids
  - Bases of pyramids typically remain hypoechoic
  - Diffuse involvement of entire medullary pyramid uncommon
  - Discrete hyperechoic foci may be seen with denser precipitates
  - May demonstrate layering gradient of ↑ echogenicity, brightest at tip of pyramid
  - Posterior acoustic shadowing usually absent
- Renal cortical echogenicity & size normal
- Associated bladder debris may represent excreted protein precipitates
- ↑ echogenicity of pyramids usually resolves by 10 days
  - Rarely persists a few days longer

Imaging Recommendations

- Best imaging tool
  - US
    - Commonly found incidentally on US performed for other reasons
    - Generally considered normal up to 10 days of life; repeat US not indicated
    - Another diagnosis should be considered if
      - Hyperechoic pyramids seen beyond 10-14 days of life
      - Renal dysfunction present

DIFFERENTIAL DIAGNOSIS

Medullary Nephrocalcinosis

- Loop diuretics (e.g., furosemide)
- Distal renal tubular acidosis
- Williams syndrome: Supravalvular aortic stenosis & hypercalcemia
- Bartter syndrome: Ion transport channel mutation → ↓ Ca^{2+} resorption & hypercalciuria
- Neonatal primary hyperparathyroidism

Hypernatremic Dehydration

- Seen in some solely breastfed neonates due to poor milk production or inadequate intake
- More common in 2nd week of life

Perinatal Ischemia

- More common in premature infants

Neonatal Renal Vein Thrombus

- Pyramids can be echogenic in acute phase, ± linear echogenic striations of kidney
- Associated renal enlargement & Doppler abnormalities

PATHOLOGY

General Features

- Etiology
  - Classic theory: Reversible precipitation of THP in renal tubules → transient ↑ echogenicity of medullary pyramids seen by US in some neonates
  - Not definitively proven: Some suggest urate crystal deposition may account for transient ↑ echogenicity
  - Resolution by around 10th day of life may be due to physiologic ↑ in glomerular filtration

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic
  - Renal insufficiency/failure warrants search for alternative cause of ↑ medullary echogenicity

Demographics

- Age
  - Early neonatal period
  - Full-term infants > premature infants
    - Possibly due to immaturity of renal tubules in premature neonates
- Epidemiology
  - 3.9-58% of term neonates

Natural History & Prognosis

- Should resolve by 10th day of life
  - Rarely seen a few days beyond this

Treatment

- None

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Renal Vein Thrombosis

KEY FACTS

TERMINOLOGY

- Obstruction of renal vein(s) by thrombus

IMAGING

- Ultrasound with Doppler
  - Enlarged, echogenic kidney with ↓ corticomedullary differentiation
  - ± visualization of main renal vein thrombus
  - High-resistance renal arterial waveforms (RI > 0.9)
    - May see complete diastolic flow reversal
- CECT/MR
  - Heterogeneous or delayed renal enhancement
  - ± filling defect in renal vein, ± inferior vena cava extension
- Chronic appearance: Renal atrophy ± Ca^2
- Associated adrenal hemorrhage or Ca^2 may be present

TOP DIFFERENTIAL DIAGNOSES

- Acute tubular necrosis
- Pyelonephritis
- Transient neonatal renal medullary hyperechogenicity
- Tumor thrombus

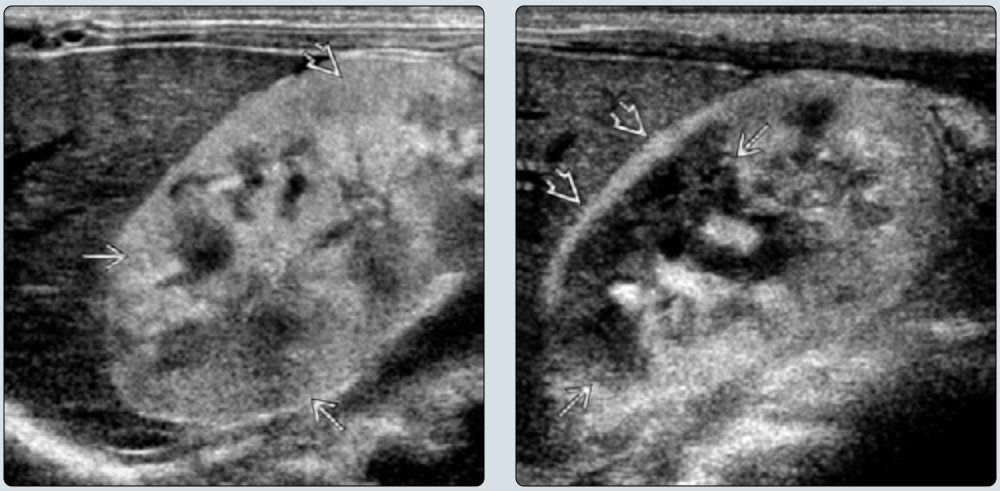
PATHOLOGY

- In neonates, thrombosis begins in small intrarenal veins → proximal extension to main renal vein
  - ↑ renal venous pressure & ↓ arterial flow
- Etiologies: Dehydration, sepsis/infection, iatrogenic

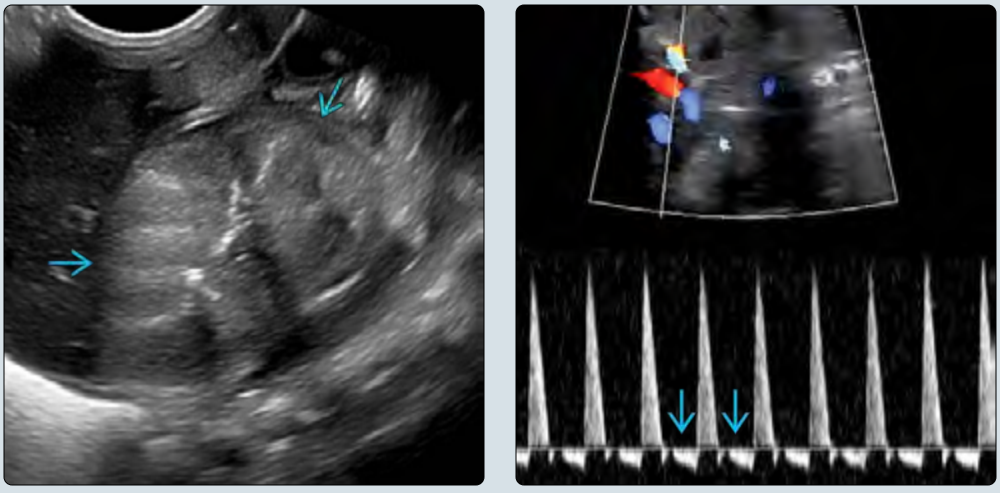
CLINICAL ISSUES

- Most commonly diagnosed in neonatal period
  - Renal dysfunction, oliguria, hypertension
  - Classic triad of palpable flank mass, hematuria, & thrombocytopenia usually absent
- Irreversible damage occurs in 70%
  - Up to 20% will have persistent hypertension
- Treatment controversial & may depend upon comorbidities
  - Supportive, ± anticoagulation, rarely thrombolysis

(Left) *Longitudinal oblique US in a newborn shows an enlarged, hyperechoic right kidney with poorly defined hypoechoic medullary pyramids in the upper pole. Corticomedullary differentiation is lost in the lower pole.* **(Right)** *Longitudinal US obtained 2 days later shows progressive ischemia in the upper pole with a new large hypoechoic focus involving the medulla & cortex. A thin rim of echogenic peripheral cortex remains.*



(Left) *Longitudinal US in a newborn with oliguria shows diffusely increased echogenicity of the right kidney with diminished corticomedullary differentiation, due to renal vein thrombosis. Scattered echogenic striations are noted.* **(Right)** *Pulsed Doppler US in the same newborn with renal vein thrombosis shows a high-resistance renal arterial waveform with complete reversal of diastolic flow.*



TERMINOLOGY

Definitions

- Obstruction of renal vein(s) by thrombus

IMAGING

General Features

- Best diagnostic clue
  - Enlarged, echogenic kidney with ↓ corticomedullary differentiation
    - Cannot rely on visualization of main renal vein thrombus for diagnosis in neonates
- Location
  - Unilateral (L > R) more common than bilateral
- Size
  - Enlarged kidney in acute stage
  - Normal size or atrophy in later stages

Ultrasonographic Findings

- Grayscale ultrasound
  - Acute phase
    - Echogenic parenchyma, predominantly cortex
      - May see echogenic striations
    - ↓ corticomedullary differentiation
    - Enlarged kidney(s)
  - Chronic phase
    - Parenchymal Ca^{2}
    - Renal size may be normal or atrophied
  - Other findings
    - Visualization of thrombus in main renal vein
      - May or may not be identified in neonates
    - Medullary pyramids can be hypo- or hyperechoic
      - Profoundly hypoechoic pyramids → poorer outcome
    - Associated adrenal hemorrhage or Ca^{2+} may be present
      - Due to impaired venous drainage of adrenal gland
- Color Doppler
  - High-resistance renal arterial waveforms (RI > 0.9) or complete reversal of diastolic flow

CT Findings

- CECT
  - Generally not indicated in neonates
  - Enlarged kidney ± perinephric stranding
  - Delayed or heterogeneous nephrogram
  - Low-attenuation thrombus within main renal vein (± inferior vena cava extension)
  - ± collateral vessels if chronic

MR Findings

- T1WI C+
  - Delayed enhancement
    - Most prominent in medullary pyramids
- MRV
  - Filling defect in main renal vein (also seen on T1/T2WI)

Imaging Recommendations

- Best imaging tool
  - Renal US with Doppler

DIFFERENTIAL DIAGNOSIS

Acute Tubular Necrosis

- Both kidneys typically affected
- Causes include shock, ischemia, infection, hemolytic-uremic syndrome, exogenous or endogenous toxins

Pyelonephritis

- Patchy ↓ in perfusion & corticomedullary differentiation
  - Can be similar to renal vein thrombosis

Transient Neonatal Renal Medullary Hyperechogenicity

- Echogenic central renal medullary pyramids
- Normal renal size, cortical echogenicity, & vascularity
- Not clearly due to Tamm-Horsfall proteinuria

Autosomal Recessive Polycystic Kidney Disease

- Markedly enlarged & echogenic kidneys with extensive fine striations from dilated tubules

Tumor Thrombus

- Clot continuous with renal mass (most commonly Wilms)
- Arterial waveforms in venous thrombus confirmatory

PATHOLOGY

General Features

- Etiology
  - In neonates, thrombosis begins in small intrarenal veins → proximal extension to main renal vein
    - ↑ renal venous pressure & ↓ arterial flow
  - Large number of potential causes
    - Dehydration, sepsis/infection, iatrogenic

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Renal dysfunction, oliguria, hypertension
  - Classic clinical triad of palpable flank mass, hematuria, & thrombocytopenia usually absent

Demographics

- Age
  - Most commonly diagnosed in neonatal period

Natural History & Prognosis

- Irreversible damage occurs in 70%
- Up to 20% will have persistent hypertension

Treatment

- Controversial & may depend upon comorbid conditions
  - Heparin therapy
  - Supportive care without anticoagulants
  - Rarely thrombolytics

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Renal Artery Stenosis

KEY FACTS

TERMINOLOGY

- Renovascular or renin-mediated hypertension
  - Arterial stenosis → ↓ renal parenchymal blood flow → activation of renin-angiotensin system → hypertension

IMAGING

- Arterial stenoses may be extrarenal (aorta, main renal artery), intrarenal, or both
  - Intrarenal more common in children
- CT & MR angiography
  - Focal arterial narrowing ± poststenotic dilation or aneurysm formation
  - Useful to evaluate aorta & main renal arteries; ↓ sensitivity for intrarenal abnormalities
- Pulsed/spectral Doppler shows abnormal renal arterial waveforms
  - Distal to stenosis: Parvus et tardus, prolonged acceleration index, low resistive index (< 0.5)
  - At stenosis: ↑ peak systolic velocity
  - Evaluate aorta if waveforms abnormal bilaterally
- Digital subtraction angiography (DSA)
  - Most sensitive & specific imaging modality
  - Only performed with high clinical suspicion

TOP DIFFERENTIAL DIAGNOSES

- Reflux nephropathy
- Essential hypertension
- Pheochromocytoma

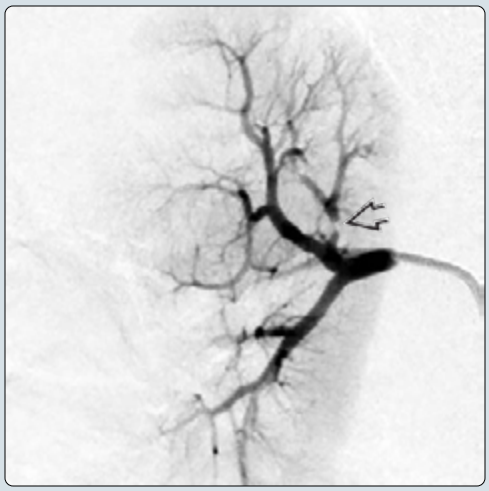
PATHOLOGY

- Common cause: Idiopathic developmental arteriopathy
  - Similar to but distinct from fibromuscular dysplasia
- Associations include neurofibromatosis type 1, Williams syndrome, tuberous sclerosis, & Marfan syndrome
- Other causes: Vasculitis, extrinsic compression, iatrogenic

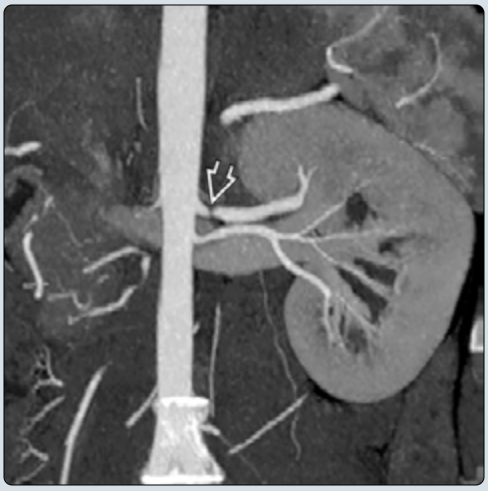
CLINICAL ISSUES

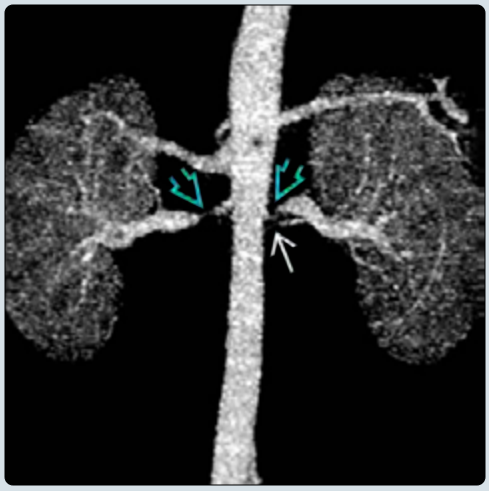
- Severe hypertension refractory to multiple medications
- Endovascular or surgical treatment

(Left) *Pulsed Doppler ultrasound of the right kidney in a 7 year old with refractory hypertension demonstrates an abnormal intrarenal arterial waveform with a diminished & delayed arterial upstroke (parvus et tardus) & an abnormally low resistive index (RI = 0.41).* **(Right)** *Anterior oblique digital subtraction angiogram (DSA) of the right kidney in the same patient reveals a tight stenosis of an intrarenal arterial branch.*



(Left) *Maximum intensity projection CTA in an 8 year old with severe hypertension shows a focal narrowing of a left upper pole accessory renal artery.* **(Right)** *Coronal 3D surface rendering of a CTA in an 11-year-old boy with hypertension demonstrates ostial narrowing of both main renal arteries. There is also a stenosis of an accessory left renal artery.*





KEY FACTS

TERMINOLOGY

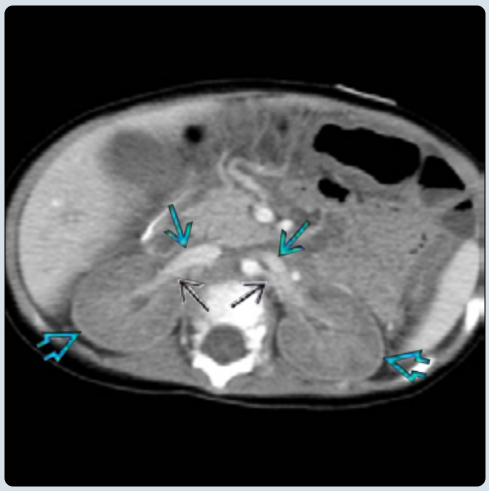
- Thrombotic microangiopathy typically occurring after diarrheal illness (hemorrhagic colitis) from Shiga toxin-producing bacteria (classically *Escherichia coli* O157:H7)
- Clinical triad of hemolytic anemia, thrombocytopenia, & acute renal failure
  - Affected organ systems: Kidneys > CNS (20-50%) > gastrointestinal tract, heart
- Atypical hemolytic uremic syndrome (HUS) often due to dysregulation in alternative complement pathway
  - Diarrhea less common; higher mortality

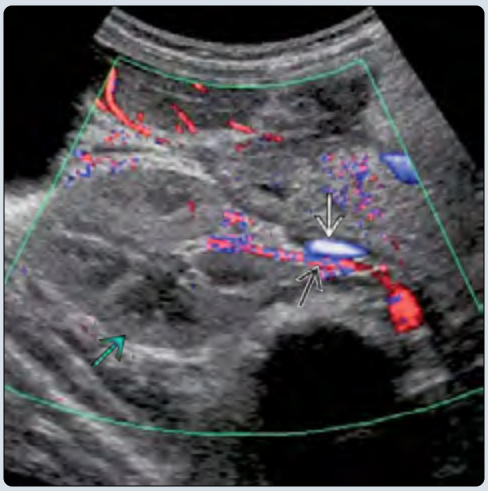
IMAGING

- Bowel: Fluid-filled loops with wall thickening
- Kidneys: ↑ renal cortical echogenicity, ↓ vascular perfusion of parenchyma early; perfusion improves in 2nd-4th weeks
  - Improved diastolic flow precedes urine production
- Brain: Potentially reversible, frequently symmetric lesions
  - Restricted diffusion > T2, FLAIR MR signal abnormalities
  - Deep gray nuclei, deep white matter > cortex
  - Foci of T1 MR shortening due to hemorrhage, necrosis

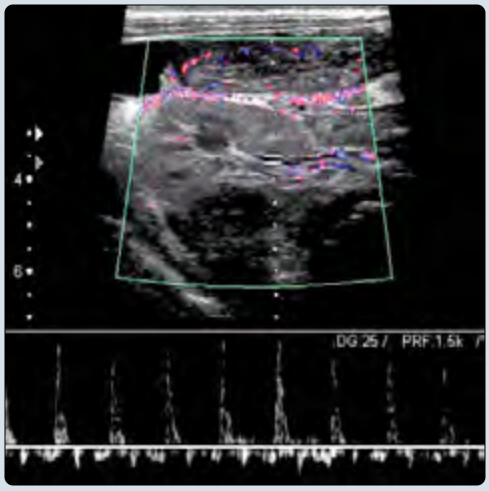
CLINICAL ISSUES

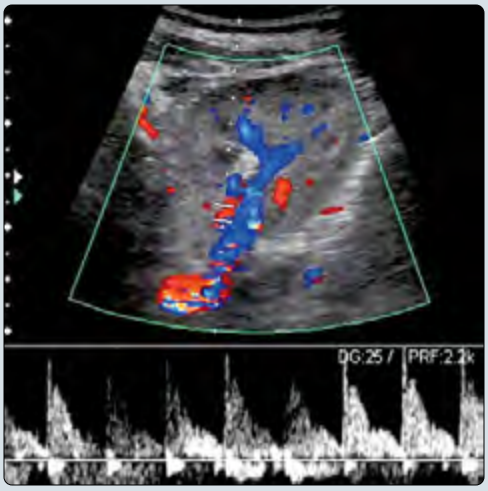
- 1 in 100,000 children affected annually
- Most patients < 5 years of age
- Gastrointestinal illness precedes renal failure by 3-14 days
- With onset of HUS: Fatigue, pallor, ↓ urine output, body/extremity edema, bruising ± seizures, altered mental status, visual disturbances
- Full recovery in 70%; 1-4% mortality
- Long-term sequelae: Renal, neurologic
  - Rarely, delayed development of hypertension & proteinuria 1-2 years after full recovery
  - Impaired neuromotor development
- Typical HUS treatment: Supportive therapy (including dialysis); ± plasma exchange; no direct treatments
- Atypical HUS treatment: As above, + anti-C5 antibody eculizumab for terminal complement blockade





(Left) *Axial CECT in a 15 month old with vomiting & altered mental status shows globally decreased enhancement of the bilateral kidneys, though capsular enhancement remains intact. The main renal veins & arteries appear patent.* **(Right)** *Transverse color Doppler US of the right kidney in the same patient shows turbulent flow in the main renal artery. The main renal vein is patent. However, no vascular flow is seen in the renal parenchyma. The renal cortex was mildly echogenic bilaterally.*





(Left) *Transverse pulsed Doppler US in the same patient shows a high-resistance waveform in the right main renal artery with complete reversal of diastolic flow. The left kidney & left main renal artery showed similar findings. However, the aortic waveforms were normal.* **(Right)** *Transverse pulsed Doppler US in the same patient 9 days later shows improved flow in the right renal parenchyma with return of diastolic flow in the main renal artery. The patient began producing urine the following day.*

KEY FACTS

TERMINOLOGY

- Bladder dysfunction secondary to neurologic disorder

IMAGING

- Voiding cystourethrogram &/or US
  - Towering, contracted bladder with thickened trabeculations
  - Bladder volume variable, ranging from small & contracted to large & atonic
  - Involuntary, uninhibited detrusor contractions
  - High filling pressure
    - Results in ↓ rate of filling or spontaneous cessation of contrast infusion
  - Voiding dysfunction: Inhibited micturition reflex
  - ↑ postvoid residual
  - Secondary bladder & upper tract abnormalities
    - Dilated upper tracts: Vesicoureteral reflux, functional obstruction, scarring

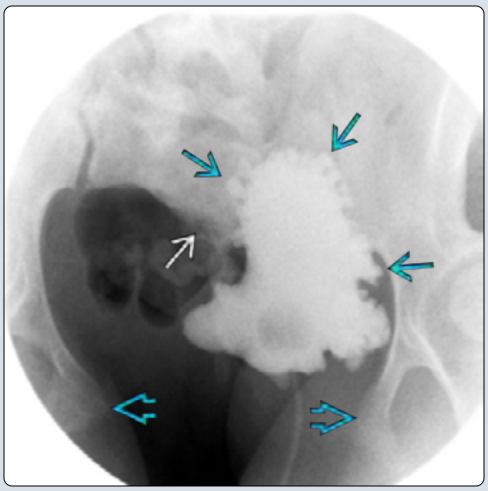
PATHOLOGY

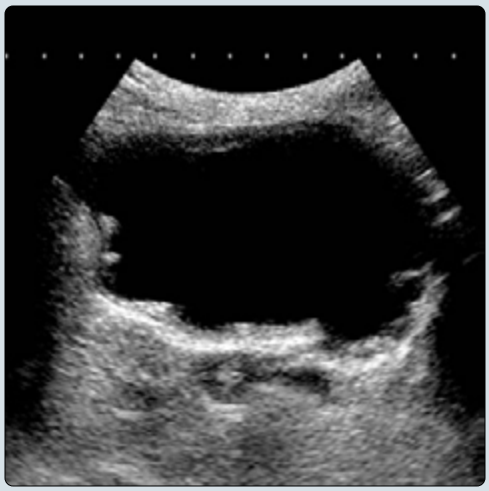
- Classification
  - Contractile bladders (hyperreflexive detrusor)
  - Intermediate (mixed) bladders
  - Acontractile bladders (detrusor areflexia)

CLINICAL ISSUES

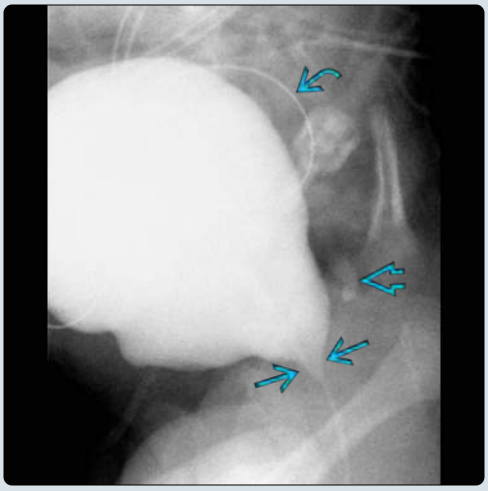
- Frequency, nocturia, urgency, incontinence
- UTI, epididymitis in boys with detrusor-sphincter dyssynergia who are allowed to void
- Upper tract deterioration & chronic renal failure related to ↑ bladder pressure
- Without intervention, 50% show upper urinary tract deterioration in first 5 years of life

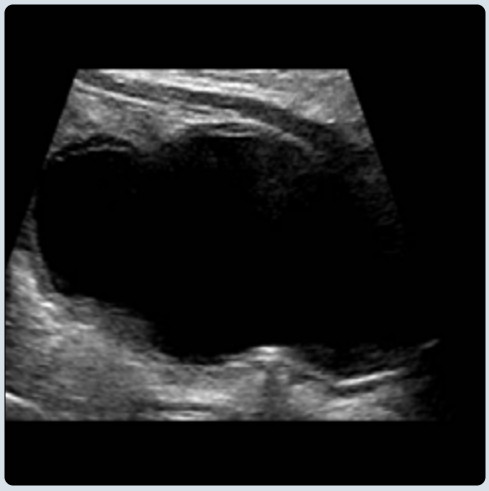
(Left) *Voiding cystourethrogram (VCUG) in a teenage girl with a cloacal exstrophy variant & a neurogenic bladder (NGB) who catheterizes 4x/day shows a small hypertonic bladder with trabeculation & multiple diverticula/pseudodiverticula, the so-called "Christmas tree" bladder. Note the pubic symphysis diastasis & sacral truncation of cloacal exstrophy.* **(Right)** *Transverse US of the bladder shows a trabeculated, thickened bladder in the same patient with cloacal exstrophy variant.*





(Left) *Oblique cystogram in a 7 day old status post myelomeningocele (MMC) repair shows an atonic, mildly lobular, large capacity bladder with no wall thickening, an open bladder neck, left vesicoureteral reflux, & no significant bladder emptying due to an atonic detrusor, consistent with NGB. Note the ventriculoperitoneal shunt catheter.* **(Right)** *Transverse US in the same patient after MMC repair shows a large, mildly lobular bladder without significant bladder wall thickening in an atonic-type NGB.*





TERMINOLOGY

Abbreviations

- Neurogenic bladder (NGB)

Definitions

- Bladder dysfunction due to neurologic disorder

IMAGING

General Features

- Best diagnostic clue
  - Towering, small, contracted bladder with thickened trabeculations ("Christmas tree" bladder)
  - Vertical, large, smooth, borderline thick, atonic bladder
  - Abnormal filling & emptying phases of modified voiding cystourethrogram (VCUG)
- Size
  - Bladder volume variable; small & contracted to large & atonic
- Morphology
  - Taller than wide, trabeculated, thickened ± diverticula

Radiographic Findings

- Sacral anomalies, spinal dysraphism, pubic symphysis diastasis, scoliosis, obstipation
- Urinary tract calculi
- Can be normal

Fluoroscopic Findings

- VCUG
  - Noncompliant bladder
    - Involuntary, uninhibited detrusor contractions
      - Thickened, trabeculated bladder wall (hypercontractile & intermediate bladders)
    - Serrated mucosa with prominent interureteric ridge (hypercontractile bladder)
    - High filling pressure
    - Results in ↓ rate or spontaneous cessation of contrast infusion
    - Relatively low bladder capacity
    - Leakage around catheter at low volume/high pressure
  - Acontractile bladders (detrusor areflexia)
    - Little if any detrusor contraction
    - Relatively high bladder capacity
    - Relatively little bladder thickening
    - Bladder leaks but does not empty spontaneously
      - Large bladder residual, incomplete emptying
  - May or may not sense need to void (inhibited micturition reflex) or have physiologic urination, depending on level of lesion
  - Voiding dysfunction
    - Detrusor-sphincter dyssynergia: Seen with contractile bladders
      - Sudden opening of bladder neck at ↑ pressure
      - Reflexive contraction of external sphincter: Functional obstruction
      - Ejaculatory duct reflux → epididymitis in boys
      - Bulging of posterior urethra
    - Gradual bladder neck opening: Intermediate (mixed) bladders
      - Bladder neck incompetent but closed on early filling
      - Opens with increasing volume/pressure
      - Funnel-like appearance: No bulging of posterior urethra
    - Persistent bladder neck opening: Acontractile bladder
      - Incompetent, open throughout filling phase
  - Secondary bladder & upper tract abnormalities
    - Diverticula/pseudodiverticula
    - Vesicoureteral reflux (VUR) in 20-25% cases
    - Dilated upper urinary tracts
    - ↑ postvoid residual
  - Complication of VCUG in NGB
    - Autonomic dysreflexia
      - Life-threatening condition
      - Spinal lesions above T6
      - Sympathetic discharge in response to bladder distention or urethral catheterization
      - Severe paroxysmal hypertension, anxiety, sweating, piloerection, headaches, bradycardia
      - Treatment: Evacuate bladder & catheter, elevate head of table, check blood pressure, pharmacologic intervention if necessary

Ultrasonographic Findings

- Grayscale ultrasound
  - Small contracted/large atonic bladder; ± wall thickening; ↑ postvoid residual
  - Diverticula, pseudodiverticula
  - ± urinary tract dilation, unilateral or bilateral
  - ± VUR or functional obstruction
  - ± renal scarring

Imaging Recommendations

- Best imaging tool
  - Modified VCUG
- Protocol advice
  - VCUG: Follow-up yearly
    - ± videourodynamics (if available)
  - Renal & bladder sonography: Follow-up every 6 months

DIFFERENTIAL DIAGNOSIS

Bladder Outlet Obstruction

- Including posterior urethral valves, stricture, hypoplasia
- Prior to ablation of valves, severity of bladder findings dependent on VUR
  - ↑ grade of VUR → less severe bladder findings

Pelvic Mass

- ± extrinsic mass or filling defect at bladder base; may lead to degree of obstruction/voiding difficulties
  - Ovarian, vaginal, or prostatic rhabdomyosarcoma
  - Sacrococcygeal teratoma
- No trabeculation; features do not change with voiding

Multiple Diverticula

- Williams syndrome
- Menkes syndrome
- Cutis laxa or Ehlers-Danlos

PATHOLOGY

General Features

- Etiology
  - Myelodysplasia
  - Sacral agenesis
  - Cerebral palsy
  - Traumatic spinal cord lesions
- Associated abnormalities
  - Anorectal malformations
  - Lipomeningocele
  - Caudal regression
  - Occult congenital spinal dysraphism
  - Spinal cord tethering

Staging, Grading, & Classification

- Multiple classification schemes
- Type of bladder dysfunction
  - Contractile bladders (hyperreflexive detrusor)
    - Upper motor neuron lesion
    - Uninhibited detrusor contraction, sudden bladder neck opening
    - Detrusor-sphincter dyssynergia → potential epididymitis in boys allowed to void
    - Functional obstruction; deterioration of upper tracts, especially with VUR
  - Intermediate (mixed) bladders
    - Lesions overlap neural pathways
  - Acontractile bladders (detrusor areflexia)
    - Lower motor neuron lesion
    - No detectable detrusor contractions
    - Sphincter weakness incontinence: Leakage of contrast around catheter, especially during coughing
- Bladder function
  - Inability to store urine properly
  - Inability to evacuate urine properly
- Level of neurologic disorder
  - Upper motor neuron lesion
  - Lower motor neuron lesion

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Failure to empty bladder
  - Frequency, nocturia, urgency, retention, incontinence
  - Urinary tract infection (UTI)
  - Bladder stones, hematuria
  - Epididymitis

Demographics

- Age
  - Neonatal to adolescence

Natural History & Prognosis

- Complications
  - Pyelonephritis
  - Hydronephrosis
  - Urolithiasis
  - Epididymitis
  - Sexual dysfunction
  - Autonomic dysreflexia
- Upper tract deterioration, UTI, & chronic renal failure related to ↑ bladder pressure
  - Without bladder management, 50% develop nephropathy in first 5 years of life
  - Predictive indicators for deterioration of renal function
    - Detrusor-sphincter dyssynergia
    - VUR
    - High bladder-filling pressure
    - Poor bladder compliance
    - High leak point pressure

Treatment

- Treatment goals
  - Preservation of renal function
  - Avoidance of UTI, epididymitis
  - Achieve appliance-free, social continence
- Therapeutic maneuvers to achieve goals
  - Clean intermittent catheterization
  - Medications: Antibiotics, anticholinergics, etc.
  - Surgical procedures
    - Operation for continence
    - Bladder augmentation
    - Artificial sphincters
- Hyperreflexia
  - ↑ volume: Cystoplasty, muscular or fascial slings, parasympatholytic drugs, botulinum A toxin
  - ↑ voiding: Catheter, transurethral sphincterotomy
- Hyporeflexia
  - Bladder training, intermittent catheterization, bladder neck resection/denervation, parasympathomimetic drugs

DIAGNOSTIC CHECKLIST

Consider

- Extraurinary findings in patients with clinical suspicion of NGB

Image Interpretation Pearls

- During VCUG
  - Monitor contrast flow
  - Evaluate bladder contour, sphincter function, bladder filling volume, & degree of emptying

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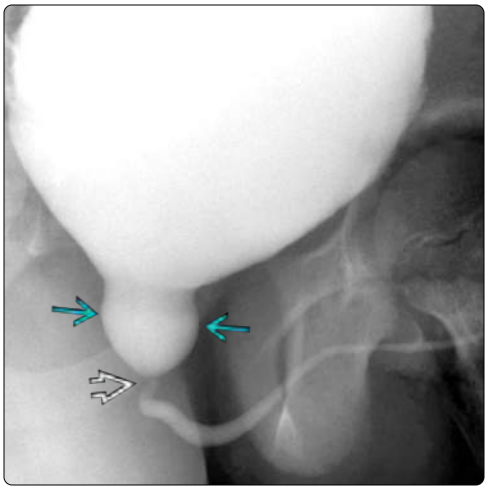
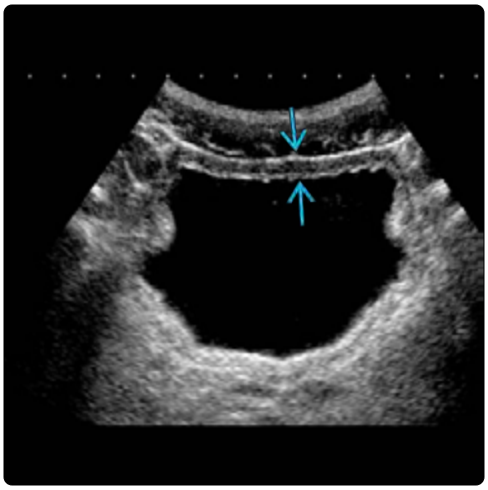
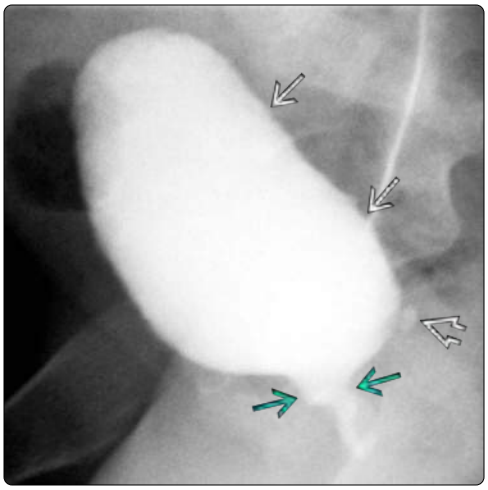
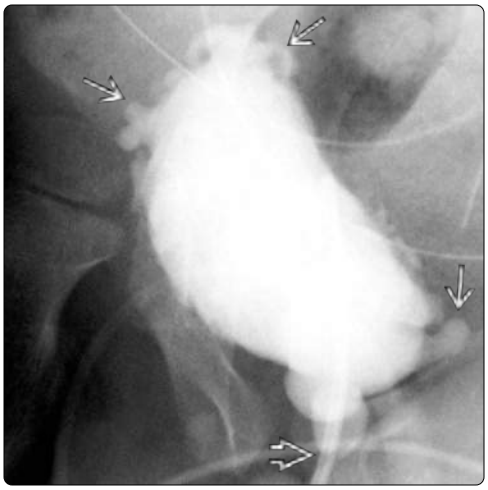


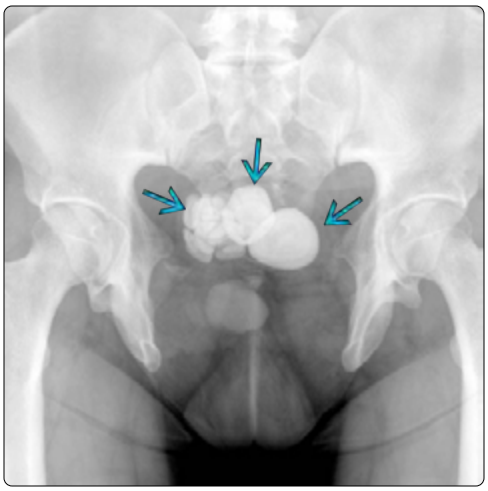


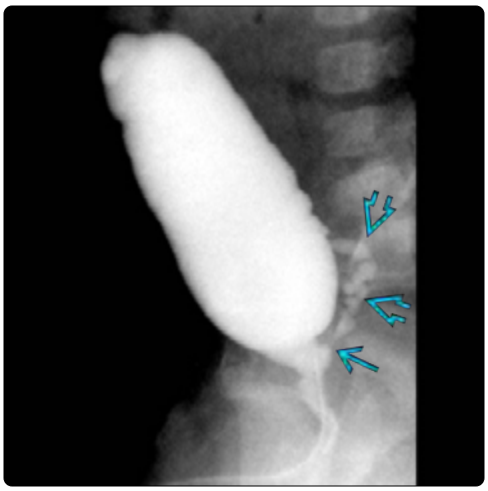
(Left) *VCUG in an 11-year-old boy with NGB on intermittent catheterization shows a caliber change of the proximal urethra ⇨ with dilation of the posterior urethra ⇨. Considerations include detrusor-sphincter dyssynergia & catheter-related urethral stricture.* **(Right)** *Transverse US in the same patient with NGB & VCUG findings suggestive of detrusor-sphincter dyssynergia or urethral stricture shows a diffusely thickened bladder wall ⇨, which is characteristic of NGB or bladder outlet obstruction.*





(Left) *VCUG of an infant with a history of MMC repair shows a towering bladder with wall thickening ⇨, a small diverticulum at the bladder base ⇨, & a lax bladder neck sphincter ⇨.* **(Right)** *VCUG obtained several years later in the same patient without appropriate follow-up shows the development of additional diverticula ⇨. Also note the bulging bladder neck with contracted external sphincter ⇨ on this nonvoiding image (due to detrusor-sphincter dyssynergia).*





(Left) *AP radiograph in a young adult male patient with a history of bladder exstrophy (suggested by pubic symphysis diastasis & a normal sacrum) shows multiple variable-sized lamellated bladder calculi ⇨ in this patient who had bladder augmentation.* **(Right)** *VCUG in an 8-month-old boy status post repaired anorectal malformation shows a tall, trabeculated NGB. During voiding, there is reflux into the left ejaculatory duct ⇨ & seminal vesicle ⇨, suggesting high posterior urethral pressures & increasing the risk of left epididymitis.*

Bladder Diverticula

KEY FACTS

TERMINOLOGY

- Herniation of urinary bladder mucosa through bladder detrusor muscle
  - Primary (congenital): Poor muscular backing near ureterovesical junction (UVJ)
    - Periureteral (Hutch): Most common congenital diverticulum (90%)
  - Secondary (acquired)
    - Chronically elevated bladder pressures
    - Weak bladder wall in connective tissue disorders
    - Iatrogenic/traumatic causes (prior surgery/catheter)

IMAGING

- Best diagnostic clues
  - Round cystic focus directly adjacent to bladder
    - Changes size with bladder filling &/or voiding
    - Anechoic on ultrasound; ± jet of urine into bladder
    - Fills with contrast on VCUG
- Diverticula with narrow neck may be small or absent during bladder filling & only seen with voiding
  - Contrast may remain after bladder emptying
- Vesicoureteral reflux (VUR) in 50%
  - Large diverticulum incorporates & distorts UVJ → VUR
- Without history of neurogenic bladder or bladder outlet obstruction, multiple diverticula should suggest syndromes
  - Williams, Menkes, Ehlers-Danlos, cutis laxa

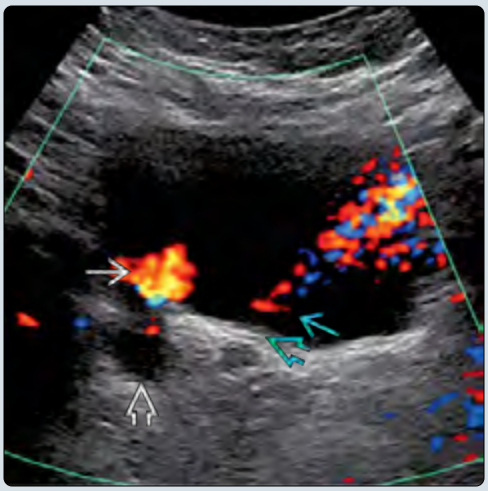
TOP DIFFERENTIAL DIAGNOSES

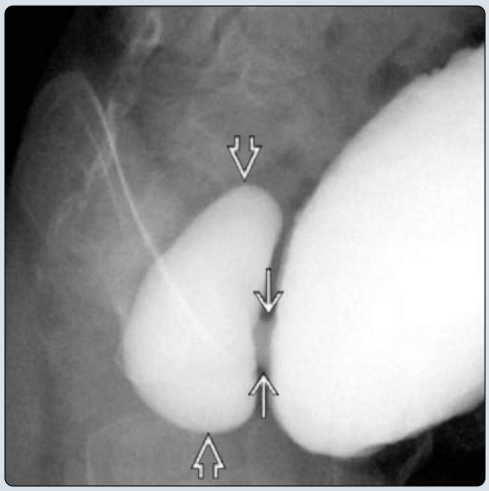
- Everting ureterocele, ureteral stump, ovarian/paraovarian cyst, gastrointestinal duplication cyst

CLINICAL ISSUES

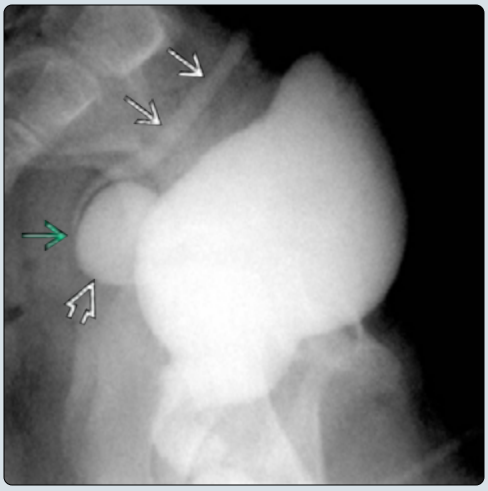
- Asymptomatic vs. urinary tract infection, hematuria, voiding dysfunction
- Surgery for complications: Resection of diverticulum ± ureteral reimplantation

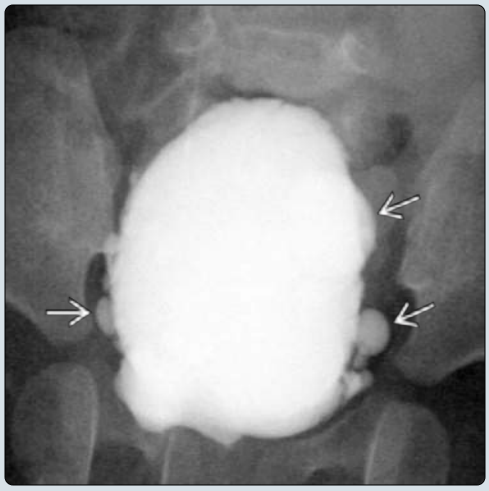
(Left) *Transverse color Doppler ultrasound of the urinary bladder shows not only a jet of urine ➡ draining from a diverticulum ➡ but also a jet of urine ⇨ emanating from the adjacent ureteral orifice ⇨ in this patient with a periureteral (Hutch) diverticulum.* **(Right)** *Oblique fluoroscopic voiding cystourethrogram was performed tangential to the neck ➡ of a moderate-sized bladder diverticulum ➡. A projection demonstrating the neck should be acquired when a diverticulum is encountered.*





(Left) *Lateral fluoroscopic VCUG shows VUR ➡ accompanying a periureteral diverticulum ➡. In this case, the ureter inserts directly into the diverticulum ⇨, which is important information for the urologist making management decisions.* **(Right)** *Frontal fluoroscopic VCUG of an infant girl's bladder at capacity shows multiple diverticula ➡. If there are no clinical findings of neurogenic bladder or bladder outlet obstruction, considerations should include Williams, Menkes, Ehlers-Danlos, or cutis laxa syndromes.*





TERMINOLOGY

Definitions

- Herniation of urinary bladder mucosa through bladder detrusor muscle
  - Primary (congenital) due to poor muscular backing near ureterovesical junction (UVJ)
  - Secondary (acquired) due to
    - Chronically elevated bladder pressures
    - Weak bladder wall in various connective tissue disorders
    - Iatrogenic/traumatic causes (prior surgery/catheter)

IMAGING

General Features

- Best diagnostic clue
  - Focal round or ovoid outpouching of urinary bladder
- Location
  - Periureteral (Hutch): Most common primary diverticulum (90%)
  - Nonperiureteral: Often secondary

Ultrasonographic Findings

- Grayscale ultrasound
  - Normal: Diverticulum may be decompressed & not visible
  - Focal bladder wall thickening adjacent to ureteral orifice
    - Can appear as soft tissue nodule within or just outside bladder wall
  - Round, anechoic structure adjacent to bladder
    - ± echogenic shadowing calculus
  - Ureteral dilation if ipsilateral vesicoureteral reflux (VUR) present
- Color Doppler
  - Turbulence within diverticulum
  - Color jet of urine into bladder lumen
    - Especially helpful when communication with bladder not apparent on grayscale images

Fluoroscopic Findings

- Voiding cystourethrogram
  - Outpouching of contrast at any bladder location
    - Periureteral most common
    - Oblique images help visualize neck of diverticulum
  - Can enlarge during voiding as bladder pressure ↑
  - Contrast persists in diverticulum after bladder emptying
  - VUR in 50%: Larger diverticula predispose to VUR due to UVJ distortion
    - Ureter may appear to insert into diverticulum
    - Diverticula rarely obstruct ureter
  - Urethral obstruction from large diverticulum (uncommon)
  - Multiple diverticula seen with syndromes
    - Williams, Menkes, cutis laxa, Ehlers-Danlos

Imaging Recommendations

- Protocol advice
  - Bladder sonography including kidneys
    - Scan bladder in full & postvoid states
  - Fluoroscopic VCUG
    - Be sure to image during & after voiding
      - Diverticulum may only fill during voiding
      - Document drainage pattern of diverticulum
    - Oblique images help visualize diverticulum neck & site of UVJ relative to diverticulum (if VUR occurs)

DIFFERENTIAL DIAGNOSIS

Everting Ureterocele

- Round, smooth-walled intraluminal cyst at UVJ
- Everts near bladder capacity; inverts postvoid
- Duplex kidney ± obstructed upper pole, lower pole VUR

Ureteral Stump

- Often associated with ipsilateral multicystic dysplastic kidney/nephrectomy

Urachal Diverticulum

- Located at bladder dome pointing toward umbilicus

Ovarian or Paraovarian Cyst

- No communication of cyst with bladder
- Ovarian parenchyma/vascularity may surround cyst

Gastrointestinal Duplication Cyst

- Sonographic trilaminar wall may appear similar to bladder
- No communication of cyst with bladder
- Peristalsis strongly suggests duplication cyst

PATHOLOGY

General Features

- Associated abnormalities
  - Williams syndrome
  - Menkes syndrome
  - Cutis laxa
  - Ehlers-Danlos syndrome

CLINICAL ISSUES

Presentation

- Most found incidentally (asymptomatic)
- Other cases: Urinary tract infection, hematuria, voiding dysfunction, pain

Natural History & Prognosis

- Complications determine need for operative treatment
  - Stagnant urine: Infection, hematuria, calculi
  - Deformed UVJ: VUR or obstruction
  - Urethral obstruction: Uncommon

Treatment

- Conservative: Observation ± chemoprophylaxis
- Surgery: Resection of diverticulum ± ureteral reimplant

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Post-Deflux Procedure Appearance

KEY FACTS

TERMINOLOGY

- Bulking agent injected near ureterovesical junction (UVJ) to prevent vesicoureteral reflux (VUR)
  - Deflux is dextranomer-hyaluronic acid copolymer
    - Biodegradable, does not migrate
  - Other brand agents: Dexell, Macroplastique, etc.
- Deflux procedure
  - Injected submucosally near UVJ
  - Techniques: STING, hydrodistention implantation technique (HIT), double HIT injections
  - Sufficient agent used to create mass effect
  - Effectively alters angle of intramural ureter
  - Favors detrusor muscle creating effective 1-way valve

IMAGING

- Focal round mass at UVJ in patient with history of antireflux procedure; uniformly echogenic on US
- Ca^{2+} can be seen months to years after injection
- Follow-up fluoroscopic VCUG or nuclear cystogram weeks to months later to assess change in VUR

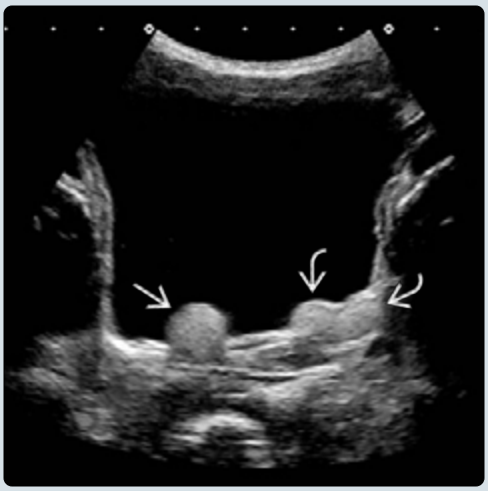
TOP DIFFERENTIAL DIAGNOSES

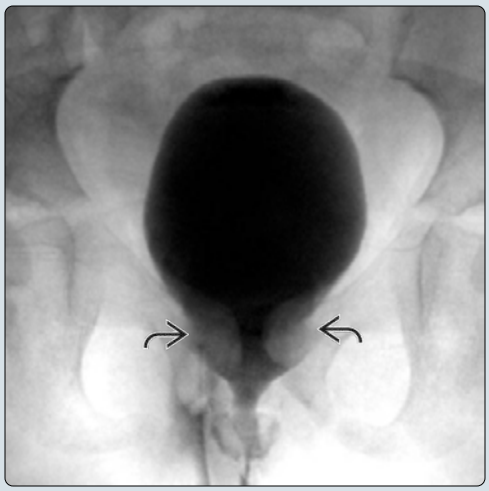
- Urinary tract calculus
- Rhabdomyosarcoma
- Ureterocele
- Fungus ball
- Bladder clot

CLINICAL ISSUES

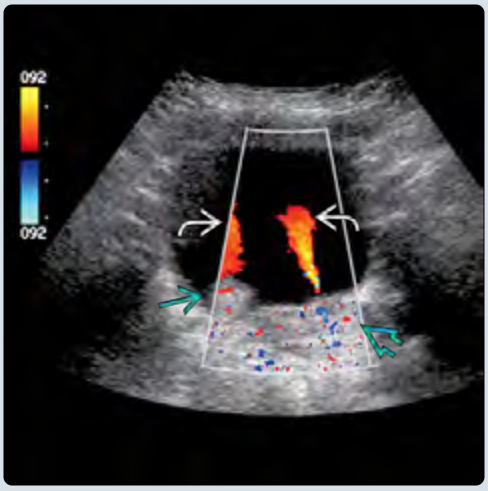
- Hydroureteronephrosis may be seen transiently following Deflux procedure
- Ultrasound useful to confirm ureteral jet/patency
- Overall success rates of procedure high
- Rare complications include obstruction, urosepsis, acute renal failure
- Recovery time shorter than with open procedures

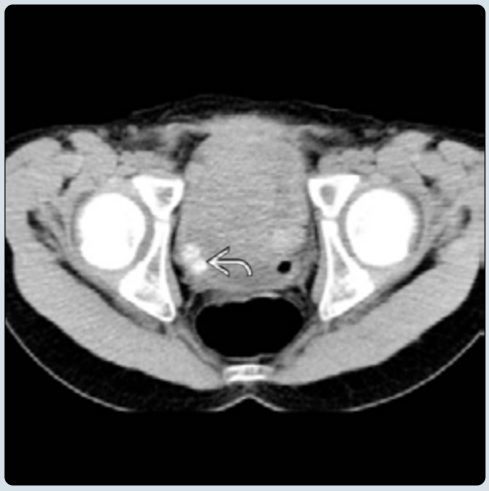
(Left) *Transverse US through a well-filled urinary bladder shows 2 echogenic mounds at the bladder base. The right mound is rounded while the left is bilobed. Both represent Deflux material injected at the ureterovesical junctions (UVJ).* **(Right)** *Voiding cystourethrogram (VCUG) in the same patient following Deflux injections shows expected filling defects at the UVJ bilaterally. A VCUG or nuclear cystogram is often performed several weeks after the procedure to confirm successful treatment.*





(Left) *Transverse color Doppler US shows a right Deflux mound within the bladder lumen & the left mound projecting posterior to the bladder wall. Bilateral ureteral jets (with mildly altered angles of the urine streams) confirm UVJ patency.* **(Right)** *Axial NECT in a teenage patient shows dystrophic calcifications along the bladder wall at a site of remote Deflux injection.*





TERMINOLOGY

Abbreviations

- DHA, DxHA, copolymer of hyaluronic acid
- Deflux most commonly used in USA; Dexell & Macroplastique also used in Europe/Asia

Definitions

- Bulking agent injected near ureterovesical junction (UVJ) to alter UVJ orientation & prevent vesicoureteral reflux (VUR)
  - Deflux (Q-Med; Uppsala, Sweden): Dextranomer-hyaluronic acid copolymer
    - In use clinically since 1995
    - Relatively large particle size inhibits migration
    - Biodegradable, stable in vivo
  - Dexell (Istem Medical; Ankara, Turkey)
    - Manufactured since 2007
    - Polycationic derivative of Dextran & cross-linked hyaluronic acid
    - Similar size & biologic profile to Deflux
  - Macroplastique (Congentix; Geleen, Netherlands)
    - FDA approved in 2006
    - Flexible, soft, textured implants of heat-vulcanized polydimethylsiloxane (solid silicone elastomer)
    - Suspended in bioexcretable polyvinylpyrrolidone gel

IMAGING

General Features

- Best diagnostic clue
  - Focal mass at UVJ in patient with history of anti-VUR procedure
- Location
  - Periureteral at UVJ
  - Submucosal location desired
    - Subureteral transurethral injection: STING technique initially widely used
    - Hydrodistention implantation technique [(HIT) with intraluminal submucosal ureteral injection] was then developed
    - Double HIT (proximal & distal intraluminal submucosal injection) now most often used
  - Intramuscular & subserosal spread common
- Size
  - 1-2 mL per ureter
- Morphology
  - Round or ovoid
  - Bilobed appearance common, may be due to
    - Intramuscular or subserosal spread
    - 2 injections in tandem (double HIT)
  - Can calcify after years: 2% after 4 years

Radiographic Findings

- Dystrophic calcium occasionally seen in mound on radiograph

Ultrasonographic Findings

- Round or ovoid
- Homogeneously echogenic
- Posterior shadowing if calcified, mimics UVJ stone
- Avascular on Doppler imaging; "twinkle artifact" if calcified

Imaging Recommendations

- Best imaging tool
  - Ultrasound encounters these "masses" most often
  - Also seen by VCUG, MR, CT, & possibly radiograph (if calcified)
  - VCUG or nuclear cystogram performed several weeks following procedure to document success
    - ↓ in grade of VUR, even if not completely resolved, considered procedural success

DIFFERENTIAL DIAGNOSIS

Urinary Tract Calculus

- Most smaller, less round, & more uniformly echogenic (with ↑ posterior shadowing or "twinkle artifact") compared to Deflux mound
- Typical renal colic history ± hematuria

Rhabdomyosarcoma

- More lobulated & heterogeneous than Deflux mound
- Doppler flow expected in bladder neoplasm

Ureterocele

- Cystic protrusion of distal ureter into bladder on cross-sectional modalities

Fungus Ball

- Typically in immunocompromised patients
- Mobile & more heterogeneous on imaging

Bladder Clot

- Expect history of hematuria & mobile debris in bladder
- Doppler should not show flow in clot

Inflammatory Myofibroblastic Pseudotumor

- Nonspecific lobulated mass with internal vascularity

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - History of Deflux procedure
- Other signs/symptoms
  - Hydronephrosis & hydroureter may be seen transiently
  - Ultrasound useful to confirm ureteral jet/UVJ patency

Natural History & Prognosis

- Success rates ~ 70-80% for VUR grades 2-3 & ~ 50-60% for VUR grades 4-5
- Reinjection vs. reimplantation if initial injection fails

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KEY FACTS

TERMINOLOGY

- Malignant tumor of striated muscle originating from any of pelvic organ (but paratesticular discussed separately)

IMAGING

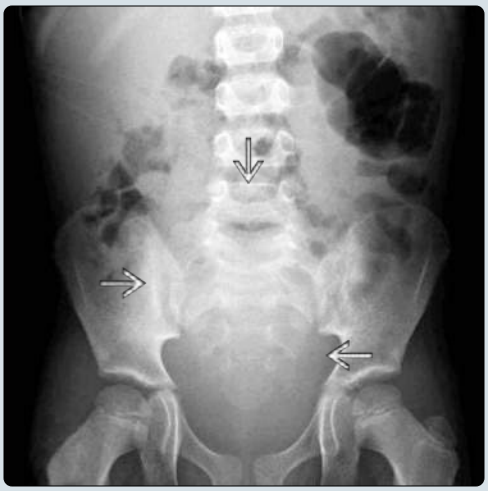
- Best clue: Large, heterogeneous, predominantly solid pelvic mass in child with symptoms of urinary tract obstruction
  - Variable cystic components
  - Botryoid variety resembles "bunch of grapes" with cysts protruding into lumen of vagina or urinary bladder
- May originate from bladder, vagina, cervix, uterus, pelvic side walls, prostate, & paratesticular tissues
  - RMS may also occur in adjacent non-GU soft tissues
- Tumors spread by local extension as well as lymphatic & hematogenous routes, sending metastases to lungs, liver, & bone
  - 15-20% have metastases at diagnosis
- Work-up typically includes
  - US for initial investigation of urinary tract symptoms or palpable mass
  - CECT or MR for further tumor characterization & localization
  - Staging with chest CT & PET

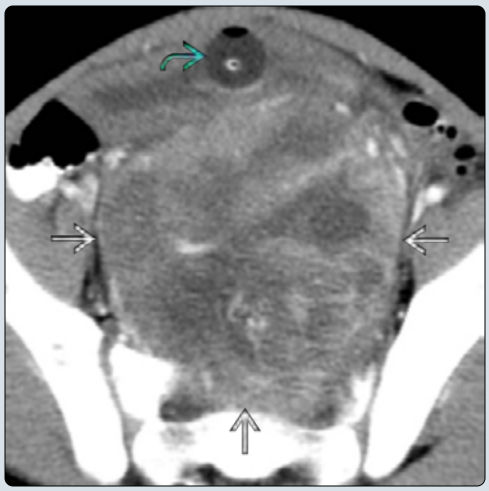
PATHOLOGY

- Small round blue cell tumor of primitive muscle cells
- Major histologic types: Embryonal (majority, especially in GU sites), alveolar, & undifferentiated (adults)
- Bladder & prostate sites unfavorable, automatically at least stage II

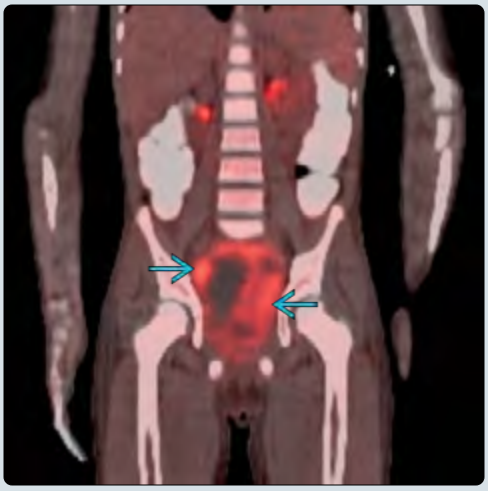
CLINICAL ISSUES

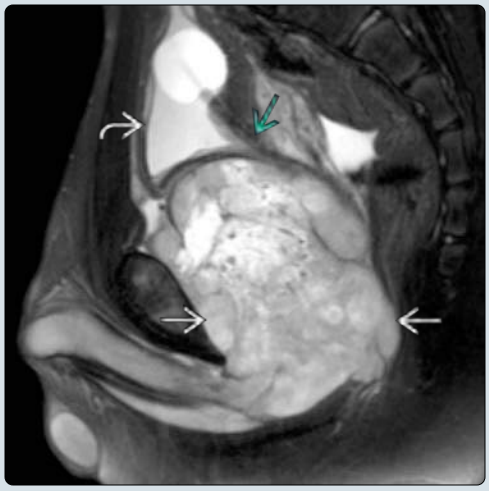
- Peak incidence: 2-6 years old
  - 75% < 5 years old at diagnosis
- Surgery, chemotherapy, & radiation therapy combined
- 5-year survival: Stage I (93%) vs. stage IV (~ 30%)
  - Embryonal type has better prognosis than alveolar





(Left) *AP radiograph of a 6-year-old boy shows displacement of bowel by a soft tissue mass in the pelvis, initially suspected to be due to urinary retention but not relieved by bladder catheterization.* **(Right)** *Axial CECT in the same 6 year old with urinary retention shows a large, heterogeneously enhancing mass displacing the urinary bladder (with Foley balloon in place) anteriorly.*





(Left) *Coronal FDG PET/CT in the same patient shows predominantly peripheral uptake in the pelvic tumor but no evidence of metastatic disease. Biopsy confirmed a rhabdomyosarcoma originating from the bladder base/prostate.* **(Right)** *Sagittal T2 FS MR in a 7-year-old boy shows a large, heterogeneous, intermediate to high signal intensity rhabdomyosarcoma arising from the prostate. Note the superior displacement of the bladder & the altered course of the catheter.*

TERMINOLOGY

Abbreviations

- Genitourinary (GU) rhabdomyosarcoma (RMS)

Synonyms

- Botryoid tumor, sarcoma botryoides, embryonal RMS

Definitions

- Malignant tumor of striated muscle originating from any pelvic organ (but paratesticular discussed in separate chapter)

IMAGING

General Features

- Best diagnostic clue
  - Large heterogeneous predominantly solid pelvic mass in child with symptoms of urinary tract obstruction
- Location
  - May originate from urinary bladder, vagina, cervix, uterus, pelvic side walls, prostate, & paratesticular tissues
    - RMS may also occur in adjacent non-GU soft tissues
  - Tumors spread by local extension as well as lymphatic & hematogenous routes, sending metastases to lungs, liver, & bone
- Morphology
  - Heterogeneous, round or lobulated, solid tumor
  - Botryoid variety resembles "bunch of grapes" protruding into lumen of vagina or urinary bladder

Radiographic Findings

- Radiography
  - Soft tissue mass displacing bowel out of pelvis; can mimic distended urinary bladder

Ultrasonographic Findings

- Grayscale ultrasound
  - Round or lobulated tumor distorting &/or extending into urinary bladder
  - Mass typically large & heterogeneous
    - Most commonly solid with variable cystic components
  - Look for evidence of urinary tract obstruction & adenopathy
- Color Doppler
  - Internal vascularity variable in degree; presence of internal flow helps confirm solid neoplasm
  - Useful to trace displaced & compressed vessels
  - Vascular invasion unusual

Fluoroscopic Findings

- Voiding cystourethrogram
  - Distortion of urinary bladder lumen by intrinsic or extrinsic mass
  - Filling defect may be round, lobulated, or irregular

CT Findings

- CECT
  - Heterogeneously enhancing, predominantly solid mass
  - Frequently locally invasive; look for disruption of fat planes
  - Search for adjacent adenopathy
  - Include liver due to frequency of metastatic disease

MR Findings

- Intermediate to low T1, intermediate to high T2 signal intensity
- Variable degrees of enhancement, ranging from mild & heterogeneous to intense & uniform
- DWI
  - Tumor may restrict diffusion
  - DWI ↑ conspicuity of all lymph nodes (normal & pathologic), helping draw attention to mildly enlarged nodes that might otherwise go unnoticed

Nuclear Medicine Findings

- Bone scan
  - Traditionally used for bony metastatic disease
- PET/CT
  - Improves staging with ↑ sensitivity for nodal spread & distant metastases
  - May improve assessment of therapeutic response

Imaging Recommendations

- Best imaging tool
  - US typically used for initial investigation of urinary tract symptoms or palpable mass
  - CECT or MR for further tumor characterization & localization
  - Staging with chest CT & PET

DIFFERENTIAL DIAGNOSIS

Ureterocele

- Cystic protrusion of distal ureter into bladder; ureteral jet &/or continuity with dilated ureter may help confirm

Bladder Hematoma

- Clinical history of instrumentation, trauma, cystitis, or chemotherapy
- Heterogeneous filling defect lacking Doppler flow

Ovarian Tumor

- Differentiation can be difficult; attempt to identify organ of origin
- Ovarian malignancies often larger with higher likelihood of peritoneal spread (nodules, ascites)

Organ of Zuckerkandl Neuroblastoma

- Younger age, Ca^{2+}, & encasement of vessels

Burkitt Lymphoma

- Differentiation can be difficult
- Bowel wall involvement more likely

Hematometrocolpos

- Look for layering or swirling debris on US
- Blood products may be bright on T1WI MR

Sacrococcygeal Teratoma

- Heterogeneous solid &/or cystic presacral mass associated with coccyx
- Most commonly exophytic from perineum in newborn with variable size of internal components

Pelvic Inflammatory Disease/Abscess

- Complex mass with peripheral hyperemia, internal debris
- Search for pertinent clinical history

PATHOLOGY

General Features

- Etiology
  - RMS can occur virtually anywhere in body
  - Thought to arise from primitive muscle cells
- Associated abnormalities
  - ↑ incidence in
    - Neurofibromatosis type 1
    - Li-Fraumeni syndrome
    - Rubinstein-Taybi syndrome
    - Beckwith-Wiedemann syndrome
    - *DICER1* mutation
  - Environmental factors linked to RMS
    - Parental use of marijuana & cocaine
    - In utero radiation exposure
    - Exposure to alkylating agents

Staging, Grading, & Classification

- Based on tumor invasiveness, tumor size, nodal disease, & metastases
  - Stage I* = T1 or T2; T size a or b; nodes N0, N1, or NX; M0
  - Stage II = T1 or T2; T size a; nodes N0 or NX; M0
  - Stage III = T1 or T2; T size a with nodes N1 or T size b with N0, N1, or NX; M0
  - Stage IV = T1 or T2; T size a or b; nodes N0 or N1; & M1
    - *GU site excluding bladder & prostate
      - Bladder & prostate unfavorable, automatically at least stage II
    - T (tumor): T1 = confined to organ of origin; T2 = local extension
    - T size: a ≤ 5 cm diameter; b > 5 cm diameter
    - N (regional nodes): N0 = not clinically involved; N1 = involved; NX = unknown
    - M (metastases): M0 = no distant metastases; M1 = distant metastases

Gross Pathologic & Surgical Features

- Botryoid subtype has cysts of mucosanguineous fluid
  - Can have transparent, gelatinous appearance

Microscopic Features

- Small round blue cell tumor
- Rhabdomyoblasts are hallmark
  - Not always present, especially if poorly differentiated
- Histochemical markers for muscle cells helpful: Desmin, myoglobin, actin
- Disseminated rhabdomyoblasts in bone marrow mimic leukemia
- 4 major histologic types
  - Embryonal (majority, especially in GU sites)
    - Botryoid variant of embryonal type (5%)
  - Alveolar
  - Undifferentiated (adults)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Large pelvic mass
  - Urinary symptoms such as dysuria, hematuria, frequency, urinary retention
  - Pain variably present
- Other signs/symptoms
  - Vaginal discharge or constipation

Demographics

- Age
  - Peak incidence: 2-6 years old
  - 75% < 5 years old at diagnosis
  - Paratesticular tumors more common in adolescents
- Gender
  - M:F = 2-3:1 for genitourinary tumors
  - M = F for head, neck, & extremity tumors
- Epidemiology
  - 250 new cases per year in USA
  - 4.5 cases per million

Natural History & Prognosis

- 5-year survival rates
  - Stage I (93%)
  - Stage II (81%)
  - Stage III (~ 50%)
  - Stage IV (~ 30%)
- Embryonal cell type has better prognosis than alveolar
- RMS of paratesticular tissues & vas deferens have best prognoses
- 15-20% have metastases at diagnosis

Treatment

- Surgery, chemotherapy, & radiation therapy
  - Initial surgery includes wide margins (when possible) & lymph node sampling
  - Pelvic exenteration type surgeries have fallen out of favor
    - Organ sparing surgery now performed
    - Gonads often moved out of radiation field temporarily
  - Secondary cancers in XRT field not uncommon 10-20 years later
- Protocols through Children's Oncology Group Soft Tissue Sarcoma Committee
  - Formerly Intergroup RMS Study Group (IRSG)

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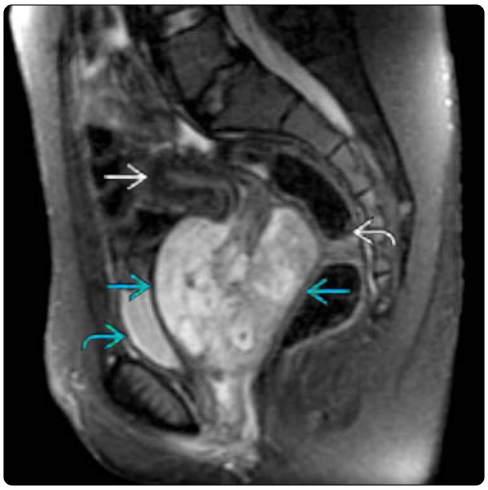
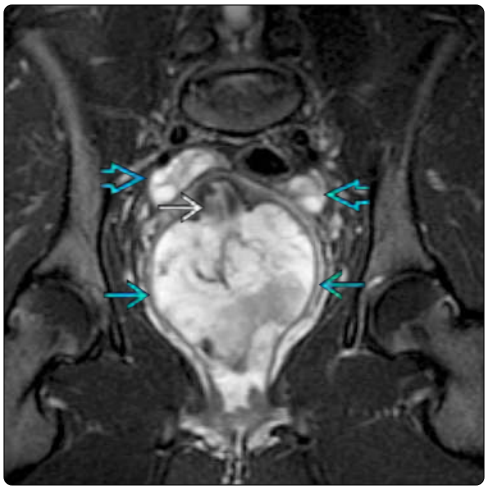
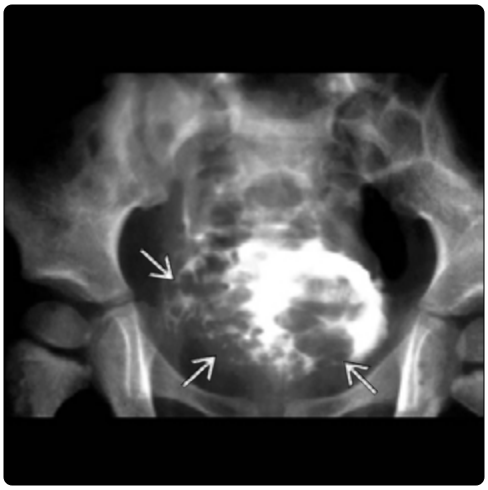
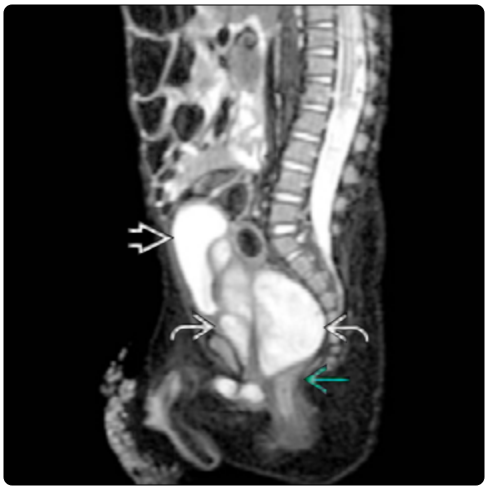


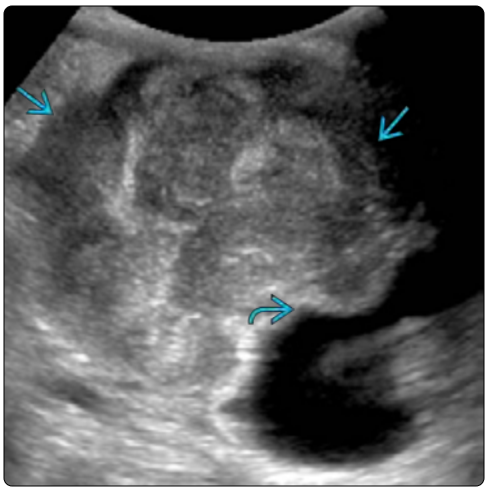


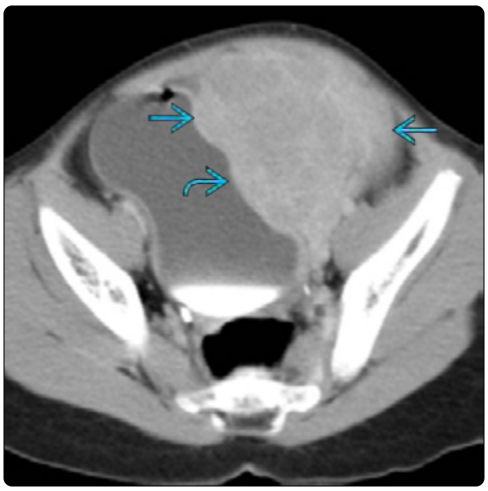
(Left) *Sagittal T2 FS MR in a 14-year-old girl shows a heterogeneous mass filling the vagina. The mass displaces the uterus superiorly, the bladder anteriorly, & the rectum posteriorly. Rhabdomyosarcoma was confirmed on biopsy.* **(Right)** *Coronal T2 FS MR in the same patient shows the vaginal rhabdomyosarcoma displacing the cervix & ovaries superiorly.*





(Left) *Frontal image from an IVP shows clusters of small, round filling defects in the contrast-filled urinary bladder in this patient with sarcoma botryoides, a grape-like protrusion of rhabdomyosarcoma that classically occurs in the urinary bladder, vagina, nasopharynx, or biliary system.* **(Right)** *Sagittal STIR MR in a young boy shows a rhabdomyosarcoma growing up & out of the pelvis between the urinary bladder & rectum. The mass likely originated from the prostate or bladder base.*





(Left) *Longitudinal US in a 4-year-old girl shows a lobulated, heterogeneous mass distorting the anterior wall of the urinary bladder.* **(Right)** *Axial CECT of the pelvis in the same patient shows the heterogeneously enhancing mass involving the lateral wall of the urinary bladder. Biopsy confirmed a rhabdomyosarcoma.*

Neonatal Adrenal Hemorrhage

KEY FACTS

TERMINOLOGY

- Perinatal bleeding into normal adrenal gland
  - Associated with many perinatal stressors: Asphyxia, sepsis, birth trauma, coagulopathies
  - ↑ frequency in full-term & large infants

IMAGING

- Right > left; bilateral in 5-10%
- US: Echogenic avascular mass replacing or expanding newborn adrenal gland
  - Appearance varies with timing of imaging
    - Acute: Hemorrhage appears echogenic & mass-like
    - Subacute: Blood products liquefy & contract, creating mixed echotexture mass
    - Chronic: Adrenal resumes normal size, ± Ca^{2+} or cyst
- CT, MR: Nonenhancing ± rim of enhancing adrenal
  - MR may show high T1 signal intensity &/or "blooming" on GRE, depending on age of blood products
- Radiographs (months to years later): Small unilateral or bilateral adreniform Ca^{2+}

TOP DIFFERENTIAL DIAGNOSES

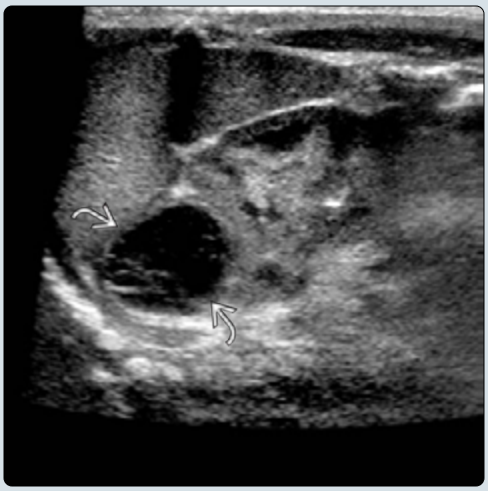
- Neuroblastoma
- Congenital adrenal hyperplasia
- Extralobar bronchopulmonary sequestration

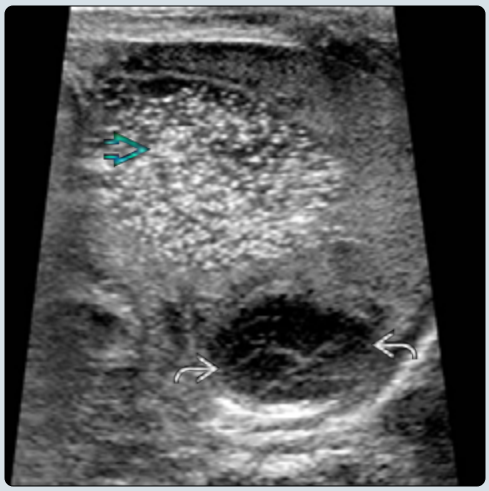
CLINICAL ISSUES

- Newborns may present with anemia, dropping hematocrit, jaundice, palpable mass, or adrenal insufficiency
  - Medical therapy for adrenal insufficiency rarely needed

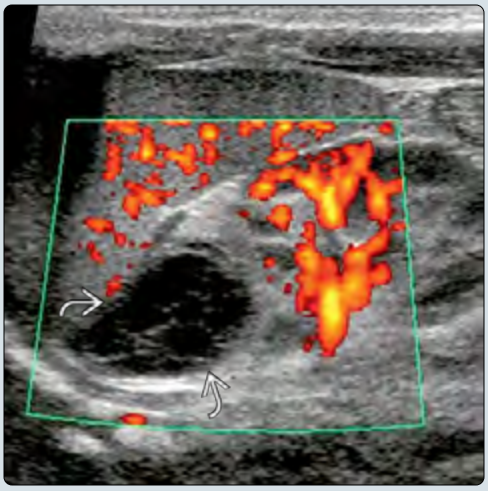
DIAGNOSTIC CHECKLIST

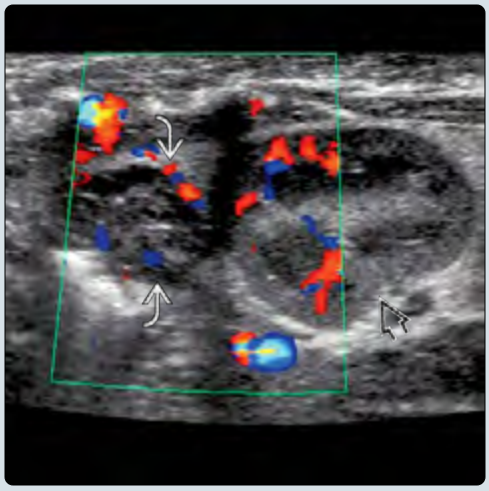
- In neonate: Follow-up US in 2-6 weeks to confirm expected evolution with ↓ size
  - If larger or more solid, work-up for neuroblastoma
- In older child with incidental radiographic paraspinal Ca^{2+}
  - If morphology & extent unclear, start with abdominal ultrasound to exclude neuroblastoma





(Left) *Longitudinal oblique ultrasound shows a complex cystic lesion in the adrenal gland of a neonate. There are many thin internal septations in this case of subacute neonatal adrenal hemorrhage. Adrenal hemorrhages are fairly common in neonates who are stressed by congenital heart disease, surgery, ECMO, sepsis, acidosis, etc.* **(Right)** *Transverse ultrasound of the same lesion shows fine internal septations in this evolving, partially liquefied hemorrhage. Note the tiny gas bubbles in the stomach.*





(Left) *Longitudinal oblique power Doppler ultrasound of the same neonatal adrenal hemorrhage shows absence of blood flow in in the lesion with normal perfusion of the adjacent kidney & spleen.* **(Right)** *Transverse oblique color Doppler ultrasound performed 2 months later shows partial resorption of the same hematoma with improved blood flow in the adrenal gland. Mass effect on the left renal upper pole has resolved.*

TERMINOLOGY

Synonyms

- Adrenal hemorrhage, adrenal cortical hematoma

Definitions

- Perinatal bleeding into normal adrenal gland
- Associated with many perinatal stressors
  - Asphyxia, sepsis, birth trauma, coagulopathies

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous, avascular, well-defined mass replacing or expanding newborn adrenal gland
  - Bleeding seldom extends outside of adrenal
    - Case reports of concurrent scrotal hematoma
- Location
  - Suprarenal; right > left; bilateral in 5-10%

Radiographic Findings

- Radiography
  - May see Ca^{2+} months to years after hemorrhage

CT Findings

- CECT
  - Dense (if acute) or hypodense (if subacute) mass enlarging/splaying/compressing adrenal gland
    - No enhancement of hematoma
    - Residual rim of compressed adrenal enhances
  - May see Ca^{2+} months to years after hemorrhage

MR Findings

- Variable signal intensity depending on age of blood products
  - T1 hyperintensity suggests hemorrhage
  - ± "blooming" of hemosiderin on GRE
  - May restrict diffusion
- Enhancement of residual adrenal surrounding nonenhancing hematoma

Ultrasonographic Findings

- Grayscale ultrasound
  - Imaging appearance varies with timing of exam
    - Acute: Hemorrhage appears echogenic & mass-like
      - Acute hemorrhage may be less echogenic in patients on anticoagulants, ECMO, etc.
    - Subacute: Blood products liquefy & contract, creating mixed echotexture mass ± internal septations
    - Chronic: Gland resumes normal size, may calcify
- Color Doppler
  - Avascular hematoma
  - Doppler useful to assess adjacent renal veins
    - Renal vein thrombosis associated with adrenal hemorrhage
      - Causal relationship not established

Nuclear Medicine Findings

- MIBG or PET may show photopenic focus in adrenal
- ± nonspecific flattening of ipsilateral renal upper pole

Imaging Recommendations

- Best imaging tool
  - Ultrasound for initial diagnosis & follow-up
  - MR & MIBG useful if lesion shows internal flow or interval ↑ in size (concerning for neuroblastoma)
    - MR documents characteristic signal intensity of aging blood products
    - MIBG shows no ↑ in uptake
- Protocol advice
  - Doppler ultrasound confirms lack of vascular flow in mass-like hematomas

DIFFERENTIAL DIAGNOSIS

Neuroblastoma

- Most common malignancy in 1st month of life
- More likely to be cystic in neonates than older children
- Ca^{2+} in ~ 85% of all neuroblastomas
- May extend beyond adrenal
  - Paraspinal masses crossing midline, invading spine, encasing vessels
  - Distant metastases to liver, bone, skin
- Doppler shows internal tumor vascularity
  - Can have concurrent avascular hemorrhage
- Many cases of spontaneous regression in congenital neuroblastoma

Congenital Adrenal Hyperplasia

- Bilateral "cerebriform" adrenal enlargement
  - Single limb thickness > 4 mm; length of gland > 20 mm
- Autosomal recessive inheritance
  - 1 in 15,000-20,000 live births
- Various enzyme deficiencies lead to lack of aldosterone & cortisol production
- Low cortisol triggers pituitary to secrete corticotropin → enlarged adrenal gland
- Females: Present at birth with ambiguous genitalia
- Males: Usually present at 5-14 days of life with severe electrolyte abnormalities

Wolman Disease

- Rare autosomal recessive disorder of lipid metabolism
  - ↓ levels of lysosomal acid lipase → bilateral adrenal Ca^{2+}
- Occurs in infancy; historically fatal in most cases before 1 year of age
- Bone marrow transplant & enzyme replacement have changed prognosis
- Affected infants show signs of lipid storage in most tissues, causing
  - Hepatosplenomegaly
  - Abdominal distension
  - Vomiting
  - Steatorrhea
  - Failure to thrive
- Case reports of isolated fetal ascites in Wolman disease

Extralobar Bronchopulmonary Sequestration

- Congenital anomaly of lung formation
  - No connection to tracheobronchial tree
- May be intra- or subdiaphragmatic above kidney
- Homogeneous solid mass ± systemic vessel feeding

- Cystic components with "hybrid" CPAM lesion

Adrenal Cysts

- Well-defined round anechoic avascular mass
- Associated with Beckwith-Wiedemann syndrome
  - May have hypertrophied adrenal

Lymphatic Malformation

- Multicystic congenital mass crossing soft tissue planes
- May have suprarenal components

PATHOLOGY

General Features

- Etiology
  - Several proposed mechanisms
    - Fetal compression during birth causes ↑ venous pressure & rupture of small venules
    - Transient hypoxia or hypotension causes hemorrhage
    - Normal involution of fetal cortex & vacuolization ↑ tendency for bleeding
- Genetics
  - Not inherited
- Embryology & anatomy
  - Relative size of neonatal adrenal glands large compared to adults
    - Fetal adrenal cortex functions in utero to produce corticosteroids
      - Quite thick at birth, creating convex borders
    - Central medullary portion of gland brightly echogenic in comparison to cortex
    - Fetal cortex & overall size of gland ↓ after birth: Almost inapparent by 6 months

Gross Pathologic & Surgical Features

- Hemorrhage into otherwise normal adrenal
- More common on right side; up to 85% in one study
  - One theoretic risk for greater right-sided incidence: Relatively short right adrenal vein

Microscopic Features

- Ischemic necrosis, supporting theory of involuting fetal cortex as predisposing factor

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Anemia, dropping hematocrit, jaundice, adrenal insufficiency, or palpable mass in perinatal period
- Other signs/symptoms
  - Occasionally discovered incidentally during work-up of
    - Unrelated antenatal hydronephrosis
    - Unrelated abdominal pain in older child
      - Incidental paraspinal Ca^{2+} from remote hemorrhages
- Clinical profile
  - Occurs more often in full-term infants & large for gestational age (LGA) babies

Demographics

- Age
  - Newborn infants
- Epidemiology
  - 1-2 per 1,000 births

Natural History & Prognosis

- Blood products gradually retract & liquefy
- Cysts or dystrophic Ca^{2+} may develop
- Adrenal function typically preserved, especially in unilateral cases
- Prognosis excellent

Treatment

- Observation in most cases
- Adrenal insufficiency extremely rare, requiring damage to > 90% of adrenal tissue
  - Medical therapy may be required transiently
    - Exogenous steroid support of hypotension

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Localized avascular suprarenal mass in neonate
  - Follow-up imaging in 2-6 weeks
  - Growth or lack of liquefaction requires oncology work-up for neuroblastoma
- Incidental radiographic detection of paraspinal adrenal Ca^{2+} in older child
  - If clearly adreniform & not crossing midline, no further work-up required
  - If morphology & extent unclear, start with abdominal ultrasound to exclude neuroblastoma

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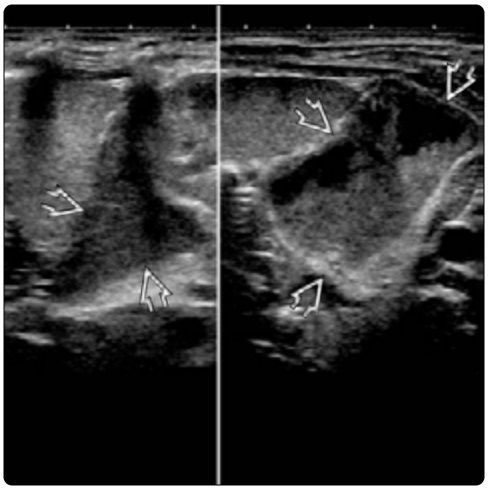
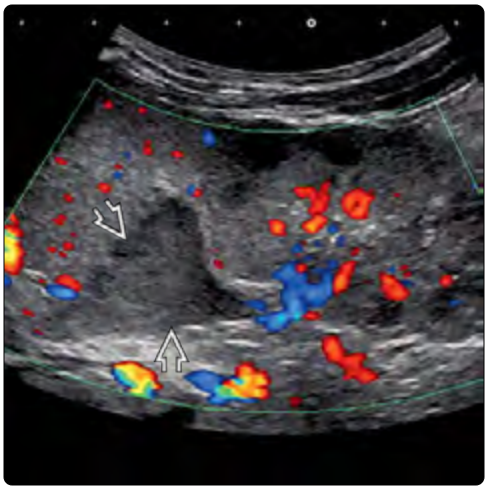
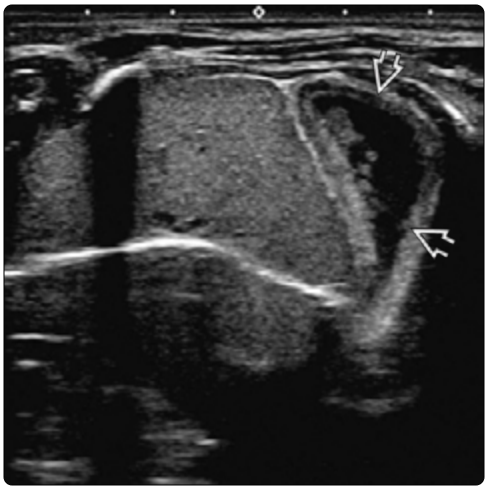
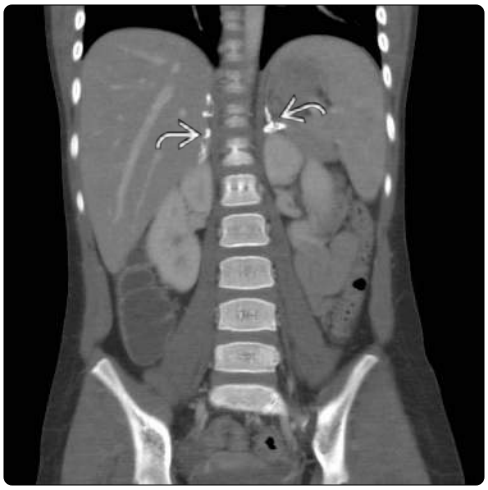


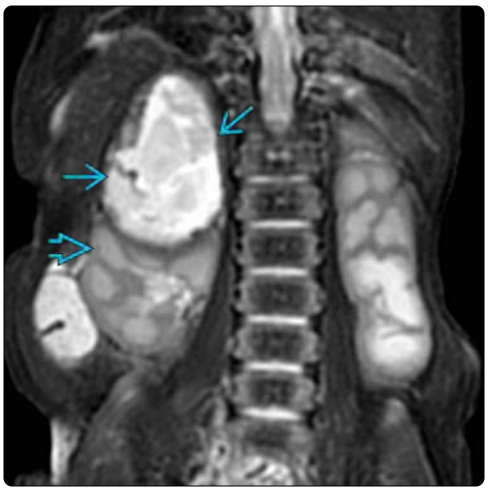


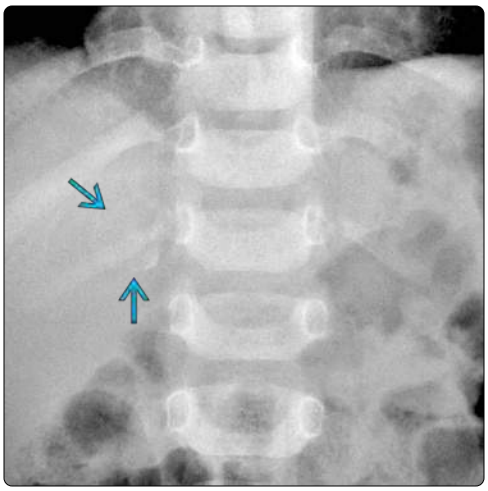
(Left) *Longitudinal (left) & transverse (right) images of the left suprarenal region in a 9-day-old infant with congenital heart disease show a mixed echogenicity lesion with well-defined margins between the spleen & kidney.* **(Right)** *Color Doppler US in the same patient shows absence of blood flow in the lesion, typical of adrenal hemorrhage.*





(Left) *Transverse ultrasound of the same lesion a week later shows progressive liquefaction, consistent with evolving hemorrhage & confirming the benign nature of the "mass."* **(Right)** *Coronal CECT in an elementary school age patient (who had been a premature infant) shows Ca^{2+} in both adrenal regions, likely due to remote neonatal adrenal hemorrhages.*





(Left) *Coronal T2 FS MR in a 20 day old with left prenatal hydronephrosis shows a heterogeneous right suprarenal mass deforming the right kidney. The mass was T1 bright with a few foci of "blooming" on GRE (not shown). Serial ultrasounds showed progressive decrease in size of the mass over the next 5 weeks.* **(Right)** *AP radiograph in the same patient 18 months later shows a small residual suprarenal Ca^{2+}, consistent with a remote adrenal hemorrhage.*

KEY FACTS

TERMINOLOGY

- Inherited disorder of cortisol (± aldosterone) biosynthesis, which leads to hypertrophy of adrenal glands & excess androgen production

IMAGING

- Adrenal glands
  - Bilateral enlargement with cerebriform morphology
  - Usually maintain overall adreniform shape & corticomedullary distinction
    - Hypoechoic cortex & hyperechoic medulla
- Females
  - US used to identify normal female uterus
    - Excess androgen stimulation may make ovaries small & difficult to find on US
  - US shows no testes in masculinized labioscrotal folds
- Males
  - Testicular adrenal rests occur in ~ 95% of adult males with congenital adrenal hyperplasia (CAH)
    - Hypoechoic mass in each testis with ↑ vascularity but no architectural distortion

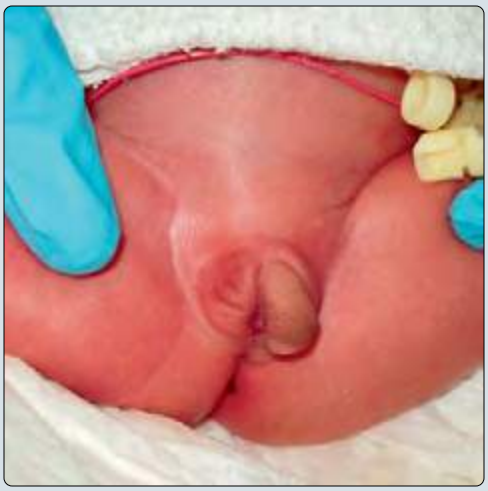
PATHOLOGY

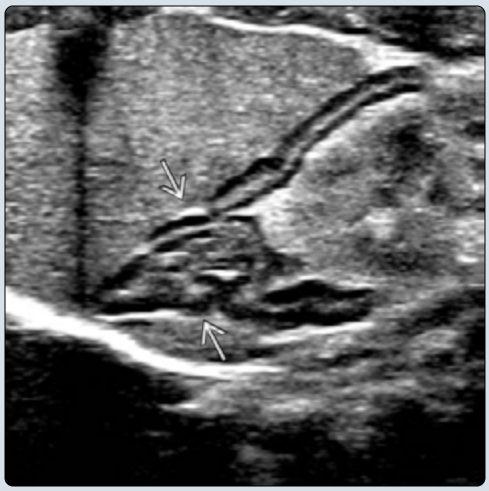
- Autosomal recessive disorder
- Caused by deficiency in 1 of 5 enzymes required to synthesize cortisol from cholesterol in adrenal cortex
  - 21-hydroxylase deficiency most common

CLINICAL ISSUES

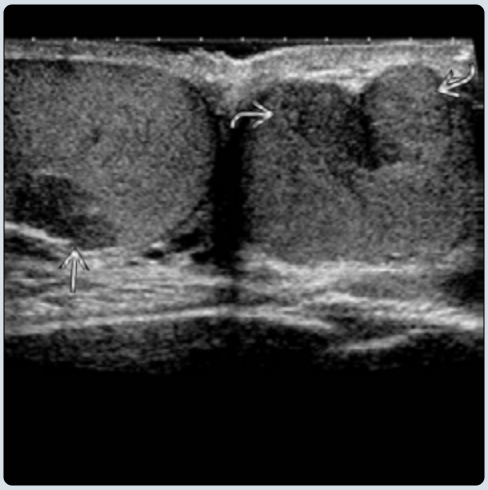
- In most of USA, neonatal screening panels test for severe forms of CAH
- Females: Present at birth with ambiguous genitalia
  - Most common cause of ambiguous genitalia in females
- Males: Usually present at 5-14 days of life with acute salt-wasting crisis
- Treated with lifelong glucocorticoids to replace cortisol & suppress androgen production
  - ± mineralocorticoid & salt supplementation if needed

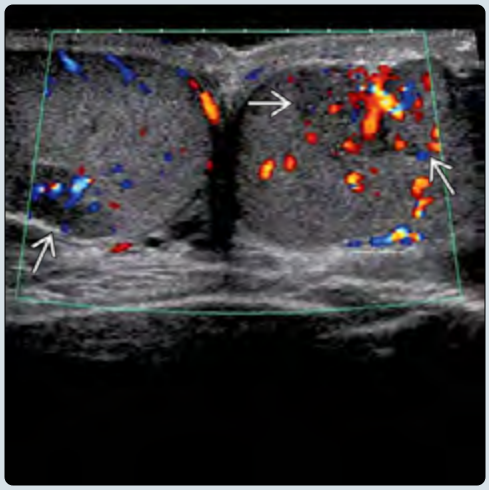
(Left) *Photograph of the perineum shows ambiguous genitalia in a genetic female (XX) with congenital adrenal hyperplasia (CAH). There is clitoromegaly & masculinization of the labioscrotal folds.* **(Right)** *Longitudinal US of the right adrenal gland shows the wrinkled, cerebriform morphology typical of CAH.*





(Left) *Transverse US of the scrotum in a patient with CAH shows round masses in both testes, consistent with congenital testicular adrenal rests. The lesion in the right testis is smaller & more uniformly hypoechoic compared to the lesion in the left testis.* **(Right)** *Transverse color Doppler US of the testes in the same patient shows hyperemia of the testicular adrenal rests, a typical finding in CAH. The vessels are not distorted as they pass through the lesions (as would be expected with testicular neoplasms).*





TERMINOLOGY

Abbreviations

- Congenital adrenal hyperplasia (CAH), 21-hydroxylase deficiency (21-OHD)

Definitions

- Inherited disorder of cortisol (± aldosterone) biosynthesis, which leads to hypertrophy of adrenal glands

IMAGING

General Features

- Best diagnostic clue
  - Enlarged, wrinkled appearance of adrenal glands
- Morphology
  - Markedly enlarged adrenal glands folded back upon themselves, giving characteristic wrinkled or cerebriform appearance

Ultrasonographic Findings

- Adrenal glands
  - Diffuse enlargement of both adrenal glands with wrinkled, cerebriform appearance
    - Single limb of adrenal gland ≥ 4 mm
    - Cerebriform appearance caused by hypertrophied adrenal gland folding back upon itself
      - Hypoechoic cortex & hyperechoic medulla usually maintained
      - Creates appearance similar to cerebral gyri & sulci
  - Left adrenal gland usually larger than right
    - Potential space for right adrenal gland smaller due to liver
  - Normal adrenal glands do not exclude CAH
- Kidneys may show pelvocaliectasis, duplex collecting systems, scarring
- Female reproductive organs
  - Females with CAH present with ambiguous genitalia/pseudohermaphroditism
  - US used to identify normal female uterus
    - Excess androgen stimulation may make ovaries small & difficult to find on US
  - US confirms absence of testes in masculinized labioscrotal folds
  - Hydrocolpos or hydrometrocolpos may be present
  - In postpubertal females, ovaries may be enlarged with polycystic appearance
  - ± ovarian adrenal rest tumors appearing as small, hypoechoic nodules
- Male reproductive organs
  - Normal appearance of testes at birth
  - Testicular adrenal rests occur in ~ 95% of adolescent & adult males with CAH
    - Ovoid hypoechoic foci within testes without associated architectural distortion
    - Echogenicity varies by size
      - Small lesions (< 2 cm): Hypoechoic lesions surrounding echogenic mediastinum testis
      - Larger lesions (> 2 cm): Heterogeneous or slightly hyperechoic lesions that may extend beyond mediastinum testis
    - Lesions show ↑ in vascularity on color Doppler

Radiographic Findings

- If untreated or undertreated, can have advanced bone age

Fluoroscopic Findings

- Voiding cystourethrogram
  - High prevalence of upper tract genitourinary malformations in girls, including vesicoureteral reflux, hydronephrosis, & duplex collecting systems
- Genitography can be performed with ambiguous genitalia prior to corrective surgery
  - Females with CAH & ambiguous genitalia may have common orifice for urethra & vagina
  - Genitography shows urethra, external sphincter, presence or absence of vagina, urethrovaginal confluence, & cervix

CT Findings

- Bilateral, symmetric enlargement of adrenal glands
  - Usually maintains normal adreniform shape
  - Occasionally, adrenal gland has ovoid mass-like configuration
- In older patients, high prevalence of adrenal nodules

MR Findings

- Used to evaluate female reproductive organs in setting of ambiguous genitalia
- Adrenal nodules seen in older patients
- Testicular adrenal rests
  - T1: Isointense to normal testicle
  - T2: Hypointense to testicle with well-defined margins
  - T1 + C: Homogeneous enhancement with well-defined margins
- MR used to evaluate brain during acute adrenal crisis
  - May help to detect changes related to acute encephalopathy vs. stroke
  - MR also used to detect chronic changes, including white matter abnormalities, smaller amygdala, & temporal lobe atrophy

Imaging Recommendations

- Best imaging tool
  - US for patients with ambiguous genitalia or salt-wasting crisis
    - US can confirm presence of uterus & absence of testes
    - US confirms cerebriform appearance of enlarged adrenal glands
  - MR can be used for problem solving & to image brain during adrenal crisis

DIFFERENTIAL DIAGNOSIS

Normal Neonatal Adrenal Gland

- Normally enlarged due to hyperplasia of adrenal cortex

Neonatal Adrenal Hemorrhage

- Variably echogenic, heterogeneous, or cystic avascular mass due to perinatal bleeding into adrenal gland from stress

Neuroblastoma

- Most common adrenal mass in children
- Solid, heterogeneous, lobulated mass that often calcifies, encases & displaces vessels, & crosses midline
- Internal vascularity variable

Beckwith-Wiedemann Syndrome

- Disorder characterized by omphalocele, macroglossia, & hemihypertrophy
- ± unilateral adrenal enlargement with normal configuration

Wolman Disease

- Rare, autosomal recessive lipid storage disorder
- Markedly enlarged adrenal glands with dystrophic Ca^{2+}

PATHOLOGY

General Features

- Etiology
  - Caused by deficiency in 1 of 5 enzymes required to synthesize cortisol from cholesterol in adrenal cortex
    - Deficiency of 21-hydroxylase most common cause of CAH
      - Accounts for 90-95% of cases of CAH
      - Caused by mutation in *CYP21A2* gene
      - Aldosterone & cortisol cannot be produced
      - Adrenal gland chronically stimulated, leading to enlargement
    - Deficiency of 11β-hydroxylase next most common enzyme affected
- Genetics
  - Autosomal recessive disorder

Gross Pathologic & Surgical Features

- Enlarged adrenal gland appears folded on itself
- Adrenal gland weighs 2-4x more than normal
- Glands darker than normal due to depletion of lipid-rich cells of zona fasciculata

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - CAH part of normal neonatal screen
  - 3 main types: Salt wasting, simple virilizing, & nonclassical or nonsalt wasting
    - Males: Usually present at 5-14 days of life with acute salt-wasting crisis
      - Vomiting, weight loss, lethargy, severe dehydration, electrolyte imbalance, hypoglycemia, hypovolemia, & anion gap acidosis
      - May initially be misdiagnosed as hypertrophic pyloric stenosis
      - May present with precocious puberty in nonsalt-wasting type
    - Females: Present at birth with ambiguous genitalia
      - Clitoromegaly, posterior fusion of labia majora, & masculinization of labioscrotal folds
      - May mimic penis with varying degree of hypospadias

Demographics

- Epidemiology
  - Occurs in 1 in 15,000-20,000 live births
  - Most common cause of ambiguous genitalia in females

Natural History & Prognosis

- Adrenal crisis most common cause of early death
- Quality of life scores ↓ compared to population
  - Major factor related to ↓ quality of life: Sexual function & ↓ fertility
- High prevalence of adrenal tumors in adults (73-83% of patients)
  - Most common: Adenomas & myelolipomas

Treatment

- Treated with lifelong glucocorticoids to suppress secretion of corticotropin-releasing hormone + ACTH as well as to reduce androgen levels
- Patients require hydrocortisone in times of stress, such as illness or surgery

DIAGNOSTIC CHECKLIST

Consider

- CAH in patients with symmetrically enlarged, wrinkled adrenal glands

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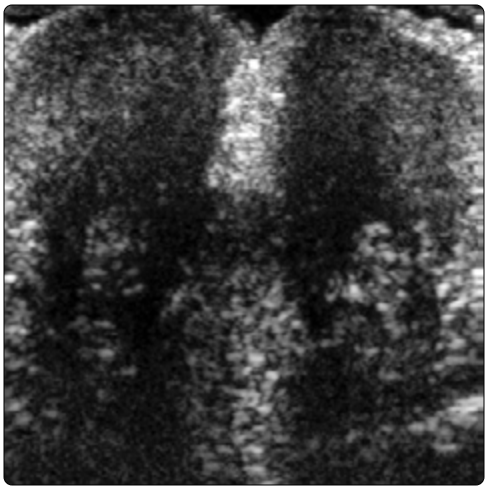
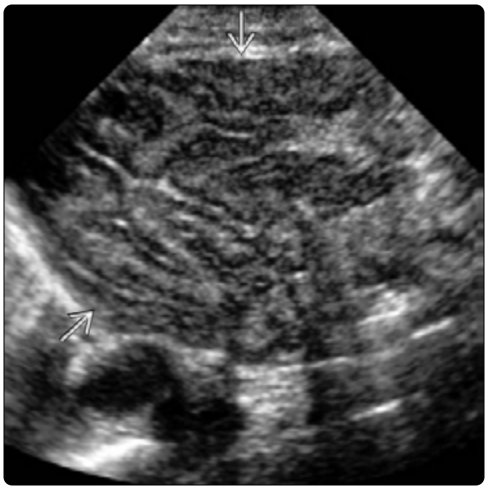
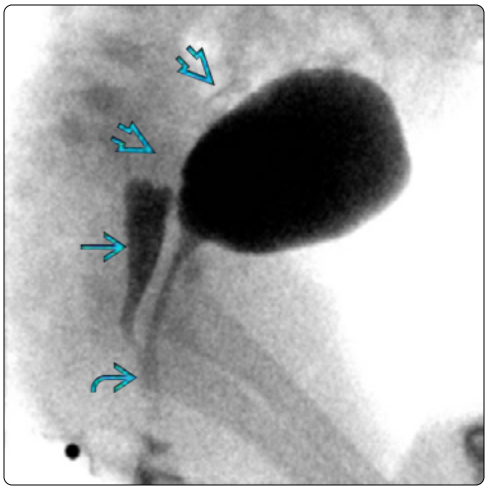
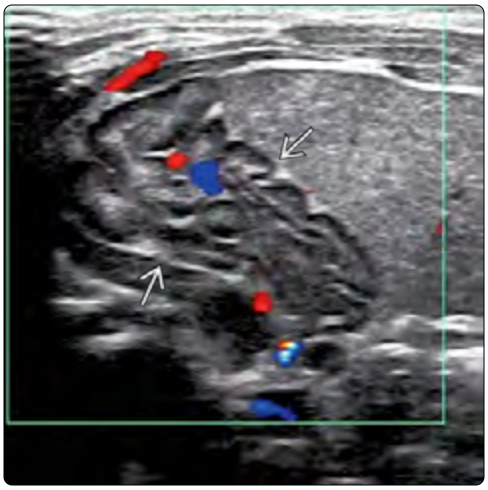


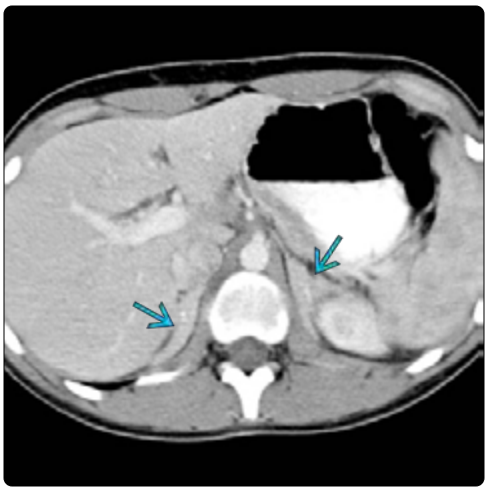


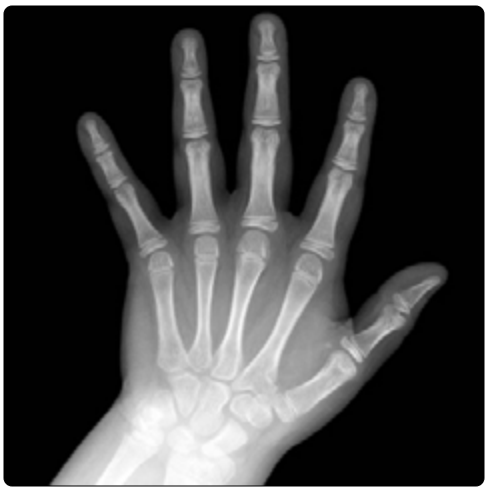
(Left) *Transverse US of the labioscrotal folds in a patient with ambiguous genitalia shows no testes. Imaging of the pelvis demonstrated a normal uterus (not shown). CAH is the most common cause of ambiguous genitalia in females.* **(Right)** *Longitudinal oblique US shows a very enlarged adrenal gland with a characteristic wrinkled or cerebriform appearance ➡. The adrenal gland has lost its normal adreniform shape but maintains corticomedullary distinction.*





(Left) *Lateral voiding cystourethrogram in a 1-week-old girl with CAH & ambiguous genitalia shows a genitourinary sinus ⇨ with reflux of voided contrast into the vagina ⇨ & uterus ⇨.* **(Right)** *Color Doppler US of the left adrenal gland in a patient with CAH shows an enlarged gland ➡ with a cerebriform appearance & mild hyperemia.*





(Left) *Axial CECT (performed after a motor vehicle accident) in a 14-year-old girl with CAH shows mild persistent enlargement & homogeneous enhancement of the bilateral adrenal glands ⇨.* **(Right)** *PA bone age radiograph of the left hand & wrist in an 8-year-old boy with CAH shows an advanced bone age of 13 years.*

Neuroblastoma, Adrenal/Retroperitoneal

KEY FACTS

TERMINOLOGY

- Malignant tumor of sympathetic chain primitive neural crest cells
- Increasing degrees of cellular differentiation/benignity along spectrum: Neuroblastoma (malignant) → ganglioneuroblastoma → ganglioneuroma (benign)

IMAGING

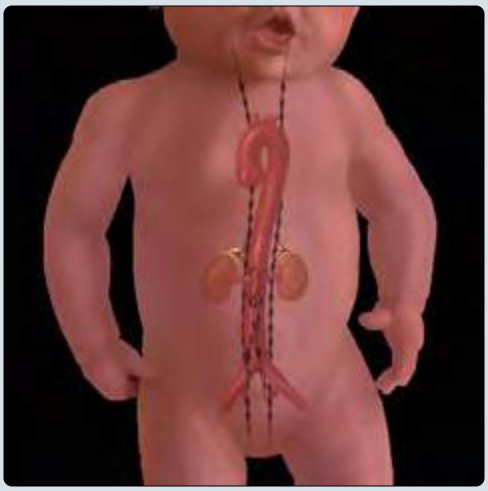
- Location
  - Adrenal (35-48%)
  - Extraadrenal retroperitoneum (25-35%)
  - Posterior mediastinum (16-20%)
- Small round solitary mass vs. large multilobulated lesion
- Aggressive tumor with tendency to invade adjacent tissues
- Frequently engulfs & displaces adjacent vascular structures (rather than just displacing)
- Ca^{2+} in up to 90% by CT
- Metastases in 50-60% at diagnosis, most commonly to bone, lymph nodes, liver, soft tissues

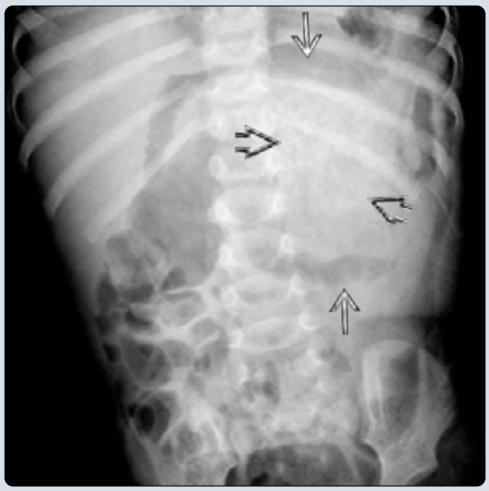
TOP DIFFERENTIAL DIAGNOSES

- Wilms tumor
- Neonatal adrenal hemorrhage
- Less common adrenal tumors
- Other cystic/solid suprarenal lesions

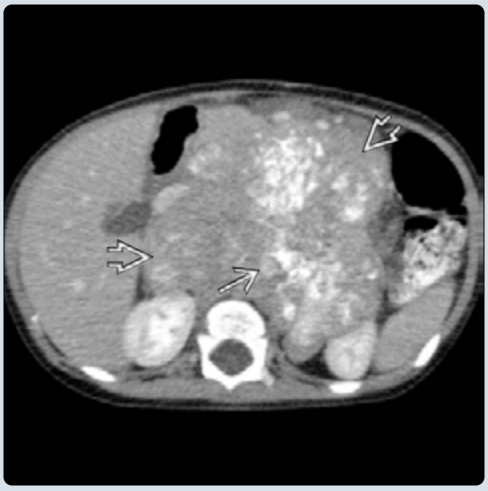
CLINICAL ISSUES

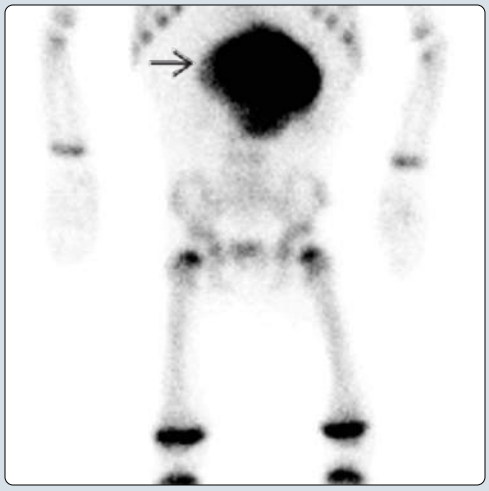
- Most common extracranial solid malignancy in children
- Median age at diagnosis: 15-17 months
- Wide variety of clinical presentations; most commonly presents as palpable abdominal mass
- Features associated with better prognosis
  - Age at diagnosis < 18 months
  - Stage 4S/MS
  - Localized tumor not involving vital structures
  - Absent *MYCN* (*N-myc*) oncogene amplification





(Left) *This graphic shows the anatomic extent of the sympathetic chain ganglia (including the adrenal glands) from the cervical region to the pelvis. Neuroblastoma (NBL) can arise anywhere along the sympathetic chain.* **(Right)** *Supine AP abdominal radiograph in a 1-year-old boy with a palpable abdominal mass shows displacement of bowel loops by a large, heterogeneously calcified mass in the left abdomen.*





(Left) *Axial CECT in the same patient shows the large, lobulated, calcified mass crossing the midline. The mass encases & lifts the aorta off of the spine. These features are typical of NBL.* **(Right)** *Anterior Tc-99m nuclear medicine bone scan in the same patient shows radiotracer uptake throughout the heavily calcified abdominal mass. No cortical bone metastases were detected in this study. Note the intense (but normal) radiotracer uptake at the primary growth centers of the visualized bones.*

TERMINOLOGY

Definitions

- Malignant tumor of sympathetic chain primitive neural crest cells
- Increasing degrees of cellular differentiation/benignity along spectrum: Neuroblastoma [(NBL), malignant] → ganglioneuroblastoma (GNBL) → ganglioneuroma [(GN), benign]

IMAGING

General Features

- Best diagnostic clue
  - Partially calcified, lobulated suprarenal/paraspinal mass in infant
- Location
  - Anywhere along sympathetic chain from neck to pelvis
    - Adrenal (35-48%)
      - 90% adrenal in prenatally detected cases
    - Extraadrenal retroperitoneum (25-35%)
    - Posterior mediastinum (16-20%)
    - Pelvis (2-3%)
    - Neck (1-5%)
    - Metastatic disease with no primary identified (1%)
- General imaging features
  - Small, round, solitary suprarenal/paraspinal mass vs. large, lobulated lesion crossing midline
  - Aggressive tumor, may invade adjacent tissues
    - Intraspinal invasion via neural foramina
    - Kidney, muscle
  - Frequently engulfs & displaces adjacent vascular structures (rather than just displacing/compressing)
  - Ca^{2+} in up to 90% by CT
  - Metastases in 50-60% at diagnosis, most commonly to bone, lymph nodes, liver, soft tissues
    - Liver: Well-defined focal vs. extensive poorly defined lesions with hepatomegaly
    - Bone: Focally destructive cortical &/or well-defined or confluent intramedullary lesions
    - Soft tissues: Cutaneous/subcutaneous lesions may be visible on physical exam & imaging

Radiographic Findings

- Often occult or subtle by radiographs
  - Ca^{2+} in only 30% by radiography
- Displacement of bowel by soft tissue mass
- Widening of inferior thoracic paraspinal soft tissues
  - May be only finding of retrocrural extension of upper abdominal mass
- Subtle bony clues of soft tissue mass
  - Splaying/remodeling of adjacent ribs
  - Vertebral body scalloping, small pedicle
- Bone metastasis
  - May be extensive with little radiographic presence (especially marrow disease)
  - May be only presenting clinical/imaging finding

CT Findings

- Mass often heterogeneous from necrosis, hemorrhage
  - Ca^{2+} in up to 90% by CT

MR Findings

- Generally intermediate to high T2/intermediate to low T1 signal intensity
  - Heterogeneity from Ca^{2+}, hemorrhage, necrosis
- Variable enhancement from none to intermediate
- Excellent depiction of intraspinal extension
- High sensitivity/specificity for marrow disease (but ↓ specificity posttherapy)

Ultrasonographic Findings

- Grayscale ultrasound
  - Mass often heterogeneous
    - Ca^{2+} causing echogenic foci ± posterior acoustic shadowing
  - Suprarenal location displaces/distorts kidney
- Color Doppler
  - Variable internal tumor vascularity
  - Vessels engulfed/lifted by tumor

Nuclear Medicine Findings

- Bone scan
  - Tc-99m MDP
  - ↑ uptake in bony metastasis (cortical > marrow)
  - May be useful in follow-up of non-MIBG avid NBL
  - Uptake in primary mass in up to 74% of cases
- PET
  - 18-FDG remains primary radiotracer
  - High sensitivity for soft tissue & bony NBL, though generally less than MIBG
    - Select populations may benefit from PET, particularly non-MIBG avid disease
- MIBG scintigraphy
  - I-123 MIBG for diagnosis, staging, follow-up imaging
  - MIBG related to norepinephrine, therefore avid uptake in catecholamine production process
  - ↑ uptake at any site of active NBL
    - Sensitivity, specificity ~ 90% in NBL
  - Evaluates bony cortical & marrow disease
  - In MIBG-avid tumors, posttherapy evaluation more specific than MR or FDG-PET

Imaging Recommendations

- Best imaging tool
  - US excellent 1st-line modality for palpable abdominal mass in child
  - MR increasingly used over CT for characterizing tumor & defining extent at diagnosis & follow-up
  - MIBG remains favored nuclear medicine study for diagnosis, staging, follow-up

DIFFERENTIAL DIAGNOSIS

Wilms Tumor

- Mean age: 3 years
- Ca^{2+} uncommon
- Grows like ball, displacing vessels
- Arises from kidney: Claw sign of residual renal parenchyma along tumor
- Lung metastases in 20%
- Invasion of renal vein + inferior vena cava

Neonatal Adrenal Hemorrhage

- Cystic &/or solid-appearing avascular suprarenal mass
- Serial US show gradual ↓ in size with ↑ Ca^{2+}

Less Common Adrenal Tumors

- Pheochromocytoma (uncommon in young children)
- Adrenal cortical neoplasms (usually hormonally active)

Other Cystic/Solid Suprarenal Lesions

- Congenital adrenal hyperplasia
- Extralobar bronchopulmonary sequestration
- Foregut duplication cyst
- Lymphatic malformation

PATHOLOGY

General Features

- Genetics
  - Increased copies of *MYCN* oncogene (*MYCN* or *N-myc* amplification): Poorer prognosis (even stage MS)
  - Deletion of 1p, 11q alleles: Poorer prognosis
  - DNA index (measure of ploidy): Better prognosis from 1.26-1.76 (near triploid)
  - Only 1-2% of cases familial, often with multiple primary tumors in infants
- Associated abnormalities
  - Neurofibromatosis type 1, Beckwith-Wiedemann syndrome, Hirschsprung disease
  - Most NBL cases occur in children without associations

Staging, Grading, & Classification

- International NBL Staging System
  - Original 1-4S system based on resection, pathology (1988, 1993)
  - Used for risk groups by Children's Oncology Group (COG)
- International NBL Risk Group Staging System
  - More comprehensive, imaging-based system (2009)
  - Utilizes modifying image-defined risk factors
    - L1: Tumor in 1 body compartment, no vital structures involved
    - L2: Tumor in 2 body compartments **or** encasing/invading major structures
    - M: Distant metastases
    - MS: Age < 18 months; metastases confined to skin, liver, bone marrow
      - Bones (including marrow) must be clear by MIBG to qualify for stage 4S/MS (with marrow disease limited to less than 10% involvement by biopsy)
  - Used to stratify as very low, low, intermediate, or high risk in conjunction with age, genetics, histology
    - May replace COG risk stratification

Microscopic Features

- Malignant tumor of primitive neural crest cells
- Composed of neuroblastic cells (small round blue cells) + schwannian stroma cells
  - Shimada classification combines histology + age
- Homer Wright rosettes: Circular orientation of NBL cells around neuropil

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless abdominal mass
- Other signs/symptoms
  - Malaise, irritability, weight loss, limping, opsoclonus-myoclonus, Horner syndrome, cerebellar ataxia, neurologic symptoms related to compression, hypertension, watery diarrhea with hypokalemia
  - Classic presentations
    - Skin metastases: Blueberry muffin syndrome
    - Skull base metastases: "Raccoon eyes"
    - Massive liver metastases: Pepper syndrome
  - 90-95% of NBL patients have elevated levels of catecholamines/metabolites (VMA, HVA) in urine

Demographics

- Age
  - Median age at presentation: 15-17 months
  - 95% diagnosed by 7 years
- Epidemiology
  - Most common extracranial solid malignancy in children
    - Most common malignancy of infancy

Natural History & Prognosis

- COG risk stratification
  - Low risk (30% of all NBL, 70% of neonatal NBL): 5-year survival > 95% with observation (select cases) or surgery
    - Spontaneous regression most likely in
      - Newborns with small adrenal lesions
      - Non-*MYCN* amplified infants with localized disease or asymptomatic 4S/MS disease
  - Intermediate risk (20% of all NBL): 5-year survival > 90% with surgery + chemotherapy
  - High risk (50% of all NBL): 5-year survival of 30-40% with intensive multimodality therapy
    - May include myeloablative therapy with stem cell rescue, biologic agents, I-131 MIBG therapy, especially for refractory/recurrent disease

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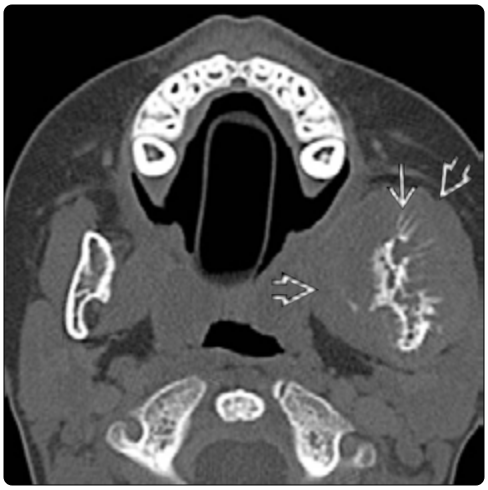
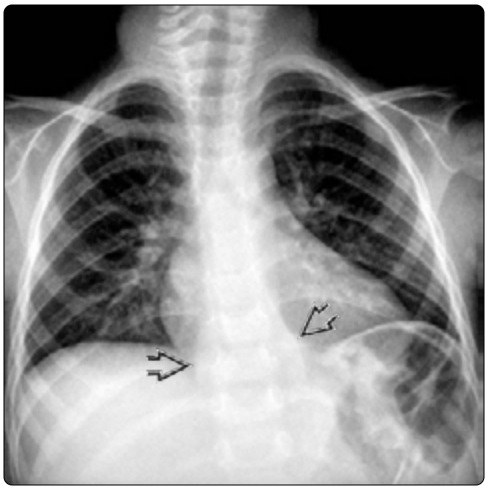
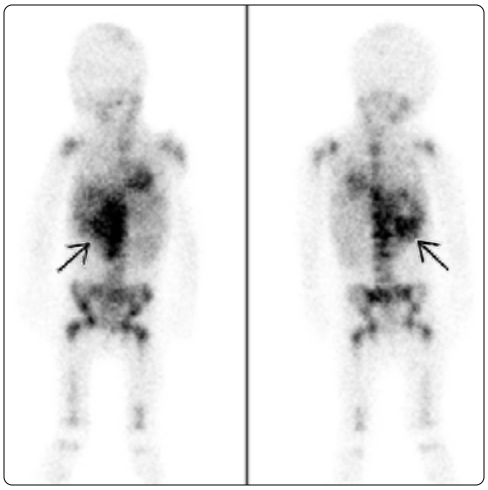
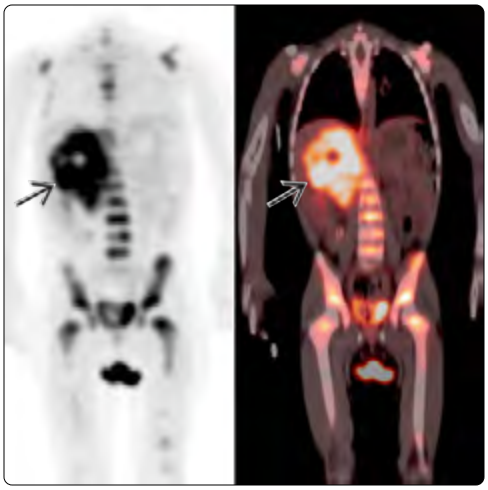


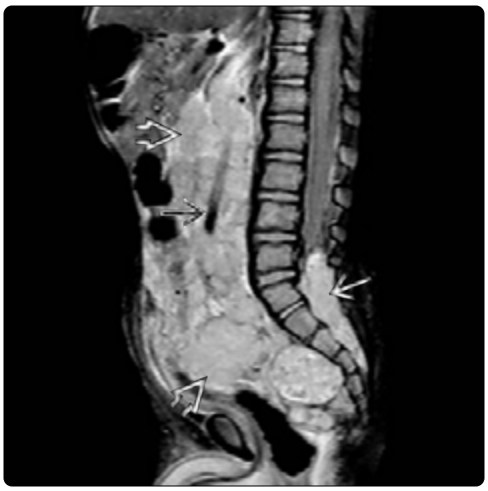


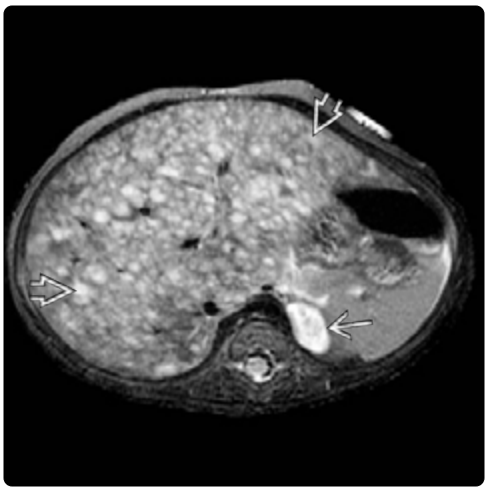
(Left) *Axial bone CT in a 3-year-old boy with a hard left facial mass shows a soft tissue tumor ➔ arising from an abnormal left mandibular ramus. Note the cortical destruction & permeation of the ramus with an aggressive periosteal reaction of radiating perpendicular spicules ➔. This finding is commonly seen in NBL metastases to the skull.* **(Right)** *AP chest radiograph in the same patient shows widening of the lower thoracic paraspinal stripes ⇨, a common finding in NBL due to its sympathetic chain origin.*





(Left) *Anterior & posterior I-123 MIBG scans in a 2-year-old NBL patient show uptake throughout the primary tumor ⇨ in the right abdomen as well as numerous foci of skeletal metastases. Note that the skeleton is not normally visualized with MIBG, making all foci of bony uptake in these images consistent with metastatic disease.* **(Right)** *F-18 FDG-PET & fused PET/CT images in the same patient show increased metabolic activity within the primary tumor ⇨ as well as numerous sites of skeletal disease.*





(Left) *Sagittal STIR MR in a 2-year-old NBL patient shows a large retroperitoneal mass ➔ encasing & displacing the aorta ⇨. Intraspinal extension ➔ is also seen distally.* **(Right)** *Axial T2 FS MR in a 2-month-old boy with hepatomegaly shows innumerable hyperintense lesions ➔ throughout the liver. The hyperintense left suprarenal mass ➔ essentially confirms metastatic NBL, though specific histologic tumor analysis is required for complete determination of diagnosis, staging, & therapy.*

Adrenocortical Carcinoma

KEY FACTS

TERMINOLOGY

- Malignant adrenal cortical neoplasm

IMAGING

- Best clue: Suprarenal mass in young child with virilization or Cushing syndrome
- More common on right side
- Average size: 7-10 cm (median: 9.5 cm)
- Well-defined mass with thin, enhancing rim
- Heterogeneous internal enhancement
- Central stellate focus: Low attenuation (CT) or high signal (T2 FS MR) without enhancement after contrast
- Ca^{2+} in ~ 70% of masses
- May invade IVC
- Sites of metastasis: Lungs, liver, lymph nodes (up to 50%)

TOP DIFFERENTIAL DIAGNOSES

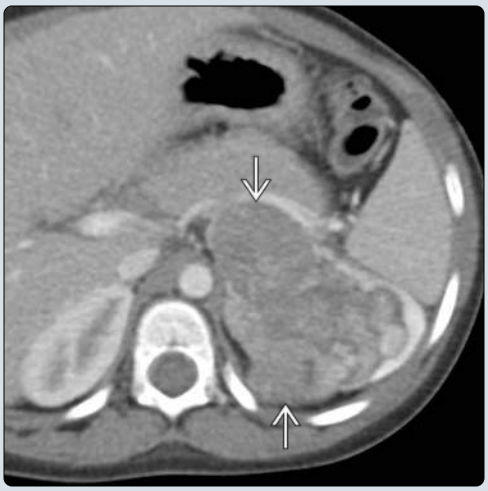
- Neuroblastoma
- Adrenal hemorrhage
- Adrenal adenoma
- Pheochromocytoma

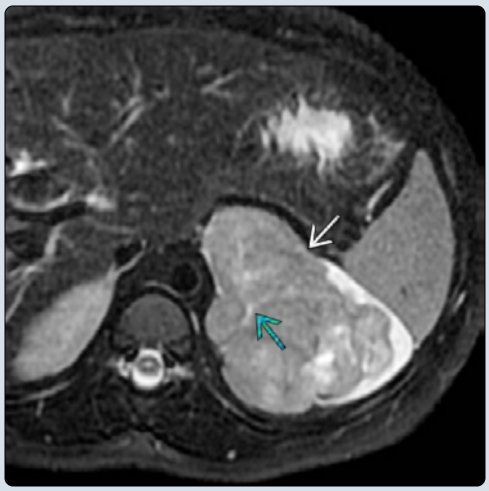
PATHOLOGY

- Associated with Li-Fraumeni & Beckwith-Wiedemann syndromes
- Up to 80% have mutation of *TP53* gene
- Weight of tumor most important predictor of clinical outcome

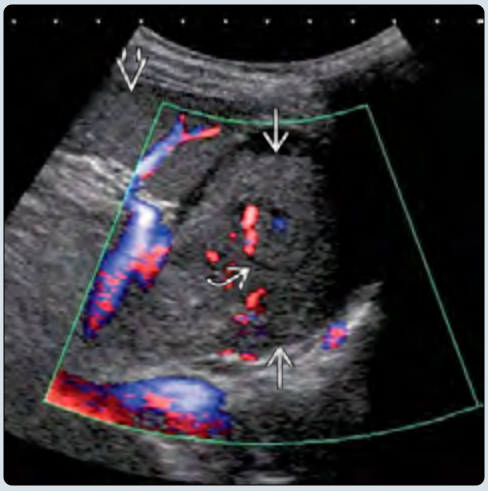
CLINICAL ISSUES

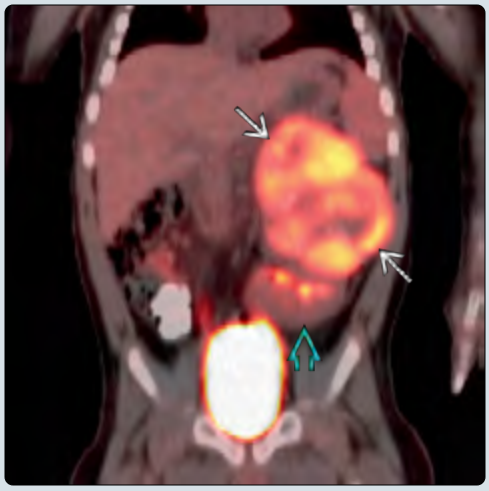
- Median age: 4 years; bimodal distribution in children
  - < 3 years of age vs. 2nd peak in adolescence
- Hormonally active
  - Excess androgen production with virilization present in 80-95% of patients
  - Cushing syndrome present in up to 75%
- Prognosis better than in adults
- Treatment of choice: Complete surgical excision





(Left) *Axial CECT of the abdomen shows a large mass arising from the left adrenal gland. The mass is typical of adrenocortical carcinoma (ACC) with a heterogeneous enhancement pattern & a thin rim of tissue that enhances more than the central components.* **(Right)** *Axial T2 FS MR of the abdomen in the same patient with ACC shows a heterogeneous mass that is hyperintense to the liver. The tumor has a poorly defined central stellate focus that is hyperintense to the remainder of the tumor.*





(Left) *Longitudinal color Doppler ultrasound in the same patient with ACC shows a large left suprarenal mass that is isoechoic to the adjacent spleen. There is a subtle central stellate focus that is hypoechoic compared to the remainder of the tumor.* **(Right)** *Coronal PET/CT in a toddler with virilization shows intense FDG uptake throughout the majority of a left suprarenal mass that is displacing the left kidney inferiorly. Histology confirmed an ACC.*

TERMINOLOGY

Abbreviations

- Adrenocortical carcinoma (ACC)

Definitions

- Malignant adrenal cortical neoplasm

IMAGING

General Features

- Best diagnostic clue
  - Suprarenal mass in young child with virilization or Cushing syndrome
- Location
  - Adrenal cortex
  - Metastasis to lungs, liver, lymph nodes (in up to 50%)
- Size
  - At presentation: 7-10 cm (median: 9.5 cm)

CT Findings

- Well-defined mass with thin, enhancing rim
- Heterogeneous internal enhancement
  - Central stellate focus of low attenuation common
- Ca^{2+} in ~ 70%

MR Findings

- T1WI
  - Heterogeneous mass, isointense to muscle
- T2WI FS
  - Variable signal intensity: Iso-/hyperintense to fat
  - ± central stellate focus: Hyperintense to fat
- T1WI C+
  - Heterogeneous enhancement pattern
  - Central stellate focus does not enhance

Ultrasonographic Findings

- Slightly hyperechoic suprarenal mass
- Central stellate focus often hypoechoic
- Inferior vena cava (IVC) compression or invasion

DIFFERENTIAL DIAGNOSIS

Neuroblastoma

- Most common pediatric adrenal mass
  - Arises from medulla, not cortex
- No hormonal changes
- Uptake on I-123 MIBG scans

Adrenal Hemorrhage

- Most common in neonatal age group (due to birth stress)
- No internal vascularity
- Regresses with time

Adrenal Adenoma

- Benign cortical neoplasm; uncommon in children
  - ACC 8-9x more likely than adenoma in children
- Pathologic/radiologic spectrum with ACC; malignancy favored with
  - Larger, heterogeneously enhancing tumors with Ca^{2+}
  - FDG avidity on PET/CT

Pheochromocytoma

- Uncommon in younger children
- Typically causes hypertension
- Small, round suprarenal mass; hyperintense on T2 MR
- Uptake on I-123 MIBG scans

PATHOLOGY

General Features

- Associated abnormalities
  - Li-Fraumeni syndrome
    - Autosomal dominant tumor predisposition syndrome due to *TP53* mutation
    - Osteosarcoma, ACC, CNS neoplasms, leukemia, soft tissue sarcomas most common in children
  - Beckwith-Wiedemann syndrome
    - Macroglossia, omphalocele, overgrowth
    - Associated with hepatoblastoma, Wilms tumor, ACC

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Excess androgen production with virilization: 80-95%
- Other signs/symptoms
  - Cushing syndrome: Up to 75%
  - Other symptoms: Abdominal mass, hypertension

Demographics

- Epidemiology
  - 0.2% of all childhood cancers
  - Median age: 4 years; bimodal distribution in children
    - < 3 years of age vs. 2nd peak in adolescence

Natural History & Prognosis

- 5-year survival rates: 30-70%
  - 80-90% survival with complete resection
  - 0-10% survival with metastases
- Factors improving prognosis
  - Age < 4 years
  - Tumor size < 10 cm
  - Tumor weight < 200 g
  - Lack of local tumor extension or distant metastases
  - Negative surgical margins

Treatment

- Complete surgical excision
- Chemotherapy if complete resection not possible

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Pheochromocytoma

KEY FACTS

TERMINOLOGY

- Definition
  - Paraganglioma arising from catecholamine-producing chromaffin cells of adrenal medulla
- Associated with
  - von Hippel-Lindau
  - Multiple endocrine neoplasia syndrome type 2
  - Neurofibromatosis type 1

IMAGING

- Best diagnostic clue: Suprarenal mass in child with hypertension
- CECT: Avidly enhancing adrenal mass
- MR
  - T1WI: Isointense to muscle
  - T2WI: Mildly to moderately hyperintense vs. "light bulb bright"
  - T1WI C+: Avidly enhancing mass
- MIBG: ↑ uptake in tumor

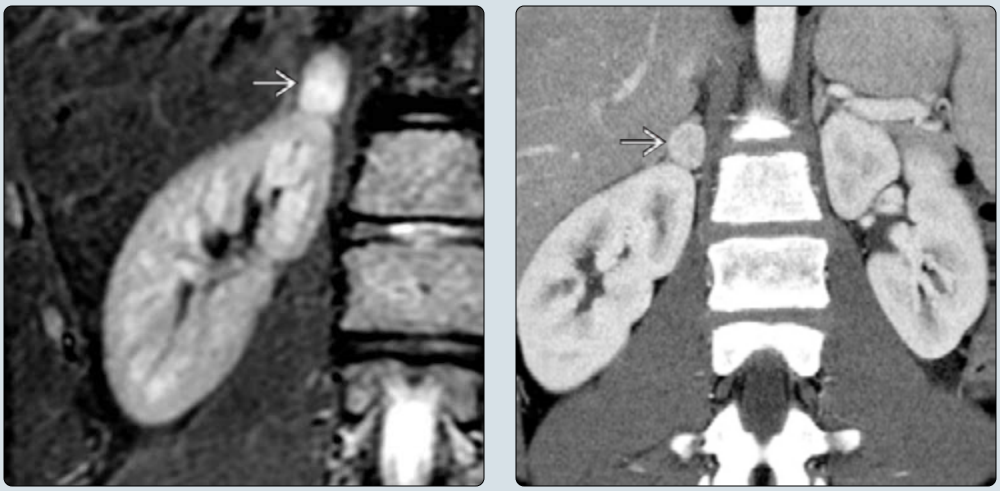
TOP DIFFERENTIAL DIAGNOSES

- Neuroblastoma
- Adrenocortical carcinoma
- Adrenal adenoma

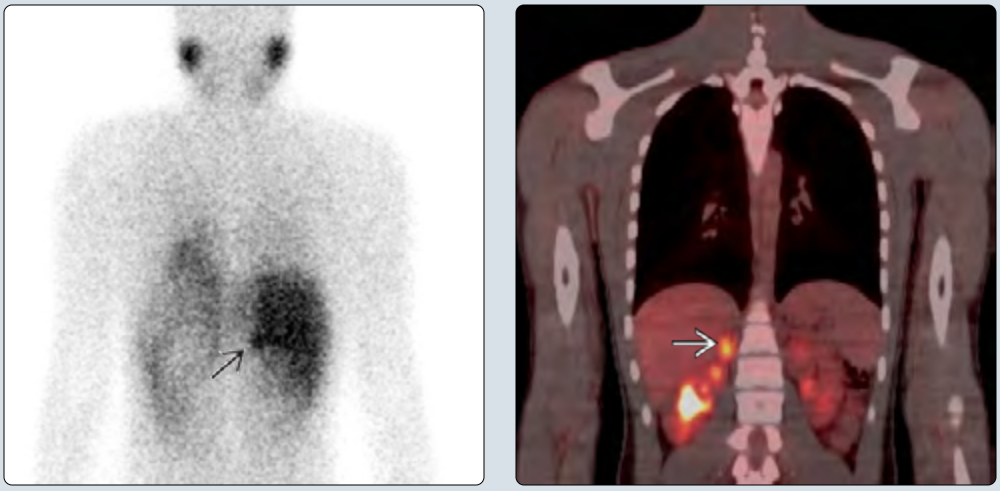
CLINICAL ISSUES

- Responsible for ~ 1% of pediatric hypertension
  - Sustained hypertension present in 63%
  - Headache, sweating, flushing, palpitations, blurred vision, syncope, panic attacks, tremor, & weight loss
- Mean age: 11 years
- Diagnostic test of choice: Plasma or urine metanephrines
- Bilateral tumors in 10-25% of patients
- Malignant in 10-47% of pediatric patients
  - No histologic criteria for malignancy
- Treatment: Surgical excision of primary mass
  - α- & β-blockers to block effect of catecholamines

(Left) *Coronal T2 FS MR of the right kidney in a patient with von Hippel-Lindau syndrome shows a moderately hyperintense right adrenal mass ➡. **(Right)** Coronal CECT in the same patient shows fairly homogeneous enhancement of the small mass ➡ of the right adrenal gland. Premedication is not required when using nonionic contrast agents to image patients with pheochromocytoma or paraganglioma.*



(Left) *Posterior projection MIBG scintigraphy in the same patient shows faint uptake in the right adrenal mass ⇨. MIBG is a specific modality used to identify tumors that manufacture catecholamines. This study will also detect such tumors outside of the adrenal glands (i.e., extraadrenal primary tumors or metastatic disease). **(Right)** Coronal fused PET/CT shows FDG avidity of the right adrenal pheochromocytoma ➡. PET/CT can be useful for staging or following MIBG-negative disease.*



TERMINOLOGY

Definitions

- Paraganglioma arising from catecholamine-producing chromaffin cells of adrenal medulla

Associated Syndromes

- von Hippel-Lindau, multiple endocrine neoplasia syndrome type 2, neurofibromatosis type 1
- Identifiable germline mutation in 59% of patients < 18 years of age & 70% of patients < 10 years of age

IMAGING

General Features

- Best diagnostic clue
  - Suprarenal mass in child with hypertension
- Location
  - Adrenal gland

CT Findings

- NECT
  - Adrenal mass of soft tissue density
  - With size, may become heterogeneous due to intratumoral hemorrhage
- CECT
  - Homogeneously enhancing adrenal mass
  - Nonionic contrast material does not incite hypertensive crisis → premedication not required

MR Findings

- T1WI: Isointense to muscle
  - No signal drop on opposed-phase images
- T2WI: Mildly to moderately hyperintense vs. "light bulb bright"
- T1WI C+: Avid enhancement, prolonged washout
  - May have salt & pepper appearance with enhancing tumor & vascular flow voids
  - Larger lesions more likely heterogeneous

Ultrasonographic Findings

- Suprarenal soft tissue mass
- Iso- to hypoechoic to liver

Nuclear Medicine Findings

- PET/CT
  - Useful in searching for extraadrenal tumors or metastases
  - Less sensitive & specific than MIBG scintigraphy
- MIBG scintigraphy
  - I-123 used in children
  - ↑ uptake in tumor
  - Very specific but less sensitive than CT or MR
  - Useful in searching for extraadrenal tumors or metastases

Imaging Recommendations

- Best imaging tool
  - MR due to lack of ionizing radiation
  - MIBG if MR negative to identify extraadrenal tumor

DIFFERENTIAL DIAGNOSIS

Neuroblastoma

- Most common adrenal tumor in children
- Median age of presentation: 15-17 months
- Often large, multilobulated mass crossing midline; displaces & encases vessels; Ca^{2+} in ~ 85-90%
- MIBG for diagnosis & staging

Adrenocortical Carcinoma

- Large heterogeneous adrenal mass causing virilization or precocious puberty; ± inferior vena cava invasion

Adrenal Adenoma

- Smaller homogeneous mass with intracellular lipid
  - Signal drop-out on opposed-phase MR imaging

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Sustained hypertension present in 63%
  - Symptoms of mass effect in up to 30%
- Other signs/symptoms
  - Headache, sweating, flushing, palpitations, blurred vision, syncope, panic attacks, tremor, & weight loss
  - Paroxysmal episodes: Headaches, palpitations, diaphoresis
  - Complications: Cardiomyopathy, hypertensive crisis, cardiovascular accidents, & seizures
- Laboratory tests
  - Diagnostic test of choice: Plasma or urine metanephrines

Demographics

- Age
  - Mean age: 11 years
- Epidemiology
  - Most common pediatric endocrine tumor
  - Responsible for ~ 1% of pediatric hypertension
  - Bilateral tumors in ~ 10-25% of patients

Natural History & Prognosis

- Malignant in 10-47% of pediatric patients
- No histologic criteria for malignancy
  - Malignancy determined by local tumor invasion or metastases (bones, lungs, liver)

Treatment

- Surgical excision of primary mass
  - Pretreatment with α- & β-blockers required to block effect of catecholamines

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Hydrometrocolpos

KEY FACTS

TERMINOLOGY

- Dilation of vagina ± uterus secondary to distal stenosis, atresia, transverse vaginal septum, or imperforate hymen
  - Prefix: Hydro- meaning fluid, hemato- meaning blood
  - Suffix: -metra meaning uterine cavity
  - Suffix: -metrocolpos meaning uterus & vagina

IMAGING

- Well-defined cystic or debris-filled mass in female pelvis between urinary bladder & rectum
- Vertically oriented in sagittal plane, round in axial plane
- Uterus frequently visible arising from dome of collection; may be normal or mildly distended (much less than vagina)
- US may show trilaminar appearance of vaginal wall, swirling internal debris, lack of internal vascularity
- MR often shows aging blood product signal intensities (↑ T1, ↓ T2); can help discern uterine & vaginal anomalies
- Urinary bladder or ureters may be obstructed
- Check for associated renal anomalies: Unilateral renal agenesis or secondary hydronephrosis
- Primary imaging modality for female GU anomalies: US
- MR used when uterine & complex GU anomalies cannot be clearly defined with US

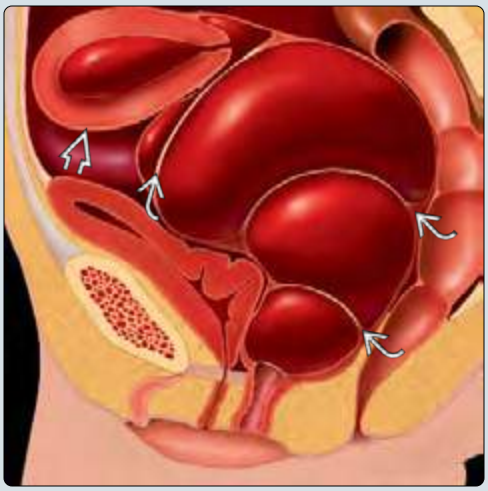
PATHOLOGY

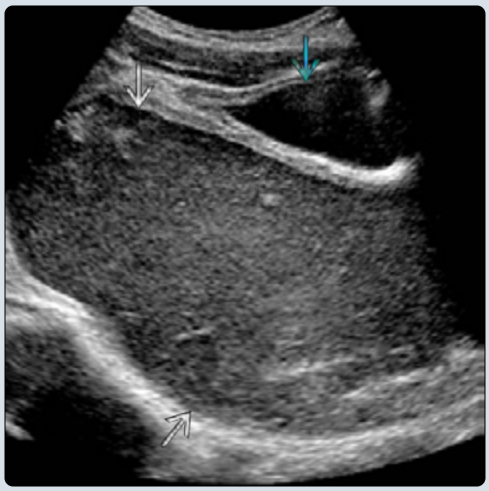
- Imperforate hymen more common cause than vaginal septum
- ± associated anal, renal, vertebral, & cardiac anomalies

CLINICAL ISSUES

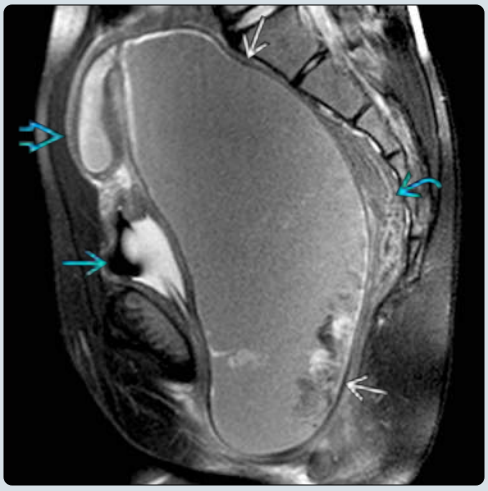
- Bimodal age of presentation
  - Infants (due to maternal hormonal stimulation): Pelvic mass, sepsis, or urinary tract obstruction
    - Vaginal contents more likely mucous, fluid
  - Adolescent girls (pubertal onset): Delayed menarche, cyclic pelvic pain, mass, &/or urinary tract obstruction
    - Vaginal contents more likely blood

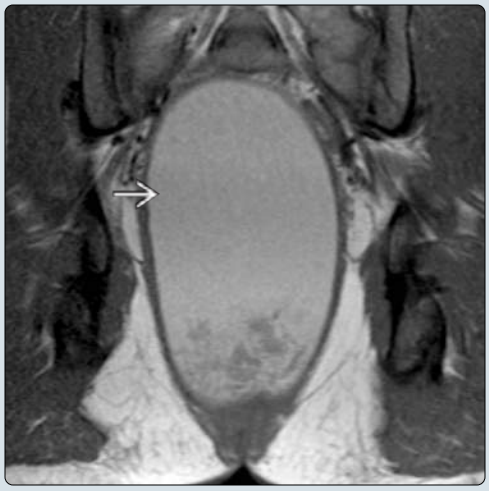
(Left) *Graphic shows potential levels of vaginal septa causing obstruction & hydro-/hematometrocolpos. Note that the vagina distends with trapped secretions & blood to a much greater degree than the uterus.* **(Right)** *Longitudinal US through the pelvis of a 15-year-old girl with urinary retention, cramping, & no history of menses shows a large well-defined, vertically oriented, heterogeneous collection posterior to the urinary bladder, suggestive of hematometrocolpos.*





(Left) *Sagittal T2 FS MR in the same patient shows a markedly distended vagina containing heterogeneous fluid & layering debris. The uterus projects anteriorly & is much less distended than the vagina, findings typical of hematometrocolpos. Note the mass effect on the urinary bladder & rectum.* **(Right)** *Coronal T1 MR in the same patient shows that the fluid in the vagina is predominantly hyperintense, typical of blood products. A vaginal septum was resected, relieving the obstruction.*





TERMINOLOGY

Synonyms

- Hematometrocolpos, hydrometra, hematometra, HMC

Definitions

- Dilation of vagina or vagina & uterus secondary to distal stenosis, atresia, transverse vaginal septum, or imperforate membrane
  - Prefix: Hydro- meaning fluid, hemato- meaning blood
  - Suffix: -metra meaning uterine cavity
  - Suffix: -metrocolpos meaning uterus & vagina

IMAGING

General Features

- Best diagnostic clue
  - Well-defined, cystic or debris-filled, vertically oriented mass in pelvis, between bladder & rectum
  - Can cause secondary bladder outlet obstruction with hydronephrosis
- Location
  - Between bladder & rectum
- Size
  - Variable; can be very large & simulate early pregnancy in teenage girls
- Morphology
  - Vertically oriented on sagittal & coronal images, round on axial images
- Classic imaging appearance: Echogenic debris filling dilated vagina & (to lesser extent) uterus, creating mass effect in pelvis
  - Vagina has elastic walls & can dilate more than uterus
- Can be associated with müllerian duct fusion anomalies, particularly uterus didelphys

Radiographic Findings

- Soft tissue mass in pelvis displacing bowel loops
- Case reports of peritoneal Ca^{2+}, presumably from debris spilling out fallopian tubes

Ultrasonographic Findings

- Grayscale ultrasound
  - Echogenic, layering debris in large well-defined cavity between urinary bladder & rectum
    - Distended vaginal walls may have trilaminar appearance, mimicking "gut signature"
    - Fluid in vagina will typically swirl with mild compression
  - Uterus frequently visible arising from dome of collection
    - Look for variable uterine distention (typically much less than vagina)
    - Look for associated müllerian duct fusion anomalies
  - Urinary bladder & ureters may be dilated from secondary bladder outlet or ureteral compression
  - Also confirm presence of both kidneys, look for secondary hydronephrosis
- Color Doppler
  - Useful to confirm lack of blood flow within debris-filled cavities as complex blood (especially acute) may appear solid on US

CT Findings

- CECT
  - Vertically elongated fluid-filled cavity with enhancing walls originating deep in pelvis
    - Appears round on axial images
  - Displaces bladder, rectum, & small bowel
  - Enhancing uterus extends from cephalad aspect of collection
    - Scrutinize uterus for associated malformations

MR Findings

- MR findings similar to CECT
- Aging blood components may have characteristic signal intensity (↑ T1, ↓ T2)
- Multiplanar imaging useful to optimally profile uterine & cervical anomalies
  - Consider planes oriented to uterus

Other Modality Findings

- Rarely hysterosalpingography or sonohysterography performed in convalescent phase to reevaluate uterine morphology

Imaging Recommendations

- Best imaging tool
  - US best initial imaging modality for suspected GU pathology in female
  - MR used when uterine & complex GU anomalies cannot be clearly defined with US
    - Consider delaying MR exam to convalescent phase, after fluid & debris have been drained

DIFFERENTIAL DIAGNOSIS

Perforated Appendicitis

- Poorly defined inflammatory collection of right lower quadrant
- May visualize appendiceal remnant, appendicolith

Meconium Pseudocyst

- Heterogeneous peritoneal collection (± Ca^{2+}) in newborn, typically secondary to in utero bowel perforation
- Dilated bowel usually still present

Lymphatic Malformation

- Infiltrative multicystic mass with thin septations
- Individual compartments show varying complexity, typically due to hemorrhage

Pelvic Abscess

- Heterogeneous adnexal collection with surrounding inflammation
- Unlikely in newborn
- In adolescents, consider pelvic inflammatory disease

Fallopian Tube Torsion, Cyst, or Obstruction

- Tubular fluid collection with folds

Pelvic Rhabdomyosarcoma

- Sarcoma botryoides protrudes into vagina ("cluster of grapes")

Other Pelvic Masses

- Sacrococcygeal teratoma

- Burkitt lymphoma
- Pelvic neuroblastoma

PATHOLOGY

General Features

- Etiology
  - Embryology-anatomy: Failure of canalization, stenosis, or atresia along lumen
  - In infancy, maternal hormone effects stimulate neonatal uterus & vagina
- Genetics
  - Generally sporadic; not inherited
  - McKusick-Kaufman syndrome
    - Rare multiple autosomal recessive syndrome
      - Hydrometrocolpos
      - Postaxial polydactyly
      - Congenital heart malformation
  - Bardet-Biedl syndrome, a.k.a. Laurence-Moon syndrome
    - Also has hydrometrocolpos & postaxial polydactyly with retinitis pigmentosa, obesity, & learning disability becoming apparent by early school age
    - Inherited as autosomal recessive trait
- Associated abnormalities
  - Most often associated with anal, renal, vertebral, & cardiac anomalies
  - Also associated with intestinal aganglionosis, imperforate anus, urogenital sinus, cloacal anomalies
  - Can be associated with müllerian duct fusion anomalies, particularly uterus didelphys
  - Iatrogenic cases reported due to malposition of artificial urinary sphincter in prepubertal girls
- Site of obstruction can be
  - Imperforate hymen
  - Vaginal stenosis or atresia
  - Cervical stenosis or atresia
  - Mass effect from duplications of uterus & vagina (didelphys)
  - Transverse vaginal septum
    - Most common location: Between middle & upper 1/3 of vagina
    - These patients have functional uteri, though fertility can be compromised
- Secondary urinary obstruction can occur at level of
  - Urethra, ureterovesical junction, ureter

Gross Pathologic & Surgical Features

- Debris contents
  - In fetal life & infancy, contents primarily of cervical mucus (mucocolpos or hydrocolpos)
    - Maternal estrogen stimulates cervical mucus production & causes swelling of labia minora
  - Peripubertal contents typically blood, sloughed endometrial lining, & cervical/vaginal mucus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - In infancy: Presents as pelvic mass, sepsis, or urinary tract obstruction
  - In adolescent girls: Presents as delayed menarche, cyclic pelvic pain, mass, &/or urinary tract obstruction
- Other signs/symptoms
  - Occasionally presents as prolapsing interlabial mass
  - In utero urinary tract obstruction from massive hydrometrocolpos can lead to fetal anuria, oligohydramnios with pulmonary hypoplasia, & renal dysplasia with renal failure in newborn
    - Intrauterine aspiration of obstructed bladder or vagina may be attempted

Demographics

- Age
  - Bimodal age presentation: Infancy or peripubertal
- Gender
  - Females only
- Epidemiology
  - Transverse vaginal septum incidence: 1 in 80,000
  - Imperforate hymen more common
    - Note: Female hymen is homolog of plica colicularis (valve tissue) in males

Natural History & Prognosis

- Immediate prognosis excellent
- Compromised fertility & endometriosis can be long-term complications

Treatment

- Typically drained & septum or stenotic segment excised from inferior approach with minimal tissue resected
- Stenoses & focal atresias may require primary anastomosis & perioperative stenting
- Secondary hydronephrosis typically resolves spontaneously without additional intervention

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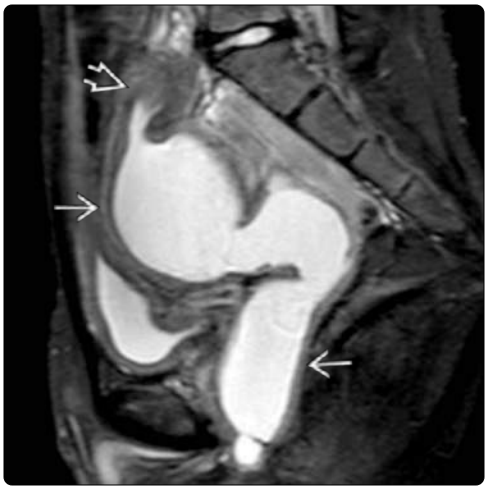
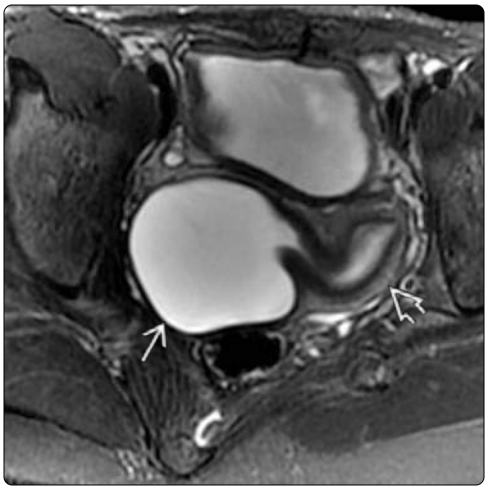
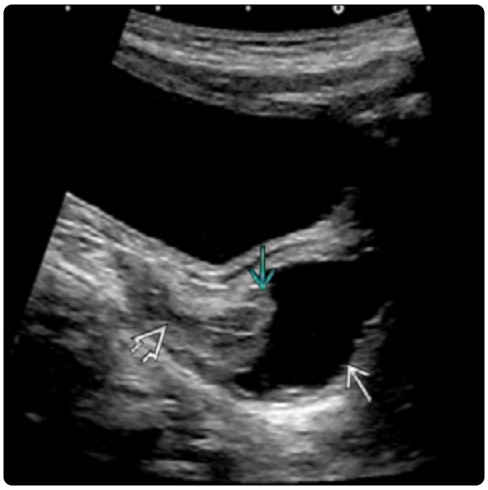
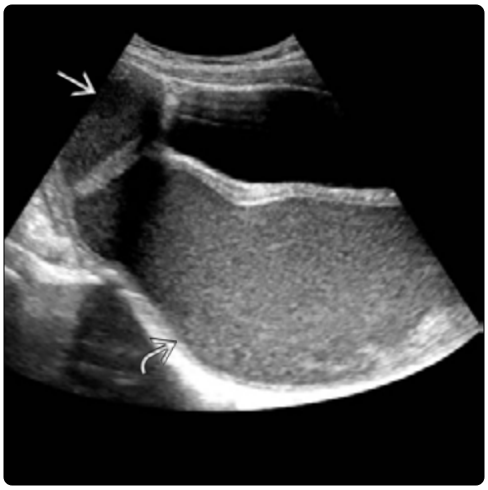


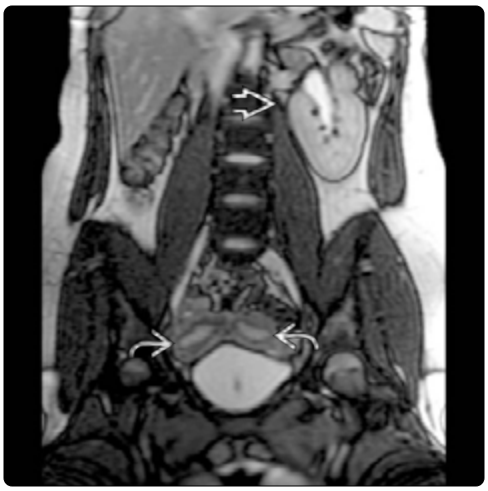


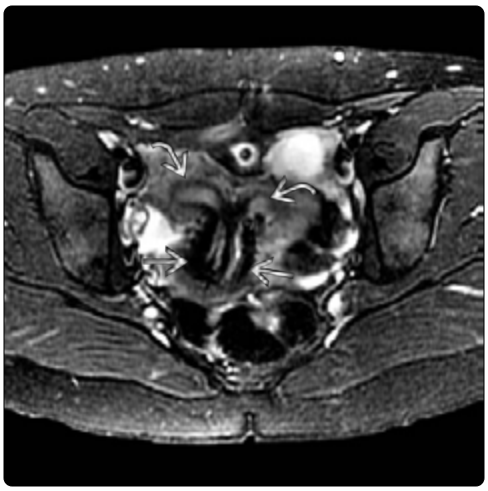
(Left) *Sagittal STIR MR in a 14 year old who had a cloacal repair in infancy shows fluid distending her tortuous neovagina &, to a lesser degree, her endometrial cavity. This was due to a stricture at the introitus.* **(Right)** *Axial T2 FS MR in a 12 year old with pelvic pain shows a very distended vagina with high-signal fluid extending into the uterus, consistent with hydrometrocolpos.*





(Left) *Longitudinal US in a 5 month old with vaginal discharge shows fluid distending the vagina but not the endometrial cavity. This case of hydrocolpos was due to hymen stenosis. Note the prominence of the cervix relative to the fundus, a typical infantile appearance of the uterus.* **(Right)** *Longitudinal oblique US in a case of hematometrocolpos in a teenager with cyclic pain shows marked distention of a debris-filled vagina posterior to the bladder. Note the mildly distended uterus.*





(Left) *Coronal SSFP localizer MR in a patient who had a "vaginal collection" drained at an outside hospital shows 2 endometrial cavities draped over the bladder. A solitary left kidney is noted, a commonly associated abnormality.* **(Right)** *Axial T2 FS MR shows the paired cervices & endometrial cavities in this same patient with uterine didelphys & an obstructing septum of a hemivagina (that led to hematometrocolpos). This combination of findings is typical of Herlyn-Werner-Wunderlich syndrome.*

KEY FACTS

TERMINOLOGY

- Abnormal development, improper fusion, or failure of resorption of müllerian (paramesonephric) duct structures

IMAGING

- Abnormal contour of uterus &/or structural abnormality of endometrial cavity or vagina
- Septate uterus most common müllerian duct anomaly (55%)
- MR best for definitive diagnosis
- Always image kidneys to look for renal anomaly

PATHOLOGY

- MDAs occur in 1 of 3 phases
  - Organogenesis: Agenesis, hypoplasia, unicornuate
  - Fusion: Didelphys, bicornuate
  - Septal resorption: Septate, arcuate
- Renal anomaly present in ~ 30%
  - Renal agenesis most common (~ 2/3)

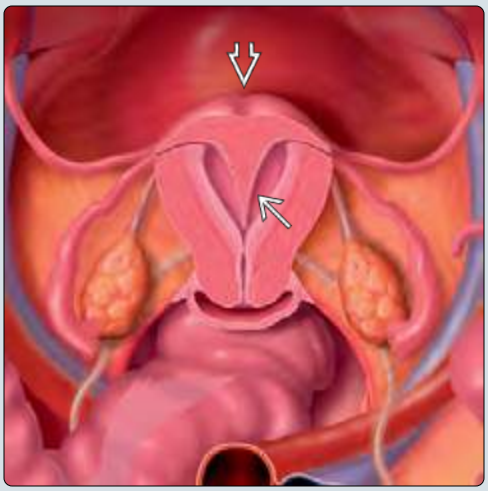
CLINICAL ISSUES

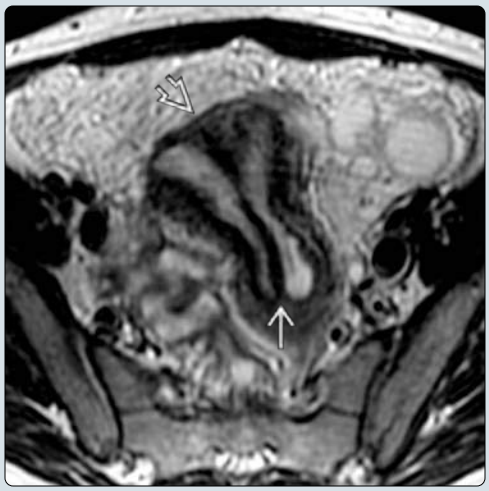
- Asymptomatic
- Symptoms may develop at menarche
  - Primary amenorrhea
  - Dysmenorrhea
  - Cyclic abdominal pain
- Incidence estimated at 1%
- Differentiation of septate from bicornuate uterus important
  - Septate treated with hysteroscopic resection of septum
  - Bicornuate rarely requires surgery
- Treatment
  - Surgery to remove rudimentary horn, resect septum, or relieve obstruction

DIAGNOSTIC CHECKLIST

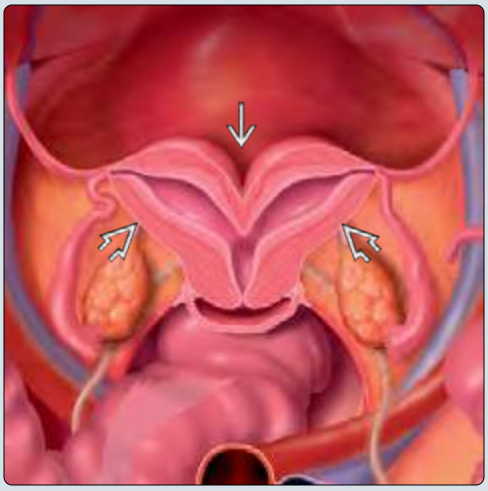
- Outer fundal contour important for differentiating bicornuate from septate uterus

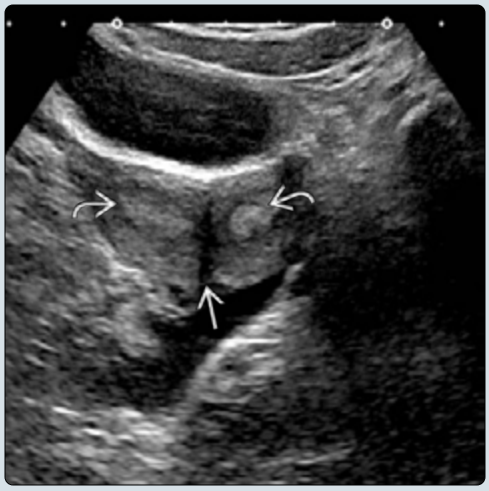
(Left) *Graphic of a septate uterus shows minimal indentation of the uterine fundus ➡. There is myometrium in the superior aspect of the septum, though normal zonal anatomy is not present in this portion ➡.* **(Right)** *Axial T2 MR of a septate uterus shows that the outer fundal contour is smooth ➡ & the intercornual angle is < 75°. There is a low-signal fibrous septum extending along the length of the endometrial cavity ➡ to a single cervix.*





(Left) *Graphic of a bicornuate uterus demonstrates a deep external fundal cleft ➡ & 2 symmetric cornua ➡ that are fused inferiorly.* **(Right)** *Transverse US shows the outer contour/notch ➡ of this bicornuate uterus due to a small amount of free fluid in the pelvis. The 2 endometrial cavities ➡ are separated by myometrium.*





TERMINOLOGY

Abbreviations

- Müllerian duct anomalies (MDA)

Synonyms

- Uterine fusion anomalies

Definitions

- Abnormal development, improper fusion, or failure of resorption of müllerian (paramesonephric) duct structures
  - Müllerian agenesis or hypoplasia (class I in American Fertility Society classification system)
    - Complete or segmental agenesis or variable degrees of uterovaginal hypoplasia
  - Unicornuate uterus (class II)
    - Partial or complete unilateral hypoplasia
  - Uterus didelphys (class III)
    - Duplication of uterus
  - Bicornuate uterus (class IV)
    - Incomplete fusion of superior uterovaginal canal
  - Septate uterus (class V)
    - Incomplete resorption of uterine septum
  - Arcuate uterus (class VI)
    - Near complete resorption of uterine septum

IMAGING

General Features

- Best diagnostic clue
  - Abnormal contour of uterus &/or structural abnormality of endometrial cavity or vagina
- **Müllerian agenesis or hypoplasia**
  - Mayer-Rokitansky-Kuster-Hauser syndrome
    - Complete vaginal agenesis
    - 90% have uterine agenesis
    - 10% have rudimentary uterus
  - ± hydrometrocolpos
- **Unicornuate uterus**
  - 20% of MDAs
  - Single horned uterus
  - Asymmetric curved & elongated (banana-shaped)
  - Hypoplastic uterine horn of variable size usually present
    - May be cavitary or noncavitary
      - Cavity may communicate with contralateral endometrium
  - ± hydrometrocolpos with obstruction
- **Uterus didelphys**
  - 5% of MDAs
  - Bilateral hemiuteri with noncommunicating endometrial cavities
  - Divergent uterine horns with deep fundal cleft
    - Each horn has normal zonal anatomy
  - 2 cervices
  - Vaginal septum present in 75%
  - Obstructed hemivagina-ipsilateral renal agenesis
    - Unilateral obstructing transverse vaginal septum in hematometrocolpos
    - Absent ipsilateral kidney
  - ± hematometrocolpos with obstruction
- **Bicornuate uterus**
  - 10% of MDAs
  - 2 symmetric cornua fused inferiorly
    - Normal zonal anatomy maintained
    - ± septum containing fibrous tissue (low signal on T2WI)
  - External fundal cleft
    - Must be at least 1 cm in depth
    - Extends to internal cervical os in complete bicornuate uterus
    - Differentiates bicornuate from septate uterus
  - Intercornual angle variable
    - > 105° suggestive of bicornuate (rather than septate) uterus
  - Usually single cervix though variations exist
    - Bicornuate bicollis: Duplicated cervix though there is some degree of communication between horns
  - ± hydrometrocolpos with obstruction
- **Septate uterus**
  - Most common MDA (55%)
  - Persistent uterovaginal septum of variable length
    - Septum extends into upper vagina in 25%
    - Inferior portion of septum composed of fibrous tissue (low signal on T2WI)
    - Cervical duplication rare
  - External fundal contour usually mildly convex or flat though can be indented < 1 cm
  - Intercornual angle variable
    - < 75° suggestive of septate (rather than bicornuate) uterus
  - Overall uterine size normal though each endometrial cavity smaller than normal
  - ± hydrometrocolpos with obstruction
- **Arcuate uterus**
  - Mild convex indentation on fundal endometrium
  - Normal external fundal contour
  - No fibrous component
- **Vaginal septum**
  - Can be associated with any MDA
  - ± transverse or longitudinal

MR Findings

- T1WI
  - When obstructed, high signal intensity contents indicate hemorrhagic byproducts
- T2WI
  - Normal zonal anatomy
    - High-signal endometrium & intermediate-/high-signal myometrium
    - Junctional zone seen as low-signal line separating endometrium & myometrium
  - Imaging parallel to long axis of uterus best for determining uterine morphology
  - In unicornuate & uterine hypoplasia, hypoplastic uterine remnant best seen on sagittal or axial T2WI
    - Usually intermediate or low signal intensity myometrium with small endometrial cavity
  - Fibrous septum low signal intensity (septate, bicornuate)

Ultrasonographic Findings

- Usually 1st imaging test performed, so important to recognize abnormality

- Alterations to uterine contour & endometrial cavity as above
- Limitations
  - Uterine characterization limited from 2-3 months until puberty due to small size
    - Anomalies can be seen prenatally & in neonate under influence of maternal hormones
  - Can be difficult to image outer fundal contour
    - Need transverse view of fundus in orthogonal plane
  - Demonstrating communication between endometrial cavities can also be challenging
  - 3D can be helpful
- Always image kidneys when MDA identified

Radiographic Findings

- Hysterosalpingography
  - Classification based on contour/morphology of endometrial cavity or cavities
  - Outer fundal contour not visible

Imaging Recommendations

- Best imaging tool
  - Ultrasound as initial screening exam
  - MR for definitive diagnosis
- Protocol advice
  - Image kidneys when MDA identified on ultrasound
  - MR
    - Combination of T1/T2WI for external contour & endometrial cavity
      - Image parallel to long axis of uterus for fundal contour
    - T2WI best for distinguishing zonal anatomy
    - Obtain at least 1 coronal series to cover renal fossa

DIFFERENTIAL DIAGNOSIS

Imperforate Hymen

- Not associated with MDA
- Differentiation from low transverse vaginal septum can be difficult by imaging

Cervical Stenosis

- ± inflammatory, iatrogenic, or secondary to mass effect
- Can cause obstruction & endometrial distortion mimicking MDA

PATHOLOGY

General Features

- Etiology
  - Müllerian duct structures
    - Fallopian tubes
    - Uterus
    - Cervix
    - Upper 2/3 of vagina
  - MDAs occur in 1 of 3 phases
    - Organogenesis: Agenesis, hypoplasia, unicornuate
    - Fusion: Didelphys, bicornuate
    - Septal resorption: Septate, arcuate
- Associated abnormalities
  - Renal anomaly present in ~ 30%
    - Renal agenesis most common (~ 2/3)
    - Ectopic kidney
    - Horseshoe kidney
    - Renal dysplasia
    - Duplicated collecting system

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic
  - Symptoms may develop at menarche
    - Primary amenorrhea
    - Dysmenorrhea
    - Cyclic abdominal pain

Demographics

- Epidemiology
  - Incidence estimated at 1%

Natural History & Prognosis

- Fertility issues
- Spontaneous abortions
- Ectopic pregnancy

Treatment

- Differentiation of septate from bicornuate uterus important
  - Septate treated with hysteroscopic resection of septum
  - Bicornuate rarely requires surgery
- Medical: Suppression of menses
- Surgical
  - Removal of rudimentary horn
  - Relieve obstruction
  - Resection of uterine septum

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Outer fundal contour important for differentiating bicornuate from septate uterus
- Define endometrial cavity & presence of communication when there are 2 uterine horns
- 1 or 2 cervices: Complete bicornuate vs. bicornuate bicollis vs. didelphys
  - Differentiate low-signal septum extending through single cervical os from 2 separate cervices

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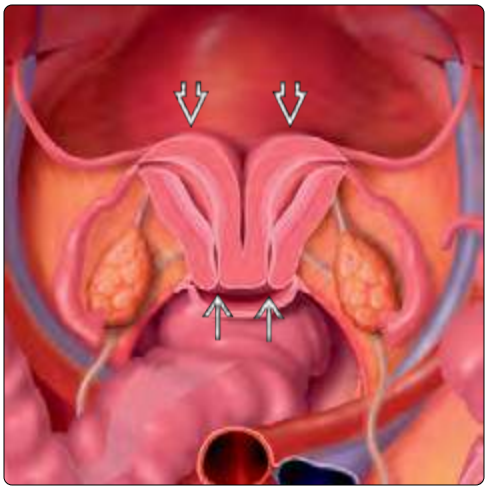
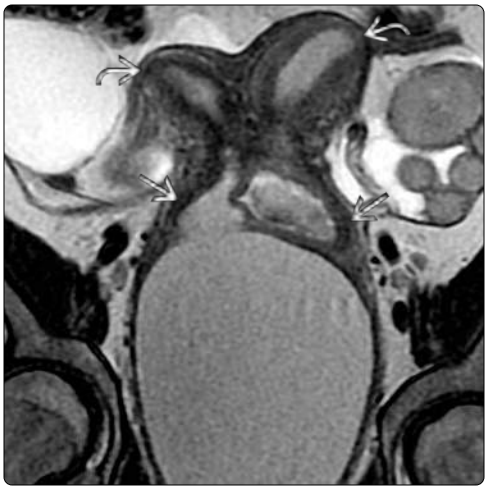
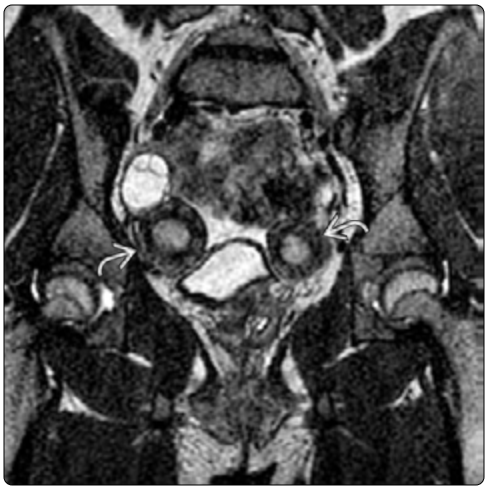
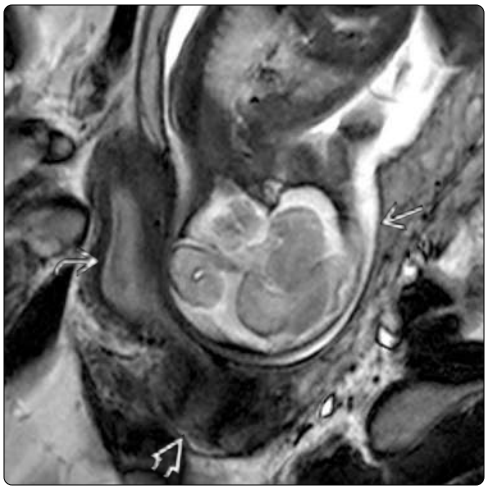


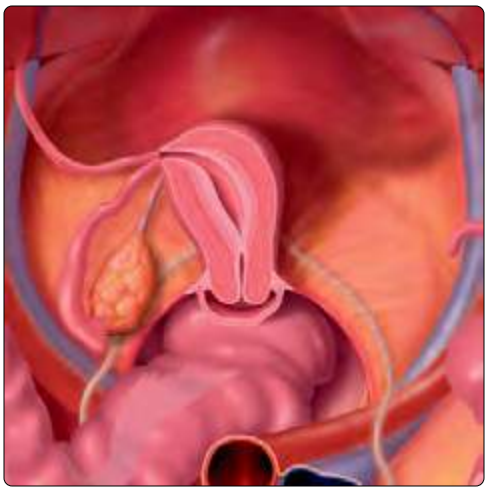


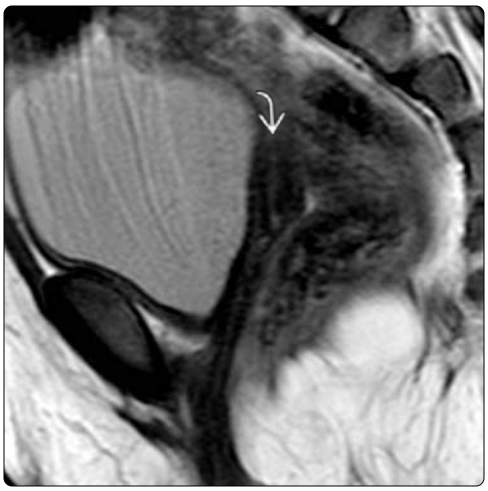
(Left) *Graphic of uterus didelphys shows 2 separate uterine horns, each with normal zonal anatomy as well as 2 cervices.* **(Right)** *Coronal oblique T2 MR shows 2 separate uterine horns, 2 cervices, & a massively distended vaginal vault in this patient with hydrometrocolpos & obstructed didelphys.*





(Left) *Coronal T2 MR shows 2 uterine horns that are widely separated in the pelvis in a patient with didelphys & an anorectal malformation.* **(Right)** *Coronal oblique T2 MR shows a uterus didelphys with a 25-week fetus in the left uterine horn as compared to endometrial proliferation in the right horn. Both cervices are closed.*





(Left) *Graphic of a unicornuate uterus illustrates the typical banana shape. There is complete absence of left-sided müllerian duct structures, although this is not always the case.* **(Right)** *Sagittal T2 MR in a patient with a complex cloacal malformation shows a unicornuate uterus located just right of the midline.*

KEY FACTS

TERMINOLOGY

- Follicle: Normal physiologic cyst < 1 cm in diameter
  - Dominant follicle may measure up to 3 cm
- Functional cysts: Can measure up to 3-10 cm
  - Corpus luteal cyst: Dominant follicle after ovulation
  - Follicular cyst: Normal mature follicle fails to involute
- Hemorrhagic cyst: Hemorrhage into functional cyst

IMAGING

- US mainstay of ovarian imaging; MR in limited circumstances
- Well-marginated round or ovoid structure within borders of ovary & having no solid component
  - Thin wall (< 3 mm)
  - No internal septa, nodule, fat, Ca^{2+}, or vascularity
  - ↑ heterogeneity with hemorrhage: Internal reticulations (or lace-like pattern) of clot mixed with tiny cystic spaces vs. layering debris

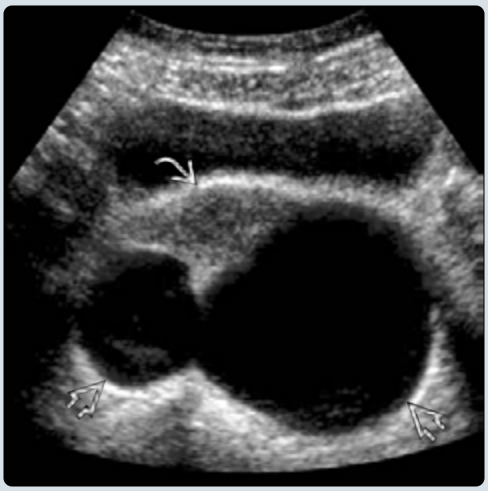
CLINICAL ISSUES

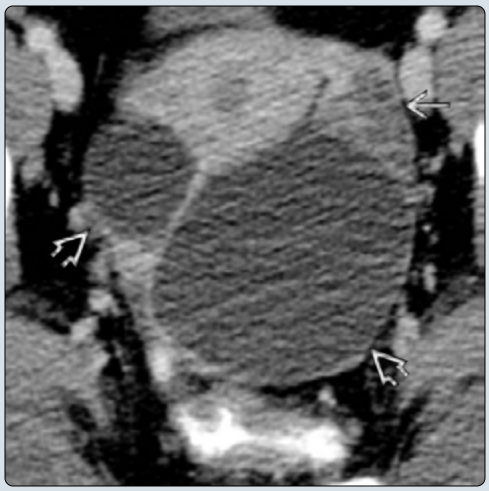
- Usually asymptomatic; pain if large or complicated by rupture, hemorrhage, or torsion
- Treatment/prognosis
  - > 90% of all functional cysts resolve spontaneously
  - Cysts < 3 cm should be considered physiologic in pre- & postmenarchal children
  - Cysts up to 4-5 cm usually monitored with surveillance ultrasound
  - Surgery vs. further imaging considered in larger cysts due to risk of torsion or neoplasm
    - For resection, ovarian sparing approach preferred if benign etiology suspected

DIAGNOSTIC CHECKLIST

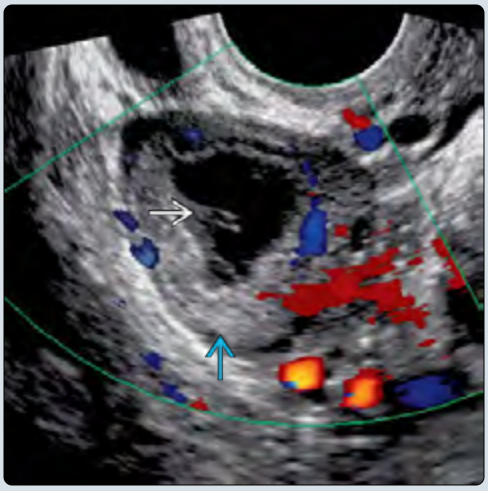
- With cystic ovarian lesion in child, radiologist must consider: Could this be torsion, neoplasm, or other pathology?
- Low threshold for follow-up US of asymptomatic cyst in 4-6 weeks if initial study unclear (due to size or mild complexity)

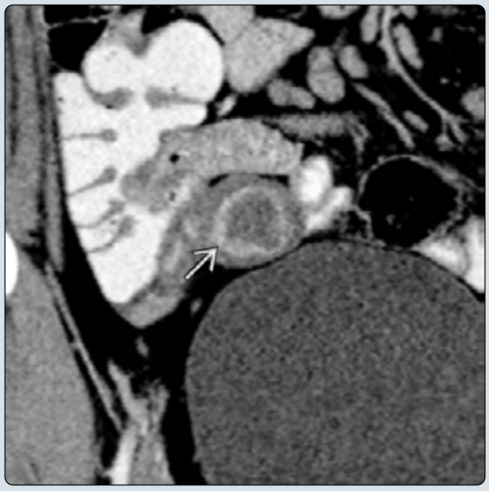
(Left) *Transverse US shows 2 well-defined, round, anechoic structures abutting the uterus, both of which have imperceptible walls & enhanced through transmission, consistent with simple cysts.* **(Right)** *Axial CECT obtained in the same patient also demonstrates typical characteristics of simple cysts. The normal-appearing left ovary lies directly adjacent to the larger cyst on this image. Both cysts resolved by the time of a follow-up ultrasound obtained several weeks later, consistent with follicular cysts.*





(Left) *Transverse endovaginal color Doppler US shows a hypoechoic structure within the right ovary that has an irregular contour, a hyperechoic rim, & a small amount of echogenic material centrally. There is mild adjacent hyperemia. The findings are characteristic of a corpus luteal cyst.* **(Right)** *Coronal CECT of the same patient on the same day shows the typical mildly thickened, irregular, hypervascular wall indicative of a corpus luteal cyst.*





TERMINOLOGY

Definitions

- Well-marginated, round or ovoid structure within or at borders of ovary; no solid component
  - No fat, Ca^{2+}, or septa
  - Thin, imperceptible wall (< 3 mm thick)
  - May appear more complex after internal hemorrhage
  - No internal vascularity on color Doppler imaging
- Definitions of different cyst types of cysts & their sizes vary
  - Follicle
    - Generally < 1 cm in diameter
    - Dominant follicle may measure up to 3 cm
  - Functional cysts
    - Corpus luteal cyst
      - After ovulation, dominant follicle becomes corpus luteal cyst
      - Usually < 3 cm, though can be much larger
    - Follicular cyst
      - Occurs when normal mature follicle fails to involute
      - Usually 3-10 cm
  - Hemorrhagic cyst
    - Hemorrhage occurs into one of above

IMAGING

General Features

- Best diagnostic clue
  - Simple cyst: Unilocular with imperceptible wall & near water echogenicity/attenuation/signal intensity internally with no central complexity or vascularity
- Size
  - Cysts under 3 cm should be considered physiologic in pre- & postmenarchal children
  - Cysts > 4-5 cm may require additional consideration
- Morphology
  - Round or ovoid with well-defined margins

Ultrasonographic Findings

- Differentiation of follicle & functional cyst mainly by size
- Follicles & follicular cysts without hemorrhage
  - Unilocular with smooth margin
  - No internal septations, though adjacent follicles can sometimes simulate septations
  - Anechoic with ↑ through transmission
    - May have minimal internal debris
- Corpus luteal cysts
  - Unilocular though contour may be somewhat irregular
  - Range from anechoic to isoechoic
  - Wall thickness variable due to vascularization
  - Classically have ↑ peripheral vascularity
  - Appearance can be identical to follicular cyst
- Hemorrhagic cysts
  - Contain heterogeneously echogenic debris, which becomes more hypo- or anechoic as clot lysis occurs
  - Reticular or lace-like pattern of internal echoes classic
  - Fluid-debris level sometimes present
  - ↑ through transmission maintained despite echogenicity

CT Findings

- CECT
  - Hypoattenuating, well-marginated mass within ovary without enhancement
  - Ovarian stroma usually low density, though normal tissue surrounding cyst can simulate soft tissue rim
    - Punctate hypodense follicles may be visualized in mildly enhancing ovarian parenchyma
  - Fat density or Ca^{2+} always pathologic
  - Corpus luteal cysts
    - Unilocular
    - Thicker wall, usually 2-4 mm, often hyperenhancing & crenulated (irregular), especially if ruptured
  - Hemorrhagic cysts more dense than simple cysts
    - Most commonly arise in follicular or corpus luteal cysts
    - Fluid-fluid level rarely seen on CT
    - Cyst rupture can cause free peritoneal fluid, which may be hyperdense if due to hemorrhage
      - Corpus luteal cysts more commonly result in hemoperitoneum due to their vascular wall

MR Findings

- May be performed if US diagnosis unclear
  - Accuracy for MR in characterizing sonographically indeterminate lesions ranges from 83-93%
  - Findings more indicative of neoplasm include
    - Thick, enhancing wall or septations > 3 mm
    - Enhancing mural nodule, papillary projections, or other solid components
    - Size > 5 cm
- Functional cysts: Homogeneously low T1, high T2 signal
- Hemorrhagic cyst: Typically high T1 & T2 signal
  - Fluid-debris level sometimes seen on T2
  - No loss of high T1 signal with fat suppression
  - Hemorrhagic components may restrict diffusion
- T1 FS & in-/opposed-phase GRE sequences aid in looking for fat of dermoid
- Mild rim enhancement with hemorrhagic or corpus luteal cyst on T1 C+ FS
  - Subtracted pre- from postcontrast T1 FS images helpful to confirm true enhancement rather than pseudoenhancement of blood products

Imaging Recommendations

- Best imaging tool
  - US mainstay for evaluation of ovaries
    - Characterization of cyst wall & internal components
    - Color Doppler gives additional information, especially in evaluating for presence of solid component
  - MR considered when US indeterminate or if neoplasm suggested but full extent not discernible by US
  - CT use minimized due to radiation concerns

DIFFERENTIAL DIAGNOSIS

Dermoid/Teratoma

- 10-15% may appear as entirely cystic by imaging
- MR much more sensitive/specific (due to fat component)

Ovarian Malignancy

- Growing, mildly complex cyst or higher degrees of complexity (solid nodular components, internal vascularity) should raise concern
- MR if question of hemorrhage vs. solid components

Ovarian Torsion

- Asymmetric, edematous, ovarian parenchyma with peripheral follicles, ± midline/contralateral location of ovary
- ± twisted vascular pedicle (whirlpool), ↓ or absent blood flow
- Frequently associated with cysts > 5 cm

Paraovarian Cyst

- Long list of potential causes including
  - Enteric duplication cyst
  - Peritoneal inclusion cyst
  - Lymphatic malformation

Tuboovarian Abscess

- Wide range of appearances from relatively simple-appearing cyst to complex, heterogeneous, cystic mass
- Clinical history & exam findings crucial

Ectopic Pregnancy

- History & pregnancy test results critical

PATHOLOGY

General Features

- Etiology
  - Infancy
    - In fetus, follicular stimulation occurs from maternal estrogen, placental hCG, & fetal gonadotropins
    - More common in maternal diabetes, toxemia, or Rh isoimmunization during pregnancy
    - Small cysts may be seen as early as 28-weeks gestation
    - Newborn estrogen & hCG levels fall, but serum LH & FSH levels ↑, peaking at 3-4 months
  - Premenarchal
    - Cysts > 1 cm least common in this age group due to low hormone levels
    - Follicle maturation occurs at low rate
  - Postmenarchal
    - Hormone levels approach adult levels, & cysts become more common
    - Usually result of dysfunctional ovulation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually asymptomatic; pain if large or complicated by rupture, hemorrhage, or torsion
  - Corpus luteal cyst more commonly symptomatic, even without significant hemorrhage

Demographics

- Epidemiology
  - Neonates: Small cysts present in up to 98% at birth
    - ~ 20% ≥ 1 cm
  - Premenarchal: Cysts > 1 cm in 2-5%
  - Postmenarchal: Cysts very common
    - Nonfunctional cysts & malignant ovarian neoplasms also occur at greater rate

Natural History & Prognosis

- Over 90% of all functional cysts resolve spontaneously
  - Larger cysts may take longer to resolve

Treatment

- Cysts < 4-5 cm usually monitored with surveillance US
- Surgery considered in larger cysts due to risk of torsion or neoplasm
  - Options include aspiration, fenestration, & resection
  - For resection, ovarian sparing approach preferred if benign etiology suspected

DIAGNOSTIC CHECKLIST

Consider

- 3 main concerns for radiologist when cystic ovarian lesion encountered in child
  - Could this be torsed?
  - Could this be neoplastic?
  - Could this be other pathology (e.g., tuboovarian abscess, ectopic pregnancy, ruptured appendicitis, etc.)?

Image Interpretation Pearls

- Superimposed torsion more likely with cysts > 5 cm
- Clinical judgment overrides imaging: Simple or hemorrhagic cyst with otherwise normal-appearing ovarian parenchyma (by grayscale & Doppler US) can still be torsed
  - Does not give license to use phrase "cannot rule out torsion" in every report
  - Simply means that strong clinical suspicion + any ovarian abnormality on imaging may require laparoscopy

Reporting Tips

- Close follow-up if complexity or size of asymptomatic cyst not entirely typical for most simple or hemorrhagic cysts
  - US in 4-6 weeks; subsequent MR &/or gynecology referral if questions persist

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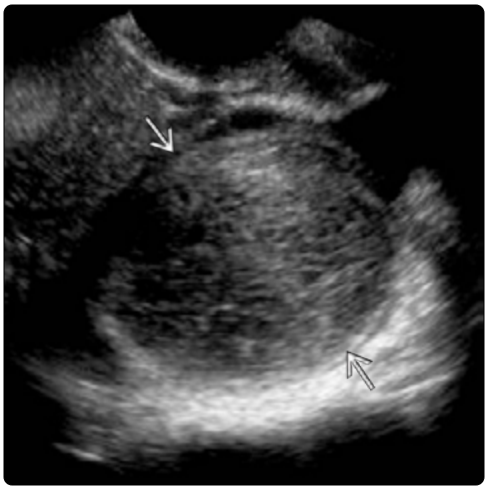
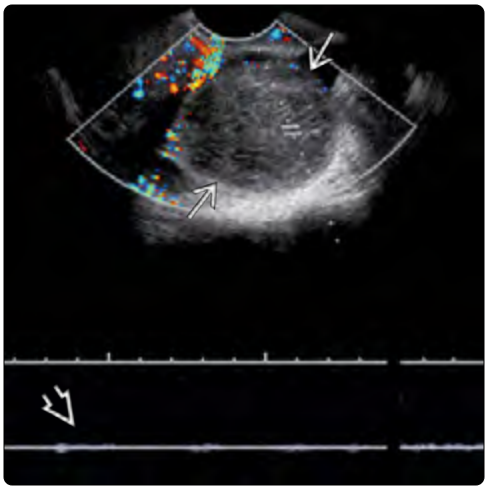
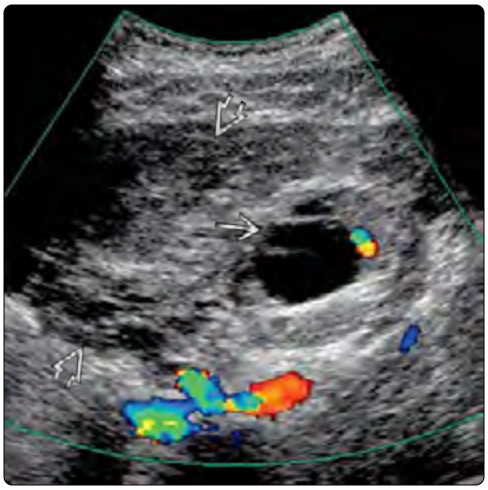
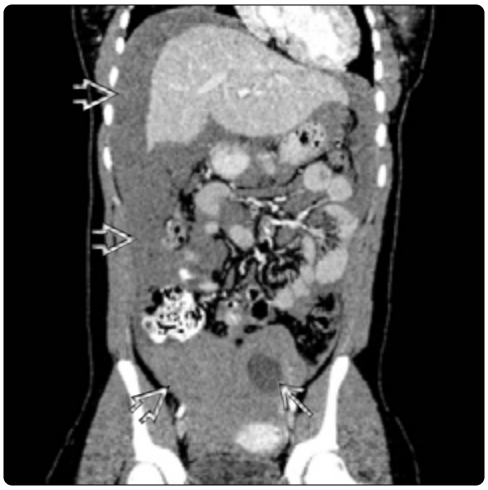


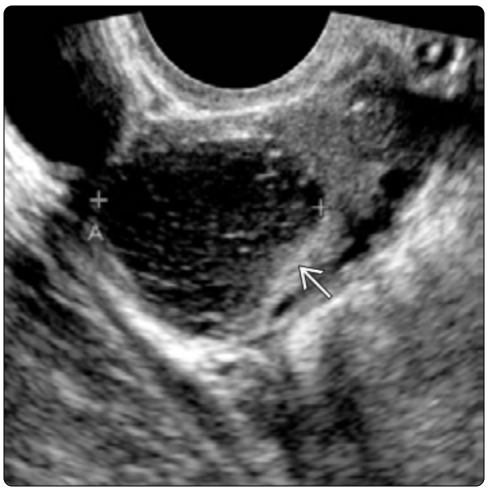


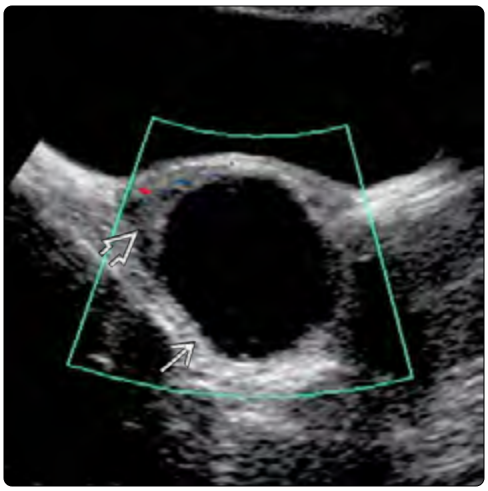
(Left) *Grayscale endovaginal US shows a large, hemorrhagic ovarian cyst ⇨ with internal lace-like echoes & tiny cystic spaces, stretching the ovarian capsule & causing pain.* **(Right)** *Pulsed Doppler US through the same hemorrhagic cyst ⇨ shows no internal vascularity ⇨ to suggest a tumor. It is helpful in such cases to look for normal ovarian tissue splayed along the cyst as cysts > 5 cm can predispose to torsion. If the clinical presentation is particularly concerning for torsion, then laparoscopy may be necessary regardless.*





(Left) *Transverse color Doppler US shows an irregular cystic structure containing septations ⇨ within the left ovary. The ovary is surrounded by a large amount of heterogeneously echogenic material ⇨, which was mobile in real time. The findings are indicative of a ruptured hemorrhagic cyst.* **(Right)** *Coronal CECT in the same patient also demonstrates the left adnexal cyst ⇨ surrounded by a large volume of hemoperitoneum that extends to the subdiaphragmatic region ⇨.*





(Left) *Transverse endovaginal US shows a well-defined, hypoechoic mass ⇨ in the right ovary with a reticular, lace-like internal structure suggestive of a hemorrhagic cyst.* **(Right)** *Transverse color Doppler US of a patient with a simple cyst ⇨ shows eccentric but otherwise normal-appearing ovarian tissue ⇨ splayed along the cyst. Despite the unalarming sonographic appearance, the patient's symptoms warranted laparoscopy, where the ovary was found to be torsed.*

Ovarian Teratoma

KEY FACTS

TERMINOLOGY

- Dermoid tumor, dermoid cyst, mature cystic teratoma
- Teratomas made up of variety of parenchymal cell types from > 1 germ cell layer, usually all 3

IMAGING

- Best clue: Heterogeneous pelvic mass containing Ca^{2+}, hair, fat, & cystic components
- Typically well-defined margins without surrounding inflammatory changes
- US 1st-line modality for female pelvic pain &/or mass
  - Multiple classic signs described for teratoma
    - Dermoid plug: Echogenic nodule protruding into cyst
    - Dermoid mesh: Linear/punctate echogenic foci of hair
    - Tip of iceberg: Echogenic superficial interfaces obscure deeper components of mass
- Radiographs showing tooth-like Ca^{2} strongly suggestive
- MR & CT best demonstrate fat, confirming teratoma

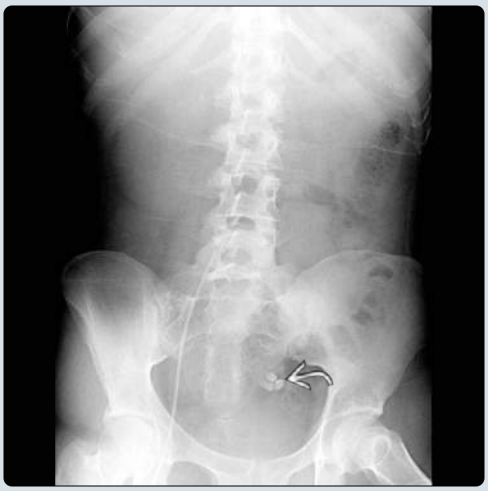
PATHOLOGY

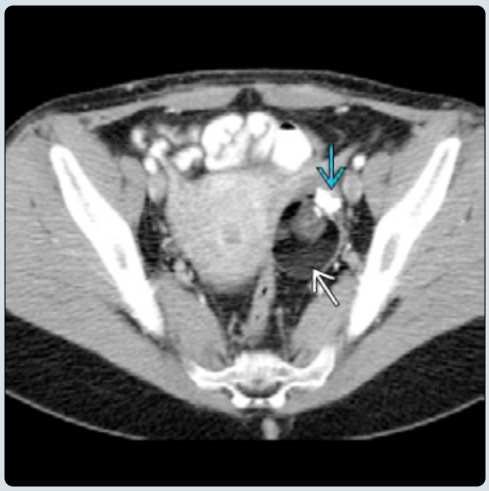
- Complications of ovarian teratoma include
  - Ovarian torsion
  - Rupture, causing chemical peritonitis
    - May lead to severe adhesions
  - Anti-NMDA receptor encephalitis
  - Malignancy (2%)

CLINICAL ISSUES

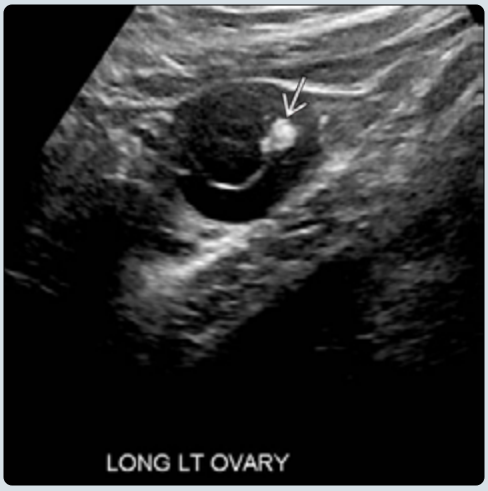
- Teratoma most common ovarian germ cell tumor
- Teratoma most common ovarian neoplasm < 20 years old
  - Found in any age; mean age of presentation: 30 years
- Often incidental finding on physical exam or during imaging for unrelated symptoms
  - With torsion or rupture, acute onset of pain typical
- Treatment: Surgical resection, ovary-sparing surgery
  - Laparoscopic surgery preferred

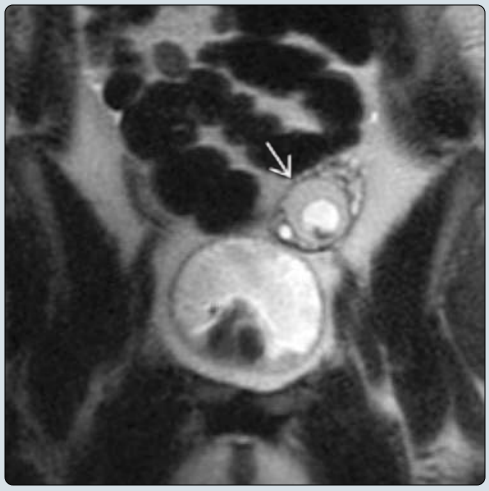
(Left) *AP radiograph of the abdomen shows several Ca^{2+} resembling teeth in the pelvis of a 22-year-old woman with no current abdominal complaints. The right groin central venous catheter is for treatment of an unrelated chronic health condition.* **(Right)** *Axial CECT in the same patient shows the well-encapsulated, mixed density mass in the left adnexa between the iliac vessels & the uterus. This combination of fat & Ca^{2+} in the adnexal region is typical of an ovarian teratoma.*





(Left) *Longitudinal US of the left ovary in a 14-year-old girl shows a mixed cystic & solid mass with hyperechoic foci & curvilinear septations.* **(Right)** *Coronal SSFSE T2 MR in the same patient (performed for other medical issues) shows the heterogenous lesion centrally within the left ovary. It was subsequently surgically shelled out of the ovary (i.e., an ovarian-sparing resection) & found to be a mature teratoma.*





TERMINOLOGY

Synonyms

- Dermoid tumor, dermoid cyst, cystic teratoma

Definitions

- Teratomas are made up of various parenchymal cell types from > 1 germ layer, usually all 3
- Teratomas generally lie in midline or paraxial & arise from totipotential cells
- Term dermoid comes from skin-like lining found in many of these tumors

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous pelvic mass containing calcium, fat, fluid, & other internal debris
  - Typically well-defined margins without surrounding inflammatory changes
- Location
  - Pelvis & lower abdomen
- Size
  - Variable, from 1-45 cm diameter
  - Mean diameter of 6 cm
- Morphology
  - Mixed tissue types surrounded by capsule

Radiographic Findings

- May be occult
- ± Ca^{2+}; those resembling teeth or bone strongly suggest teratoma
- ± mass effect on bowel

Ultrasonographic Findings

- Grayscale ultrasound
  - Heterogeneous mass with cystic & solid components
  - Ca^{2+} may show posterior shadowing or ring-down artifact when very small
  - Fat & hair appear echogenic; hair may appear reticular
  - ± fluid-fat-debris levels &/or floating debris within cysts
  - Dermoid mesh sign: Linear & punctate hyperechoic foci (due to hair) within cystic mass
  - Dermoid plug (Rokitansky nodule): Echogenic solid focus bulging into lesion from cyst wall
  - Tip of iceberg sign refers to strong echogenic interface at leading edge of teratoma that blocks deeper components from view
    - May be caused by Ca^{2+}, hair, fat, etc.
- Color Doppler
  - Flow useful in differentiating solid perfused tissue from solid avascular hair, teeth, etc.

CT Findings

- Exquisite for identifying fat & Ca^{2+}
- Otherwise heterogeneous tissue components with septations & fluid-debris levels
- Residual ovary tissue giving rise to teratoma may be difficult to identify if tumor large

MR Findings

- Heterogeneous signal due to mix of elements
  - Low signal intensity foci with blooming on T2* GRE suggests Ca^{2+}
  - Identification of fat key to diagnosis
    - T1WI ± FS
      - Signal loss upon FS application confirms presence of macroscopic fat (as do other T1 bright material such as blood, protein, etc.); may show fat-fluid level
    - In-/opposed-phase GRE sequences to look for microscopic fat
      - Signal loss on opposed-phase confirms fatty lesion

Imaging Recommendations

- Best imaging tool
  - US primary investigative tool for female pelvic pain or palpable mass
  - MR reserved for complex cases or patients ill-suited to sonography
  - CT minimized due to radiation concerns
- Protocol advice
  - Scan mass in orthogonal planes looking for variable tissue & cyst contents
  - Endovaginal scanning optimal when tolerated
  - Larger masses may require transabdominal scanning & extended field of view

DIFFERENTIAL DIAGNOSIS

Other Ovarian Neoplasms

- Benign
  - Simple/follicular cysts
  - Cystadenomas
    - Mucinous
    - Serous
  - Gonadoblastoma
- Malignant
  - Germ cell tumors
  - Sex cord-stromal tumors
  - Epithelial tumors
  - Malignant teratomas

Endometrioma

- Cyclic pain history useful

Perforated Appendicitis With Appendicolith

- Heterogeneous collection in lower abdomen, can be close mimicker of ovarian teratoma

Tuboovarian Abscess

- Fever, cervical tenderness, & vaginal discharge typical

Ovarian Torsion

- May require surgical exploration to distinguish
- May coexist with ovarian teratoma

Ectopic Pregnancy

- Correlate with β-hCG

Peritoneal Inclusion Cyst

- Lacks complex contents seen in teratomas

Bladder Calculi

- Ca^{2} in pelvis could reside in urinary bladder

- Especially in patients with Mitrofanoff, other bladder surgeries, & those performing bladder catheterization

Stool Impaction

- Stool & gas in rectum can mimic tip of iceberg sign

PATHOLOGY

General Features

- Etiology
  - Arise from primordial germ cells, which migrate during embryogenesis from yolk sac to gonads
- Associated abnormalities
  - Complications of ovarian teratoma include
    - Ovarian torsion
    - Rupture, causing chemical peritonitis
      - Severe adhesions can result from perforation of teratoma
    - Infection
    - Hemolytic anemia
    - Malignancy in ~ 2%
- 3 types of ovarian teratomas
  - Mature cystic teratoma (dermoid cyst)
  - Monodermal teratomas (struma ovarii, carcinoid tumors, & neural tumors)
  - Immature teratomas
- Teratoma distribution
  - Sacrococcygeal (57%)
  - Gonadal (29%), ovarian > testicular
  - Mediastinal > retroperitoneal > cervical > intracranial
- Cells differentiate along various germ lines, essentially recapitulating any tissue of body
- Ectoderm, mesoderm, & endodermal elements may all be present
  - Because ectodermal components tend to predominate term dermoid cyst has been applied
- Tissues include hair, teeth, fat, skin, muscle, & endocrine tissue

Gross Pathologic & Surgical Features

- Heterogeneous mass surrounded by well-defined capsule
- Cyst contents may be oily, milky, or have serous fluid, hair, teeth, cartilage, etc.

Microscopic Features

- Variable well-differentiated tissues, including bone, cartilage, muscle, thyroid follicles, gastrointestinal lining, respiratory epithelium, etc.
- Immature teratomas graded from 0 to 3 based on amount of immature neural tissue found in tumor specimen
  - Higher grades more likely to have yolk sac tumor components

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often incidental finding on physical exam or during imaging for unrelated symptoms
  - With torsion or rupture, acute onset of pain typical
- Other signs/symptoms
  - Abdominal pain, abdominal mass, or swelling
  - Abnormal uterine bleeding
  - Urinary symptoms
  - Gastrointestinal complaints
  - Back pain less common
  - Elevated α-fetoprotein or hCG more likely with germ cell tumors/immature teratomas
  - Rarely associated with anti-NMDA encephalitis

Demographics

- Age
  - Any age; mean of 30 years
- Gender
  - Females only for ovarian lesions
- Epidemiology
  - Teratoma most common ovarian germ cell tumor
  - Also most common ovarian neoplasm in patients < 20 years old
  - 10% of teratomas are diagnosed during pregnancy
  - Bilateral in up to 15%
- Right-sided teratomas slightly more common than left

Natural History & Prognosis

- Teratomas enlarge, spontaneously hemorrhage, or twist & come to medical attention
- Prognosis generally excellent following resection
  - Prognosis poor in small minority with malignant foci
  - Malignant teratomas tend to metastasize widely
  - 5-year survival for malignant teratomas is < 30%
- Fertility issues may arise
  - When entire ovary is removed
  - When chemical peritonitis impairs ovulatory function

Treatment

- Treatment is surgical resection, ovary-sparing surgery
- Laparoscopic surgery preferred
- Malignant teratomas treated with surgery, hyperthermic intraperitoneal chemotherapy, neoadjuvant chemotherapy

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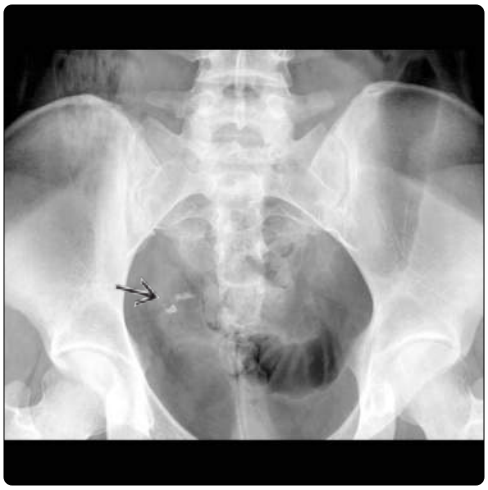
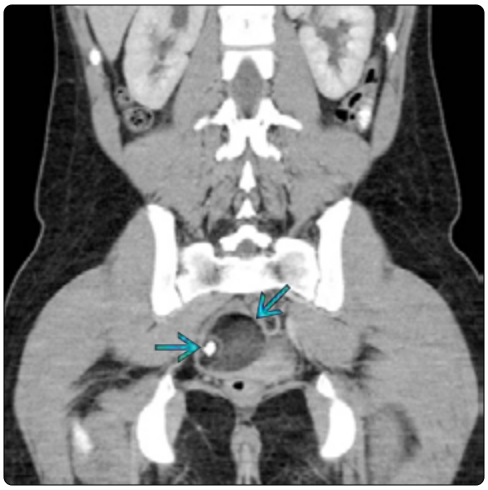
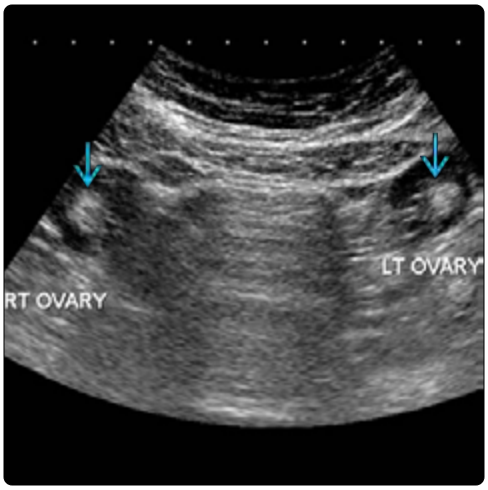
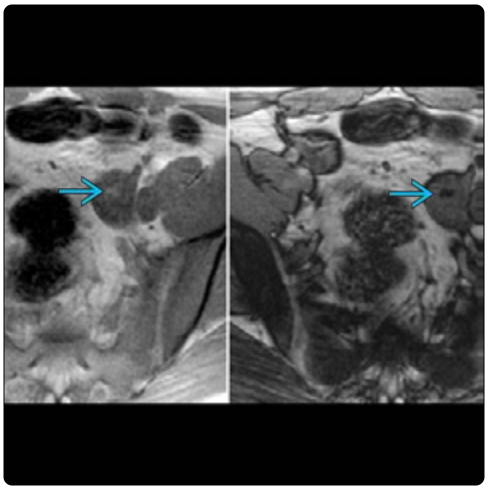


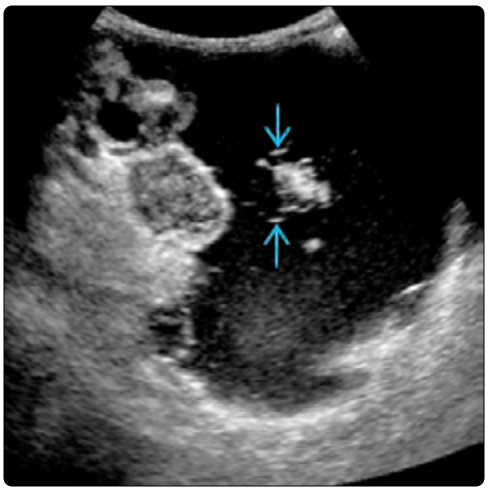


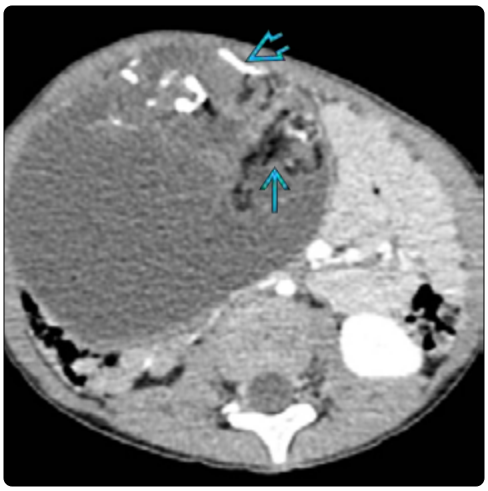
(Left) *AP radiograph in a teenage girl with pelvic pain & normal menstrual cycles shows chunky Ca^{2+} in the pelvis ⇨ that are suspicious for an ovarian teratoma (or possibly appendicoliths).* **(Right)** *Coronal CECT in the same girl shows a heterogeneous mass ⇨ in the right adnexa containing Ca^{2+}, fat, fluid (not shown), & nonlayering debris. The presence of all these tissues confirms ovarian teratoma as the diagnosis.*





(Left) *Tranverse US in an 11-year-old girl with anti-NMDA receptor encephalitis shows bilateral 1-cm echogenic ovarian lesions ⇨, potentially teratomas.* **(Right)** *Axial in- (left) & opposed- (right) phase T1 GRE MR images in the same patient show a left ovarian lesion ⇨ containing macro- & microscopic fat with signal drop on the opposed-phase image. A similar finding was seen in the right ovary, & T1 FS images (not shown) also confirmed fat in both lesions. Ovarian-sparing teratoma resection was performed bilaterally.*





(Left) *Transverse abdominal US in a 3-year-old girl with a palpable mass shows a mixed cystic & solid lesion with nodular echogenic foci protruding into the cyst. Several linear echogenic strands (appearing punctate in cross section ⇨) correspond to hair (the dermoid mesh sign).* **(Right)** *Axial CECT in the same patient shows numerous foci of fat ⇨ & Ca^{2+} ⇨ within the mixed cystic & solid mass, typical of a teratoma.*

Ovarian Malignancies of Childhood

KEY FACTS

IMAGING

- **Germ cell tumors**
  - Mature ovarian teratoma: Cystic mass with fat & Ca^{2+}
  - Immature ovarian teratoma: Cannot differentiate from mature teratoma on imaging
  - Ovarian dysgerminoma: Multilobulated, solid mass with Ca^{2+}
  - Yolk sac tumor: Large, complex cystic & solid mass that extends into abdomen
  - Choriocarcinoma: Solid, hypervascular mass in female with elevated β-hCG
- **Sex cord-stromal tumors**
  - Juvenile granulosa-theca cell tumor: Large, multilocular mass with septations & solid component
  - Sertoli-Leydig cell tumor: Heterogeneous cystic & solid mass
- **Epithelial tumors**
  - Serous tumor: Homogeneous cystic mass with thin walls & no mural nodule
  - Mucinous tumor: Large, heterogeneous, multilocular cystic mass

PATHOLOGY

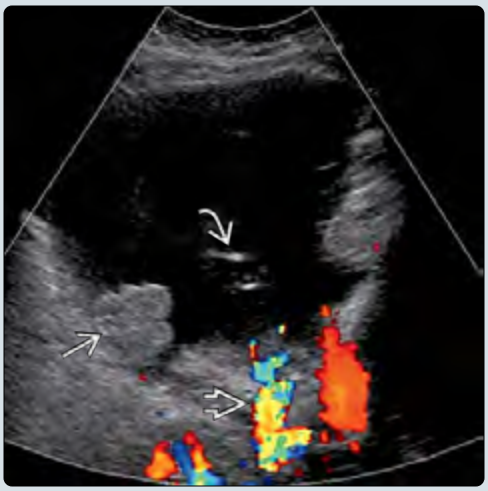
- 2 main staging systems for ovarian neoplasms
  - International Federation of Gynecology & Obstetrics
  - Children's Oncology Group

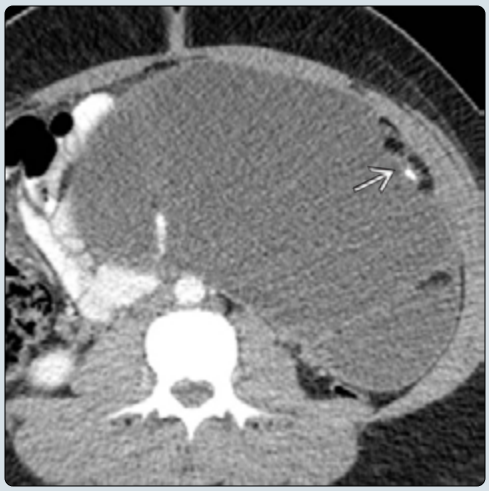
CLINICAL ISSUES

- Abdominal pain &/or distention, mass, ↓ appetite
- Hormonally active tumors
  - Juvenile granulosa-theca cell tumor: 80% present with pseudoprecocious puberty due to hyperestrogenism
  - Sertoli-Leydig cell tumor: ~ 30% virilizing
- Torsion more likely with benign neoplasms
- Current treatment: Unilateral salpingo-oophorectomy
- Chemotherapy for stage II disease & higher

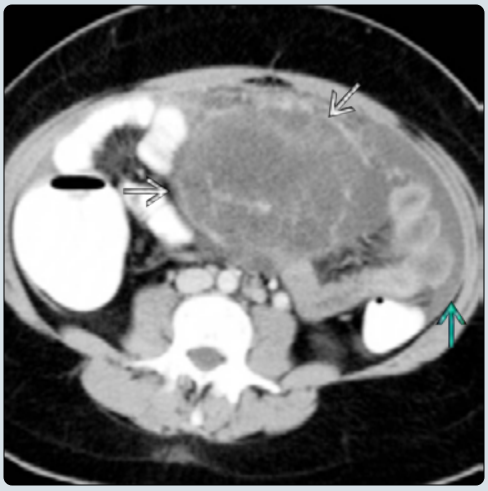
DIAGNOSTIC CHECKLIST

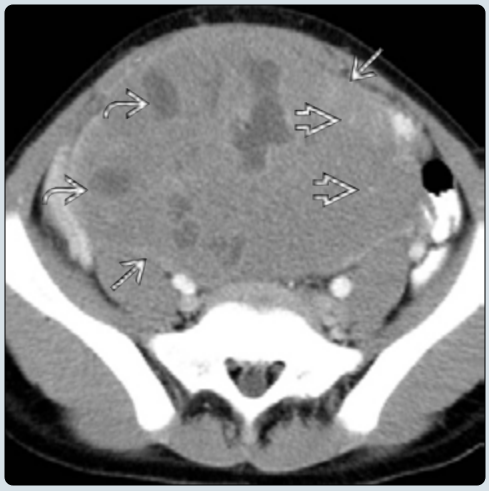
- Look for signs of tumor rupture/peritoneal seeding





(Left) *Transverse US in an adolescent with a mature cystic teratoma shows a large, predominantly anechoic mass in the pelvis. Solid nodules, linear echogenic foci of hair, & a peripheral twinkle artifact from Ca^{2+} are noted in the mass.* **(Right)** *Axial CECT in the same patient shows a large cystic mass with peripheral fat & calcification. The presence of soft tissue/fluid, fat, & calcium is diagnostic of a teratoma, but it is not possible to differentiate between mature & immature tumors by imaging.*





(Left) *Axial CECT of the abdomen in an adolescent with a yolk sac tumor shows a large heterogeneous mass. There is nearby ascites & peritoneal enhancement concerning for peritoneal seeding.* **(Right)** *Axial CECT in an adolescent with a dysgerminoma shows a large, heterogeneous mass in the pelvis. The mass is mostly solid with small cystic foci. There are faint areas of increased density in the mass due to small Ca^{2+}. Dysgerminomas typically appear as solid masses with speckled Ca^{2+}.*

TERMINOLOGY

Definitions

- Germ cell tumors
  - Mature ovarian teratoma, immature ovarian teratoma, ovarian dysgerminoma, yolk sac tumor, choriocarcinoma
- Sex cord-stromal tumors
  - Juvenile granulosa-theca cell tumor, Sertoli-Leydig cell tumor
- Epithelial tumors
  - Serous & mucinous tumors
  - Can be differentiated into benign, borderline, & malignant, depending on histology

IMAGING

General Features

- Best diagnostic clue
  - Germ cell tumors
    - Mature ovarian teratoma: Cystic mass with intratumoral fat & Ca^{2+}
    - Immature ovarian teratoma: Cannot reliably differentiate from mature teratoma on imaging; immature teratomas tend to
      - Be larger
      - Have smaller, speckled Ca^{2+}
      - Have more prominent solid components
    - Dysgerminoma: Multilobulated, solid mass with Ca^{2+}
    - Yolk sac tumor: Large, complex cystic & solid mass that extends into abdomen
    - Choriocarcinoma: Solid, hypervascular mass with elevated β-hCG
  - Sex cord-stromal tumors
    - Juvenile granulosa-theca cell tumor: Large, multilocular mass with septations & solid component
    - Sertoli-Leydig cell tumor: Heterogeneous, cystic, & solid
  - Epithelial tumors
    - Serous tumor: Homogeneous cystic mass with thin walls & no mural nodule
    - Mucinous tumor: Large, heterogeneous, multilocular cystic mass
- Location
  - Most ovarian tumors are unilateral
    - Slightly more common on right
    - Bilateral tumors in 5-15% depending on tumor type
      - More common with borderline malignant potential epithelial tumors & dysgerminomas
- Size
  - Variable; small (< 1 cm) to very large (> 20 cm)

Radiographic Findings

- Mature teratoma: May see tooth-like Ca^{2+}
- Other ovarian tumors: If large enough, tumor can cause mass effect upon bowel, displacing it superiorly & laterally
- Separate masses or complex ascites from tumor rupture & peritoneal seeding can also displace & narrow bowel

CT Findings

- Germ cell tumors
  - Mature ovarian teratoma: Cystic mass + fat & Ca^{2+}
  - Immature ovarian teratoma: Mass with prominent solid component + cystic foci & intratumoral fat
    - Ca^{2+} can be scattered rather than confined to mural nodules as in mature teratomas
  - Dysgerminoma: Multilobulated solid mass with enhancing fibrovascular septa
    - May have speckled Ca^{2+}
  - Yolk sac tumor: Large, heterogeneous mass with cystic & enhancing solid components
  - Choriocarcinoma: Solid enhancing tumor with areas of hemorrhage or necrosis
- Sex cord-stromal tumors
  - Juvenile granulosa-theca cell tumor: Large, multilocular mass with septations & solid component
  - Sertoli-Leydig cell tumor: Heterogeneous mass with cystic & solid components
- Epithelial tumors
  - Serous: Homogeneous mass with thin walls/septations
  - Mucinous: Large, multilocular cystic mass with internal contents containing different densities

MR Findings

- Germ cell tumors
  - Mature ovarian teratoma: Intratumoral fat confirmed with fat-saturated images
  - Immature ovarian teratoma: Mass with prominent solid component, cystic areas, & intratumoral fat
  - Dysgerminoma: Multilobulated mass with fibrovascular septa
    - Septa low signal intensity on all sequences but enhance intensely
  - Yolk sac tumor: Large, heterogeneous cystic & solid mass
  - Choriocarcinoma: Solid enhancing tumor with areas of hemorrhage or necrosis
- Sex cord-stromal tumors
  - Juvenile granulosa-theca cell tumor: Large, multilocular mass with septations & solid component
    - Foci of ↑ T1 signal due to intratumoral hemorrhage
    - Sponge-like appearance with solid areas interspersed with many cystic areas on T2WI
  - Sertoli-Leydig cell tumor: On T2WI, mass predominantly low signal with scattered foci of ↑ signal
- Epithelial tumors
  - Serous: Unilocular cystic mass with homogeneous signal intensity & thin wall
  - Mucinous: Multilocular cystic mass with locules containing fluid of differing signal intensity

Ultrasonographic Findings

- Germ cell tumors
  - Mature ovarian teratoma: Cystic mass with echogenic nodule that shadows (Rokitansky nodule)
    - May have multiple thin, echogenic bands from intratumoral hair
  - Immature ovarian teratoma: Cannot reliably distinguish from mature teratoma
  - Ovarian dysgerminoma: Multilobulated mass with anechoic areas representing hemorrhage or necrosis
  - Yolk sac tumor: Large, heterogeneous mass with cystic & solid components
  - Choriocarcinoma: Mostly solid mass with cystic areas of hemorrhage or necrosis

- Sex cord-stromal tumors
  - Juvenile granulosa-theca cell tumor: Large, multilocular mass with septations & solid component
  - Sertoli-Leydig cell tumor: Heterogeneous mass with cystic & solid components
- Epithelial tumors
  - Serous: Simple cystic mass
  - Mucinous: Large cystic mass with multiple loculations

Imaging Recommendations

- Best imaging tool
  - Ultrasound: 1st-line imaging modality for abdominal distention or palpable mass in young female patient
  - MR for further characterization & localization

DIFFERENTIAL DIAGNOSIS

Functional Cyst

- Occurs during follicular phase of menstrual cycle
- Simple ovarian cyst that resolves over time
- Usually < 3 cm

Corpus Luteum Cyst

- Hormone-dependent ovarian cyst
- Enhancing wall or solid-appearing mass in ovulating woman

Ovarian Torsion

- Presents with acute lower quadrant pain & asymmetric ↑ in ovarian size
- ± swirl or "whirlpool" of twisted ovarian pedicle, ↓ ovarian enhancement, peripheralization of follicles, surrounding edema, lack of or diminished internal flow
- Torsion may occur secondary to mass acting as lead point
  - Best suggested by size of lesion & new onset pain

PATHOLOGY

Staging, Grading, & Classification

- 2 main staging systems for ovarian neoplasms
  - International Federation of Gynecology & Obstetrics (FIGO)
    - Stage I: Limited to 1 ovary (IA), both ovaries (IB), or either ovary with ascites (IC)
    - Stage II: Tumor of 1 or both ovaries with pelvic extension to uterus or tubes (IIA), other pelvic tissue (IIB), or any pelvic extension with ascites (IIC)
    - Stage III: Ovarian mass with microscopic peritoneal implants (IIIA), macroscopic peritoneal implants < 2 cm (IIIB), or macroscopic implants > 2 cm or retroperitoneal or inguinal nodes (IIIC)
    - Stage IV: Distant metastases
  - Children's Oncology Group
    - Stage I: Limited to ovary
    - Stage II: Microscopic residual disease or disease in lymph nodes < 2 cm
    - Stage III: Gross residual disease or disease in lymph nodes > 2 cm or contiguous spread
    - Stage IV: Metastatic disease

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Abdominal pain &/or distention
- Other signs/symptoms
  - Palpable mass, fever, ↓ appetite
  - Torsion uncommon presentation for ovarian malignancy
    - Torsion more likely with benign neoplasms
  - Mature teratoma rarely presents with limbic encephalitis
  - Hormonally active tumors
    - Juvenile granulosa-theca cell tumor
      - 80% present with pseudoprecocious puberty due to hyperestrogenism
      - Accounts for 10% of cases of isosexual precocious puberty in females
    - Sertoli-Leydig cell tumor: ~ 30% virilizing; associated with *DICER1* mutation

Demographics

- Epidemiology
  - Incidence of ovarian masses in childhood: 2.6 cases per 100,000 girls per year
  - 16-55% malignant; account for 1% of all pediatric cancers

Natural History & Prognosis

- Most tumors are FIGO stage I at diagnosis
- Different tumors have different markers
  - Dysgerminoma: Lactic dehydrogenase
  - Yolk sac tumor: α-fetoprotein
  - Choriocarcinoma: β-hCG
  - Epithelial tumors: CA125
  - Juvenile granulosa cell tumor: Inhibin

Treatment

- Current treatment: Unilateral salpingo-oophorectomy
  - Fertility-sparing surgery if likely benign (teratoma)
- Chemotherapy for stage II disease & higher

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Fat-containing ovarian mass: Teratoma
- Other ovarian masses often diagnosed at resection
  - More important to suggest neoplasm & evaluate for complication than name specific tumor type
- Complications include
  - Ovarian torsion (typically in benign lesions)
  - Tumor rupture/peritoneal seeding
    - Irregular or discontinuous tumor margins
    - Ascites ± dependently layering debris
    - Smooth or nodular peritoneal thickening
    - Nodularity & abnormal enhancement of omentum
  - Distant metastases

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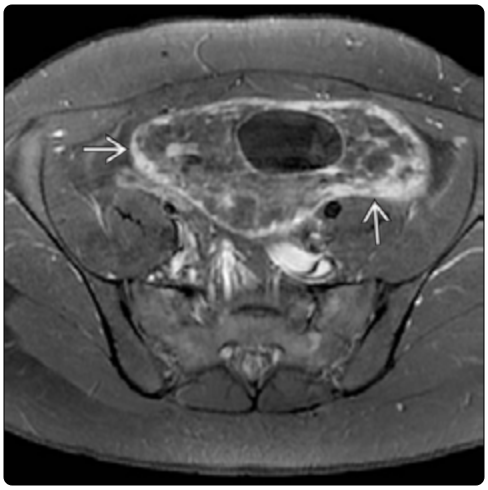
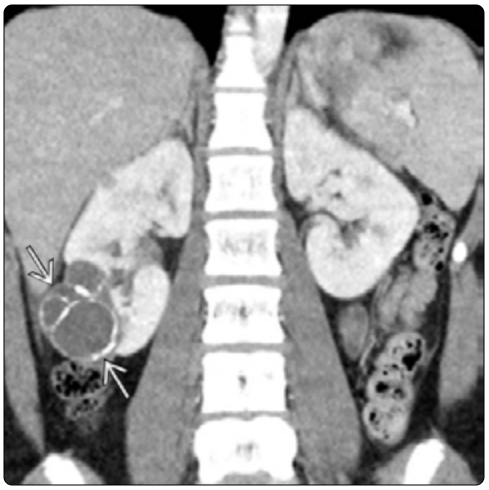
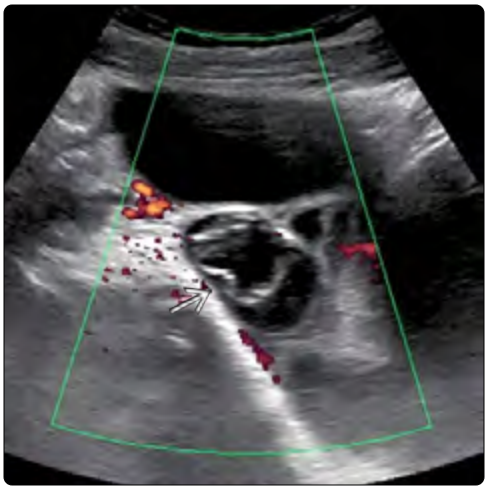
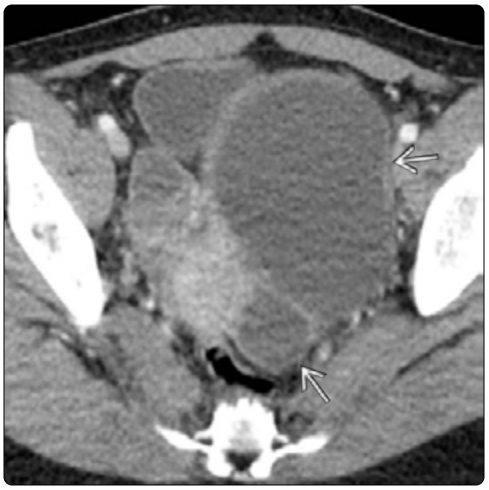


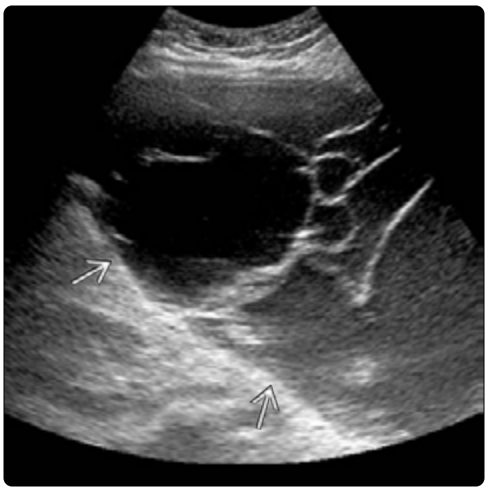


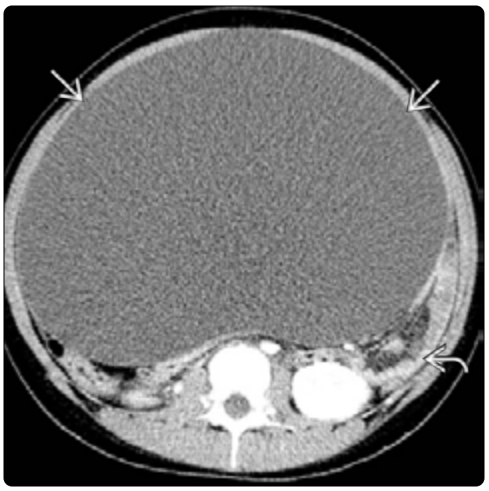
(Left) *Axial T1 C+ FS MR in a young adult with a Sertoli-Leydig cell tumor & a DICER1 gene mutation shows a large, heterogeneous mass ➡ with variable enhancement. Sertoli-Leydig cell tumors cause virilizing symptoms in 30% of patients.* **(Right)** *Coronal CECT in the same patient shows a multiloculated cystic nephroma ➡ in the lower pole of the right kidney. DICER1 gene mutations are associated with pleuropulmonary blastoma, multilocular cystic nephroma, Sertoli-Leydig cell tumor, & thyroid carcinoma.*





(Left) *Transverse US in a young adult with a borderline serous tumor shows a complex mass ➡ deep in the pelvis. Borderline epithelial ovarian tumors are more common in children than adults.* **(Right)** *Axial CECT in the same patient shows a multilocular cystic mass ➡ arising from the left ovary. Epithelial tumors typically appear as unilocular or multilocular cystic masses with variable amounts of papillary projections.*





(Left) *Longitudinal ultrasound in an adolescent with a mucinous cystadenoma shows a large, multiseptated cystic mass ➡. Mucinous cystadenomas are benign lesions that cannot be differentiated from malignant lesions on imaging.* **(Right)** *Axial CECT in a patient with a serous cystadenoma shows a massive cystic mass ➡ of the abdomen. The mass displaces the bowel ➡ posteriorly. Serous cystadenomas are epithelial tumors, which characteristically appear as a homogeneous cystic mass with thin walls or septations.*

Ovarian Torsion

KEY FACTS

TERMINOLOGY

- Definition: Twisting of vascular pedicle of ovary, fallopian tube, or both → venous obstruction → edema → arterial compromise → ischemia → hemorrhagic infarction

IMAGING

- Unilaterally enlarged ovary
  - Ovarian volume > 100 mL highly suggestive of torsion
  - Ovarian volume < 20 mL in postpubertal patient never torsed in 1 series
  - Ratio of abnormal to normal ovarian volumes ≥ 5:1 strongly correlated with torsion in same series
- Scattered predominantly peripheral follicles of 8-12 mm
- Ovary often displaced to midline
  - May extend into abdomen or contralateral pelvis
- Sonographic whirlpool sign of twisted vascular pedicle
- Variable patterns of Doppler flow within twisted ovary ranging from normal to completely absent
- Pelvic free fluid or hemoperitoneum

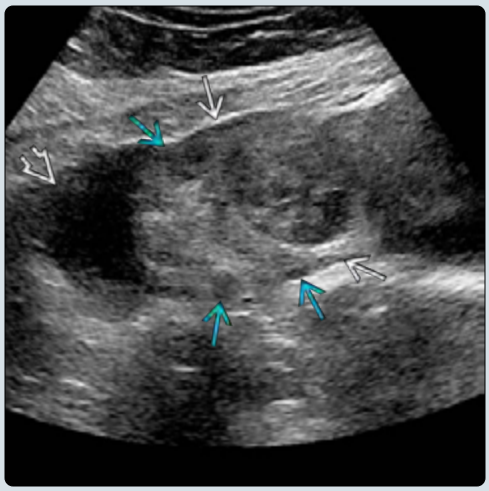
CLINICAL ISSUES

- Urgent surgical detorsion
  - Conservation of ovarian tissue if not frankly necrotic
  - > 90% salvage rate of ovarian function
- Variable rates of retorsion; oophoropexy controversial

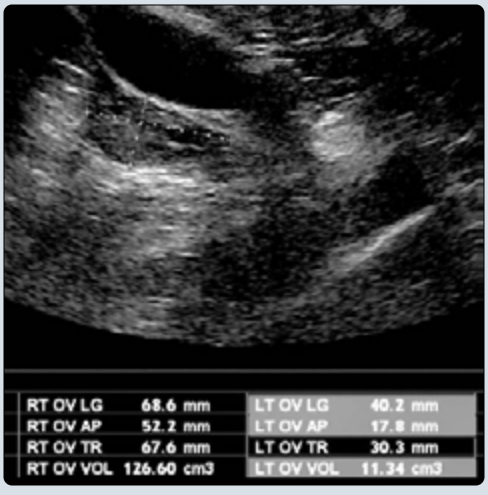
DIAGNOSTIC CHECKLIST

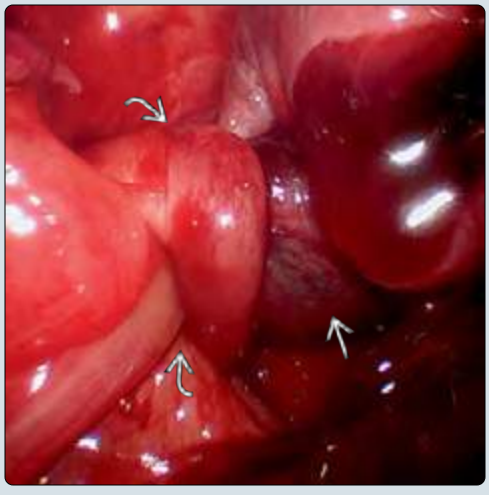
- Painful midline pelvic mass may represent torsed ovary if both normal ovaries not confidently visualized
- Doppler exam of ovarian parenchyma may be normal despite true adnexal torsion
  - Grayscale findings & high clinical suspicion more predictive
- Presence of underlying ovarian cyst/mass can create diagnostic dilemma
  - Experienced clinical evaluation key in these cases
- Normal symmetric grayscale & Doppler appearance of ovaries makes ovarian torsion highly unlikely

(Left) *Graphic shows torsion of the ovarian vascular pedicle & fallopian tube, which results in ischemia of the ovary & distention of the distal segment of the fallopian tube.* **(Right)** *Transverse pelvic ultrasound for acute pain in a 13-year-old girl shows a large 6- to 7-cm heterogeneous mass with adjacent free fluid. A few cysts are present at the outer margin of the mass.*



(Left) *Comparison image of the left adnexa in the same 13-year-old girl shows the normal left ovary between the cursors, with a volume of 11 mL. The painful right-sided mass (her right ovary) was over 10x as large & was torsed 3x at laparoscopy.* **(Right)** *Intraoperative photograph during laparoscopy shows the typical twisting & spiral appearance of adnexal supporting structures. The darker structure deep to the twisted tissue is the torsed ovary.*





TERMINOLOGY

Synonyms

- Adnexal torsion, twisted ovary

Definitions

- Twisting of vascular pedicle of ovary, fallopian tube, or both → venous obstruction → edema → arterial compromise → ischemia → hemorrhagic infarction

IMAGING

General Features

- Best diagnostic clue
  - Unilateral ovarian enlargement with scattered peripheral follicles ± discrete cyst or tumor in setting of acute pain
  - Painful midline pelvic mass with failure to identify ipsilateral normal ovary also very suspicious
  - Doppler exam not sensitive for ovarian torsion
    - Abnormal flow can be useful adjunct to grayscale findings
  - Sensitivity: 79-92% ultrasound; 42% CT (recent meta analysis)
- Location
  - Ovaries usually on either side of uterus; torsed ovary often displaced to midline
    - May extend into abdomen or contralateral pelvis
  - Right ovary affected slightly more often than left
- Size
  - Twisted adnexa: 7- to 200-mL volume (mean 24 mL in recent series); rarely > 2000 mL
  - Twisted ovary generally > 2x size of contralateral ovary
    - Can be much larger, especially with underlying mass
- Morphology
  - Unilateral enlarged ovary with scattered predominantly peripheral follicles ± complex mass or cyst

CT Findings

- Bland, rounded, soft tissue density mass ± cyst
  - Smooth thickening of wall suggests torsion
- Uterine deviation towards twisted adnexa
- Tubal thickening (> 10 mm) + surrounding edema/fluid
- Ascites, which may be hemorrhagic

MR Findings

- Tubal thickening, enlargement of ovarian stroma
- Scattered T2 hyperintense peripheral follicles
- ± T2 hyperintensity of ovarian stroma
- ± foci of T1 hyperintensity from hemorrhage
- ↓ or absent enhancement of ovarian parenchyma
- Surrounding soft tissue edema & fluid

Ultrasonographic Findings

- Grayscale ultrasound
  - Unilateral ovarian enlargement
    - Ovarian volume > 100 mL highly suggestive of torsion
    - Ovarian volume < 20 mL in postpubertal patient never torsed in 1 series
      - Ratio of abnormal to normal ovarian volumes ≥ 5:1 strongly correlated with torsion in same series
      - Median torsed to normal ovarian volume ratio of 12:1 in another series
  - Scattered, predominantly peripheral follicles of 8-12 mm in enlarged ovary
    - Moderately sensitive, highly specific for torsion
    - Different than numerous "string of pearls" peripheral follicles bilaterally in polycystic ovarian syndrome
    - ± fluid or debris in follicles
  - Variable ovarian parenchymal echotexture depending on presence of
    - Edema vs. hemorrhage/infarction
    - Underlying cystic or solid mass
  - Failure to confidently visualize bilateral normal ovaries
  - Fallopian tube thickening > 10 mm
  - Pelvic free fluid or hemoperitoneum
- Color Doppler
  - Sonographic whirlpool sign of twisted vascular pedicle
    - Round, swirling bull's eye or target appearance of hyper- & hypoechoic rings or stripes ± Doppler flow
    - May only be achieved with endovaginal scanning
  - Variable patterns of flow within twisted ovary ranging from normal to completely absent
    - View any asymmetry in flow with suspicion

Imaging Recommendations

- Best imaging tool
  - Ultrasound: Grayscale findings more reliable (92% sensitive, 96% specific) than Doppler (50% sensitive)
  - Doppler exam of ovary may be normal despite true adnexal torsion
    - Theories include: Dual blood supply (ovarian & uterine arteries), venous thrombosis with secondary arterial compromise, intermittent torsion, partial torsion that impairs only venous outflow
- Protocol advice
  - Endovaginal scanning in patients who are sexually active
  - Transabdominal scanning via well-distended urinary bladder in patients who are not sexually active
    - May need to fill bladder via Foley if patient vomiting & dehydrated

DIFFERENTIAL DIAGNOSIS

Appendicitis

- Noncompressible thick-walled blind-ending tubular structure in right lower quadrant originating from cecum & measuring > 6 mm in diameter with surrounding fat induration
- Appendix more difficult to define with perforation

Ovarian Cyst

- Simple or hemorrhagic cyst may cause pain without torsion
- Avascular internally by Doppler
- Hemorrhagic cyst with reticular septations, debris, nodular clot
- Cyst > 5 cm more likely to be associated with torsion

Isolated Fallopian Tube Torsion

- Dilated fluid-filled tube or paraovarian cystic mass, often with normal adjacent ovary

Pelvic Inflammatory Disease

- Complex tuboovarian fluid collection in setting of cervical tenderness, discharge, & generalized pain typical of pelvic inflammatory disease

Ovarian Tumor

- Discrete heterogeneous cystic &/or solid mass
- Painful rarely from rupture or necrosis vs. true torsion

Ectopic Pregnancy

- Cystic tubal mass ± yolk sac, extrauterine embryonic cardiac activity, surrounding hyperemia, free fluid
- Serum β-hCG result crucial

Distal Ureteral Calculus

- Echogenic stone ± Doppler twinkle artifact, posterior acoustic shadowing, thick-walled hydroureter, abnormal ureteral jet, variable hydronephrosis

PATHOLOGY

General Features

- Etiology
  - Usually spontaneous twist of ovary & fallopian tube
    - Torsion of normal adnexal structures more common in children than adults
  - Developmental abnormalities of fallopian tubes or mesosalpinx (such as excessively long tube or absent mesosalpinx) can precipitate torsion
  - Intrinsic ovarian or tubal disease, tumors, cysts, trauma, or recent surgery also predispose to torsion
    - ~ 50% of pediatric cases associated with dominant cyst or tumor
    - Torsion most likely if mass > 5 cm

Microscopic Features

- Peripheral cysts reflect ovarian congestion & transudation of fluid into follicles

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute, severe unilateral lower abdominal/pelvic pain, constant or intermittent
  - Nausea & vomiting (often synchronous with pain)
- Other signs/symptoms
  - Low-grade fever
  - Tender palpable mass
  - Incarcerated inguinal hernia

Demographics

- Age
  - Mean: 10-11 years
  - 50% of cases occur in premenarchal girls
  - ~ 10% occur in perinatal period
- Epidemiology
  - Annual incidence: 1 in 20,000 females ages 1-20 years

Natural History & Prognosis

- Infarction → nonfunctioning ovary → infertility risk
- Variable retorsion rate after detorsion
  - Higher retorsion rate in prepubertal girls
- Asynchronous bilateral ovarian torsion in 5-10%

Treatment

- Urgent surgical detorsion
  - Conservation of ovarian tissue increasingly adopted
    - Oophorectomy now uncommon, even if necrotic-appearing
  - Oophoropexy (to prevent retorsion) controversial
- Salvage rates better for ovarian than testicular torsion
  - > 90% salvage rate for ovarian function
  - Length of symptoms prior to surgery does not always predict viability
  - Appearance at surgery also not predictive of salvage
- Aspiration ± biopsy, resection of cyst or solid lesion

DIAGNOSTIC CHECKLIST

Consider

- Painful midline pelvic mass may represent torsed ovary if 2 normal ovaries are not confidently visualized

Image Interpretation Pearls

- Documenting normal arterial & venous flow in adnexa does not exclude torsion
  - Grayscale findings & high clinical suspicion more predictive
- Presence of underlying ovarian cyst/mass can create diagnostic dilemma
  - Hemorrhagic cyst may be painful in absence of torsion despite causing overall ↑ ovarian volume
  - However, cyst/mass also predisposes to torsion & may obscure other classic imaging findings of torsion
  - Experienced clinical evaluation key in these cases
- Completely normal & symmetric grayscale & Doppler appearance of ovaries makes ovarian torsion highly unlikely

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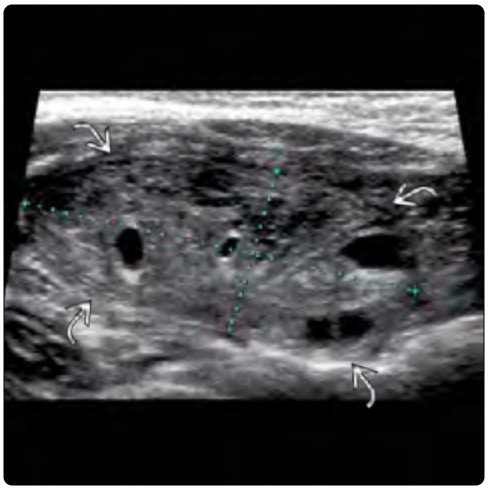
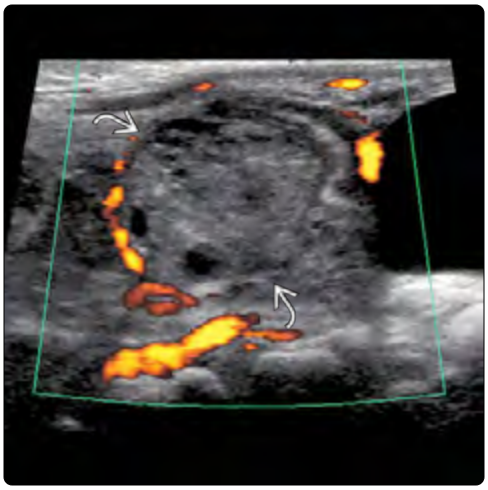
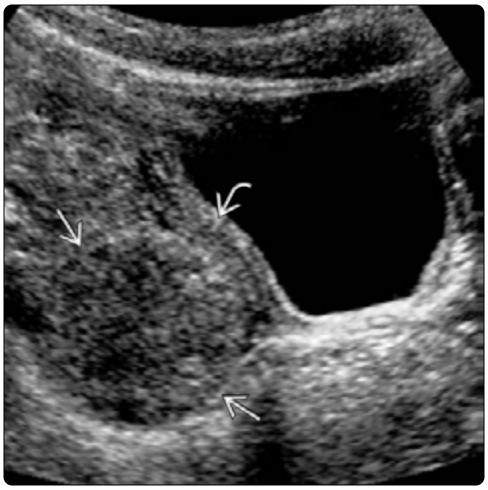
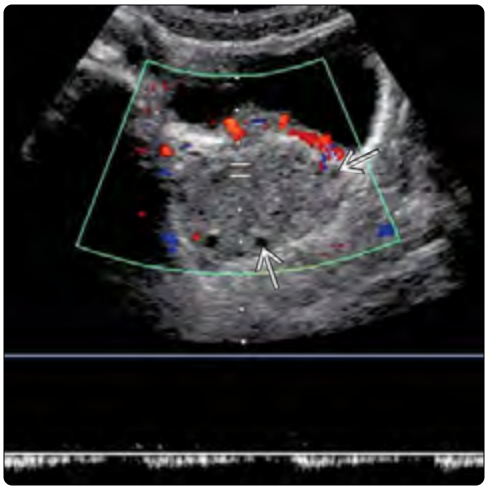


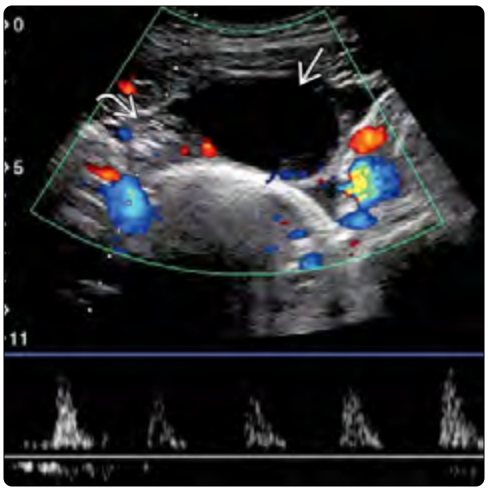


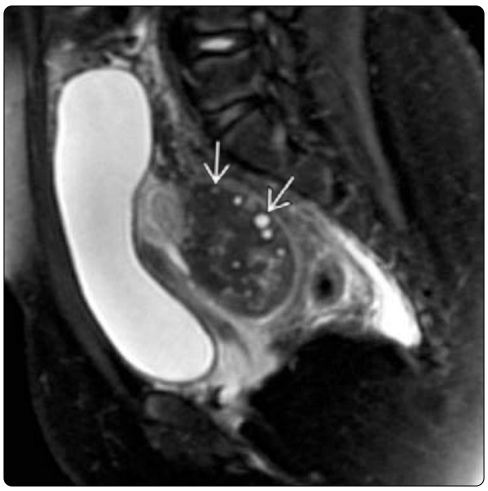
(Left) *Longitudinal oblique ultrasound of the right lower quadrant in an 18-month-old girl experiencing abdominal pain & vomiting was performed to exclude intussusception but showed this mass ➪ containing several cysts just to the right of the bladder.* **(Right)** *Transverse power Doppler ultrasound shows the position of this mass ➪ adjacent to the bladder. No blood flow is seen within the lesion. Since a normal right ovary could not be identified, ovarian torsion was suspected & confirmed surgically.*





(Left) *Midline longitudinal ultrasound in a 7-year-old girl with acute pain shows a round solid mass ➪ posterior to the uterus ➪. No normal right ovary was identified.* **(Right)** *Transverse color Doppler ultrasound in the same patient shows numerous small peripheral follicles ➪ & minimal blood flow within this displaced edematous right ovary. Ovarian torsion was confirmed at surgery.*





(Left) *Longitudinal color Doppler in a 14-year-old girl with acute pelvic pain shows a septated hypoechoic mass ➪ of the right ovary, typical of a hemorrhagic cyst. The ovarian parenchyma ➪ is not edematous but shows high-resistance arterial flow. A single twist was confirmed surgically.* **(Right)** *Sagittal T2 FS MR in a 12-year-old girl with pain & a reported "uterine mass" shows an enlarged, displaced, low signal intensity ovary with peripheral follicles ➪ & surrounding soft tissue edema, consistent with torsion.*

Ectopic Ovary

KEY FACTS

TERMINOLOGY

- Ectopic ovary: Abnormally located ovary
  - Inguinal ovarian hernia (95%): Presents as labial mass
    - Canal of Nuck hernia
  - Intraabdominal ectopia may be incidental or iatrogenic
  - Retroperitoneal location extremely rare

IMAGING

- Labial/groin soft tissue mass with numerous follicles
  - Right > left (60% vs. 30%); 10% bilateral
- US study of choice for palpable inguinal mass (± specific suspicion for ovarian herniation)
  - Use highest frequency transducer available
  - Step-off pad improves visualization of superficial structures (including herniated ovarian follicles)

TOP DIFFERENTIAL DIAGNOSES

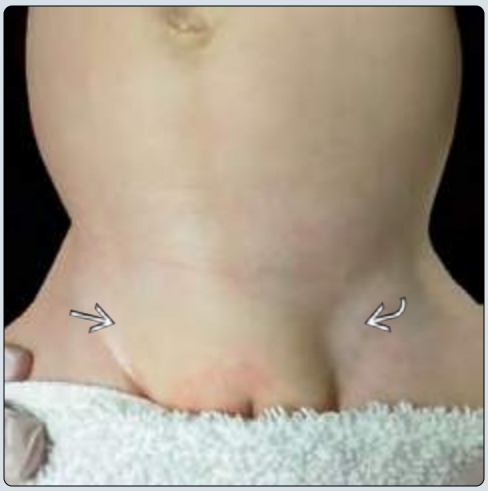
- Abscess
- Inguinal lymphadenopathy
- Bowel-containing inguinal hernia
- Mesothelial cyst of round ligament
- Ovarian torsion

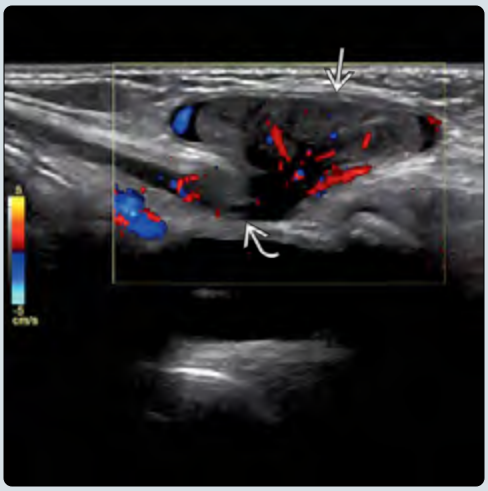
PATHOLOGY

- Canal of Nuck analogous to processus vaginalis in boys
  - May remain patent in up to 10% of term newborns
- Up to 20% with ovarian herniation also have herniation of salpinx, urinary bladder, bowel, &/or uterus
- ~ 20% of herniated ovaries develop strangulation or torsion
- Small association of ovarian herniation with müllerian anomalies

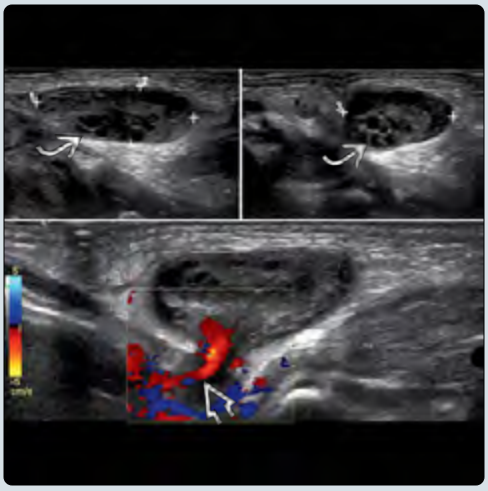
CLINICAL ISSUES

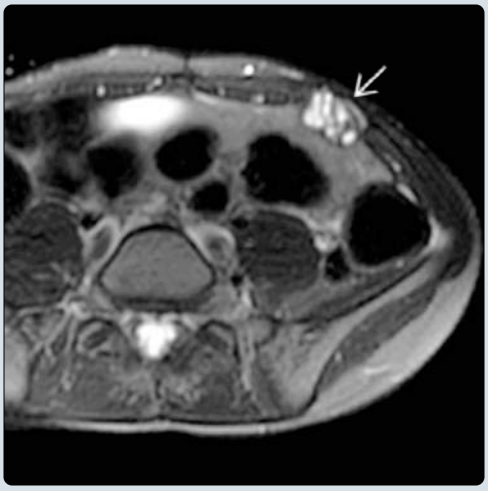
- Inguinal ovarian hernia should be top consideration in any newborn/infant girl with labial or groin mass
- Little morbidity without superimposed strangulation/torsion





(Left) *Clinical photo shows an 8-month-old girl with a new, nonpainful right inguinal mass due to herniation of the right ovary through the canal of Nuck. Note the relative concavity on the normal, contralateral side.* **(Right)** *Longitudinal color Doppler US of the right groin in the same patient shows a normal-appearing ovary in the superficial soft tissues. Note the adnexal vessels extending through the canal of Nuck. Transverse US (not shown) better demonstrated anechoic follicles, confirming that this was the ovary.*





(Left) *Longitudinal (top left) & transverse (top right) US of a palpable labial mass in a newborn shows a normal-appearing ovary (calipers) with numerous anechoic follicles. Oblique longitudinal color Doppler US (bottom) better shows the adnexal vessels of the herniated ovary extending though the canal of Nuck.* **(Right)** *Axial T2 FS MR in a 12-year-old girl with several prior surgeries for an anorectal malformation shows an ectopic left ovary positioned in a prior ostomy site.*

TERMINOLOGY

Definitions

- Ectopic ovary: Abnormally located ovary
  - Inguinal ovarian hernia (95%): Presents as labial mass
  - Intraabdominal ectopia: Incidental or iatrogenic
    - Nonstandard ovarian location common & usually incidental (due to long ovarian ligaments)
    - Nonpelvic location sometimes seen after numerous or complex abdominopelvic surgeries
    - Ovarian transposition sometimes performed to preserve fertility prior to radiation therapy
  - Retroperitoneal location extremely rare

IMAGING

General Features

- Best diagnostic clue
  - Oval soft tissue mass with numerous follicles in unexpected location for ovary
- Location
  - Inguinal, groin, or labial mass (95%)
    - Right > left (60% vs. 30%); 10% bilateral
- Size
  - Ovarian size remains normal for age (0.5-3.0 cm)

Ultrasonographic Findings

- Grayscale ultrasound
  - Typical ovarian morphology: Oval soft tissue mass with several round anechoic follicles scattered throughout
- Color Doppler
  - Look for ↓, absent, or otherwise abnormal blood flow as sign of ovarian torsion/strangulation
    - Grayscale findings actually more sensitive

MR Findings

- T2 bright follicles easily recognized on MR

Imaging Recommendations

- Protocol advice
  - Newborn or infant with groin or labial mass
    - US with highest frequency transducer available
    - Step-off pad further ↑ visualization of superficial structures (such as herniated ovarian follicles)
- Suspected intraabdominal ovary in older child
  - Multiplanar T2 MR through pelvis
  - Preceding ovarian stimulation aids identification

DIFFERENTIAL DIAGNOSIS

Abscess

- Complex fluid collection without internal blood flow
- Look for other signs of inflammation/infection: Cellulitis, erythema, exquisite pain, fever, etc.

Lymphadenopathy

- Usually multiple hypoechoic oval/reniform masses
- Look for hilar vessels & echogenic fatty hilum
  - May be absent with necrosis or infiltration

Bowel-Containing Inguinal Hernia

- Most common labial mass, often easily reducible
- Look for classic bowel wall US signature & peristalsis

Mesothelial Cyst of Round Ligament

- Benign cyst of round ligament within inguinal canal
- Simple, unilocular cystic mass; may be complex if infected
- Rare; analogous to hydrocele of spermatic cord in boys

Ovarian Torsion

- Swollen, torsed ovary (without herniation) may be displaced to midline or contralateral pelvis
- Failure to visualize 2 normal ovaries in patient with pain & "pelvic mass" → torsion until proven otherwise

PATHOLOGY

General Features

- Canal of Nuck analogous to processus vaginalis in boys
  - Evagination of parietal peritoneum into inguinal canal
  - Normally loses connection with peritoneal cavity but may remain patent in up to 10% of term newborns
  - Up to 20% with ovarian herniation also have herniation of salpinx, urinary bladder, bowel, &/or uterus

Gross Pathologic & Surgical Features

- Small association with müllerian anomalies
- ~ 20% of herniated ovaries strangulated or torsed

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless, palpable, mobile mass in groin or labia majora
  - Pain & vomiting common with superimposed strangulation or torsion

Natural History & Prognosis

- Good prognosis with little morbidity if not strangulated

Treatment

- Relatively urgent reduction, hernia repair, & oophoropexy
- Emergent reduction for ovarian strangulation/torsion

DIAGNOSTIC CHECKLIST

Consider

- Always consider ectopic ovary in girls with labial/groin mass
- ~ 10% of term newborns have metachronous hernias

Image Interpretation Pearls

- Look for rare associated müllerian duct anomalies (solitary or pelvic kidney, uterine anomalies, etc.)

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Epididymoorchitis

KEY FACTS

TERMINOLOGY

- Infectious inflammation of epididymis, testicle, or both
- Orchitis alone much less common than epididymoorchitis

IMAGING

- Enlargement of affected tissues (i.e., testicle, epididymis, or both) with accompanying ↑ blood flow
  - ↑ blood flow best demonstrated on transverse side-by-side comparison view
  - Arterial waveforms typically remain low resistance
- Echotexture may be ↑ or ↓, often heterogeneous
- Reactive hydrocele
- Scrotal wall also thickened

TOP DIFFERENTIAL DIAGNOSES

- Torsion of appendage testis
- Testicular torsion
- Scrotal cellulitis
- Hernia

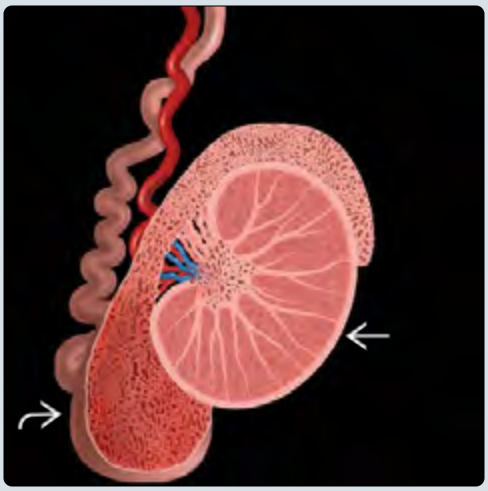
PATHOLOGY

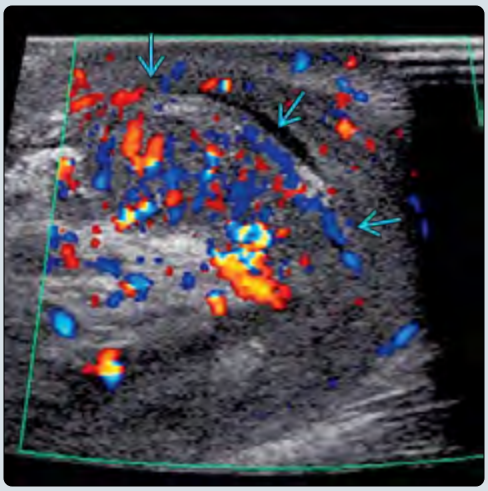
- Bacterial infections may be due to ascending infection (in sexually active adolescents), direct seeding of infected urine with genitourinary (GU) anomalies (especially in young children), or hematogenous seeding
- Can also be viral (typically mumps) or posttraumatic
- Some cases of epididymitis likely due to unrecognized appendage torsion

CLINICAL ISSUES

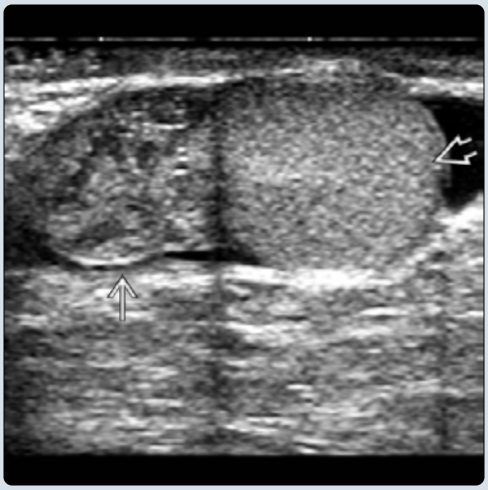
- Gradual onset of painful scrotum, swelling, erythema ± dysuria, enuresis, frequency
  - Prehn sign: Elevation of affected hemiscrotum relieves pain of epididymitis & exacerbates pain of torsion
- Primary therapy: Antibiotics
- Bedrest, scrotal support & elevation, ice packs, antiinflammatory agents, & analgesics also used
- Consider work-up for GU anomalies in younger children & recurrent cases

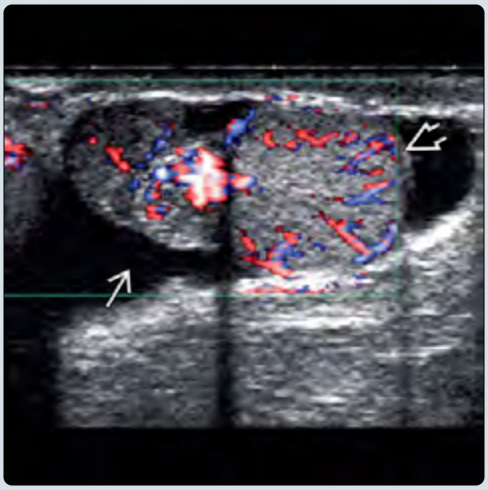
(Left) *Longitudinal section through the testis & epididymis shows a focally enlarged inferior aspect of the epididymis due to epididymitis. If the testicle were also enlarged & inflamed, this would be epididymoorchitis.* **(Right)** *Longitudinal oblique color Doppler US shows a thickened, hyperemic, & heterogeneous epididymis in a teenager with several days of pain, consistent with epididymitis.*





(Left) *Grayscale US in a 5-year-old boy with acute left scrotal swelling shows marked enlargement & heterogeneous echotexture of the epididymis. The testicle has a normal echotexture but was larger than the contralateral side.* **(Right)** *Longitudinal color Doppler US of the same 5-year-old boy with left scrotal swelling shows enlargement of the epididymis & testis with increased blood flow & a surrounding hydrocele, consistent with epididymoorchitis.*





TERMINOLOGY

Synonyms

- Acute scrotum, epididymitis, orchitis, mumps

Definitions

- Infectious inflammation of epididymis, testicle, or both
- Orchitis alone much less common than epididymoorchitis

IMAGING

General Features

- Best diagnostic clue
  - Enlargement of affected tissues (i.e., testicle, epididymis, or both) with accompanying ↑ blood flow & small reactive hydrocele
- Location
  - Entire hemiscrotum may show inflammatory changes
  - Acute epididymitis bilateral in 5-10% of patients

Ultrasonographic Findings

- Grayscale ultrasound
  - Inflamed organs typically show ↑ size when compared to asymptomatic side
  - Echotexture may be ↑ or ↓ from normal, often heterogeneous
  - Reactive hydrocele common
  - Scrotal wall also thickened
- Color Doppler
  - Hyperemia of involved tissues
    - Flow in testis typically remains low resistance
  - ↑ blood flow often best demonstrated on transverse side-by-side comparison view
  - Testicular blood flow can sometimes be difficult to demonstrate in very young males
    - Flow easy to see in cases of epididymoorchitis
- Power Doppler
  - Doppler flow dramatically ↑

Fluoroscopic Findings

- Voiding cystourethrogram
  - May be performed in infants & nonsexually active boys to exclude underlying genitourinary (GU) anomaly
    - Ectopic ureter, vesicoureteral reflux
    - Urethral abnormality with reflux into vas deferens
    - Voiding dysfunction/high-pressure voiding pattern
  - Trend toward less imaging work-up with presumed viral etiology

Nuclear Medicine Findings

- Nuclear scintigraphy
  - Historical interest only (now replaced by US)
  - Radiotracer of choice: Tc-99m pertechnetate
  - Scan shows hyperemia with ↑ radiotracer accumulation & enlargement; occasional cold rim of large hydrocele

MR Findings

- MR urography if complex GU anomalies suspected & not fully elucidated by US & VCUG

Radiographic Findings

- No role for radiography except in cases of penetrating injury or suspected foreign body

Imaging Recommendations

- Best imaging tool
  - Ultrasound with Doppler
- Protocol advice
  - High-frequency linear transducers best
  - Supporting scrotum on towel(s) may be helpful

DIFFERENTIAL DIAGNOSIS

Torsion of Appendage Testis

- Nodular hypo- or hyperechoic avascular mass adjacent to testis with surrounding hyperemia
- May be most common cause of acute scrotum in childhood
  - Some cases of epididymitis likely due to unrecognized appendage torsion

Testicular Torsion

- Unilateral testicular blood flow diminished or absent
- May see twist or knot of spermatic cord above testis
- Surgical emergency
- Note that intermittent torsion may show hyperemia; look for other clues including
  - Abnormal lie of testis
  - Hydrocele completely surrounding testis (suggesting bell-clapper deformity)
  - High-resistance arterial waveforms in setting of "improved pain"

Traumatic Rupture of Testicle

- Focal parenchymal heterogeneity with contour deformity & interruption of echogenic tunica albuginea
- History typically obvious

Scrotal Cellulitis

- Generalized wall thickening & hyperemia
- Normal testicle & epididymis
- May be infectious, allergic, related to insect bite, or sign of Henoch-Schönlein purpura

Scrotal Hernia

- Herniated contents continuous with peritoneal cavity through inguinal canal
- Look for bowel wall signature & peristalsis

Leukemia

- Unilateral or bilateral enlargement
- Parenchyma may be hypoechoic diffusely with palisading vessels

PATHOLOGY

General Features

- Etiology
  - Ascending GU tract infection in sexually active adolescents
    - In males ages 14-35 years, disease most frequently caused by *Neisseria gonorrhoeae* & *Chlamydia trachomatis*
    - Retrograde passage of infected urine from prostatic urethra to epididymis via ejaculatory ducts & vas deferens
  - Bacterial seeding may also occur
    - Directly in cases with GU anomaly

- Hematogenously in cases without demonstrable anomaly
- Most often caused by *Staphylococcus aureus*, *Escherichia coli*, or viruses, especially mumps
  - Mumps orchitis often has fever, malaise, & myalgia
  - Parotiditis precedes onset of orchitis by 3-5 days
  - Subclinical infections occur in 30-40% of patients
  - Outbreaks of mumps reported in school age children, despite mumps, measles, & rubella (MMR) vaccination
  - Mumps epididymoorchitis most often has focal swelling of epididymal head with head:tail ratio > 2
- Associated abnormalities
  - When seen in infants & children, search for predisposing GU anomaly
    - Ectopic ureter
    - Ectopic vas deferens
    - Prostatic utricle
    - Urethral duplication
    - Posterior urethral valves
    - Urethrorectal fistula
    - Detrusor sphincter dyssynergia
    - Vesicoureteral reflux
    - Neurogenic bladder
    - Anorectal malformation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Gradual onset of painful scrotum, swelling, erythema
  - Systemic symptoms of fever, nausea, vomiting
  - Urinary symptoms of dysuria, enuresis, frequency
- Other signs/symptoms
  - Prehn sign: Elevation of affected hemiscrotum relieves pain of epididymitis & exacerbates pain of torsion
  - Instrumentation & indwelling catheters common risk factors for acute epididymitis
  - Urethritis or prostatitis can also coexist

Demographics

- Age
  - Adolescents beginning sexual activity
  - In infants & children, consider work-up for underlying urinary tract anomalies
- Gender
  - Males
- Ethnicity
  - No ethnic or racial predilection
- Epidemiology
  - MMR vaccine has reduced incidence of mumps orchitis
  - 1 in 1,000 men affected yearly; frequency in pediatric patients is lower
  - Epididymoorchitis & torsion of testicular appendage > testicular torsion
    - Some cases of epididymitis likely due to unrecognized appendage torsion

Natural History & Prognosis

- Prognosis generally excellent
- Can lead to abscess if not treated
- Can recur in ~ 25%
  - Recurrent episodes can lead to long-term fertility problems
- Term "chronic epididymitis" refers to patients with symptoms lasting ≥ 6 months

Treatment

- Antibiotics mainstay of therapy
  - Antibiotics reserved for culture positive cases; otherwise, viral etiology presumed
  - Bedrest, scrotal support & elevation, ice packs, antiinflammatory agents, & analgesics also used
  - Follow-up scans to exclude abscess if not improving
- Work-up for GU anomalies in younger children & recurrent cases
- Surgical exploration performed
  - If testicular torsion cannot be ruled out
  - If complications occur from acute epididymitis & orchitis
    - Abscess, pyocele, testicular infarction

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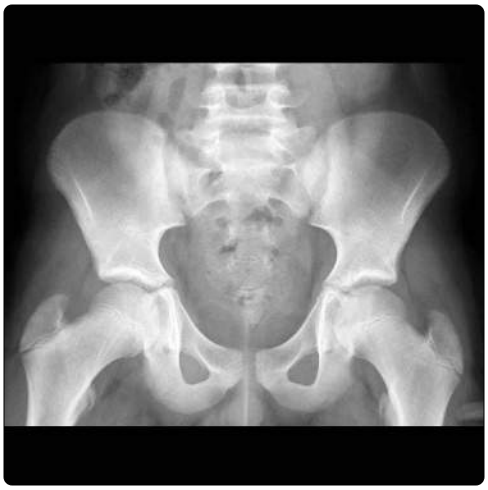
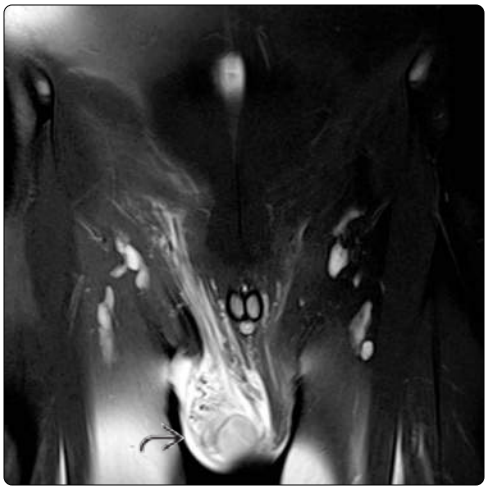
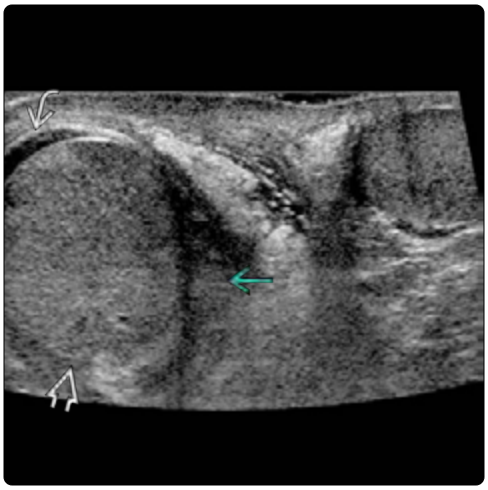
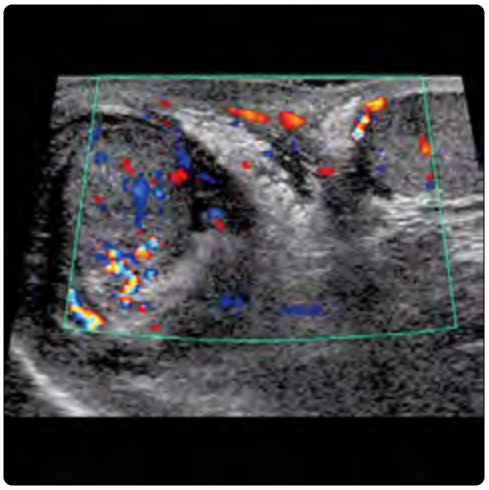


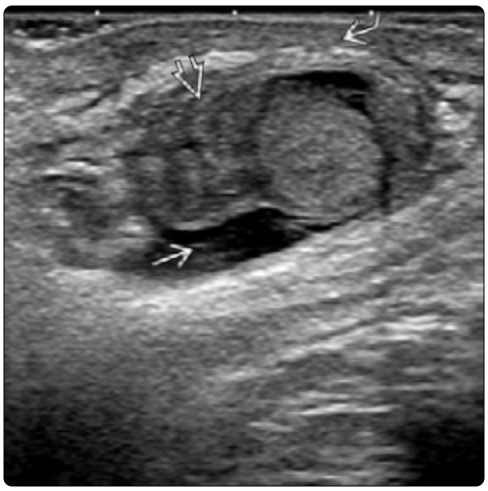


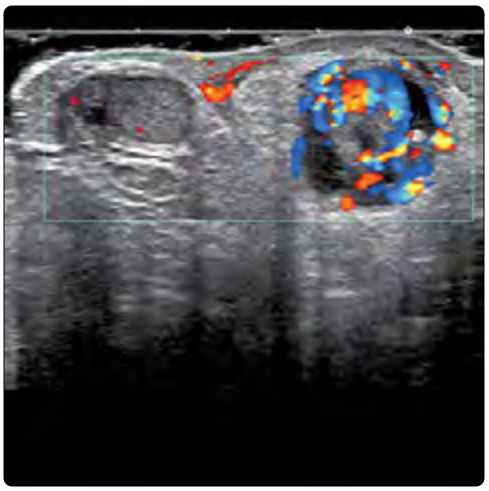
(Left) *AP radiograph in a 12-year-old boy complaining of 1 week of right-sided hip pain is normal. However, the referring service found an elevated erythrocyte sedimentation rate & WBC, so an MR was ordered.* **(Right)** *Coronal T2 FS MR in the same boy shows marked high signal intensity throughout the right hemiscrotum, suspicious for epididymoorchitis or subacute torsion given the duration of his symptoms. The right hip (not shown) was normal.*





(Left) *Transverse side-by-side US performed following the MR shows an enlarged right testicle, hydrocele, enlarged heterogeneous epididymis, & right-sided scrotal wall thickening.* **(Right)** *Transverse side-by-side color Doppler US in the same patient shows hyperemia in all of the right-sided scrotal structures compared to the left, confirming right epididymoorchitis. There was no evidence of testicular torsion.*





(Left) *Transverse US in a 6-year-old boy who had marked scrotal swelling on the left side shows a hydrocele, an enlarged heterogeneous epididymis, & scrotal wall thickening.* **(Right)** *Transverse side-by-side color Doppler US in the same patient shows marked hyperemia of all the left-sided scrotal contents, consistent with epididymoorchitis.*

Testicular Torsion

KEY FACTS

TERMINOLOGY

- Spontaneous or traumatic twisting of testis & spermatic cord within scrotum → vascular occlusion/infarction

IMAGING

- ↓ or absent blood flow in testicle on Doppler US
  - Transverse side-by-side comparison image of asymptomatic & symptomatic testicles very helpful
- "Spiral" twist of spermatic cord just above testis
- ± abnormal lie of testicle within scrotal sac
- Enlarged testis ± altered echotexture
- May see hyperemia after detorsion

TOP DIFFERENTIAL DIAGNOSES

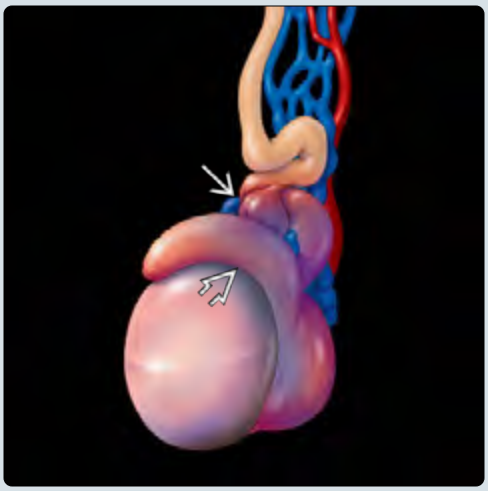
- Epididymoorchitis or orchitis
- Torsion of testicular appendage or appendix epididymis
- Testicular trauma
- Hernia complications

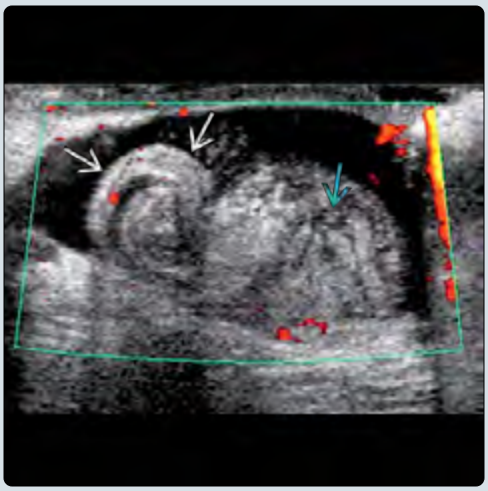
PATHOLOGY

- Intravaginal torsion of spermatic cord
  - Abnormally high attachment of tunica vaginalis → bell clapper deformity
- Extravaginal torsion of spermatic cord
  - Occurs proximal to attachments of tunica vaginalis
  - More common in neonates; rarely salvageable

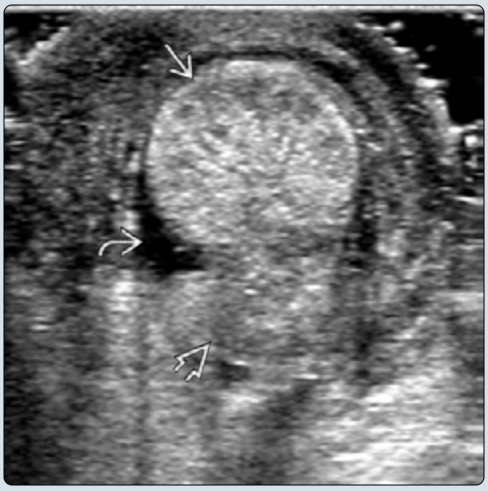
CLINICAL ISSUES

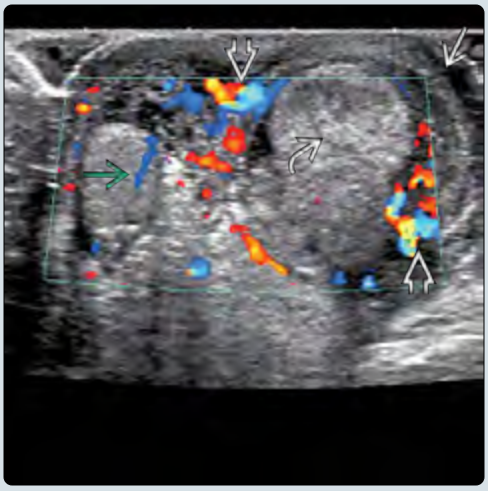
- Presents with acute scrotal &/or inguinal pain
- Surgical emergency to prevent testicular infarction
- Surgical exploration with detorsion & bilateral orchiopexy if testicle viable
  - Nonviable testicle usually removed
- Salvage rates
  - 80-100% within 6 hours of pain onset
  - Virtually 0% after 12 hours of pain onset





(Left) *Anatomic drawing of testicular torsion shows a twisted cord (resembling a snail shell) & an enlarged epididymis. (Right) Longitudinal power Doppler US at the inguinal-scrotal junction shows a twisted spermatic cord surrounded by a hydrocele in a teenager with several hours of left-sided scrotal pain. The epididymis is enlarged & heterogeneous, & no flow was seen in the testicle (not shown), consistent with torsion.*





(Left) *Transverse US of the left scrotum in a 3-year-old boy with swelling & pain shows a heterogeneous echotexture of the left testicle with an enlarged epididymis & a small hydrocele. (Right) Transverse color Doppler US of both testes in the same 3-year-old boy shows scrotal wall thickening & surrounding hyperemia with no left testicular blood flow, confirming left testicular torsion. Flow is seen in the right testicle.*

TERMINOLOGY

Synonyms

- Torsion, late or "missed" torsion, acute scrotum

Definitions

- Spontaneous or traumatic twisting of testis & spermatic cord within scrotum → vascular occlusion/infarction
- Testicle twists medially in 2/3 of cases, so manual detorsion laterally more likely to be effective
  - Direction of manual detorsion described as "opening book"

IMAGING

General Features

- Best diagnostic clue
  - ↓ or absent blood flow in testicle on color Doppler US
    - Easiest to appreciate when compared side by side to normal flow in asymptomatic testicle
- Location
  - Unilateral in 95% of patients
- Size
  - Normal testicular volume
    - 1 mL in newborn
    - 15-20 mL in postpubertal males
  - May ↑ in torsion depending on severity & length of ischemia

Ultrasonographic Findings

- Grayscale ultrasound
  - Testicular parenchyma may be normal early
    - Gradual development of hypoechoic &/or heterogeneous testicular parenchyma
    - Intratesticular necrosis, hemorrhage, or fragmentation seen if delayed diagnosis
  - Enlarged testicle & epididymis
    - Remote torsion shows small testicle ± Ca^{2+}
  - Testicle may lie in abnormal plane
  - Spiral, whirlpool, or knot twist of spermatic cord just above testicle (below inguinal canal)
    - Snail shell-shaped mass measuring 11-33 mm
  - Reactive or secondary hydrocele
  - Scrotal wall thickening
- Color Doppler
  - Absent or ↓ blood flow throughout testicle
    - Compare side by side with asymptomatic testicle
    - Small percentage of patients with early or partial torsion have normal exam
  - ± high resistance residual flow in partially or intermittently torsed testicle
  - Peripheral capsular & scrotal wall hyperemia
  - Hyperemia may be seen in testicle after detorsion
- Sensitivity of 86%, specificity of 100%, accuracy of 97% in diagnosis of testicular torsion when presence of intratesticular flow is sole criterion for diagnosis

Nuclear Medicine Findings

- Tc-99m pertechnetate: Sensitivity 80-90%
  - Technique
    - Dynamic flow imaging at 2- to 5-second intervals for 1 minute (vascular phase)
    - 5-minute intervals for tissue phase
    - Pinhole collimation useful, especially in young patients
    - Penis should be positioned out of field of view, usually secured to anterior abdominal wall
    - Scrotum supported symmetrically on towels
  - "Cold" spot of torsed testicle with surrounding hyperemia

Imaging Recommendations

- Best imaging tool
  - US with high-frequency linear transducer & color Doppler
- Protocol advice
  - Power Doppler with comparison to contralateral normal testis
    - Particularly helpful in neonates & young boys

DIFFERENTIAL DIAGNOSIS

Epididymoorchitis or Orchitis

- Enlarged hypoechoic epididymis with ↑ flow on color Doppler

Torsion of Testicular Appendage or Appendix Epididymis

- Look for round, devascularized, enlarged, heterogeneous appendix with surrounding hyperemia

Testicular Tumor

- Focal intratesticular mass on US; abnormal flow within tumor

Testicular Trauma

- Hematocele, irregular contours, heterogeneous parenchymal echogenicity, interruption of echogenic ± capsule

Inguinal Hernia

- Incarcerated/strangulated hernia can mimic testicular torsion

PATHOLOGY

General Features

- Etiology
  - Most occur spontaneously; occasionally due to trauma
- Embryology/anatomy
  - Intravaginal torsion of spermatic cord
    - Abnormally high attachment of tunica vaginalis → bell clapper deformity
      - Testicle rotates freely within scrotum → spermatic cord twists → venous flow occludes → arterial flow occludes
    - Present in 12% of male population at autopsy
    - Much higher incidence of bell clapper deformity than testicular torsion
  - Extravaginal torsion of spermatic cord
    - Occurs proximal to attachments of tunica vaginalis
    - More common in neonates
    - Accounts for only 5% of all cases of testicular torsion
    - Bilateral in 20%

Staging, Grading, & Classification

- Previously classified as acute, subacute, or delayed based on duration of symptoms
  - Duration of symptoms not always predictive of salvage rate, especially in partial or intermittent torsion

Gross Pathologic & Surgical Features

- Purple, edematous, ischemic testicle; may rapidly reperfuse when manually untwisted
- Varying degrees of ischemic necrosis & fibrosis depending on duration of symptoms

Microscopic Features

- Hemorrhage, interstitial edema, necrosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute scrotal &/or inguinal pain
  - Swollen erythematous hemiscrotum without recognized trauma
  - Physical exam findings highly predictive of testicular torsion include
    - Elevation of affected testicle
    - Transverse position of testicle
    - Anterior rotation of epididymis
    - Absence of cremasteric reflex
    - Pain relief with successful manual detorsion
    - TWIST score 0-6
  - In neonates, purple discoloration of swollen scrotum may indicate extravaginal testicular torsion
    - More common in high birth weight babies
    - Can be confused with scrotal hematoma due to birth trauma
- Other signs/symptoms
  - Nausea & vomiting common
  - Low-grade torsion may be tolerated for long periods
  - Almost 1/2 of patients have history of similar symptoms previously that resolved spontaneously
    - Indicates spontaneous torsion & detorsion

Demographics

- Age
  - Bimodal peak: Teenagers during puberty vs. neonates
- Gender
  - Males only
- Epidemiology
  - Epididymoorchitis & torsion of testicular appendage > testicular torsion
  - 4.5/100,000 males < 25 years of age per year
  - Left side slightly more commonly affected
  - Incidence ↑ in cold weather months: December & January

Natural History & Prognosis

- Surgical emergency: Testicular infarction if not treated promptly
- Testicular viability depends on
  - Degree of torsion: > 540° twist is worse
  - Duration of symptoms & time to surgical intervention
    - ~ 1/3 of testes are not salvageable at operation
- Unilateral testicular loss typically does not lead to infertility problems

Treatment

- Surgical exploration + detorsion + bilateral orchiopexy if viable testicle
  - Nonviable testicle usually removed (due to antisperm antibody theory)
  - Higher risk of subsequent torsion on contralateral side justifies contralateral pexy
- Salvage rates
  - 80-100% within 6 hours of pain onset
  - Virtually 0% after 12 hours
  - Only 9% salvage rate in neonates
- Bilateral pexy advocated in children with intermittent scrotal pain
  - Risk of testicular loss approaches 80% in patients with intermittent torsion
- Note that orchiopexy does not 100% guarantee against future torsion
- Antioxidant medications, heparin, steroids being tested to ↓ ischemia-reperfusion injury

DIAGNOSTIC CHECKLIST

Consider

- Early/partial/intermittent torsion may not show complete absence of flow

Image Interpretation Pearls

- Heterogeneity of avascular testicular parenchyma on US ↓ likelihood of salvageability

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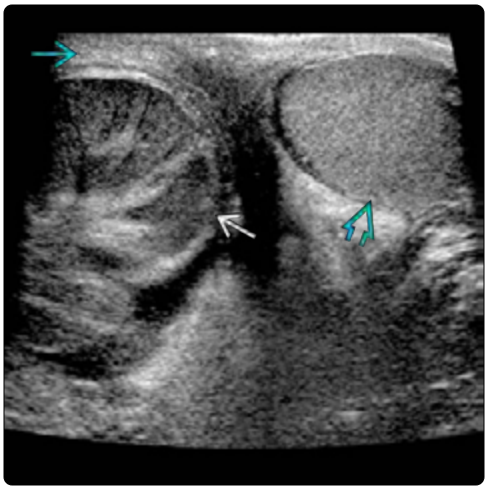
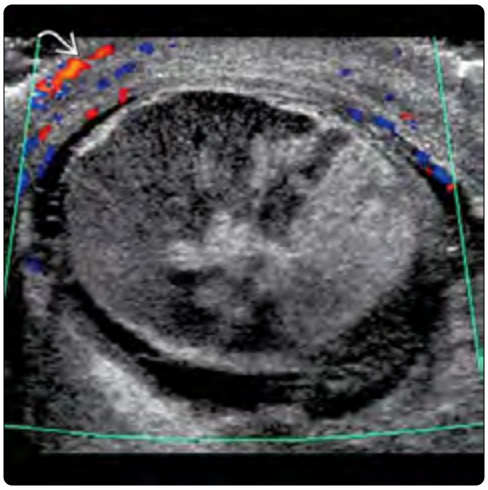
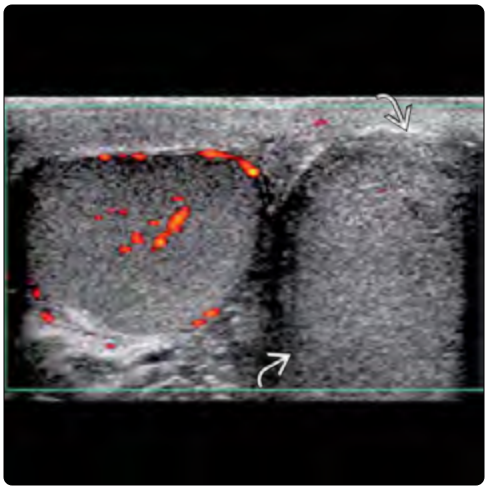
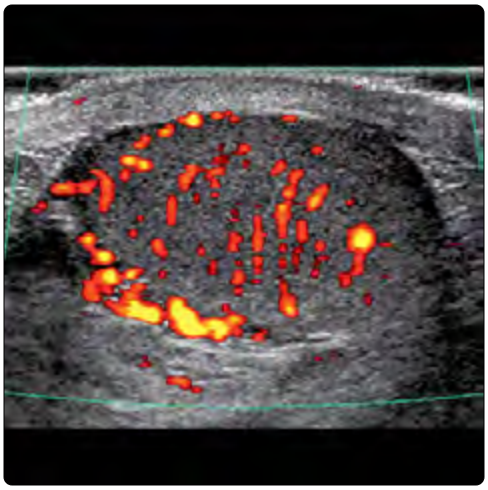


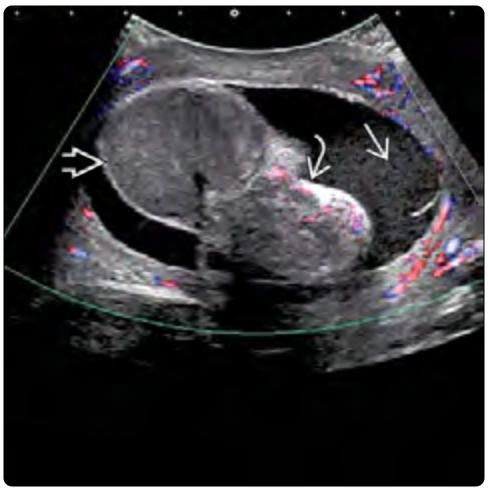


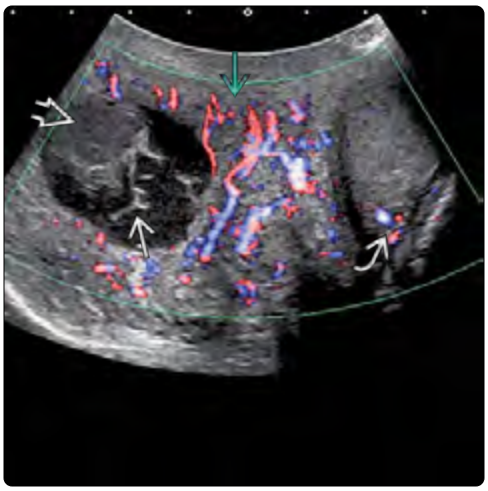
(Left) *Transverse US in a 13-year-old boy with 2 weeks of waxing & waning pain in the right scrotum shows an enlarged right testis with heterogeneous echotexture. There is also a small right hydrocele with scrotal wall thickening. The left testis has a normal homogeneous echotexture.* **(Right)** *Transverse color Doppler US of the symptomatic right testicle shows absent blood flow within the testicle, confirming subacute testicular torsion. Note the mild hyperemia in the thickened scrotal wall.*





(Left) *Transverse power Doppler US in a 14-year-old boy with acute left-sided pain shows absent blood flow in the left testicle, consistent with testicular torsion. After the patient repositioned his scrotal contents on the exam cushions, he exclaimed that he felt much better.* **(Right)** *Repeat power Doppler US of the same patient shows hyperemia in the previously avascular symptomatic testicle, indicating spontaneous/manual detorsion. This testicle remains at risk for retorsion & requires orchiopexy.*





(Left) *Longitudinal color Doppler US in a teenage boy with several hours of right-sided pain & swelling shows a large hydrocele with mobile debris & an avascular enlarged right testis. Note the enlarged epididymis.* **(Right)** *Transverse color Doppler US of both testes in same teenager shows a complex hydrocele, avascular right testis, hyperemia of the thickened scrotal wall, & blood flow in the left testicle. At surgery, the right testis was necrotic & could not be salvaged.*

Torsion of Testicular Appendage

KEY FACTS

TERMINOLOGY

- Synonyms: Twisted appendage, torsed appendix testis, torsion of appendix epididymis, appendiceal torsion
- Definitions
  - Spontaneous twisting of pedunculated vestigial remnant along testicle or epididymis, causing ischemia & pain
  - Most common etiology of acute scrotal pain in children
    - Incidence exceeds epididymoorchitis & testicular torsion

IMAGING

- Ultrasound with Doppler best imaging modality
- Appendage size best indicator of torsion (> 5-6 mm acutely)
- Spherical shape suggests swelling (normally vermiform)
- Duration of symptoms determines echogenicity
  - < 24 hours: Hypoechoic with salt & pepper pattern
  - > 24 hours: Hypo-, iso-, or hyperechoic
- Classically, ↓ or absent internal vascularity of torsed appendix with periappendiceal hyperemia
- Reactive hydrocele & scrotal wall edema common

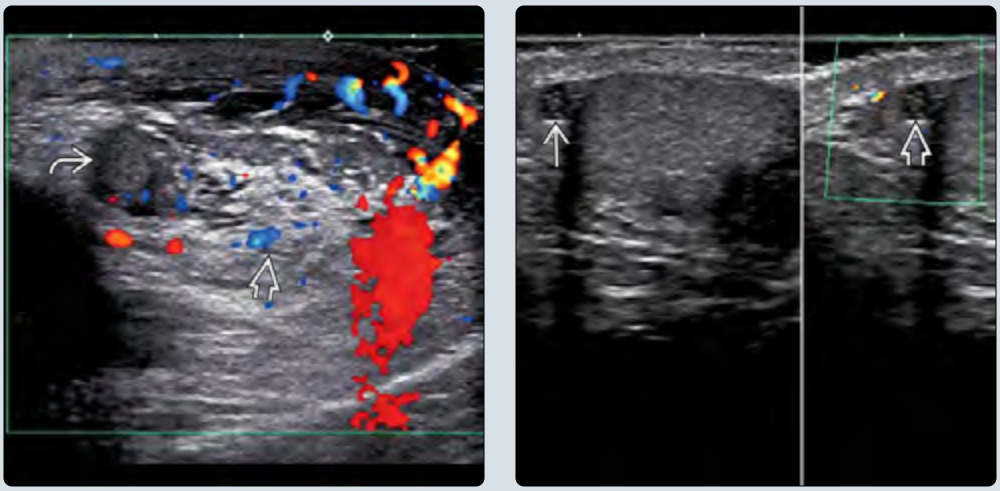
TOP DIFFERENTIAL DIAGNOSES

- Testicular torsion
- Epididymoorchitis or orchitis
- Isolated scrotal wall edema
- Inguinal hernia complications
- Testicular or paratesticular tumor
- Testicular trauma

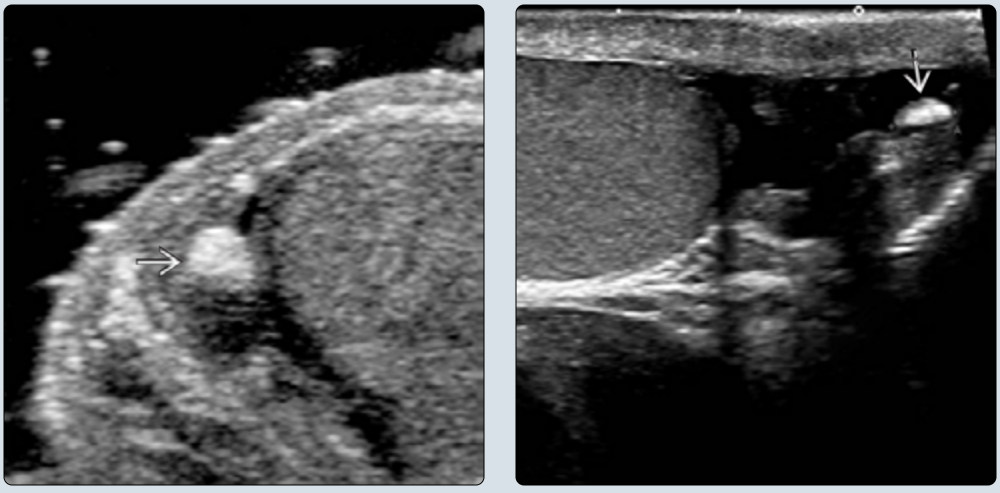
CLINICAL ISSUES

- Acute scrotal pain, swelling
- ± small, tender, mobile lump at upper pole of testis
- ± blue dot sign of ischemic appendage seen through scrotal wall in minority of patients (< 30%)
- 80% of cases 7-14 years old; mean age: 9 years
  - vs. mean age of 14 years for testicular torsion, epididymoorchitis
- Self-limited illness, excellent prognosis

(Left) *Longitudinal color Doppler US in a 7 year old with scrotal pain & swelling shows a lack of blood flow in a small round nodule ➔ adjacent to an inflamed epididymis ➔. The testicular blood flow (not shown) was normal. The findings are consistent with a torsed appendage.* **(Right)** *Longitudinal US images of the epididymis in a 6 year old show a hypoechoic nodule ➔ (superior to the testis) which is avascular on Doppler ➔. This was the point of maximal discomfort & likely represented a torsed appendage.*



(Left) *Oblique scrotal US shows an echogenic appendage ➔ in a patient with a subacute history of pain. The increased echogenicity may reflect acute hemorrhage vs. more chronic fibrotic change or Ca^{2+} in a torsed appendage.* **(Right)** *Longitudinal US shows a hyperechoic ovoid focus ➔ surrounded by a small hydrocele, consistent with a partially calcified, chronically torsed appendage. At this point in time, the patient was asymptomatic.*



TERMINOLOGY

Synonyms

- Twisted appendage, torsed appendix testis, torsion of appendix epididymis, appendiceal torsion

Definitions

- Spontaneous twisting of pedunculated vestigial remnant along testicle or epididymis, causing ischemia & pain
- Most common etiology of acute scrotal pain in children
  - Incidence >> epididymoorchitis & testicular torsion

IMAGING

General Features

- Best diagnostic clue
  - Enlarged, spherical, hypoechoic, avascular nodule along testis or epididymis at site of patient pain + hyperemia of surrounding tissues
    - Reactive hydrocele very common
- Location
  - Most common appendage to twist: Testicular remnant of paramesonephric (müllerian) duct
    - Located between superior pole of testis & epididymis
  - Appendix epididymis is remnant of mesonephric (wolffian) duct
    - Most often seen projecting from head of epididymis
  - Other minor appendages of testicle & epididymis have variable locations & may also twist
  - Can be difficult to differentiate testicular vs. epididymal appendage (not clinically relevant)
- Size
  - Appendage > 5.6 mm in size: Diagnostic of acute torsion
    - Sensitivity of 68.2% & specificity of 100%
    - Normal appendix usually tubular, pedunculated, & 4 mm or smaller
  - Chronically torsed appendage also small
- Morphology
  - Spherical shape & enlargement most reliable indicators of appendiceal torsion
  - Echotexture not predictive of torsion, but ↑ echotexture is associated with longer chronicity

Ultrasonographic Findings

- Grayscale ultrasound
  - Appendix size best indicator of torsion (> 5-6 mm acutely)
  - Spherical shape suggests swelling (normally vermiform)
  - Duration of symptoms determines echogenicity
    - < 24 hours: Hypoechoic with salt & pepper pattern
      - Echogenic dots & septa
    - > 24 hours: Hypo-, iso-, or hyperechoic
  - Reactive hydrocele
  - Scrotal wall edema
  - Epididymal head enlargement
- Color Doppler
  - Classically, ↓ or absent internal vascularity of torsed appendix with periappendiceal hyperemia
    - Findings can be variable
  - Testicular blood flow may be normal or ↑
- Power Doppler
  - Useful in younger, uncooperative patients
    - Sensitive for low flow; no directional information

Nuclear Medicine Findings

- Tc-99m pertechnetate scan
  - Historical interest only: Replaced by ultrasound
  - Normal testicular uptake, excluding diagnosis of testicular torsion
  - May show focal ↑ or ↓ uptake in region of superior testicle at site of twisted appendix
    - Focal "hot" spot equivalent to blue dot sign seen on physical exam
  - Technique same as scan for testicular torsion
    - Pinhole or low-energy, high-resolution collimator
    - Scrotum supported on towels, penis secured up & out of field of view
    - Dynamic acquisition during blood flow or perfusion phase
    - Static delayed imaging ± markers

Imaging Recommendations

- Best imaging tool
  - Ultrasound with Doppler
    - More sensitive & specific for diagnosing appendage torsion than
      - Clinical signs alone
      - Imaging in other causes of acute scrotal pain (i.e., testicular torsion or epididymoorchitis)
- Protocol advice
  - High-frequency linear transducer
  - Thick layer of ultrasound gel ↓ discomfort during scan
  - Support scrotum on towels
  - Compare with asymptomatic side

DIFFERENTIAL DIAGNOSIS

Testicular Torsion

- Abnormal Doppler (absent, diminished, or high-resistance flow) in painful testis compared to asymptomatic side
- Whirlpool or knot sign of twisted spermatic cord above testis
- Normal grayscale appearance of testicular parenchyma early with subsequent changes of frank infarction

Epididymoorchitis/Orchitis

- Hyperemia & enlargement of affected side ± tissue heterogeneity
- Global tenderness on physical exam

Isolated Scrotal Wall Edema

- Thickening, heterogeneity, & hyperemia of scrotal wall
- May be isolated & self-limited or due to cellulitis, insect bite, allergic reaction, or Henoch-Schönlein purpura

Inguinal Hernia Complications

- Normal peristalsing bowel vs. abnormal, thick-walled ischemic bowel tracking from peritoneal cavity through inguinal canal into scrotum
- Pressure effects may cause testicular ischemia in young infants

Testicular or Paratesticular Tumor

- Distortion of normal sonographic anatomy by focal intratesticular or paratesticular mass
- Scrotal tumors range from small & homogeneous to large & heterogeneous
- Usually present with gradual swelling or palpable mass, not pain

Testicular Trauma

- Hematocele, altered testicular echotexture, irregular testicular contour with capsular disruption

PATHOLOGY

General Features

- Etiology
  - Spontaneous twisting of appendage most common, occasionally associated with trauma or tumor
  - Rising levels of estrogen & androgens early in puberty may account for appendiceal enlargement & predisposition to appendiceal torsion in this age group
    - Testicular appendage contains variable numbers of both androgen & estrogen receptors
- Associated abnormalities
  - Rare case reports of tumors arising in scrotal appendages
    - Appendix testis contains müllerian epithelium
    - May produce epithelial tumors similar to those occurring in female genital tract
  - Paratesticular tumors (e.g., rhabdomyosarcoma) likely arise from stromal tissues rather than appendages

Microscopic Features

- Variable degrees of interstitial edema, hemorrhage, & necrosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute scrotal pain & swelling
  - ± small, tender, mobile lump at upper pole of testis
  - ± blue dot sign of ischemic appendage seen through scrotal wall in minority of patients (< 30%)
- Other signs/symptoms
  - Metachronous & bilaterally synchronous cases of appendiceal torsion rarely reported

Demographics

- Age
  - 80% of cases occur between ages 7-14 years
  - Mean age: 9 years
    - vs. mean age of 14 years for testicular torsion & epididymoorchitis
- Gender
  - Males only
- Epidemiology
  - Most common cause (35-67%) of acute scrotal pain
    - Appendix torsion 2.5x more common than testicular/spermatic cord torsion
  - Incidence of 1 in 2,000 males

Natural History & Prognosis

- Self-limited illness, excellent prognosis
- Pain usually resolves within 1 week
- Consider repeat imaging if symptoms persist
  - Rare reports of secondary infection in infarcted, necrotic tissue

Treatment

- Analgesics & antiinflammatory agents for symptom relief
  - Antibiotics not indicated in routine cases
- Reports of manual detorsion with ultrasound guidance
  - Reduction by pulling or squeezing appendage
  - Success if pain relieved, appendix size ↓, Doppler flow restored
- Resection of torsed appendage if scrotum explored due to concern for testicular/spermatic cord torsion
  - Surgery not indicated if torsed appendix diagnosis clear

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Can be difficult to distinguish from epididymitis if no discrete nodule visualized
  - Appendage torsion more common, especially in prepubertal population

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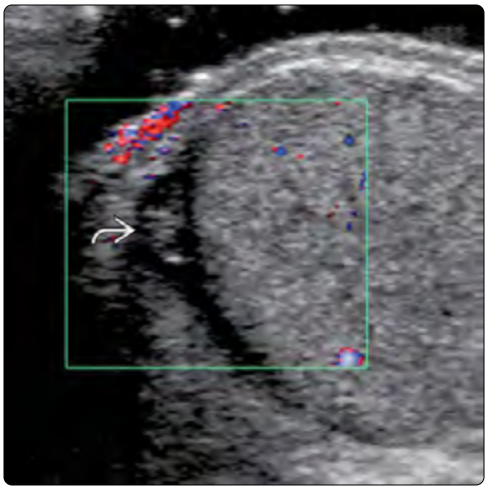
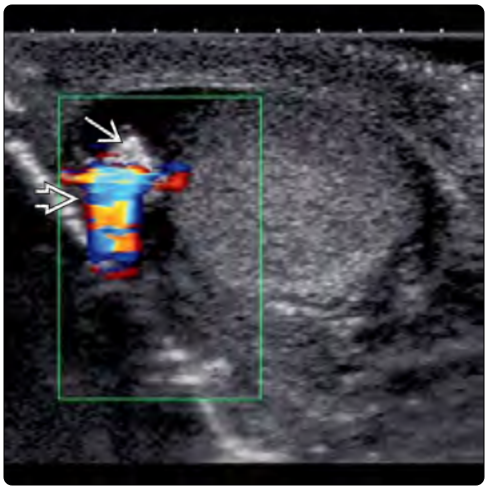
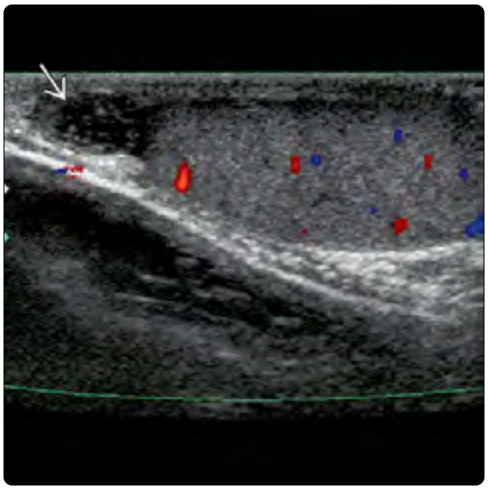
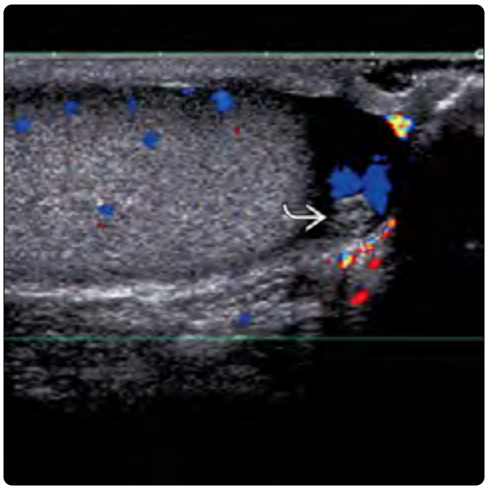


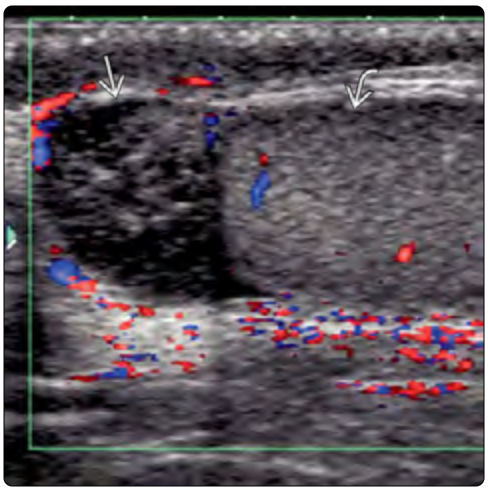


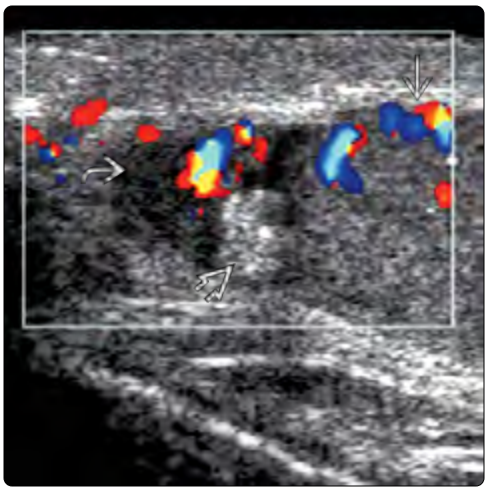
(Left) *Transverse color Doppler scrotal US shows absent flow in a small nodule ➡ along the testicle at the site of patient tenderness, confirming acute torsion of a testicular appendage.* **(Right)** *Transverse color Doppler US through the scrotum in a patient with prior pain shows an echogenic nodule ➡ along the testis with a tiny adjacent hydrocele. There is associated "twinkle" artifact posteriorly ➡. This calcified nodule is consistent with a previously torsed appendage.*





(Left) *Longitudinal color Doppler US in a 12-year-old boy with acute right-sided scrotal pain shows an ovoid, heterogeneously hypoechoic, avascular nodule ➡ along the superior pole of the testicle, consistent with a torsed appendage.* **(Right)** *Longitudinal color Doppler US of the scrotum in a 15-year-old boy with acute left-sided scrotal pain shows a small discrete avascular nodule ➡ at the lower pole of the left testis. There is surrounding hyperemia & an adjacent hydrocele, consistent with appendage torsion.*





(Left) *Longitudinal color Doppler US in a 5-year-old boy with scrotal pain shows a round heterogeneously hypoechoic nodule ➡ along the superior pole of the testicle ➡. Note the internal salt & pepper appearance with surrounding hyperemia but no internal vascularity, consistent with a torsed appendage.* **(Right)** *Sagittal color Doppler US in a patient with pain shows absent blood flow in an echogenic focus ➡ adjacent to a normal-appearing epididymis ➡ & testicle ➡, consistent with a torsed appendage.*

Paratesticular Rhabdomyosarcoma

KEY FACTS

TERMINOLOGY

- Malignant solid tumor of mesenchymal origin; arises from skeletal muscle precursors
- Paratesticular rhabdomyosarcoma (PT-RMS) refers only to intrascrotal rhabdomyosarcoma (RMS), not other GU sites

IMAGING

- Large, lobulated paratesticular mass with variable echogenicity; often heterogeneous & hypervascular compared to testis
  - Usually arises in epididymis; may arise from or invade tunica, testis, or spermatic cord
  - Can be ill defined or well defined; may encase testicle
- Look for retroperitoneal adenopathy

TOP DIFFERENTIAL DIAGNOSES

- Epididymitis
- Inguinal hernia
- Hematoma
- Lipoma
- Polyorchidism
- Testicular adrenal rest tumors

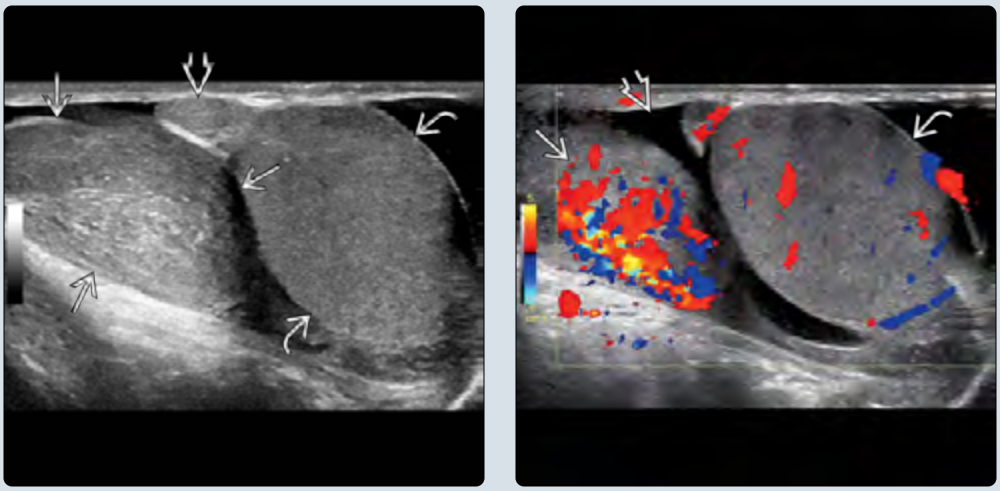
PATHOLOGY

- Most PT-RMS of embryonal RMS subtypes
- Most cases sporadic
- ↑ risk with *TP53* mutations, *DICER1* mutations, RASopathies

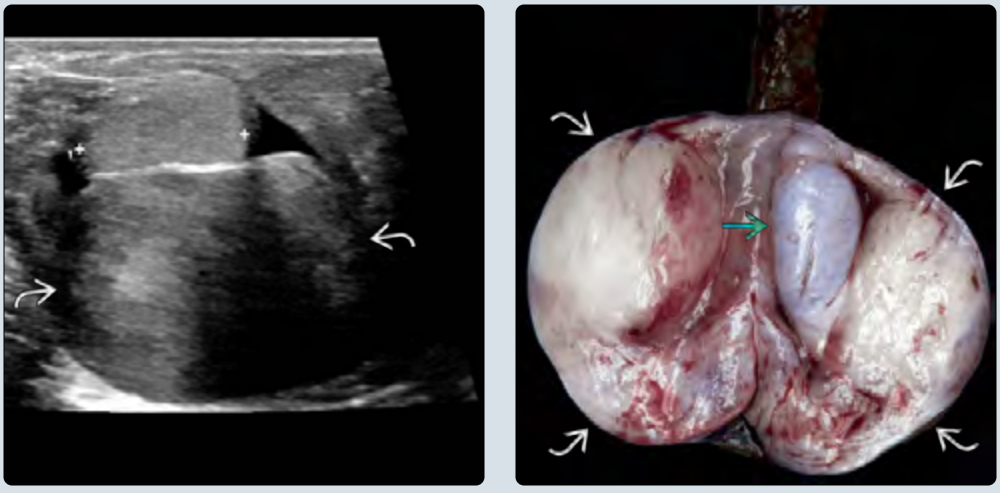
CLINICAL ISSUES

- Gradual, painless scrotal swelling, ± palpable mass
- 2 age peaks: < 5 years vs. 2nd decade
- Most common extratesticular solid mass in boys
- PT-RMS accounts for ~ 5% of childhood RMS but has much better prognosis than other forms of genitourinary RMS
  - > 90% survival rate for localized (nonmetastatic) PT-RMS
  - Age < 1 or > 10 years, tumor size ≥ 5 cm, & lymph node involvement portend worse prognosis

(Left) *Longitudinal oblique US through the left hemiscrotum shows a slightly heterogeneous & hyperechoic paratesticular soft tissue mass. Note the normal testis & epididymis.* **(Right)** *Color Doppler US from the same study reveals markedly increased blood flow within the mass compared to the testis; also note the small hydrocele. This mass was a localized paratesticular rhabdomyosarcoma (PT-RMS).*



(Left) *Scrotal US in a 5 year old with 1 week of right scrotal swelling shows a large, slightly heterogeneous paratesticular mass, ultimately proven to be a PT-RMS. Real-time imaging showed the mass to almost completely surround the testis (shown with calipers).* **(Right)** *Gross pathologic bi-valved specimen from the same patient status post orchiectomy shows a large, bland, tan-white PT-RMS almost completely surrounding, but not invading, the testis.*



TERMINOLOGY

Abbreviations

- Rhabdomyosarcoma (RMS)
- Paratesticular rhabdomyosarcoma (PT-RMS)

Definitions

- Malignant solid tumor of mesenchymal origin arising from skeletal muscle precursors

IMAGING

General Features

- Best diagnostic clue
  - Large, vascular paratesticular soft tissue mass
- Location
  - PT-RMS refers only to intrascrotal RMS; other genitourinary locations of RMS considered separately
  - Usually arises within epididymis but can occur within testicular tunica, testis, or spermatic cord
- Morphology
  - Can be ill defined or well defined, may encase testicle, may invade tunica or testis itself

Ultrasonographic Findings

- Lobulated paratesticular mass with variable echogenicity, often heterogeneous
- Doppler shows ↑ blood flow compared to testes
- May have associated hydrocele

CT Findings

- Chest/abdomen/pelvis CT used to evaluate for lung metastases & retroperitoneal lymphadenopathy (LAD)
- PET/CT improves sensitivity for nodal disease

MR Findings

- Generally not indicated for PT-RMS (vs. RMS) but may be used to evaluate retroperitoneal LAD

DIFFERENTIAL DIAGNOSIS

Epididymitis

- Enlarged, hypervascular epididymis; no distinct mass
- History usually involves painful scrotal swelling

Inguinal Hernia

- Peristalsing bowel loops, omental fat
- Continuity with peritoneal cavity through inguinal canal

Lipoma

- Most common benign paratesticular neoplasm
- Usually within spermatic cord; homogeneously hyperechoic on US & of fat attenuation/signal on CT/MR

Adenomatoid Tumor

- Most common benign tumor of epididymis
- Usually affects young adult men aged 20-30 years
- Typically well-circumscribed, round to oval, homogeneous mass with variable echogenicity

Polyorchidism

- Well-defined mass resembling normal testicle
- Ipsilateral testis often small, may be contiguous
- Identical appearance to testis on all imaging modalities

Testicular Adrenal Rest Tumors

- Multiple, bilateral (usually peripheral) intratesticular masses with variable echogenicity & ↑ vascularity
- Adrenal rests trapped in developing fetal testis
  - 10-15% of newborns; < 1% of adults
  - Enlarge if exposed to ↑ ACTH; otherwise regress

Splenogonadal Fusion

- Extremely rare, almost always left-sided; occurs in association with limb deficiency & micrognathia
- May be in continuity with spleen (through inguinal canal)
- Splenic scintigraphy can be diagnostic

PATHOLOGY

General Features

- Etiology
  - Primitive skeletal muscle precursors within testes, epididymis, & spermatic cord
- Associated abnormalities
  - Most cases sporadic
  - Several genetic diseases have predisposition for RMS

Staging, Grading, & Classification

- RMS consists of 6 histologic subtypes; most PT-RMS of embryonal subtypes
- Staging complex: PT-RMS stratified into 1 of 4 risk groups based on TNM stage, histologic subtype, extent of postoperative residual tumor, & age at diagnosis

Gross Pathologic & Surgical Features

- Tumor spread mostly by lymphatics → retroperitoneal LAD
- Hematogenous spread less common: Liver, lungs, & bones
- Direct invasion of tunica or testicle rare

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Gradual, painless scrotal swelling, ± palpable mass

Demographics

- 2 age peaks: < 5 years vs. 2nd decade
  - Median age 5 years

Natural History & Prognosis

- Most common extratesticular solid mass in boys
- PT-RMS accounts for ~ 5% of childhood RMS but has much better prognosis than other forms of genitourinary RMS
  - > 90% survival rate for localized (nonmetastatic) PT-RMS
  - Age < 1 or > 10 years, tumor size ≥ 5 cm, & lymph node involvement portend worse prognosis

Treatment

- Multimodal treatment based on local control & assessment of any distant involvement: Radical orchiectomy, chemotherapy, ± radiation

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KEY FACTS

TERMINOLOGY

- Testicular neoplasms divided into germ cell & nongerm cell tumors
  - Germ cell tumors: ~ 2/3 of pediatric tumors
  - Nongerm cell tumors: ~ 1/3 of pediatric tumors
  - Lymphoma & leukemia can also involve testes

IMAGING

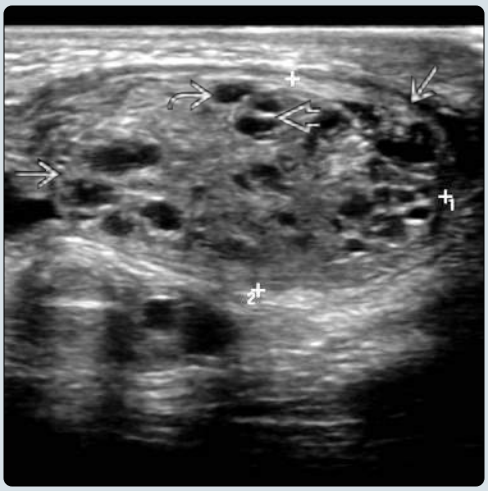
- Intratesticular mass of variable echogenicity & vascularity
- Vary widely in size, from imperceptible to testis-replacing
- Teratomas characteristically have extremely complex appearance with cystic areas, Ca^{2+}, solid components, & punctate or linear echogenic hairs
- Epidermoids (true cysts, not neoplasms) appear solid, targetoid, or have "onion skin" from laminations of keratinizing stratified squamous epithelium
- Regressed MGCTs may appear as subtle architectural distortion with ill-defined microcalcification
- Sex cord-stromal tumors often small & well defined

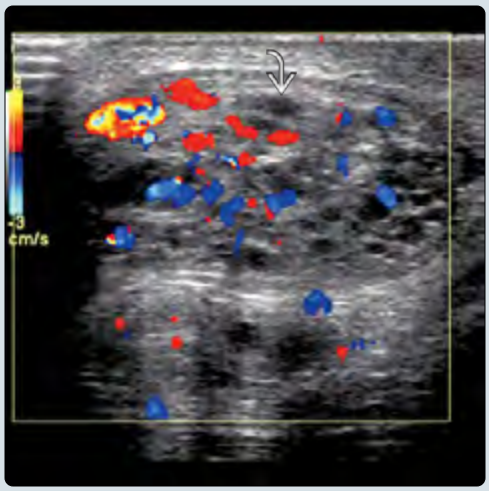
CLINICAL ISSUES

- Represent 1-2% of childhood solid tumors
- Presentations: Asymptomatic or painless mass most common; mild discomfort or pain in abdomen, groin, or testicle; perceived scrotal heaviness
- 10% associated with cryptorchidism
- Most prepubertal intratesticular masses benign (teratoma, epidermoid), though YSTs 2nd most common tumor
  - YSTs: 90% have ↑ AFP, used as tumor marker
- Most postpubertal intratesticular masses malignant: Embryonal carcinoma & MGCT most common
- Standard of care: Orchiectomy; testis-sparing enucleation increasingly performed in newborns & prepubertal boys without ↑ AFP; ± adjuvant chemotherapy depending on tumor type & stage

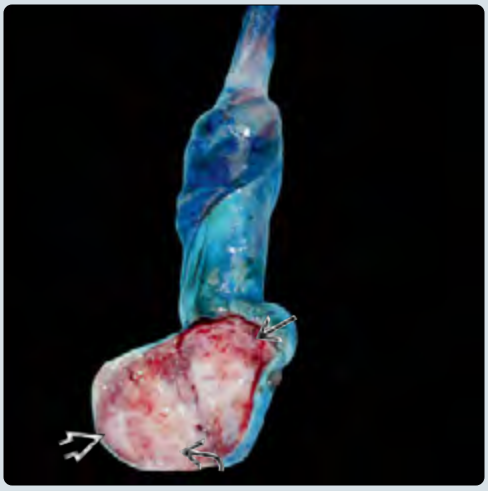
DIAGNOSTIC CHECKLIST

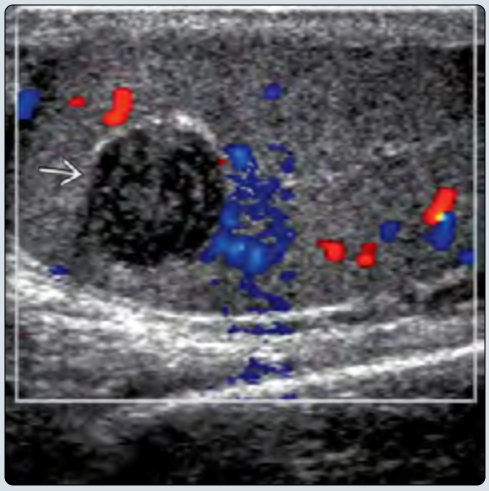
- Use highest frequency linear-array US transducer available





(Left) *Longitudinal US of a juvenile granulosa cell tumor in a newborn with firm testicular enlargement shows a heterogeneous mass replacing the testis. There are numerous hypoechoic foci separated by thin septations, a classic appearance for this rare tumor.* **(Right)** *Color Doppler US in the same patient shows the hypoechoic foci to be cystic, not vascular. As is common in this tumor, low-resistance vascular flow was demonstrated (spectral tracing not shown) within the solid portions & septations.*





(Left) *Gross pathological photograph status post orchiectomy in the same patient shows complete replacement of the testis by a gray-white tumor with scattered cystic spaces & foci of hemorrhagic staining.* **(Right)** *Longitudinal color Doppler US shows a well-defined, spherical, avascular intratesticular mass with a multilaminated onion skin appearance, characteristic of an epidermoid. The mass was treated in this case by enucleation rather than orchiectomy because of the confident imaging diagnosis.*

TERMINOLOGY

Synonyms

- Yolk sac tumor (YST): Endodermal sinus tumor, orchioblastoma
- Teratoma: Differentiated or mature teratoma, undifferentiated or immature teratoma
- Mixed germ cell tumor (MGCT): Polyembryoma
- Leydig cell tumor: Interstitial cell tumor
- Sertoli cell tumor: Androblastoma

Definitions

- Primary testicular tumors: Germ cell vs. nongerm cell tumors
- Germ cell tumors (GCTs): ~ 2/3 of pediatric testicular tumors
  - YST: Histologically recapitulates yolk sac, allantois, & extraembryonic mesenchyme
  - Teratoma: Contains elements of all 3 germinal layers (endoderm, mesoderm, & ectoderm)
    - Mature contains only well-differentiated tissues; immature adds fetal elements
    - Epidermoids: Monodermal cysts (not neoplasms) filled with keratinizing stratified squamous epithelium
  - Embryonal carcinoma: Malignant tumor of anaplastic undifferentiated cells similar to those seen in very early embryo
  - Choriocarcinoma: Rare, highly malignant tumor composed of placenta-like tissues
  - Mixed GCTs: Malignant tumor of 2 or more germ cell types
  - Seminoma: Malignant tumor of germ cell origin lacking any embryonic elements
- Nongerm cell tumors: ~ 1/3 of pediatric testicular tumors
  - Leydig cell tumors: Recapitulate normal development of Leydig cells (stromal cells that produce androgens)
  - Sertoli cell tumors: Recapitulate normal development of Sertoli cells (sex cord cells that support spermatogonial stem cells)
  - Juvenile granulosa cell tumors: Extremely rare stromal tumor that resembles ovarian graafian follicles
- Lymphoma & leukemia can also involve testes

IMAGING

General Features

- Best diagnostic clue
  - Intratesticular mass ± internal vascularity
- Morphology
  - Epidermoids, YSTs, & sex cord-stromal tumors tend to be well defined; others tend to be ill-defined

Ultrasonographic Findings

- Nonspecific appearance for most testicular masses, necessitating surgery
  - Most nonseminomatous germ cell tumors have variable echogenicity
    - ± hypo-, iso-, or hyperechoic; homogeneous vs. heterogeneous
    - Commonly have cystic areas from necrosis & echogenic areas from Ca^{2+}, fibrosis, or hemorrhage
    - More aggressive tumors (especially embryonal carcinoma) may invade tunica albuginea
    - YSTs often well defined, but may simply enlarge testis without discernible mass
  - Teratomas characteristically have extremely complex appearance with cystic areas, Ca^{2+}, solid components, & punctate or linear echogenic hairs
  - Epidermoids (true cyst, not neoplasm) appear solid, targetoid, or have "onion skin" from laminations of keratinizing stratified squamous epithelium
  - Regressed MGCTs may appear as subtle architectural distortion with ill-defined microcalcification
  - Seminomas usually hypoechoic, homogeneous, well-defined intratesticular masses with lobulated margins
  - Sex cord/stromal tumors may be small & well defined
    - Most (2/3) of juvenile granulosa cell tumors are multicystic, multiseptated; remaining 1/3 appear solid
    - Leydig cell tumors usually small (< 1 cm diameter) & very well defined; markedly hypervascular on color Doppler & contrast-enhanced US; stiffer than surrounding parenchyma by elastography

MR Findings

- Nonseminomatous GCTs usually heterogeneous, T1 iso- or hyperintense, T2 hypointense
- Regressed MGCTs have ill-defined areas of low T2 signal or architectural distortion without visible mass
- Seminomas usually homogeneous, T1 isointense, & T2 hypointense
- Look for pelvic & retroperitoneal adenopathy

Imaging Recommendations

- Best imaging tool
  - US ~ 100% sensitive, MR for problem solving
  - Chest (CT) + abdomen & pelvis (CT or MR) screening for metastases, depending on tumor type
- Protocol advice
  - US: Supine positioning with warm folded towel between thighs to support scrotum; use highest frequency linear-array transducer available; color Doppler setting should be sensitive to low-velocity flow
  - MR: Large FOV axial T2 FS & DWI from kidneys through pelvis; small FOV high-resolution T2 FS images in 3 planes, axial GRE, & T1 FS pre- & post gadolinium of scrotum

DIFFERENTIAL DIAGNOSIS

Hematoma

- History crucial, but trauma does not exclude hemorrhage of preexisting neoplasm
- Can be multifocal & any size; color Doppler demonstrates no internal vascularity
- Hyperacute/acute → isoechoic to parenchyma; chronic → becomes hypoechoic/anechoic; 24-hour repeat US can be useful to document evolution

Focal Orchitis

- Present with acute scrotal pain & swelling; concomitant epididymitis common
- Altered echotexture, swelling, hypervascularity
- Appearance improves with short-term follow-up

Epidermoid Cyst

- Ectodermal inclusion cyst; most common benign intratesticular mass
- Laminated, concentric rings of low & high echogenicity or signal, a.k.a. onion skin pattern; no blood flow
- Surgeon may remove by enucleation, not orchiectomy

Testicular Adrenal Rest Tumors

- Adrenal rests (not neoplasm) trapped in developing fetal testis, found in 10-15% of newborns
- Enlarge if exposed to ↑ adrenocorticotrophic hormone, otherwise regress; seen in < 1% of adults
- Multiple, bilateral (usually peripheral), intratesticular nodules
- Variable echogenicity (usually hypoechoic), low T1 & T2 signal
- May be hypervascular but do not distort vessels

Dilated Rete Testes

- Benign dilation of tubules near mediastinum, rare in boys
- Often bilateral; associated with spermatoceles; geographic margins, no mass effect

Segmental Infarction

- Uncommon, may see in sickle cell disease
- Wedge-shaped hypoechogenicity with absent or diminished flow; MR may show hemorrhagic signal around avascular region

PATHOLOGY

Staging, Grading, & Classification

- No universal staging system for pediatric testicular tumors
- Children's Oncology Group stages germ cell tumors
  - Stage I: Limited to testis; complete resection, normal tumor markers
  - Stage II: Invasion of scrotum or spermatic cord, incomplete resection (microscopically), persistent elevation of tumor markers
  - Stage III: Incomplete resection (macroscopically); retroperitoneal lymph node involvement, but no visceral or extraabdominal metastases
  - Stage IV: Distant metastases

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic; painless (rarely painful) testicular mass; mild discomfort or pain in abdomen, groin, or testicle; perceived scrotal heaviness
- Other signs/symptoms
  - YST: 90% have ↑ α-fetoprotein, used as tumor marker
  - Choriocarcinoma: Most have ↑ β-hCG; diagnosis usually due to result of hematogenous metastasis
  - Sertoli cell tumors: Painless mass in infants; may have gynecomastia
  - Leydig cell tumors: Boys aged 5-10 years; may elaborate testosterone or have ↑ estrogen & estradiol levels → precocious puberty or gynecomastia
  - Juvenile granulosa cell tumors: Most frequent congenital testicular neoplasm

Demographics

- Represent 1-2% of childhood solid tumors
- ~ 1 per 100,000 boys; most common malignancy in postpubertal young men (15-34)
- 2 age peaks: First 2 years of life & late adolescence
  - Most prepubertal masses benign (teratoma, epidermoid), though YSTs 2nd most common tumor
    - YST: Most common pure germ cell tumor
    - Teratoma: Increasingly reported as most common prepubertal tumor; most present in 1st year of life
      - Prognosis depends age rather than presence of mature or immature histologic features
  - Most postpubertal masses malignant: Embryonal carcinoma & MGCT most common
    - Embryonal carcinoma: Very rare prior to puberty
    - Seminoma: Extremely rare prior to puberty; usually men
- 10% associated with cryptorchidism
  - If orchiopexy before puberty, then relative risk 2x compared to general population
  - If orchiopexy after puberty, then relative risk 5x compared to general population

Natural History & Prognosis

- YST: Most in infants are stage I → orchiectomy only, > 80% disease free at 6 years; recurrence has excellent response to platinum therapy
- Teratoma: Age predicts prognosis → benign in prepubertal testes, even if immature; 1/3 malignant in postpubertal teratomas, even if mature
- Choriocarcinoma: Seen in older adolescents & young men, usually with advanced disease
- Juvenile granulosa cell tumors: Benign & hormonally inactive; 20% associated with Y-chromosomal &/or urogenital abnormalities

Treatment

- Standard of care: Orchiectomy; testis-sparing enucleation may be performed in newborns & prepubertal boys without ↑ AFP
- YST & teratomas in prepubertal boys may need no further treatment beyond orchiectomy

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Use highest frequency linear array US transducer available
- MR for problem-solving or questionable masses

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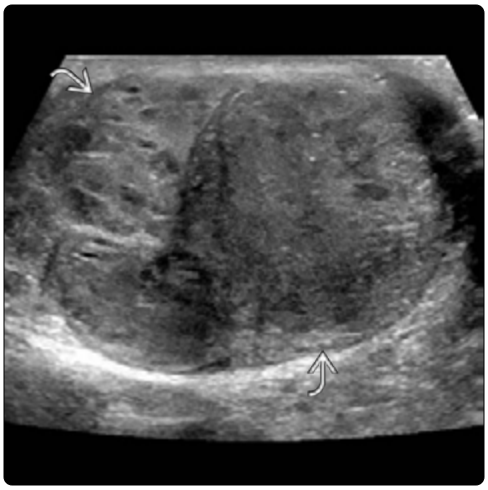
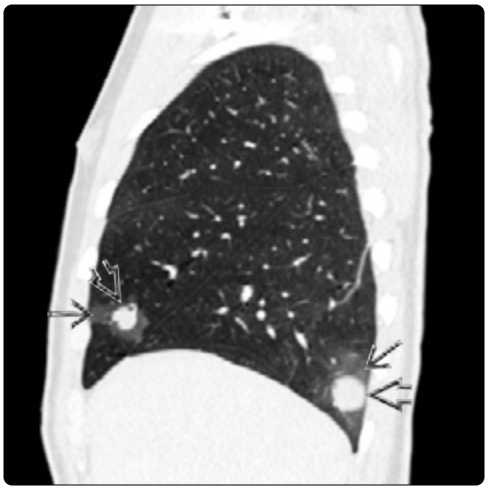
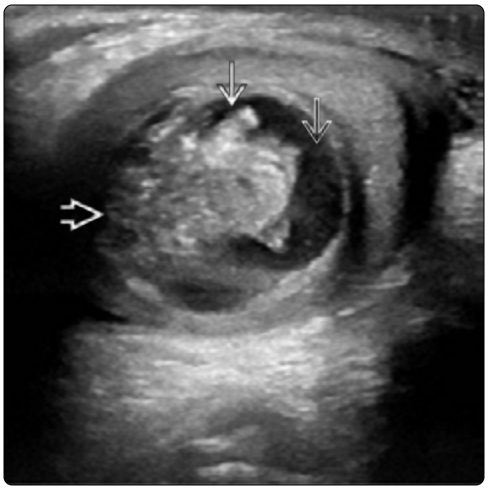
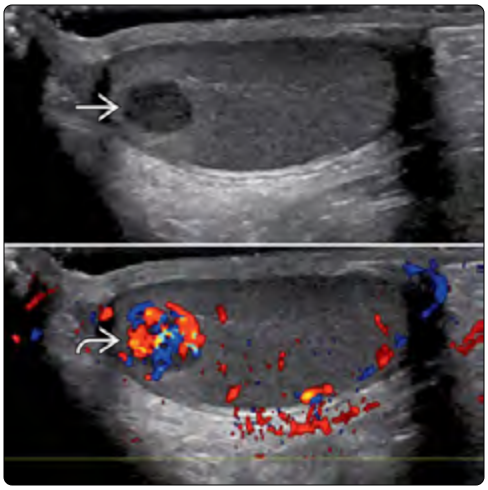


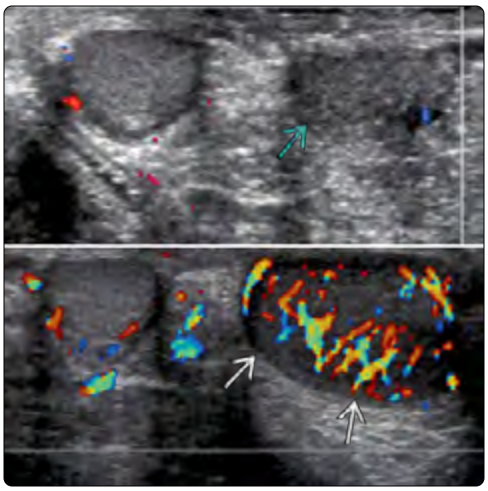


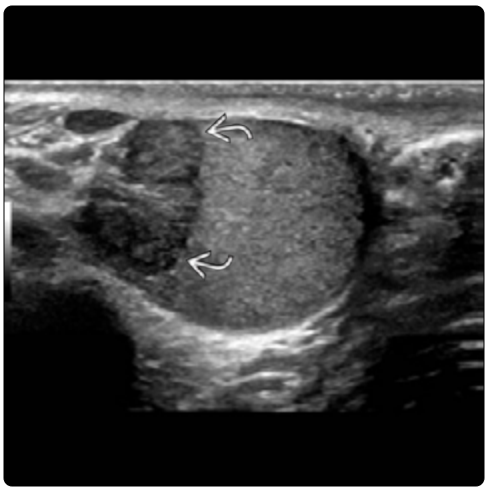
(Left) *Longitudinal US in a 17 year old who presented with chronic fatigue & a large, firm testicular mass shows a lobulated, complex, & heterogeneous lesion replacing the right testis. Blood tests revealed anemia & elevated levels of LDH & β-hCG. Metastatic choriocarcinoma was ultimately diagnosed.* **(Right)** *Sagittal lung CECT in the same patient with choriocarcinoma demonstrates well-defined pulmonary metastases with surrounding areas of ground-glass attenuation due to hemorrhage.*





(Left) *Transverse US in an infant with nonpainful testicular swelling shows a well-defined, complex testicular mass. Note the echogenic, solid component surrounded by fluid. A mature teratoma was found at resection.* **(Right)** *Longitudinal grayscale & color Doppler US images in a 7 year old with precocious puberty, ↑ testosterone, & a bone age 8 standard deviations above the mean shows a well-defined hypoechoic testicular mass with marked hypervascularity, ultimately proven to be a Leydig cell tumor.*





(Left) *Transverse color Doppler US (bottom) shows an enlarged, uniformly hypoechoic & hypervascular left testis with palisading vessels due to leukemia. Six weeks after chemotherapy (top), the testicle shows normal size & vascularity.* **(Right)** *Transverse US of a testicular adrenal rest in a 17 year old with congenital adrenal hyperplasia demonstrates a well-defined, slightly lobulated, hypoechoic mass in the periphery of the right testis. The left testis had a similar appearance (not shown).*

Testicular Trauma

KEY FACTS

TERMINOLOGY

- Hydrocele: Simple fluid between layers of tunica vaginalis
- Hematocele: Blood between tunica vaginalis layers
- Hematoma: Contained collection of blood products within testis, epididymis, or scrotal wall; may involve ≥ 1 site
- Testicular fracture: Disruption of testicular parenchyma
- Testicular rupture: Disruption of tunica albuginea, often with extrusion of testicular parenchyma
- Devascularization: Vascular pedicle injury causing absent or diminished blood flow without true torsion (twisting)
- Traumatic epididymitis: Epididymal contusion causing inflammation, enlargement, & hypervascularity
- Traumatic testicular ectopia: Traumatic dislocation of testis into inguinal canal, abdominal cavity, or perineum

IMAGING

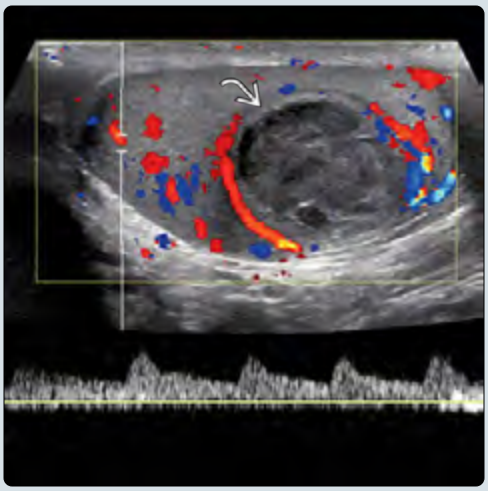
- Look for alteration of normal testicular echotexture, disruption of tunica albuginea, absent or diminished blood flow, complex hydrocele, &/or scrotal wall thickening

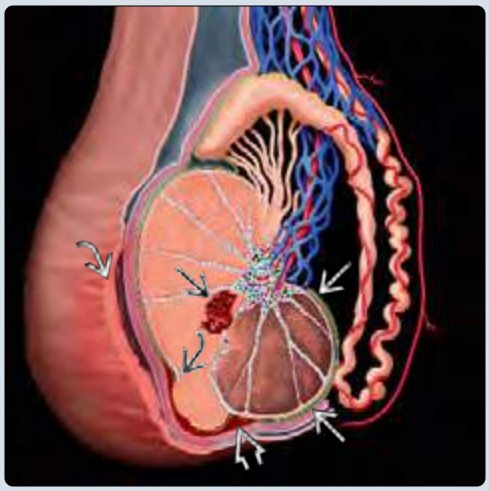
TOP DIFFERENTIAL DIAGNOSES

- Viral epididymitis
  - Epididymal enlargement, heterogeneity, & hyperemia
  - Imaging indistinguishable from trauma without history
- Testicular torsion
  - No blood flow to testis; twisting of spermatic cord
  - May occur spontaneously or after minor trauma
- Torsion of testicular appendage
  - Leading cause of acute scrotum in boys
  - Enlarged, spherical, echogenic, pedunculated remnant of tissue, ± periappendiceal hyperemia, reactive hydrocele
- Neoplasm
  - Rare, but 10-15% are found during US for scrotal trauma
  - Intratesticular mass with vascular flow (vs. testicular hematoma, which will not have blood flow)

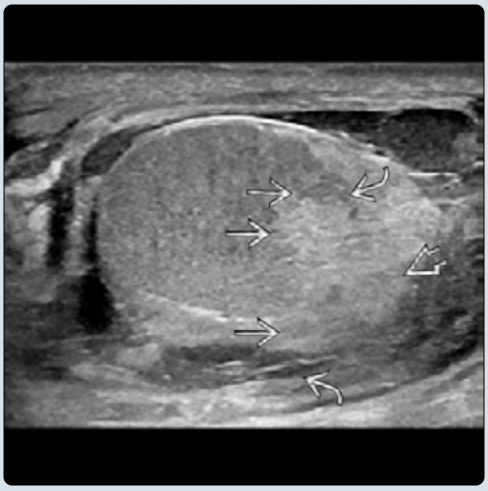
CLINICAL ISSUES

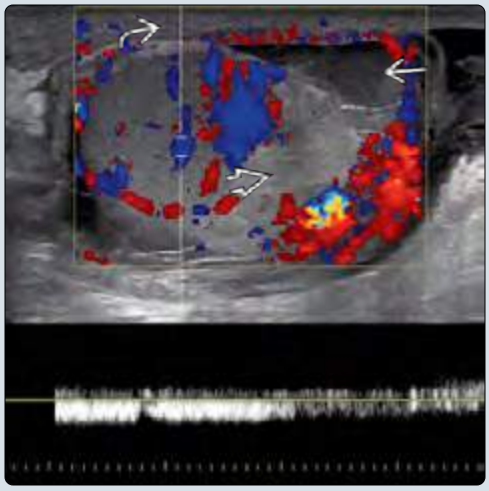
- Testicular fracture/rupture → urologic emergencies → prompt treatment reduces infection, atrophy, necrosis





(Left) *US in a teenager with hemophilia A who hit a tree while skiing shows a well-defined, heterogeneous, avascular intratesticular hematoma, which was smaller on follow-up US (not shown). Note that a neoplasm would have internal blood flow & would not be smaller on follow-up imaging.* **(Right)** *Composite graphic shows various manifestations of testicular trauma, including a scrotal hematoma, rupture of the tunica albuginea, segmental testicular infarction, parenchymal hematoma, & small hematocele.*





(Left) *US in an 11 year old who was kneed during a basketball game shows a sharp, jagged demarcation through the parenchyma, representing a testicular fracture, as well as hyperechogenicity in the lower pole due to segmental infarction. The tunica albuginea is interrupted.* **(Right)** *Doppler US in the same boy shows avascularity in the lower testis due to segmental infarction (with reactive hypervascularity of the adjacent parenchyma). There is also a surrounding hematocele as well as a scrotal wall hematoma.*

TERMINOLOGY

Definitions

- Hydrocele: Simple fluid between layers of tunica vaginalis
- Hematocele: Blood products between layers of tunica vaginalis
- Hematoma: Contained collection of blood products within testis, epididymis, or scrotal wall; can be multifocal
- Testicular fracture: Disruption of testicular parenchyma
- Testicular rupture: Disruption of tunica albuginea, often with extrusion of testicular parenchyma
- Devascularization: Vascular pedicle injury causing absent or diminished blood flow without true torsion (twisting)
- Traumatic epididymitis: Epididymal contusion causing inflammation, enlargement, & hypervascularity
- Traumatic testicular ectopia: Traumatic dislocation of testis into inguinal canal, abdominal cavity, or perineum

IMAGING

Ultrasonographic Findings

- Hydrocele, hematocele, epididymitis, scrotal hematoma
  - Echogenic/complex fluid of hematocele most common finding (after hydrocele)
- Fracture: Hypoechoic line through testicular parenchyma with focally altered echotexture ± disruption of echogenic tunica albuginea
  - Testicular parenchyma may extrude through tunica (resulting in contour deformity)
- Hematoma
  - Scrotal: Echogenic thickening of scrotal wall ± focal areas of complex fluid
  - Epididymal: Mixed echogenicity, nonvascular mass
  - Testicular: Mixed echogenicity, nonvascular mass
- Devascularization: Absent/diminished testicular blood flow
- Testicular dislocation: Rare, consider if testis not in scrotum

Imaging Recommendations

- Best imaging tool
  - Grayscale & color Doppler US with highest frequency (10-14 MHz) linear-array transducer available

DIFFERENTIAL DIAGNOSIS

Viral Epididymitis

- Epididymal enlargement, heterogeneity, & hyperemia
- Indistinguishable from traumatic cause without history

Testicular Torsion

- Absent or diminished vascularity in testis
- Look for twisting of spermatic cord just above testicle
- May occur spontaneously or after minor trauma

Torsion of Testicular Appendage

- Echogenic or salt & pepper appearance of enlarged nodular tissue along margin of testis, usually with periappendiceal hyperemia
- May cause reactive hydrocele & scrotal wall thickening
- Leading cause of acute scrotum in boys

Neoplasm

- Intratesticular mass with internal vascular flow
- 10-15% found during US for trauma

PATHOLOGY

General Features

- Etiology
  - Blunt trauma & compression most common
  - Traumatic injury to scrotal contents overall rare because of several protective features

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Intense scrotal pain in setting of recent trauma
  - Ecchymosis, swelling, skin abrasion, or laceration

Natural History & Prognosis

- Possible complications: Infarction, infection, atrophy, ↓ sex hormones & spermatogenesis, infertility

Treatment

- Physical exam has poor correlation to degree of injury, therefore US important to guide management (conservative vs. surgical repair or debridement)
- Fracture & rupture = urologic emergency
- Salvage rate for rupture: 90% → 45% after 72 hours
- Testis can usually be repaired to avoid orchiectomy
- Large hematocele or hematoma may require evacuation

DIAGNOSTIC CHECKLIST

Consider

- Scrotal, epididymal, & testicular injuries may occur in concert
- US helps exclude testicular fracture or rupture (both typically necessitate urgent surgery; other injuries managed conservatively)
- In cases of penetrating trauma, US may be limited by air artifact but can be helpful to identify possible foreign body

Image Interpretation Pearls

- Testicular rupture may violate tunica vasculosa → high rate of devascularization
- Do not confuse testicular hematoma with neoplasm; do remember that hemorrhagic neoplasms can cause false-positive US for trauma
- Hematoceles have high association with rupture (but lack of hematocele does not rule out testicular rupture or fracture)
- Testicular dislocation extremely rare, but do not assume cryptorchism in cases of trauma with missing testicle

Reporting Tips

- Patients with large, expanding hematocele may need surgery secondary to pain or possible compromise to testis
- Large hematomas may need follow-up US to exclude superinfection or testicular necrosis

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2. Wright S et al: Emergency ultrasound of acute scrotal pain. Eur J Emerg Med. 22(1):2-9, 2015
3. Gómez RG et al: Traumatic testicular dislocation. Int Urol Nephrol. 46(10):1883-7, 2014

Ectopic Testicle

KEY FACTS

TERMINOLOGY

- Undescended testis (UDT): Found along normal pathway of descent, but outside scrotum
- Ectopic testis: Found outside normal pathway of descent
  - Very rare, < 5% of boys worked up for UDT
- Testicular retraction: Physiologic, reducible retraction of testis out of scrotum due to hyperactive cremasteric reflex
- Ascending testis: Nonphysiologic, nonreducible retraction of testis out of scrotum (acquired UDT)

IMAGING

- Many pediatric urologists prefer to skip imaging for UDT
- UDT: Empty hemiscrotum, ± testis along pathway of descent; no history of testis having been within scrotum
- Ectopic testes: Empty hemiscrotum & testis abnormally located in perineum, femoral canal, superficial inguinal pouch, suprapubic area, or contralateral hemiscrotum

TOP DIFFERENTIAL DIAGNOSES

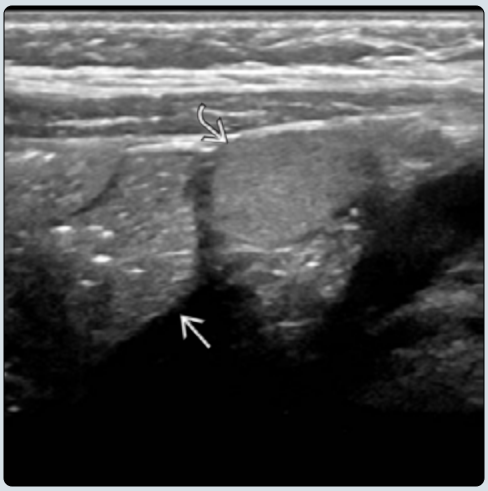
- Inguinal lymphadenopathy
- Female with congenital adrenal hyperplasia

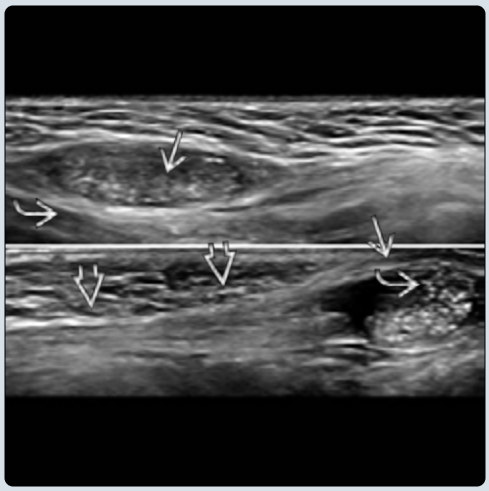
PATHOLOGY

- Pathogenesis of testicular ectopia & UDT influenced by hormonal, genetic, environmental, & anatomic factors
- UDT: Testis intrinsically abnormal with altered spermatogenesis
- Ectopic: Testis normally developed with normal spermatogenesis (if treated)

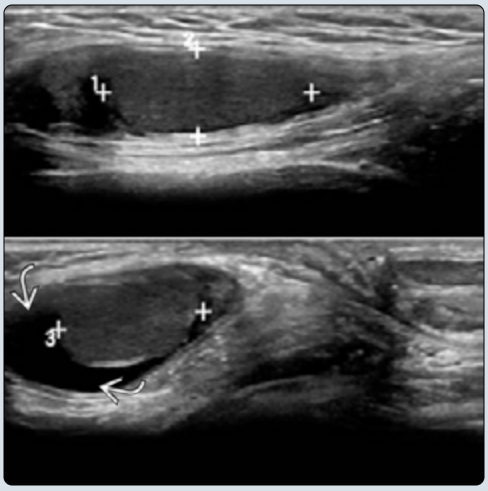
CLINICAL ISSUES

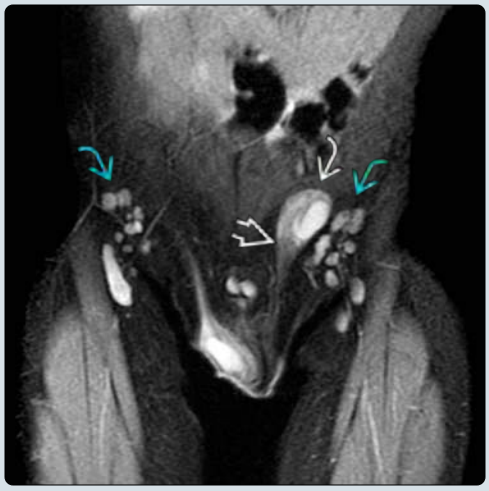
- UDT: Most spontaneously descend in 1st year of life; testis will not descend after 12 months; ↑ risk of infertility, testicular tumors, torsion, inguinal hernia if not treated
- Ectopic: ↑ risk of trauma, ± altered spermatogenesis
- Testicular retraction very common in 1st decade, but may herald ascending testis (↑ risk of infertility, trauma, tumor)





(Left) *Right lower quadrant US of an infant with cryptorchidism shows a normal-appearing testis next to bowel in the abdomen. Testes located outside the normal pathway of descent are termed ectopic testes.* **(Right)** *Trans (top) & long (bottom) US in a boy with cryptorchidism show the right testis in the inguinal canal. The normal spermatic cord course implies that it followed a normal pathway of descent. Unlike ectopic testes, UDTs are intrinsically abnormal & have a higher rate of microlithiasis.*





(Left) *Long (top) & trans (bottom) US in a 13 year old reveal a small but otherwise normal-appearing testis in the right inguinal canal (shown by calipers with a volume of 0.8 mL; normal mean testicular volume at this age is 4.4 mL). Note the small hydrocele, a common finding with canalicular UDTs.* **(Right)** *Coronal STIR MR in a toddler shows a retracted testis next to inguinal lymph nodes. The spermatic cord entering the testicular pole helps distinguish the testis from lymph nodes, which have vessels entering at the hilum.*

TERMINOLOGY

Synonyms

- Undescended testis (UDT), cryptorchidism

Definitions

- UDT: Along normal route of descent, but outside scrotum
- Ectopic testis: Outside normal pathway of descent
- Testicular retraction: Physiologic, reducible retraction of testis out of scrotum due to hyperactive cremasteric reflex
- Ascending testis: Nonphysiologic, nonreducible retraction of testis out of scrotum (acquired UDT)

IMAGING

General Features

- UDT: Empty hemiscrotum, testis along pathway of descent in 80% (or not found); no history of testis ever in scrotum
  - Differentiation of UDT from testicular retraction & ascending testis based on history & physical exam
- Ectopic testes: Empty hemiscrotum & testis abnormally located in perineum, femoral canal, superficial inguinal pouch, suprapubic area, or contralateral hemiscrotum

Ultrasonographic Findings

- Ectopic testis usually normally appearing; UDTs may be normal or small with hydrocele & microlithiasis common

CT Findings

- Testicular retraction often noted on CT as 1or both testes "riding high" in inguinal canal(s)

MR Findings

- Useful problem-solving tool for ambiguous genitalia or ectopic testes; no added value for UDT

Points to Consider

- Many pediatric urologists prefer to skip imaging for UDT

DIFFERENTIAL DIAGNOSIS

Lymphadenopathy

- Look for typical hilar fat & vessels + classic reniform shape
- Lymph nodes will be lateral to inguinal canal

Female With Congenital Adrenal Hyperplasia

- Ambiguous external genitalia with normal ovaries & uterus
- Abnormal electrolytes & endocrinologic studies

PATHOLOGY

General Features

- Pathogenesis of testicular ectopia & UDT multifactorial
  - Hormonal factors (hypothalamic-pituitary dysfunction, low androgens) & genetics play major role in UDT
  - Environmental & anatomic factors (short vas deferens, ventral wall defects, etc.) play role in ectopia & UDT
- Associated abnormalities
  - Boys with UDT have higher rates of inguinal hernia, other GU abnormalities, & endocrine abnormalities
  - Prader-Willi syndrome, Kallmann syndrome, pituitary hypoplasia, ventral wall defects → high rates of UDT

Staging, Grading, & Classification

- Testicular ectopia can be congenital or posttraumatic
  - Congenital: Femoral canal, base of penis, or perineum
  - Posttraumatic: Dislocation into inguinal canal, abdominal cavity, or perineum
- UDT: Classified based on location
  - 80% superficial & palpable (upper scrotum, superficial inguinal pouch, or inguinal canal)
  - 20% not palpable (intraabdominal)

Gross Pathologic & Surgical Features

- UDT: Testis intrinsically abnormal with altered spermatogenesis; ↑ risk of inguinal hernia, testicular neoplasm, torsion, & infertility
- Ectopic: Testis normally developed with normal spermatogenesis (if treated); no ↑ risk of inguinal hernia, testicular neoplasm, torsion, or infertility

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - UDT: Empty hemiscrotum, ± palpable testis along pathway of descent; right 50%, left 30%, bilateral 20%
  - Ectopic testes: Unilateral empty scrotal sac & palpable mass in location characteristic for ectopic testis

Demographics

- UDT most common gonadal abnormality in boys; incidence 30% if preterm, 5% if term, & 1% by 1 year
- Ectopic testis very rare, < 5% of boys worked up for UDT

Natural History & Prognosis

- UDT: Most spontaneously descend in 1st year of life; testis will not descend after 12 months; ↑ risk of infertility, testicular tumors, torsion, inguinal hernia if not treated
- Ectopic: ↑ risk of trauma ± altered spermatogenesis
- Testicular retraction very common in 1st decade but may herald ascending testis (↑ risk of infertility, trauma, tumor)

Treatment

- Orchiopexy by 1 or 2 years of age; reduces (but does not remove) risk of testicular cancer

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Make sure inguinal "mass" not actually testicle prior to biopsy of "enlarged node"

SELECTED REFERENCES

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2. Vos A et al: The value of ultrasonography in boys with a non-palpable testis. J Pediatr Surg. 49(7):1153-5, 2014
3. Ramareddy RS et al: Ectopic testis in children: experience with seven cases. J Pediatr Surg. 48(3):538-41, 2013

Variations of Hydroceles

KEY FACTS

TERMINOLOGY

- Hydrocele: Abnormal fluid in scrotal tunica vaginalis &/or spermatic cord/inguinal canal
  - Communicating: Fluid extends freely between scrotum & peritoneal cavity via patent processus vaginalis
  - Noncommunicating: No connection between scrotum & peritoneal cavity → implies closed processus vaginalis
- Spermatic cord hydrocele: Fluid in inguinal canal
  - Encysted: Loculated fluid in inguinal canal with no connection to scrotum or peritoneal cavity
  - Funicular: Fluid in inguinal canal with connection to peritoneal cavity (via open internal ring)
- Abdominoscrotal hydrocele: Loculated, dumbbell-shaped fluid collection with abdominal & scrotal components
  - More frequent in adolescents & adults
- Hydrocele of canal of Nuck: Girls only → localized fluid collection in groin &/or labia majora
- Complex hydrocele: Complicated fluid, which may be due to blood (hematocele) or pus (pyocele)
  - Older children, usually with history of trauma, infection, testicular torsion, or malignancy

IMAGING

- Simple, anechoic fluid surrounding testicle
  - Sparing of "bare area": Site of testicular attachment to epididymis; lacks tunica vaginalis
- ± connection to inguinal canal & peritoneal cavity
- ± mild debris or thin septations

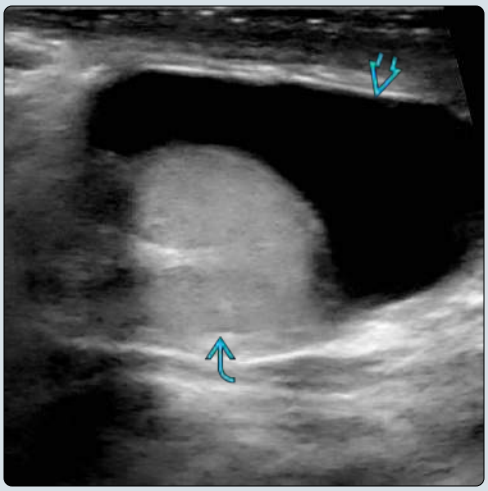
TOP DIFFERENTIAL DIAGNOSES

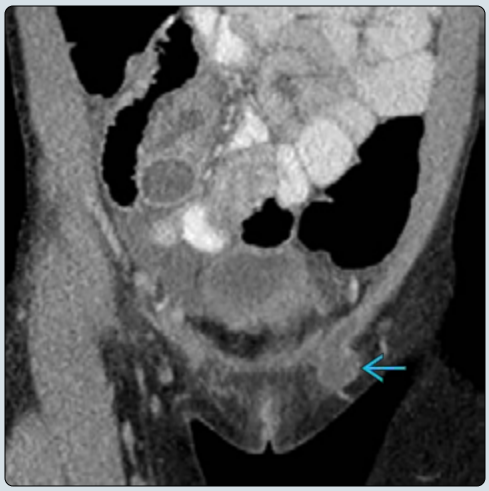
- Indirect inguinal hernia
- Paratesticular rhabdomyosarcoma
- Lymphatic malformation

CLINICAL ISSUES

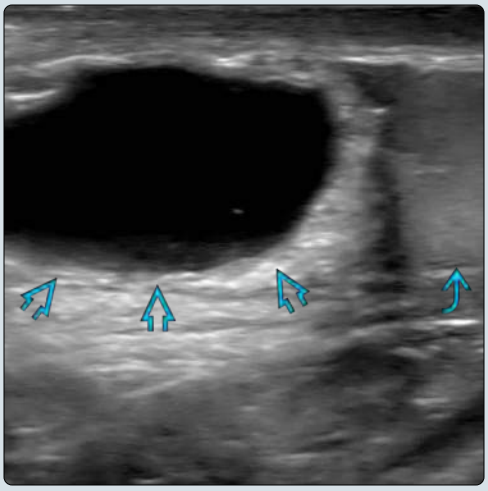
- Most common < 1 year of age
- Patent processus vaginalis often spontaneously closes ≤ 2 years of age; surgery often required > 2 years

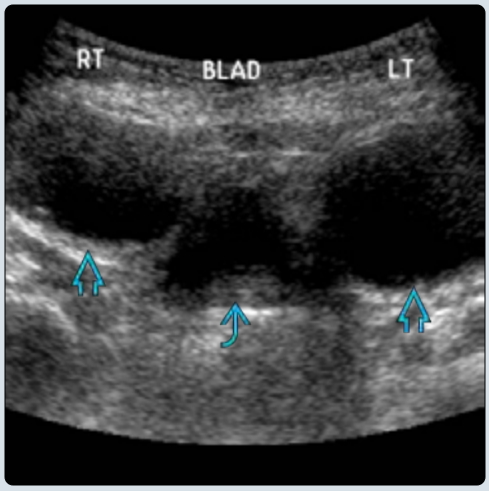
(Left) *Transverse ultrasound in an 18-month-old boy with scrotal swelling demonstrates simple fluid within the tunica vaginalis surrounding the left testicle, consistent with a noncommunicating hydrocele.* **(Right)** *Coronal CECT in a 7-year-old girl being evaluated for acute appendicitis shows a fluid attenuation structure in the left inguinal canal, consistent with a small hydrocele of the canal of Nuck.*





(Left) *Longitudinal ultrasound of the left inguinal canal in a 5-year-old boy with groin swelling shows fluid in the inguinal canal above the left testicle. The fluid increased when the patient was upright, indicating a connection to the peritoneal cavity (i.e., a funicular hydrocele of the spermatic cord).* **(Right)** *Transverse ultrasound of the pelvis in a 7-month-old boy with groin swelling shows a localized, cystic appearance to the abdominal portions of bilateral abdominoscrotal hydroceles on either side of the urinary bladder.*





TERMINOLOGY

Definitions

- Hydrocele: Abnormal fluid in scrotal tunica vaginalis &/or spermatic cord/inguinal canal
- Communicating hydrocele: Fluid extends freely between scrotum & peritoneal cavity via patent processus vaginalis
- Noncommunicating hydrocele: No connection between scrotum & peritoneal cavity → implies closed processus vaginalis
- Spermatic cord hydrocele: Fluid in inguinal canal
  - Encysted hydrocele: Loculated fluid in inguinal canal with no connection to scrotum or peritoneal cavity
  - Funicular hydrocele: Fluid in inguinal canal with connection to peritoneal cavity (via open internal ring)
- Abdominoscrotal hydrocele: Loculated, dumbbell-shaped fluid collection with abdominal & scrotal components
  - More frequent in adolescents & adults
- Hydrocele of canal of Nuck: Girls only → localized fluid collection in groin &/or labia majora
  - Also called female hydrocele
- Complex hydrocele: Complicated fluid, which may be due to blood (hematocele) or pus (pyocele)
  - Older children, usually with history of trauma, infection, testicular torsion, or malignancy

IMAGING

General Features

- Best diagnostic clue
  - US showing simple fluid around testicle with extension into inguinal canal
- Size
  - Variable size; can be quite large
  - Canal of Nuck hydroceles tend to be smaller (< 3 cm)

Ultrasonographic Findings

- Grayscale ultrasound
  - Simple, anechoic fluid surrounding testicle except for "bare area" (site of testicular attachment to epididymis; lacks tunica vaginalis)
  - ± connection to inguinal canal & peritoneal cavity
    - With proximal inguinal canal extension, fluid may
      - Freely enter peritoneal cavity
      - Be contained as cystic intraabdominal mass
    - Fluid may be localized entirely to inguinal canal
  - ± mild debris or thin septations
  - Hydrocele may deform or resolve with compression

MR Findings

- T2WI
  - High fluid signal surrounding lower signal testes

Imaging Recommendations

- Best imaging tool
  - Grayscale ultrasound
- Protocol advice
  - Must image inguinal canal to determine communication with peritoneal cavity
  - Consider imaging with different positions (upright, supine) & Valsalva maneuver

DIFFERENTIAL DIAGNOSIS

Indirect Inguinal Hernia

- Dynamic protrusion of bowel &/or echogenic mesenteric fat into inguinal canal ± scrotum

Paratesticular Rhabdomyosarcoma

- Solid heterogeneous extratesticular malignant mass with variable internal vascularity

Lymphatic Malformation

- Multicystic slow flow vascular malformation infiltrating different tissue compartments
- Rarely isolated to scrotum (though scrotum often involved by more extensive lymphatic malformations)

Enlarged Lymph Nodes

- Inguinal adenopathy common, ranging from
  - Enlarged reactive nodes with normal architecture
  - Necrotic/suppurative adenitis with cellulitis → abscess
  - Malignant infiltration with loss of architecture, round morphology, & distorted internal vascularity

PATHOLOGY

General Features

- Etiology
  - Failure of involution of normal developmental peritoneal fold extending into scrotum during testicular descent → patent processus vaginalis
  - Occurs at labia majora in girls along round ligament

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Scrotal or inguinal swelling, typically painless
  - May change with position & Valsalva (if communicating)

Demographics

- More common in infants
- Boys >> girls
- ↑ incidence in premature babies & children with peritoneal dialysis, ventriculoperitoneal shunts, or ascites

Natural History & Prognosis

- Patent processus vaginalis often spontaneously closes ≤ 2 years of age → observation considered
- > 2 years of age, spontaneous resolution unlikely → surgery usually required
  - Closure of patent processus vaginalis + opening or drainage of hydrocele sac

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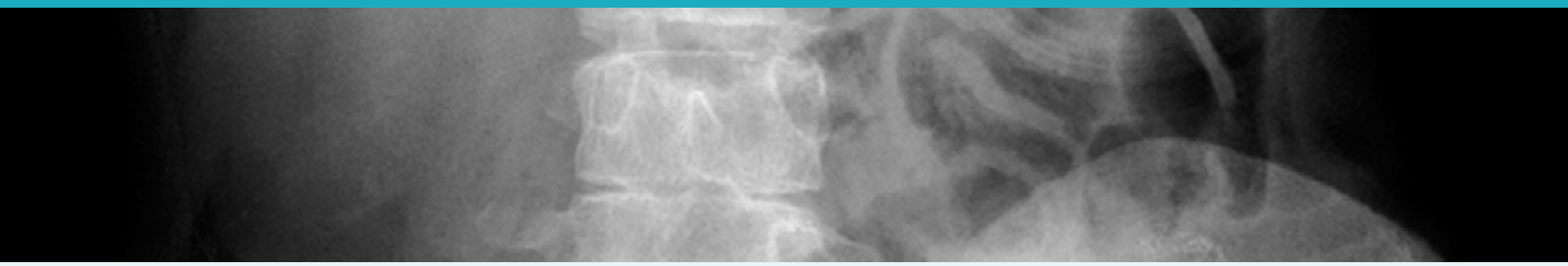
SECTION 6

Musculoskeletal



Musculoskeletal Imaging Modalities

Radiography

Although there have been great advances in imaging technology, radiography remains the most important imaging test in most circumstances of suspected musculoskeletal pathology. With appropriate techniques, the associated radiation exposure is usually minimal, & radiography is less expensive than other more advanced imaging techniques. Most cases of trauma require radiographs & no other imaging. With suspicion of nonaccidental trauma (or an underlying skeletal dysplasia), a full skeletal survey should be performed.

Computed Tomography

CT is fast but utilizes ionizing radiation & must be employed judiciously. It allows multiplanar assessments & provides better soft tissue contrast than radiographs. Cortical bone detail is excellent by CT, though marrow evaluation is limited. CT is helpful in the detection & characterization of specific fractures that are intraarticular or complex, or for fractures that are often subtle but risk significant consequences if the diagnosis is delayed. Additional CT indications include the evaluations of osteoid osteoma, sequestra of osteomyelitis, tarsal coalitions, & union of complicated fractures.

Ultrasound

Ultrasonography uses no radiation, is relatively inexpensive, requires no sedation, & provides excellent superficial spatial resolution. It also enables dynamic assessments, differentiates cystic vs. solid masses, & characterizes vascular flow. Specific indications include the evaluations of infant hips for developmental dysplasia, retained nonradiopaque foreign bodies, soft tissue masses & fluid collections, & tendon injuries.

Magnetic Resonance

MR imaging uses no ionizing radiation; however, it is costly, has a relatively long exam time, & may require sedation. MR provides excellent soft tissue contrast & is multiplanar. When there is a high suspicion of pathology at a targeted musculoskeletal site, MR is the primary tool for investigation. Such indications include joint trauma, infection, soft tissue masses, & bone tumors. Other indications are stress injuries, physeal osseous bridges, & myopathy/myositis.

Nuclear Medicine

In isolation, nuclear medicine scans have excellent sensitivity (but low specificity) for whole body screening of bone pathology. Bone scan indications include metastatic disease, nonaccidental trauma, stress fracture, spondylolysis, osteomyelitis, osteoid osteoma, & avascular necrosis. Sensitivity is further increased by bone scan SPECT, & excellent anatomic localization is achieved in combination with bone CT (e.g., for the detection of spondylolysis). Gallium-67 citrate or indium-111 WBC scans may be used in complex cases of osteomyelitis. FDG PET indications are largely centered on cancer staging.

Congenital Abnormalities

These abnormalities are typically discovered prenatally, at birth, or within the 1st few months of life. The diagnosis of such entities mainly relies on radiographs.

Proximal focal femoral deficiency (PFFD) is a malformation in which complete growth & development of the upper femur fails to occur. The Aitken classification, from class A (the least severe form with a short femur & common subtrochanteric varus deformity) to class D (in which both the femoral head & acetabulum are absent, with a short distal femoral segment), guides treatment in PFFD. However, classification can be difficult with radiographs at an early age (due to unossified segments).

Tarsal coalition is the congenital or acquired fusion of 2 or more tarsal bones & is most commonly found at the calcaneonavicular or talocalcaneal levels.

Congenital tibial dysplasia is a congenital pseudoarthrosis with anterolateral tibial bowing or fracture. Seventy percent of patients will eventually be diagnosed with neurofibromatosis type 1 (NF1).

Congenital tibial bowing is otherwise typically convex posteromedially & due to in utero positioning. It tends to resolve.

Other malformations include syndactyly, radial clubhand, radioulnar synostosis, arthrogryposis, & amniotic band syndrome.

Trauma

Pediatric fractures may be complete (i.e., all the way through the bone from one surface to the opposite surface) or incomplete. The elastic properties of the developing pediatric skeleton increase the potential for incomplete fractures, including plastic deformation (bowing), buckle, or greenstick fractures. It is important to obtain orthogonal views for a complete evaluation of the fracture. When describing the fracture, remember to include the location, extension, translation, & angulation.

Salter-Harris Classification

Approximately 1 in 5 pediatric fractures involves an adjacent growth plate.

In a type I fracture, the fracture is purely through the growth plate or physis (e.g., slipped capital femoral epiphysis).

In a type II fracture [the most common Salter-Harris (SH) fracture], the physeal fracture extends through a portion of the metaphysis.

In a type III fracture, the physeal fracture extends the through epiphysis (e.g., juvenile Tillaux fracture).

In a type IV fracture, the fracture extends across the physis & involves both the epiphysis & metaphysis (e.g., triplane & lateral condylar fractures).

In a type V fracture, there is a crush injury of the physis.

Common Elbow Fractures

The supracondylar fracture is the most common pediatric elbow fracture. It most commonly occurs at 5-7 years of age. An elbow effusion may suggest an occult supracondylar fracture.

The medial epicondyle avulsion comprises 10% of elbow fractures in pediatrics. The avulsed fragment may become entrapped in the elbow joint.

The lateral condylar fracture is typically an SH type IV fracture with the distal component extending through the unossified distal humeral cartilage.

The radial neck fracture accounts for 5% of pediatric elbow fractures with an average age of 10 years. Ninety percent are SH type II fractures.

Important Ankle Fractures

The juvenile Tillaux fracture is an SH type III fracture through the anterolateral distal tibial epiphysis. The average age is 12-15 years. Mortise or oblique imaging views are helpful, but NECT with coronal & sagittal reformation helps determine the degree of articular displacement & associated therapy (e.g., when there is > 2 mm of displacement, operative treatment is employed).

The triplane fracture is typically an SH type IV fracture in 3 planes (transverse physeal, sagittal epiphyseal, & coronal metadiaphyseal planes). The average age is 12-15 years. NECT with coronal & sagittal reformation helps determine the degree of displacement & categorize the fracture. When there is > 2 mm of displacement, operative treatment is employed.

Soft Tissue Masses

Some soft tissue masses can be diagnosed by clinical exam, such as lipomas (which are superficial & doughy by palpation) & ganglion cysts (which transilluminate & lie near a joint or tendon). Others warrant further evaluation by US &/or MR. MR is particularly helpful in determining the deep extent of a lesion & recognizing characteristic features that may elude US (including the presence of fat, muscle edema, & layering blood products). Firm, round, solid masses are particularly concerning for malignancy & almost always require biopsy. Nonspecific but entirely superficial lesions on US may be considered for excision without MR.

The ganglion cyst is a homogeneously hyperintense round or lobulated mass with peripheral enhancement on MR; it communicates with an adjacent tendon or joint space.

A lipoma follows fat signal on all MR sequences & has thin septations with no significant enhancement.

Vascular malformations are congenital anomalies that are present at birth but may not manifest until later in life. Slow-flow venous or lymphatic malformations may show layering fluid-fluid levels of stagnant blood products. Venous lesions will show gradual patchy to diffuse enhancement while lymphatic malformations will only show rim or septal enhancement. Fast flow arteriovenous malformations show a tangle of vascular flow voids with or without a soft tissue mass.

A hemangioma is a benign neoplasm that is typically small or absent at birth with rapid growth over the 1st few months of life before involuting over months to years. On MR, this lobulated lesion is typically subcutaneous & shows hyperintense T2 signal, vascular flow voids, & intense contrast enhancement. On Doppler US, there are > 5 vessels/cm² in the lesion with numerous low-resistance arterial waveforms.

Fat necrosis most commonly follows blunt trauma (though the patient may not remember the traumatic event) & is typically a subcutaneous, elongated fluid collection with angular peripheral margins (± central fat signal) & no soft tissue mass.

Plexiform neurofibromas are found in NF1 & may show a target or bag-of-worms appearance. There is concern for a malignant peripheral nerve sheath tumor when the mass shows disproportionate enlargement, loses the target sign, invades the surrounding structures, or becomes painful.

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children. Embryonal RMS accounts for 60-70% of childhood RMS & typically occurs in the GU tract & head/neck (but may occur in the extremities). Alveolar RMS occurs in adolescents, most commonly in the extremities, trunk, & perianal/perirectal area.

Synovial sarcoma is the 2nd most common soft tissue sarcoma of childhood. It most commonly presents at 15-35 years of age. There is calcification in 1/3 of cases, & the lesion often occurs near a joint. The lesion may be small & indolent-appearing on MR.

Desmoid-type fibromatosis was previously known as aggressive fibromatosis. On T2 MR, the mass is frequently hyperintense with bands of hypointensity & variable enhancement. The mass may erode or scallop the adjacent bone. It is often infiltrating & slow growing with a high rate of local recurrence.

Focal Bone Lesions

In the evaluation of extremity bone tumors, begin the exam with T1 & STIR MR joint-to-joint imaging to exclude skip metastases of the marrow. Smaller field-of-view images targeted at the main tumor can then be obtained with dedicated surface coils.

Osteosarcoma is the most common malignant primary bone tumor in children & young adults. The bimodal age distribution is 10-30 years followed by > 60 years. It classically shows aggressive destruction plus new osteoid formation with 55-80% occurring around the knee. It is typically metaphyseal (90%).

Ewing sarcoma is the 2nd most common primary bone malignancy in children. The typical age range is 5-25 years. It has an aggressive appearance that is permeative or moth eaten, though sclerosis (in the bone) may occur in up to 25%. Its aggressive periosteal reaction may include Codman triangles, a spiculated (i.e., sunburst or hair standing on end) appearance, or a lamellated onion-skin appearance. There is often a disproportionately larger soft tissue mass than the amount of bone destruction.

Langerhans cell histiocytosis has a variable radiographic appearance. It occurs between 0-30 years of age with a peak at 5-10 years. It is classically a lucent round or geographic punched-out lesion with a narrow zone of transition.

Infection

The absence of radiographic findings early does not exclude osteomyelitis. It shows periosteal reaction &/or bony destruction with an aggressive look after 7-10 days. In subacute osteomyelitis, an intraosseous Brodie abscess may show the penumbra sign with a high T1 signal rim marginating a more hypointense center.

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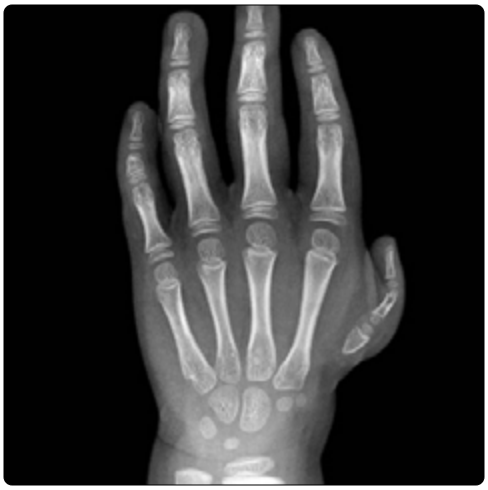
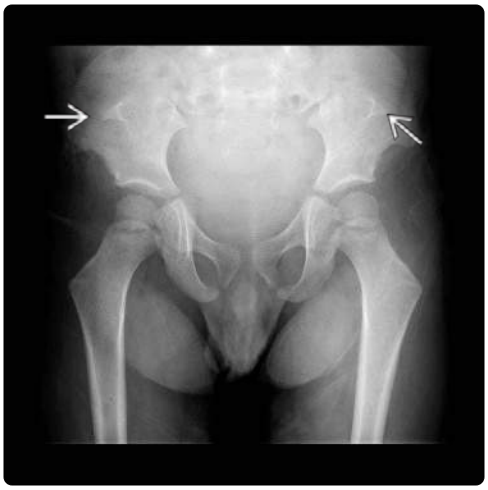
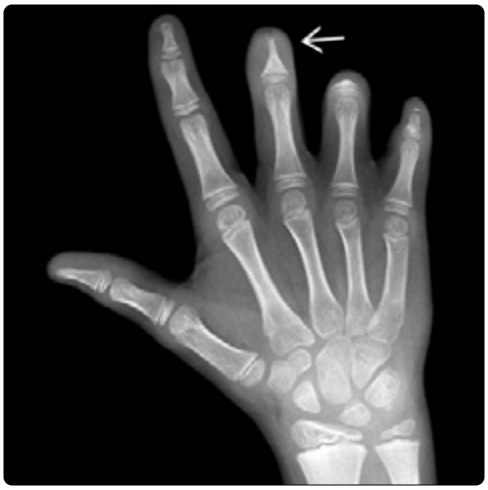
7. Navarro OM: Soft tissue masses in children. Radiol Clin North Am. 49(6):1235-59, vi-vii, 2011

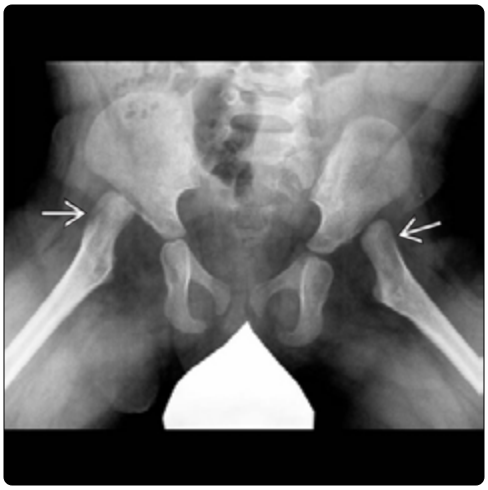
(Left) *PA radiograph of the left hand in a 4 year old with Fanconi anemia shows a hypoplastic thumb with a truncated, triangular-shaped distal metacarpal & hypoplastic proximal & distal phalanges. Fanconi anemia is associated with radial ray dysplasia, which may be isolated to the thumb.* **(Right)** *AP radiograph in a 3 year old shows bilateral posterior iliac horns ("Fong prongs") ➡ in a child with nail-patella syndrome or Fong disease. The iliac horns are considered pathognomonic for this syndrome.*



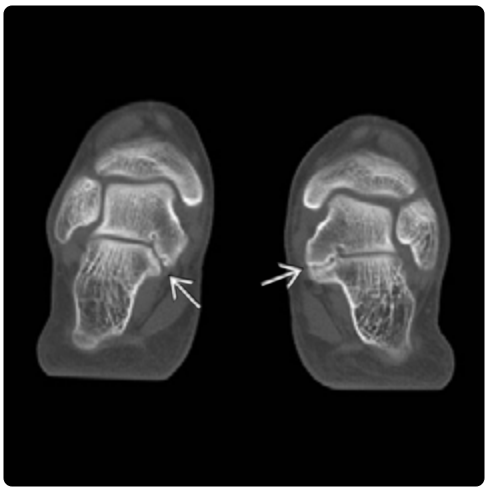


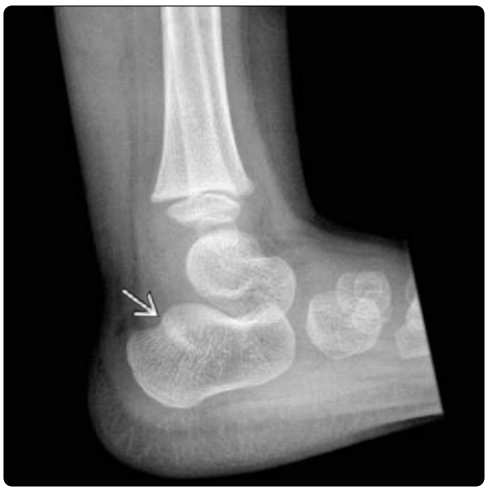
(Left) *PA radiograph of the hand in an 8 year old with middle finger swelling shows loss of the distal portions of the 3rd-5th digits due to amniotic band syndrome. There is soft tissue swelling & irregularity of the tip of the middle finger ➡ in this child with cellulitis.* **(Right)** *Frog leg lateral radiograph in a 17 month old with hip pain shows severe changes of developmental dysplasia of the hips with bilateral dislocations of the proximal femurs ➡ & shallow dysplastic acetabula.*

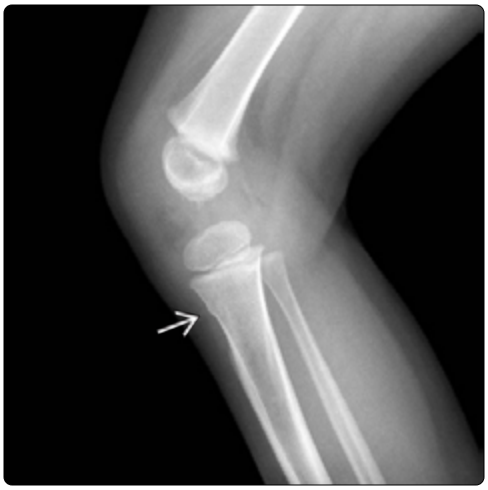


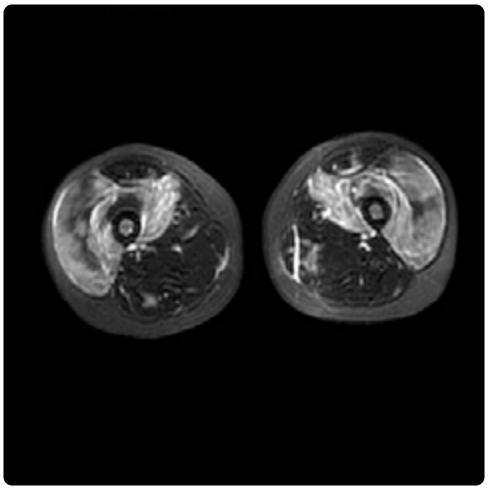


(Left) *Coronal NECT in a 12 year old with bilateral foot pain & flat feet shows bilateral middle facet nonosseous talocalcaneal coalitions ➡.* **(Right)** *Lateral radiograph in a 23 month old who fell down steps & is not weight-bearing on the left leg presents for follow-up radiographs 10 days after an initially normal exam. The current image shows a band of sclerosis or stress fracture within the calcaneus ➡.*

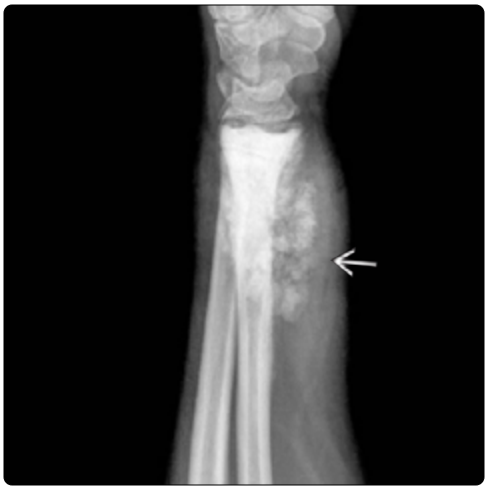


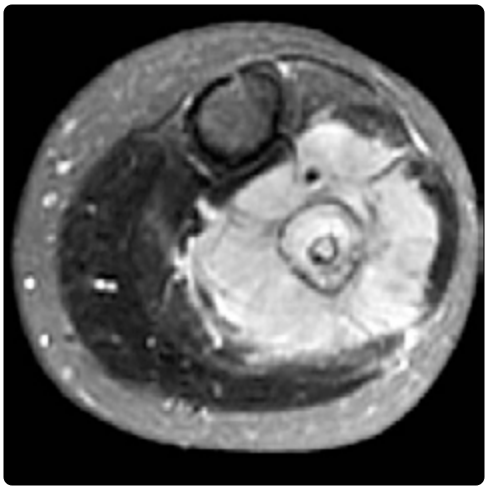




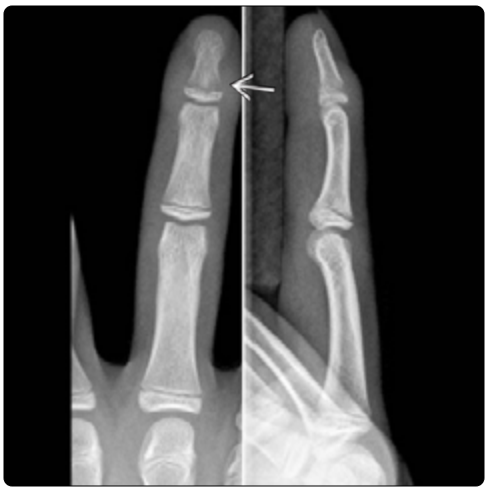


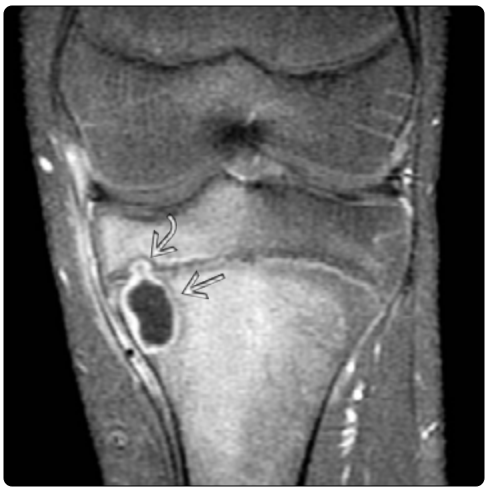
(Left) *Lateral radiograph in a 2.5 year old after an injury on a trampoline shows a buckle fracture of the anterior tibia* ➡. **(Right)** *Axial T2 FS MR in an 8 year old with weakness shows relatively symmetric hyperintense T2 signal within the anterior compartments of the thighs. Five major criteria for juvenile dermatomyositis include: Symmetric proximal muscle weakness, characteristic changes on muscle biopsy, ↑ muscle enzymes in serum, EMG abnormality of myopathy & denervation, & a characteristic rash.*





(Left) *Lateral radiograph in a 13 year old with osteosarcoma shows increased sclerosis of the distal radius with cloud-like Ca^{2+} (or osteoid)* ➡ *about the bone.* **(Right)** *Axial T2 FS MR in a 9 year old with a mass in the lower leg shows a hyperintense T2 fibular lesion with a large soft tissue component. The anterior neurovascular bundle was completely encased by the mass. This lesion was a biopsy-proven Ewing sarcoma.*





(Left) *Frontal & lateral radiographs of the middle finger in a 14 year old after a basketball injury 2 weeks prior show increased lucency* ➡ *of the metaphysis of the distal phalanx. This is a "stubbed finger" fracture complicated by osteomyelitis.* **(Right)** *Coronal T1 C+ FS MR in a 14 year old with proximal tibial pain shows a rim-enhancing intraosseous Brodie abscess* ➡ *with surrounding marrow edema. The abscess ends across the proximal tibial physis* ➡.

Primary and Secondary Growth Centers

KEY FACTS

TERMINOLOGY

- Primary growth center = primary ossification center
- Secondary growth center = secondary ossification center
- Endochondral ossification: Bone formation on preexisting cartilage model at primary & secondary growth centers
- 1° growth centers: Where majority of ossification occurs
  - Predominately at long bone physes in childhood
- 2° growth centers: Ossification centers that do not significantly contribute to longitudinal growth; in general
  - Surrounded by growth plate & unossified cartilage
  - Found at articulations of long bones (epiphyses) & nonarticular equivalents (apophyses, carpals, tarsals, etc.)

IMAGING

- Radiographs: Multiple &/or irregular growth centers normal at some locations but can be confused with pathology
  - Upper extremity: Trochlea, pisiform
  - Lower extremity: Femoral condyles, tibial tubercle, medial malleolus, calcaneal apophysis, cuneiforms
- MR: Normal ossification centers (± normal surrounding cartilage) without abnormal edema/fluid signal

TOP DIFFERENTIAL DIAGNOSES

- Osteochondroses
- Osteochondritis dissecans
- Osteomyelitis
- Osteonecrosis

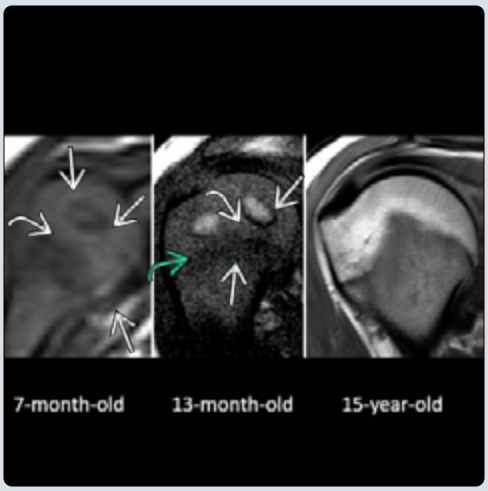
CLINICAL ISSUES

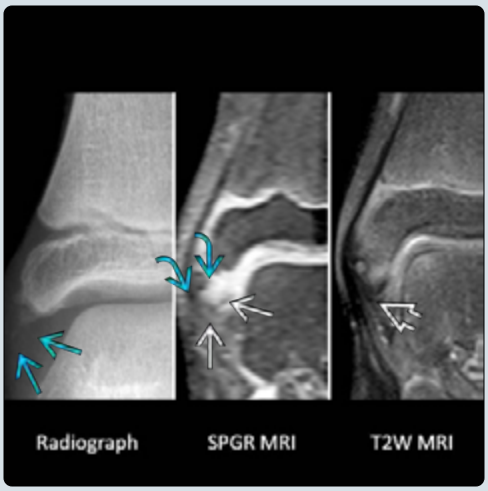
- 1° & 2° growth centers normally asymptomatic
- Pathology can occur at growth centers (e.g., acute/chronic trauma, infection, & osteonecrosis)

DIAGNOSTIC CHECKLIST

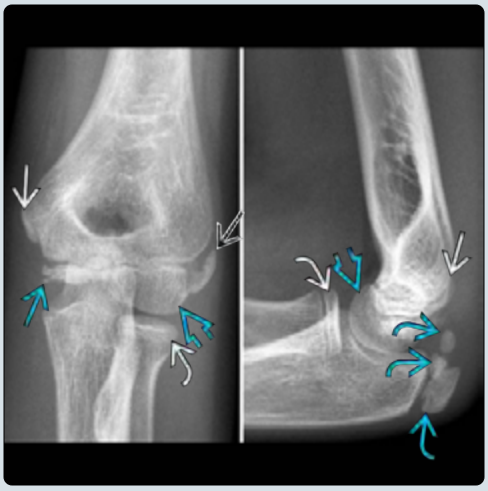
- Familiarity with normal irregular ossification prevents mistaking multiple/irregular growth centers with pathology
- Comparison with contralateral side or prior studies of similar-aged children can be helpful
- MR useful in difficult cases

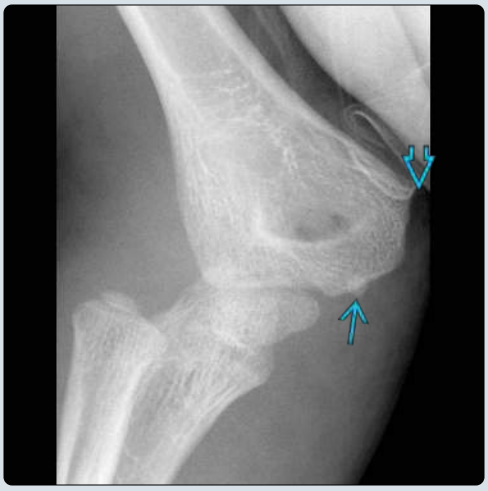
(Left) *Coronal T1 MR shows changes of the humeral head at 3 ages. The secondary growth center of the epiphysis is intermediate signal (red marrow) at 7 months & ↑ signal (yellow marrow) at 13 months. Note the greater tuberosity center at 13 months. Epiphyseal cartilage surrounds these centers.* **(Right)** *Radiograph shows multiple medial malleolar ossification centers. SPGR MR shows the ossified centers as low signal within bright epiphyseal cartilage. No edema is seen on the T2 FS MR.*





(Left) *Note the elbow secondary growth centers: Capitellum, radial head, internal (medial) epicondyle, trochlea, olecranon, & external (lateral) epicondyle. The trochlear irregular ossification & the multiple olecranon growth centers are normal findings.* **(Right)** *Elbow dislocation in a 7 year old shows a growth center at the expected location of the trochlea but with no medial epicondylar (ME) growth center, which should form 1st. This pattern indicates that the ME has been avulsed & resides in the joint.*





TERMINOLOGY

Synonyms

- Primary (1°) growth (ossification) center
- Secondary (2°) growth (ossification) center

Definitions

- Endochondral ossification: Bone formation of preexisting cartilage model at 1° & 2° growth centers
- 1° growth centers: Where majority of ossification occurs
  - Predominately at long bone physes in childhood
    - Account for majority of longitudinal growth
- 2° growth centers: Ossification centers that do not significantly contribute to longitudinal growth; in general
  - Surrounded by growth plate & unossified cartilage
  - Found at articulations of long bones (epiphyses) & nonarticular equivalents (apophyses, carpals, tarsals, etc.)

IMAGING

General Features

- Location
  - Skull base: 1° growth centers develop ~ 12-17 weeks
  - Vertebrae: 1° growth centers present at birth, 2° growth centers form in C2-L1 after birth
  - Elbow: Order of appearance of 2° growth centers remembered by acronym **CRITOE**: **C**apitellum → **r**adial epiphysis → **i**nternal (medial) epicondyle → **t**rochlea → **o**lecranon → **e**xternal (lateral) epicondyle
  - Hand/wrist: 1° centers of carpals & 2° centers of phalanges form after birth
  - Pelvis: 2° growth centers form in teens, most fuse in 20s
  - Femur: 2° growth centers form in utero & in childhood
  - Tibia: 2° growth centers form at birth & in childhood
  - Fibula: 2° growth centers form throughout childhood
  - Foot: 1° centers of tarsals form before & after birth, 2° centers form after birth
- Morphology
  - Multiple &/or irregular growth centers at some locations
  - Upper extremity
    - Trochlea of distal humerus: Irregular ossification
    - Pisiform: Irregular ossification
  - Lower extremity
    - Proximal femoral epiphyses: Minimal irregularity < 3 years
    - Femoral condyles: Irregular ossification
      - Radiographically difficult to distinguish from osteochondritis dissecans (OCD)
      - Irregular ossification typically in younger patients, more posteriorly & not in intercondylar region
    - Tibial tubercle: Multiple irregular centers
    - Medial Malleolus: Multiple irregular centers
    - Calcaneus apophysis: Multiple irregular centers
    - Cuneiforms: Often have irregular ossification
    - Apophysis of 5th metatarsal base: Lateral & longitudinally oriented

Radiographic Findings

- Multiple &/or irregular growth centers can be confused with pathology
- Elbow: 2° growth center appearance (**CRITOE**)
  - **I**nternal (medial) epicondyle typically appears before **t**rochlea
  - If ossification center seen in region of trochlea but medial epicondyle not present, then suspect fracture/intraarticular dislocation of medial epicondyle mimicking trochlear growth center
- Growth centers at hand/wrist used to assess skeletal maturation (i.e., Greulich & Pyle standards)

MR Findings

- Helpful in difficult cases to distinguish irregular ossification & pathologic processes
- Normal development will show multiple &/or irregular dark ossification centers in (± normal bright surrounding cartilage) without abnormal edema/fluid signal
  - PD FS or T2 * GRE best for cartilage; T2 FS best for fluid

DIFFERENTIAL DIAGNOSIS

Osteochondroses

- Poorly defined group of painful growth disturbances (usually due to repetitive microtrauma &/or avascular necrosis) that occur at epiphyses & apophyses

Osteochondritis Dissecans

- Overuse injury in femoral condyles, talus, & capitellum

Osteomyelitis

- Can involve epiphysis: May be primary site in infants vs. spread from metaphysis or joint in any child
- Typically signs/symptoms of infection

Osteonecrosis

- May see in sickle cell & steroid use or primary idiopathic hip disease

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 1° & 2° growth centers are normal part of endochondral ossification & are asymptomatic
  - Growth center pathology can occur (e.g., acute/chronic trauma, infection, osteochondroses, & osteonecrosis)

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Familiarity with normal irregular ossification prevents mistaking multiple/irregular growth centers with pathology
- Comparison with contralateral side (in limited circumstances), prior studies of similar-aged children, & reference texts can be helpful
- MR may be useful in difficult cases

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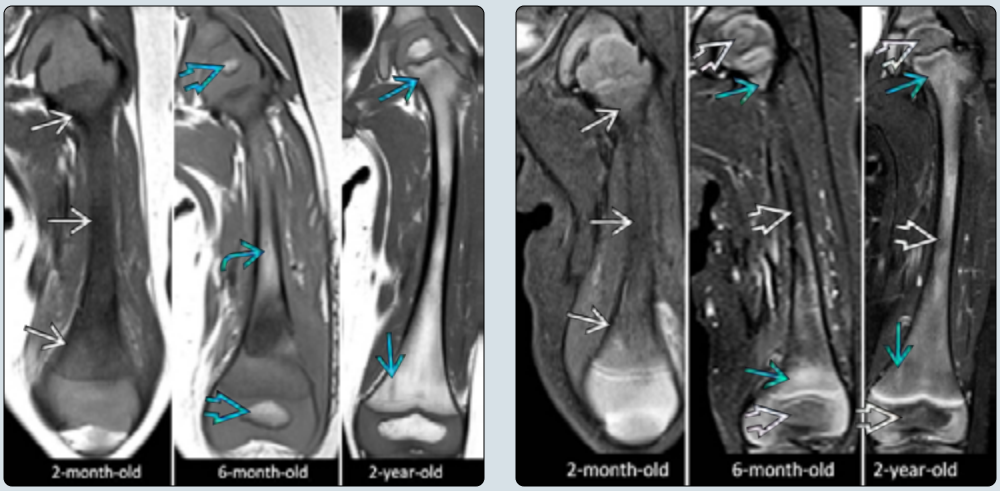
Red to Yellow Marrow Conversion

KEY FACTS

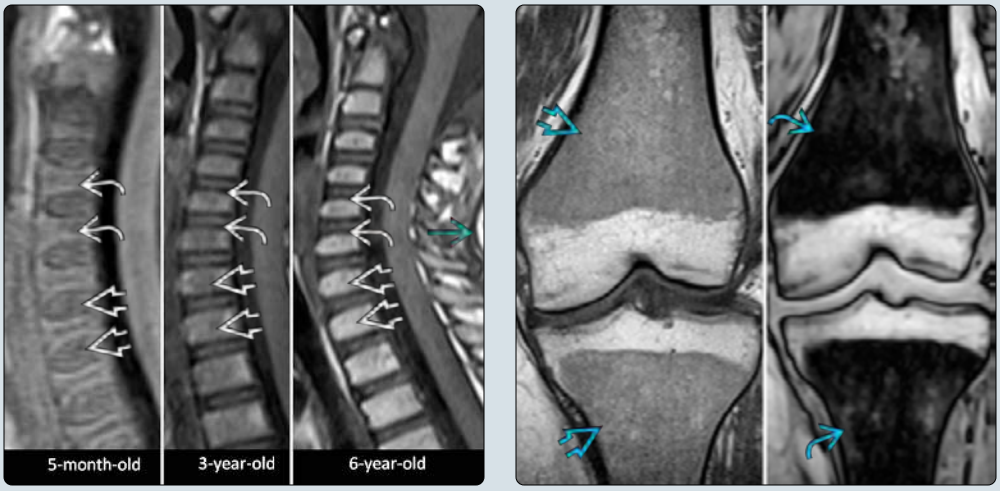
IMAGING

- At birth: Red marrow present throughout entire skeleton
- Conversion of hematopoietic red to fatty yellow marrow begins shortly after birth & follows predictable pattern
  - Appendicular before axial skeleton
  - Distal before proximal extremities (e.g., phalanges of fingers begin conversion before humeri)
  - Within individual long bones
    - Epiphyses convert 1st: Within 6 months of appearance, epiphyseal ossification center should contain almost entirely yellow marrow
    - Central diaphyses next, spreading toward metaphyses
    - Metaphyseal red marrow often persists in adults
      - Typically flame-shaped or poorly defined
  - Conversion of red to yellow marrow typically symmetric (i.e., similar appearance of right vs. left extremities)
- By 12-24 months, most appendicular marrow is approaching MR signal intensity of subcutaneous fat on spin-echo T1WI (i.e., concordance of signal intensities)
  - On FS T2WI or STIR, greatest concordance in epiphyses
  - Diffuse or multifocal discordance after this age (especially on spin-echo T1WI) implies pathology
    - Multifocality particularly concerning if asymmetric & well defined
- Variability among individuals as to exact timeframe
- Degree of red marrow often persists in spine & pelvis
- On spin-echo T1WI MR (best marrow sequence): Red marrow shows low to intermediate signal intensity
  - Typically iso- or slightly hyperintense as compared to muscle & intervertebral discs
- In- & opposed-phase T1 GRE MR
  - Normal red marrow drops signal intensity on opposed-phase images compared to in-phase images
    - Due to water & fat protons in same voxel
  - Infiltrating processes (such as leukemia & metastases) generally do not drop signal on opposed-phase images
- Reconversion of yellow to red marrow (from variety of causes) occurs in reverse order from initial conversion

(Left) *Coronal T1 MRs of femurs in normal children of various ages: Initially, low signal intensity red marrow is seen diffusely. In infancy, yellow marrow appears 1st in the epiphyses & central diaphysis, later progressing to the metaphyses. Note the persistent areas of metaphyseal red marrow.* **(Right)** *Corresponding coronal STIR MRs show homogeneous low signal intensity early. Later, epiphyseal & diaphyseal yellow marrow is of lower signal intensity than metaphyseal red marrow.*



(Left) *Sagittal T1 MRs of the cervical spine at various ages. The signal intensity of vertebral body marrow compared to the intervertebral discs is initially hypointense, gradually becoming iso- & hyperintense. In the 6 year old, the marrow signal approaches but remains lower than subcutaneous fat.* **(Right)** *Coronal in- (left) & opposed-phase (right) GRE T1 MRs of a child with sickle cell disease. Chronically stimulated red marrow within the diaphyses & metaphyses drops signal on the opposed-phase image.*



TERMINOLOGY

Synonyms

- Red (hematopoietic) marrow, yellow (fatty) marrow

Definitions

- At birth, red marrow is present throughout entire skeleton
- Conversion to yellow marrow begins shortly after birth
- Reconversion from yellow to red marrow occurs secondary to variety of causes & must be distinguished from pathologic marrow infiltration

IMAGING

General Features

- Best diagnostic clue
  - In predictable pattern, appendicular skeleton converts from red to yellow marrow before axial skeleton
  - Conversion of red to yellow marrow is typically symmetric (i.e., right vs. left)
  - By 12-24 months, most appendicular marrow is generally approaching signal intensity of subcutaneous fat on T1-weighted spin-echo MR pulse sequences
    - On FS T2WI or STIR MR, epiphyseal yellow marrow has greatest concordance with subcutaneous fat
  - Regions of red marrow often persist in spine, pelvis, & long bone metaphyses of normal adults
- Location
  - Appendicular skeleton conversion
    - Begins in bones of distal extremities (i.e., phalanges of fingers & toes) & progresses proximally
    - Within individual long bones
      - Epiphyses convert 1st: Within 6 months of appearance, epiphyseal ossification center should contain almost entirely yellow marrow
      - Central diaphyseal conversion occurs next, spreading proximally & distally toward metaphyses
      - Residual red marrow often persists in adult metaphyses, particularly proximal femurs & humeri
  - Spine: Marrow signal intensity of vertebral bodies changes on spin-echo T1WI MR by approximate ages
    - 0-1 year: Hypointense to intervertebral disc
    - 1-5 years: Isointense to mildly hyperintense to intervertebral disc
    - > 5 years: Hyperintense to intervertebral disc, hypointense to subcutaneous fat
  - Reconversion of yellow to red marrow generally occurs in reverse order as compared to initial conversion
    - Axial → appendicular skeleton
    - Proximal → distal extremities
    - Long bone metaphyses → diaphyses → epiphyses

MR Findings

- T1WI (spin-echo)
  - Yellow marrow shows ↑ signal intensity
  - Red marrow shows ↓ to intermediate signal intensity
    - Mildly ↑ compared to muscle & intervertebral discs
    - Neonatal red marrow may be iso- or ↓ in signal intensity compared to muscle & discs
- In- & opposed-phase T1 GRE
  - Red marrow drops signal intensity on opposed-phase images due to water & fat protons in same voxel
  - Helps distinguish red marrow from infiltrating processes (such as leukemia & metastases), which generally do not drop signal on opposed-phase images
- T2WI FS & STIR
  - Yellow marrow shows ↓ signal intensity, most pronounced in epiphyses
  - Red marrow generally shows signal intensity similar to or mildly ↑ compared to skeletal muscle

DIFFERENTIAL DIAGNOSIS

Residual Red Marrow or Red Marrow Reconversion

- Iso- to mildly hyperintense on T1WI compared to skeletal muscle & intervertebral discs
- Drops signal on opposed-phase MRs
- In long bones, normal residual red marrow often flame-shaped in metaphyses

Systemic Marrow Infiltrating Processes

- Primarily leukemia or metastases
- Diffuse confluent or multifocal well-defined marrow lesions ± cortical destruction, periosteal reaction
- Iso- to hypointense to skeletal muscle & intervertebral discs on T1WI; usually hyperintense on T2WI FS/STIR MR
- No drop of signal intensity on opposed-phase MRs (as cells completely replace fat)

Marrow Deposition Diseases

- Gaucher disease: Most common lysosomal storage disease
  - Distribution & signal intensity can be similar to red marrow reconversion
  - Other imaging findings typically present: Endosteal scalloping, Erlenmeyer flask deformities (undertubulation), bone infarcts, hepatosplenomegaly
- Iron deposition disease
  - Decreased signal intensity on all pulse sequences

PATHOLOGY

General Features

- Differences in fat & water content account for imaging appearances of red & yellow marrow
- Red marrow ~ 40% adipocytes, 60% hematopoietic cells
- Yellow marrow ~ 95% adipocytes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Conversion of red to yellow marrow is asymptomatic normal physiologic process
  - Physiologic stresses, disease processes, & medications may cause reconversion to red marrow

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3. Shah LM et al: MRI of spinal bone marrow: part I, techniques and normal age-related appearances. AJR Am J Roentgenol. 197(6):1298-308, 2011
4. Laor T et al: MR imaging insights into skeletal maturation: what is normal? Radiology. 250(1):28-38, 2009

KEY FACTS

IMAGING

- Growing skeleton demonstrates range of normal age-related radiographic appearances due to
  - Abundant radiolucent growth cartilages
  - Gradual endochondral ossification of such cartilages
- Normal developmental variants have
  - Typical orientation, site, & patient age
  - No overlying swelling or point tenderness
    - May be clouded by isolated soft tissue injury
- MR useful when source of symptoms unclear (e.g., is fragmented growth center incidental normal variant or pathologic due to fracture, infarction, or infection)
  - Marrow edema on T2 FS/STIR MR favors pathology
  - ± soft tissue edema &/or joint effusion

TOP DIFFERENTIAL DIAGNOSES

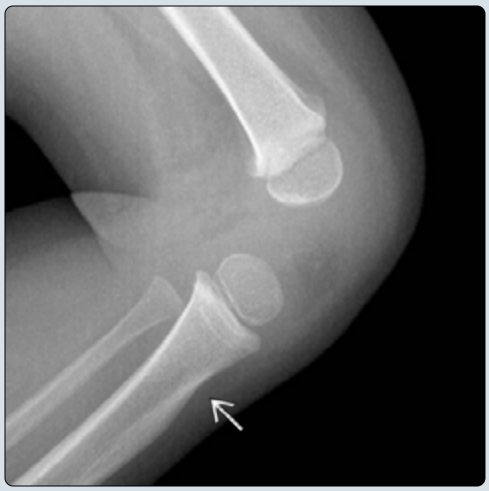
- Physeal fractures
- Incomplete fractures
- Remote trauma
- Child abuse
- Ligamentous disruption
- Avascular necrosis

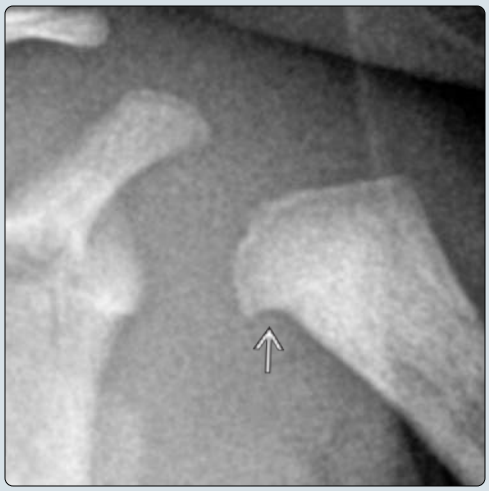
CLINICAL ISSUES

- Usually asymptomatic; come to attention incidentally by radiographs (often with history of regional trauma)

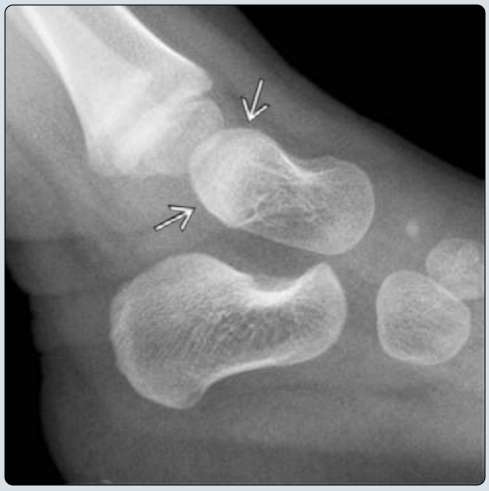
DIAGNOSTIC CHECKLIST

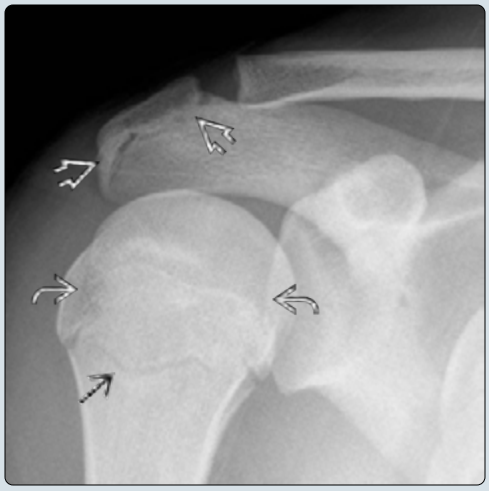
- If unclear whether or not bony appearance is abnormal vs. incidental normal developmental variant in regionally symptomatic child
  - Review prior radiographs
  - Discuss with referring clinician regarding exact site of symptoms (including point tenderness)
  - Consider
    - Contralateral radiographs of asymptomatic side
    - Splinting with follow-up radiographs in 10-14 days to assess for healing changes





(Left) *Lateral radiograph of the knee in a 2-year-old patient shows a normal anterior tibial metadiaphyseal concavity ➔ at the site of the unossified tibial tubercle. Transversely oriented fractures may be seen at this level, but show focal buckling or cortical interruption.* **(Right)** *AP radiograph in a 6-day-old boy shows an accentuated metaphyseal curvature ➔ in the medial proximal humerus, a normal finding in the young child at this level. The curve still flows smoothly with the diaphyseal cortex (passing the "marble test").*





(Left) *Lateral radiograph in a 2-year-old girl shows poorly defined sclerosis in the posterior talus ➔, a commonly seen normal finding in this age group. Linear sclerosis in the posterior calcaneus or cuboid (not present here) would suggest a fracture in this age group.* **(Right)** *AP internal rotation radiograph of the right shoulder in a 15-year-old boy shows an unfused acromion ossification center ➔. The proximal humeral physis is viewed at different heights anteriorly ➔ & posteriorly ⇨ (a common fracture mimic).*

TERMINOLOGY

Definitions

- Skeletally immature patients have numerous radiolucent growth centers composed of cartilage
  - Primary physis: Site of majority of longitudinal growth; lies between epiphysis & metaphysis
  - Secondary physis: Surrounds secondary ossification center (SOC) in epiphysis or equivalent bone
    - Interface where cartilaginous precursor is transformed into bone from central to peripheral
    - Process is uniform in some centers (capitellum, femoral head), nonuniform in others (fragmented trochlea, irregular medial femoral condyle)
  - Apophysis: Nonarticular SOC with muscle/tendon attachment (ischial & tibial tuberosities)
  - Zone of provisional calcification (ZPC): Thin, radiodense line at interface of physeal growth cartilage & newly mineralized bone
- Accessory ossification centers: Separate small rounded ossicles vs. SOCs in same unossified cartilage as adjacent epiphysis or equivalent
- Pseudoepiphysis: False appearance of growth centers at distal 1st & proximal 2nd-5th metatarsals & metacarpals
- Physiologic periosteal reaction (PPR): Smooth, solid, new bone being laid down rapidly along entire lengths of long bone diaphyses in infants

IMAGING

General Features

- Best diagnostic clue
  - No overlying swelling or point tenderness
    - May be clouded by isolated soft tissue injury
  - Orientation, site, & patient age typical for variant
- Morphology
  - SOCs widely spaced at carpals & tarsals before maturity (due to remaining unossified cartilage)
  - Some long bones (radius, ulna) have normal mild degrees of generalized curvature

Radiographic Findings

- Radiography
  - Growth cartilages normally lucent
  - Bony metaphyseal & epiphyseal margins often undulating; may be irregular in some instances
  - Lack of interrupted cortex or ZPC
  - No soft tissue edema (unless isolated soft tissue injury)
  - PPR: Smooth, solid periosteal reaction along shafts of certain long bones (humeri, femurs, tibias)
    - Typical age: 1-6 months
    - Most commonly symmetric

MR Findings

- Useful when source of symptoms unclear
  - Is fragmented growth center incidental normal variant or pathologic (fractured, infarcted, infected)
- Marrow edema on T2 FS/STIR MR favors pathology
  - ± soft tissue edema, joint effusion

DIFFERENTIAL DIAGNOSIS

Physeal Fractures

- Soft tissue edema ± physeal widening & displaced metaphyseal or epiphyseal fragment

Incomplete Fractures

- Buckle fractures: Abnormal focal cortical bump or angulation
  - Imagine marble rolling along cortex: Normally rolls smoothly on diaphysis & metaphysis without bump
- Plastic/bowing deformities: ↑ curvature

Remote Trauma

- Sclerotic margins; soft tissue edema absent
- Differentiation from normal variant not always possible

Child Abuse

- Subtle metaphyseal corner fracture in infant may lead to extensive diaphyseal periosteal reaction
- Typically unilateral

Ligamentous Disruption

- ↑ separation of bones with overlying soft tissue edema
- Stress views helpful

Avascular Necrosis

- Sclerosis, fragmentation, &/or collapse of SOC
- In otherwise healthy child, certain sites & ages typical (such as Legg-Calve-Perthes at hip)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Normal variants usually asymptomatic; come to attention incidentally on imaging (often with history of regional trauma)

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- If unclear whether or not bony appearance is abnormal vs. incidental normal developmental variant in regionally symptomatic child
  - Review prior radiographs
  - Discuss with referring clinician regarding exact site of symptoms (including point tenderness)
- Consider
  - Contralateral radiographs of asymptomatic side
  - Splinting + follow-up radiographs in 10-14 days to assess for healing changes

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2. Oestreich AE: Concave distal end of ulna metaphysis alone is not a sign of rickets. Pediatr Radiol. 45(7):998-1000, 2015
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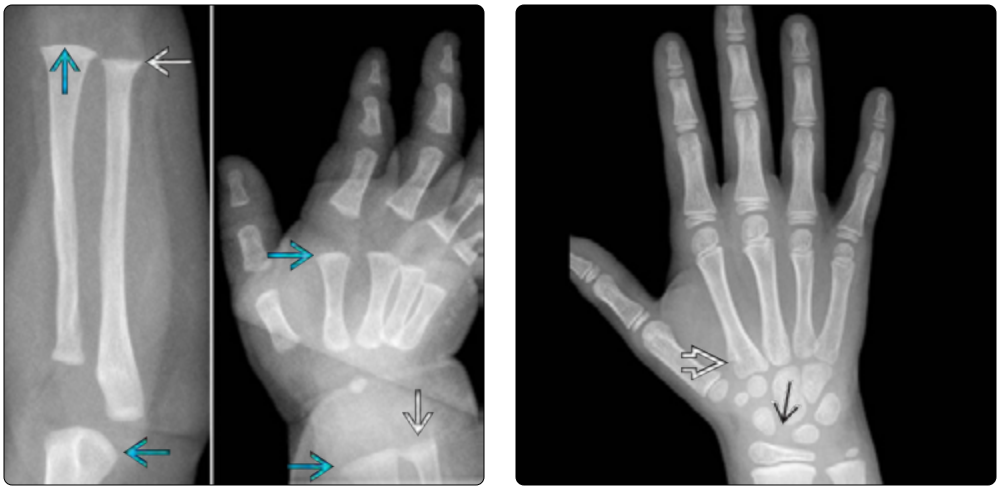
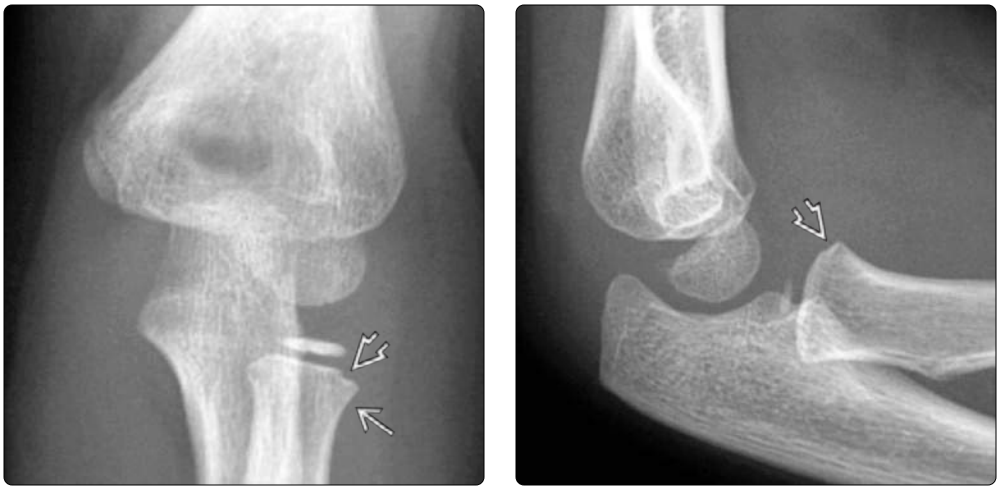
5. Varich LJ et al: Normal maturation of the distal femoral epiphyseal cartilage: age-related changes at MR imaging. Radiology. 214(3):705-9, 2000

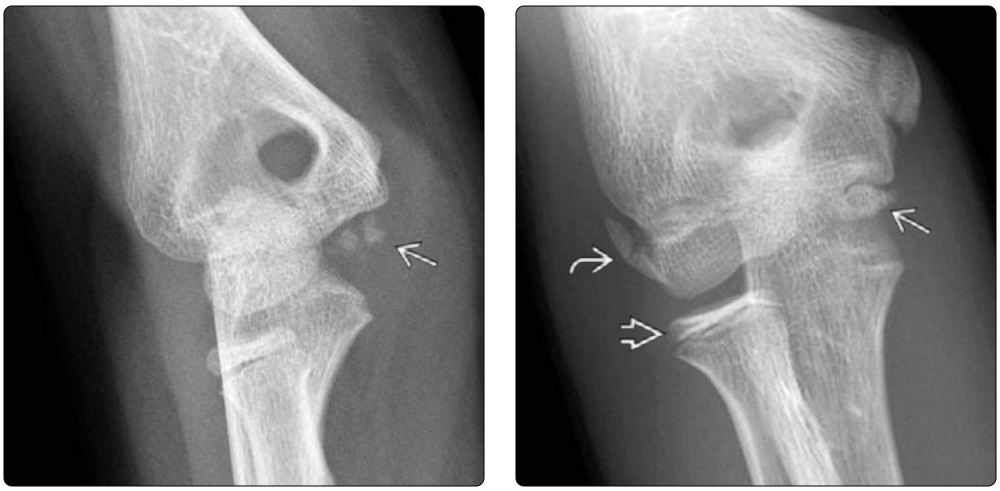
(Left) *AP left forearm & oblique right hand radiographs in a 3 month old with a femur fracture show a normal variant appearance of the distal ulnar metaphyses ➡ mimicking "fraying & cupping." As the remaining growth centers show normal zones of provisional calcification ⇨, underlying rickets is excluded.* **(Right)** *PA radiograph of a 7-year-old girl shows false widening of the scapholunate interval ⇨ due to incomplete carpal ossification in this age. Note the pseudoepiphysis ➡ at the 2nd metacarpal.*

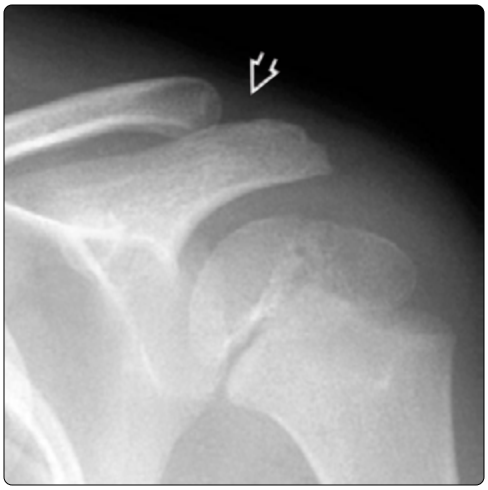


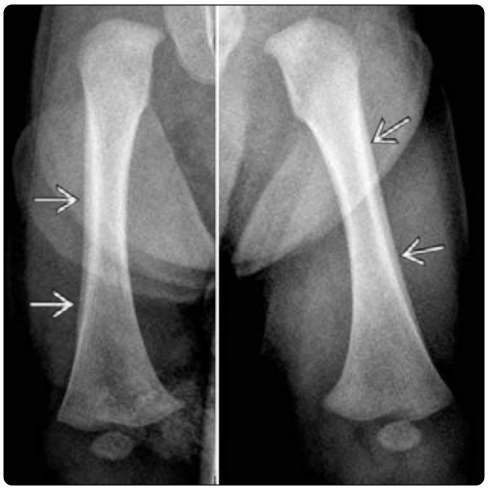
(Left) *AP radiograph of a 7-year-old patient shows a normal location for step-off at the lateral physeal margin of the proximal radial metaphysis ➡. Note the smooth radial metaphyseal neck laterally ➡ (which is a common site for Salter-Harris II injuries).* **(Right)** *Lateral elbow radiograph in a 4-year-old girl shows the commonly seen normal step-off at the anterior & lateral physeal margin of the proximal radial metaphysis ➡.*



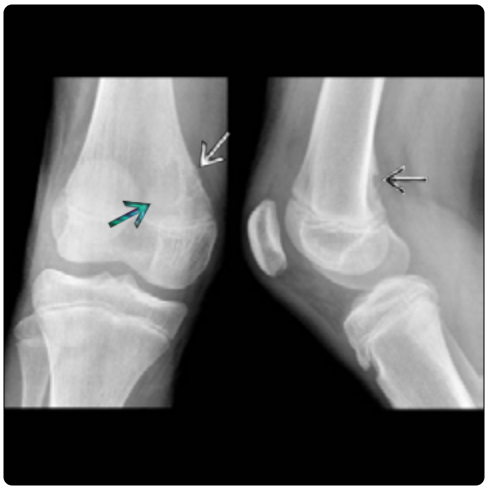
(Left) *Oblique radiograph of the elbow in an 8-year-old girl shows fragmentation of the trochlear ossification center ➡, a normal finding.* **(Right)** *AP radiograph in a 12-year-old patient shows the normal ossification centers about the elbow. The lateral or internal epicondyle ➡ may be fragmented, the radial head ➡ may be sclerotic, & the trochlea ➡ may be irregular, sclerotic, &/or fragmented.*

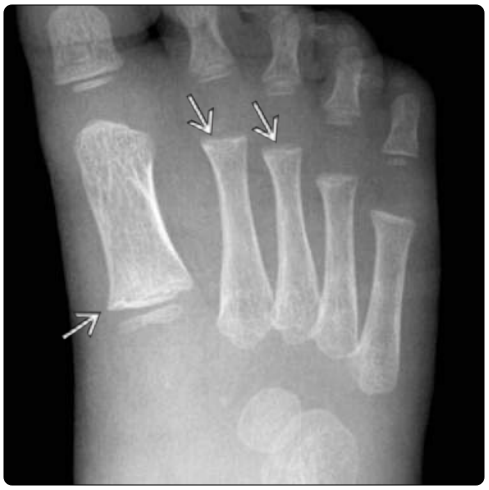




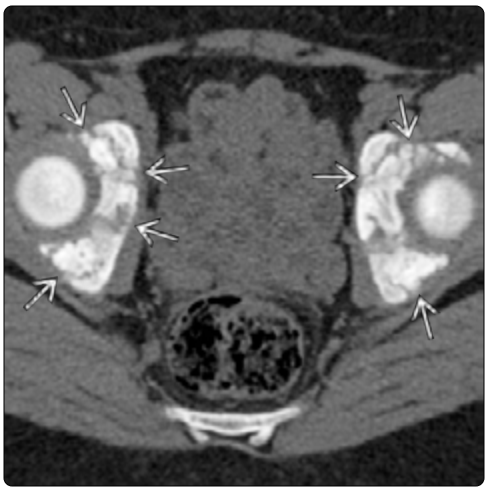


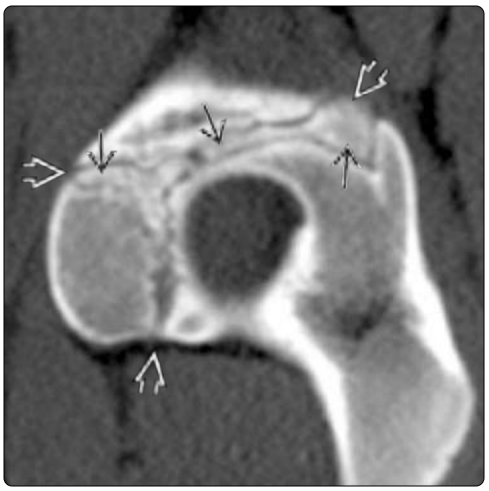
(Left) *AP radiograph in a 4-year-old patient shows normal "widening" of the acromioclavicular (AC) joint ➡ due to lack of acromion ossification in this age. Any suspicion for AC joint separation should prompt bilateral stress views for asymmetry (though this injury is uncommon in young patients).* **(Right)** *AP femur radiographs in a 1-month-old patient show thin, smooth, solid periosteal reaction along the lateral aspects of the femoral diaphyses bilaterally ➡, typical of physiologic periosteal reaction.*





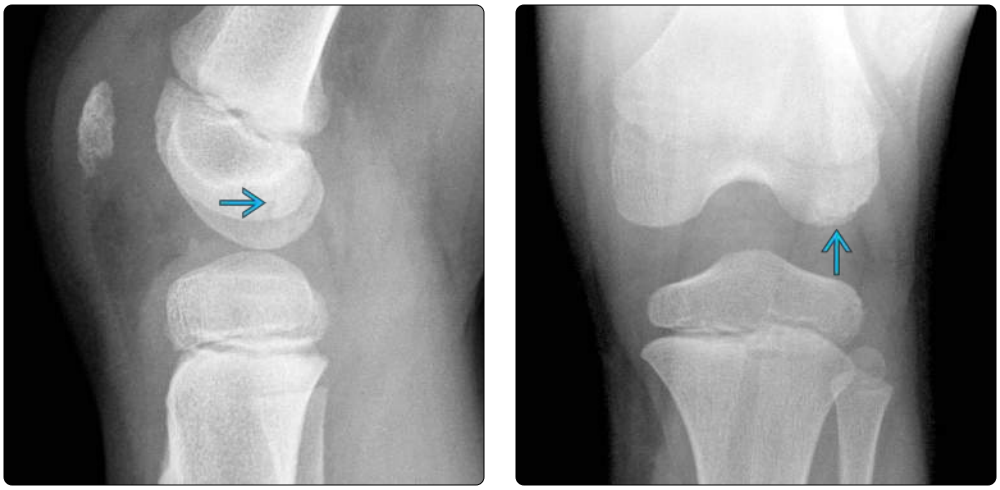
(Left) *AP (left) & lateral (right) radiographs show a typical location & configuration of avulsive irregularity of the posteromedial distal femur. A well-defined sclerotic margin ➡ is often present with mild medial ➡ & posterior ➡ irregularity being less frequent. Any lesion at this location appearing more aggressive than this should get further work-up.* **(Right)** *AP foot radiograph in a 2-year-old boy shows normal metaphyseal undulation ➡ at the base of the 1st metatarsal as well as the distal 2nd & 3rd metatarsals.*



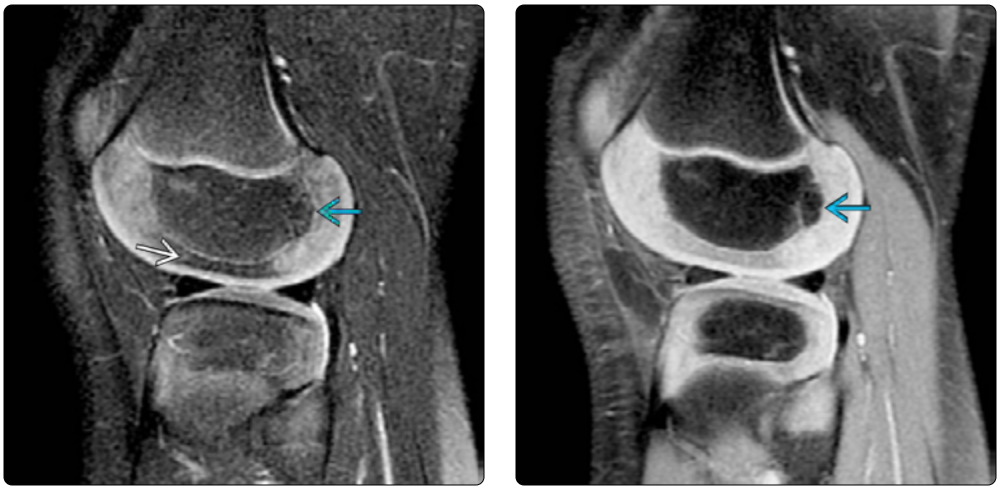


(Left) *Axial NECT in a 7-year-old patient shows multiple ossification centers within the triradiate cartilages of the medial acetabula bilaterally ➡.* **(Right)** *Sagittal NECT image of the same patient more clearly displays the arrangement of the accessory ossification centers ➡ within the triradiate cartilage ➡ of the medial acetabular wall.*

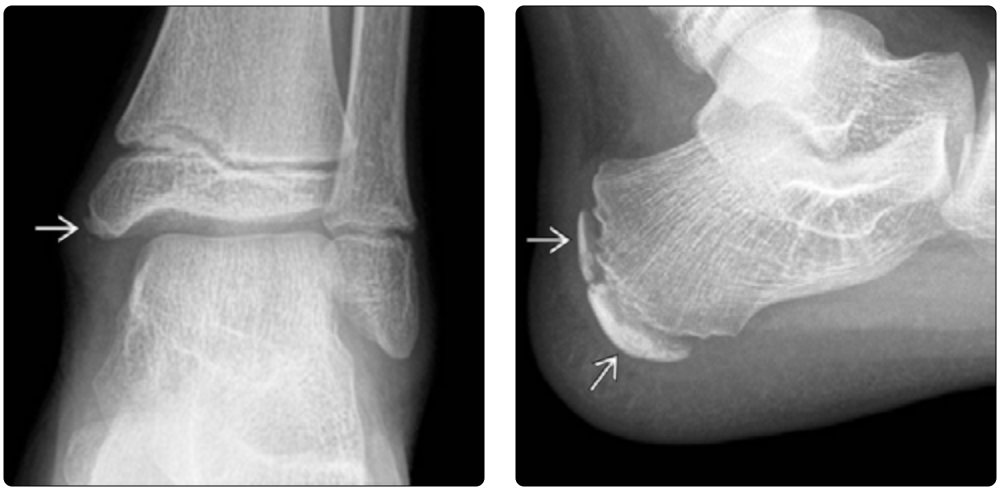
(Left) *Lateral radiograph in a 6 year old with knee pain shows an irregular lucency in the posterior lateral femoral condyle ⇨. (Right) Tunnel view of the same knee shows the irregularity/fragmentation ⇨ of the posterior non-weight-bearing aspect of the lateral femoral condyle, a common location for developmental irregular ossification. This is in contrast to osteochondritis dissecans (OCD), which more classically affects the lateral & central aspects of the medial femoral condyle.*

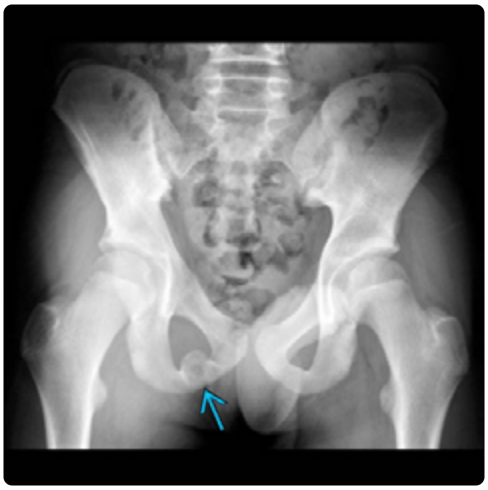


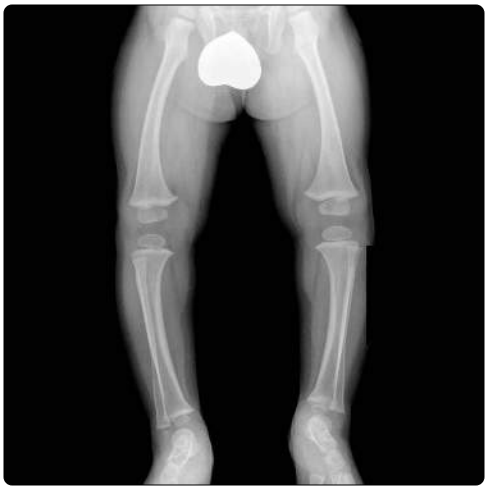
(Left) *Sagittal T2 FS MR in the same patient shows minimal ossific fragmentation posteriorly ⇨ without adjacent marrow edema, cystic change, cartilage fissuring, or secondary growth plate interruption to suggest a true OCD lesion. Also note the normal dark signal of the weight-bearing unossified femoral condyle ➡ due to water displacement in the cartilage. (Right) Sagittal PD FS MR in the same patient shows the ossific puzzle piece fragmentation of the posterior lateral femoral condyle ⇨, a normal variant.*



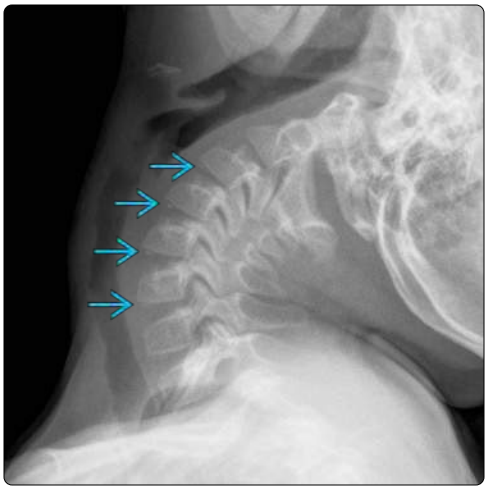
(Left) *AP ankle radiograph in a 9-year-old boy shows an ovoid ossification center with sclerotic margins at the medial malleolus ➡. This is a very common location for accessory ossification centers (which may be fragmented). (Right) Lateral radiograph of the calcaneus in a 10-year-old patient shows a normal fragmented & sclerotic appearance of the calcaneal apophysis ➡. Patients may have symptoms related to recurrent traction here (Sever disease), but this entity is not diagnosed by bony radiographic changes.*

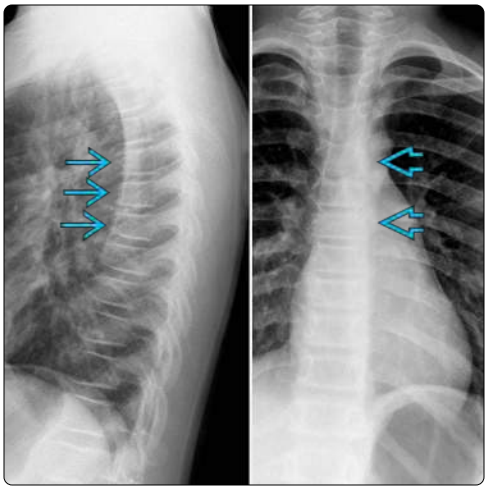




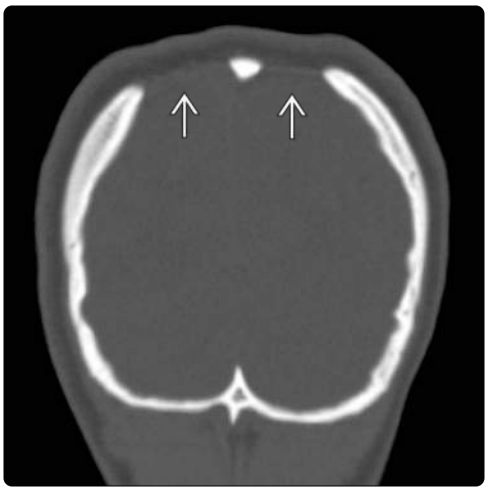


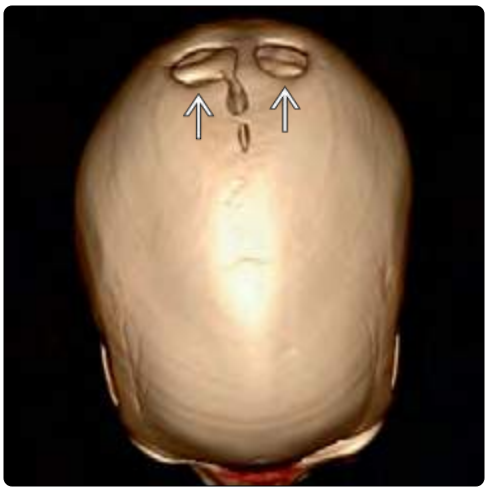
(Left) *AP radiograph of the pelvis in a 14-year-old boy with a coccygeal injury shows a mixed lucent & sclerotic expansile lesion at the right ischiopubic synchondrosis. This is a recognized & typically asymptomatic normal variant occurring in some patients around puberty.* **(Right)** *AP radiograph of the lower extremities in a child with genu varum shows physiologic bowing of the tibias & femurs. Note that there is only mild medial down-sloping of the proximal tibial metaphyses without fragmentation (in contrast to Blount disease).*





(Left) *Lateral cervical spine extension radiograph in a 6 year old without trauma shows normal mild anterior wedging of vertebral bodies due to incomplete ossification. Note the small ring apophyses.* **(Right)** *Chest radiographs in a 10 year old with chest pain but no trauma show very mild gradual anterior wedging of several mid-thoracic vertebral bodies without focal concavity. The vertebral body heights are maintained from left to right on the AP view. This normal wedging can be difficult to separate from traumatic compression.*





(Left) *Coronal NECT in an 11 year old with a history of headaches shows a typical location & morphology of giant parietal foramina, an appearance which was stable from a CT scan 2 years prior. Note the smoothly tapered bony margins without permeation or periosteal reaction.* **(Right)** *Surface-rendered 3D vertex (face down) view of the skull from the same NECT shows the relatively symmetric paramidline giant parietal foramina. These normal variants are often smaller than the example seen here.*

Distal Femoral Avulsive Irregularity

KEY FACTS

TERMINOLOGY

- Common pediatric finding at posterior, medial distal femoral metaphysis: Cortical defect deep to medial gastrocnemius or adductor magnus attachments
- Likely due to chronic avulsive forces
- Typically incidental lesion; no treatment needed
- Synonyms
  - Avulsive cortical irregularity or tug lesion
  - Cortical desmoid: Misnomer as biology & histology differ from true desmoid tumors (which are locally aggressive benign neoplasms)

IMAGING

- Lucent focus of cortical interruption/irregularity at posterior, medial distal femoral metaphysis
- Deep margin commonly concave ± sclerosis
- May have adjacent cortical thickening
- Frontal view may show well-defined round/ovoid lucent focus with sclerotic rim or mild medial cortical irregularity
- Bilateral: 25-100%

TOP DIFFERENTIAL DIAGNOSES

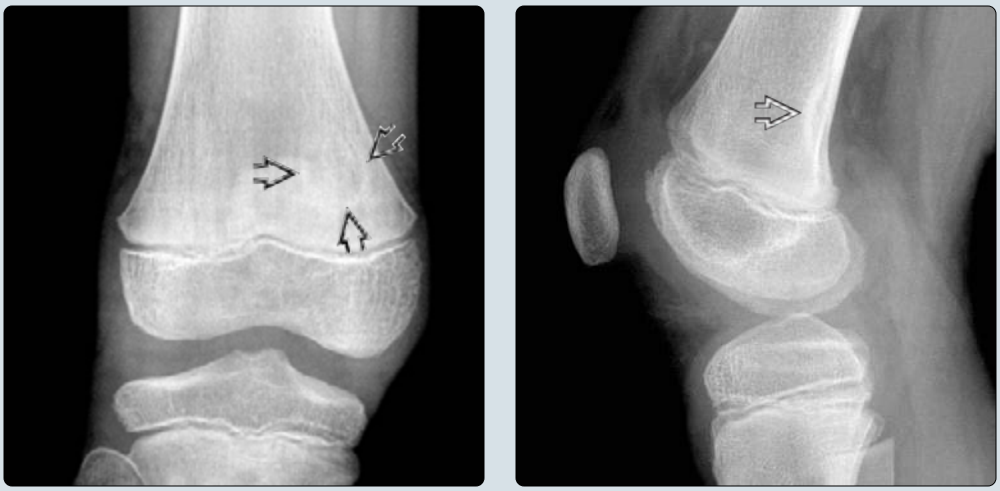
- Osteosarcoma
- Osteomyelitis
- Langerhans cell histiocytosis
- Fibroxanthoma
- Periosteal/juxtacortical chondroma
- Osteoid osteoma
- Metastases

PATHOLOGY

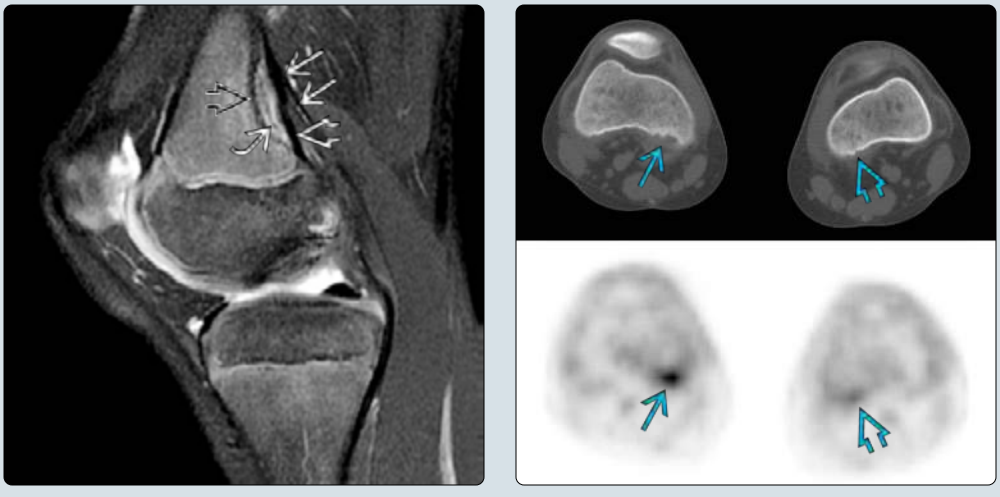
- Benign, self-limited reactive process (not neoplasm)
- Needle biopsy may lead to inappropriate treatment
  - Therefore a "don't touch" lesion

DIAGNOSTIC CHECKLIST

- Location of this entity very typical
- More aggressive entities also occur here
  - Get MR or contralateral radiographs if unclear



(Left) *AP radiograph from a 10-year-old boy shows a common appearance of distal femoral avulsive irregularity (DFAI) in the medial distal femoral metaphysis. It is well circumscribed, ovoid, & centrally lucent with mildly sclerotic margins. (Right) Lateral radiograph in the same patient shows a scalloped appearance of the posterior distal femoral cortex at the site of the lesion. There is no periosteal reaction.*



(Left) *Sagittal T2 FS MR through the same lesion shows heterogeneous signal intensity centrally. There is typical, concave low signal intensity at the sclerotic deep margin. The overlying periosteum (which is normally thickened at this level) is intact deep to the medial gastrocnemius muscle attachment. (Right) Axial CT (top) & FDG PET (bottom) images in a teenager with a known chest wall Ewing sarcoma (not shown) demonstrate increased metabolic activity at bilateral foci of DFAI, right > left.*

TERMINOLOGY

Synonyms

- Benign cortical irregularity of distal femur
- Avulsive cortical irregularity or tug lesion
- Cortical desmoid
  - Misnomer as biology & histology differ from true desmoid tumors (which are locally aggressive benign neoplasms)

Definitions

- Irregularity of posterior, medial distal femoral metaphyseal cortex deep to attachments of gastrocnemius muscle medial head or adductor magnus aponeurosis
- Benign self-limited lesion likely due to chronic avulsion

IMAGING

General Features

- Best diagnostic clue
  - Typically incidental radiographic finding at classic location without soft tissue mass or symptoms
- Location
  - Bilateral: 25-100%
  - Left:right = 2:1
- Morphology
  - Round vs. elongated with femoral long axis
  - May appear as
    - Scalloped, concave, saucer-shaped cortical defect with well-defined border
    - Bulging, convex cortical lesion with irregular surface
    - Divergent lesion splitting cortex

Radiographic Findings

- Lateral view (key to visualization)
  - Lucent focus of cortical interruption/irregularity
    - Deep margin commonly concave ± sclerosis
    - Solid periosteal reaction or cortical thickening may abut lesion
    - Lesion may contain bony spicules
- Frontal view may show round or ovoid lucent focus with sclerotic rim &/or subtle medial cortical irregularity
- Important negative findings
  - No associated soft tissue abnormality
  - No aggressive periosteal reaction

MR Findings

- Interruption of contiguous smooth cortex by intermediate T1/heterogeneous T2 signal intensity lesion
- Lesion may enhance on T1 C+ sequences
- Thin, concave, T1/T2 hypointense deep rim
- Overlying periosteum intact
- ± mild adjacent marrow edema
  - More likely symptomatic
- No overlying soft tissue mass

Nuclear Medicine Findings

- Bone scan
  - Healing lesions show mildly ↑ uptake
  - Inactive lesions show no uptake
- PET/CT
  - ↑ metabolic activity; accompanying CT diagnostic

DIFFERENTIAL DIAGNOSIS

Osteosarcoma

- Destructive bone lesion most commonly arising from intramedullary distal femoral metadiaphysis
- Aggressive features are rule, not exception

Osteomyelitis

- Overlying soft tissue edema early
- Radiographic bone changes in 10-14 days
  - Lucent cortical destruction/permeation

Langerhans Cell Histiocytosis

- Flat bones > metaphyseal/diaphyseal long bones
- Classically: Geographic lytic lesion
- Sclerotic margins with healing

Fibroxanthoma/Nonossifying Fibroma

- Overlaps distal femoral avulsive irregularity in some cases
- Eccentric multilobulated lucent metaphyseal/metadiaphyseal lesion
- Thin sclerotic margin; narrow zone of transition

Periosteal/Juxtacortical Chondroma

- Uncommon benign surface lesion with scalloping & buttressing of cortex ± chondroid matrix

Bone Metastases

- Neuroblastoma, leukemia most common < 10 years of age
- Lucent metaphyseal bands ± permeative destruction, aggressive periosteal reaction

Osteoid Osteoma

- Small lucent, highly vascular nidus ± central calcification
- Adjacent periosteal reaction, cortical thickening, soft tissue & marrow edema

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually asymptomatic; local pain uncommon

Demographics

- Age: 3-17 years; most common: 10-15 years
- Gender: M:F = 1.4-3:1

Treatment

- None if asymptomatic
- With localizing symptoms & no other etiologies, conservative therapy may relieve repetitive stress injury
- Avoid unnecessary biopsy

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VACTERL Association

KEY FACTS

TERMINOLOGY

- Nonrandom association of anomalies involving multiple organ systems (except brain)
  - **V**ertebral/vascular
  - **A**nal atresia/auricular
  - **C**ardiac
  - **T**racheoesophageal fistula
  - **E**sophageal atresia
  - **R**enal/radial/rib
  - **L**imb
- VACTERL association diagnosed when ≥ 3 of above malformations present; causative gene unknown

IMAGING

- Actively seek other features of VACTERL association when 1-2 components present
- Initial imaging in suspected cases: Radiographs & US
  - Radiographs: Spine & limbs (if limb anomaly on physical exam)
  - US: Head, spine, renal/bladder, echocardiography
- Additional & advanced imaging depending on imaging & clinical exam findings

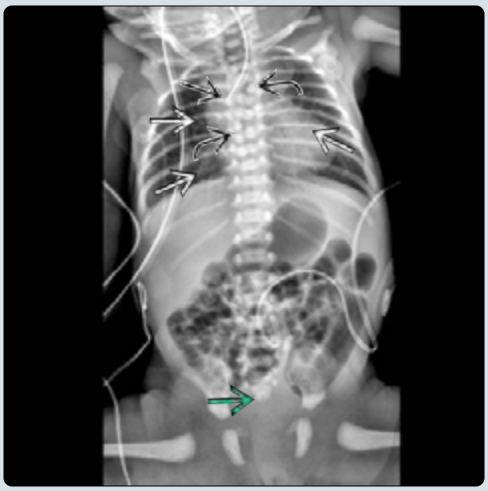
CLINICAL ISSUES

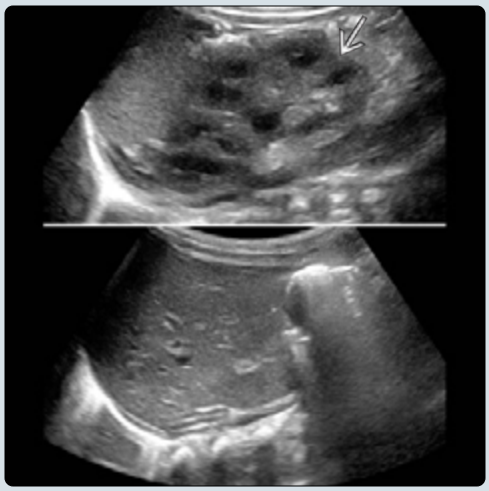
- Incidence of VACTERL association: 1/10,000 to 40,000 liveborn infants
- Children with VACTERL: 72% have 3 anomalies, 24% have 4 anomalies, 8% have 5 anomalies
- Frequency of anomalies in VACTERL
  - Cardiac: 40-80%
  - Renal: 50-80%
  - Anal: 55-90%
  - Tracheoesophageal: 50-80%
  - Vertebral: 60-80%
  - Limb: 40-50%

DIAGNOSTIC CHECKLIST

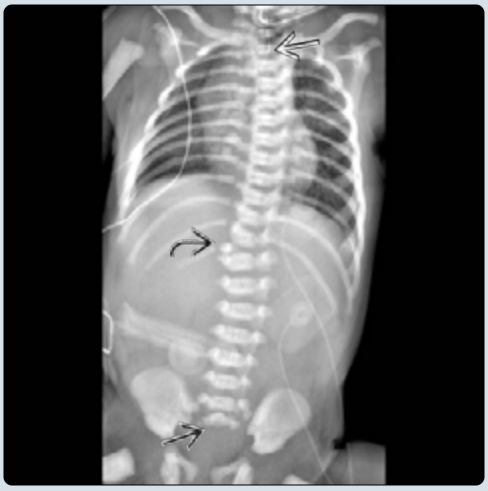
- Consider VACTERL in child with vertebral & other anomalies

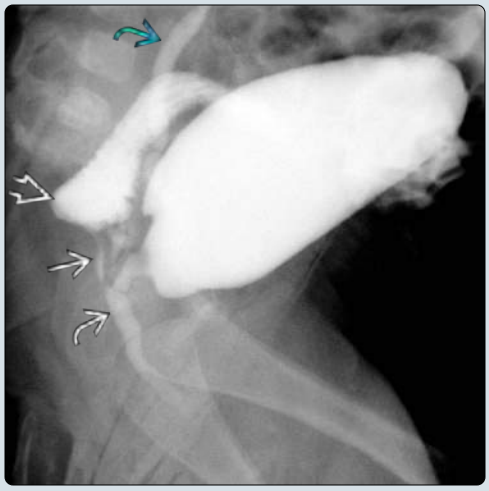
(Left) *Frontal radiograph in a newborn shows vertebral segmentation anomalies. The nasogastric tube (NG) could not be advanced beyond the upper esophagus due to esophageal atresia (with the bowel gas indicating an associated tracheoesophageal fistula). Mild central pulmonary vascular congestion is secondary to a VSD. There is partial sacral agenesis in this child with an ARM.* **(Right)** *Longitudinal US images of the left (top) & right (bottom) renal fossae in the same patient show a solitary left kidney.*





(Left) *Frontal radiograph in a newborn shows vertebral segmentation anomalies & partial agenesis of the sacrum. The NG tube could not be passed beyond the upper esophagus due to esophageal atresia (with the lack of abdominal gas indicating the absence of a tracheoesophageal fistula).* **(Right)** *Lateral view from a VCUG in the same child shows a rectourethral fistula from the rectal pouch to the posterior urethra. Left vesicoureteral reflux is also seen in this child with an ARM.*





TERMINOLOGY

Abbreviations

- Anorectal malformation (ARM)
- Tracheoesophageal fistula (TEF)
- Esophageal atresia (EA)

Synonyms

- VATER, VACTER, VACTEL association, TREACLE, ARTICLE, ARTICLE V, axial mesodermal dysplasia spectrum

Definitions

- Nonrandom association of anomalies involving multiple organ systems (except brain)
  - **V**ertebral/vascular
  - **A**nal atresia/auricular
  - **C**ardiac
  - **T**racheoesophageal fistula
  - **E**sophageal atresia
  - **R**enal/radial/rib
  - **L**imb
- VACTERL association diagnosed when ≥ 3 of above malformations present
  - Clubfoot & hip dysplasia excluded if no other limb anomalies present
- No clinical or laboratory evidence of alternative diagnosis

IMAGING

General Features

- Best diagnostic clue
  - Vertebral anomalies in presence of other malformations

Radiographic Findings

- Radiography
  - Axial skeleton
    - Vertebral anomalies
      - Cleft, block, butterfly vertebrae; hemivertebrae; hypersegmentation; vertebral bars; caudal regression
      - Secondary scoliosis, kyphosis
    - Other spine issues: Cord tethering (8-78%)
    - Ribs: Fused; bifid; hypoplastic; supernumerary/cervical
  - Limbs or extremities
    - Radial ray: Dysplastic or absent radius; radioulnar synostosis; thumb hypoplasia; radial polydactyly; absent scaphoid; radial artery hypoplasia
    - Hands: Polydactyly (20%) most common; syndactyly
    - Reduction deformities (34%): Aplasia/hypoplasia of humerus, radius, femur, tibia, or fibula
  - Head & neck
    - Choanal atresia, cleft lip/palate, auricular defects
  - Chest
    - Congenital heart disease: Ventricular septal defect (VSD) (30%); patent ductus arteriosus (26%); atrial septal defect (ASD) (20%)
    - EA/TEF
    - Lung agenesis, horseshoe lung (posterior lung fusion), ectopic bronchus
  - Abdomen & pelvis
    - Imperforate anus ± fistula
    - Microgastria, duodenal atresia, malrotation, Meckel diverticulum

Ultrasonographic Findings

- Prenatal US may suggest diagnosis
- 1 study showed detection rate on standard prenatal US ~ 50%
- Prenatal detection rates of VACTERL anomalies
  - Renal malformations: 45%; TEF: 44%; cardiac malformations: 20%; vertebral: 13%; limb: 11%
- Neonatal head US to evaluate for findings suggestive of other diagnoses (e.g., hydrocephalus)
- Spinal US to evaluate for tethered cord
- Renal/bladder US to evaluate for renal/GU anomalies
  - Renal agenesis most common (bilateral in ~ 13%); multicystic dysplasia; horseshoe kidney; ectopia; hydronephrosis
  - Persistent urachus; cryptorchidism

Imaging Recommendations

- Actively seek other features of VACTERL when 1-2 components present
- Multidisciplinary group suggests specific work-up for VACTERL in
  - All infants with 2 features of VACTERL association
  - All infants with TEF/EA
  - All infants with ARM
- Radiographs & US used initially for imaging in suspected cases
  - Radiographs: Spine & limbs (if limb anomaly on physical exam)
  - US: Head, spine, renal/bladder, echocardiography
  - Additional & advanced imaging depending on radiographic & clinical exam findings

DIFFERENTIAL DIAGNOSIS

Alagille Syndrome

- Butterfly vertebra & cardiac anomalies ± renal anomalies
- Differs from VACTERL: Bile duct paucity/cholestasis, ophthalmologic anomalies, neurological anomalies, characteristic facies, *JAG1* mutations in > 90%

CHARGE Syndrome

- Cardiac malformations, GU anomalies ± TEF
- Differs from VACTERL: Coloboma, cranial nerve dysfunction, characteristic facial features, *CHD7* mutations in > 50%

Currarino Syndrome

- Partial sacral agenesis, presacral mass, & ARM
- Differs from VACTERL: Presacral mass, *MNX1* mutations

Fanconi Anemia

- Radial deficiency & many other features of VACTERL
- Differs from VACTERL: Pancytopenia; pigmentation anomalies

Holt-Oram Syndrome (Heart-Hand Syndrome)

- Cardiac conduction defect ± ASD/VSD; thumb, wrist, & forearm abnormalities
- Differs from VACTERL: Heterozygous mutations in *TBX5* gene

Oculoauriculovertebral Syndrome (Goldenhar Syndrome)

- Vertebral anomalies, cardiac abnormalities, limb abnormalities, urogenital anomalies
- Differs from VACTERL: Ear anomalies, hemifacial microsomia

Thrombocytopenia/Absent Radius Syndrome

- Differs from VACTERL: Thrombocytopenia

VACTERL-H (VACTERL + Hydrocephalus)

- Severe mental retardation; poor prognosis
- Differs from VACTERL: Hydrocephalus, various known mutations

22q11.2 Deletion Syndrome (DiGeorge/Velocardiofacial Syndrome)

- Cardiac malformations, renal anomalies
- Differs from VACTERL: Hypocalcemia, immune dysfunction, characteristic facial features, deletion of 1 copy of chromosome 22q11.2

PATHOLOGY

General Features

- Etiology
  - Classified as association due to no unifying causative gene identified
    - Association: Grouping of anomalies more frequently than expected by chance
  - Current theory: Malformations occur during blastogenesis → developmental field defect → polytopic anomalies affecting multiple organ systems
  - Exact etiology unknown
- Genetics
  - *SHH* mutations theorized based on animal models
    - Not established in humans
  - Associated with trisomies 13 & 18, *cri du chat* syndrome, Fanconi anemia
- Associated abnormalities
  - Cleft palate: 18%
  - Neural tube defect: 10%
  - Diaphragmatic hernia: 8%
  - Omphalocele: 6%
  - Exstrophy of cloaca
  - Congenital pulmonary airway malformation
  - Sirenomelia

Staging, Grading, & Classification

- No definitive consensus of criteria for VACTERL
  - Most require ≥ 3 components for diagnosis
- Other etiologies in differential should be excluded based on clinical & laboratory work-up

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Neonatal: Depends on anomaly constellation
- Other signs/symptoms
  - Prenatal imaging
    - Polyhydramnios (EA), kyphoscoliosis, absent radius, cardiac &/or renal anomalies, single umbilical artery
  - Prematurity: ~ 1/3
  - Stillborn: 12%

Demographics

- Epidemiology
  - 1/10,000 to 40,000 liveborn infants
  - Frequency of anomalies in VACTERL
    - Cardiac: 40-80%
    - Renal: 50-80%
    - Anal: 55-90%
    - Tracheoesophageal: 50-80%
    - Vertebral: 60-80%
    - Limb: 40-50%
  - Concurrence of 2 specific VACTERL anomalies 11x more frequent than expected by chance
    - Probably belong to VACTERL continuum
  - Concurrence of ≥ 3 specific VACTERL anomalies 95x more frequent than expected by chance
  - Children with VACTERL: 72% have 3 anomalies, 24% have 4 anomalies, 8% have 5 anomalies
    - Most common 3-anomaly combinations: Cardiac-renal-limb & cardiac-renal-anal
    - Most common 5-anomaly combination: Cardiac-renal-limb-anal-tracheoesophageal fistula

Natural History & Prognosis

- Mortality (not due to any specific defect)
  - 28% neonatal mortality
  - 48% mortality in 1st year
- Intelligence usually normal

Treatment

- If prenatal diagnosis: Delivery at tertiary care facility

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Consider VACTERL in child with vertebral & other anomalies

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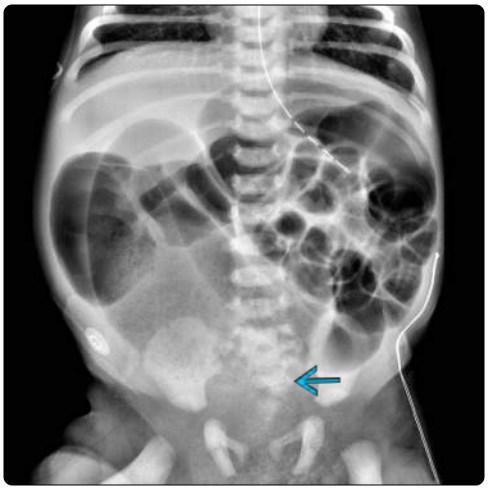
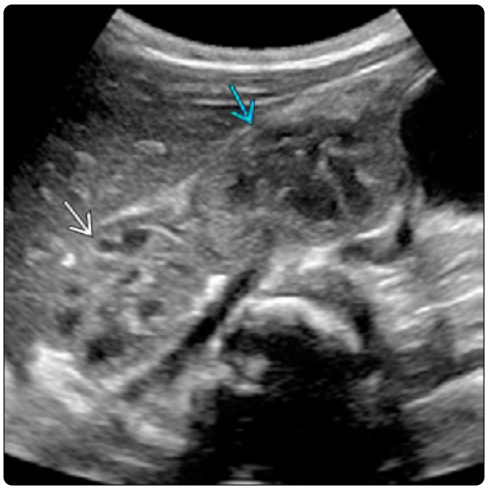
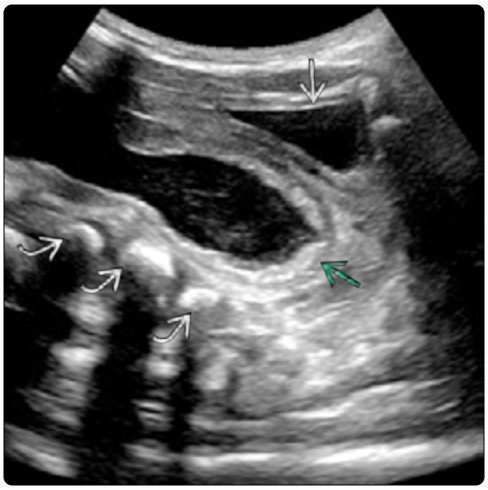
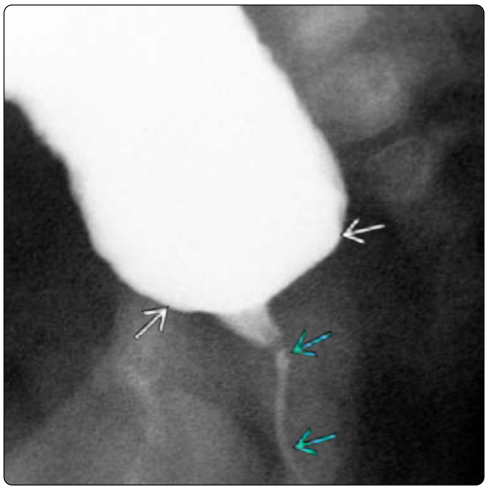


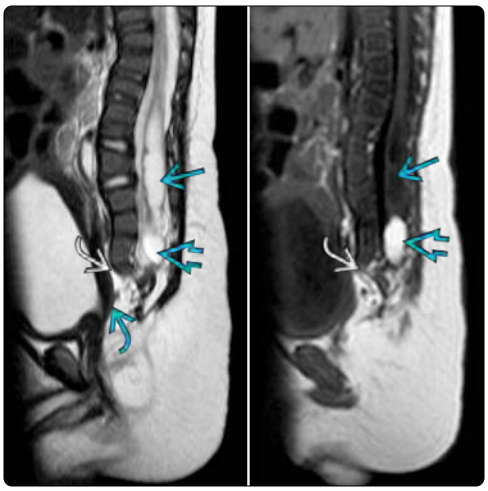


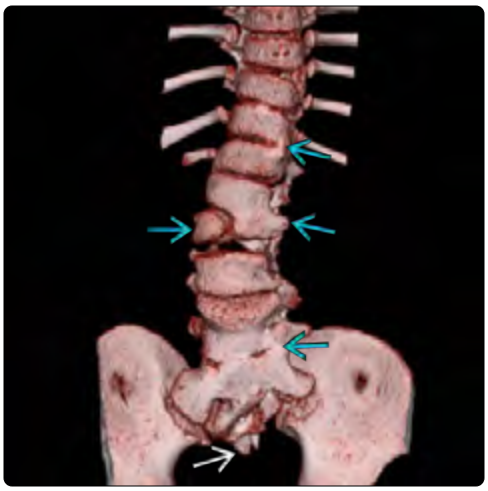
(Left) *Frontal abdominal radiograph in a newborn boy with an ARM shows diffusely dilated bowel loops, consistent with a distal bowel obstruction. The sacrum has an anomalous morphology, nearly scimitar.* **(Right)** *Transverse US of the right renal fossa in the same patient shows a cross-fused ectopic left kidney, which is inferior to & fused to the normally positioned right kidney.*





(Left) *Sagittal US of the pelvis in the same boy shows the blind-ending distal rectum, consistent with an ARM. The urinary bladder is seen anteriorly, & echogenic sacral vertebral bodies are seen posteriorly.* **(Right)** *Lateral fluoroscopic image of the pelvis from a distal colostogram in the same boy with an ARM shows a long rectoperineal fistula extending from the distal rectal pouch to the perineum.*





(Left) *Sagittal T2 (left) & T1 MRs in a 3-month-old boy with VACTERL show partial agenesis of the sacrum. There is a low-lying conus with associated hydrosyringomyelia & a terminal lipoma. The decompressed blind-ending rectum does not extend to the perineum, consistent with this child's known ARM.* **(Right)** *3D surface-rendered CT of the same patient at 10 years of age shows multiple vertebral anomalies & partial sacral agenesis.*

Polydactyly

KEY FACTS

TERMINOLOGY

- Polydactyly: Extra digits of hands or feet
  - Preaxial: Radial side of hand, tibial side of foot
  - Postaxial: Ulnar side of hand, fibular side of foot
  - Central (mesoaxial): Involves central digits
  - Mirror-image polydactyly: Central thumb/great toe-like digit with variable duplication of 2nd-5th digits
- Syndactyly: Fusion of digits
  - Simple (soft tissue fusion) vs. complex (osseous fusion)
  - Complete (entire digit) vs. incomplete (spares distal digit)
- Polysyndactyly: Polydactyly + syndactyly (soft tissue ± osseous fusion of digits)

IMAGING

- Radiographs of affected hand/foot done primarily to detect skeletal elements within extra digit(s)
- Extra digit can range from small entirely soft tissue skin tag → rudimentary digit with hypoplastic bones → fully developed extra digit
- Affected phalanges, metacarpals/metatarsals may be bifurcated or duplicated
- Skeletal survey indicated if syndromic association suspected

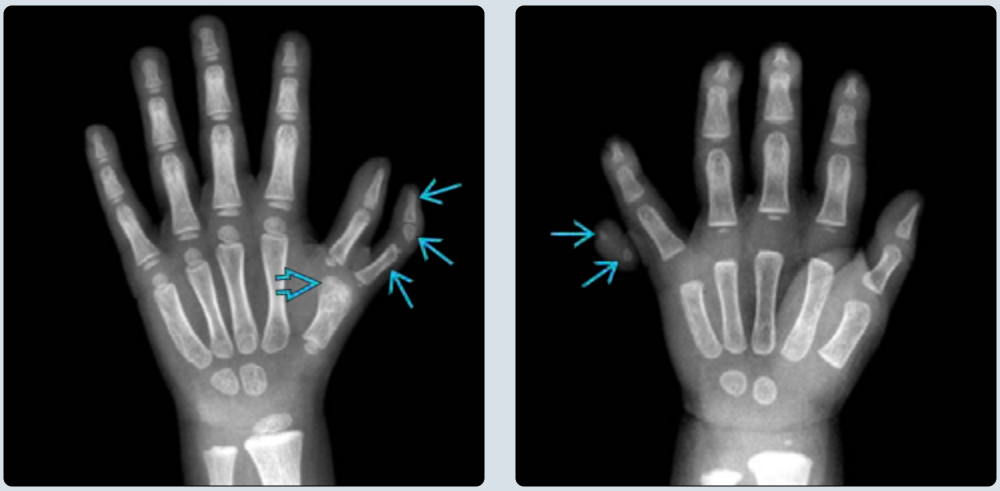
PATHOLOGY

- Most cases sporadic & isolated
- However, ~ 300 syndromes associated with polydactyly
  - Trisomies (13, 18, 21), Meckel-Gruber, VACTERL, Ellis-van Creveld, etc.

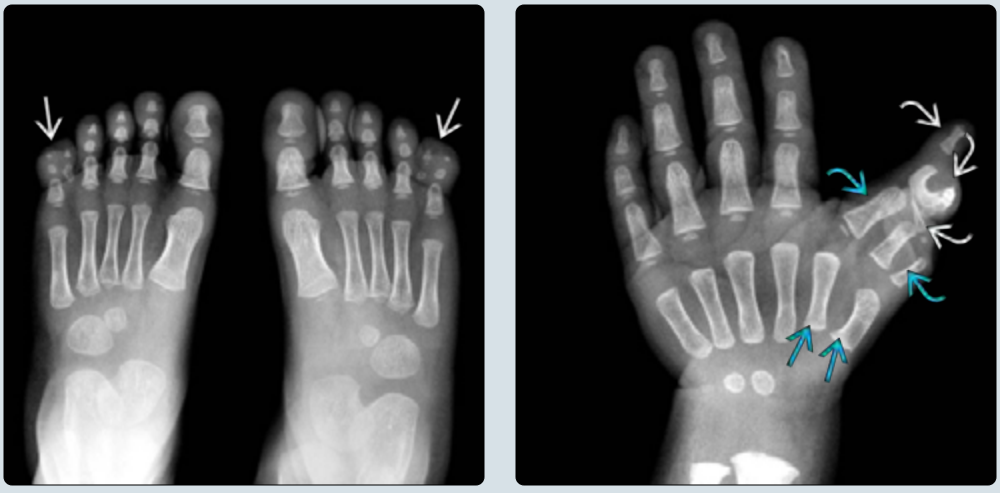
DIAGNOSTIC CHECKLIST

- If discovered on prenatal US or MR, search for other anomalies required
- Description of each digital anomaly should include
  - Degree of bifurcation/duplication of each phalanx
  - Morphology of metacarpal/metatarsal (duplicated, bifurcated "Y" or "T", broadening of head)
  - Any associated soft tissue (± osseous) syndactyly

(Left) *PA radiograph shows the left hand of a 2-year-old girl with preaxial polydactyly. The duplicated thumb is triphalangeal ⇨ (Wassel type VII). The distal end of the 1st metacarpal is broad ⇨ but not duplicated.* **(Right)** *PA radiograph shows the left hand of a 2-year-old boy with postaxial polydactyly (type B). There is an incompletely formed 6th digit, which contains 2 small, incompletely formed phalanges ⇨.*



(Left) *AP radiograph of both feet in a 2-year-old girl shows bilateral postaxial polydactyly (type B) with duplication of the 5th middle & distal phalanges ⇨.* **(Right)** *PA radiograph of the left hand in a girl with multiple limb anomalies shows a complex preaxial polydactyly with 2 metacarpals ⇨, 3 proximal phalanges ⇨, & a complex array of middle & distal phalanges ⇨ with soft tissue & osseous fusion.*



KEY FACTS

TERMINOLOGY

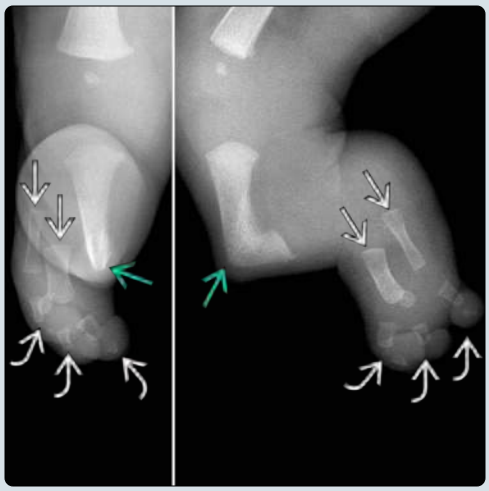
- Hemimelia: Absence of all or portion of distal limb
  - Transverse hemimelia: Defects of both tibia & fibula
  - Paraxial hemimelia: Defect of only tibia (preaxial) or fibula (postaxial)
  - Terminal hemimelia: Foot bones absent to some degree
  - Intercalary hemimelia: Foot spared

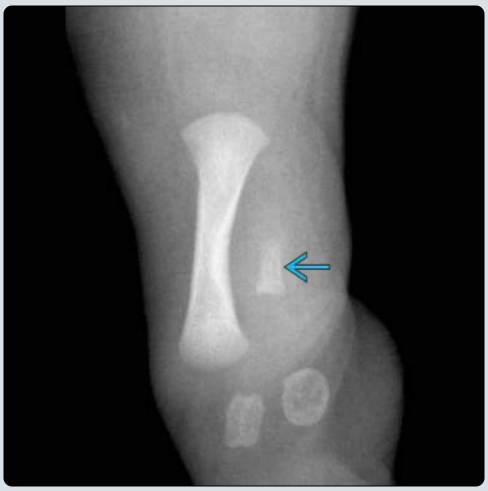
IMAGING

- Radiographs remain primary imaging modality for detection & characterization of hemimelia
- Both lower extremities should be imaged from hips to feet with AP & lateral views to characterize
  - Specific hemimelia defects
  - Associated limb shortening/limb length discrepancy
  - Other lower extremity bony anomalies
- US & MR more accurately define extent of
  - Cartilaginous deficiencies
  - Soft tissue anomalies

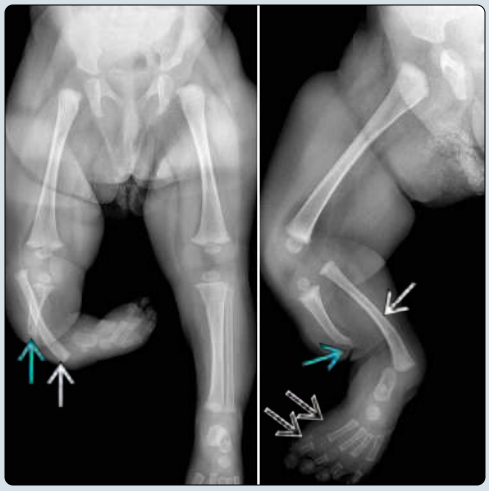
DIAGNOSTIC CHECKLIST

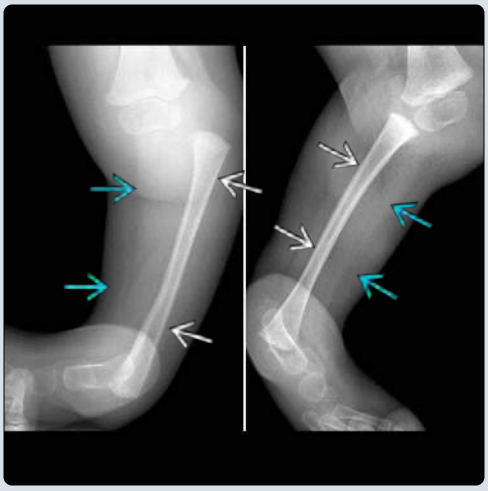
- Differentiating fibular from tibial hemimelia
  - Fibular hemimelia associations
    - Tibial bowing (typically with anteromedial apex)
    - Absent lateral digits
    - Valgus foot deformity
  - Tibial hemimelia associations
    - Increased fibular width
    - Absent medial digits
    - Varus foot deformity
- Report details should include descriptions of
  - Complete vs. partial absence of each leg bone
    - If partial: Approximate fraction & position of affected segment
  - Specifics of foot involvement
    - Be aware of radiographically limited tarsal assessment in young children due to ossification timing
  - Associated lower extremity anomalies





(Left) *Frontal (left) & lateral (right) radiographs in a 2-week-old girl with fibular hemimelia show complete absence of the fibula. The tibia is shortened with anteromedial bowing. There is an associated foot deformity with only 2 well-formed metatarsals & 3 digits visualized.* **(Right)** *Frontal radiograph in a 2 day old with partial fibular hemimelia shows only a small hypoplastic fibula. All the digits in the foot were present (not shown).*





(Left) *Frontal (left) & lateral (right) radiographs in a 3 month old with partial tibial hemimelia show absence of the distal right tibia. The right fibula is thickened. There is an associated preaxial foot anomaly with absence of a normal 1st metatarsal & great toe.* **(Right)** *Frontal (left) & lateral (right) radiographs of the left leg in a 1 year old with tibial hemimelia show complete absence of the tibia. The left fibula is intact but thickened. An associated foot anomaly is partially visualized.*

Tibial Bowing

KEY FACTS

TERMINOLOGY

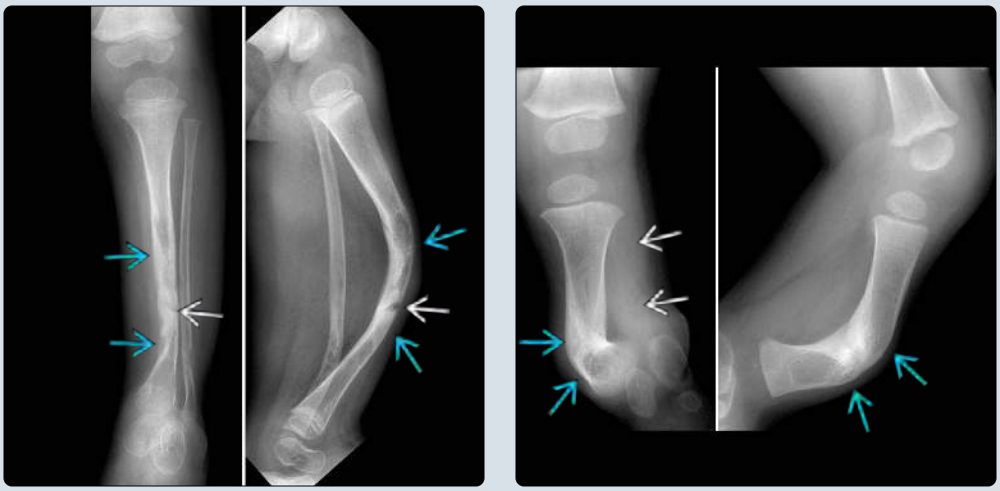
- Tibial bowing: Typically unilateral congenital or infantile diaphyseal deformity characterized by direction of apex
  - Posteromedial: Physiologic bowing frequently secondary to intrauterine positioning
  - Anteromedial: Associated with fibular hemimelia
  - Anterolateral: High association with neurofibromatosis type 1

IMAGING

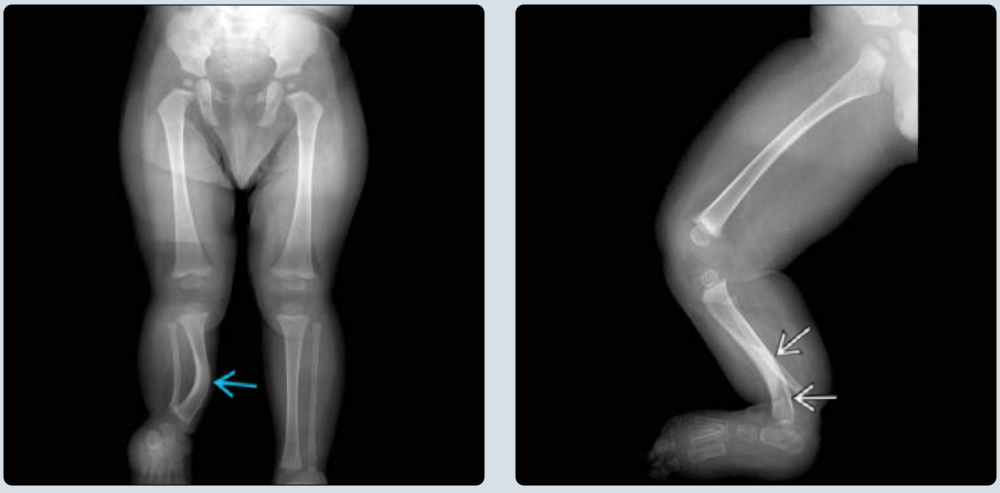
- Frontal & lateral leg radiographs necessary to characterize apex of deformity
  - If tibial bowing present, evaluate for
    - Cortical thickening or thinning
    - Fracture or pseudarthrosis
    - Underlying lesion or abnormal mineralization
    - Fibular hemimelia
    - Associated knee, ankle, & foot deformities
- Standing leg length radiograph
  - Characterizes limb length discrepancy
- Additional radiographs to evaluate for associated anomalies
  - Foot radiographs
    - Posteromedial bowing: Calcaneovalgus
    - Anteromedial bowing with fibular hemimelia: Absent rays, talipes equinovalgus, tarsal coalitions
- Advanced imaging may be used to evaluate associated conditions affecting management
  - Knee MR in fibular hemimelia may show congenital ligamentous & meniscal abnormalities

DIAGNOSTIC CHECKLIST

- Depending on apex direction/type, tibial bowing should prompt search for associated anomalies
  - Affects local diagnosis, prognosis, & therapy
  - May impart underlying systemic diagnosis



(Left) *Frontal (left) & lateral (right) radiographs in a 2-year-old boy with neurofibromatosis type 1 show anterolateral bowing of the mid tibial diaphysis with associated increased sclerosis & cortical thickening. There is a developing fracture, which ultimately progressed to a pseudarthrosis.* **(Right)** *Frontal (left) & lateral (right) radiographs in an 8 month old with fibular hemimelia show anteromedial tibial bowing associated with absence of the fibula.*



(Left) *This frontal radiograph of the lower extremities in a 1 year old shows medial tibial bowing on the right. There is shortening of the right tibia & fibula with an associated limb length discrepancy.* **(Right)** *Lateral radiograph in the same child as in the prior image shows posterior bowing of the tibia. An associated flatfoot deformity is suggested, though weight-bearing radiographs are required to adequately assess disorders of foot alignment.*

KEY FACTS

TERMINOLOGY

- Arthrogryposis multiplex congenita (AMC): Descriptive term for infant born with ≥ 2 joint contractures
  - **Not** specific diagnosis (> 300 underlying causes)

IMAGING

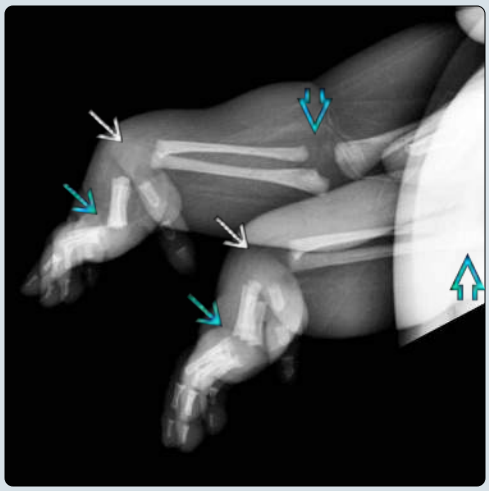
- Radiographs of affected limbs typically performed
  - Static/fixed abnormal deviations/orientations of joints
    - Positioning for standard views difficult
  - Gracile & osteoporotic bones with muscle wasting
  - Foot radiographs should simulate weight-bearing to accurately diagnose associated alignment disorders
- MR
  - Brain/spine evaluation if neurologic exam abnormal
  - Extremities show ↓ muscle bulk with fatty replacement in many conditions
  - May aid in preoperative assessment of joints (e.g., soft tissue bands, abnormalities of unossified cartilage)

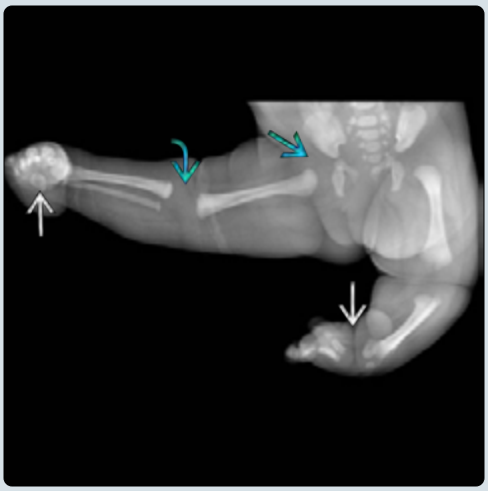
PATHOLOGY

- Broad categories of AMC etiologies
  - Neurologic (70-80%): Various CNS/PNS anomalies
    - Forebrain malformations, spinal muscular atrophy
  - Primary myopathies (~ 20%)
    - Amyoplasia (most common cause of AMC overall)
    - Distal arthrogryposis, congenital muscular dystrophies
  - Connective tissue disorders
    - Skeletal dysplasias, multiple pterygium syndromes
  - Maternal exposures: Teratogens, infections
  - Intrauterine compression: Oligohydramnios, multiple gestations, uterine anomalies

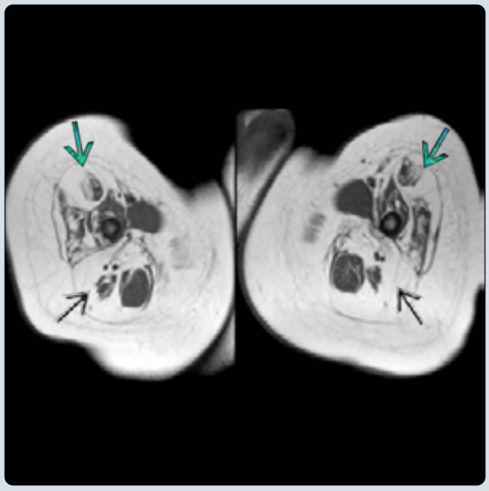
CLINICAL ISSUES

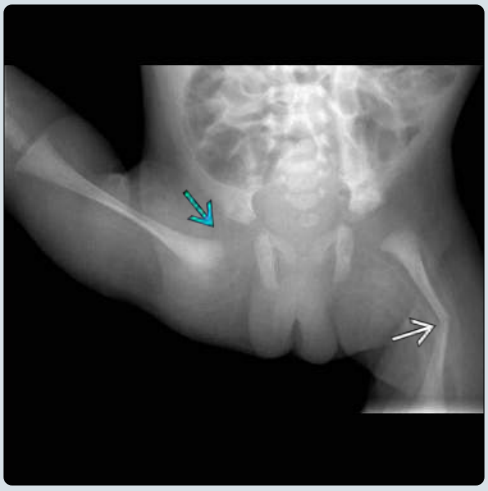
- Perinatal fractures common in AMC
- Initial treatment of contractures typically nonoperative: Passive manipulation & serial casting
- Operative treatments: Soft tissue release, tendon transfer, various osteotomies





(Left) *Lateral radiograph of both arms in a newborn with arthrogryposis multiplex congenita (AMC) due to amyoplasia shows persistent extension at the elbows with the forearms held straight out from the body. The wrists show hyperflexion with hyperextension at the MCP joints.* **(Right)** *Lower extremity radiograph in the same patient shows right hip flexion & right knee hyperextension. Bilateral talipes equinovarus deformities are incompletely evaluated on this non-weight-bearing exam.*





(Left) *Axial T1 MR in a 5 month old with AMC due to amyoplasia shows severe loss of muscle bulk of the bilateral quadriceps & hamstring muscles with associated fatty replacement. Fatty & fibrous replacement of muscles is typical of amyoplasia.* **(Right)** *Frontal radiograph of the pelvis in a newborn with AMC shows right hip hyperflexion. Both femurs are gracile (overtubulated) with muscle wasting of the bilateral thighs. There is a fracture of the left femoral diaphysis. Perinatal fractures are common in infants with AMC.*

KEY FACTS

TERMINOLOGY

- Synonym: Talipes equinovarus
- **C**avus, forefoot **a**dductus, hindfoot **v**arus & **e**quinus (CAVE)
- Plantarflexion of calcaneus relative to tibia (equinus) + hindfoot inversion (varus) + forefoot adduction (varus)

IMAGING

- Talus: Lateral rotation within ankle joint
  - Talus point of reference for hindfoot
- Calcaneus: Relative medial rotation + equinus
- Navicular: Medial subluxation on talus
- Cuboid: Medial subluxation on calcaneus
- Metatarsals: Inverted, appear parallel on lateral view
- AP view: "Laterally pointing" hindfoot + adducted forefoot
  - Long axis of talus very lateral to 1st metatarsal
  - Long axis of calcaneus lateral to 5th metatarsal
- Measurements made on weightbearing views
  - ↑ tibiocalcaneal angle on lateral view (> 90° = calcaneal equinus)
  - ↓ talocalcaneal angle on lateral & AP views (hindfoot varus)
  - ↑ talus-1st metatarsal angle (forefoot varus)
- Frequently detected on screening 2nd-trimester ultrasound
- Prenatal MR performed for other abnormalities (e.g., myelomeningocele) may detect clubfoot

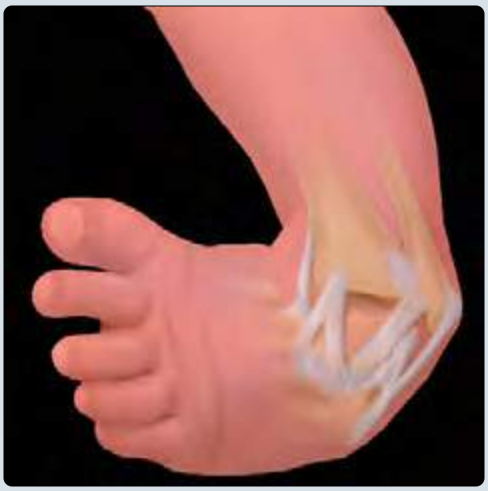
PATHOLOGY

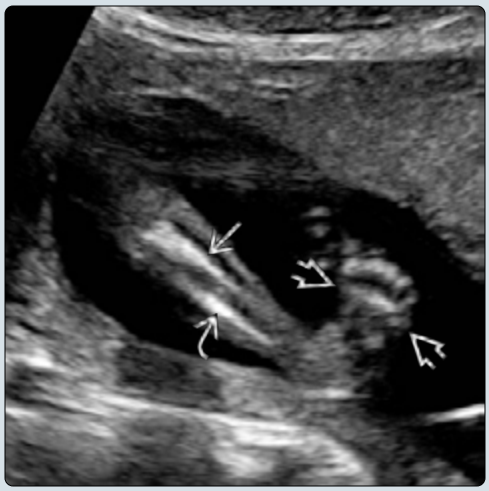
- Isolated, idiopathic, congenital form most common
- Additional anomalies in 24-50%
  - Myelomeningocele, arthrogryposis, myotonic dystrophy
  - Various syndromes (trisomies 18, 21)
- Association with intrauterine "packing disorders" (e.g., oligohydramnios, twinning)

CLINICAL ISSUES

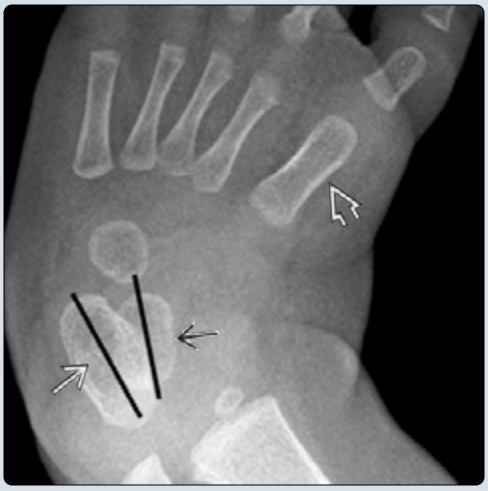
- 50% bilateral
- Treatment primarily conservative with manipulation & casting; selective use of surgery

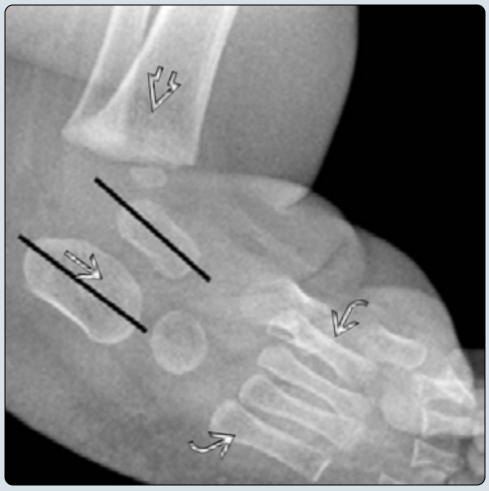
(Left) *Oblique graphic shows a clubfoot with equinus, inversion, & forefoot adduction.* **(Right)** *Sagittal US through the lower leg of a 27-week gestational age fetus with amniotic band syndrome shows abnormal positioning of the foot relative to the foreleg with the tibia ➡, fibula ➡, & all metatarsals ➡ visible in their long axes on a single image (consistent with clubfoot).*





(Left) *AP radiograph in a 9 month old with clubfoot shows hindfoot varus with a reduced talocalcaneal angle (black lines). There is varus angulation of the forefoot ➡ with the long axis of the talus ➡ far lateral to the 1st metatarsal & the long axis of the calcaneus ➡ lateral to the 5th metatarsal.* **(Right)** *Lateral view shows hindfoot varus with a reduced talocalcaneal angle (black lines). The angle between the long axes of the tibia ➡ & calcaneus ➡ is > 90° (hindfoot equinus). The inverted metatarsals ➡ appear parallel.*





TERMINOLOGY

Abbreviations

- **C**avus, forefoot **a**dductus, hindfoot **v**arus, hindfoot **e**quinus (CAVE)

Synonyms

- Talipes equinovarus

Definitions

- Plantarflexion of calcaneus relative to tibia (equinus) + hindfoot inversion (varus) + forefoot adduction (varus)

IMAGING

General Features

- Best diagnostic clue
  - AP view: "Laterally pointing" hindfoot + adducted forefoot
    - Long axis of talus very lateral to 1st metatarsal; long axis of calcaneus lateral to 5th metatarsal

Radiographic Findings

- Radiography
  - Angles measured on weightbearing images
  - ↑ tibiocalcaneal angle (calcaneal equinus)
    - Lateral view: Normal 70-90°, > 90° in clubfoot
  - ↓ talocalcaneal angle (hindfoot varus)
    - AP view: Normal 20-40°, ~ 0-10° in clubfoot
    - Lateral view: Normal 35-50°, ~ 10-20° in clubfoot
  - ↑ talus-1st metatarsal angle (forefoot varus)
    - AP view normally 0-15°, ~ 20-40° in clubfoot
  - Talus: Lateral rotation within ankle joint
    - Talus point of reference for hindfoot
  - Calcaneus: Relative medial rotation + equinus
  - Navicular: Medial subluxation on talus
  - Cuboid: Medial subluxation on calcaneus
  - Metatarsals: Inverted, appear parallel on lateral view

MR Findings

- Prenatal MR can detect clubfoot

Ultrasonographic Findings

- Prenatal
  - Affected foot short, plantarflexed, bent medially
- Postnatal
  - Dynamic visualization of unossified cartilage

Imaging Recommendations

- Protocol advice
  - Measurements must be made on weightbearing or simulated weightbearing views

DIFFERENTIAL DIAGNOSIS

Metatarsus Adductus

- Forefoot varus without other findings of clubfoot

Congenital Vertical Talus

- Talar plantarflexion, navicular displaced, hindfoot equinus

Rocker Bottom Foot

- Foot foreshortened & convex ("Persian slipper")
- May be associated with clubfoot repair

Amniotic Band Syndrome

- Ranges from extremity ring constrictions (edema → fracture → amputation) to major craniofacial & visceral defects

Vertical Calcaneus in Myelodysplasia

- Imbalance of ankle/foot dorsi/plantar flexion → vertical calcaneal rotation

PATHOLOGY

General Features

- Etiology
  - Isolated, idiopathic congenital most common
  - Additional anomalies: 24-50%
    - Neuromuscular etiologies: Myelomeningocele, arthrogryposis, myotonic dystrophy
    - Numerous syndromic associations
  - Intrauterine factors with ↑ risk: Intrauterine growth restriction, twinning, oligohydramnios, amniotic bands, 1st-trimester amniocentesis
  - Acquired/postnatal onset seen in cerebral palsy (> 5 years)
- Genetics
  - Genetic basis of isolated cases suspected: ~ 25% family history & ↑ risk in mono (33%) vs. dizygotic (3%) twins
  - Likely multifactorial &/or polygenic in nature

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Hindfoot: Varus & equinus
  - Forefoot: Adduction
  - Stiffness: Ankle & foot
  - 50% bilateral

Demographics

- Age: Most commonly congenital
- Gender: M:F = 2.5:1

Natural History & Prognosis

- Untreated → lateral weightbearing → ↑ equinus & inversion → ↑ lateral column growth → ↑ deformity/stiffness

Treatment

- Goals: ↑ mobility, ↓ pain, ↓ stiffness
  - Nonsurgical: Birth to 12 months (better if earlier)
    - Ponseti method: Serial casting ± Achilles tenotomy
  - Surgery: Not primary therapy, used to correct deformities after conservative treatments

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Discoid Meniscus

KEY FACTS

IMAGING

- Modality of choice: MR
  - Sagittal images with confirmation on coronal data sets
    - Sagittal: Evaluate periphery
      - Complete "bow ties" on ≥ 3 consecutive images (with 4- to 5-mm slices) suggestive
    - Coronal: Central images key
      - > 50% coverage of lateral tibial plateau
      - > 13- to 14-mm transverse width
      - ≥ 2 mm in height > medial meniscus
  - Frequent linear or amorphous ↑ intrameniscal signal (intermediate to hyperintense on PD, T2, or T2* GRE) in slab-like meniscus
    - Difficult to determine if intrameniscal signal indicates tear, vascularity, mucoid degeneration, or cyst

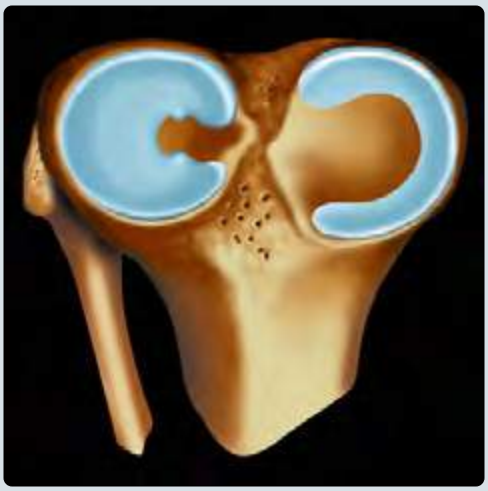
PATHOLOGY

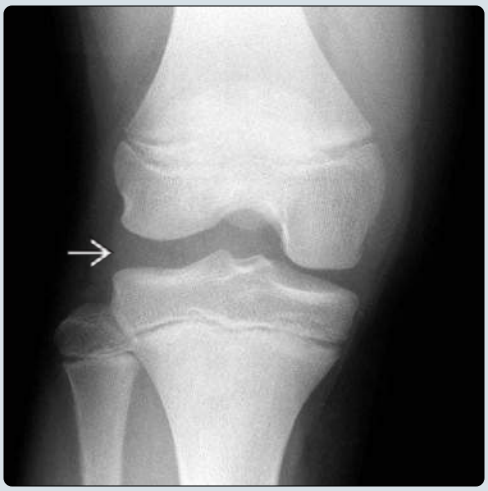
- Watanabe classification
  - Complete discoid meniscus: Stable
    - Extends into intercondylar notch on coronal images
  - Incomplete: Stable
    - No extension into intercondylar notch
  - Wrisberg ligament type: Unstable, hypermobile
    - Lacks posterior meniscal attachments

CLINICAL ISSUES

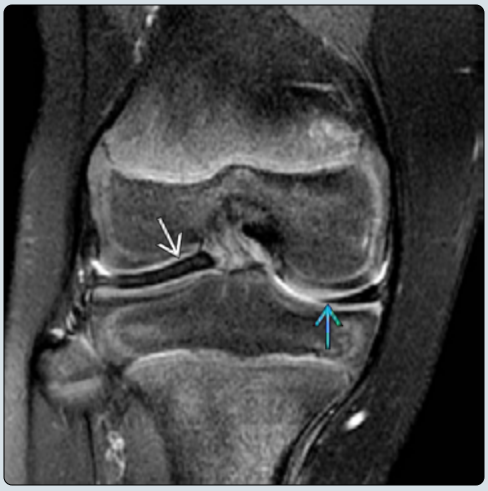
- Most commonly asymptomatic in children
  - Symptoms may not develop until adolescence or later
  - Snapping knee syndrome: Wrisberg type
    - < 10 years old
    - Snaps in flexion & extension
  - Symptomatic in older children from tears or unstable variant: Locking, pain, clicking
- Treatment
  - Asymptomatic: Typically observe, no surgical treatment
  - Symptomatic: Saucerization & repair
    - Arthroscopic goal: Width of peripheral rim remaining at 5-8 mm

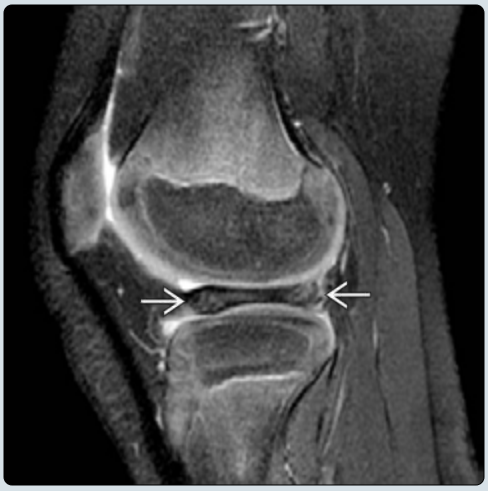
(Left) *Graphic shows a discoid lateral meniscus with minimal resorption of the central portion & > 50% coverage of the lateral tibial plateau.* **(Right)** *AP standing radiograph shows widening of the lateral joint compartment. There is subtle cupping of the lateral tibial plateau. This appearance proved to be a discoid lateral meniscus on MR & subsequent arthroscopy.*





(Left) *Coronal T2 FS MR in an 8 year old with knee pain & popping after playing basketball shows a complete discoid lateral meniscus. Note the slab-like appearance with the lack of tapering of the inner margin of the lateral meniscus compared to the normal tapering of the medial meniscus.* **(Right)** *Sagittal T2 FS MR in the same child shows the slab-like appearance of the discoid lateral meniscus.*





TERMINOLOGY

Definitions

- Large, congenitally dysplastic meniscus with loss of normal semilunar shape

IMAGING

General Features

- Best diagnostic clue
  - Continuity of anterior & posterior horns on ≥ 3 consecutive MR sagittal images (with 4- to 5-mm slices)
    - Central meniscal portion covers > 50% of articular surface of lateral tibial plateau
    - Loss of normal semilunar morphology
    - Coronal images most accurate
- Location
  - Lateral > > medial discoid meniscus
  - Bilateral: 20%
- Size
  - ≥ 2 mm in height > medial meniscus
  - > 13-14 mm in cross section on central coronal image (for both partial & complete discoid meniscus)
    - Normal: 5-13 mm from capsular margin to free edge on central coronal image
  - Ratio of minimal meniscal width:maximal tibial width of ≥ 20% on coronal
- Morphology
  - Large slab/pancake-like complete discoid meniscus
  - Partial discoid meniscus tapers centrally

Radiographic Findings

- Discoid lateral meniscus
  - Normal radiographs in most
  - Widened lateral joint space (best on weight-bearing view)
  - High fibular head
  - Hypoplastic or squared femoral condyle
    - Condylar cutoff sign: ↓ prominence of lateral condyle adjacent to intercondylar notch on tunnel view (with complete discoid meniscus)
  - Hypoplastic lateral tibial spine
  - Cupping of lateral tibial plateau

MR Findings

- Continuous body with continuity of anterior & posterior horns ("bow ties") on ≥ 3 consecutive sagittal images (with 4- to 5-mm thick slices)
- Meniscal size > 13-14 mm in cross section on central coronal image
  - Normal hypointense meniscus: 5-13 mm from capsular margin to free edge on central coronal image
- Complete discoid meniscus has pancake-like appearance (does not taper medially)
  - Hypointense slab-like meniscus
  - Extending from intercondylar notch to periphery of compartment
- Partial discoid meniscus: Tapers centrally but too wide & tall peripherally
- Frequent linear or amorphous increased intrameniscal signal (intermediate to hyperintense on PD, T2, or T2* GRE)
  - Difficult to determine if intrameniscal signal indicates tear, vascularity, mucoid degeneration, or cyst
    - Within substance of meniscus (intrameniscal tear) vs. extending to articular surface (tear)
      - Extensive signal abnormality may reflect tear
    - Discoid menisci more prone to more complex tears
- Prominent ligament of Wrisberg
- May see cord-like intermeniscal ligament

Imaging Recommendations

- Best imaging tool
  - MR modality of choice
  - MR sagittal images suggestive with confirmation on coronal data sets

DIFFERENTIAL DIAGNOSIS

Vacuum Phenomenon

- Typically in hyperextended knee
- ↓ signal intensity in joint between weight-bearing surfaces
- Rarely homogeneous like discoid meniscus

Flipped Meniscus

- Abnormal morphology to "donor site" of torn meniscus
  - Portions of meniscus missing, allowing distinction from discoid meniscus
- Displaced meniscal fragment may be found in various locations
  - Double anterior horn sign
    - Usually posterior horn flipped anteriorly
- < 2 "bow ties" on consecutive sagittal images (4- to 5-mm slices)

Bucket-Handle Tear

- Longitudinal vertical peripheral tear with displacement of meniscal fragment into intercondylar notch
  - Displaced fragment remains continuous with nondisplaced torn meniscus at anterior & posterior horns
- Double posterior cruciate ligament (PCL) sign
  - Displaced meniscal fragment anterior to PCL creating appearance of 2 PCLs
- Foreshortened, truncated, & abnormal-sized meniscus on sagittal images
- < 2 "bow ties" on consecutive sagittal images (4- to 5-mm slices)

Osteochondritis Dissecans/Osteochondral Lesion

- Focal ovoid irregularity/defect of subchondral femoral condyle
  - ± fissuring with fluid undercutting lesion, cystic change, surrounding marrow edema
  - ± detached osteochondral fragment in joint space
- Most common at lateral aspect of medial femoral condyle
- May be due to acute trauma or repetitive microtrauma

PATHOLOGY

General Features

- Etiology
  - Failure of fetal discoid meniscal form to involute
    - Derived from mesenchyme that is initially disc-shaped then forms semilunar shape

Staging, Grading, & Classification

- Watanabe classification
  - Complete discoid meniscus (type I)
    - Stable
    - Extending into intercondylar notch on coronal images (fills entire lateral compartment)
  - Incomplete (type II)
    - Stable
    - Covers ≤ 80% of tibial plateau on coronal image
    - Does not extend into intercondylar notch on coronal images
  - Wrisberg ligament-type (type III)
    - Unstable
    - Least common
    - Lacks posterior meniscal attachments
    - Hypermobile, can extend into intercondylar notch with knee extension

Gross Pathologic & Surgical Features

- Pancake-like or large, otherwise normal-appearing meniscus in lateral > > medial compartment
- Wrisberg ligament-type discoid meniscus lacks posterior meniscotibial attachment
- More prone to tears
  - Due to meniscal thickness & mechanical relationships
  - Most common: Horizontal cleavage tear
- Peripheral rim instability patterns
  - 28% of discoid lateral meniscus patients at arthroscopy → requires repair
  - Most commonly involves anterior 1/3 of meniscus

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Often asymptomatic in children (most common), especially < 10 years of age
  - Snapping knee syndrome may be seen < 10 years
    - Snap in flexion & extension due to displacing meniscus
      - Wrisberg type
    - Virtually pathognomic for discoid lateral meniscus
  - Symptomatic in older children from tears or unstable variant
    - Patients often present with pain, clicking, & snapping
    - Locking common in children
    - "Giving way"
    - Lateral joint line tenderness
    - Effusion
- Other signs/symptoms
  - McMurray & Apley grinding test may have pain & audible snap or click

Demographics

- Age
  - Uncommonly symptomatic < 10 years old
    - If symptomatic < 10 years old, snapping knee most common
  - Adults usually symptomatic
    - Peak incidence for injury
      - Female: 2nd decade
      - Male: 4th decade
- Gender
  - M > F
- Ethnicity
  - Higher incidence in Asians
- Epidemiology
  - 5-17%
    - Japan: 15%

Natural History & Prognosis

- When discoid meniscus present, more prone to be torn

Treatment

- Asymptomatic: Debatable, but most observe without surgical treatment
- Symptomatic: Arthroscopic partial central meniscectomy (i.e., saucerization)
  - Stabilization to capsule if unstable
  - Repair tear if present
- Attempt to leave peripheral rim width of 5-8 mm
- Case reports of meniscal regrowth after repair

DIAGNOSTIC CHECKLIST

Consider

- Displaced meniscal tear may cause same symptoms

Image Interpretation Pearls

- Sagittal images: ≥ 3 consecutive images show "bow tie" configuration → think discoid meniscus
- Coronal images: Slab-like shape with meniscus not tapering centrally towards intercondylar notch
- Meniscus displacement & deformation → ↑ risk of tear
  - True tears of discoid meniscus may be difficult to diagnose on MR

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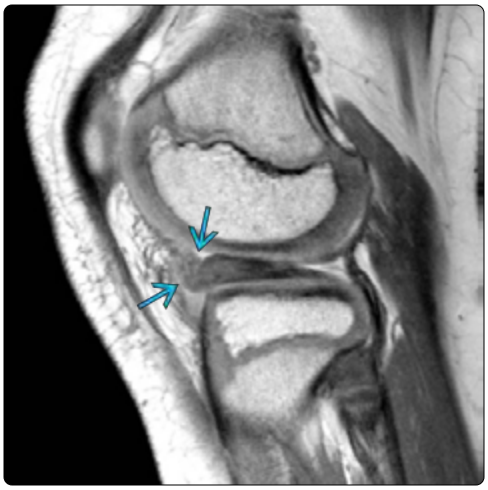
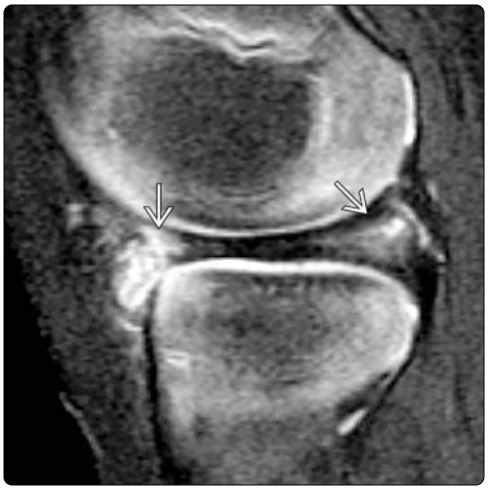
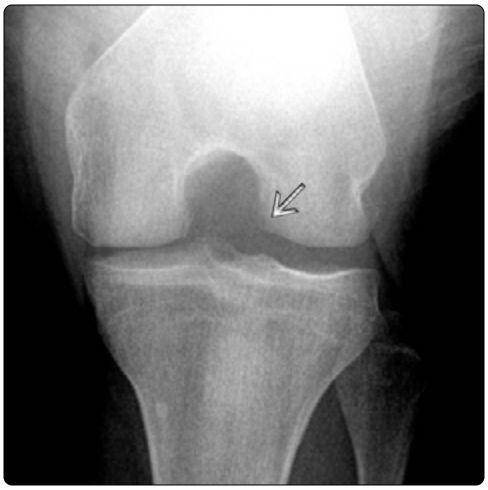
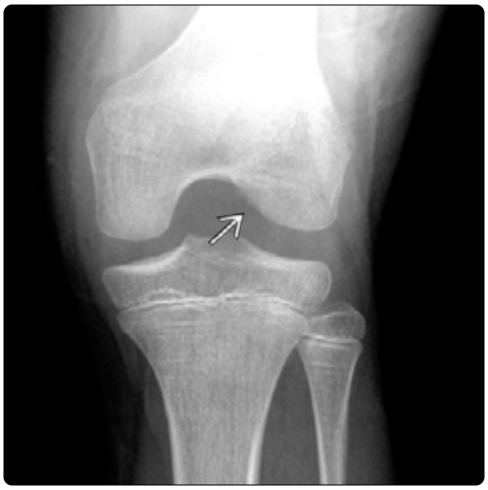


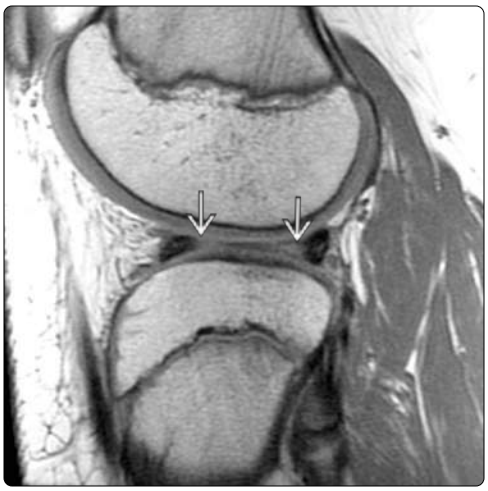


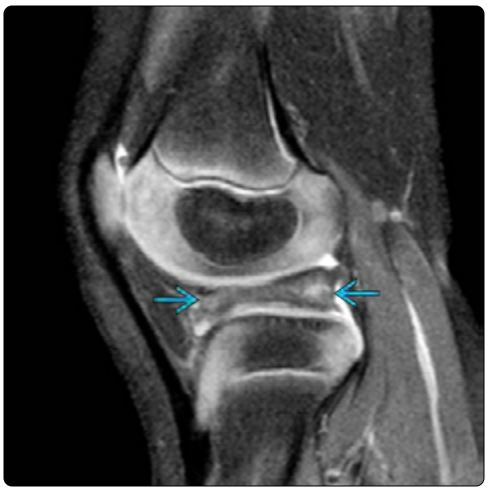
(Left) *Sagittal PD MR of the knee in a 9 year old with a Wrisberg variant of a discoid meniscus shows anterior buckling of the meniscus. At arthroscopy, no posterior capsular attachment of the discoid lateral meniscus was found.* **(Right)** *Sagittal T2 FS MR shows hyperintense signal within the anterior & posterior horns of this discoid lateral meniscus. The meniscus was found to be degenerated & torn at arthroscopy.*





(Left) *Tunnel radiograph of the knee shows a normal prominence of the medial margin of the lateral femoral condyle adjacent to the intercondylar notch in a patient without a discoid meniscus.* **(Right)** *Tunnel radiograph in a 9 year old with a discoid lateral meniscus shows decreased prominence of the lateral femoral condyle, the positive cutoff sign. This child had undergone a discoid lateral meniscus repair on the contralateral knee in the past.*





(Left) *Sagittal PD MR from a 12 year old presenting with lateral knee pain shows a complex tear of a partial discoid lateral meniscus.* **(Right)** *Sagittal T2 FS MR in a 3 year old with pain, clicking, & popping shows enlargement of a discoid lateral meniscus with internal hyperintense signal. A radial tear was found at arthroscopy.*

KEY FACTS

TERMINOLOGY

- Fracture of immature skeleton involving cartilaginous primary growth plate (physis)

IMAGING

- Most fractures detected & managed by radiographs alone
  - Widening or interruption of normally uniform undulating lucent physis
  - Translation &/or angulation of bony fragment adjacent to physis with overlying soft tissue swelling
  - Persistent physeal widening > 3 mm post reduction suggests tissue entrapment requiring open reduction
- CT: Helps evaluate comminution, displacement, articular surface step-off, loose intraarticular fragment(s)
- MR: Can detect nondisplaced fractures, assess cartilaginous & soft tissue injury or entrapment

TOP DIFFERENTIAL DIAGNOSES

- Incomplete fracture
- Chronic physeal stress injury
- Rickets
- Osteomyelitis

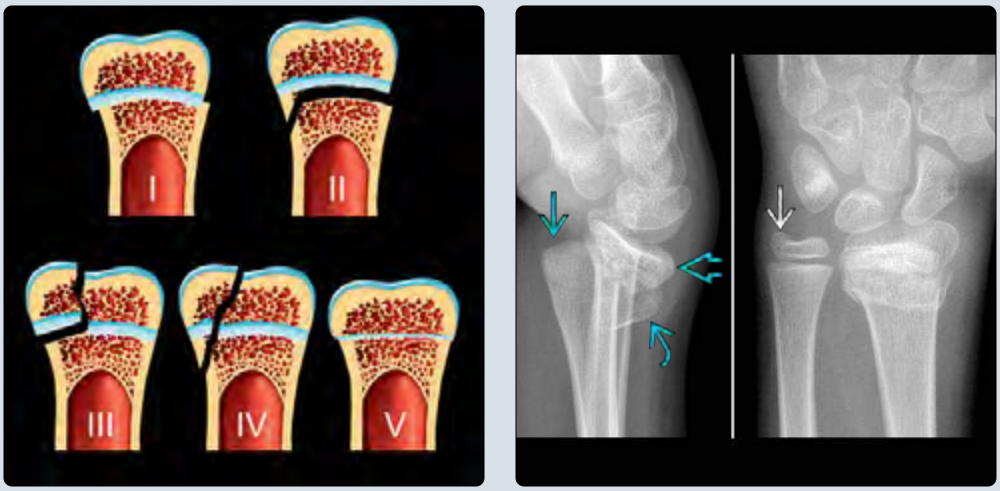
CLINICAL ISSUES

- Peak age: 11-12 years
- 6-30% of childhood fractures involve physis
- Overall complication rate: ~ 14% (but varies by site)
  - Premature physeal closure with limb shortening or angulation
    - Growth disturbance risk highest in distal femur, tibia
  - Joint incongruity due to intraarticular extension → degenerative arthritis
  - Osteomyelitis (particularly with nailbed injury)

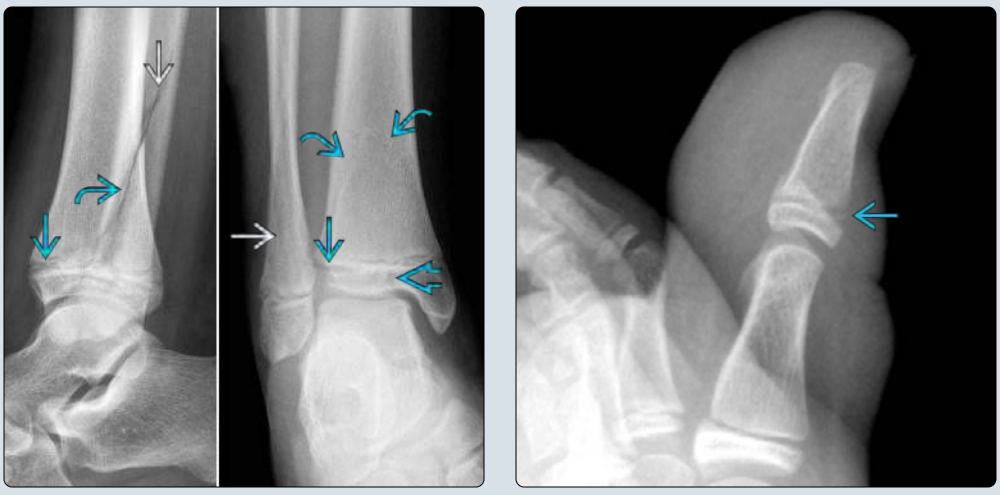
DIAGNOSTIC CHECKLIST

- Always evaluate involved growth plate for premature closure on follow-up studies

(Left) *Graphic shows the relationship of the epiphyseal, physeal, & metaphyseal components of the 5 main types of Salter-Harris (SH) fractures.* **(Right)** *Lateral (left) & PA (right) radiographs of the wrist in a 9 year old show a SH II fracture of the distal radius with ~ 60% dorsal translation of the epiphyseal ⇨ & metaphyseal ⇨ fracture fragments with ~ 45° apex volar angulation. Note the uncovering of the volar metaphysis ⇨. A nondisplaced ulnar styloid fracture ➡ is noted.*



(Left) *Lateral (left) & AP (right) radiographs of the ankle in a 15 year old show a classic SH IV triplane fracture with sagittal epiphyseal ⇨, horizontal physeal ⇨, & coronal metaphyseal & diaphyseal ⇨ components. A fibular fracture ➡ is also present.* **(Right)** *Lateral radiograph in a 10 year old after a toe stubbing injury shows a subtle, nondisplaced SH II fracture ⇨ of the great toe distal phalanx. If there were a nailbed injury, the patient should be given prophylactic antibiotics to prevent osteomyelitis.*



TERMINOLOGY

Synonyms

- Salter-Harris (SH) fractures 1-5 (I-V)

Definitions

- Fracture of immature skeleton involving cartilaginous primary growth plate (physis)

IMAGING

General Features

- Best diagnostic clue
  - Widening or interruption of normally uniform undulating lucent physis
  - Translation/angulation of bony fragment adjacent to physis with overlying soft tissue swelling
- Location
  - Upper extremity
    - Distal radius: 28-50%
    - Phalanges: 26%
    - Distal humerus: 7-17%
    - Proximal radius: 5%
    - Distal ulna: 5%
    - Metacarpals: 4%
    - Proximal humerus: 2%
  - Lower extremity
    - Distal tibia: 9-11%
    - Phalanges: 7%
    - Distal fibula: 3%
    - Metatarsals: 1%
    - Proximal tibia: 1%
    - Distal femur: 1-5%

Radiographic Findings

- Upon presentation
  - Partial or complete physeal widening with metaphyseal &/or epiphyseal fracture lines
  - Varying degrees of translation &/or angulation of distal fragments
  - Overlying soft tissue edema
- Immediately after reduction
  - Persistent physeal widening > 3 mm suggests entrapment requiring open reduction
    - Overlying periosteum most common
    - Adjacent ligament or tendon
    - Bony fragment from comminution
- Long term follow-up
  - Evaluate affected physis for premature closure

CT Findings

- Bone CT
  - Helps evaluate comminution, displacement, articular surface step-off, loose intraarticular fragment(s)
    - ≥ 2 mm articular surface step-off generally requires open reduction + internal fixation (though some advocate at less)

MR Findings

- T1WI
  - Low signal intensity fracture line
- T2WI FS
  - High signal intensity fracture line due to fluid
  - Surrounding marrow & soft tissue edema
  - Elevation of loose metadiaphyseal periosteum by subperiosteal hemorrhage
  - Low signal intensity periosteum, ligament, or tendon rarely entrapped in fracture
- STIR
  - Similar to T2WI FS (both fluid-sensitive sequences)
- T2* GRE
  - Cartilage-sensitive sequence
  - Most useful in chronic setting to detect growth-arrest (physeal bar/bridge) due to prior injury
- MR may alter clinical management in acute setting by
  - Detecting nondisplaced fractures
  - Visualizing fracture relationship to radiolucent cartilage (changing fracture classification)
  - Visualizing entrapped structures (requiring open reduction)
  - Visualizing adjacent neurovascular injury
- MR can alter clinical management of long-term complications by
  - Detecting & quantifying bony bridges
  - Detecting degenerative changes

Ultrasonographic Findings

- Useful in neonates/infants/toddlers with limited ossification of epiphyses
- May visualize displacement of cartilaginous epiphyses, physeal &/or cortical interruption, subperiosteal fluid
- Sonographic comparison of contralateral side helpful

Imaging Recommendations

- Best imaging tool
  - Typically detected & managed by radiographs alone
  - CT & MR have limited indications
- Protocol advice
  - Opposite-side comparison radiographs may help (particularly for SH I)
  - Fluid-sensitive sequences critical if MR requested

DIFFERENTIAL DIAGNOSIS

Incomplete Fracture

- Cortical buckle deformity of metadiaphysis shows no fracture line extension to physis

Chronic Physeal Stress Injury

- Partially or completely widened physis mimics nondisplaced SH I fracture
  - Interruption of endochondral ossification results in persistence of growth cartilage without fracture
- Metaphyseal irregularity & sclerosis
- Chronic pain in adolescent high-level athletes

Epiphysiodesis

- Extremity may show physeal widening & irregularity after drilling
  - Sometimes performed to halt growth in setting of growth arrest of contralateral long bone from remote insult (therefore preventing limb length discrepancy)

Rickets

- Variety of metabolic abnormalities can inhibit endochondral ossification with physeal widening
- Multiple physes symmetrically involved
- Classic rickets has metaphyseal fraying, cupping, widening, & loss of zone of provisional calcification

Osteomyelitis

- Commonly affects metaphysis in children
- Soft tissue abnormalities precede bony radiographic findings
  - Permeative lytic bony destruction &/or periosteal reaction after 10 days
  - May cause physeal widening
- Pathologic fracture may result

PATHOLOGY

General Features

- Etiology
  - Structure of normal physis
    - Germinal zone (closest to epiphysis): Small active chondrocytes emerge from resting chondrocytes
    - Proliferative zone: Flattened chondrocytes arranged in columns
    - Hypertrophic zone: Swollen chondrocytes arranged in columns
    - Zone of provisional calcification (closest to metaphysis): Chondrocytes die, cartilage matrix calcifies, osteoblasts form osteoid
- Associated abnormalities
  - ± injuries of neurovascular bundle, ligaments, cartilage, other bones
    - Depends on mechanism, fracture location, fracture type, & degree of displacement

Staging, Grading, & Classification

- Type I (~ 8.5%): Involves only physis
- Type II (~ 73%): Involves physis & metaphysis
- Type III (~ 6.5%): Involves physis & epiphysis
- Type IV (~ 12%): Involves physis, metaphysis, & epiphysis
- Type V (< 1%): Crush fracture involving physis
  - Usually recognized when cone epiphyses or partial physeal arrest becomes apparent later
- Types VI-IX, as described by Ogden 1981 (rare)
  - Infrequently used classification

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, swelling, point tenderness, limited range of motion, inability to bear weight

Demographics

- Age
  - Peak: 11-12 years
- Gender
  - Skeletal maturity earlier in girls than in boys
    - Mean age of physeal injuries 1-2 years earlier in girls
- Epidemiology
  - 6-30% of childhood fractures involve physis

Natural History & Prognosis

- Overall complication rate ~ 15%
  - Premature physeal closure with limb shortening or angulation
    - Much more common in lower than upper extremities (regardless of SH type)
      - 40-90% of distal femur, 25% of proximal tibia, 5-50% of distal tibia physeal fractures
      - 5-7% of distal radial physeal fractures
    - Twice as likely in displaced vs. nondisplaced fractures
  - Joint incongruity due to articular surface involvement → degenerative arthritis
    - Up to 29% of SH III or IV ankle injuries
  - Osteomyelitis with adjacent nailbed trauma ("stubbed toe osteomyelitis") or penetrating injury

Treatment

- Closed reduction & casting for low SH categories (unless unstable post-reduction)
  - < 2 mm displacement → ↓ risk of growth arrest
  - Follow-up at 5-7 days to ensure stability
- Open reduction & internal fixation often required with higher categories (due to articular surface involvement)
- Subsequent bony bridges resulting in angular deformities or limb-length discrepancies → bone bridge resection vs. contralateral epiphysiodesis

DIAGNOSTIC CHECKLIST

Consider

- Always evaluate involved growth plate for premature closure on follow-up studies
  - Follow knee & ankle fractures for ≥ 1 year or until skeletal maturity due to risk of growth disturbances

Reporting Tips

- Include type, direction, & magnitude of displacement
  - Translation (including shortening & distraction), angulation, & rotation
- Presence of physeal &/or articular extension

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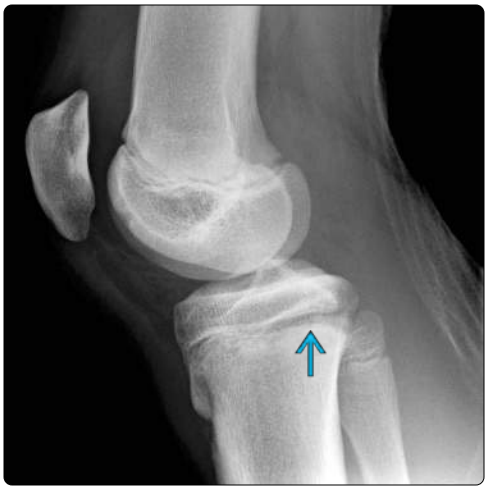
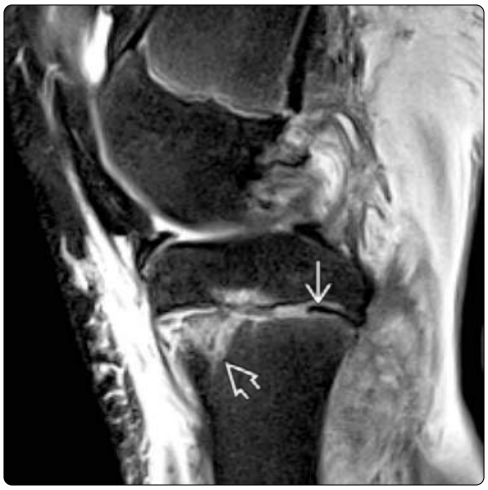
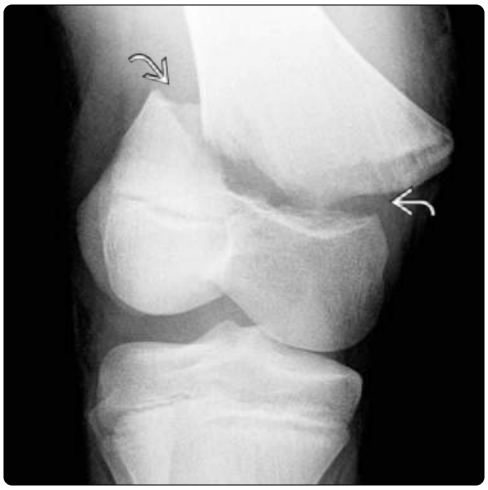
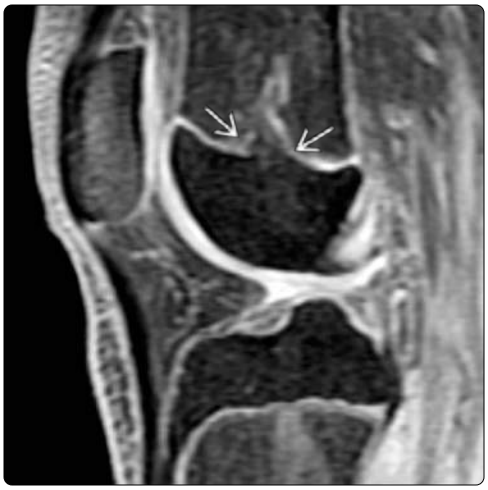


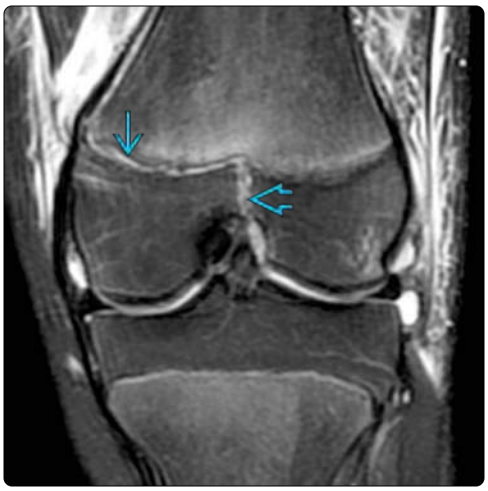


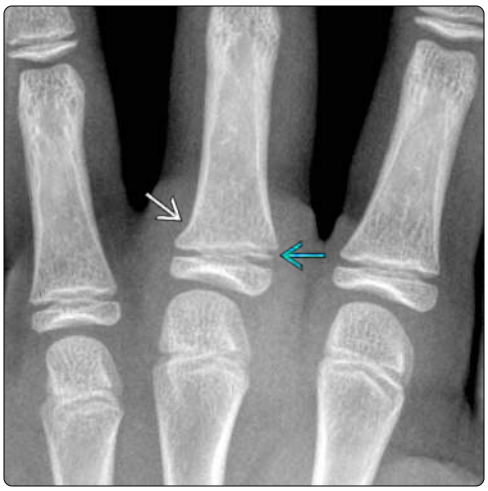
(Left) *Lateral radiograph in a 15 year old with pain from a gymnastics injury shows widening of the posterior physis of the proximal tibia.* **(Right)** *Sagittal T2 FS MR in the same patient shows low signal intensity periosteum trapped in the widened physis. SH II metaphyseal fracture components are present anteriorly. Subsequent growth arrest is more likely when periosteal entrapment occurs.*





(Left) *AP radiograph of the distal femur in a 12 year old shows a displaced SH II fracture.* **(Right)** *Sagittal 3D GRE MR of the same patient 20 months later shows focal, central interruption of the distal femoral growth plate by a bony bridge. MIP reconstructions parallel to the growth plate will allow calculation of the bone bridge size relative to the intact physis for operative planning.*





(Left) *Coronal T2 FS MR in a 12 year old shows a SH III fracture of the distal femur with vertical intraarticular epiphyseal & horizontal medial physeal components. As in this case, these fractures can be radiographically occult.* **(Right)** *PA radiograph of the hand in a 10 year old shows slightly increased angulation of the ulnar-sided cortex of the middle finger proximal phalanx. A metaphyseal fragment confirms a SH II fracture.*

Apophyseal Injuries

KEY FACTS

TERMINOLOGY

- Apophysis: Nonarticular secondary center of ossification that serves as attachment site for muscle or tendon
  - Considered an epiphyseal equivalent
- Acute injury: Avulsion fracture of osseous &/or cartilaginous apophysis through subjacent physis
- Chronic injury: Repetitive submaximal tensile forces (avulsive microtrauma) exceed rate of repair, leading to local growth plate disturbance (± symptoms)

IMAGING

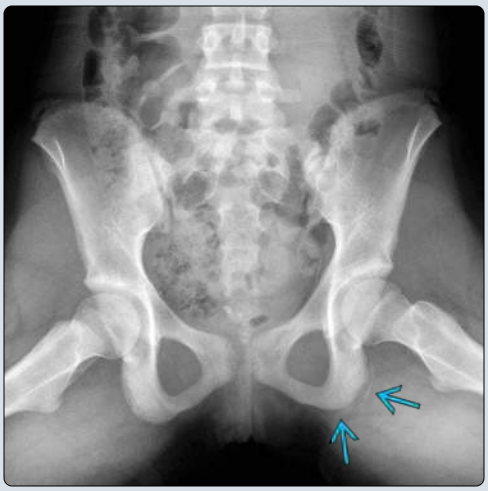
- Acute injury: Displaced apophyseal ossification center
- Chronic injury: Soft tissue swelling with ossific irregularity at tendon attachment site
- Radiographs usually diagnostic of acute avulsion
- Further imaging may be required if fragment nondisplaced, ossification center not yet present, or chronic apophysitis suspected
  - MR more sensitive & specific than US

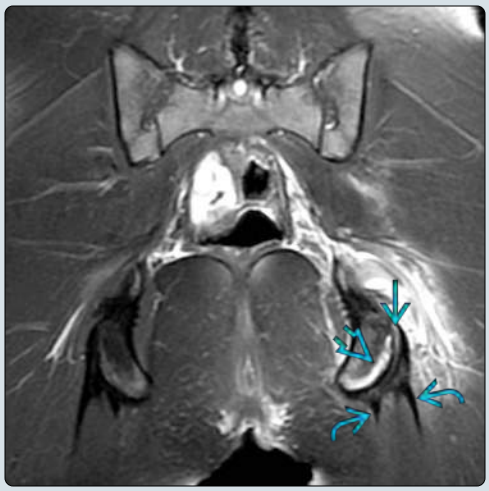
TOP DIFFERENTIAL DIAGNOSES

- Osteomyelitis
- Osteosarcoma
- Muscle injury
- Stress injury of bone

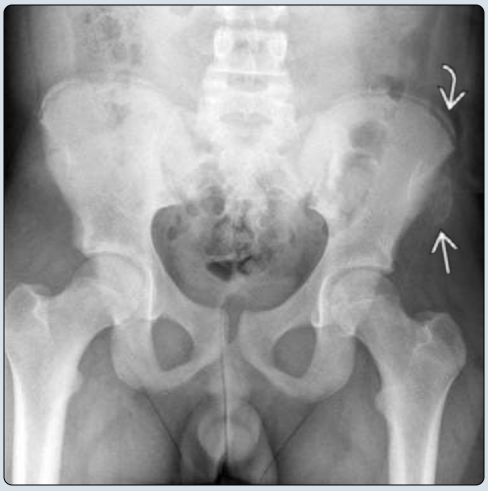
CLINICAL ISSUES

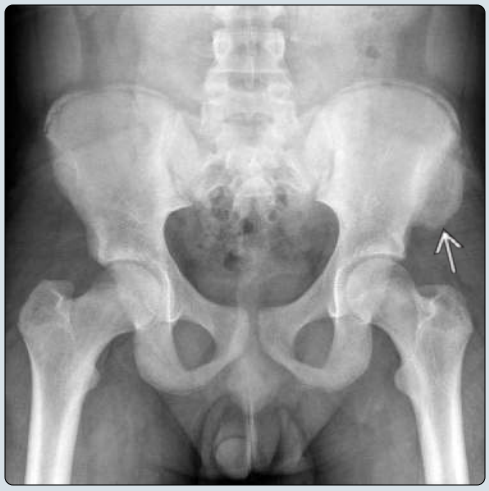
- Acute injury: Sudden onset of pain with sensation of "pop" & instant ↓ of muscle function during athletic activity
- Chronic injury: Insidious onset of pain & swelling without specific event or associated bruising
- Majority of acute pelvic avulsions occur from ages 12-18 years; mean of 13.8-15.2 years
  - Most occur during kicking or sprinting
    - Soccer, gymnastics, rugby, track & field
  - AIIS, ASIS, ischial tuberosity > iliac crest, pubic symphysis
- Conservative (nonsurgical) therapy: Highly successful
  - Complications (nonunion, chronic pain) uncommon
- Surgical fixation reserved for displacement > 1.5-2.0 cm





(Left) *Frog leg lateral radiograph in a 13-year-old girl with acute onset of left hip pain & the sensation of a "pop" during cheerleading shows displaced bony fragments adjacent to the left ischial tuberosity.* **(Right)** *Coronal T2 FS MR in the same patient shows the attachments of the hamstring tendons to the displaced curvilinear osteocartilaginous apophysis at the ischial tuberosity. Fluid undercuts the displaced fragment, typical of an acute avulsion injury.*





(Left) *AP radiograph in a 16-year-old boy with a history of pain & a "pop" while kicking a ball shows an inferiorly displaced bony fragment that was avulsed from the anterior superior iliac spine (ASIS). The lateral left iliac wing apophysis also shows fragmentation & growth plate widening, suggesting an additional avulsion component.* **(Right)** *AP radiograph in the same patient 3 months later shows progressive healing at the site of injury with callus now seen between the fragment & the underlying ilium.*

TERMINOLOGY

Synonyms

- Acute injury: Avulsion fracture
- Chronic injury: Apophysitis, physeal stress fracture, osteochondrosis, various eponyms specific to site

Definitions

- Apophysis: Nonarticular secondary center of ossification that serves as attachment site for muscle or tendon
  - Contributes to bony shape but not length
  - Considered an epiphyseal equivalent
- Acute injury: Avulsion fracture of osseous &/or cartilaginous apophysis through subjacent physis
- Chronic injury: Repetitive submaximal tensile forces (avulsive microtrauma) exceed rate of repair, leading to local growth plate disturbance (± symptoms)

IMAGING

General Features

- Best diagnostic clue
  - Acute injury: Displaced apophyseal ossification center
  - Chronic injury: Soft tissue swelling with ossific irregularity at tendon attachment site
- Location
  - Acute avulsion injury
    - Pelvis/hips: High concentration of muscle attachments
      - Ischial tuberosity (hamstrings: Biceps femoris, semimembranosus, semitendinosus)
      - Anterior inferior iliac spine (AIIS) (rectus femoris)
      - Anterior superior iliac spine (ASIS) (sartorius)
      - Iliac crest (tensor fascia lata, abdominal wall muscles)
      - Pubic symphysis (adductors)
      - Greater trochanter (gluteus medius & minimus)
      - Lesser trochanter (iliopsoas)
    - Tibial tubercle (patellar tendon)
    - Medial epicondyle of humerus (flexor group & ulnar collateral ligament)
  - Chronic stress injury
    - Tibial tuberosity: Osgood-Schlatter disease
    - Humeral medial epicondyle: Little Leaguer elbow
    - Iliac crest
    - Calcaneal apophysis: Sever disease

Radiographic Findings

- Radiography
  - Acute avulsion injury
    - Displacement of apophyseal ossification center
      - Tangential view: Crescentic, triangular, or irregular bone fragment
      - En face view: Poorly defined or ovoid fragment
    - Moderate to marked soft tissue edema
      - May be only sign if ossification has not yet developed in apophysis
      - May be occult at pelvis due to overlapping structures
  - Healed/remote acute avulsion fracture
    - Bony remodeling from prior injury
    - Heterotopic ossification of soft tissues may develop at site of prior injury
  - Chronic stress injury
    - Ossific irregularity or fragmentation at site of tendon attachment
    - Underlying physeal widening without true fragment displacement
    - Mild overlying soft tissue swelling

MR Findings

- T1WI
  - Dark signal intensity of marrow & soft tissue edema
  - ± bright fatty marrow in avulsed bone
- T2WI FS
  - Long TE decreases conspicuity of cartilage, especially unossified "epiphyseal" cartilage (appears dark)
  - Acute injury
    - Bright fluid deep to displaced apophysis
    - Dark tendon remains attached to osteocartilaginous fragment
    - Bright marrow edema of apophysis & adjacent metaphyseal equivalent donor site
    - Moderate/marked surrounding soft tissue edema
      - Fluid &/or hemorrhage at avulsion site
      - Muscle edema from strain ± laxity
  - Chronic injury
    - Mild edema of nondisplaced apophysis & metaphysis
    - Mild ↑ signal, widening, & irregularity of physis without discrete fluid
- PD/intermediate FS
  - Bright cartilage fragment
    - Short TE improves cartilage visualization
- STIR
  - Findings similar to T2WI FS
- GRE FS
  - Bright cartilage fragment, dark bone fragment

Ultrasonographic Findings

- Grayscale ultrasound
  - Distortion of normal soft tissue planes & osteocartilaginous interfaces
    - Comparison to contralateral normal side is key
  - Acute injury: Displaced osteocartilaginous fragment attached to muscle/tendon
    - Hypoechoic cartilage surrounding hyperechoic, shadowing ossification center
    - Dynamic maneuvers can move fragment
  - Chronic: Soft tissue edema, tendon thickening, bony fragmentation

Imaging Recommendations

- Best imaging tool
  - Radiographs usually diagnostic of acute avulsion
  - MR useful in some settings
    - Acute injury: Nondisplaced fragment or purely cartilaginous fragment (prior to ossification)
    - Chronic apophysitis
- Protocol advice
  - FS T2 or STIR MR: Best for marrow & soft tissue edema
  - FS PD or FS GRE MR: Best for cartilaginous fragments

DIFFERENTIAL DIAGNOSIS

Osteomyelitis

- Pelvic bones subjacent to growth cartilage (i.e., metaphyseal equivalents) frequently involved
- Marrow & soft tissue edema ± fluid collections

Osteosarcoma

- Most commonly originates in long bone metaphysis
- Bony destructive features intermixed with cloud-like intramedullary & soft tissue osteoid

Muscle Injury

- Musculotendinous junction becomes weak link of musculoskeletal system after physeal closure
- Edema, fluid, interruption of muscle fibers ± tendon; muscle retraction if tear complete

Stress Injury of Bone

- Solid periosteal reaction ± linear sclerosis or lucency (often perpendicular to bone long axis)
- Dark T1 MR signal fracture line precedes radiographic findings
- Surrounding marrow, soft tissue edema on FS T2 MR

PATHOLOGY

General Features

- Etiology
  - Osteochondral junction: Weakest point of immature musculoskeletal system
  - ↑ muscle strength of adolescence causes ↑ forces on relatively weak sites of muscle/tendon attachments
  - Acute apophyseal avulsions: Essentially Salter-Harris growth plate fractures
  - Chronic injury (apophysitis): Repetitive submaximal tensile stresses exceed rate of bone repair
    - Physeal widening due to impaired endochondral ossification or chondrocyte hypertrophy
    - Chronic stress injury may weaken physis & predispose to acute avulsion

Staging, Grading, & Classification

- Pelvic avulsions classified by McKinney, Nelson, & Carrion
  - Type I: Not displaced
  - Type II: < 2-cm displacement
  - Type III: > 2-cm displacement
  - Type IV: Symptomatic nonunion or exostosis
  - Majority of cases displaced < 2 cm (type I or II)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute: Sudden onset of pain with sensation of "pop" & instant ↓ of muscle function during athletic activity
  - Chronic: Insidious onset of pain & swelling without specific event or associated bruising

Demographics

- Age
  - Acute pelvic avulsions: Majority occur from ages 12-18 years; mean of 13.8-15.2 years
    - Ossification centers about pelvis appear around ages 12-14 years, fuse by 25 years
    - Sites of radiographically diagnosed injuries correlate with timing of ossification center appearance
  - Medial epicondyle avulsions: 9-14 years
  - Tibial tubercle avulsions: 13-16 years
- Gender
  - M:F = 3:1
- Epidemiology
  - Acute
    - Most pelvic avulsions occur with kicking or sprinting
      - Soccer, gymnastics, rugby, track & field
      - AIIS, ASIS, ischial tuberosity > iliac crest, pubic symphysis
      - Multiple fractures in 6%
    - Tibial tubercle avulsions common in basketball
    - Medial epicondyle avulsions common with throwing or dislocation
  - Chronic
    - Little leaguer: Valgus stress of overhead throwing
    - Osgood-Schlatter: Repetitive traction of jumping
    - Sever disease: Traction by Achilles tendon

Natural History & Prognosis

- Complications uncommon
  - Nonunion: Much more likely in type III vs. type II
  - Chronic pain
    - Most common with AIIS avulsions
      - Concomitant or subsequent labral injury
      - Exuberant heterotopic ossification
    - Sciatic nerve irritation from displaced ischial tuberosity avulsions
  - Not at risk for clinically significant growth arrest (unlike long-bone physeal injuries)

Treatment

- Most therapy conservative (nonsurgical) & highly successful
  - Initial rest followed by physical therapy
- Surgical fixation reserved for higher grade injuries
  - More likely if displacement > 1.5-2.0 cm

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Check pelvic apophyses closely in teenager with acute hip pain & pop during activity
- Radiographs may be negative initially in younger patients who have not yet ossified their apophyses

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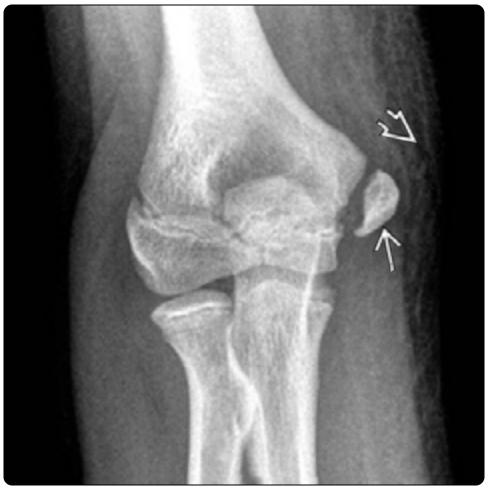
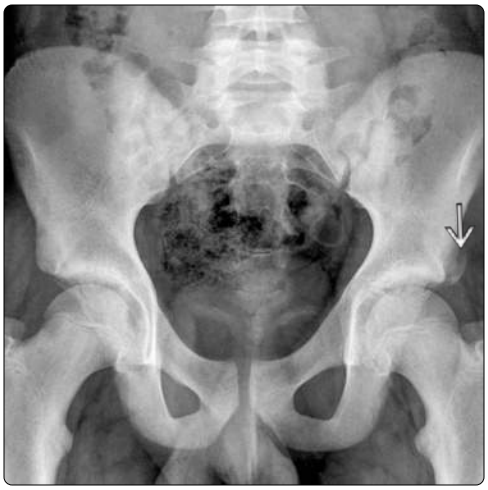
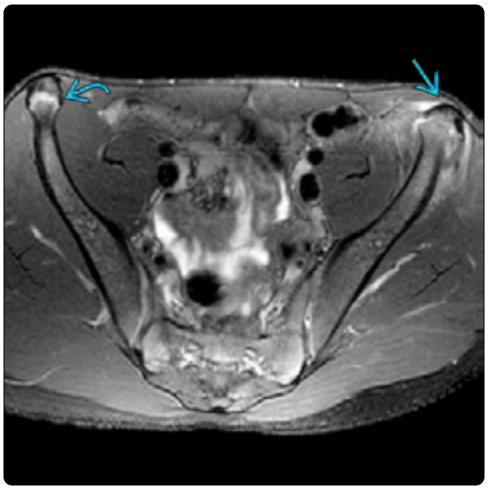
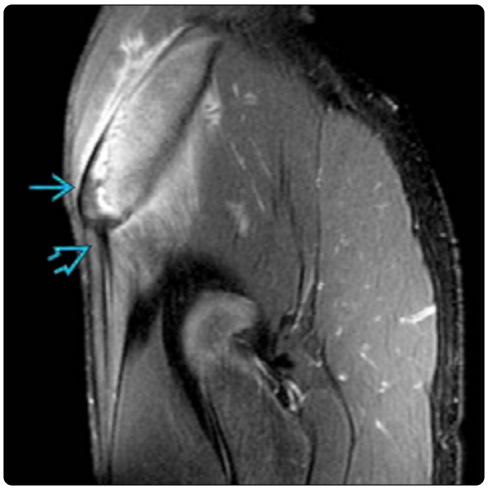


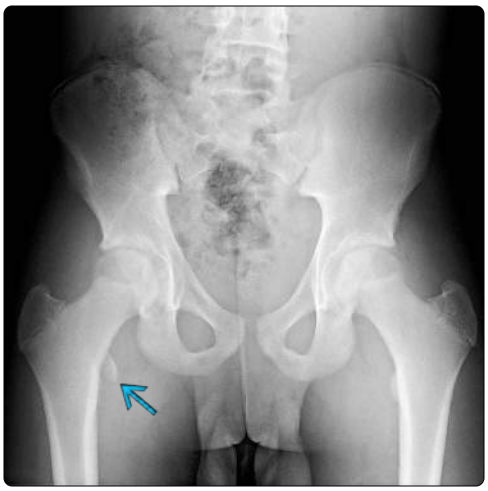


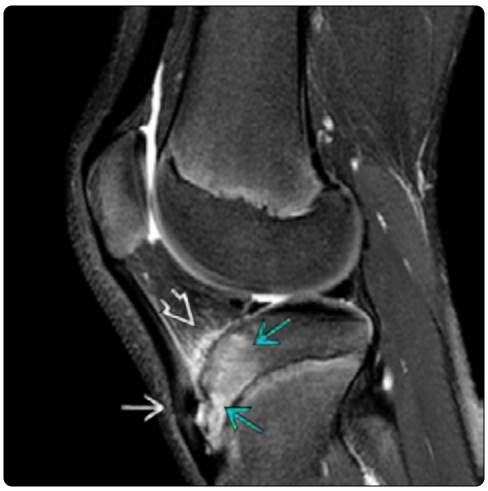
(Left) *AP radiograph of the elbow in a 13-year-old boy shows avulsion of the medial epicondyle ossification center with overlying soft tissue edema.* **(Right)** *AP radiograph of a 12 year old with hip pain shows mild lateral displacement of an avulsed anterior inferior iliac spine (AIIS) ossification center.*





(Left) *Axial T2 FS MR in a 14-year-old boy who felt a "pop" while running shows a displaced apophyseal fragment due to sartorius avulsion of the left ASIS. The right ASIS shows chronic physeal stress injury with growth plate widening.* **(Right)** *Sagittal T2 FS MR in the same patient shows cartilaginous apophyseal avulsion at the sartorius attachment to the left ASIS. This fracture was radiographically occult.*





(Left) *AP radiograph in a 14 year old who felt a "pop" while swimming shows avulsion of the unfused right lesser trochanter. This fracture does not implicate underlying pathology in children.* **(Right)** *Sagittal T2 FS MR of a 13 year old with Osgood-Schlatter disease shows tibial tubercle ossification center edema extending proximally into the tibial plateau. Mild fluid signal intensity is noted in & overlying the thickened patellar tendon. There is also edema inferiorly in the Hoffa fat pad.*

Incomplete Fractures

KEY FACTS

TERMINOLOGY

- Incomplete fracture: Macroscopic fracture line does not traverse entire bony diameter
  - Pediatric bones more elastic than adult bones
  - Greater propensity to bow or bend before breaking
- Buckle fracture: Focal outward bulge of cortex (without frank interruption) on compression side; cortex usually intact on tension side
- Plastic deformation: Smooth but accentuated bending of shaft without visible fracture line
- Greenstick fracture: Discrete fracture line on tension side does not extend through opposite cortex

IMAGING

- 2 tangential views show at least 1 unbroken cortex
- Occurs in diaphysis or metadiaphysis
- Typically diagnosed & managed by radiographs alone
- Contralateral comparison views may be helpful
  - Especially in plastic deformation

TOP DIFFERENTIAL DIAGNOSES

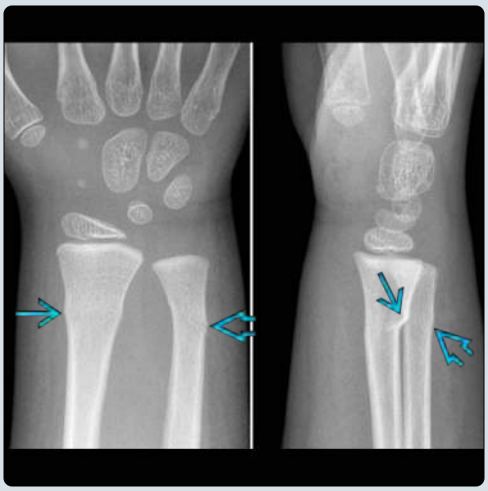
- Bowing due to underlying skeletal disease
  - Systemic or localized bony dysplasias
  - Metabolic bone diseases
- Normal developmental variants
- Salter-Harris type II fracture

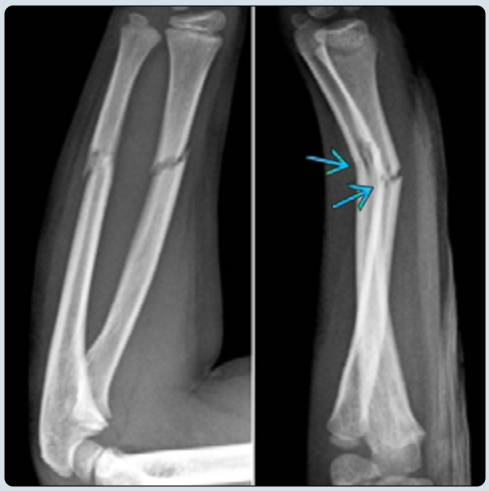
CLINICAL ISSUES

- Pain, swelling, tenderness, disuse of limb after fall
- Refracture in 7-20% of greenstick fractures

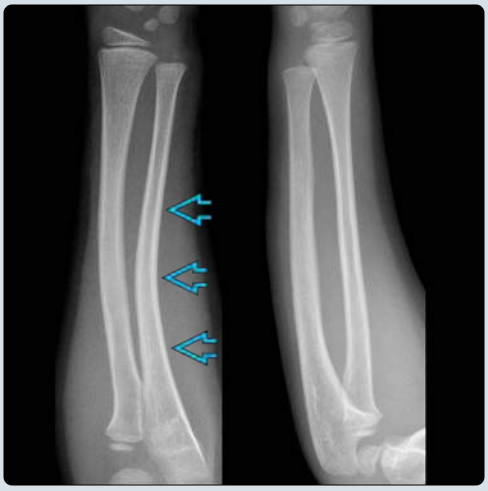
DIAGNOSTIC CHECKLIST

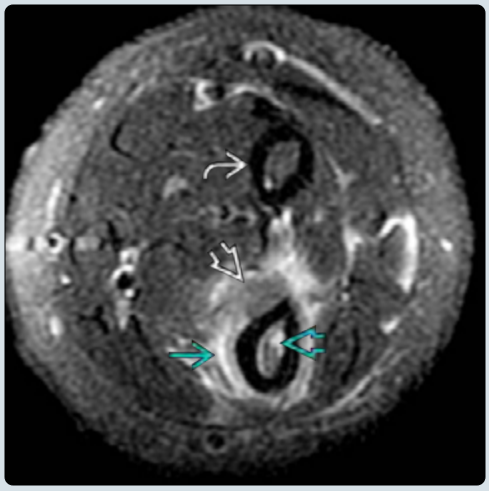
- Imaginary marble should smoothly roll down diaphyseal & metaphyseal cortex on radiograph
  - If it dips or bounces, strongly consider incomplete fracture (especially with overlying swelling)
- Look carefully for metaphyseal fracture line extending to physis (implying Salter-Harris type II fracture)
  - Complications & follow-up different from buckle





(Left) *PA & lateral radiographs of the wrist in a 5 year old after a fall show a buckle deformity of the distal radial metadiaphyseal junction as well as an incomplete fracture (with cortical interruption) of the distal ulna.* **(Right)** *AP & lateral radiographs of the forearm in a 5 year old after a fall show incomplete fractures of the distal radial & ulnar diaphyses. Note the intact "bent but not broken" posterior cortex of each bone, typical of greenstick fractures.*





(Left) *AP radiograph of the forearm in a 5 year old after a fall shows a plastic (bowing) deformity of the ulnar diaphysis. The radial curvature is within normal limits. The ulnar deformity is not readily visible on the lateral view.* **(Right)** *Axial T2 FS MR in the same patient 4 days later (performed for specific elbow complaints) shows marrow, periosteal, & soft tissue edema in/about the ulnar diaphysis with no cortical break. Subperiosteal hemorrhage is noted. The radius is normal.*

TERMINOLOGY

Definitions

- Incomplete fracture: Macroscopic fracture line does not traverse entire bony diameter
  - Pediatric bones more elastic than adult bones
    - Greater propensity to bow or bend before breaking
- Greenstick fracture: Discrete fracture line on tension side does not extend through opposite cortex
  - Appearance of bent immature tree branch ("green stick")
- Plastic deformation: Smooth but accentuated bending of shaft without visible fracture line
  - "Bowing fracture"
- Buckle fracture: Focal outward bulge of cortex (without frank interruption) on compression side; cortex usually intact on tension side
  - "Torus" term no longer in favor (as it implies circumferential bulging)
- Incomplete fracture with cortical disruption: Frank cortical disruption/angulation on compression side

IMAGING

General Features

- Best diagnostic clue
  - Cortical bump or angulation at site of pain + overlying soft tissue swelling after fall
- Location
  - Buckle fractures: Most often at distal diaphysis or metadiaphysis of radius & ulna or proximal tibia
  - Plastic deformation: Most common in diaphysis of ulna, fibula, or radius
  - Greenstick fractures: Most common in forearm diaphyses

Radiographic Findings

- Buckle fracture
  - Focal protuberance of cortex on compression side
- Plastic deformation
  - Subtle increased curvature of long bone diaphysis
  - Adjacent bone of forearm or lower leg often fractured
    - May be different type or dislocation
  - Periosteal reaction may be limited during healing
- Greenstick fracture
  - Visible hairline (or larger) fracture on convex tension side of cortex

Ultrasonographic Findings

- Focal interruption/bulging of echogenic cortex ± subperiosteal hemorrhage, soft tissue edema

DIFFERENTIAL DIAGNOSIS

Bowing Due to Underlying Skeletal Disease

- Systemic or localized bony dysplasias
- Metabolic bone diseases

Normal Developmental Variants

- Few sites of normal cortical angulation/protuberance
- Comparison views of contralateral side may be helpful

Salter-Harris Type II Fracture

- Fracture line extends to physis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, swelling, tenderness, disuse of limb, or limp; typically after fall

Natural History & Prognosis

- Buckle fracture: Complete healing typical
- Plastic deformation: Does not typically remodel
- Greenstick fracture
  - Refracture in 7-20%
  - Rare median nerve entrapment or transection
  - Rare development of small subperiosteal cortical "cyst" during healing (due to entrapped medullary fat)

Treatment

- Buckle fracture
  - 3-week immobilization with splint
  - Follow-up not required in some circumstances
- Greenstick fracture
  - Depending on angulation, may require closed reduction & casting
  - Some advocate completing fracture for improved alignment
  - Prolonged immobilization may reduce refractures
- Plastic deformation
  - ± closed reduction
    - Requires significant force, adequate sedation
    - Prevents limitations of pronation-supination
    - Correction of rotational component may be required

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Imaginary marble should smoothly roll down diaphyseal & metaphyseal cortex on radiograph
  - If it dips or bounces, strongly consider incomplete fracture (especially with overlying swelling)
- Look carefully for metaphyseal fracture line extending to physis (implying Salter-Harris type II fracture)
  - Complications & follow-up different from buckle

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Child Abuse, Metaphyseal Fracture

KEY FACTS

TERMINOLOGY

- Classic metaphyseal lesion (CML) or metaphyseal corner fracture: Transverse fracture of subphyseal metaphysis that undercuts subperiosteal bone collar (SPBC) peripherally
  - Fracture of infants with high specificity for child abuse

IMAGING

- Most common at distal femur, proximal & distal tibia
- Radiographic appearance
  - Triangular fragment at metaphyseal corner when x-ray beam perpendicular to bone long axis
  - Bucket-handle fragment adjacent to metaphysis when x-ray beam angled caudal or cranial relative to physis
  - Healing fractures more conspicuous
    - Extension of physeal lucency into metaphysis
    - May see sclerosis along fracture margin
    - Subperiosteal new bone formation & callus
- Tc-99m MDP bone scan & 18F-NaF PET complementary to initial skeletal survey
  - Sensitivity greater for most fractures except: CML, skull, scapula, & remote

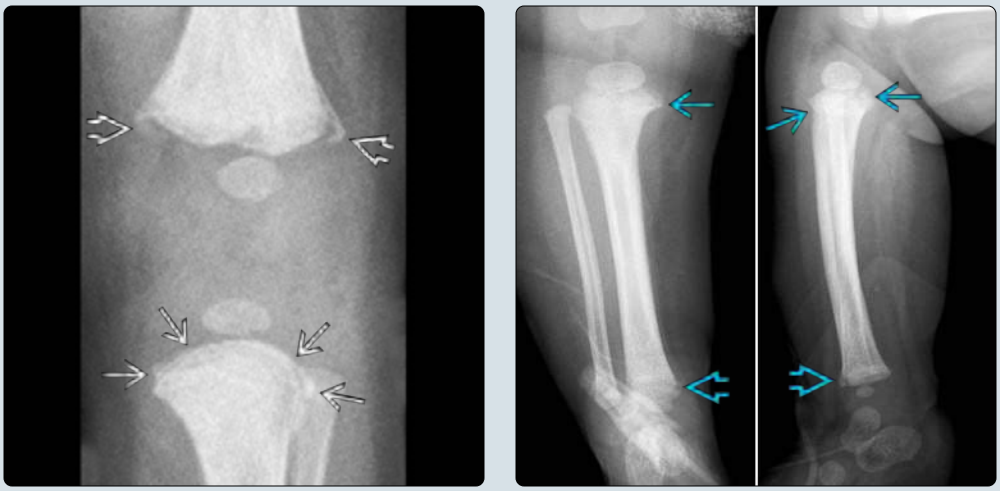
PATHOLOGY

- Planar fracture through junction of primary & secondary spongiosa at trabecular transition zone; fracture undercuts SPBC peripherally
- Due to tensile & torsional forces from twisting or pulling extremity or from acceleration/deceleration of shaking
- No established evidence that rickets or metabolic bone disease can cause CML

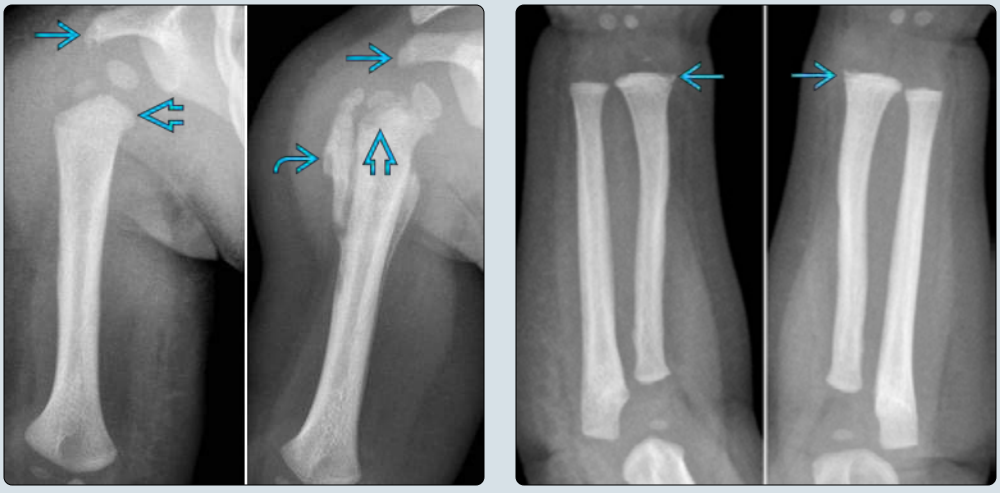
CLINICAL ISSUES

- Initial skeletal survey obtained for
  - < 2 years old with suspicion of NAT
  - < 5 years old with suspicious fracture
  - Concern for NAT in any child unable to communicate
- Obtain follow-up skeletal survey in 2 weeks: Healing increases fracture conspicuity
- 95% of cases with CML have ≥ 1 additional injury

(Left) *AP radiograph in a 1-month-old girl shows a typical metaphyseal corner fracture at the distal femoral metaphysis with a bucket-handle appearance of a proximal tibial metaphyseal fracture.* **(Right)** *AP (left) & lateral (right) radiographs of the right lower leg in a 6 month old with bruising show lucent irregularities of the subphyseal tibial metaphyses proximally & distally. A corner fragment is best appreciated anteriorly at the distal tibia on the lateral view.*



(Left) *AP radiographs of the right humerus in the same patient at the time of presentation (left) & 2 weeks later (right) show an evolving acromion fracture (which has a high specificity for NAT). The initial image shows minimal proximal humeral metaphyseal abnormality that becomes much more conspicuous on the follow-up study with abundant periosteal reaction.* **(Right)** *AP radiographs in the same patient (at presentation) show bilateral corner fractures of the distal radial metaphyses.*



TERMINOLOGY

Synonyms

- Nonaccidental trauma (NAT), battered child syndrome
- Metaphyseal corner fracture, classic metaphyseal lesion (CML), bucket-handle fracture

Definitions

- Fracture of infants that extends though subphyseal metaphysis & undercuts subperiosteal bone collar (SPBC) peripherally

IMAGING

General Features

- Best diagnostic clue
  - Triangular bone fragments at corners of metaphysis or bucket-handle rim-like fragment of subphyseal metaphysis
- Location
  - Metaphyses of long bones, most commonly: Distal femur, proximal & distal tibia, proximal humerus

Radiographic Findings

- Acute fractures subtle, often difficult to identify on initial skeletal survey
- Transverse subphyseal lucency separates thin bone fragment from metaphysis & undercuts SPBC peripherally
  - May fully or partially extend across metaphysis
- Appearance depends on radiographic projection
  - Triangular fragment at metaphyseal corner when x-ray beam perpendicular to metaphysis long axis
  - Bucket-handle fragment adjacent to metaphysis when x-ray beam angled caudal or cranial relative to physis
- Healing fractures more conspicuous
  - Focal extension of physeal lucency into metaphysis
    - Most common at site of SPBC undercutting
    - May be broad with extensive fracture
  - Callus & subperiosteal new bone formation (SPNBF)
  - May see sclerosis along fracture margin
- Most common at distal femur, proximal & distal tibia
  - More commonly seen medially at these locations
  - Lateral & anterior fractures always accompanied by medial fractures in series of cases
  - Proximal tibial fracture may extend anteriorly along tibial tubercle region, best seen in healing phase with sclerosis on lateral view
  - Distal tibial fracture vertical component undercutting SPBC typically longer than in proximal tibia
- General comments on dating fractures
  - SPNBF appears after 7-10 days
  - SPNBF thickness ↑ with time
  - Callus usually by 15 days, not before 9 days
  - Callus thickness ↓ with age

CT Findings

- Not advocated for identification of fractures
- Evaluate proximal humeri & femurs on CT obtained to evaluate for intrathoracic or intraabdominal injuries

MR Findings

- Whole-body MR shows very low sensitivity for signal abnormalities at sites of CML seen on skeletal survey: 31%
- May be complementary given ability to detect soft tissue signal abnormalities

Ultrasonographic Findings

- Grayscale ultrasound
  - Rarely used for NAT evaluation
  - Disruption of echogenic metaphyseal cortex
  - Irregularity of chondro-osseous junction
  - Displacement &/or distortion of normally well-defined echogenic perichondral ring
  - Periosteal elevation & early Ca^{2+}
  - May see subtle edema in adjacent soft tissues

Nuclear Medicine Findings

- Tc-99m methylene diphosphonate (MDP) skeletal scintigraphy or 18F-NaF PET complementary to initial skeletal survey
  - Focal ↑ in radionuclide activity with 24 hours, normalizing within 6 months
  - Overall, sensitivity of scintigraphy for detecting fractures > initial skeletal survey, with exception of CML, skull & scapular fractures, remote injuries
    - ↓ sensitivity for CML due to physiologic physeal uptake
      - Tc-99m MDP bone scan 35%, 18F-NaF PET 67%
  - Scintigraphy specificity lower than initial skeletal survey

Imaging Recommendations

- Initial skeletal survey
  - Indications
    - < 2 years old with suspicion of NAT
    - < 5 years old with suspicious fracture
    - Concern for NAT in any child unable to communicate
  - Images include: AP & lateral skull, lateral cervical & lumbar spine, AP & lateral & both obliques thorax, AP pelvis, AP humeri, AP forearms, PA hands, AP femurs, AP lower legs, AP feet
  - Additional views based on clinical & imaging findings
- Follow-up skeletal survey
  - Minimum of 10 days from initial evaluation
  - Typically 2 weeks from initial evaluation
  - Indications
    - Concerning fractures on initial study
    - Normal initial study with persistent suspicion based on clinical or imaging findings
  - Used to confirm suspected fractures & identify additional fractures
  - In 1 study, clarified questionable fractures or identified new fractures in 48% of cases
    - Of 27 new fractures, 2 were CML
    - Of 29 questionable fractures, 6 were confirmed CML
- Tc-99m MDP skeletal scintigraphy or 18F-NaF PET as complementary or problem-solving tools
- CECT for suspected intrathoracic, intraabdominal, or intracranial injury

DIFFERENTIAL DIAGNOSIS

Osteogenesis Imperfecta

- Multiple fractures ± Wormian bones, blue sclera
- ± osteoporosis

Metaphyseal Spur

- Distal extension of SPBC beyond chondroosseous junction, may be seen at lateral distal femur & other sites

Rickets

- Metaphyseal fraying, cupping, widening ± fractures
- Demineralization

Birth Trauma

- CML-like injuries very rarely reported with C-section

Leukemia

- Metaphyseal lucent bands ± fractures, more aggressive permeative lesions

Menkes Syndrome

- Osteoporosis, metaphyseal corner spurs, Wormian bones, tortuous intracranial arteries, & brittle hair from abnormal copper metabolism

Spondylometaphyseal Dysplasia

- Metaphyseal irregularities resembling corner fractures due to abnormal endochondral ossification
- Scoliosis, platyspondyly, coxa vara, pectus carinatum

Metaphyseal Chondrodysplasia

- Early metaphyseal cupping → severe expansion → fragmentation or cystic changes
- Frontonasal hyperplasia, hypertelorism, proptosis, micrognathia, curved forearms & legs

PATHOLOGY

General Features

- Planar fracture through junction of primary & secondary spongiosa at trabecular transition zone; fracture undercuts SPBC peripherally
- Due to tensile & torsional forces from twisting or pulling extremity or from acceleration/deceleration of shaking
- No established evidence that rickets/metabolic bone disease can cause CML

CLINICAL ISSUES

Presentation

- Wide range of clinical presentations for NAT
  - Injury inconsistent with history or stage of development
  - Multiple injuries in various stages of healing
  - Bruising in nonmobile infant
  - Retinal hemorrhages
- CML
  - 1 study evaluating bruising as indicator for fracture
    - 25% had bruising at sites other than fracture site
    - Only 13% had bruising at/near fracture site
  - Another study found that 95% of cases with CML had at least 1 additional injury
    - 84% had additional non-CML fractures, most commonly long bone & rib
    - 43% had cutaneous injuries (bruising or burns)
    - 28% had traumatic brain injury

Demographics

- Epidemiology
  - ~ 702,000 child maltreatment victims in USA in 2014
    - Infants ≤ 1 year old most at risk, account for 36.7%
    - Physical abuse in 41% of child maltreatment cases
  - CML has high specificity for NAT
    - Most common fracture in fatal NAT cases in 1 study
    - Present in 50% of infants at high risk for NAT compared to 0% in infants at low risk for NAT

Natural History & Prognosis

- ~ 1,580 fatalities from child maltreatment in USA in 2014
  - Death rate greatest in infants < 1 year old, 17.96/100,000 same-aged children in population
  - 71% of all deaths occurred in children < 3 years old
  - 79% of fatalities involved parent as perpetrator

Treatment

- Multidisciplinary investigation of maltreatment allegation
- Ensure "at risk" child & siblings placed in safe environment

DIAGNOSTIC CHECKLIST

Reporting Tips

- Concern for child abuse raised by imaging must be discussed with referring clinician immediately
- Report represents legal document: Review carefully
- Provide clear, detailed description of each fracture, including location, fragments, & estimated age

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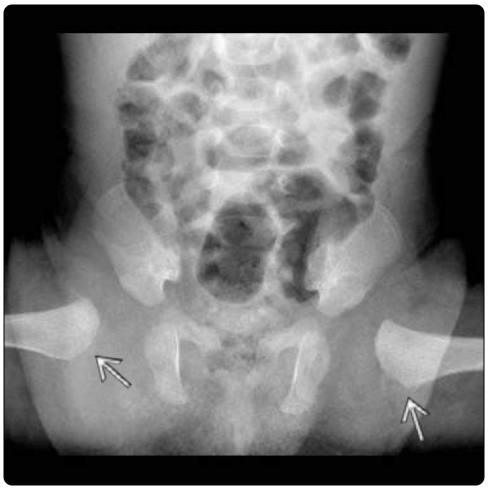
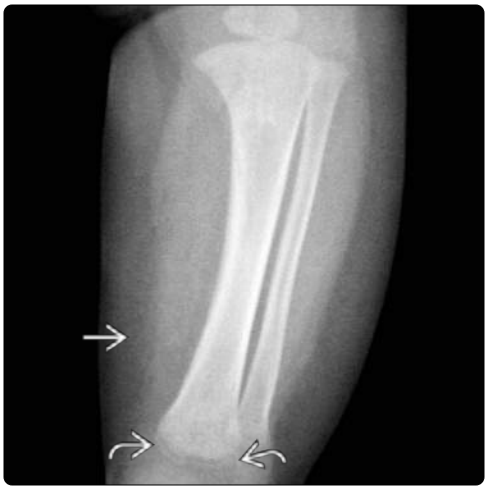
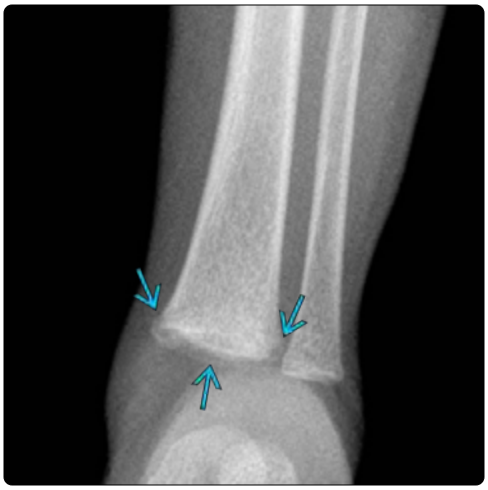
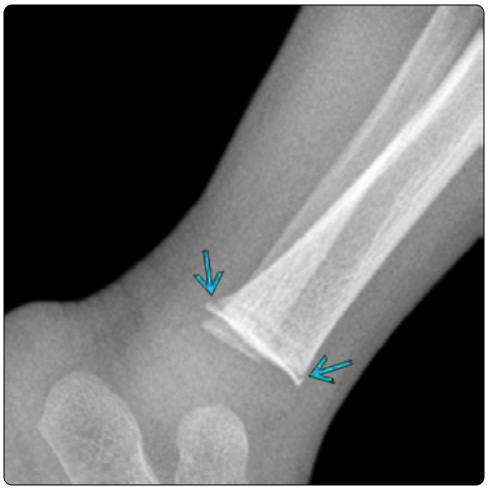


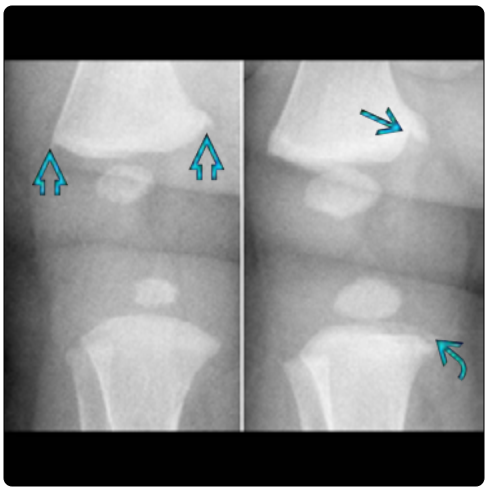


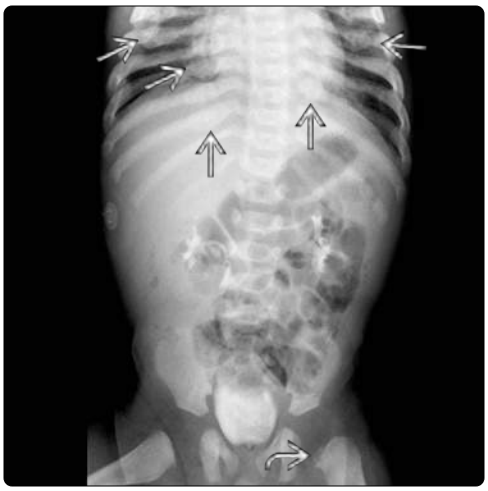
(Left) *AP radiograph in a 2-month-old patient shows bilateral metaphyseal corner/bucket-handle fractures of the proximal femurs. **(Right)** AP radiograph in a 3-month-old infant shows a bucket-handle fracture of the distal tibial metaphysis with overlying soft tissue swelling.*





(Left) *AP radiograph of the tibia of a 6 month old with failure to thrive demonstrates the bucket-handle appearance of an acute classic metaphyseal lesion. This is a high-specificity indicator of NAT. **(Right)** Lateral view of the tibia in the same patient shows the metaphyseal fracture. The periphery of the fracture undercuts the subperiosteal bone collar, which is thicker than the thin central component.*





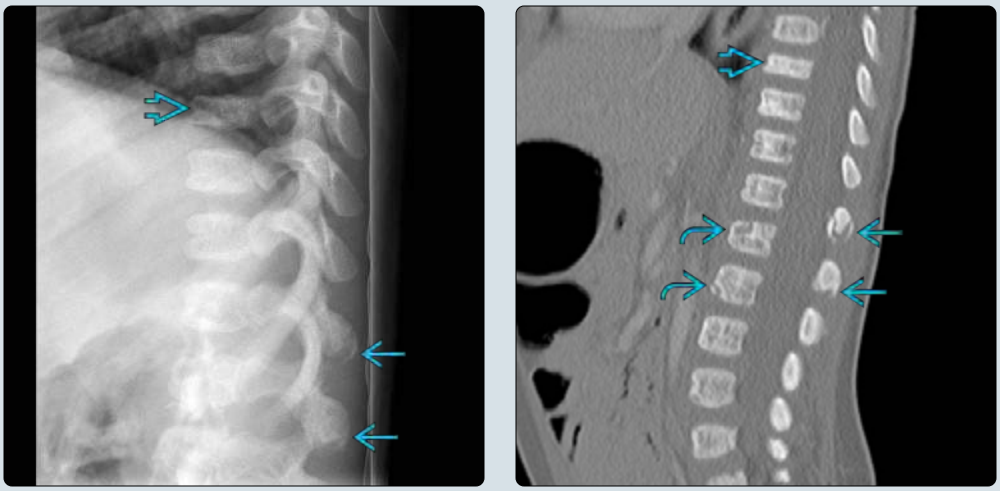
(Left) *AP radiographs [at presentation (left) & 7 weeks later (right)] of the right knee in a 3 month old with a skull fracture show an evolving appearance of a distal femoral metaphyseal corner fracture with focal physeal extension on the follow-up exam. Note the bucket-handle fracture of the tibia on the 2nd study. **(Right)** AP abdominal radiograph in a 4-month-old infant shows numerous healing posterior & lateral rib fractures. A bucket-handle metaphyseal fracture is also seen at the proximal left femur.*

KEY FACTS

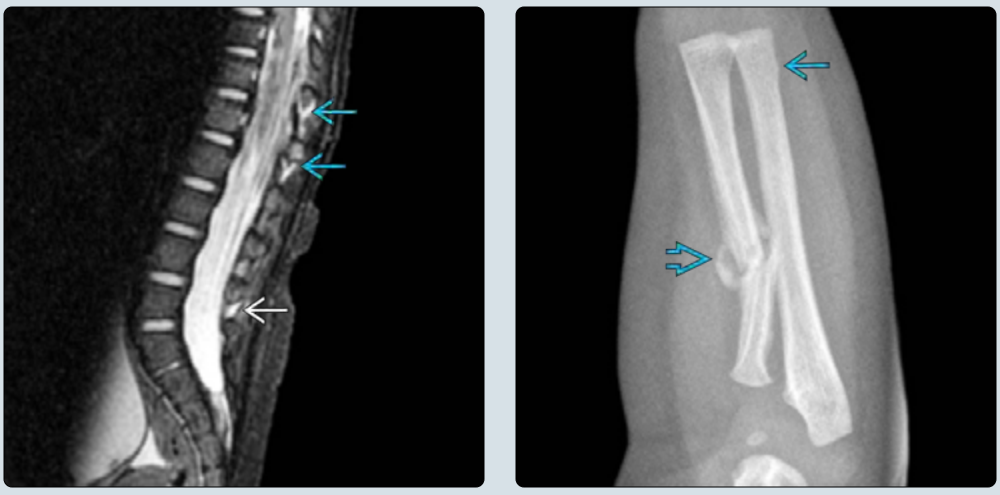
IMAGING

- High specificity for child abuse
  - Classic metaphyseal lesions, posterior rib fractures
  - Scapular fractures
    - Transverse or oblique fractures of midacromion process most common
    - Acromion tip fracture mimics ossification center
  - Sternal fractures
    - Linear lucency or buckling of anterior cortex
    - Widened sternal synchondrosis or malalignment of sternal segments
  - Spinous process fractures
    - Cartilage/bone avulsion at interspinous ligamentous attachment due to hyperflexion & shaking
    - Ossific density adjacent to spinous process may represent acute or remote injury
- Moderate specificity for child abuse
  - Vertebral body fractures
    - Compression deformity &/or anterosuperior endplate fracture
  - Vertebral fracture-dislocations
    - Neurocentral synchondrosis fracture extending through endplate apophyses with retropulsion of vertebral centrum
    - Facet dislocation ± fracture
  - Transphyseal fracture/distal humeral epiphyseal separation
    - Capitellar ossification center, radius, & ulna displaced posteriorly & medially relative to distal humerus
    - Distal humeral cartilage maintains alignment with radius & ulna (not true dislocation)
  - Complex skull fractures, hand & foot fractures, pelvic fractures
- Low specificity for child abuse
  - Clavicle, long bone shaft, & linear skull fractures common but have low specificity for nonaccidental trauma

(Left) *Lateral radiograph of the spine in an 8 month old with bruising & a subdural hematoma shows irregularity, lucency, & sclerosis of the T12 & L1 spinous processes ⇨ as well as compression of the T9 vertebral body ⇨. Spinous process fractures are highly suggestive of child abuse.* **(Right)** *Sagittal CECT (in bone algorithm & windows) in the same patient shows the vertebral spinous process ⇨ & compression ⇨ fractures as well as other levels of possible fracture ⇨.*



(Left) *Sagittal STIR MR in the same child shows abnormal fluid signal intensity ⇨ at the spinous process fractures & adjacent soft tissues. Additional interspinous ligament injury is seen distally ⇨.* **(Right)** *Oblique forearm radiograph in the same patient shows a healing, angulated proximal radial diaphyseal fracture ⇨ as well as a mild buckle deformity of the distal ulnar metadiaphyseal junction ⇨.*



TERMINOLOGY

Synonyms

- Nonaccidental trauma (NAT), battered child syndrome

IMAGING

General Features

- Best diagnostic clue
  - Classic metaphyseal lesions (CML) & posterior rib fractures: High specificity for NAT
    - Other high-specificity injuries include scapular, spinous process, & sternal fractures
  - Multiple fractures, bilateral fractures, & fractures of varying ages: Moderate specificity for NAT
    - Other moderate-specificity injuries include epiphyseal separation, vertebral body, digital, complex skull, & pelvic fractures
  - Injury incompatible with age & history suspicious for NAT
- Distribution of fractures due to NAT
  - Skull (27%), ribs & sternum (18%), vertebra (2%), pelvis (< 1%)
  - Clavicle (4%), humerus (11%), forearm (7%), hand (< 1%)
  - Femur (18%), tibia & fibula & ankle (12%), tarsal & metatarsal (< 1%)

High-Specificity Fractures

- Scapular fractures
  - Acromion most commonly involved
    - Transverse or oblique fractures of midprocess
    - Tip fractures less common, can mimic ossification center
  - Coracoid, inferior glenoid, scapula body fractures rare
  - Best evaluated on humerus & oblique rib views
  - CT or radionuclide exam for challenging cases
- Sternal fractures
  - Rare, result from direct force to sternum
  - Linear lucency or buckling of anterior cortex
  - Widened sternal synchondrosis or malalignment of sternal segments
  - Irregularity & sclerosis of synchondrosis osseous margins with healing
  - May require dedicated views or CT for evaluation
  - Likelihood of accidental cause ↑ > 1 year of age
- Spinous process fractures
  - Avulsion of cartilage &/or bone at interspinous ligamentous attachments due to shaking, hyperflexion
  - Ossific density adjacent to spinous process may represent
    - Acute avulsed bone
    - Remote avulsion with failure of fragment to reunite
    - Ossification within old avulsed cartilaginous fragment
  - ± vertebral body compression deformity

Moderate-Specificity Fractures

- Vertebral body fractures
  - Compression deformity &/or anterosuperior endplate fracture
  - Healing
    - Central vertebral body sclerosis
    - Anterosuperior endplate notching/lucency + marginal sclerosis with remote injuries
  - Findings may be subtle, underestimate injuries
    - MR & F-18 NaF PET more sensitive
- Vertebral fracture-dislocations
  - Neurocentral synchondrosis injury
    - Fracture extending through superior & inferior endplate apophyses & neurocentral synchondrosis posteriorly due to hyperflexion
    - Extrusion of vertebral centrum, typically posteriorly
      - Retropulsion of vertebral body on lateral view
    - Widened interpediculate distance & disc space narrowing on AP view
    - ± vertebral body compression deformity
    - ± mass effect on thecal sac on MR
  - Facet dislocation ± fracture
    - Widened interspinous distance on AP & lateral views
    - Retrolisthesis on lateral view
    - Facet joint disruption/widening
    - ± compression deformity &/or fractures involving facets, posterior elements, endplates
  - Paraspinal Ca^{2+} with healing
- Epiphyseal separations
  - Transphyseal fracture of distal humerus
    - Largely unossified capitellar ossification center translated posteriorly & medially relative to distal humerus but maintains alignment with radius
    - In absence of epiphyseal/capitellar ossification, posterior & medial translation of radius & ulna suggested if
      - Radius aligns with central & medial humeral metaphysis; ulna medial to humerus
      - Mimics dislocation (rare in infants); most dislocations occur laterally
    - ± metaphyseal fracture fragment
    - MR, US, or arthrogram helpful to confirm diagnosis & evaluate degree of displacement
      - Discontinuity of distal humeral epiphyseal cartilage from bony metaphysis
      - Posterior & medial translation of epiphysis, which maintains alignment with radius & ulna
    - Up to 50% due to NAT but can occur with birth or accidental trauma
  - Proximal femoral epiphyseal separation
    - Proximal femoral diaphysis translated laterally & proximally relative to ossified capital femoral epiphysis (or acetabulum if epiphysis unossified)
    - May develop coxa vara deformity
    - Birth trauma may appear identical
- Complex skull fractures
  - > 1 fracture line, stellate or branching, comminuted
- Digital fractures of hands & feet
  - Relatively uncommon, present in 4.9% of all positive skeletal surveys obtained for NAT
    - Buckle fractures of metatarsals, usually medial 1st metatarsal
    - Buckle fractures of metacarpals & phalanges
    - Likely result from twisting or bending forces
- Pelvic fractures
  - Subtle, typically involve superior pubic ramus in infants

- ± association with sexual assault in older children
- MR & F-18 NaF PET may be helpful

Low-Specificity Fractures

- Clavicle, long bone shaft, & linear skull fractures common but low specificity for NAT

Dating Fractures

- Soft tissue swelling only in first 1-2 days, ↓ by 7 days; seen again (less pronounced) from 15-35 days
- Subperiosteal new bone formation after 7-10 days, ↑ thickness until 25 days
- Soft callus usually by 15 days, ↓ by 35 days
- Bridging & remodeling by 14-21 days
- Hard callus by > 35 days

DIFFERENTIAL DIAGNOSIS

Accidental Trauma

- Age & clinical history consistent with fracture

Osteogenesis Imperfecta

- Multiple fractures, wormian bones, diffuse osteoporosis, ± blue sclera

Birth Trauma

- Correlates with age, birth history, & typical location

Rickets

- Demineralization with metaphyseal fraying, cupping, widening; loss of normal zone of provisional calcification

Metabolic Disorder-Related Bone Changes

- Mucopolysaccharidoses: Vertebral body beaking, oar-shaped ribs, short thick metacarpals with proximal pointing
- Menkes disease: Metaphyseal spurs, brittle hair, tortuous intracranial arteries

Normal Variants Confused With Fractures

- Normal variant vertebral notching typically at 1 level; no disc space narrowing or malalignment
- Minimal anterior sloping of midthoracic vertebrae

PATHOLOGY

General Features

- Hyperextension/hyperflexion with acceleration-deceleration forces from shaking account for many NAT-related fractures, including CML, posterior rib fractures, spinal fractures, avulsion-type injuries of scapula
- Grabbing & twisting/torsional forces: Transcondylar & digital fractures
- Direct impact mechanism: Sternal, skull, & scapular body fractures

CLINICAL ISSUES

Presentation

- Wide range of clinical presentations for NAT
  - Injury inconsistent with history or stage of development
  - Multiple injuries in various stages of healing
  - Bruising in nonmobile infant
  - Burns, retinal hemorrhages
- Majority of spinal fractures clinically inapparent
  - Severe & undiagnosed may result in cord injury
  - Significant association with intracranial injury

Demographics

- Epidemiology
  - Estimated 702,000 child maltreatment victims in 2014
    - Infants ≤ 1 year old most at risk, account for 36.7%
    - Physical abuse in 41% of child maltreatment cases

Natural History & Prognosis

- Estimated 1,580 fatalities from child maltreatment in 2014
  - Death rate greatest in infants < 1 year old, 17.96/100,000 same-aged children in population
  - 71% of all deaths occurred in children < 3 years old
  - 79% of fatalities involved parent as perpetrator

Treatment

- Multidisciplinary investigation of maltreatment alligations must involve physicians, social worker, Child Protective Services, legal authorities
- Ensure "at risk" child & siblings given safe environment
  - May involve removal of child from home, temporary/protective custody
  - 23% of maltreatment victims placed in foster care

DIAGNOSTIC CHECKLIST

Consider

- Imaging findings & their specificity for NAT must be correlated with clinical history & physical exam findings

Reporting Tips

- Concern for child abuse raised by imaging must be discussed with referring clinician ASAP
- Report represents legal document: Review carefully
- Provide clear, detailed description of each fracture, including location, fragments, & approximate age

SELECTED REFERENCES

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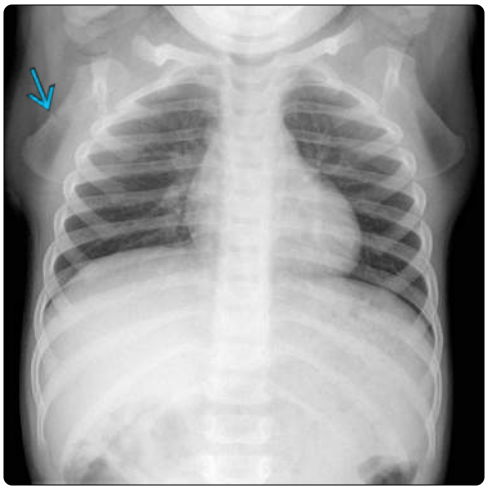
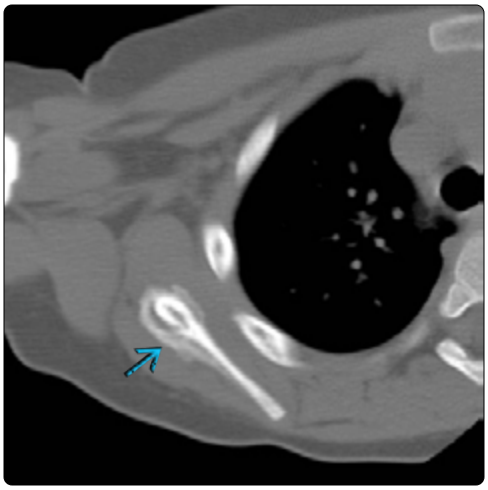
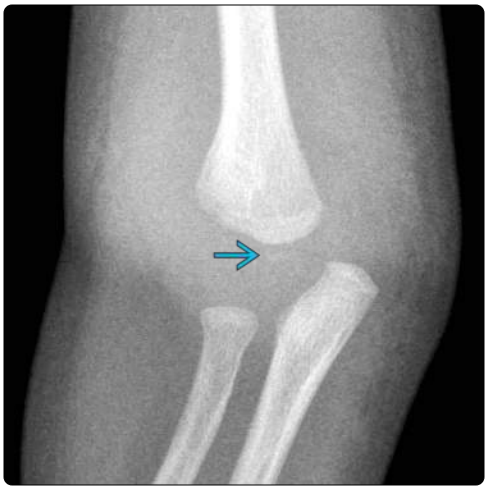
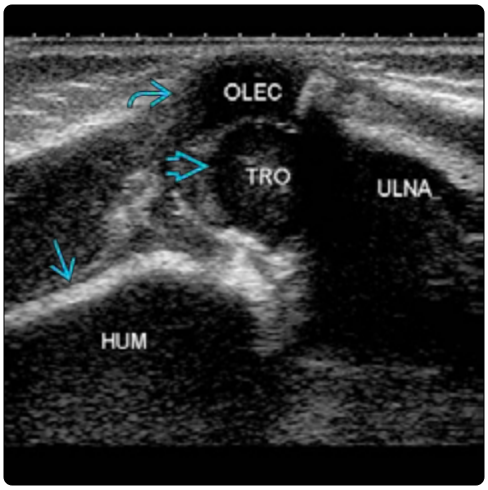


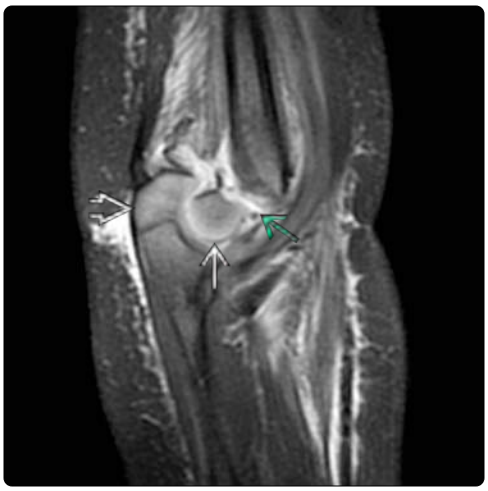


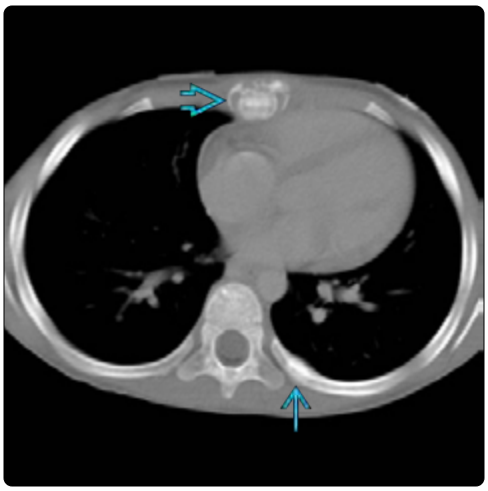
(Left) *AP chest radiograph in a 2 year old with bruising shows a fracture of the right scapular body.* **(Right)** *Axial bone CT in the same 2 year old with bruising shows callus formation about the right scapular body fracture. Scapular fractures in young children have a high specificity for child abuse.*





(Left) *External oblique radiograph in a 1 month old not using his arm shows medial translation of the capitellar ossification center, radius, & ulna relative to the distal humerus.* **(Right)** *Posterior longitudinal US of the elbow in the same patient shows posterior translation of the hypoechoic trochlear cartilage relative to the humerus. The olecranon still articulates with the trochlea. These findings are consistent with a distal humeral epiphyseal separation fracture rather than an elbow dislocation.*





(Left) *Sagittal T2 FS MR in a 6 month old after a reported fall shows a fracture through the distal humerus with posterior translation & angulation of the unossified trochlear cartilage & articulating olecranon cartilage. These findings are consistent with a distal humeral epiphyseal separation.* **(Right)** *Axial CECT (in bone windows) in a young child with abdominal pain shows healing fractures of the sternum & a posterior left rib. Both of these fractures are highly suggestive of child abuse.*

KEY FACTS

TERMINOLOGY

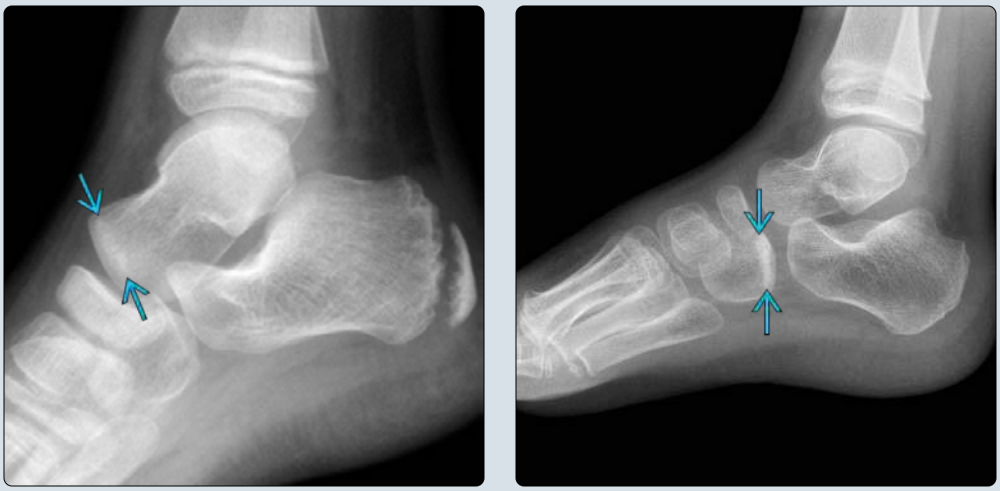
- Fatigue fracture: Fracture due to abnormal stresses applied (over time) to normal bone
- Insufficiency fracture: Fracture due to normal stresses applied to abnormal bone
- Stress response/reaction: Result of stresses upon bone prior to development of macroscopic fracture
- Chronic physeal stress injury: Repetitive stress to growth plate interrupts normal endochondral ossification

IMAGING

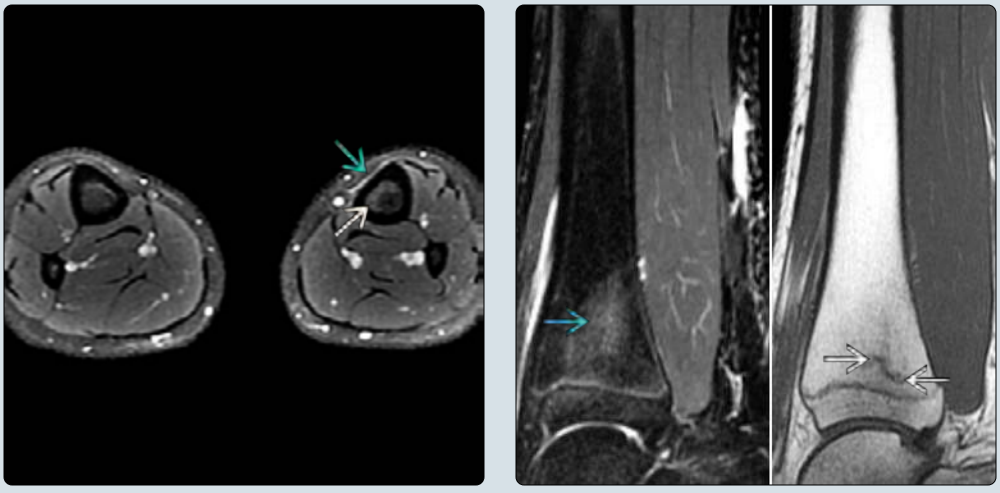
- Stress injury of formed bone
  - Periosteal new bone ± transversely oriented lucent cortical fracture or band of sclerosis on radiographs
  - Transverse linear focus of unicortical & medullary ↓ T1 & T2 signal (fracture line) + poorly defined surrounding ↑ T2 signal (edema) on MR = stress fracture
  - Limited marrow, periosteal, & soft tissue abnormalities without discrete fracture line = stress response
- Chronic physeal stress injury
  - Asymmetric lengthening ("widening") of lucent growth plate with metaphyseal irregularity on radiographs
  - Broad metaphyseal extension of cartilaginous physeal signal intensity on MR (especially PD/T2 FS or T2* GRE)

CLINICAL ISSUES

- Fatigue fractures occur in
  - Athletes with recent changes in routine
  - Newly ambulating children
  - Children with malalignment or altered weight bearing
  - Preadolescent children without ↑ activity intensity
- Insufficiency fractures occur in children with focal or systemic processes leading to bone weakening
- Treatment includes cessation of stresses with adequate time for bone repair & recovery of normal ossification
  - Rest & immobilization to prevent completion of fracture or permanent growth disturbance from physeal injury



(Left) *Lateral radiograph of the ankle in an 8 year old with limping shows a band of vertically oriented sclerosis in the talar head, typical of a stress injury.* **(Right)** *Lateral radiograph of the foot in a 3 year old shows a band of vertically oriented sclerosis in the posterior aspect of the cuboid, a common location for a stress injury.*



(Left) *Axial T2 FS MR in a 15-year-old female track athlete with left anterior lower leg pain shows mild edema of the tibial diaphyseal marrow plus increased fluid signal intensity overlying the anteromedial tibial cortex, consistent with grade 2 medial stress injury.* **(Right)** *Sagittal T2 FS (left) & T1 (right) MR images in the same patient show mild poorly defined marrow edema surrounding a low signal intensity stress fracture of the distal tibia (remote from the more proximal diaphyseal abnormality).*

TERMINOLOGY

Definitions

- Fatigue fracture: Fracture due to abnormal stresses applied (over time) to normal bone
- Insufficiency fracture: Fracture due to normal stresses applied to abnormal bone
  - Demineralized or otherwise weakened bone from focal or systemic process
- Stress response/reaction: Result of stresses upon bone prior to development of macroscopic fracture
- Chronic physeal stress injury: Repetitive stress to metaphyseal vasculature & cartilaginous growth plate interrupts normal endochondral ossification

IMAGING

Radiographic Findings

- Stress fracture
  - Poor cortical definition with varying degrees of cortical thickening/periosteal new bone
  - ± transversely oriented
    - Hairline fracture lucency extending centrally from involved cortex
    - Sclerotic band of medullary cavity
- Chronic physeal stress injury
  - Broad or focal lengthening ("widening") of lucent physis
  - Metaphyseal irregularity with loss of normal thin radiodense zone of provisional calcification

CT Findings

- Hairline transversely oriented unicortical lucency extending centrally with adjacent solid smooth periosteal reaction

MR Findings

- T1WI
  - Transverse hypointense stress fracture line extending from cortex into medullary cavity
  - Varying degrees of surrounding poorly defined hypointense marrow edema
- T2WI FS
  - Poorly defined hyperintense signal of marrow, periosteum, & soft tissues without discrete fracture line: Stress response
  - Transverse linear hypointense focus of medullary signal: Stress fracture
  - Metaphyseal extension of cartilaginous physeal signal intensity: Chronic physeal stress injury
    - Broad vs. small "tongue" of unossified cartilage
- T2* GRE
  - Cartilage-sensitive sequences nicely demonstrate abnormalities at interface of physis-metaphysis (i.e., chronic physeal stress injuries & rare secondary bony bridges)
- T1WI C+ FS
  - Enhancing marrow, periosteal, & soft tissue edema

Nuclear Medicine Findings

- Bone scan: Intense cortical uptake; sensitivity ~ 100%
- SPECT-CT: ↑ specificity for location & etiology
  - May help separate fracture from osteoid osteoma
  - Excellent for pars interarticularis stress fracture

DIFFERENTIAL DIAGNOSIS

Focal Periosteal Reaction, Marrow Edema

- Osteoid osteoma
- Osteomyelitis
- Bone malignancy
  - Ewing sarcoma, metastatic neuroblastoma, leukemia

Linear Sclerosis Within Bone

- Infarction

Lucent & Irregular Physes/Metaphyses

- Rickets
- Epiphysiodesis (drill type)
- Leukemia

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Stress fracture: Pain, swelling with appropriate history
  - Chronic physeal stress injury: Pain; rarely asymptomatic
  - Symptoms typically for weeks prior to presentation
- Clinical profile
  - Fatigue fractures in
    - Athletes with recent changes in routine
      - Medial tibial stress syndrome: Overuse or repetitive stress injury to shin area; may progress to fracture
    - Newly ambulating children
      - Young child with new refusal to bear weight
    - Children with malalignment or altered weight bearing
    - Preadolescent children with normal play activities & no elevation of activity intensity
  - Insufficiency fractures in
    - Children with focal or systemic processes leading to bone weakening
      - New stress reaction/insufficiency fracture in osteoporotic hindfoot due to ↑ ambulation after weeks of casting for preceding distal tibial fracture

Treatment

- Stress fracture
  - Prevention paramount: Gradual ↑ in new activity intensity + prompt activity reduction when pain occurs
  - Combination of reduced activity, rest, immobilization, casting, & (rarely) internal fixation
- Chronic physeal stress injury
  - Rest & immobilization typically sufficient for endochondral ossification to resume

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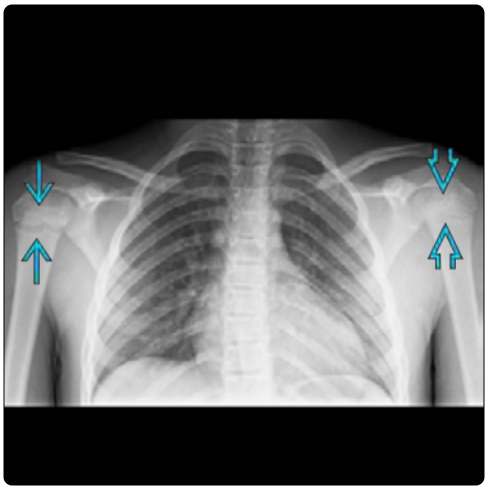
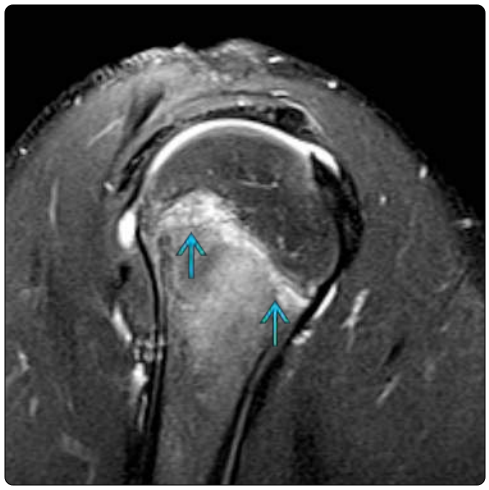
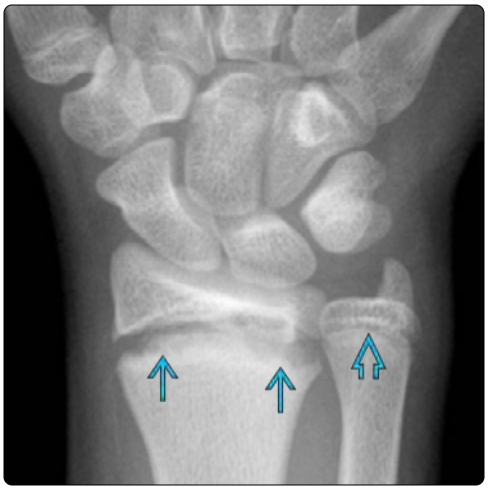
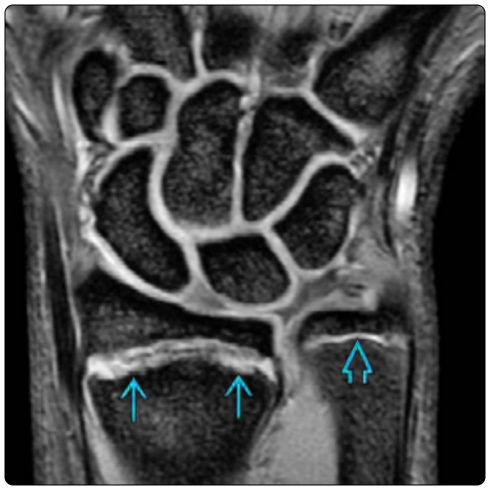
(Left) *AP radiograph of the shoulders in a 14-year-old pitcher with weeks of right shoulder pain shows lengthening of the proximal right humeral physis as compared to the normal left side. Note the pseudofracture appearance of both proximal humeri due to the normal obliquity of the growth plates.* **(Right)** *Sagittal T2 FS MR in the same patient shows an abnormally long, irregular, & hyperintense physis, typical of a chronic repetitive stress injury of the growth plate in this pitcher ("little league shoulder").*



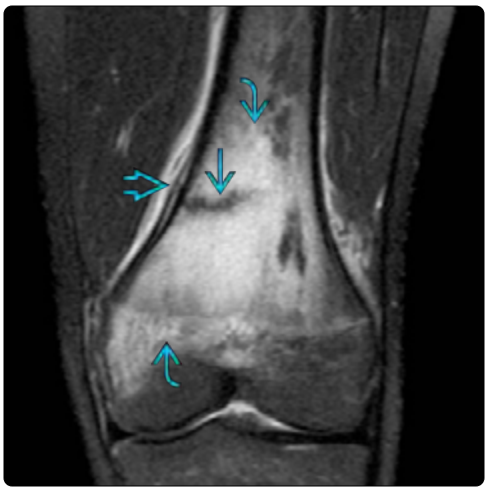


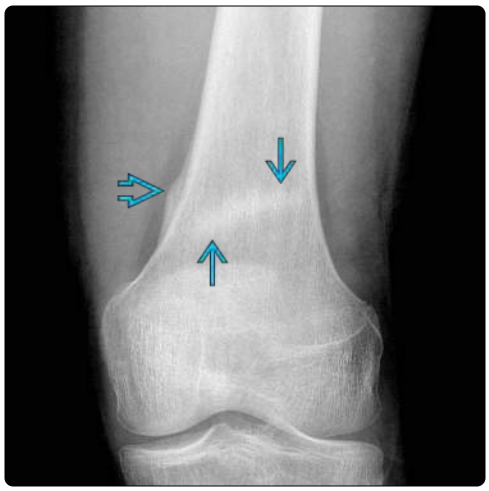
(Left) *PA wrist radiograph in a 13-year-old female gymnast with pain shows abnormal lengthening of the cartilaginous distal radial physis with metaphyseal irregularity & loss of the normal thin dense zone of provisional calcification. Note the normal distal ulnar physis.* **(Right)** *Coronal T2* GRE MR in this patient confirms abnormal lengthening of the radial physis due to repetitive microtrauma ("gymnast wrist"). The ulnar physis is normal as it does not endure the same physical stresses as the radius.*

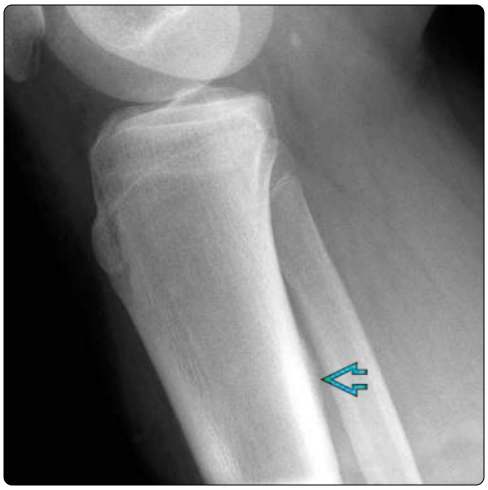


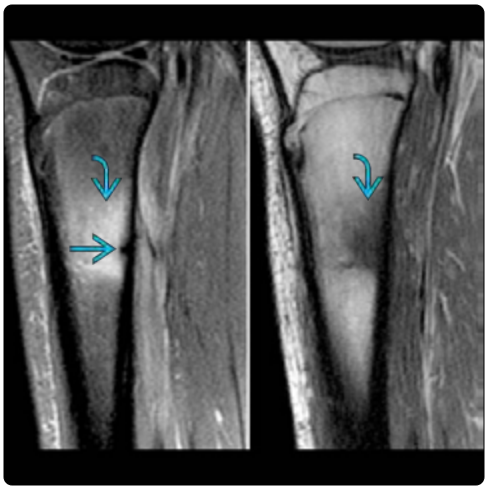


(Left) *Coronal T2 FS MR in a 16 year old with knee pain shows a transverse band of low signal intensity in the distal femoral diaphysis, consistent with an incomplete stress fracture. There is surrounding marrow edema & overlying periosteal reaction.* **(Right)** *Follow-up AP radiograph in the same patient 4 weeks later shows sclerosis of the healing stress fracture with focal solid periosteal reaction at this level.*

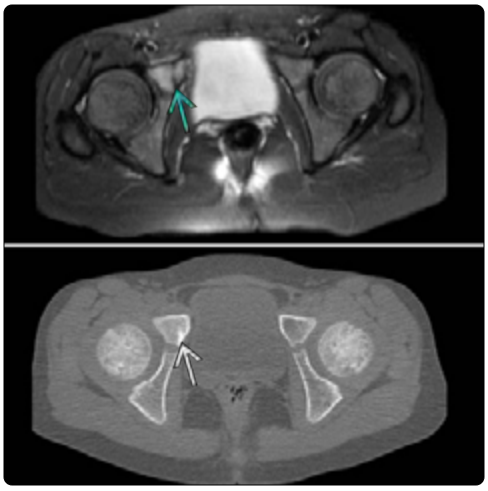


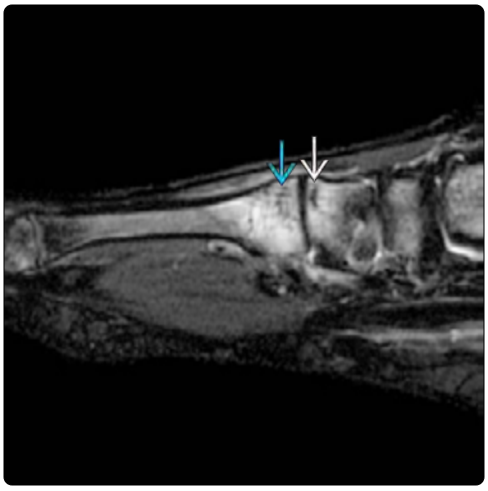




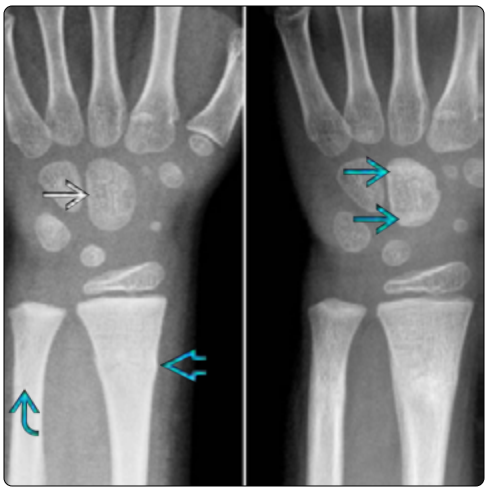


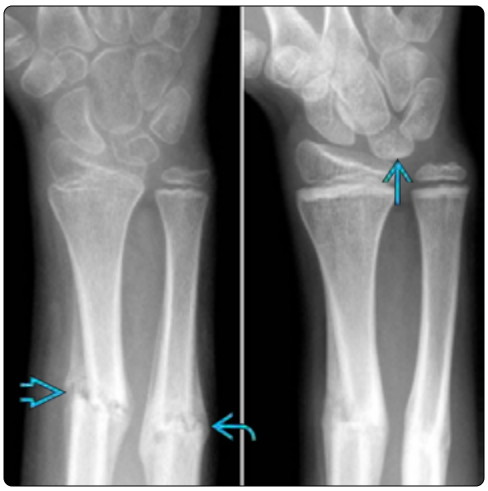
(Left) *Lateral radiograph in a 14 year old with lower leg pain shows subtle solid periosteal reaction focally at the posterior proximal right tibial diaphysis. **(Right)** Sagittal T2 FS (left) & T1 (right) MR images of the same patient show poorly defined marrow edema surrounding a small linear corticomedullary focus of low signal intensity, consistent with a stress fracture.*





(Left) *Axial T2 FS MR (top) of the pelvis in a 6 year old with pain shows a low signal intensity fracture line of the superior pubic ramus with surrounding marrow edema. Axial bone CT (bottom) shows sclerosis at this same level, consistent with a healing stress fracture.* **(Right)** *Sagittal STIR MR in a 16-year-old cross country runner with pain shows linear low signal intensity foci in the 2nd metatarsal base & middle cuneiform with surrounding marrow edema, consistent with stress fractures.*





(Left) *PA wrist radiographs taken 6 weeks apart in a 6 year old after a fall show incomplete fractures of the distal radius & ulna initially (left). The capitate is normal at that time. Six weeks later (right), there is disuse osteoporosis distally with horizontal stress reaction of the capitate.* **(Right)** *PA wrist radiographs in a 12 year old taken at 2 (left) & 3 (right) months after distal radial & ulnar fractures show new sclerosis in the osteoporotic lunate, consistent with stress reaction.*

KEY FACTS

TERMINOLOGY

- Collection of poorly understood "disorders" of immature skeleton
  - > 70 entities, many with eponyms
- Many entities reflect symptomatic growth disturbances with elements of idiopathic osteonecrosis &/or overuse injury
- Symptoms range from asymptomatic → pain → growth disturbances
- Outcomes range from resolution of self-limited process → permanent deformity if untreated

IMAGING

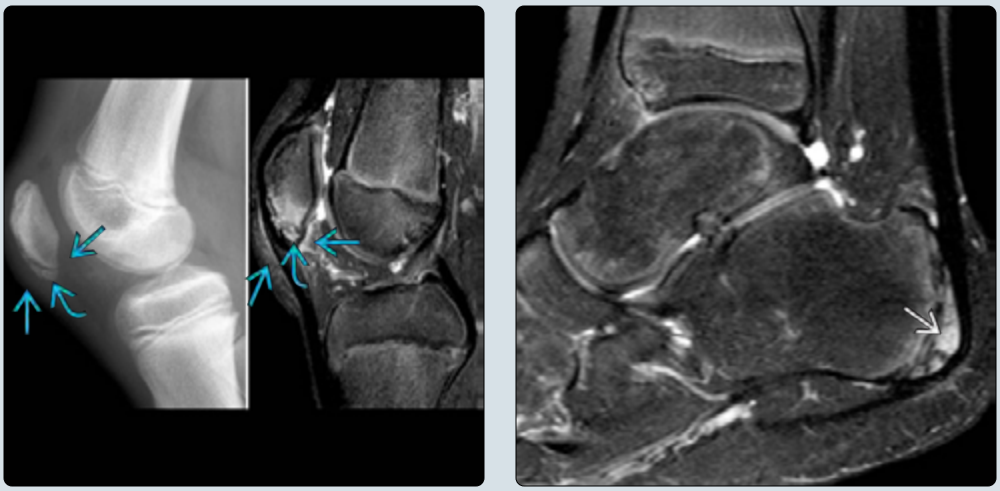
- Affect certain sites where bone growth occurs by endochondral ossification (epiphyses, apophyses > metaphyses)
- Radiographs: Fragmentation, sclerosis, flattening
  - Depending on location, symptoms, & patient age, such findings do not necessarily connote disease
  - Normal variants of irregular epiphyseal ossification can occur at certain locations
- MR: Marrow & soft tissue edema typically correlate with symptoms
  - Subtracted pre/postcontrast T1 FS images may show ↓ enhancement in setting of ischemia/necrosis
  - DWI/ADC may also reflect ischemia

CLINICAL ISSUES

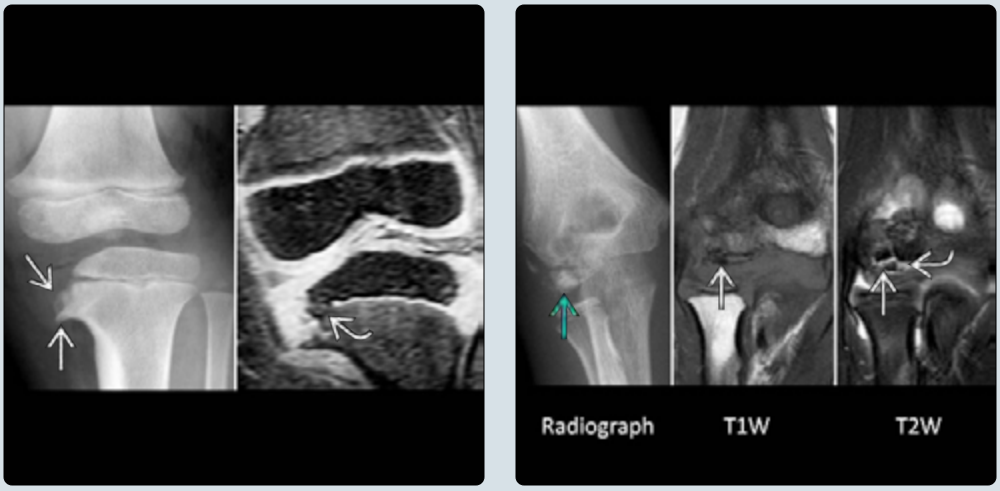
- Conservative therapy with rest effective for many
- Certain entities (e.g., Blount, LCP) often require surgical intervention to preserve long-term function

DIAGNOSTIC CHECKLIST

- Not every irregular ossification center abnormal
  - Correlate with site, patient age, & symptoms
    - Helpful to know locations where irregular ossification normally occurs vs. never occurs
  - MR useful adjunct for determining source of pain



(Left) *Lateral radiograph (left) & sagittal T2 FS MR (right) of a 12-year-old boy with anterior knee pain show fragmentation of the inferior pole of the patella with adjacent soft tissue swelling. The findings are consistent with Sinding-Larsen-Johansson.* **(Right)** *Sagittal T2 FS MR shows edema in the calcaneal apophysis, consistent with Sever disease in this 8-year-old boy with posterior heel pain. Fragmentation & sclerosis may normally be seen at this level but edema should not.*



(Left) *AP radiograph (left) shows downsloping, beaking, & irregularity of medial tibial metaphysis (Blount disease). Coronal GRE MR (right) shows abnormal ossification of medial tibial epiphysis & metaphysis with an osseous bridge crossing the tibial physis.* **(Right)** *AP radiograph (left) of an 8-year-old girl with Panner disease shows fragmentation & sclerosis of the capitellum. There is associated low signal intensity at this level on the T1 (middle) & T2 FS (right) MR images with interspersed fluid signal intensity.*

TERMINOLOGY

Synonyms

- > 70 entities, many with eponyms

Definitions

- Collection of poorly understood "disorders" of immature skeleton
- Many entities reflect symptomatic growth disturbances with elements of idiopathic osteonecrosis &/or overuse
- Symptoms range from asymptomatic → pain → chronic manifestations of growth disturbances
- Outcomes range from resolution of self-limited process → permanent deformity if untreated

IMAGING

General Features

- Best diagnostic clue
  - Radiographic fragmentation, sclerosis, flattening
    - Depending on location, symptoms, & patient age, such findings do not necessarily connote disease
    - Normal variants of irregular epiphyseal ossification can occur at certain locations
  - Bone marrow & soft tissue edema by MR
    - Typically correlate with symptoms
- Location
  - Affect certain sites where bone growth occurs by endochondral ossification (epiphyses, apophyses > metaphyses)
  - > 1 osteochondrosis can occur at some locations with differing clinical implications
  - Most frequent sites
    - Thoracic spine: Scheuermann disease
    - Capitellum: Panner disease, osteochondritis dissecans (OCD)
    - Lunate: Kienbock disease
    - Hip: Legg-Calvé-Perthes, Meyer dysplasia
    - Inferior patella: Sinding-Larsen-Johansson
    - Proximal, medial tibia: Blount disease
    - Tibial tubercle: Osgood-Schlatter disease
    - Calcaneal apophysis: Sever disease
    - Tarsal navicular: Köhler disease
    - 2nd or 3rd metatarsal head: Freiberg disease

Radiographic Findings

- Irregularity, fragmentation, sclerosis, flattening at site of endochondral ossification
  - Chronic changes may lead to deformity (e.g., LCP) &/or osteoarthritis (e.g., Freiberg)
- Bony bridges rarely form across physes in this setting, leading to angular deformities or limb length discrepancy (e.g., Blount)
- ± adjacent soft tissue edema

MR Findings

- Abnormal sclerosis: Low T1 & T2 signal
- Marrow & soft tissue edema: Poorly defined high T2 signal
- Fragmentation/collapse: Discrete linear fluid signal intensity
- Osteonecrosis: Variable depending on stages
  - Subtracted pre/postcontrast T1 FS images show earliest findings of ischemia & necrosis (↓ enhancement) & subsequent revascularization (↑ enhancement)
  - DWI can show early ischemia

Imaging Recommendations

- Best imaging tool
  - Radiographs correlated with specific symptoms
  - MR useful adjunct if clinically uncertain

DIFFERENTIAL DIAGNOSIS

Normal Developmental Variants

- Asymptomatic
- Radiographic findings only with essentially normal MR
  - No marrow edema
- Characteristic locations (e.g., humeral trochlea)

Osteonecrosis of Systemic Disorders

- Sclerosis, fragmentation, collapse of epiphyses
- Often widespread with predisposing conditions

Spondyloepiphyseal Dysplasia

- Epiphyseal dysplasias may or may not involve spine
- Epiphyses affected throughout body

Septic Arthritis

- Joint effusion, synovitis, marrow & soft tissue edema
- Fever, leukocytosis, altered weight bearing typical

Juvenile Idiopathic Arthritis

- Swelling, pain, joint effusion, synovitis acutely
- Epiphyseal overgrowth, erosions, joint space loss chronically

CLINICAL ISSUES

Presentation

- Site-dependent: Pain, limp, or limb deformity possible

Treatment

- Conservative therapy with rest effective for many
- Certain entities (e.g., Blount, LCP) often require surgical intervention to preserve long-term function

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Not every irregular ossification center abnormal
- Helpful to know locations where irregular ossification
  - Occurs normally (e.g., trochlea of humerus)
  - May occur normally with unrelated, superimposed symptoms (e.g., Sever disease)
  - Never occurs normally (e.g., capitellum of elbow)
- MR useful adjunct for determining source of pain

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Osteochondritis Dissecans

KEY FACTS

TERMINOLOGY

- Focal joint disorder with progressive changes in subchondral bone & overlying articular cartilage that may lead to early joint degeneration
- Divided into juvenile & adult osteochondritis dissecans (OCD) based on skeletal maturity (with open or closed physes, respectively)

IMAGING

- MR predictors of instability in juvenile OCD (JOCD) include
  - High T2 signal rim of fluid surrounding OCD + 2nd deeper rim of low T2 signal
  - Multiple breaks in subchondral bone
  - Multiple cysts or single cyst larger than 5 mm
  - Fluid-filled OCD defect
- MR predictors of instability in adult OCD include
  - Displaced fragment or loose body
  - High T2 signal rim surrounding fragment
  - High T2 signal fracture line through articular cartilage

TOP DIFFERENTIAL DIAGNOSES

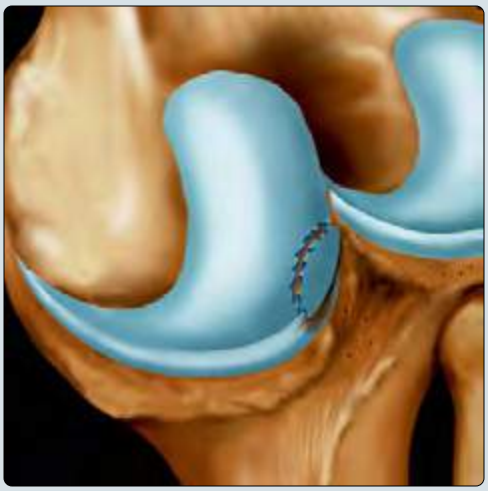
- Normal irregular distal femoral epiphyseal ossification
- Avascular necrosis
- Osteochondral impaction fracture
- Stress or insufficiency fracture

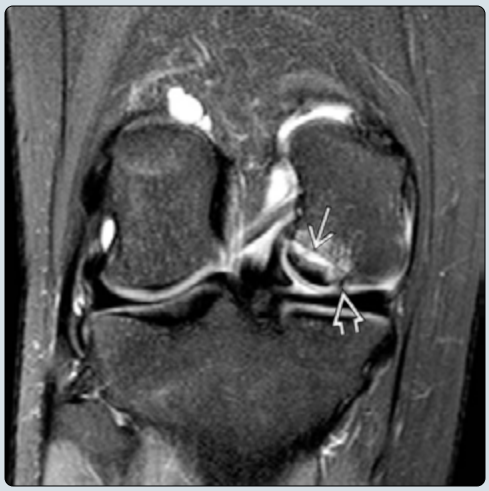
PATHOLOGY

- Etiology likely multifactorial (traumatic, ischemic, genetic)
  - Favored mechanism: Repetitive microtrauma
  - JOCD may represent growth disturbance of secondary physis; little necrosis or inflammation on histology

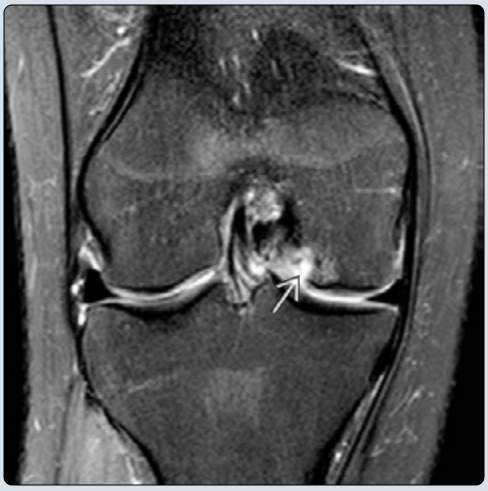
CLINICAL ISSUES

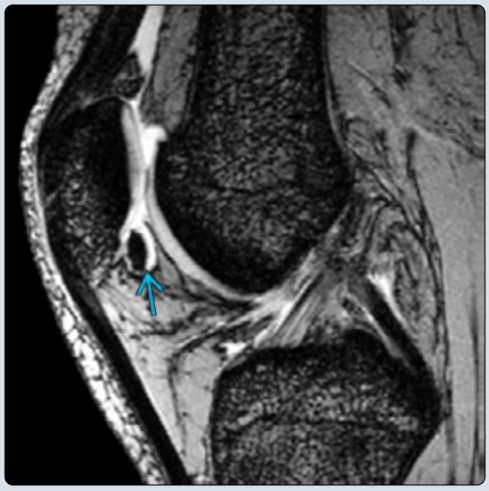
- Most common in adolescent athletes
- Traditional treatment schemes lack strong evidence
  - Stable JOCD lesions often heal over 6-18 months with conservative therapy
  - Variety of surgical treatment options for stable OCD failing conservative therapy or unstable OCD





(Left) *Coronal oblique graphic shows circumferential fissuring of the articular cartilage of the posterolateral aspect of the medial femoral condyle.* **(Right)** *Coronal T2 FS MR in a 15-year-old female patient shows the in situ portion of a complex osteochondral lesion of the medial femoral condyle. Fluid ➔ undercuts the fragment with an overlying T2-hypointense cartilage crack ➔ noted, consistent with an unstable lesion.*





(Left) *Slightly more anterior T2 FS MR slice from the same patient shows fluid filling an osteochondral defect ➔ of the "parent" femoral condyle.* **(Right)** *Sagittal midline T2* GRE MR in the same patient shows the displaced osteochondral fragment ➔ along the inferior patellar articular surface. The fragment was shown to be loose (rather than synovialized & fixed) by interval changes in position on radiographs (not shown).*

TERMINOLOGY

Abbreviations

- Osteochondritis dissecans (OCD)

Synonyms

- Osteochondral lesion, juvenile OCD (JOCD), adult OCD

Definitions

- Focal joint disorder with progressive changes in subchondral bone & overlying articular cartilage that may lead to early joint degeneration
- JOCD limited to skeletally immature patients (open physes)

IMAGING

General Features

- Best diagnostic clue
  - Osteochondral lesion at lateral aspect of medial femoral condyle ± loose fragment in young adolescent athlete
- Location
  - Knee most frequently affected
    - Lateral aspect of medial femoral condyle (69%)
    - Lateral femoral condyle (15%), patella (5%), femoral trochlea (1%)
    - Bilateral: 15-33%
  - Less frequent in elbow, ankle, hip
  - Very rare in shoulder, wrist
- Morphology
  - Crescent-/oval-shaped osteochondral lesion contiguous with donor/parent bone (in situ) vs. detached intraarticular fragment

Radiographic Findings

- Crescentic/ovoid lucent subchondral bone lesion surrounded by sclerotic margin
- ± in situ or displaced intraarticular osseous fragment
- Underestimates lesion size, does not assess cartilage integrity

MR Findings

- T1WI
  - Hypointense fragment
  - ± hypointense subchondral marrow edema
  - ± hyperintense fibrovascular tissue on 3D T1 SPGR FS
- T2WI
  - Variable hyperintense signal in osteochondral fragment & adjacent marrow
  - Overlying fissures/defects in articular cartilage best appreciated on PD/T2 ± FS
  - PD/T2 FS may demonstrate direct cartilaginous &/or bony extension of fluid
  - ± hyperintense cysts or fibrovascular tissue deep to lesion
  - ± hyperintense synovial thickening or joint fluid
  - Specifically in JOCD
    - Laminar "Oreo cookie" hyperintensity surrounded by hypointensity at sites of osteochondral clefts
    - Disruption of thin, circumferential hyperintense secondary physis overlying OCD lesion
    - Thickened overlying unossified epiphyseal cartilage
- T2* GRE
  - May help visualize displaced osteochondral fragment
    - Purely chondral fragments may be best seen on FSE FS images
- MR arthrography with gadolinium-based agent
  - Contrast between OCD fragment & parent bone = unstable fragment
  - Intraarticular body loose/free if surrounded by contrast (rather than synovialized/fixed)
- **Key MR features for treatment planning**
  - Predictors of instability in JOCD
    - High T2 signal intensity rim (fluid) surrounding fragment + 2nd deeper rim of low T2 signal intensity
    - High T2 signal intensity fracture line/fissure of articular cartilage
      - Cartilage crack may be of low T2 signal intensity
    - Multiple breaks in subchondral bone
    - Multiple cysts or single cyst larger than 5 mm
    - Fluid-filled OCD defect
  - Predictors of instability in adult OCD
    - Displaced/detached fragment
    - High T2 signal intensity rim (fluid) at fragment/parent bone interface
    - High T2 signal intensity line (fluid) in articular cartilage + subchondral bone
    - Cysts surrounding OCD
  - Predictors of stability
    - Fragment continuity with parent bone without linear fluid signal intensity interface, cyst, or cartilage fissuring
  - MR accuracy for predicting stable vs. unstable OCD lesion as compared to arthroscopy: 30-92%
  - MR findings of healing after therapy
    - Variable osteochondral changes depending on type of intervention
    - In general, healing indicated by
      - ↓ lesion size, cysts, marrow edema
      - ↑ bone filling OCD lesion bed
      - Improved morphology of articular cartilage surface

Imaging Recommendations

- Best imaging tool
  - MR or MR arthrography
- Protocol advice
  - PD/T2 FS MR sagittal, coronal ± axial sequences (depending on OCD site)
  - 3D GRE FS MR cartilage-sensitive sequence
  - MR arthrography with gadolinium-based contrast agent can aid instability assessment

DIFFERENTIAL DIAGNOSIS

Normal Irregular Distal Femoral Epiphyseal Ossification

- Asymptomatic younger patients
- Most commonly in posterior condyle
- Often has "puzzle piece" fragment-parent bone interface
- Deep rather than flat
- No adjacent bone marrow edema
- Overlying cartilage intact, including thin, T2-hyperintense secondary physis

Avascular Necrosis

- Serpentine sclerotic foci with characteristic double-line appearance on T2 MR
  - May lead to subchondral collapse
- History of steroid therapy, lupus, sickle cell disease, other predisposing condition

Osteochondral Impaction Fracture

- Single traumatic event
- Different location than typical OCD lesion

Stress or Insufficiency Fracture

- Sclerotic band, often horizontal to bone long axis
- Not usually subchondral

PATHOLOGY

General Features

- Etiology
  - Favored mechanism: Repetitive microtrauma
    - Acute trauma, ischemia, genetic predisposition may be contributory
  - JOCD may represent symptomatic growth disturbance of epiphyseal secondary physis
  - Adult OCD may be incompletely healed JOCD lesion

Staging, Grading, & Classification

- Arthroscopic grading
  - Research in Osteochondritis of the Knee (ROCK) 2013
    - Immobile lesions
      - 0: Normal
      - 1: Cartilage intact with subtle lesion demarcation
      - 2: Fissure, buckle, or wrinkle of cartilage
    - Mobile lesions
      - 3: Intact cartilage
      - 4: Peripheral fissuring, unable to hinge
      - 5: Peripheral fissuring, able to hinge
      - 6: In situ with circumferential fissuring, complete separation
      - 7: Exposed subchondral defect

Microscopic Features

- No substantial necrosis or inflammation in JOCD lesions
- Abundant fibrovascular tissue at osteochondral interface & in subchondral bone
- Clefts at osteochondral interfaces
- Thickening of epiphyseal cartilage + chondrocyte cloning

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain aggravated by activity
  - Mechanical symptoms (clicking, catching, grinding, locking) raise concern for unstable OCD
- Other signs/symptoms
  - Can be asymptomatic

Demographics

- Age
  - Primarily 10-20 years
  - Mean age of JOCD 11.3 years by one study
  - Symptoms often lasting > 1 year prior to diagnosis
- Gender
  - M:F = 2-3:1
- Epidemiology
  - 8.7/100,000 ages 6-11; 21.8/100,000 ages 12-19
  - Often seen in athletes

Natural History & Prognosis

- ↑ rate of spontaneous healing in JOCD vs. adult OCD
  - 50-67% of JOCD lesions heal in 6-18 months with conservative therapy only
    - Spontaneous healing: Stable > > unstable
- American Academy of Orthopaedic Surgeons (AAOS) practice guidelines state, "natural history of OCD of the knee remains unclear"

Treatment

- AAOS 2010 review of OCD evidence largely inconclusive regarding diagnosis + treatment recommendations
- Stable lesions generally treated with rest, casting, NSAIDs over 3-12 months
  - Failure to heal on conservative therapy → surgery
    - Drilling (transarticular or retroarticular) to create vascular channels
- Unstable lesions → surgery
  - Salvageable: Internal fixation of fragment
    - Metal screws; bioabsorbable pins, screws, or nails
    - ± bone autograft or allograft
  - Unsalvageable: Cartilage repair/restoration
    - Chondroplasty
    - Microfracture
    - Osteochondral autograft/allograft transfer system
    - Autologous chondrocyte implantation

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Careful evaluation for signs of instability ± loose body

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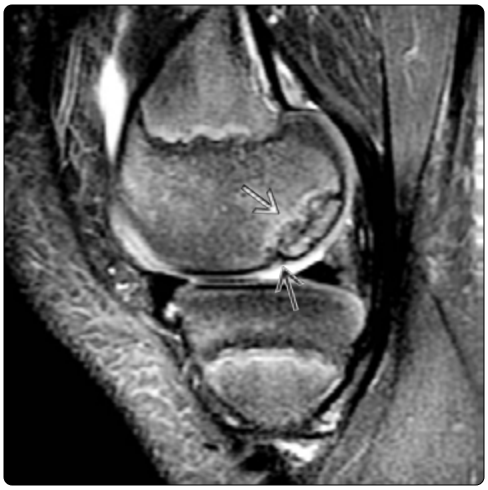
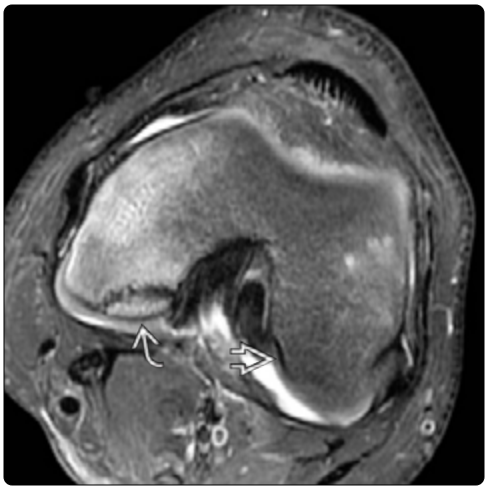
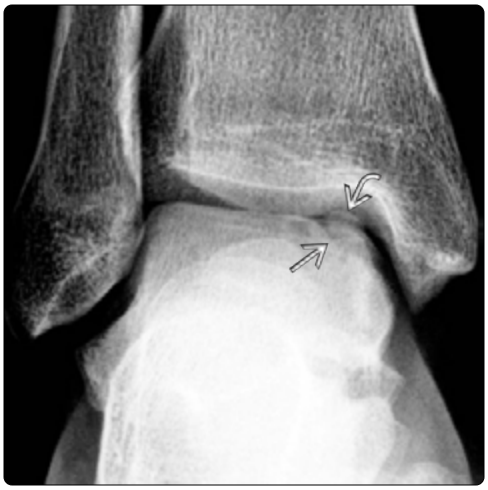
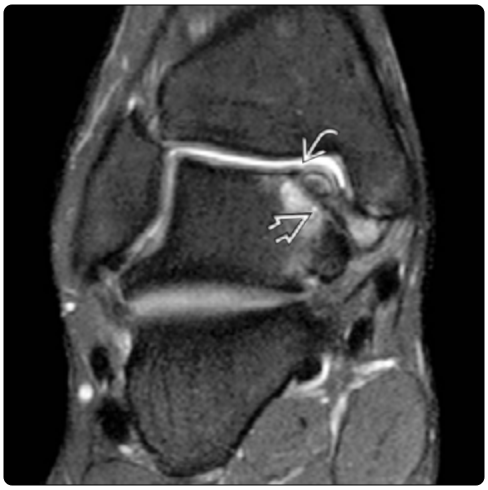


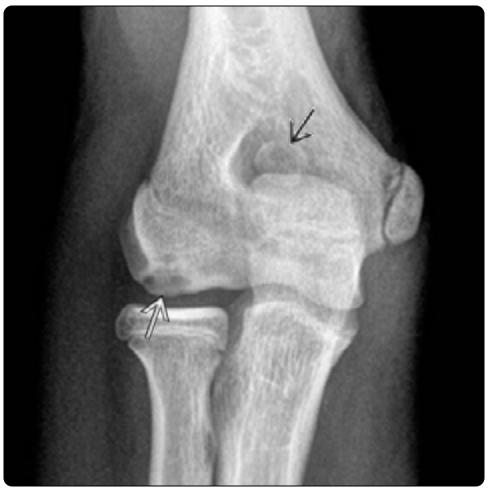


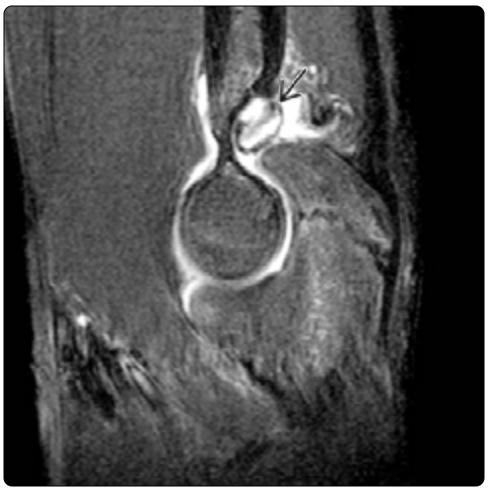
(Left) *Sagittal T2 FS MR in an 11-year-old boy with pain shows a "puzzle piece" morphology to an ossific fragment in the posterior femoral condyle, most typical for a normal ossification variant. However, the marrow edema ➡ & overlying articular cartilage contour deformity ⇨ favor juvenile OCD.* **(Right)** *Axial T2 FS MR in the same patient shows focal loss ➡ of the thin, hyperintense stripe of the normal secondary physis ➡ of the epiphyses. This feature also favors a juvenile OCD lesion.*





(Left) *AP ankle radiograph in a 16-year-old boy shows a crescentic lucent lesion in the medial talar dome ➡ with a bony fragment ➡ protruding into the joint space.* **(Right)** *Coronal T2 FS MR in the same patient confirms the OCD lesion of the medial talar dome with adjacent cartilage fissuring ➡. Cystic change ➡ & surrounding edema are noted in the parent bone. While no fluid is seen undercutting the fragment, the constellation of radiographic & MR findings remains suspicious for an unstable lesion.*





(Left) *AP elbow radiograph in a 13-year-old male patient with pain & locking shows an irregular lucent defect ➡ with sclerotic margin in the humeral capitellum, typical of OCD. An ossific body ⇨ projects over the olecranon fossa.* **(Right)** *Sagittal T2 FS MR in the same patient confirms an edematous intraarticular osteochondral body in the olecranon fossa ⇨ with surrounding joint effusion.*

KEY FACTS

TERMINOLOGY

- Nonunion: Lack of progression to union > 6 months after injury with corresponding clinical signs & symptoms
- Malunion: Nonanatomical alignment of fracture fragments
- Premature physeal closure: Osseous physeal bridge (bar) formation across cartilaginous physis

IMAGING

- Nonunion: Sclerosis at ends of bones, absence of bridging trabecula, persistence of fracture line
- Premature physeal closure
  - Progressive focal narrowing or interruption of lucent physis ± discrete bone bridge/bar
  - Abnormal (obliquely oriented or asymmetric) growth arrest/growth recovery lines
  - Peripheral bridge → angular deformity
  - Central bridge → longitudinal growth restriction
  - 3D SPGR or T2* GRE FS MR can generate physeal map
    - Assess percent of physis occupied by bridge
- Posttraumatic cyst-like lesion
  - Radiographs: Round or ovoid, lucent, eccentric, subperiosteal lesion, typically in distal radius or tibia
  - CT/MR: Fat characteristics
- Osteonecrosis: T1 C+ FS MR can help determine viability of proximal fragment in scaphoid nonunions

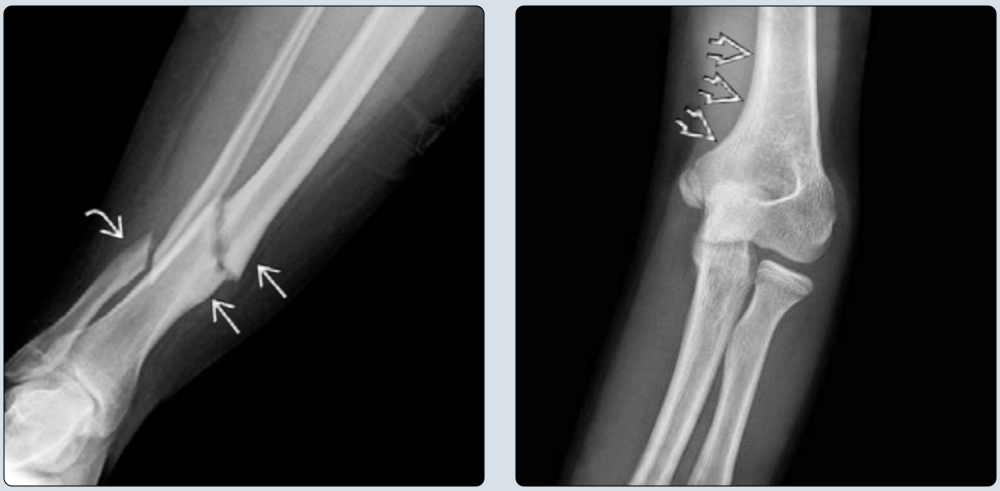
CLINICAL ISSUES

- Malunion common in pediatric fractures (but have high corrective potential due to remodeling in growing skeleton)
- Physeal bridges begin 1-2 months following injury but may not manifest until years later during adolescent growth

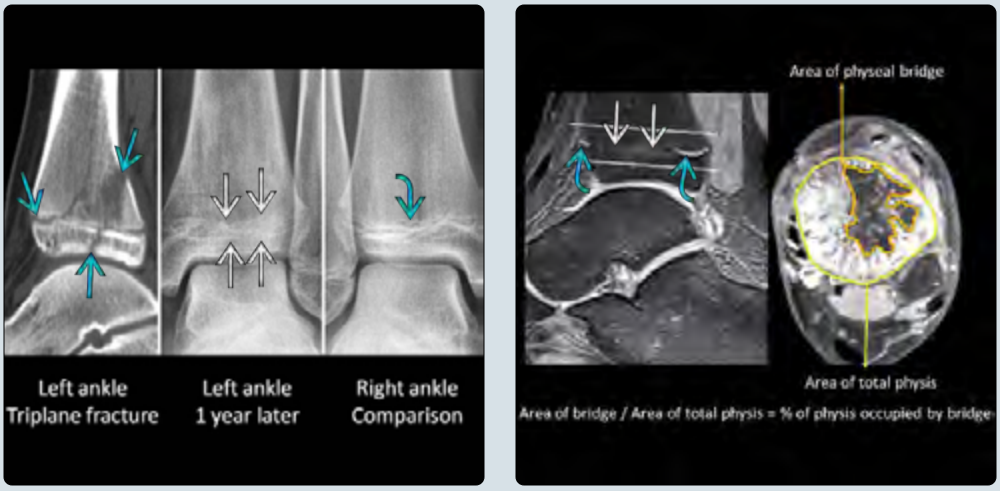
DIAGNOSTIC CHECKLIST

- Imaging findings of nonunion & malunion must be clinically correlated; from medicolegal standpoint, it may be best to be descriptive & discuss with orthopedist rather than use these terms in report

(Left) *AP radiograph of a 13-year-old boy shows new fractures superimposed on remote, incompletely remodeled fracture sites of the distal tibial & fibular diaphyses.* **(Right)** *AP radiograph in an 11-year-old girl with a deformity of her elbow (following a previously treated distal humeral supracondylar fracture) shows malunion with varus configuration to the distal humerus (cubitus varus).*



(Left) *Sagittal bone CT in a 13-year-old boy shows a triplane fracture. Follow-up frontal radiographs show poor definition of the central physis due to a physeal bridge of the distal left tibia. Note the normal open distal right tibial physis.* **(Right)** *Sagittal 3D SPGR FS MR (left) in the same patient shows a dark osseous bridge interrupting the bright cartilaginous physis. The axial MIP of the physis (right) allows calculation of the areas of the osseous bridge (outlined in orange) & total physis (outlined in yellow).*



TERMINOLOGY

Complications Encountered

- Nonunion: Lack of bony union > 6 months after injury with corresponding clinical signs/symptoms
- Delayed union: Lack of union by 6 months but with imaging &/or clinical signs of healing; eventually heals
- Malunion: Healing with nonanatomic alignment of fracture fragments
- Premature physeal closure/growth arrest: Osseous physeal bridge (bar) formation across otherwise unfused cartilaginous physis
- Osteonecrosis: Disruption of blood supply to fracture fragment with resulting infarction
- Chondrolysis: Progressive destruction of articular cartilage; may lead to premature degenerative arthritis
- Posttraumatic lipid inclusion cyst: Subperiosteal focus of marrow fat entrapped in remodeled cortex (due to transposition of fatty marrow during prior fracture)
- Others: Infection, degenerative changes, refracture

IMAGING

General Features

- Nonunion
  - Radiographic findings
    - Sclerosis around bone ends at fracture site
    - Absence of bridging trabecula with persistence of fracture line
    - Lack healing/callus progression on serial radiographs
      - Callus may not be seen normally with rigid fixation
  - Scaphoid nonunion
    - Humpback deformity
      - ↑ angulation between proximal & distal poles with settling/impaction of fragments & eventual dorsal bone formation
    - Carpal instability
      - ↑ capitolunate angle (normal 0-30°)
      - ↑ or ↓ scapholunate angle (normal 30-60°)
    - Late complications
      - Scaphoid nonunion advanced collapse: Late development of arthritis
      - Begins at radioscaphoid joint → scaphocapitate & capitolunate joints, sparing radiolunate joint
- Premature physeal closure
  - Peripheral bridge → angular deformity
    - Medial proximal femur → coxa vara
    - Anterior proximal tibia → genu recurvatum
    - Medial distal tibia → ankle varus
  - Central bridge → longitudinal growth restriction
    - Leg length discrepancy
    - Distal radial bridge → ulnar positive variance → ulnar abutment/impaction
  - Radiographs
    - May show bone bridge across physis (contralateral radiographs helpful)
      - Focal poor definition/rank interruption of lucent physis
    - Altered growth arrest/growth recovery line
      - Obliquely oriented or tethered growth recovery line extending to bony bridge at physis
      - Asymmetry of transverse growth recovery lines indicating focally diminished growth
    - Altered relative length of bone (as compared to adjacent long bone)
    - Increasing angulation of epiphysis
  - MR
    - Spin-echo T1 & PD/T2 FS sequences can demonstrate larger physeal bridges
      - Bridge may be bright on T1/dark on T2 FS if marrow present
    - 3D FSPGR or T2* GRE FS sequences (with multiplanar reformatted images) detect even small bridges & can generate axial MIP physeal map
      - Cartilaginous physis bright, bone bridge dark
      - Area of bridge/area of total physis = % of physis occupied by bridge (used to determine therapy)
- Posttraumatic cyst-like lesion
  - Radiographs: Small eccentric, round or ovoid lucent subperiosteal lesion, typically in distal radius or tibia
  - CT/MR: Fat characteristics
- Osteonecrosis
  - Scaphoid
    - Osteonecrosis in ~ 30% of mid body fractures & up to 100% of proximal pole fractures
    - Relative sclerosis of proximal pole on radiographs
    - STIR/T2 FS MR not helpful in determining viability (i.e., lack of necrosis) of proximal pole
    - T1 MR may be of value with regard to assessing viability
      - One recent study indicated diffusely ↓ T1 signal in proximal pole correlated with nonviability (sensitivity 55%, specificity 94%)
    - Other sources suggest T1 MR less helpful
      - Mummified fat may allow for ↑ T1 signal despite presence of osteonecrosis
      - ↓ T1 signal may be seen in ischemic but potentially viable bone
      - ↓ T1 signal may exaggerate necrosis because of granulation tissue
    - T1 C+ FS MR useful to assess viability of proximal fragment, especially with perfusion/subtraction techniques
      - One recent study showed that no contrast enhancement = no viability (sensitivity 76.5%, specificity 98.6%)
      - However, presence of contrast enhancement does not exclude presence of osteonecrosis
    - T1 C+ FS MR also helpful in predicting healing potential following surgical treatment & in evaluating healing response following vascularized bone graft
  - Distal humerus: Fishtail deformity
    - Osteonecrosis of distal humerus predominantly involving radial aspect of trochlea
    - Reported after fractures of various types (e.g., supracondylar, lateral condylar)

CLINICAL ISSUES

Nonunion

- Rare complication in pediatrics

- Most common in children: Diaphyseal long bone fractures & elbow (especially displaced lateral condyle) fractures
- Scaphoid nonunions rare in children
- Open fractures, infection, severe soft tissue loss, & insufficient fixation contribute to nonunion development
- Treatment options: Debridement of nonunion site, bone grafting, electrical stimulation, & rigid internal fixation
  - Determined on case-by-case basis

Malunion

- Common in children but with high corrective potential due to ↑ chance for remodeling in growing skeleton
- Recommendations regarding maximum acceptable displacement at fracture sites in children controversial & depend on age, location, & type of displacement
- Subsequent functional limitation & cosmetic deformity depend on site of fracture
  - Forearm shaft & distal radial fractures have great capacity for remodeling
  - Supracondylar fractures have less potential for remodeling
- Common sites of malunion
  - Supracondylar fracture
    - Cubitus varus deformity with medial angulation of distal fragment, not growth disturbance
    - Baumann (humerocapitellar) angle used to predict final carrying angle following reduction of supracondylar fractures to attempt to avoid cubitus varus deformities
      - Angle formed by line along long axis of humeral shaft & line along physis of capitellum
      - Normal range: 64-81°
  - Forearm fracture
    - May cause limited pronation/supination, ↑ rate of refracture, cosmetic deformity, or distal radioulnar joint pain
    - Acceptable angulation controversial
      - Some authors: > 10° in children > 8 years old = malunion
      - Other definitions: > 20° in children > 9 years old = malunion
  - Distal radius fracture
    - Usually remodel satisfactorily if < 11-12 years of age
    - Malunion may result in functional limitation, pain, or cosmetic deformity
    - Features appear similar to adults prior to remodeling
      - ↓ radial inclination; abnormal dorsal tilt, radial shortening, articular incongruity
    - Adolescents & young adults can still develop significant distal radius malunion
      - One source suggests malunion: Dorsal tilt ≥ 5°, radial inclination ≤ 10°, loss of radial height ≥ 5 mm
      - Normal values: Volar tilt (11°, range: 2-20°), radial inclination (21-25°), radial length (10-13 mm)
  - Femur fracture (definitions vary)
    - Some contend > 10° coronal plane & > 15° sagittal plane; others suggest > 5° & 10°, respectively
- Treatment may consist of osteotomy & epiphysiodesis

Premature Physeal Closure

- Secondary to fracture involving cartilaginous growth plate
- Bridges start to form 1-2 months following injury but may not manifest until years later
  - Clinical follow-up to skeletal maturity recommended in high-risk fractures due to possibility of delayed presentation during adolescent growth spurt
  - If caught early, only resection of bar may be necessary vs. correction of angular deformity
- Treatment
  - Indicated when deformity present/developing & patient has 2 years or ≥ 2 cm of growth remaining
  - May include corrective osteotomy, completion of epiphysiodesis (± contralateral extremity), lengthening of involved bone, surgical excision of physeal bridge + insertion of interposition material, or combination
  - Bridge typically not resected if > 50% of physis involved

Posttraumatic Cystic Lesions

- Seen at sites of prior fracture; thought to be due to extension of intramedullary fat through disrupted cortex with intact periosteum
- May mimic more aggressive process
- Distal radius most common, followed by distal tibia

Degenerative Change

- Intraarticular fractures at risk for posttraumatic arthrosis
- Fixation typically necessary to reestablish articular congruence if articular surface disruption > 2 mm

Osteonecrosis

- Proximal pole of scaphoid: Sclerosis & humpback deformity
- Distal humerus: Fishtail deformity

DIAGNOSTIC CHECKLIST

Reporting Tips

- Describe residual deformity; however, radiographic appearance may not correlate with clinical outcome
  - Malunion may not be clinically relevant (e.g., patient with worrisome radiograph may have no functional limitation &/or may completely remodel over time)
- Lack of fracture healing may be suggestive of nonunion if > 6 months but must be correlated with clinical findings
  - From medicolegal standpoint, best to describe & discuss with orthopedist, avoiding term "nonunion" in report

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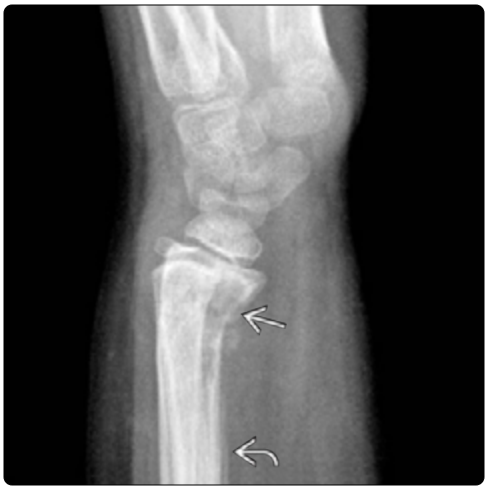
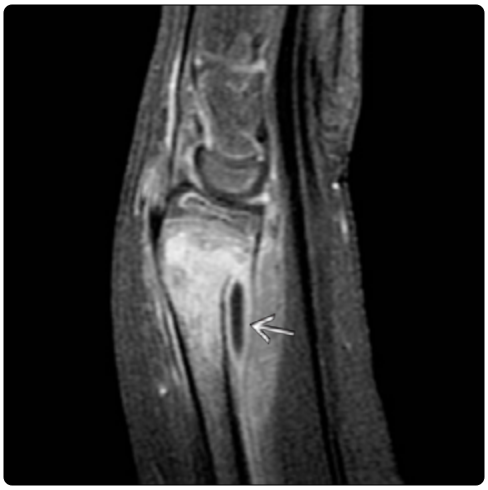
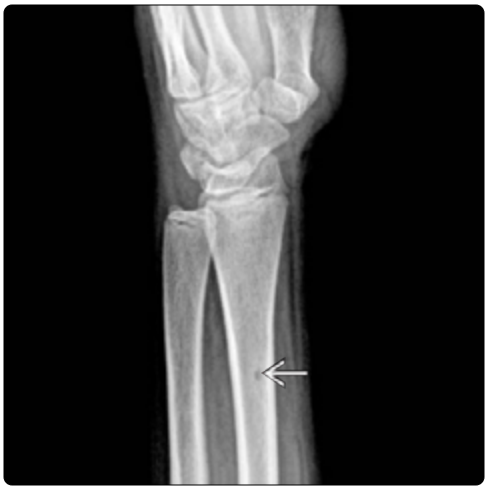
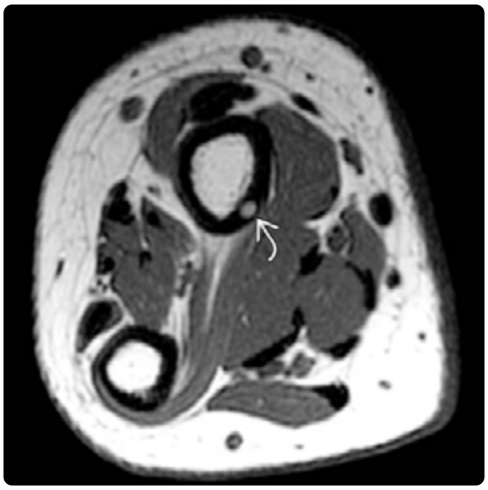


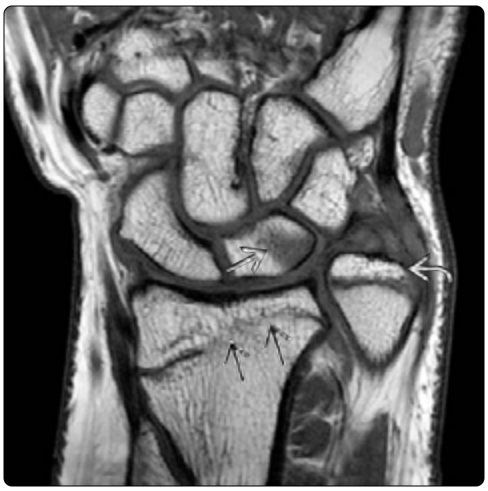


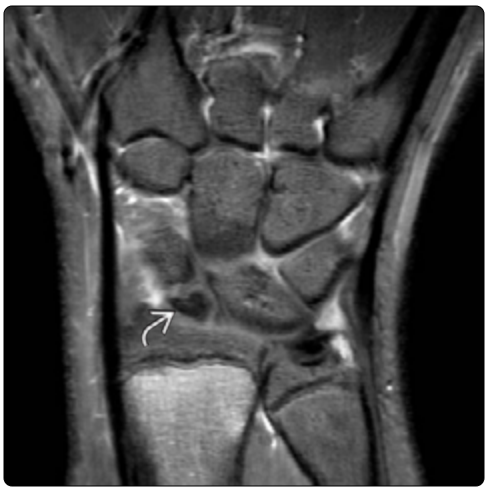
(Left) *Lateral wrist radiograph (obtained after cast removal) in a 6-year-old girl recently diagnosed with a Salter-Harris fracture shows an aggressive lytic process at the distal radius with cortical bone destruction ➡ & periosteal reaction ➡.* **(Right)** *Sagittal T1 C+ FS MR in the same patient shows a peripherally enhancing subperiosteal fluid collection ➡. Pus was found at surgery, & the patient was diagnosed with Staphylococcus aureus osteomyelitis.*





(Left) *Oblique radiograph of the distal forearm in a 14-year-old girl with a history of a fracture of the distal radial metadiaphysis (2 years prior to presentation) shows a small ovoid lucent lesion ➡. A true lateral radiograph was not obtained.* **(Right)** *Axial T1 MR in the same patient shows an eccentrically located ovoid lesion in the volar distal radial cortex with increased T1 signal intensity ➡. This followed the signal of fat on all sequences, compatible with a small lipid inclusion "cyst" related to the prior fracture.*





(Left) *Coronal T1 MR from a 14-year-old boy following a Salter-Harris II fracture of the distal radius shows bridging trabecular bone across the central growth plate of the distal radius, compatible with a physeal bar ⇨. There is associated ulnar positive variance ➡, resulting in abutment & reactive marrow change in the lunate ➡.* **(Right)** *Coronal T1 FS C+ MR in a 14-year-old girl with a scaphoid fracture shows a diffuse lack of enhancement ➡ within the proximal pole of the scaphoid, suggesting nonviability.*

KEY FACTS

TERMINOLOGY

- Orthopedic hardware: Any of innumerable devices used for fracture fixation, realignment, ligament repair/reconstruction, arthroplasty, & other procedures

IMAGING

- External vs. internal fixation
- Goal of rigid fixation: Reduce motion & callus formation, particularly at intraarticular sites
  - Look for endosteal rather than periosteal callus to indicate appropriate healing
    - Periosteal callus indicates motion at fracture site
- Radiographs remain primary evaluation of hardware
  - Malpositioning/migration or fracture of hardware
  - Fracture of bone at/adjacent to fixation site
  - Loosening or infection of hardware
  - Malunion, delayed union, or nonunion of fracture
- CT more sensitive; used in select cases of clinical/imaging concern
- Comparing nuclear In-111 leukocyte & Tc-99m sulfur colloid exams is most specific evaluation of infected hardware

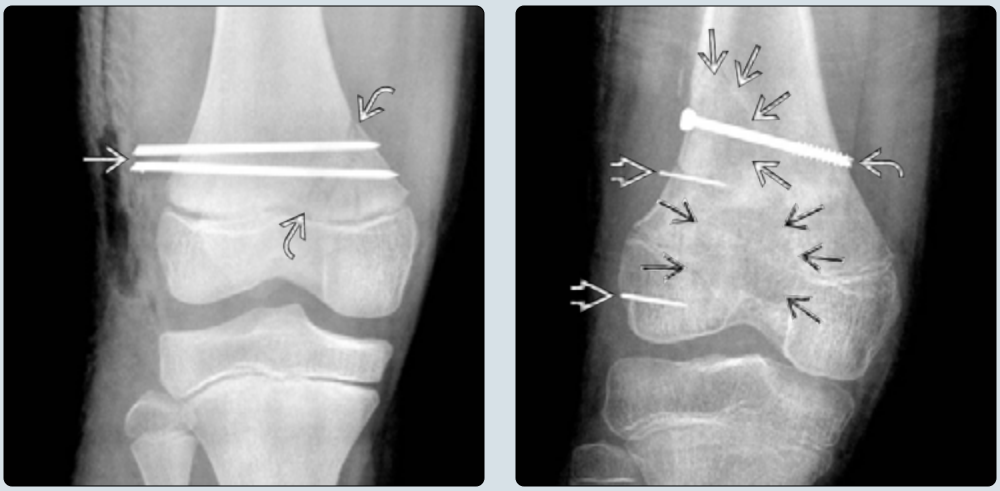
CLINICAL ISSUES

- Goal of fracture treatment is to improve alignment & stabilize fracture to aid in healing & return function
- Numerous indications for orthopedic devices in children other than fracture fixation
  - Scoliosis, congenital deformity, growth disturbance, sports-related injuries

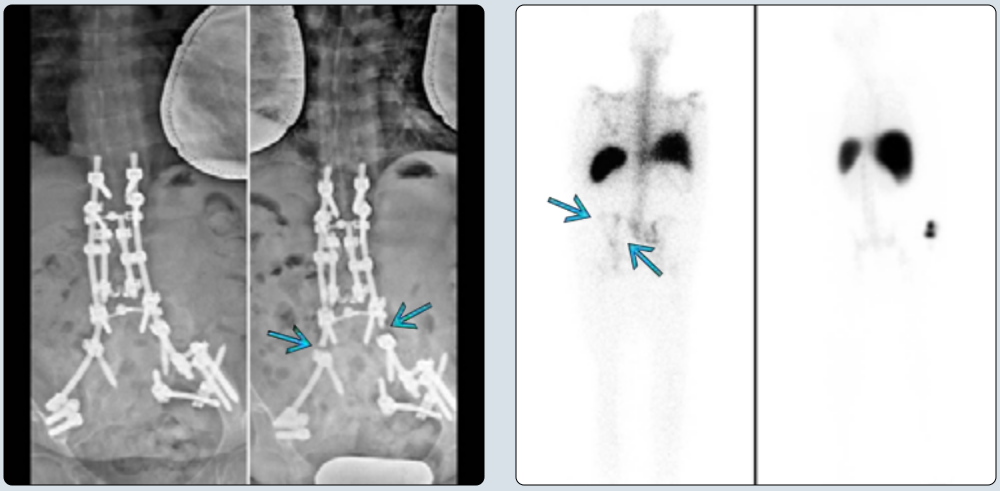
DIAGNOSTIC CHECKLIST

- Understanding purpose/function of orthopedic devices aids in evaluation of
  - Appropriate positioning of hardware
  - Hardware complications
  - Expected healing

(Left) *AP radiograph of a 12-year-old boy shows 2 fully threaded Steinmann pins transfixing a Salter-Harris II fracture.* **(Right)** *AP radiograph shows the same patient ~ 8 months later following a physeal bar resection. The bone block from the distal femoral window osteotomy is fixed with a single partially threaded cannulated screw. Steinmann pins were placed to evaluate for subsequent growth. Cranioplast material is placed across the physis to prevent new bar formation.*



(Left) *Initial (L) & follow-up (R) frontal radiographs in a boy with neurofibromatosis type 1 & posterior spinal fusion hardware for kyphoscoliosis show interval fractures of the hardware. The patient had fever at this time.* **(Right)** *Posterior images from In-111 leukocyte (L) & Tc-99m sulfur colloid (R) scans were obtained in the same patient. There is increased uptake in the right sacrum & iliac bones on the leukocyte scan with no corresponding increased uptake on the sulfur colloid scan. This discordance is consistent with infection.*



IMAGING

Radiographic Findings

- Radiographs: Primary modality for hardware assessment
- Evaluate for
  - Malpositioning/migration of hardware
  - Fracture of hardware components
  - Fracture of bone at or adjacent to fixation site
  - Loosening: Progressive lucency around hardware
  - Infection: Perihardware lucency, extensive periosteal reaction, erosions, subcutaneous emphysema
  - Malunion, delayed union, or nonunion of fracture
  - Osteonecrosis: Interruption of periosteal blood supply
  - Heterotopic bone formation

Fluoroscopic Findings

- Intraoperatively guides/confirms hardware placement

CT Findings

- More sensitive evaluation for hardware complications
- Modification of CT protocols to minimize artifacts
  - ↓ pitch setting, ↑ mAs, ↑ kVp (↑ radiation dose)
  - Dual-energy CT (no additional radiation)
  - Soft-tissue image reconstruction filters

MR Findings

- MR useful in certain postoperative situations
  - Ligament repair/reconstruction
  - Assessment of soft tissue abscesses
- Limited by metallic artifact
- Utilize techniques to ↓ artifacts

Ultrasonographic Findings

- Can be used to evaluate for fluid collections/abscesses

Nuclear Medicine Findings

- Bone scan
  - Normal bone scan can exclude hardware infection
  - ↑ radiotracer activity may be due to infection or loosening
- Labeled leukocyte scintigraphy
  - ↑ radiotracer activity on leukocyte exam
    - Osteomyelitis
    - Marrow adjacent to fracture or hardware
  - Compare leukocyte & Tc-99m sulfur colloid exams
    - ↑ activity on labeled leukocyte exam but normal sulfur colloid exam = infection
    - Any other pattern is negative for infection

PATHOLOGY

General Features

- Fractures heal by primary or secondary mechanisms
  - Primary (direct) healing
    - Direct bone healing without callus formation
    - Occurs with rigid fracture fixation
  - Secondary (indirect) healing
    - Healing via callus formation
    - Occurs with flexible fracture fixation

CLINICAL ISSUES

Treatment

- Conservative treatment
  - Closed reduction with goal of restoring bone alignment
  - Stabilization via traction or external splinting (i.e., slings, splints, casts)
- Fracture fixation
  - External or internal fixation
  - Flexible versus rigid fixation
    - Flexible: Allows interfragmentary movement under functional load
    - Rigid: Utilizes compression → ↑ stability; ↓ motion → primary healing without callus
      - Important at intraarticular sites where callus formation & incongruity → osteoarthritis
- External or internal fixation also used for nontraumatic purposes (i.e., leg lengthening, corrective osteotomy, etc.)
- External fixation advantages
  - Surgeon can control flexibility of fixation
  - ↓ injury to soft tissues, vasculature, & periosteum
- External fixation indications
  - Avoidance of growth plates in skeletally immature patients
  - Osteomyelitis (internal hardware avoided)
  - Substantial soft tissue injury requiring vascular procedures
  - Limb lengthening & other corrective osteotomies
- External fixation complications
  - Pin tract infection, breakage, or loosening
  - Delayed union, nonunion, malunion
- Internal fixation advantages
  - Potential for rapid return of function & rehabilitation
- Internal fixation indications & complications (categorized by different types of hardware)
  - Pin (Kirschner wires & Steinman pins) indications
    - Temporary fixation of fragments during reduction
    - Attachment of skeletal traction devices
    - Guide accurate placement of larger cannulated screws
    - Occasionally used for definitive fracture treatment
    - Also used in setting of external fixation
  - Pin complications
    - Migration when used for definitive fracture treatment
  - Wire indications
    - Reattach osteotomized bone fragments
    - Tension banding (in combination with pins or screws, e.g., patella, olecranon fractures)
      - Applied following certain fractures or osteotomies to convert tensile forces from adjacent tendinous insertions into compressive forces
      - Aids with stability & fracture healing
    - Suture bone & soft tissue
    - Cerclage wires (used with intramedullary fixation to stabilize long bone fragments)
  - Wire complications
    - Breakage (insignificant if fragments maintain position)
    - Interruption of periosteal blood supply with subsequent osteonecrosis or fracture nonunion
  - Screws: Types & indications
    - 2 basic types: Cortical & cancellous

        - Distance between threads = pitch
        - Screw diameter with threads = thread diameter
        - Core = diameter of screw without threads
    - Cortical screws
        - Designed for use in diaphysis
        - Typically fully threaded (threads along entire length) with smaller thread diameter & pitch
    - Cancellous screws
        - Can cross long segments of cancellous bone
        - Typically deeper threads, larger thread diameter, greater pitch, partially threaded
    - Interfragmentary screw
        - Screw that crosses fracture line
        - Purpose: Act as lag screw providing compression of fragments → ↑ stability & healing
    - Cannulated screw
        - Possesses hollow shank, so can be placed over guide pin for ↑ accuracy
        - May be inserted percutaneously under fluoroscopy
        - Used with fracture table, which provides traction & maintains reduction during fixation
    - Suture anchor
        - Anchor can be fixed into bone by screwing or lodging (e.g., press fit)
        - Used for capsular, ligamentous, or tendinous repair
    - Washer
        - Prevents screw head from sinking into bone
        - Enhances compressive area of screw in regions of thin cortex
        - Prevent fractures under screw
- Screw complications
    - Breakage, loosening, change in position
- Plates
    - Techniques for plating
        - Compression plating: Eccentric screw placement within sloped holes drives fragments into compression
        - Neutralization plating: Holds fracture fragments in place without compression; useful for severely comminuted fractures (bridging plate) & fractures with bone loss
        - Tension band plating: Placed along tension side of fracture; replaces normal tensile with compressive forces, allowing for healing
        - Buttress plating: Often used in association with metaphyseal/epiphyseal shear or split fractures; prevents shear forces across fracture from displacing fragments
    - Dynamic compression plate: Can be used in compression, neutralization, tension band, or buttress
    - Additional plates
        - Tubular plate, reconstruction plate (malleable; used for pelvic fractures), less invasive stabilization system (LISS) plate, T-plate, blade plate, other special anatomically shaped plates
    - Locking plates may contain locking screws that contain threads at screw heads, allowing construct to better act as unit
- Plate complications
    - Plates have large contact area, may disrupt periosteal capillaries → compromise cortical perfusion
    - Underlying bone resorption & old screw holes after plate removal may lead to fracture
    - Malposition, i.e., plate or screws violating articular surface or impinging upon joint motion
- Intramedullary nails or rods
    - Standard treatment for diaphyseal fractures of femur & tibia; may also be used in humerus
    - Rods locked "statically" (proximal & distal interlocking screws) or "dynamically" (fixed only at 1 end)
        - Dynamic locking ↑ compression across fracture site
        - Interlocking screws ↑ fixation stability & prevent rotation
    - Skeletally immature patients receive flexible intramedullary nails inserted through metaphysis
        - Typically 2 rods placed through multiple insertion sites diverging at metaphyseal ends
        - Require additional external stabilization with cast
- Intramedullary nail or rod complications
    - Hardware fracture, loosening, or infection
    - Violation of joint by rods or screws
    - Intramedullary reaming is associated with ↑ infection rates (due to damage to internal cortical vasculature) & pulmonary fat embolism
- Antibiotic beads
    - Used for infected fractures
- Autogenous bone graft, allograft, bone graft substitute
    - Treatment of bone defects

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- On initial preoperative studies: Carefully evaluate fractured bone for any sign of underlying aggressive lesion
    - Placement of hardware may spread tumor cells
- On fluoroscopic intraoperative images: Carefully assess for hardware malposition or iatrogenic fracture
- On follow-up postoperative studies: Carefully evaluate full complement of hardware (& adjacent bone) for interval change compared with prior studies

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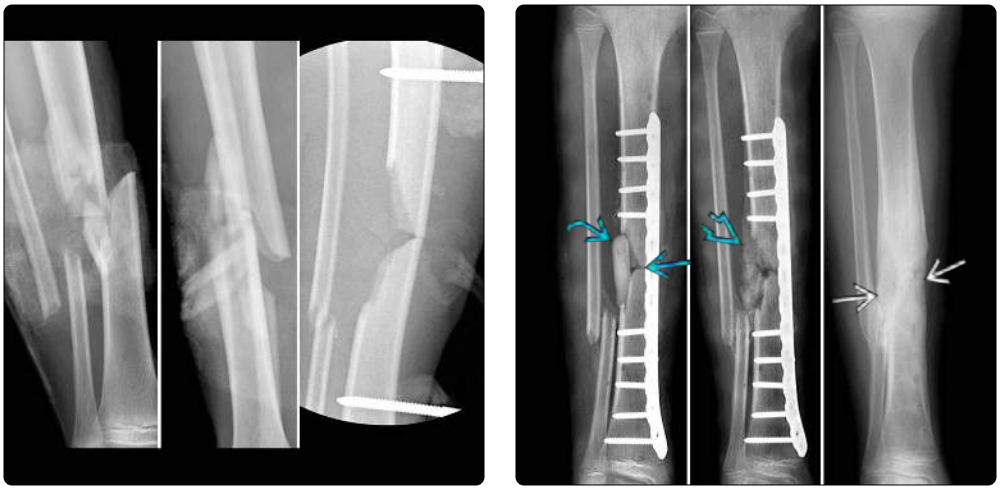
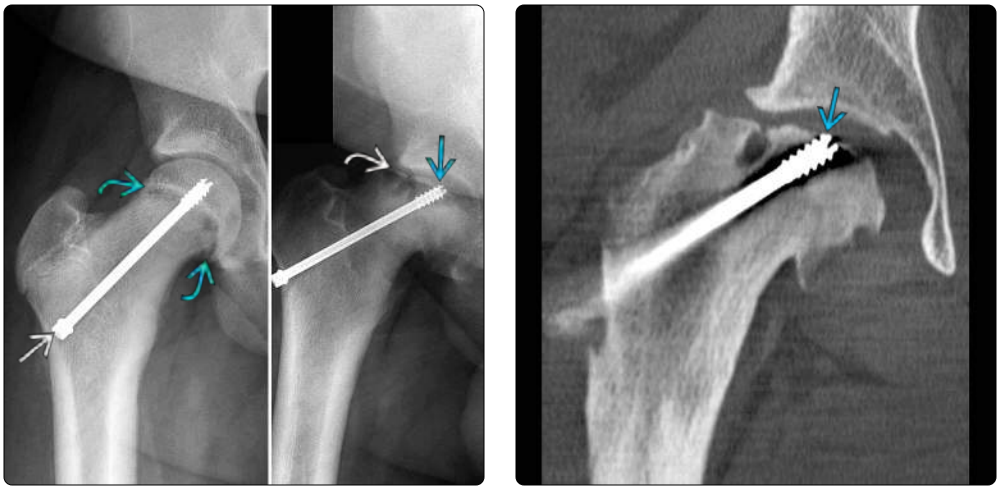
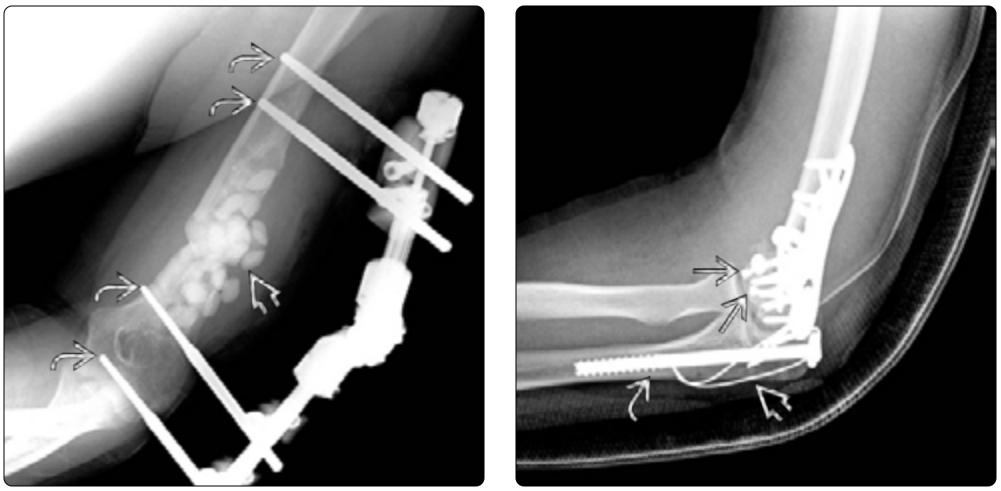


(Left) *Initial frontal (L) & lateral (M) radiographs in a boy show an open, comminuted tibial fracture. The initial treatment was with external fixation (R) due to the risk of infection.* **(Right)** *Radiograph in the same boy 7 months later (L) shows internal plate/screw fixation, placement of an antibiotic impregnated spacer ⇨, & nonunion of the tibial fracture ⇨. Follow-up image (M) after debridement & placement of bone graft ⇨ leads to incorporation & bridging callus at the fracture ➡ 1 year later (R).*



(Left) *Initial postop image (L) of a 12-year-old boy with a slipped capital femoral epiphysis (SCFE) ⇨ shows fixation with a transphyseal partially threaded cannulated screw ➡. Follow-up (R) shows collapse of the femoral head ➡ from avascular necrosis, a known complication of SCFE. The screw extends beyond the articular surface ⇨.* **(Right)** *Coronal bone CT in the same patient confirms that the screw ⇨ extends across the articular surface of the collapsed femoral head.*



(Left) *AP radiograph of the left humerus in a patient with an infected nonunion following an open fracture shows antibiotic beads ➡ as well as an external fixator with 2 half-pins ➡ proximal & distal to the fracture site.* **(Right)** *Lateral radiograph shows a partially threaded cannulated screw ➡ with washer & tension band ➡ across an olecranon osteotomy site created for reduction of a capitellar shear fracture. Two headless compression screws ➡ are seen in the capitellum.*

Soft Tissue Foreign Bodies, Acute and Chronic

KEY FACTS

TERMINOLOGY

- Penetrating injury → soft tissue foreign body (FB)
- Chronic FB → granulomatous reaction → soft tissue mass

IMAGING

- Most FBs are not radiopaque (e.g., wood splinters)
- Some FBs are radiopaque (e.g., metal, glass, bone)
- Radiographs assess radiopaque FBs & osseous changes
- US: Excellent detection of superficial FBs
  - Typically echogenic
  - ± posterior shadowing depending on composition
  - Hypoechoic rim of edema/granulomatous reaction
- MR: FB typically of low signal on all sequences
  - Nonanatomic, geographic shape (e.g., linear, triangular)
  - FB may be small or very subtle
  - GRE may show "blooming" if metallic or Ca^{2+}
  - Adjacent edema or granulomatous reaction
  - Detects associated cellulitis, abscess, osteomyelitis

TOP DIFFERENTIAL DIAGNOSES

- Posttraumatic fat necrosis
- Soft tissue sarcoma
- Benign soft tissue tumors
- Venous malformation

PATHOLOGY

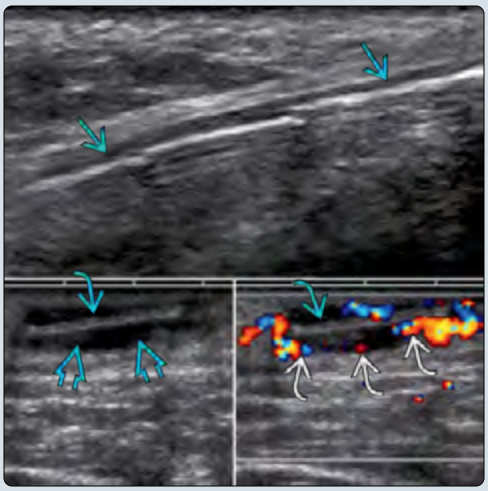
- Chronic FB → granulomatous reaction

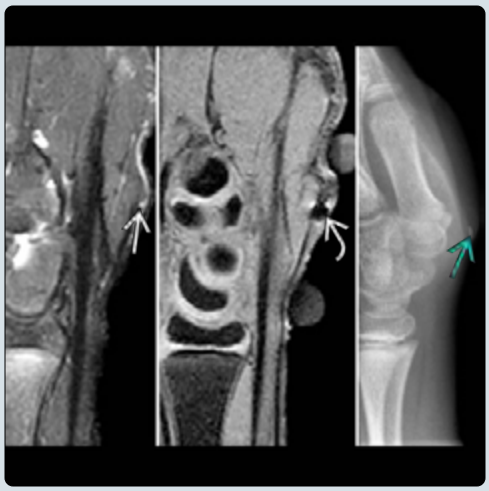
CLINICAL ISSUES

- Acute: Sensation of FB under skin after injury
- Chronic: Firm painless soft tissue mass
  - May occur long after injury that introduced FB
  - Often no specific trauma recalled
- Treatment
  - Surgical or US-guided removal

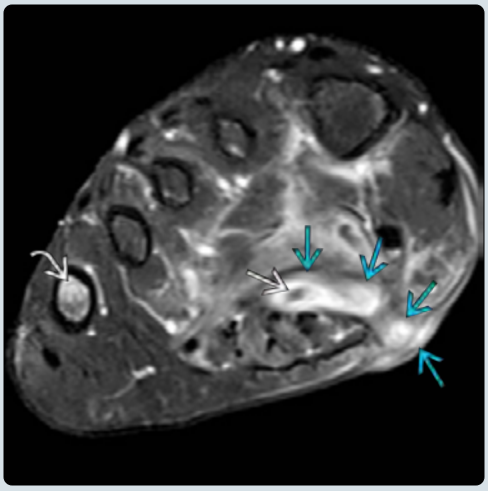
DIAGNOSTIC CHECKLIST

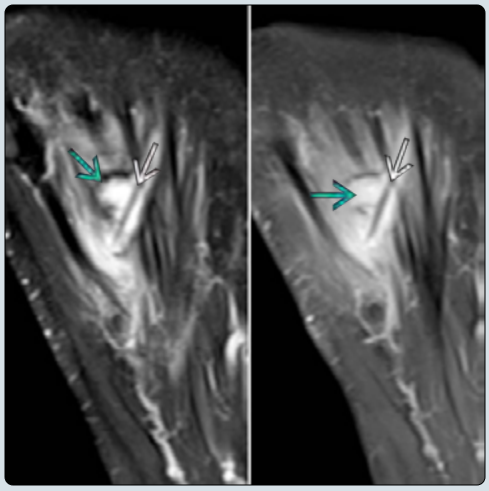
- If radiolucent superficial FB suspected → US





(Left) *Initial US (top) of the left buttock in a 5-year-old boy who fell on a plastic toy shows a linear subcutaneous echogenic foreign body (FB). Removal was performed without imaging guidance. US 6 weeks later (bottom) shows a retained portion of the FB with adjacent hypoechoic but hypervascular granulation tissue.* **(Right)** *Sagittal MRs in a 10-year-old girl with a palpable lump show a focus of low signal on T2 FS (left) that "blooms" on GRE (middle). A lateral radiograph confirmed a FB (right).*





(Left) *Axial T2 FS in a 9 year old who removed a toothpick from her foot 5 months prior shows a linear tract of increased signal extending from the plantar surface to a central focus of low signal, concerning for a retained FB. There is adjacent soft tissue edema & distant marrow edema in the 5th metatarsal from infection vs. stress changes.* **(Right)** *Coronal long-axis T2 FS (left) & T1 C+ FS (right) MRs in the same patient show the linear low-signal retained toothpick surrounded by high-signal granulation tissue.*

TERMINOLOGY

Abbreviations

- Foreign body (FB)

Synonyms

- Chronic FB: FB granuloma, giant cell granulomatous reaction, fibrohistiocytic reaction

Definitions

- Penetrating injury leads to soft tissue FB deposition
  - Injury & retention of FB may not be recognized/recalled
- Chronic FB may lead to mass of granulomatous reaction

IMAGING

General Features

- Best diagnostic clue
  - Radiopaque FBs identified on radiographs
  - Radiolucent FBs may be identified on US, MR, or CT
- Location
  - Often confined to subcutaneous fat
  - Typically in areas prone to injury
    - Plantar aspect of feet (e.g., walking barefoot)
    - Areas commonly injured from fall (e.g., anterior knee/shin, hands, elbows, buttocks)

Radiographic Findings

- Radiopacity depends upon FB composition
  - Conspicuity depends on size & degree of difference in radiodensity from surrounding soft tissues
- Most FBs not radiopaque (e.g., wood splinters, thorns, plastic)
  - May only show nonspecific soft tissue fullness
- Some FBs are radiopaque (e.g., metal, glass, bone)
- ± findings of osteomyelitis in subacute/chronic setting

MR Findings

- FB: Typically low signal intensity on all sequences
  - Nonanatomic, geographic shape (e.g., linear, square)
  - Low signal intensity may be small or very subtle
  - GRE sequence can be helpful depending on composition
    - "Blooming" artifact if metal or bone
- Acute FB: Poorly defined edema in adjacent soft tissues
  - Reticular ↑ signal on T2WI FS/STIR, ↓ signal T1WI
  - Reticular enhancement on T1WI C+ FS
- Chronic FB: May lead to granulomatous reaction
  - Mass-like ↑ signal on T2WI FS/STIR, ↓ signal T1WI
  - Diffuse enhancement on T1WI C+ FS
- Associated findings
  - Tract from skin surface → linear focus of fluid signal
  - Soft tissue abscess: Localized fluid collection with peripheral enhancement (vs. diffuse enhancement of FB granuloma)
  - Marrow edema/hyperenhancement of adjacent bone may be seen with osteomyelitis or stress changes (due to altered weight-bearing in setting of pain)

Ultrasonographic Findings

- Excellent for identifying superficial acute & chronic FBs
- FBs typically echogenic, often punctate or linear
- Variable posterior shadowing depending on composition
- Peripheral ↓ echogenicity from edema (acute) or granulomatous reaction (chronic)
- Exam may be confusing if US only performed after attempted FB removal

Imaging Recommendations

- Best imaging tool
  - Radiography for radiopaque FBs (e.g., glass, metal, bone)
    - Also shows bone involvement by penetrating trauma
  - US for suspected radiolucent superficial FB

DIFFERENTIAL DIAGNOSIS

Posttraumatic Fat Necrosis

- Injury of subcutaneous fat with associated hematoma
- No central FB seen at imaging

Soft Tissue Sarcomas

- More common in areas not prone to foreign bodies (proximal to mid extremities)
- No central FB seen at imaging
- Typically well circumscribed without surrounding edema

Benign Soft Tissue Tumors

- Variety of nonmalignant soft tissue tumors occur in children
  - e.g., fibrous lesions (fibromatosis, nodular fasciitis), subcutaneous granuloma annulare
- No central FB seen at imaging
- Can be well circumscribed or poorly defined & infiltrative

Venous Malformations

- Serpentine tangle or mass of abnormal veins
- ↑ signal on T2WI FS + contrast enhancement
- ± phleboliths, fluid-fluid levels

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute: Sensation of FB under skin after injury
  - Chronic: Firm painless soft tissue mass
    - May occur long after injury that introduced FB
    - Often no specific trauma recalled
- Other signs/symptoms
  - Erythema, swelling, &/or induration of overlying skin
  - Sinus tract to skin surface may develop

Treatment

- Surgical removal: US or fluoroscopy can localize FB
- Alternatively, US-guided removal with needle or forceps
  - Hydrodissection technique may assist procedure

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Morel-Lavallée Lesion

KEY FACTS

TERMINOLOGY

- Closed degloving injury of deep subcutaneous fat at interface with fascia, often overlying bony protuberance
- Results in chronic fluid collection (blood, lymph, fat)

IMAGING

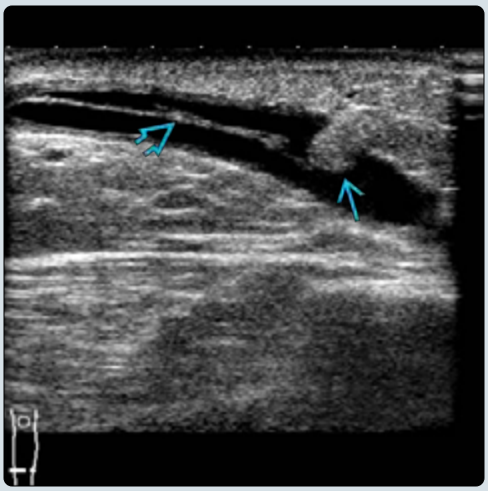
- Classic Morel-Lavallée lesion occurs at hip/proximal thigh over greater trochanter
  - Other sites: Abdominal wall, lumbar spine, buttock, sacrum, thigh, knee, lower leg, scapula, scalp
- Mean size: 8 cm; range: 3-17 cm
- Appearance depends on timing of imaging & complications
  - Acute/subacute: Elongated with irregular margins & internal heterogeneity
  - Chronic: Elongated & smooth; ↓ heterogeneity
  - Angular margins at periphery of shear plane suggestive
- MR: Collection ultimately follows fluid signal intensity internally except for fat nodules, blood products
  - Fibrous pseudocapsule hypointense on all sequences
  - Nodular enhancement, typically peripheral, may be seen with granulation tissue, inflammation, or infection
- Ultrasound: Ultimately hypoechoic to anechoic internally (due to ↓ complexity over time)
  - Echogenic fat nodules characteristic

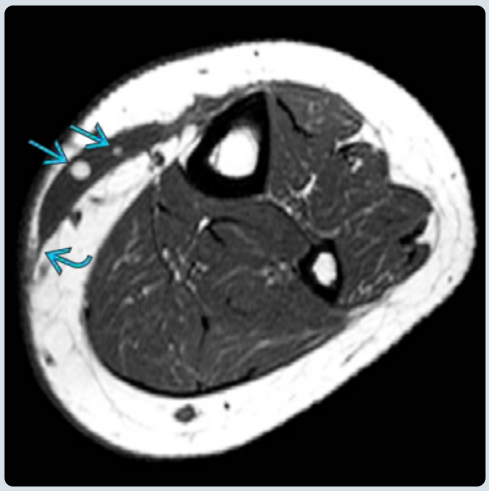
PATHOLOGY

- Blunt trauma with compressive & shear forces creates potential space with disruption of vessels & nerves

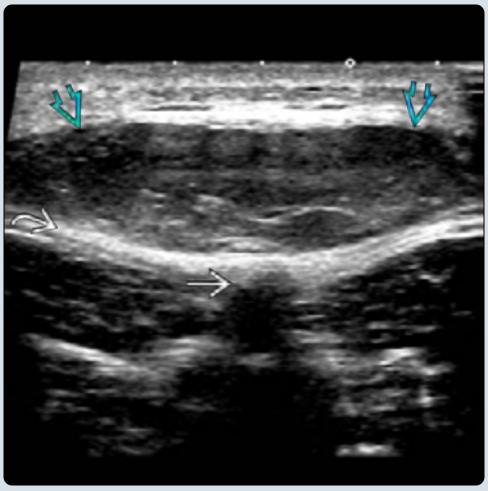
CLINICAL ISSUES

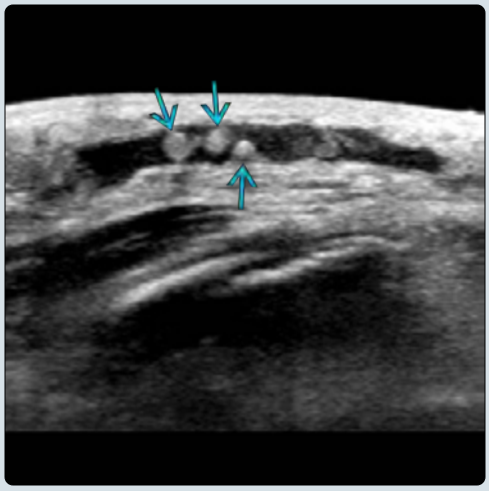
- Fluctuant mass, bruising, ↑ skin mobility, ↓ skin sensation
  - Delayed presentation after injury in 1/3 of patients
- Variable course: Resolution, persistence, enlargement
  - Risk for infection: Up to 50%
- Treatment
  - Conservative: Compression wraps, rest, physical therapy
  - Intervention: Percutaneous drainage ± steroid injection or sclerodesis; surgical debridement or excision





(Left) *Transverse ultrasound of the anteromedial lower leg in a 16-year-old soccer player with a recent injury & swelling shows an elongated, hypoechoic collection of the subcutaneous fat containing an echogenic nodule & internal septation.* **(Right)** *Axial T1 MR of the same patient shows that the nodules in the collection are of similar signal intensity to the subcutaneous fat (& followed fat signal on all sequences). A Morel-Lavallée type injury was diagnosed. Note the acute angle at the margin of the shear plane.*





(Left) *Transverse ultrasound of a 16-year-old boy with a fall onto his back 1 week prior shows an elongated, heterogeneous collection overlying the lower lumbar spine. Note the echogenic subjacent fascia. Internal swirling of debris was noted with compression.* **(Right)** *Transverse ultrasound over the right iliac crest in a 15-year-old boy with a recent fall from a bicycle shows an elongated, hypoechoic subcutaneous collection containing echogenic nodules, typical of a Morel-Lavallée lesion.*

KEY FACTS

TERMINOLOGY

- Fascial defect resulting in focal muscle protrusion, usually at interface of superficial fascia with subcutaneous fat

IMAGING

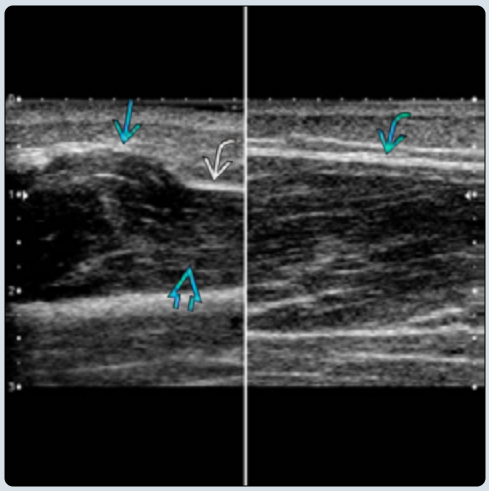
- Lower > upper extremity
  - Most commonly affected muscle: Anterior tibialis
- MR
  - Bulging nodule isointense to muscle on all sequences
- Ultrasound
  - Focal interruption of thin, linear hyperechoic fascia overlying hypoechoic, striated muscle
  - Focal bulge of hypoechoic muscle through fascial defect, creating "mushroom cap" appearance
  - Hernia accentuated by contraction or vigorous activity, reduced with relaxation or ↑ transducer pressure
    - Ankle dorsiflexion ↑ anterior tibialis muscle herniation
- Protocol advice (to optimize hernia visualization)
  - Use variety of provocative maneuvers
  - Reduce pressure applied by ultrasound transducer or MR skin marker (capsule) over site of clinical concern
  - Compare with contralateral extremity

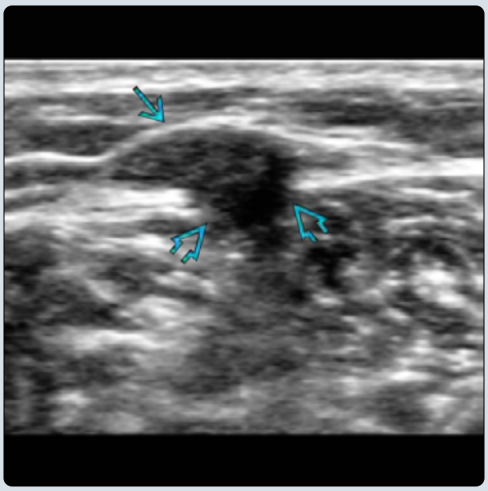
PATHOLOGY

- Congenital or constitutional fascial hernia
  - Thin fascia, focal weakness at nerve/vessel perforation
- Acquired or traumatic fascial tear/rupture
  - Penetrating injury, closed fracture, excessive contraction
- Predisposing conditions
  - Chronically increased compartmental pressure
  - Surgical manipulation of muscle/tendon (such as flap creation or tendon harvest)

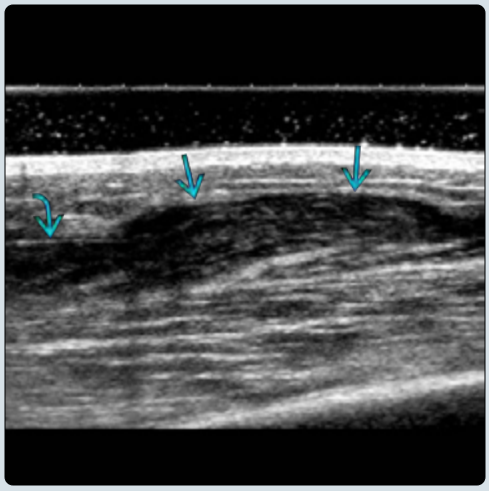
CLINICAL ISSUES

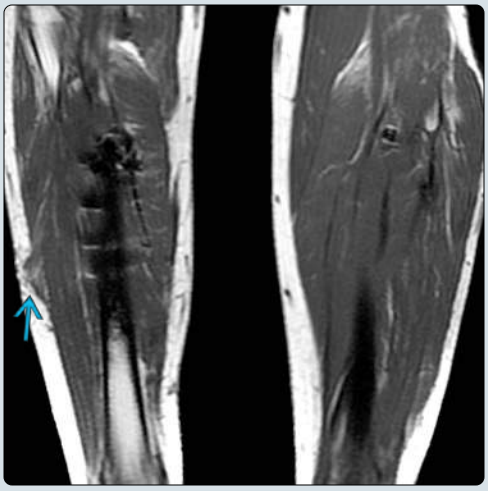
- Asymptomatic vs. intermittent painless bulge/nodule vs. painful nodule appearing after vigorous exertion
- Surgery for symptomatic cases not responding to conservative measures





(Left) *Longitudinal ultrasound of the right lower leg in an 11-year-old girl with a palpable lump shows herniation of the anterior tibialis muscle through a fascial defect. Note the thin, linear, echogenic appearance of intact fascia distally on the right & in the contralateral left leg.* **(Right)** *Transverse ultrasound through the right lower leg in a 17-year-old girl shows herniation of muscle through a fascial defect, creating a "mushroom cap" appearance. The hernia intermittently reduced with relaxation.*





(Left) *Longitudinal ultrasound through the lower leg in a 16-year-old girl shows a focal muscle herniation through the superficial fascia.* **(Right)** *Coronal T1 MR in an 18-year-old woman with a history of surgery for a right tibial osteosarcoma shows a focal herniation of peroneal muscle through a fascial defect.*

Supracondylar Fracture

KEY FACTS

IMAGING

- AP radiograph
  - Transverse fracture through distal humeral metadiaphyseal junction at level of coronoid/olecranon fossae
  - ± medial or lateral cortical buckling or angulation
- Lateral radiograph
  - Visible posterior fat pad due to joint effusion
    - May be only finding of nondisplaced fracture
  - Anterior humeral line fails to bisect capitellum due to dorsal angulation of distal fracture fragment
- Radiocapitellar alignment generally maintained on all views (in contradistinction to true dislocation)

TOP DIFFERENTIAL DIAGNOSES

- Lateral condylar fracture
- Posterior dislocation
- Distal humeral epiphyseal separation/transphyseal fracture

PATHOLOGY

- Extension/FOOSH (**f**all **o**n **o**ut**s**tretched **h**and) mechanism (98%) vs. flexion injury with direct trauma (2%)
- Modified Gartland classification (I-IV) to direct treatment
  - Classified by degree of displacement, rotation, instability
  - Treatment ranges from casting to closed reduction with percutaneous fixation to open reduction internal fixation
- Neurovascular injury much higher with completely displaced fractures (types III & IV)
  - Vascular injury requires urgent reduction + pinning at minimum; may need ORIF

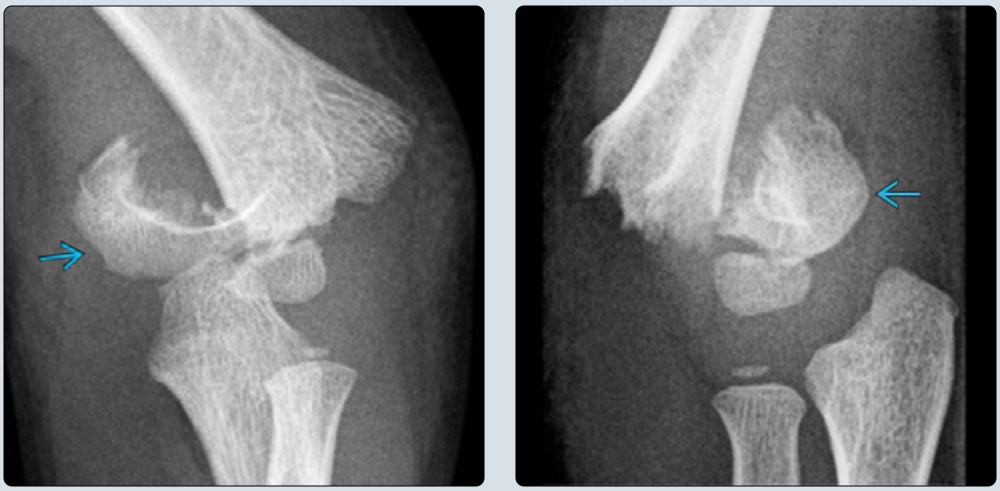
CLINICAL ISSUES

- Most common in children 5-7 years old; rare in adults (< 3%)
- Accounts for 60% of pediatric elbow fractures

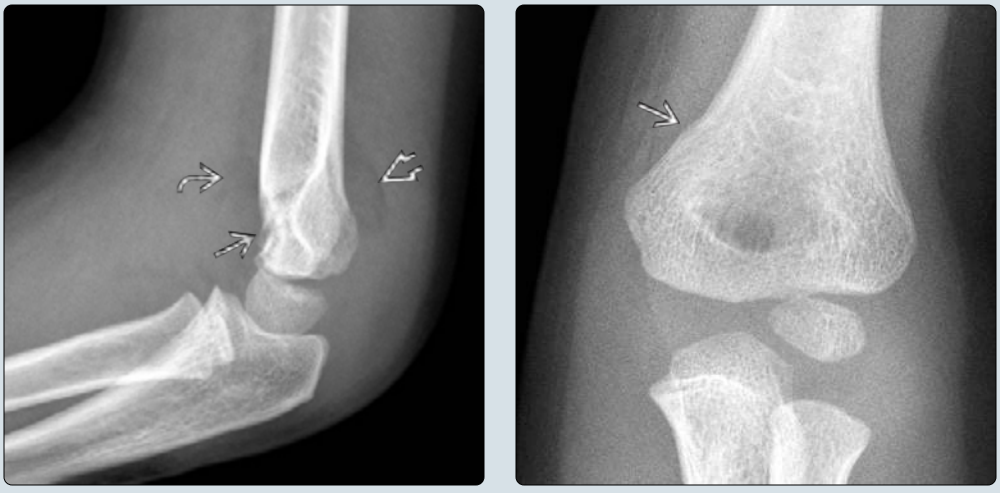
DIAGNOSTIC CHECKLIST

- Consider follow-up radiographs in 10-14 days to identify healing occult fracture if joint effusion only finding initially

(Left) *AP radiograph in a 5 year old after a fall shows a complete, transversely oriented distal humeral fracture through the olecranon & coronoid fossae with > 50% medial translation of the metaphysis.* **(Right)** *Lateral radiograph in the same patient shows no cortical contact of the posteriorly translated distal fragment with the proximal fragment (a Gartland III or IV supracondylar fracture). The elbow articulations remain intact on both views.*



(Left) *Lateral radiograph after a fall shows anterior cortical disruption in the typical location of a supracondylar fracture. There is mild dorsal angulation of the distal fragment (with hinged but intact posterior cortex) such that an anterior humeral line no longer bisects the capitellum (Gartland IIA). There is a large elbow joint effusion with elevation of the anterior & posterior fat pads.* **(Right)** *AP radiograph in the same child shows subtle medial cortical buckling of the supracondylar humerus.*



TERMINOLOGY

Synonyms

- Gartland fracture

Definitions

- Supracondylar fracture: Transverse fracture of distal humeral metadiaphyseal junction with varying degrees of displacement & comminution
- FOOSH injury: **F**all **o**n **o**ut**s**tretched **h**and
- Anterior humeral line: Vertical line drawn along anterior cortex of humeral diaphysis; normally bisects capitellum
- Radiocapitellar (RC) line: Line drawn through long axis of radius; normally bisects capitellum on any view
  - Due to eccentric ossification of capitellum, RC line may not bisect ossified capitellum < 10 years of age

IMAGING

General Features

- Best diagnostic clue
  - Lateral radiograph
    - Dorsal angulation of distal humeral metaphysis with failure of anterior humeral line to bisect capitellum
    - Displacement of anterior & posterior fat pads by joint effusion → may be only finding of nondisplaced fracture
  - AP radiograph
    - Transverse metadiaphyseal junction fracture lucency
- Location
  - Fracture occurs at thin distal humerus between olecranon (posterior) & coronoid (anterior) fossae
- Morphology
  - Various configurations depending on degree of angulation, translation, rotation, & comminution

Radiographic Findings

- Radiography
  - AP view
    - Transversely oriented linear lucency of distal humeral metadiaphyseal junction
    - Fracture line may not be visible in up to 25%
    - Subtle buckling or angulation of medial or lateral cortex may be only finding on this view
  - Lateral view
    - Displacement of fat pads due to joint effusion
      - Posterior fat pad not normally visible
    - Dorsal angulation ± translation of distal humeral fracture fragment
      - Anterior humeral line no longer bisects capitellum in majority
  - RC line typically maintained
    - Disruption would indicate radial head subluxation/dislocation (or possibly patient < 10 years old if mild)
  - Follow-up imaging
    - Baumann angle (shaft-capitellum angle) on AP view: Superolateral angle between humeral diaphyseal long axis & physis deep to capitellar ossification center
      - Normally 64-81° → mild valgus carrying angle
      - ↑ Baumann angle (> 5° vs. contralateral side): Cubitus varus (5-10%)
    - Fishtail deformity of central humeral epiphysis due to lateral trochlear avascular necrosis
      - Uncommon complication of distal humeral fractures (most frequently supracondylar)
      - May lead to proximal migration of ulna + radial head subluxation/dislocation

DIFFERENTIAL DIAGNOSIS

Lateral Condylar Fracture

- Fracture extends obliquely through lateral humeral metaphysis to central physis

Posterior Dislocation

- Ulna & radius displaced posteriorly, laterally, & proximally
  - RC line no longer intersects capitellum
- No supracondylar metadiaphyseal fracture

Medial Epicondyle Avulsion

- Avulsion ± intraarticular entrapment of medial epicondyle
- Elbow dislocation in 50%

Distal Humeral Epiphyseal Separation (Transphyseal Fracture)

- Uncommon fracture of young children (< 2 years of age) through distal humeral physis
- May be difficult to recognize due to lack of radiographically visible ossification centers
- Capitellum, radius, & ulna translate medially
  - RC line maintained (if capitellum visible)
- High association with child abuse

PATHOLOGY

General Features

- Etiology
  - Supracondylar humerus susceptible to fracture due to thin bone between medial & lateral pillars at coronoid fossa anteriorly & olecranon fossa posteriorly
  - Mechanism: Hyperextension form (98%) in FOOSH injury vs. flexion type (2%) from fall onto posterior elbow with direct trauma to olecranon
- Associated abnormalities
  - Concomitant elbow & forearm fractures (11%)
    - Olecranon, medial epicondyle, distal radius
    - Floating elbow: Forearm fractures become unstable in setting of supracondylar fracture (5%)
      - ↑ risk of neurovascular injury & compartment syndrome
  - Nerve injuries (10-20%)
    - Anterior interosseous/median > ulnar, radial nerves
      - Ulnar nerve injury more likely with flexion mechanism
      - Transection uncommon, usually radial nerve
  - Vascular injuries (3-14%)
    - Spasm, laceration, thrombus, rupture

Staging, Grading, & Classification

- Modified Gartland classification
  - Type I
    - Nondisplaced or minimally displaced (< 2 mm)
    - Intact anterior humeral line
    - Posterior fat pad sign may be only finding

- Type IIA
  - Mildly displaced (> 2 mm) anterior cortex
  - Posterior cortex intact but hinged
  - Anterior humeral line touches anterior capitellum but does not extend through middle 1/3
  - No rotational deformity on AP view
- Type IIB
  - Mildly displaced + malrotation or lateral displacement with maintenance of cortical contact
- Type III
  - Displaced fracture with no cortical contact
  - Periosteum torn
  - Often with soft tissue & neurovascular injuries
  - Subclassification into high vs. low may have prognostic implications
    - Based on fracture location relative to isthmus at midcoronoid/olecranon fossae
- Type IV
  - Multidirectional instability (determined intraoperatively)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, loss of function, swelling, discoloration
  - Varying degrees of deformity depending on displacement
- Other signs/symptoms
  - ↓ distal radial pulse, cool extremity
  - Open fracture in 3%

Demographics

- Age
  - Most common in children 5-7 years old
  - Suggestive of abuse in nonambulatory infants
- Epidemiology
  - 60% of pediatric elbow fractures
  - 3-16% of all pediatric fractures
  - Rare in adults (< 3%)

Natural History & Prognosis

- Return of function in over 90% regardless of Gartland type
- Return of range of motion may take 1 year
- In patients with cool pulseless extremity, radial pulse often returns quickly (> 50%) after closed reduction
- Nerve dysfunction typically transient; may require months

Treatment

- Conservative
  - Type I: Above elbow cast in 90° flexion for 3-4 weeks
- Surgical
  - Type II: Treatment controversial
    - Closed reduction & casting vs. pin fixation
      - ~ 70% of type II fractures maintain alignment with casting + weekly follow-up
      - Remainder lose alignment & require fixation
  - Type III/IV: Percutaneous lateral pin fixation vs. open reduction internal fixation (ORIF)
  - Prompt reduction & pinning for vascular compromise or floating elbow
    - Delayed treatment of 8-24 hours otherwise increasingly accepted
  - Lateral divergent (splayed) K-wires >> crossing K-wires (from medial & lateral approaches)
    - Crossed method shows better fixation strength but ↑ risk of ulnar nerve injury
  - ORIF may be needed in up to 20%
    - Open fractures
    - Entrapped soft tissues preventing closed reduction
    - Worsening neurovascular compromise after closed treatment
- Complications
  - Neurovascular injury, compartment syndrome, infection, loss of reduction (even with pinning), malunion, restricted motion, hyperextension, cubitus varus or cubitus valgus, fishtail deformity
    - Cubitus varus can predispose to subsequent lateral condylar fracture (mean of 18-32 months later), posterolateral rotary instability (> 20 years later), delayed ulnar nerve palsy
      - Requires corrective osteotomy
    - Fishtail deformity found at mean time of ~ 5 years after injury with pain, "cracking," ↓ mobility

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Be sure to look for other elbow & forearm fractures
- Consider follow-up radiographs in 10-14 days to identify healing occult fracture if joint effusion only finding initially

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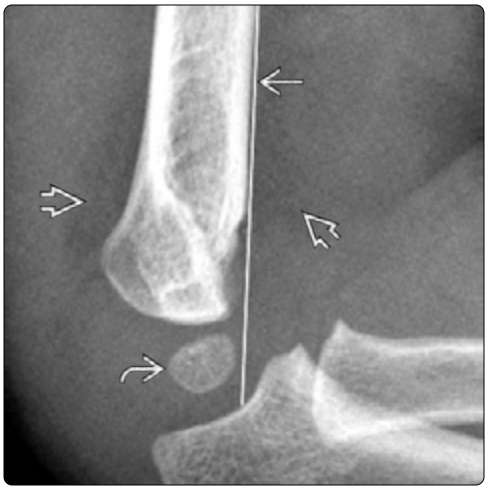
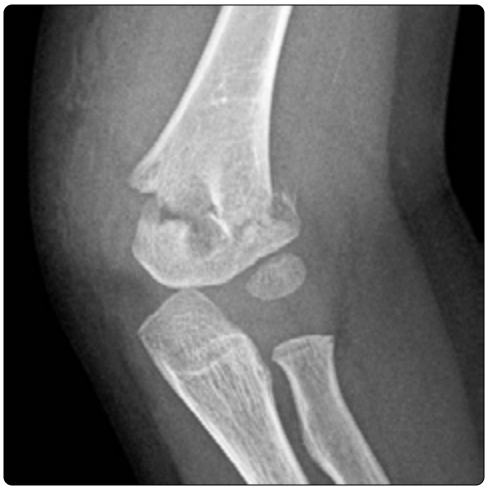
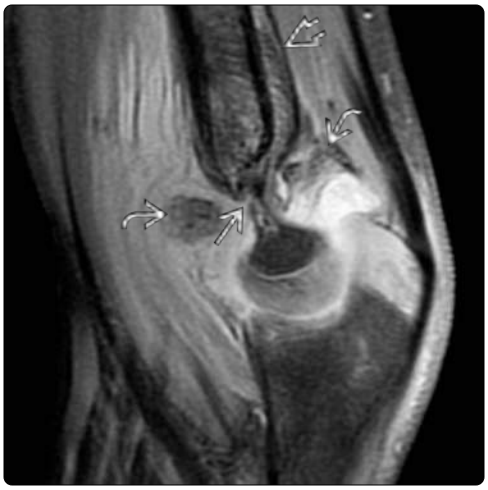
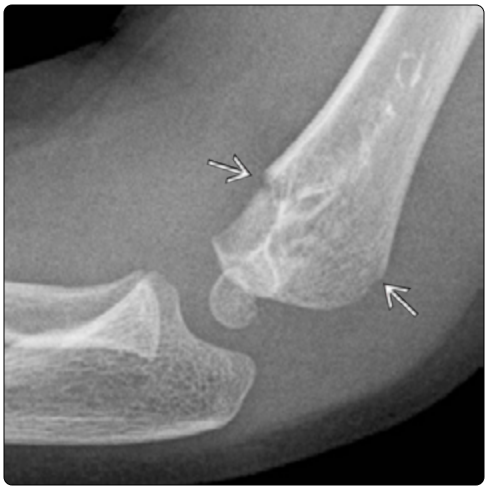


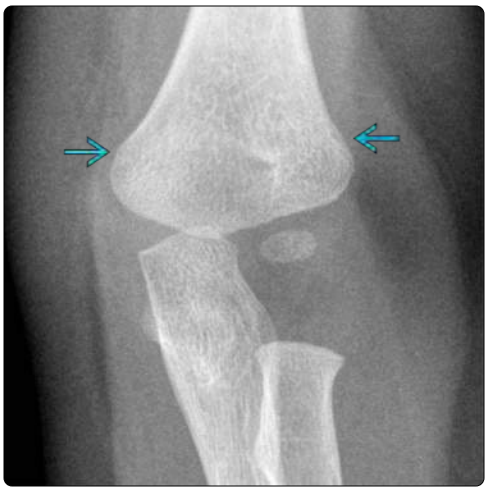


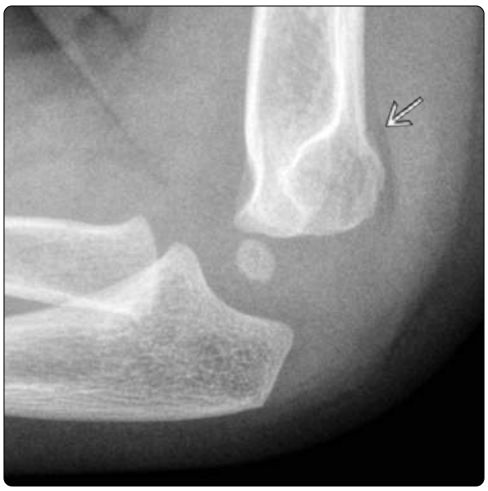
(Left) *Lateral radiograph shows that the anterior humeral line fails to intersect the capitellar ossification center. This finding is consistent with a supracondylar fracture. Note the elevation of both the anterior & posterior fat pads, indicating an elbow joint effusion.* **(Right)** *AP radiograph shows translation & angulation of a transversely oriented supracondylar fracture of the distal humerus, either a Gartland type III or IV fracture (with multidirectional instability being present clinically in the latter type).*





(Left) *Sagittal PD FS MR in a 9 year old with a healing supracondylar fracture shows anterior cortical disruption with displacement of the humeral fat pads by a joint effusion. Note the posterior humeral periosteal reaction.* **(Right)** *Lateral radiograph in a patient after a fall shows a horizontally oriented fracture lucency through the supracondylar humerus with no significant displacement.*





(Left) *AP radiograph in a 23-month-old patient shows slight cortical irregularity of the supracondylar humerus with overlying soft tissue swelling.* **(Right)** *Lateral radiograph in the same patient shows displacement of the posterior fat pad overlying a mildly angulated posterior humeral cortex. The anterior humeral line fails to bisect the capitellum.*

Lateral Condylar Fracture

KEY FACTS

TERMINOLOGY

- Posterior oblique fracture through lateral condyle of humeral metaphysis with distal extension through unossified epiphyseal cartilage (Salter-Harris IV)
  - Fracture line may dissipate within epiphyseal cartilage before reaching articular surface, leaving cartilaginous hinge/bridge

IMAGING

- AP view: Ranges from sliver of nondisplaced metaphyseal fracture fragment to marked translation & rotation of larger triangular metaphyseal fragment + capitellum
- Lateral view: Posterior oblique metaphyseal fracture plane; displacement of lucent anterior & posterior fat pads by joint effusion
- Internal oblique radiographs: More accurate for detecting fracture & determining displacement
- Cartilaginous extension not radiographically visible but implied by degree of bone fragment displacement
  - Lack of bone fragment displacement does not ensure cartilage integrity/fracture stability
  - Arthrography, MR, or US characterizes extent of cartilaginous articular surface injury

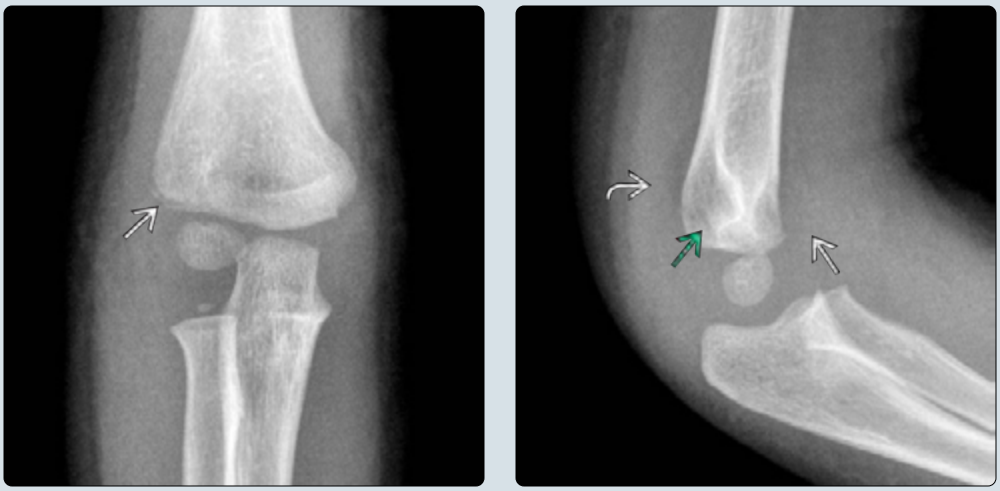
PATHOLOGY

- Jakob classification used to determine therapy
  - Type I (≤ 2 mm displacement): Typically casted
  - Type II (> 2 mm displacement without rotation): Closed reduction with percutaneous pinning
  - Type III (> 2 mm displacement with rotation): Open reduction & fixation

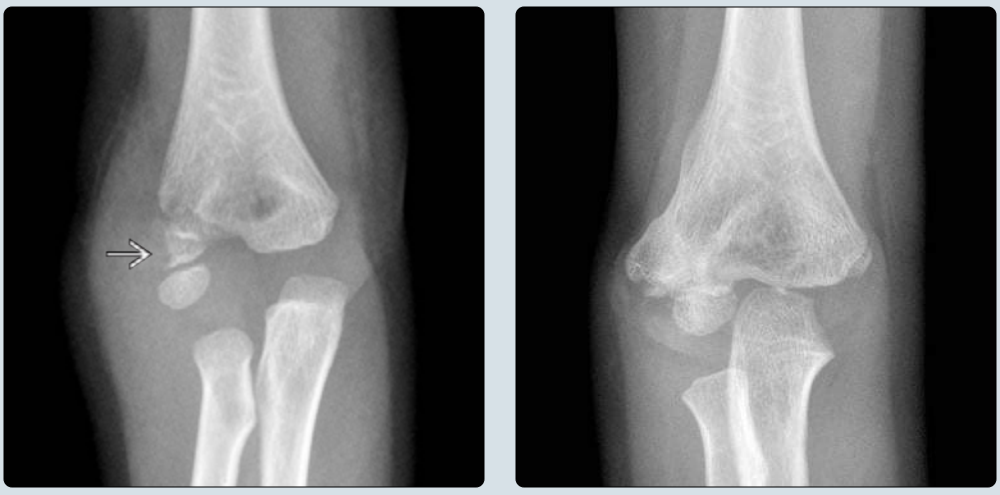
CLINICAL ISSUES

- 10-20% of pediatric elbow fractures (2nd most common)
- Typically 5-10 years old; peak age: 6 years
- Complications include stiffness, late displacement, nonunion, delayed union, malunion, prominence or spurring of lateral condyle, capitellar avascular necrosis, tardy ulnar nerve palsy

(Left) *AP radiograph in a 2 year old who fell off of a laundry basket shows an oblique fracture line through the lateral condyle with a sliver of a metaphyseal osseous fragment. This is a Jakob type I fracture.* **(Right)** *Lateral radiograph in the same child shows an elbow joint effusion with elevation of both the anterior & posterior fat pads. The fracture line has a posterior oblique course, typical of lateral condylar fractures.*



(Left) *AP radiograph in a 4 year old who injured her elbow after falling off of a scooter shows a displaced & rotated Jakob type III lateral condylar fracture.* **(Right)** *AP radiograph in the same patient 8 months later shows a healed fracture with an enlarged lateral condyle & mild cubitus varus, both of which are recognized complications of lateral condylar fractures. This patient's fracture had been previously transfixed by 3 percutaneous K-wires.*



TERMINOLOGY

Definitions

- Posterior oblique fracture through lateral condyle of humeral metaphysis with distal extension through unossified epiphyseal cartilage
  - Fracture line may dissipate within epiphyseal cartilage before reaching articular surface

IMAGING

General Features

- Morphology
  - Proximal fracture component parallels margin of physis (or slightly steeper)
  - Salter-Harris IV fracture: Distal extension through unossified epiphyseal cartilage not radiographically visible

Radiographic Findings

- AP view: Ranges from sliver of nondisplaced metaphyseal fracture fragment to marked translation & rotation of larger triangular metaphyseal fragment + capitellum
  - Nondisplaced fracture may be occult or easily missed
  - If missed &/or not treated appropriately, may lead to nonunion
- Lateral view: Posterior oblique metaphyseal fracture plane; displacement of lucent anterior & posterior fat pads by joint effusion
- Internal oblique radiographs: More accurate for detecting fracture & determining displacement
  - May still underestimate true displacement
- Arthrography characterizes extent of cartilage/articular surface injury

CT Findings

- CT with reformation: Helpful for understanding complex fracture components to determine classification & treatment

MR Findings

- Vertical/oblique linear focus of fluid signal intensity through unossified distal humeral epiphyseal cartilage
- Horizontal to oblique linear high or low T1/T2 signal intensity fracture component in lateral metaphyseal bone, ± surrounding marrow edema
- MR also useful for follow-up of complications

Ultrasonographic Findings

- May be used to assess cartilage

Imaging Recommendations

- Protocol advice
  - Radiographs: AP & lateral views initially
    - Internal oblique view: Most sensitive for occult lateral condylar fractures & determining displacement
  - MR
    - Coronal & sagittal PD & T2 FS
    - Useful in assessing cartilage integrity/disruption
    - More expensive + need of sedation in young child

DIFFERENTIAL DIAGNOSIS

Supracondylar Fracture

- Most common pediatric elbow fracture
- Transversely oriented fracture of distal humerus extending through level of olecranon fossa & coronoid fossa
  - Variable degrees of displacement
  - Disruption of anterior humeral line (normally bisects middle 1/3 of capitellum) on lateral view
  - May be occult showing only elbow joint effusion

Olecranon Fracture

- Association with lateral condylar & other elbow fractures
- Must not confuse normal ossification center for fracture

Medial Epicondyle Avulsion

- ~ 10% of elbow fractures in pediatrics
- Distraction of medial epicondyle (ME) ossification center
  - Can become entrapped in elbow joint, simulating trochlear ossification center
- Unreliable fat pad displacement: ME tends to be extracapsular > 2 years old
- 50% associated with elbow dislocations

Radial Neck Fracture

- 5% of elbow fractures in children
- Salter-Harris II fracture: 90%

Transphyseal Fracture

- Distal humeral epiphyseal separation < 2 years old
  - Displacement of largely unossified epiphyseal cartilage medially, resulting in malalignment between humerus & radius (thereby suggesting elbow joint dislocation)
    - May be difficult to distinguish from dislocation when capitellum not ossified
      - In true dislocation, radiocapitellar alignment disrupted with radius displaced laterally & posteriorly
      - In transphyseal fracture, capitellum still aligns with radial head
- > 50% associated with nonaccidental trauma

PATHOLOGY

General Features

- Etiology
  - Push-off: Fall on outstretched hand causing impaction of lateral condyle into radial head
  - Pull-off: Avulsion by lateral collateral ligamentous complex & common extensor tendons
- Associated abnormalities
  - Olecranon fractures

Staging, Grading, & Classification

- Salter-Harris IV epiphyseal fracture extension much more common than Salter-Harris II (through medial physis)
- Jakob classification: Most common system used by surgeons; based on fragment displacement on internal oblique radiograph
  - Type I
    - Nondisplaced or ≤ 2 mm of displacement
    - Fracture line presumed not to extend to articular surface

- Type II
  - > 2 mm of displacement (typically < 4 mm)
  - Partial disruption of articular surface with intact hinge/bridge of cartilage
    - No rotation of lateral condylar fragment
- Type III
  - > 2 mm of displacement
  - Complete disruption of articular surface
    - Metaphyseal fragment & capitellum rotated
    - Loss of radiocapitellar alignment → elbow instability
- Milch classification: Less commonly used due to high radiographic-surgical discordance
  - Milch type I
    - Fracture extends through capitellar ossification center lateral to trochlear groove
  - Milch type II
    - Fracture extends into trochlea rather than through capitellar ossification center

CLINICAL ISSUES

Presentation

- Lateral elbow swelling
- Lateral supracondylar ridge tenderness
- Absent pain over medial supracondylar ridge
- Ecchymosis

Demographics

- 10-20% of pediatric elbow fractures
  - 2nd in frequency after supracondylar fracture
  - Most common intraarticular pediatric elbow fracture
- Age: Typically 5-10 years old; peak: 6 years

Natural History & Prognosis

- Complications
  - Stiffness most common
  - Late fracture displacement 3 days to 3 weeks after nonoperative cast placement in ~ 11%
  - Nonunion
    - ↑ in intraarticular fractures
      - Bathed by synovial fluid leading to ↓ healing
      - Pull of extensor muscles
      - Poor circulation to distal fracture fragment
    - ↑ in inadequately treated or displaced fractures
    - ↑ in missed or unrecognized fractures
    - Fracture treated with closed reduction
  - Delayed union
    - > 6-8 weeks
  - Malunion
    - Fishtail deformity
      - Concavity with deepening of trochlear groove due to avascular necrosis
      - Rare; more common in supracondylar fractures
      - Usually minimal symptoms early; may have limited flexion & extension long term
    - Cubitus varus
      - More common than cubitus valgus
      - Most common with nondisplaced or minimally displaced fractures
    - Cubitus valgus
      - Fragment migration vs. lateral growth arrest
      - May lead to tardy ulnar nerve palsy
  - Tardy ulnar nerve palsy
    - Slow progressive paralysis of ulnar nerve
      - Atrophy of intrinsic hand muscles, sensory loss
      - Latency period of several months to many years
    - Treated by anterior transposition of ulnar nerve
  - Enlarged &/or spurred lateral condyle (nearly 50%)
    - Usually cosmetic without affecting function
  - Avascular necrosis of capitellum
    - Rare complication of extensive surgical dissection
  - Degenerative arthritis

Treatment

- Type I (≤ 2 mm of displacement)
  - Controversial: Some advocate surgical treatment
    - Occult extension to joint may occur without initial fracture displacement (thereby predisposing to late displacement if only casted initially)
  - Most commonly treated with long arm cast
    - Frequent follow-up radiographs every 7-10 days
      - Due to risk of late displacement
- Type II (> 2 mm of displacement without rotation)
  - Closed reduction with percutaneous pinning
    - Consider compression screws
  - Intraoperative arthrography helpful in determining articular surface congruity
- Type III (> 2 mm of displacement with rotation)
  - Open reduction & fixation
    - K-wires &/or compression screws
- Recent literature suggests that classic 2-mm cutoff for operative intervention too high with displacement of > 1.6 mm being at risk of complications if surgery delayed
- Cast maintained for 4-6 weeks with operative or nonoperative treatment
- Removal of hardware after healing
- Physical therapy for stiffness

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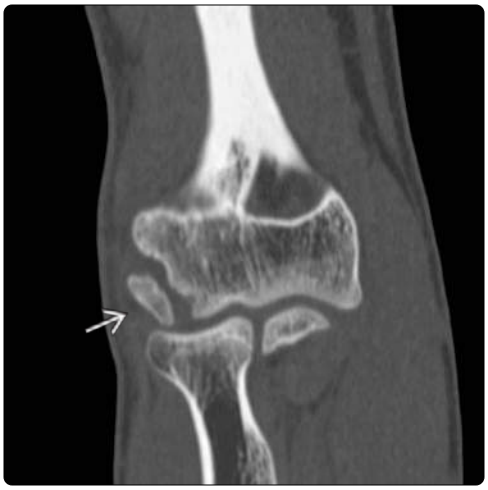
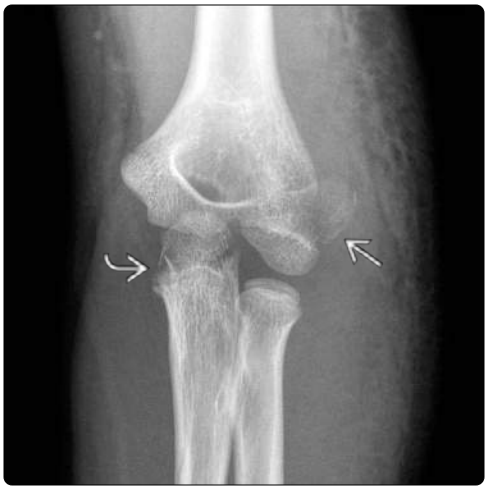
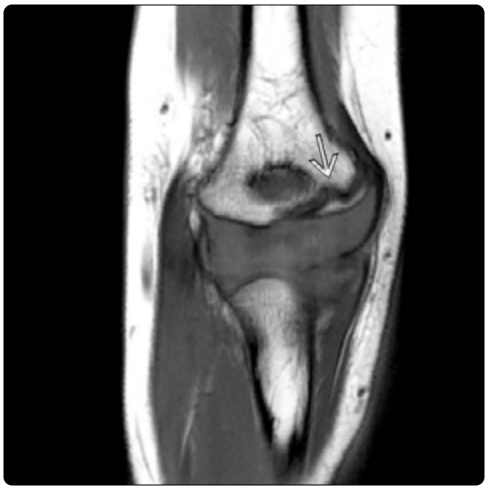
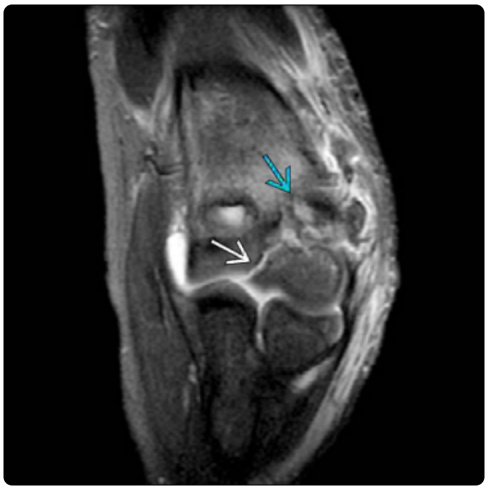


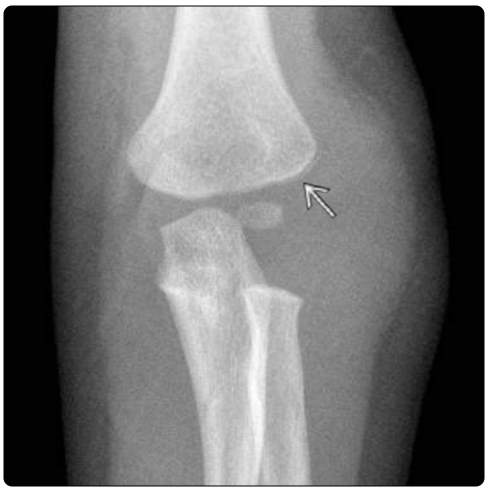


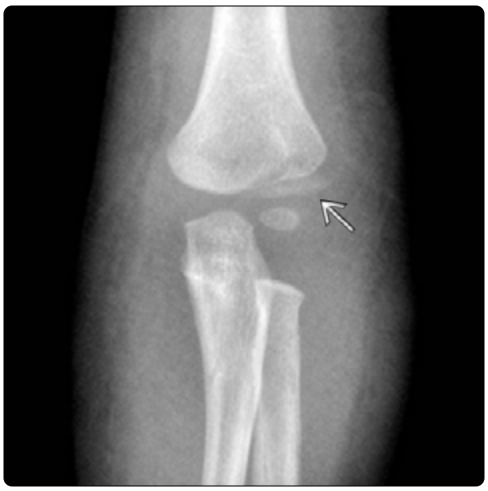
(Left) *Coronal reformatted bone CT in a 15 year old who sustained a lateral condylar fracture while playing football 3 years prior shows nonunion of a displaced fragment. This complication is seen more often in children with inadequately treated or displaced fractures.* **(Right)** *AP radiograph in a 6 year old with elbow trauma shows a displaced & rotated Jakob type III lateral condylar fracture with overlying soft tissue swelling. There is also an olecranon process fracture.*





(Left) *Coronal T1 MR in a 5 year old who sustained an elbow injury shows a hypointense oblique fracture line extending across the lateral condyle. No intraarticular extension of the fracture was identified.* **(Right)** *Coronal T2 FS MR in a patient after a fall shows an oblique hyperintense fracture line in the trochlear cartilage extending from the disrupted lateral condylar metaphysis to the articular surface.*





(Left) *AP radiograph in an 18 month old after falling out bed shows a subtle Jakob type I fracture. Note the marked lateral soft tissue swelling.* **(Right)** *AP radiograph in the same child 1 week later shows increased displacement of the fracture despite immobilization. Delayed displacement is a known risk of nonoperative management in Jakob type I fractures.*

Medial Epicondyle Avulsion

KEY FACTS

TERMINOLOGY

- Acute injury: Avulsion fracture of medial epicondyle (ME) ossification center
- Chronic stress injury: Traction apophysitis, medial epicondylitis, Little Leaguer's elbow

IMAGING

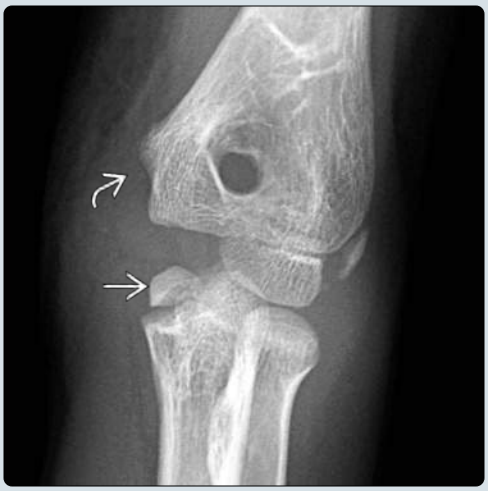
- Acute injury: Distal &/or medial displacement of ME ossification center with moderate soft tissue swelling
  - Remember CRITOE pattern of ossification at elbow
    - Should normally see ME in expected location on AP radiograph if trochlea present
      - Important observation as avulsed, entrapped ME can simulate trochlear ossification center
  - Unreliable fat pad sign: Joint effusion may be absent
    - ME may become extracapsular > 2 years of age
- Chronic injury: Widening & irregularity of cartilaginous physis deep to ME
  - Less pronounced ME separation & soft tissue swelling
  - ± fragmentation & edema of ME

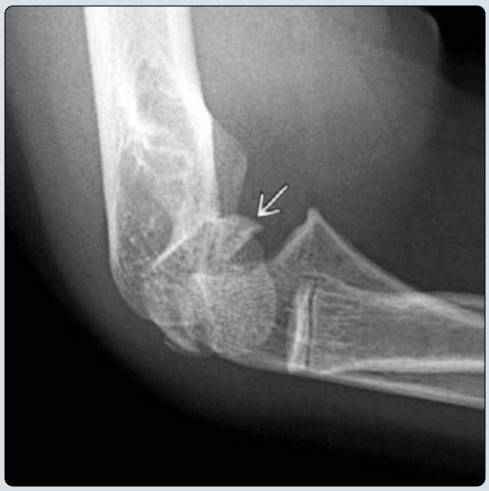
PATHOLOGY

- Weakest link of immature musculoskeletal system: Osteocartilaginous interface
  - Acute tensile force → osteocartilaginous avulsion
    - Skeletally mature patients more likely to injure ligaments, tendons, muscles
  - Chronic submaximal valgus forces (repetitive microtrauma) exceed healing capacity of system → irritation & disturbance of normal ossification

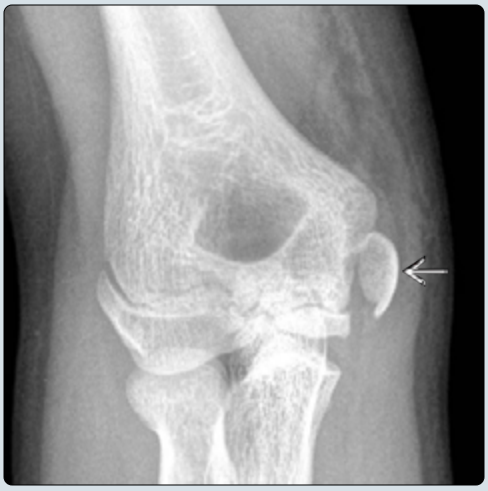
CLINICAL ISSUES

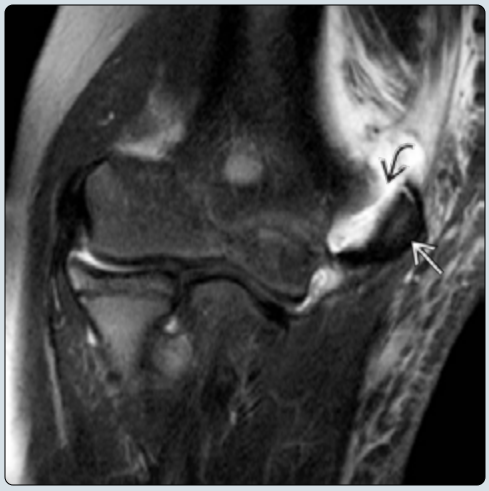
- Typical age for acute & chronic ME injuries: 8-14 years
- Acute avulsion fracture: Elbow dislocation in 50%, entrapped ME in 15-20%
- Chronic stress injury: Same mechanism predisposes to capitellar osteochondritis dissecans (OCD) & olecranon stress injuries
  - Typically overhead throwing athletes (e.g., pitchers)





(Left) *AP radiograph shows an ossific fragment ➡ adjacent to the medial margin of the olecranon of the ulna. No medial epicondyle (ME) ossification center is identified in its expected location ➡, though soft tissue swelling is noted at this level.* **(Right)** *Lateral radiograph in the same child shows that the avulsed ME ossification center ➡ is trapped in the joint.*





(Left) *AP radiograph shows an acute avulsion of the ME ossification center with mild rotation & distal displacement of the fragment ➡.* **(Right)** *Coronal T2 FS MR in a 13-year-old pitcher with an acute injury shows a displaced ME ossification center ➡. Hyperintense fluid ➡ is interposed between the fragment & the medial humeral metaphysis, & there is moderate adjacent soft tissue edema.*

TERMINOLOGY

Synonyms

- Acute medial epicondyle (ME) injury: Avulsion fracture
- Chronic injury: Stress injury, medial epicondylitis, traction apophysitis, Little Leaguer's elbow

Definitions

- Acute injury: Avulsion of ME ossification center
- Chronic stress injury: Repetitive traction microtrauma exceeds healing capacity; causes inflammation, growth disturbance, degeneration
  - Skeletally immature: Apophysitis of ME
  - Skeletally mature: Partial tearing/degeneration of common flexor tendon, medial/ulnar collateral ligament

IMAGING

General Features

- Best diagnostic clue
  - Acute: Radiographic displacement of ME ossification center + overlying soft tissue swelling after injury
  - Chronic: MR fluid-sensitive sequence showing marrow & soft tissue edema at ME + widening, irregularity of physis
- Morphology
  - Should normally see ME in expected location on AP radiograph if trochlea identified → excludes entrapped ME, which can simulate trochlear ossification center

Radiographic Findings

- Acute injury: Displacement of ME ossification center medially &/or distally
  - Or laterally into joint (especially with lateral/posterior elbow dislocation) between olecranon & trochlea
  - ± joint effusion (therefore fat pad sign unreliable)
- Chronic injury: Enlargement, sclerosis, fragmentation of ME with subjacent physeal widening & irregularity ± thickening of medial humeral cortex
- Overlying soft tissue swelling: Acute > chronic injuries

MR Findings

- Acute: Separation of osteocartilaginous ME ossification center from humerus with interposed fluid signal
  - Moderate adjacent soft tissue edema ± marrow edema
- Chronic: Widening & irregularity of cartilaginous physis mimicking mild ME separation from humerus
  - Mild marrow & soft tissue edema, ± fragmentation of ME

DIFFERENTIAL DIAGNOSIS

Ulnar/Medial Collateral Ligament Injury

- More common with skeletal maturity
- Complete tear: Disruption of taut, linear/fan-shaped hypointense band
- Sprain: Thickened ligament with intermediate/high signal

Flexor Muscle Injury/Strain

- More common with skeletal maturity
- Thickening & ↑ signal intensity with tendinopathy

Olecranon Stress Injury

- Pattern varies by age & degree of skeletal maturity
- Olecranon fragmentation, edema with appropriate history

Capitellar Osteochondritis Dissecans

- Repetitive impaction of radial head on capitellum with valgus stresses
- Lucent defect with flattening of anterior/mid capitellum
- Lateral pain & locking with loose intraarticular body

PATHOLOGY

General Features

- Etiology
  - ME serves as attachment for medial/ulnar collateral ligament & flexor-pronator muscle group of forearm
  - Osteocartilaginous interface most likely site to fail under acute tensile stress in skeletally immature patients
    - Musculotendinous junction or ligament in adults
  - Mechanism for acute avulsion
    - Forceful contraction of flexor-pronator muscle group, often during pitching or wrestling
    - Fall on outstretched hand with elbow flexed
    - Posterior/lateral elbow dislocation (50%)
  - Mechanism for chronic stress injury (traction apophysitis)
    - Overuse syndrome found in athletes participating in overhead throwing sports (e.g., baseball pitcher)
    - Repeated tensile valgus stress during cocking & acceleration phases of throwing causes irritation of ME apophysis/physis
- Associated abnormalities
  - Elbow dislocation in 50% of acute avulsions
    - Ulnar nerve injury: 10-50%
    - ME trapped in elbow joint: 15-20%
  - Chronic repetitive valgus stresses also lead to capitellar impaction with osteochondritis dissecans & olecranon stress injuries

CLINICAL ISSUES

Treatment

- Acute avulsion fracture treatment controversial
  - Operative management
    - Absolute indications: Open fracture, incarcerated ME
    - Relative indications: Elbow instability, ulnar neuropathy, ME displacement, high-level athletes
      - Degree of displacement necessitating fixation unclear: 2 vs. 5 vs. 10 mm
  - Nonoperative management
    - Good functional outcomes despite high nonunion rate
- Treatment of chronic stress injuries largely conservative
  - Rest, physical therapy, antiinflammatory medications, proper pitching techniques

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Forearm Fractures

KEY FACTS

TERMINOLOGY

- Complete fracture: Macroscopic fracture line traverses entire bony diameter (both cortices on single view)
- Incomplete fracture: Macroscopic fracture line does not traverse entire bony diameter
  - Opposite cortex often deformed

IMAGING

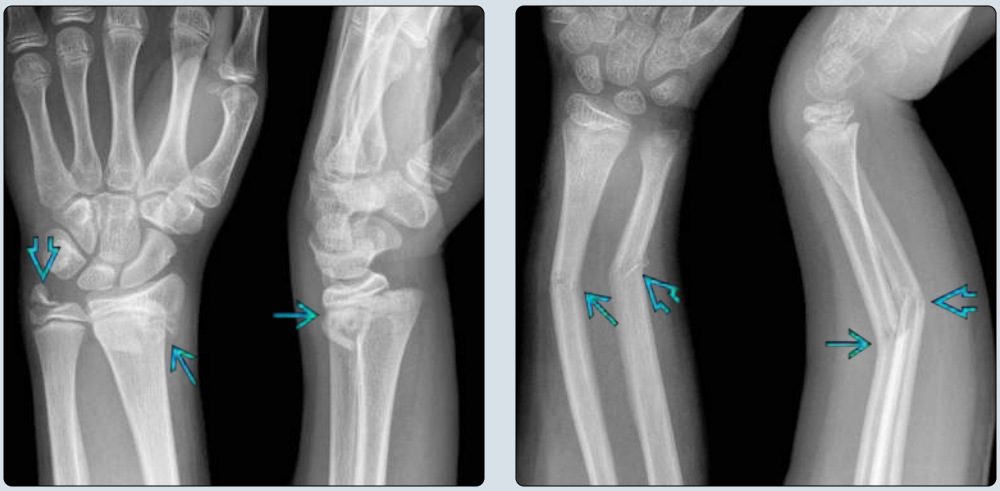
- Focal, abrupt cortical angulation, "buckling," &/or discrete fracture line with overlying soft tissue swelling
  - Few sites of normally occurring cortical angulation/protuberance in children
  - Comparison radiographs helpful if uncertain
- ± physeal extension of fracture with displacement of metaphyseal fragment & epiphysis
- Distal radial metaphysis/metadiaphyseal junction
  - Up to 85% of pediatric forearm fractures
  - Buckle (on compression side), complete oblique/transverse, or physeal fractures
  - Distal ulnar injury usually present
- Diaphysis
  - Plastic (bowing) deformity, greenstick (with cortical interruption of tension side), or complete fractures
  - Both shafts usually fractured (unless dislocated)
- Proximal radius & ulna fractures considered with elbow
  - Radial neck & ulnar olecranon most common

PATHOLOGY

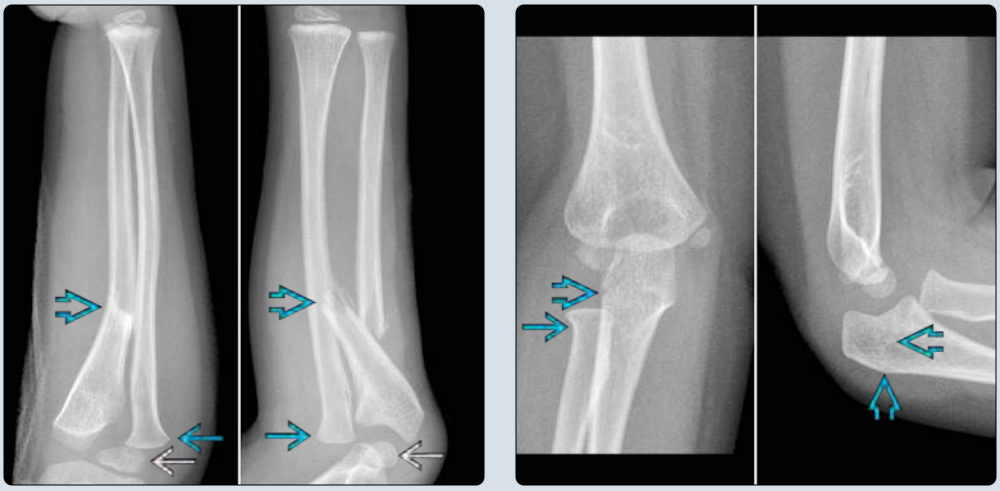
- Fall on outstretched hand > > direct blow
- ↑ elasticity of pediatric bone allows for greater deformation under stress prior to break

DIAGNOSTIC CHECKLIST

- Often subtle; must have 2 tangential views
- Report displacement (translation, angulation, & rotation), physeal &/or articular involvement, dislocation
  - Include direction & magnitude



(Left) *PA & lateral views of the wrist in a 12 year old show a comminuted Salter-Harris 2 fracture of the distal radius with ~ 50% dorsal translation of the distal fracture fragment. An ulnar styloid fracture is also present. Though not seen in this case, an ulnar metadiaphyseal fracture also frequently occurs in this setting.* **(Right)** *AP & lateral forearm radiographs in a 6 year old show a complete ulnar diaphyseal fracture with an incomplete greenstick radial diaphyseal fracture (note the intact cortex).*



(Left) *AP & lateral forearm radiographs in a 4 year old show a complete fracture of the proximal ulnar diaphysis with dislocation of the proximal radius relative to the capitellum, consistent with the Monteggia fracture-dislocation complex. The radial head has not yet ossified.* **(Right)** *AP & lateral elbow radiographs in a 5 year old show nondisplaced fractures of the radial neck (with focal angulation of the cortex) & proximal ulna (with linear lucency & cortical interruption), which often occur together.*

TERMINOLOGY

Definitions

- Complete fracture: Macroscopic fracture line traverses entire bony diameter (both cortices on single view)
  - Physeal (Salter-Harris): Fracture through cartilaginous growth plate, typically with adjacent bony involvement
  - Monteggia: Radial head dislocation + proximal ulnar shaft fracture
- Incomplete fracture: Macroscopic fracture line does not traverse entire bony diameter
  - Buckle: Focal convex (bump) deformity of cortex (compression side)
  - Plastic (bowing) deformity: Smooth, flowing abnormal curvature of bony shaft
  - Greenstick: Frank interruption of 1 cortex (tension side) without fracture line through opposite cortex

IMAGING

General Features

- Best diagnostic clue
  - Abrupt angular deformity or frank interruption of normally smooth metadiaphyseal cortical contour
- Location
  - Distal radial metaphysis/metadiaphyseal junction
    - Up to 85% of pediatric forearm fractures; 25-43% of all pediatric fractures
    - Buckle, complete transverse, or physeal
      - 15% involve physis; almost all Salter-Harris II
    - Ulnar injury usually present
      - Ulnar styloid or incomplete metaphyseal fracture > physeal injury or dislocation (Galeazzi)
      - Often subtle
  - Diaphysis
    - Plastic (bowing) deformity, greenstick, or complete
    - Both shafts usually fractured ("both bone") unless dislocated
    - Adjacent shaft fractures may be of different types
  - Radial neck + proximal ulna/olecranon
    - Typically considered as elbow fracture

Radiographic Findings

- Focal abrupt cortical angulation, protuberance ("buckling" on compression side), &/or discrete fracture line with overlying soft tissue swelling
- ± physeal extension of fracture with displacement of metaphyseal & epiphyseal fragments

Ultrasonographic Findings

- Focal interruption/bulging of echogenic cortex ± subperiosteal hemorrhage, soft tissue edema

Imaging Recommendations

- Vast majority diagnosed & managed by radiographs alone

DIFFERENTIAL DIAGNOSIS

Normal Developmental Variants

- Few sites of normally occurring mild cortical angulation/protuberance or shaft bowing
- Contralateral comparison radiographs helpful

Metabolic Bone Diseases, Skeletal Dysplasias

- Pathologically weakened bones ± multiple fractures may show chronic bowing
- Severe types have diffusely abnormal bones

PATHOLOGY

General Features

- Etiology
  - Fall on outstretched hand > > direct blow
  - ↑ elasticity of pediatric bone allows for greater deformation under stress prior to break
    - Frank cortical breaks propagate < adults

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, swelling, tenderness, ↓ use

Natural History & Prognosis

- Remodeling potential of pediatric fractures > > adults due to residual bone growth
  - Children (especially < 10 years old) generally tolerate ↑ degrees of residual displacement (within certain limits) of nonarticular fractures after closed reduction
- Buckle fractures
  - Generally require only short-term (3 weeks) immobilization by splint
- Complete or incomplete diaphyseal fractures
  - Usually closed reduction & casting sufficient
  - Up to 33% of reduced distal radial fractures will lose their reduction, require intervention
- Physeal injuries
  - < 5% develop subsequent symptomatic growth arrest at distal radius vs. ~ 50% growth arrest rate at distal ulna
  - Osteotomy, epiphysiodesis, or distraction to treat
- Ulnar styloid injuries
  - Vast majority not treated; nonunion typical
  - Few patients will have long-term symptoms

DIAGNOSTIC CHECKLIST

Reporting Tips

- Type, direction, & magnitude of displacement
  - Translation (including shortening or distraction), angulation, rotation
- Presence of physeal &/or articular involvement
- Associated dislocation
- Presence of healing or underlying pathologic lesion

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KEY FACTS

TERMINOLOGY

- Intrasubstance anterior cruciate ligament (ACL) tears may be complete or partial; differentiation can be difficult
- Avulsion fractures of tibial eminence at distal ACL attachment more frequent in skeletally immature patients

IMAGING

- Radiographs: Joint effusion most common but nonspecific
  - Less common but more specific secondary findings: Deep lateral condylar notch sign & Segond fracture
- MR: ↑ intrasubstance signal of ACL with disrupted fibers
  - Empty lateral intercondylar wall on coronal PD/T2 FS
  - "Kissing" contusions (poorly defined edema/hemorrhage in lateral femoral & posterolateral tibial condyles)
  - Additional injuries: Menisci, MCL, posterolateral corner

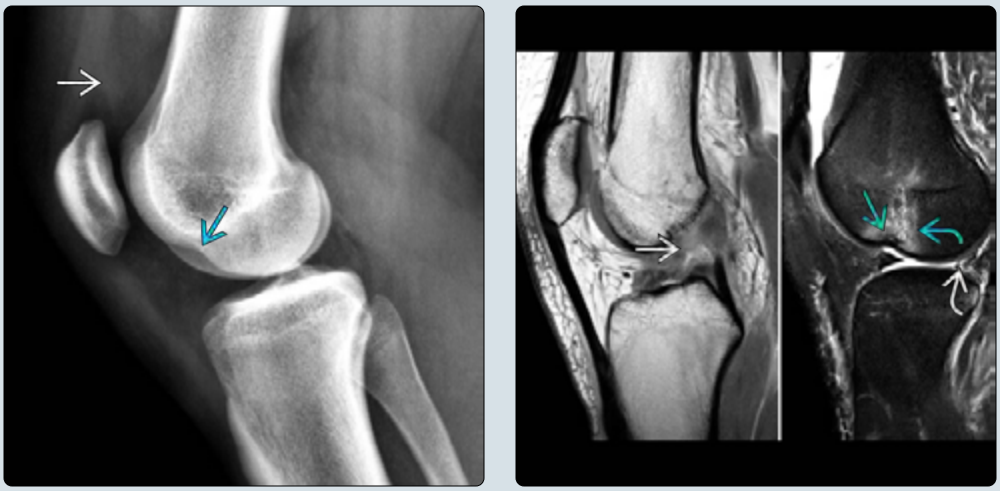
CLINICAL ISSUES

- Mid or proximal substance ACL tears typical of athletic patients at or approaching skeletal maturity (F > M)
- Tibial eminence avulsion fractures most common at 8-14 years of age (M > F); ACL intact or partially torn

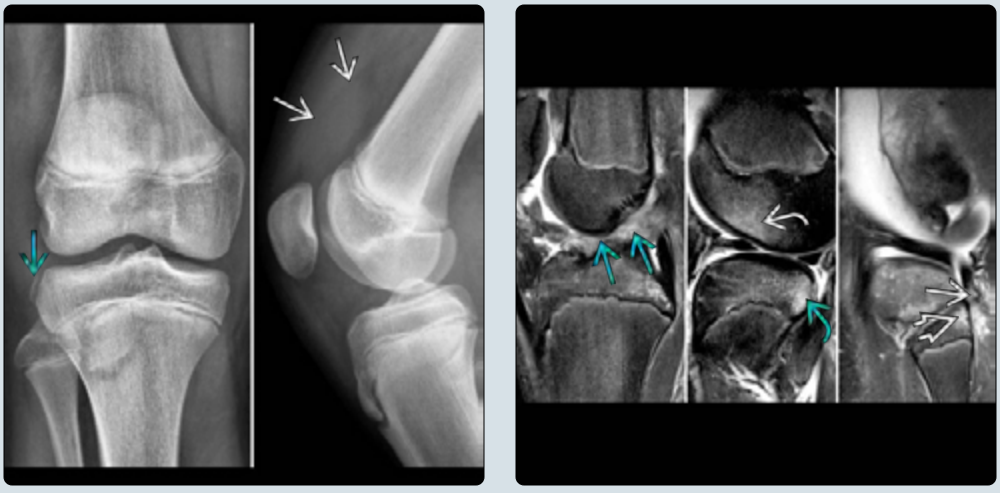
DIAGNOSTIC CHECKLIST

- Evaluate integrity of ACL in 3 standard planes
  - Oblique planes along course of ACL may improve detection of partial tears
- Important associated MR findings in setting of ACL tear
  - Meniscal tears (posterior horn lateral meniscal tears often missed)
    - Closely inspect lateral meniscal-meniscofemoral ligament attachment
    - Apparent extension of attachment ≥ 4 images (on 3-mm slices) lateral to PCL indicates tear
  - Posterolateral corner & MCL injuries
  - Meniscus or transverse ligament entrapment under avulsed tibial eminence fracture fragment

(Left) *Lateral radiograph of a 17-year-old girl with a right knee injury during a soccer game shows a joint effusion & a deep lateral condylar notch that measures > 3 mm.* **(Right)** *Sagittal PD (left) & T2 FS (right) MRs (same patient) show subchondral fracture of lateral femoral condyle (MR equivalent of lateral notch sign) with underlying edema + a complex tear of posterior horn of the lateral meniscus. A more central PD image shows a complete midsubstance anterior cruciate ligament (ACL) tear.*



(Left) *Frontal (left) & lateral (right) radiographs in a boy with a right knee injury show a joint effusion & a Segond fracture. Segond fractures are nearly always associated with ACL tears.* **(Right)** *Sagittal T2 FS MRs from central (left) to lateral (right) in the same boy show a complete intrasubstance ACL tear, "kissing contusions" in the lateral femoral & posterolateral tibial condyles, & partial tearing of the popliteofibular ligament with edema in the fibular head.*



TERMINOLOGY

Abbreviations

- Anterior cruciate ligament (ACL)

Definitions

- ACL has anteromedial & posterolateral bundles
- ACL tears can be complete or partial

IMAGING

General Features

- Best diagnostic clue
  - Radiographs: Large joint effusion
    - Less common: Deep lateral condylar notch, Segond fracture
  - MR: ↑ intrasubstance signal & disrupted fibers
    - Empty lateral intercondylar wall on coronal PD/T2 FS
    - "Kissing" contusions (lateral femoral & posterolateral tibial condyles)
- Location
  - Typically midsubstance or proximal
  - Tibial eminence avulsion fractures of distal ACL in younger (skeletally immature) children

Radiographic Findings

- Midsubstance/proximal ACL tears
  - Large joint effusion
    - Absence of joint effusion following acute injury virtually excludes ACL tear
  - Segond fracture: Vertically-oriented avulsion from lateral proximal tibia
    - Associated ACL tear: 75-100%
    - Associated meniscal tear: 66-75%
  - Deep lateral femoral notch sign
    - Suggests ACL tear if > 1.5-2 mm
  - Fibular head avulsion fracture (arcuate sign)
    - Indicates injury to arcuate ligament complex
    - Associated with cruciate ligament [posterior cruciate ligament (PCL) > ACL] tears
- Tibial eminence fractures
  - Meyers & McKeever classification (modified)
    - Type I: Minimal displacement
    - Type II: "Trap-door" configuration (elevation of anterior fragment with posterior hinge)
    - Type III: Completely displaced fragment
    - Type IV: + comminution of fragment

MR Findings

- T2WI FS
  - Primary signs
    - ↑ intrasubstance signal, diffuse thickening, & discretely disrupted fibers
      - May be partial or complete: Can be difficult to distinguish, even at 3T
    - Empty lateral intercondylar wall on coronal images
      - Fluid signal at femoral attachment of ACL
    - Abnormal horizontal or bowed orientation of ligament relative to roof of intercondylar notch
  - Secondary signs
    - "Kissing contusions": Poorly defined foci of high signal intensity in marrow of central lateral femoral & posterolateral tibial condyles
    - Anterior tibial translation
    - Uncovered lateral meniscus sign
    - Laxity of PCL
    - Posterolateral corner injury (PLC)
    - Medial collateral ligament injury
    - Meniscal tears
- PD/intermediate
  - Useful to evaluate associated meniscal injuries
  - Posterior horn lateral meniscus tear: Most commonly missed tear associated with ACL injury
    - Often confused with "pseudotear" at lateral meniscal-meniscofemoral ligament attachment
    - Apparent ↑ lateral extension of this attachment site should raise suspicion for tear
      - ≥ 4 images (on 3-mm slices) lateral to PCL = tear
- Chronic ACL tears
  - Complete: Fatty signal in notch without ACL fibers
  - Chronic partial tears: Attenuated ACL
- Tibial eminence fracture
  - MR performed to assess for associated injuries
    - Partial intrasubstance ACL tears
    - Interposition of menisci or transverse ligament under avulsed fracture fragment
      - Up to 33% of type II & 65% of type III fractures
      - Can prevent reduction & healing

Imaging Recommendations

- Best imaging tool
  - Radiographs in acute setting
    - Evaluate for joint effusion, deep lateral condylar notch, Segond fracture, or tibial eminence avulsion
  - MR confirms ACL tear & evaluates associated injuries
    - Meniscal, medial collateral ligament (MCL), PLC, & cartilaginous injuries
- Protocol advice
  - Evaluate ACL on all 3 standard planes (not just sagittal)

DIFFERENTIAL DIAGNOSIS

Congenital Absence of ACL

- Hypoplasia to complete absence of ACL
- Small/absent intercondylar notch & dysplasia of tibial eminence

ACL Ganglion

- Lobular, septated fluid signal mass along or within ACL

Synovial Cleft of ACL

- Variant herniation of fluid-containing synovium into ACL

PATHOLOGY

Gross Pathologic & Surgical Features

- ACL intraarticular but extrasynovial (i.e., encased by synovial lining such that joint fluid does not directly contact ACL)
- Tibial intercondylar eminence divided into 4 regions
  - Medial & lateral intercondylar tubercle or spine
  - Anterior & posterior intercondylar area

- ACL footprint at anterior intercondylar area, between anterior attachments of medial & lateral menisci
- Due to this proximity, menisci &/or transverse ligament can become trapped under avulsed tibial eminence fragment → prevents fracture reduction & healing
- Strong association between ACL tears & meniscal tears

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Complete ACL tear
    - Acute swelling & pain with knee "giving out"
    - Sensation of &/or audible "pop" at time of injury
- Clinical profile
  - Complete ACL tears
    - Pivot shift injury
      - Noncontact injury seen commonly in American football players & skiers
      - Valgus stress applied to flexed knee + external rotation of tibia or internal rotation of femur loads ACL → tear
      - ACL rupture → unrestrained anterior translation of tibia relative to femur → impaction of central lateral femoral & posterolateral tibial condyles
      - Various degrees of flexion may alter location of bone bruise in femoral condyle (i.e., ↑ flexion → more posterior, ↓ flexion → more anterior)
      - Can get contrecoup injury with bone contusion to posteromedial tibial condyle
  - Tibial eminence fractures
    - Seen with ↑ frequency related to ↑ childhood athletic activities
    - Mechanism of injury
      - Typically hyperextension force ± valgus or rotational component
      - May occur from direct blow to femur with knee flexed
- Clinical examination
  - ACL injury
    - Lachman test: Anterior translation force applied to proximal tibia with knee fully extended
      - Lack of solid/firm endpoint = + test
    - Anterior drawer test: Anterior translational force applied to proximal tibia with knee flexed at ~ 80°
      - ↑ translation (vs. contralateral side) = + test

Demographics

- Epidemiology
  - ↑ frequency of ACL tears in skeletally immature children
    - ACL injuries in skeletally immature children: M > F
    - Tibial eminence fractures much more common at 8-14 years of age than in older children
  - Frequency of ACL tears in skeletally mature adolescents approaches that seen in adults
    - ACL injuries in young but skeletally mature patients
      - F > M

Natural History & Prognosis

- Complete ACL tears in skeletally immature patients often have poor outcomes
  - Instability → degenerative change, meniscal tears, & chondral injury
  - Few patients return to preinjury sports level
- Partial ACL tears can progress to complete tears & symptomatic laxity within 1 year
- Tibial eminence fractures
  - May be associated with partial tear of ACL
    - Contributes to residual sagittal laxity even following "anatomic" reduction & healing

Treatment

- Complete ACL tears need ligamentous reconstruction
  - May be complicated by presence of open growth plates
    - Various physeal-sparing approaches for children with substantial growth remaining
      - Intra- & extraarticular iliotibial band autograft
      - All epiphyseal approach: Uses epiphyseal tunnels & fixation
    - Transphyseal approach with metaphyseal fixation for patients past peak growth velocity
  - Adult reconstruction techniques in patients with closed or closing growth plates
- Tibial eminence fractures
  - Type I: Immobilization in long-leg cast or fracture brace with knee flexed at ~ 10-20°
  - Type II & III: Optimal treatment controversial
    - Type II
      - Closed reduction by knee extension & aspiration of hematoma
      - Open reduction & fixation if adequate closed reduction cannot be achieved
      - No long-term difference in patient outcomes
    - Type III-IV
      - Typically open or arthroscopic reduction with many different techniques described

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Important associated MR findings in setting of ACL tear
  - Meniscal tears (posterior horn lateral meniscal tears often missed)
    - Closely inspect lateral meniscal-meniscofemoral ligament attachment
    - Apparent extension of attachment ≥ 4 images (with 3-mm slices) lateral to PCL indicates tear
  - Posterolateral corner & MCL injuries
  - Meniscus or transverse ligament trapped under avulsed tibial eminence fracture fragment
- Oblique planes along ACL may improve partial tear detection

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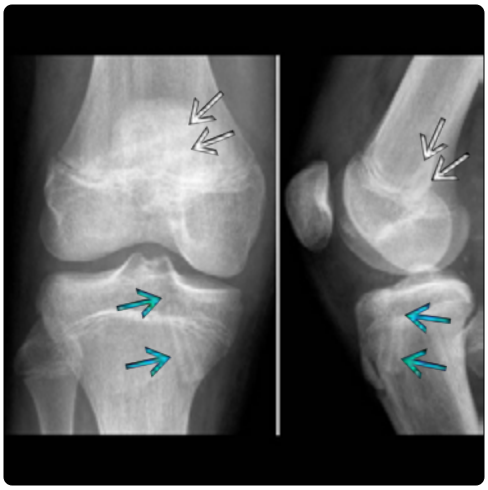
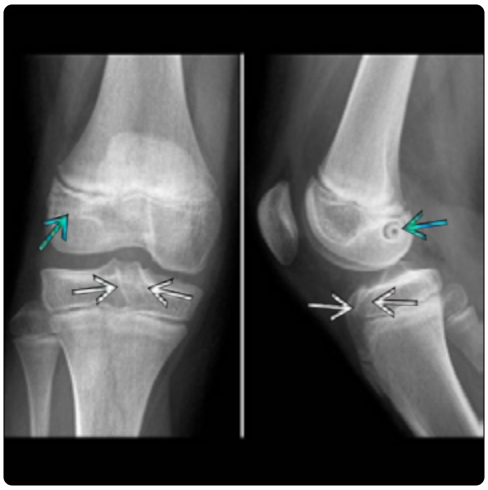
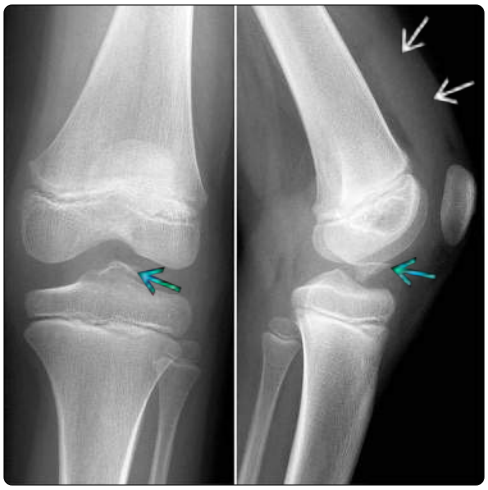
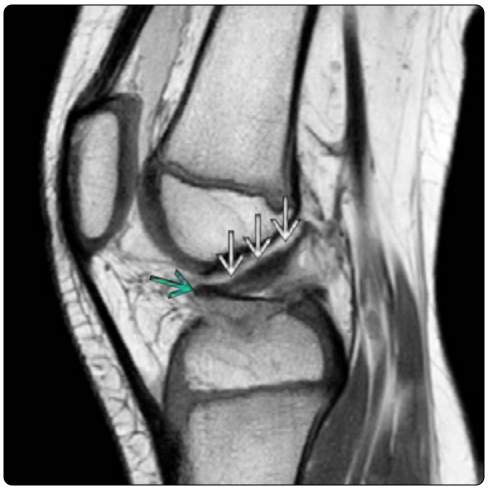


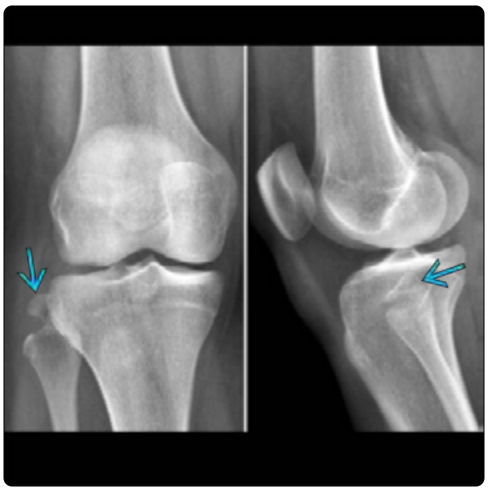


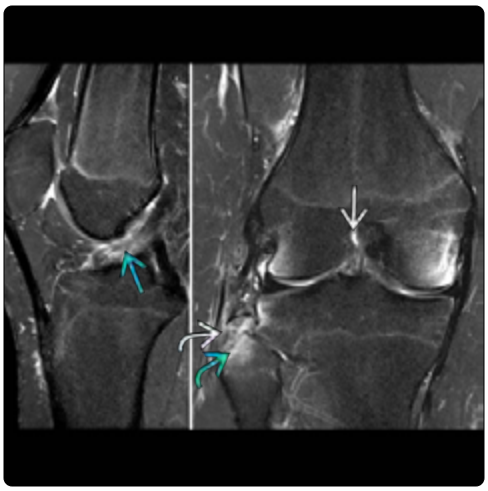
(Left) *Frontal (left) & lateral (right) radiographs after an ACL repair show that this boy was close enough to skeletal maturity to perform a transphyseal reconstruction. Note how the tibial tunnel & interference screw cross his tibial physis. Two bioabsorbable pins fix the graft to the femur.* **(Right)** *Frontal (left) & lateral (right) radiographs in a 12-year-old with a physeal-sparing ACL reconstruction show that the femoral tunnel interference screw & the tibial tunnel are completely epiphyseal & do not cross the physes.*





(Left) *Frontal (left) & lateral (right) radiographs in a 7-year-old boy with a left knee injury show an elevated tibial eminence avulsion fracture with a joint effusion.* **(Right)** *Sagittal PD MR from the same boy shows that the intact ACL is attached to the avulsed tibial eminence fragment, the anterior portion of which is elevated (consistent with a type II tibial eminence fracture). In this case, there is no meniscal tissue or transverse ligament trapped deep to the avulsed fragment.*





(Left) *Frontal (left) & lateral (right) radiographs in a 13-year-old girl with a right knee injury show an avulsion of the fibular head (arcuate sign), indicating an injury to the arcuate complex. This finding has a high association with cruciate ligament (PCL > ACL) tears.* **(Right)** *Sagittal (left) & coronal (right) T2 FS MR in the same patient show increased signal within indistinct ACL fibers as well as fluid in the lateral condylar notch. Note the avulsed fibular head fragment & adjacent fibular edema.*

Patellar Dislocation

KEY FACTS

TERMINOLOGY

- Transient patellar dislocation (TPD): Lateral dislocation & relocation of patella from direct/indirect injury
  - Shearing, tensile, compressive forces → medial patella, lateral femur, & soft tissue injuries

IMAGING

- Radiographs
  - Large joint effusion after acute TPD
  - Osseous fragments (of patella & lateral femoral condyle)
- MR
  - Medial patella & lateral femoral condyle contusions
  - Medial patellofemoral ligament (MPFL) tears
  - Chondral & osteochondral injuries

PATHOLOGY

- MPFL = condensation of medial retinaculum
  - Extends from superomedial patella to medial epicondyle of femur
  - Strongest passive medial stabilizer of patella

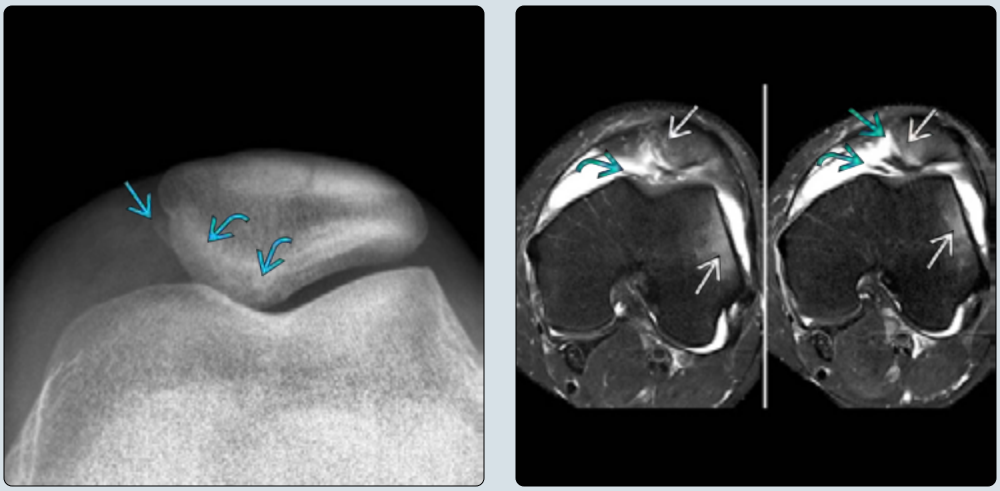
CLINICAL ISSUES

- Predisposing factors may be congenital or acquired
  - Patella: Alta, lateral subluxation/tilt, dysplasia
  - Trochlear dysplasia
  - Lateralization of tibial tubercle
  - Deficient/absent soft tissue medial stabilizers
  - Generalized ligamentous laxity (e.g., Ehlers-Danlos or Marfan syndrome)

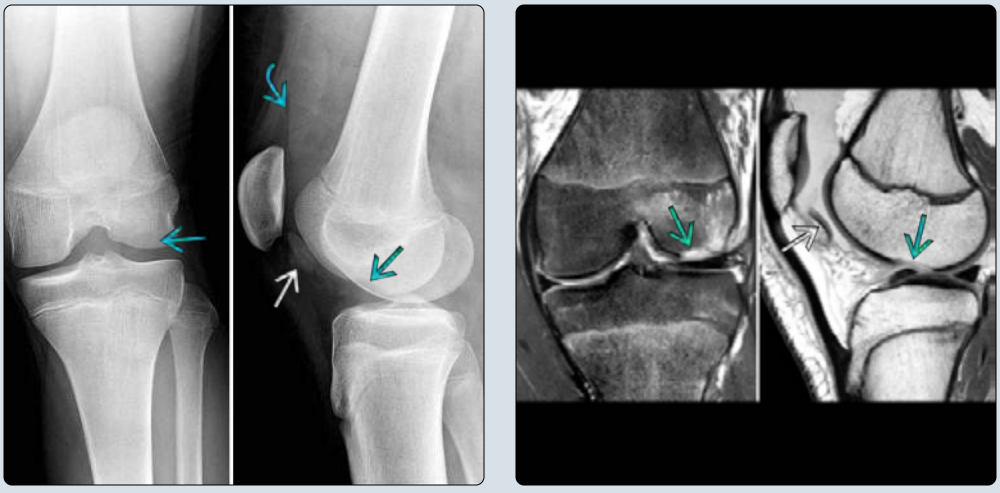
DIAGNOSTIC CHECKLIST

- Radiographic axial patellar view: Evaluate for small fragments & patellar articular surface integrity
- MR: Diagnosis (unsuspected) & characterization of injuries
  - Patellar & lateral femoral condylar articular cartilage injuries ± osteochondral/chondral intraarticular bodies
  - MPFL tears (evaluate entirety of MPFL)

(Left) *Axial radiograph of the patella in a 16-year-old boy with a recent transient patellar dislocation (TPD) shows an osseous fragment adjacent to the medial patella with irregularity of the entire medial facet.* **(Right)** *Axial T2 FS MR (superior on the left, inferior on the right) in the same patient shows bone contusions of the lateral femoral condyle & medial patella. There is also full-thickness articular cartilage loss of the medial patellar facet with an associated intraarticular osteochondral body.*



(Left) *Frontal & lateral radiographs in a 14-year-old boy with a recent TPD show a large osseous fragment in the anterior knee joint with an associated lucency in the lateral femoral condyle. A lipohemarthrosis is also present.* **(Right)** *Coronal T2 FS (left) & sagittal PD (right) MR in the same boy confirms a displaced, rotated osteochondral fracture fragment in the anterior aspect of the joint with a donor site at the lateral femoral condyle.*



TERMINOLOGY

Abbreviations

- Transient patellar dislocation (TPD)

Definitions

- Lateral dislocation of patella from direct/indirect injury
- Dislocation/relocation → shearing, tensile, & compressive forces → injuries of medial patella, lateral femur, & soft tissues

IMAGING

General Features

- Best diagnostic clue
  - Radiographs
    - Large hemarthrosis
    - Patellar or femoral osseous/osteochondral fragments
  - MR
    - Medial patella & lateral femoral condyle contusions
    - MPFL tear
- Morphology
  - Predisposing anatomic abnormalities of knee
    - Trochlear dysplasia, lateral patellar subluxation/tilt, patella alta, & tibial tubercle lateralization
  - Medial patellofemoral ligament (MPFL) normally dark, linear, oblique band extending from medial distal femur to superior 2/3 of patella

Radiographic Findings

- Large joint effusion after acute TPD
- Osseous fragments
  - Medial patellar osseous avulsion (at MPFL attachment)
  - Patellar & femoral osteochondral fracture fragments from dislocation/relocation
- Axial view of patella (e.g., Merchant, sunrise) important for
  - Osseous avulsions with osteochondral fragments along medial patella
  - Patellar articular surface irregularities
  - Medial soft tissue swelling
- Rarely see persistently dislocated patella on radiographs
  - Patella typically relocates when knee extended

MR Findings

- T1WI
  - Bone contusions: Poorly defined marrow foci of ↓ signal intensity in medial patella & lateral femoral condyle
- T2WI FS
  - Bone contusions: Poorly defined marrow foci of ↑ signal intensity in medial patella & lateral femoral condyle
  - MPFL injury can occur at femoral or patellar attachments, midsubstance, or multiple sites; similar criteria to other ligaments
    - Grade 1/sprain: ↑ signal about ligament
    - Grade 2/partial tear: Intrasubstance ↑ signal, partial fiber discontinuity
    - Grade 3/complete tear: Complete fiber discontinuity
    - Chronic tears
      - Focal or diffuse attenuation of MPFL
  - Vastus medialis obliquus (VMO) muscle tears can be seen in TPD, typically in association with MPFL tears
  - Chondral abnormalities
    - Cartilage injury: Ranges from softening (↑ signal but normal morphology) to focal defects (partial to full thickness)
    - Osteochondral fractures
    - Displaced chondral & osteochondral fragments → intraarticular bodies
      - Often laminar appearance from layered structure of cartilage & subchondral bone
    - Differentiation of chondral vs. osteochondral injuries made by assessing subchondral black line that separates cartilage from bone marrow
      - Intact black line with normal shape & thickness indicates preservation of subchondral bone
  - Joint effusion
    - Hemarthrosis may be evident with hematocrit layer
    - Lipohemarthrosis seen with osteochondral fractures
- Patella alta
  - Traditionally defined on radiographs
    - Insall-Salvati index = tendon length (TL)/patella length (PL) (> 1.2 considered patella alta)
  - Proposed MR criteria
    - Measured on single mid-sagittal image
    - Length of inner patellar tendon (TL)/anteroinferior to posterosuperior edge of patella (PL)
    - Ratios change during range of skeletal maturity
      - Values not definitively established in children
      - Some authors use MR ratio > 1.3 in children to define patella alta
- Trochlear dysplasia
  - Lateral trochlear inclination (of femur)
    - Shallow lateral inclination predisposes to TPD
    - Angle between lines along subchondral bone of lateral trochlea & posterior aspect of femoral condyles on axial image
    - Inclination angle < 11° = trochlear dysplasia
  - Trochlear facet asymmetry (of femur)
    - On axial image 3 cm above tibiofemoral joint, ratio of length of medial facet to lateral facet x 100%
    - Facet ratio < 40% = trochlear dysplasia
  - Trochlear depth (of femur)
    - Measured on axial image 3 cm above knee joint
    - Trochlear depth ≤ 3 mm = trochlear dysplasia
- Tibial tubercle to trochlear groove distance (TT-TG distance)
  - Measurement of lateral offset of extensor mechanism
  - Transverse distance from anterior-most osseous tibial tubercle to trochlear groove (deepest osseous portion)
  - Technique for measuring on axial images
    - Mark location of anterior-most tibial tubercle on inferior image
    - Scroll to superior slice with deepest trochlea & mark position of tibial tubercle on this image
      - Posterior condylar line drawn on this axial slice
    - Line perpendicular to posterior condylar line drawn between marks for
      - Deepest portion of osseous trochlea
      - Position of tibial tubercle
    - Distance between these lines = TT-TG distance
  - TT-TG distance varies with age; general guideline
    - Normal < 15 mm, borderline 15–20 mm, abnormal > 20 mm

- Patellar tendon to trochlear groove distance (PT-TG) distance
  - Measurement of lateral offset of extensor mechanism developed specifically on MR, where soft tissue & cartilage landmarks can be seen
  - Similar but not equivalent to TT-TG distance
  - Center of patellar tendon (PT) (on superior-most axial slice of tibial attachment) is used as distal landmark rather than tibial tubercle
  - Deepest portion of cartilaginous (vs. osseous) trochlea is used as proximal landmark for TG location
  - PT-TG vs. TT-TG distances may differ ≥ 4 mm in same individual
  - PT-TG distance has better inter- & intraobserver reliability
- Postoperative: MPFL reconstructions
  - Normal graft
    - Low signal intensity on all sequences with intact fibers
    - Continuous, taut, & oriented along course of native MPFL
  - Graft tear: Intrasubstance ↑ signal on T2WI FS MR + partial or complete disruption of fibers

DIFFERENTIAL DIAGNOSIS

Dorsal Patellar Defect

- Well-defined lucent lesion in superolateral aspect of patella
- Characteristic location & appearance distinguish from osteochondral fracture

Bipartite/Multipartite Patella

- Typically superolateral aspect of patella
- Characteristic location & appearance distinguish from fracture

PATHOLOGY

Staging, Grading, & Classification

- International Cartilage Reparative Society (ICRS)

Gross Pathologic & Surgical Features

- Condensations of medial patellar retinaculum form complex of medial patellar retinacular ligaments
  - MPFL most important
    - Strongest passive medial stabilizer of patella
      - Considerable variation in size & thickness of MPFL normal individuals
    - Contributes up to 60% of medial restraining force on patella

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Knee giving way, hemarthrosis/effusion, tenderness along medial retinaculum, sensation of impending dislocation with manual pressure upon patella
  - Patellar dislocation clinically occult in 45-73% of cases
    - Due to transient nature of process, patients frequently unaware that patella has dislocated
- Clinical profile
  - Majority of TPD occurs from direct or indirect trauma
  - Persons who dislocate with less forceful injuries typically have predisposing factors
  - Predisposing factors may be congenital or acquired
    - Osseous factors
      - Patella: Alta, lateral subluxation/tilt, dysplasia
      - Trochlear dysplasia: ↓ depth, ↓ lateral inclination
      - Lateralization of tibial tubercle
    - Soft tissue factors
      - Deficiency/absence of medial retinacular complex
      - Hypoplasia of vastus medialis muscle
      - Tight lateral retinaculum
      - Generalized ligamentous laxity (e.g., Ehlers-Danlos or Marfan syndrome)

Demographics

- Epidemiology
  - Overall incidence of 1st-time TPD = 5.8/100,000
  - In 10-17 year olds, incidence increases to 29/100,000

Natural History & Prognosis

- After single episode of TPD
  - 50% of patients have persistent anterior knee pain
  - If managed conservatively, 15-40% have recurrent TPD
- After 2nd TPD, chance of recurrence increases to 50%
- Recurrent dislocations ↑ risk for persistent symptoms & degenerative changes

Treatment

- Treatment of acute TPD is controversial
- Conservative management considered if
  - No osteochondral injury
  - Patella is stable on clinical exam
  - No more than partial injury to medial patellar stabilizers
- Conservative management
  - Patellar-stabilizing orthotic & early mobilization
  - Physical therapy directed rehab to strengthen medial patellar stabilizers
- Surgical management indicated for
  - 1st-time TPD with
    - Intraarticular bodies
    - Major tear of medial stabilizers
  - Recurrent TPD
- Surgical procedures (performed alone or in combination)
  - Lateral retinacular release
  - MPFL repair or reconstruction
    - Reconstruction: Single or double graft fixed to medial distal femur & medial patella
  - Distal realignment procedure
    - Tibial tubercle repositioning medial &/or distal
  - Trochleoplasty

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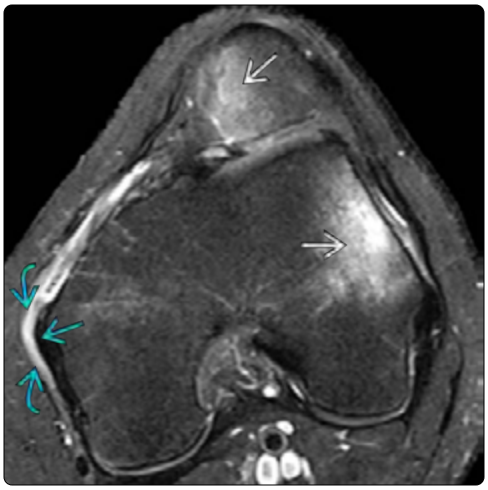
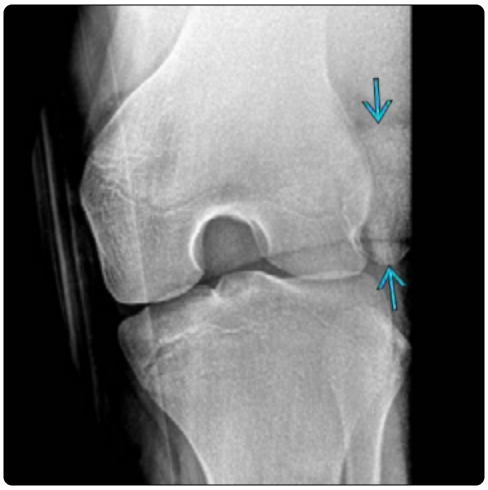
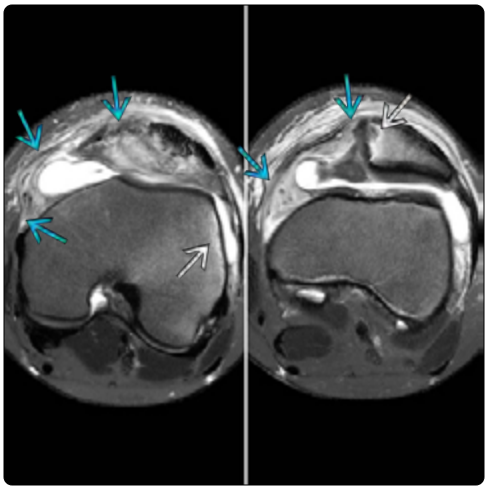
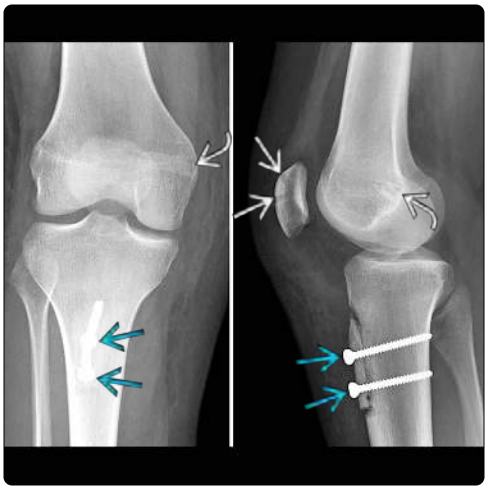


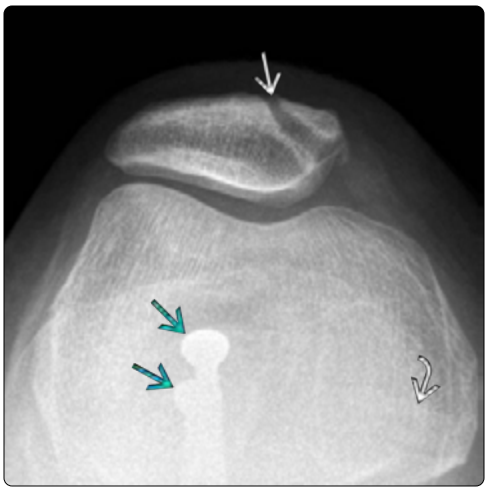


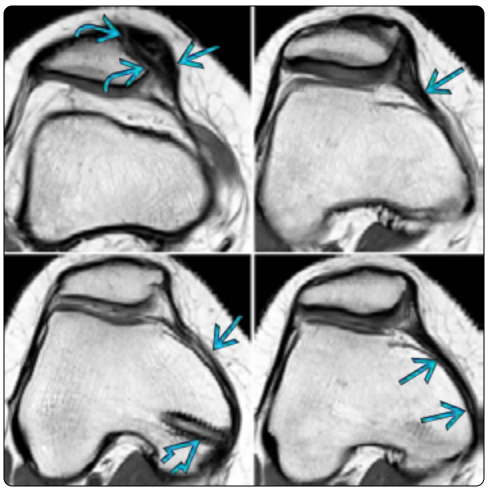
(Left) *Axial T2 FS MR in a 17-year-old boy with a recent TPD shows medial patella & lateral femoral condyle bone contusions. There is increased signal at the MPFL femoral attachment with complete disruption of the fibers. Note the normal MCL attachment deep to the torn MPFL.* **(Right)** *Tunnel radiograph in a 12 year old who was splinted immediately after a knee injury shows lateral dislocation of the patella. Typically, the patella reduces spontaneously with knee extension prior to radiographic evaluation.*





(Left) *Axial T2 FS MR (inferior on the left, superior on the right) in the same girl shows bone contusions in the lateral femoral condyle & medial patella with tearing of the MPFL throughout its course.* **(Right)** *AP & lateral radiographs in a 15-year-old girl with prior patellar dislocations show findings of MPFL reconstruction & tibial tubercle repositioning. The MPFL graft loops through a patellar tunnel & is fixed to the medial femur with an interference screw. Two screws fix the medially repositioned tibial tubercle.*





(Left) *Axial radiograph in the same girl shows the patellar tunnel through which the MPFL graft is looped. The femoral interference screw & tibial tubercle screws are also seen.* **(Right)** *Axial PD MR (clockwise superior to inferior from the upper left to the lower left) in the same patient shows the MPFL graft within the patellar tunnel extending posteriorly in an oblique course (similar to the native MPFL). The graft has normal low signal intensity & is fixed in a femoral tunnel with an interference screw.*

Patellar Sleeve Avulsion

KEY FACTS

TERMINOLOGY

- Avulsion injury of patella in skeletally immature patients
  - Small bone fragment & larger sleeve of unossified "epiphyseal" cartilage avulsed from patella at
    - Inferior pole by patellar tendon (more common)
    - Superior pole by quadriceps tendon (less common)
- **Not** periosteal sleeve avulsion (patella has no periosteum)

IMAGING

- Radiographs
  - Inferior pole: Small ossific fragment displaced distally from irregular inferior pole ± patella alta
  - Superior pole: Small ossific fragment retracted proximally from irregular superior pole ± patella baja
  - Either site: Lax quadriceps &/or patellar tendons, soft tissue swelling ± joint effusion
  - Extent of patellar injury & degree of fragment displacement underestimated by radiographs alone
- Ultrasound or MR reveals larger sleeve of unossified cartilage surrounding displaced bony fragment
  - With minimal displacement, fluid signal intensity in fracture cleft stands out against darker unossified "epiphyseal" cartilage on fluid-sensitive MR sequences

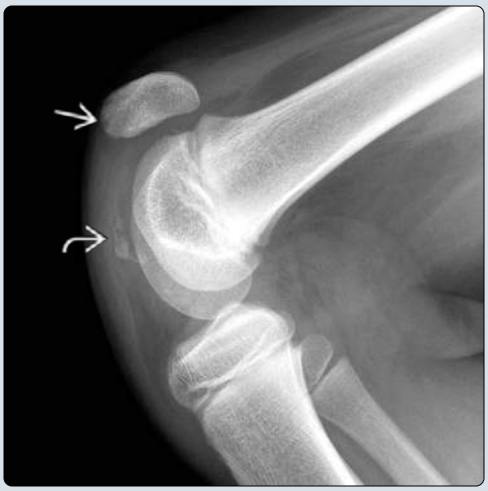
TOP DIFFERENTIAL DIAGNOSES

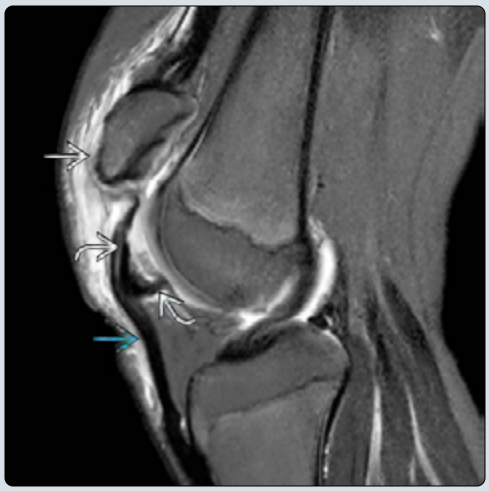
- Normal variant ossification
- Sinding-Larsen-Johansson
- Isolated extensor mechanism tendon rupture
- Tibial tubercle avulsion

CLINICAL ISSUES

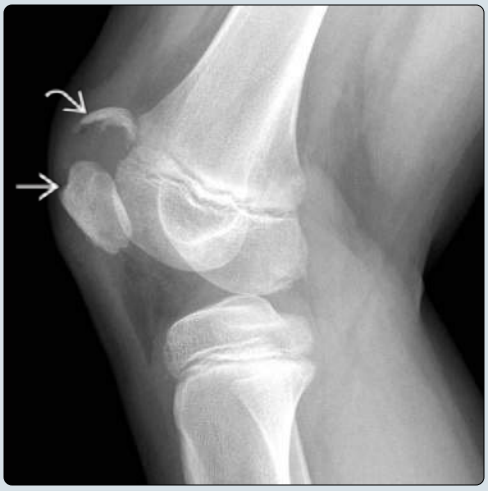
- Peak incidence: 12-13 years; range: 8-16 years
- Sudden pain with "explosive force" (such as jumping) or fall
  - Knee flexion & quadriceps contraction during injury
- Clinical findings: Swelling, extensor lag, palpable defect
- Treatment: Restore extensor mechanism function
  - Open reduction, internal fixation generally required

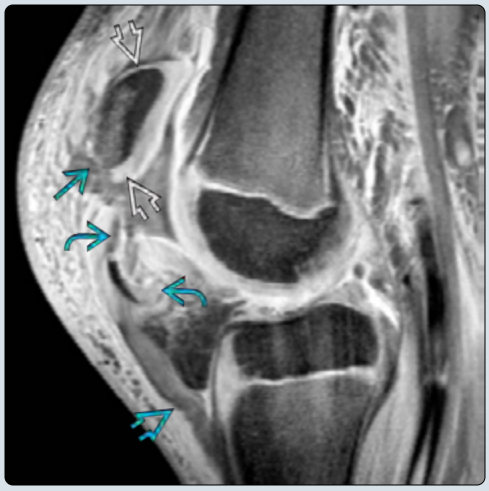
(Left) *Lateral radiograph in a 10-year-old girl after a fall shows ossific irregularity ➔ at the inferior pole of the high-riding patella (alta). The avulsed irregular ossific fragments ➔ remain attached to the proximal patellar tendon.* **(Right)** *Sagittal T2 FS MR of the same patient shows interruption of the low signal intensity unossified "epiphyseal" cartilage ➔ at the inferior pole of the superiorly retracted patella. An avulsed osteochondral sleeve ➔ remains attached to the patellar tendon ➔.*





(Left) *Lateral radiograph of the knee in a 10-year-old boy after a fall shows an ossific fragment ➔ retracted superiorly by the quadriceps tendon. This fragment was avulsed from the superior patellar pole ➔.* **(Right)** *Sagittal PD FS MR in a 10-year-old boy after a football injury shows avulsion of an osteochondral sleeve ➔ from the inferior pole of the patella ➔. Note the remaining intact unossified "epiphyseal" cartilage ➔ of the patella & the lax patellar tendon ➔.*





KEY FACTS

IMAGING

- Bone fragment retracted proximally by patellar tendon from site of anterior tibial tubercle ossification center
- Many variables, including fragment size & shape, degree of comminution & displacement, pattern of extension
- Adjacent soft tissues may become entrapped

TOP DIFFERENTIAL DIAGNOSES

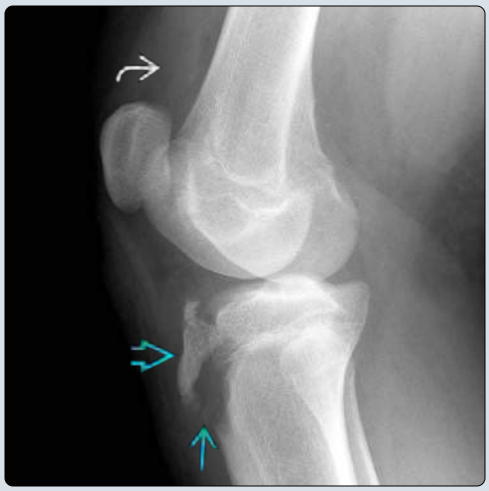
- Normal ossification variant
- Osgood-Schlatter disease
- Patellar sleeve avulsion fracture
- Patellar tendon rupture

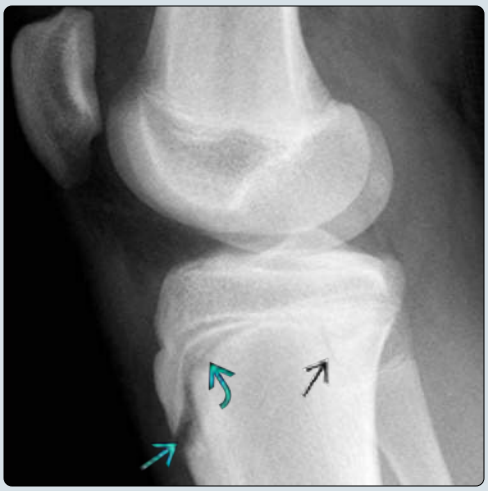
PATHOLOGY

- Predisposing anatomy
  - Portion of physis deep to tibial tubercle ossification center uniquely composed of strong fibrocartilage
  - As patient nears physeal closure, this growth plate converts to weaker hyaline cartilage
  - Timing coincides with ↑ muscle strength, athletic activity
- Mechanisms include forceful quadriceps contraction with extension (i.e., jumping) or passive knee flexion during quadriceps contraction (i.e., landing)
- Prior Osgood-Schlatter disease in 25%
- Associated injuries (< 5%): Tears of patellar tendon, ACL, collateral ligaments, & menisci; compartment syndrome

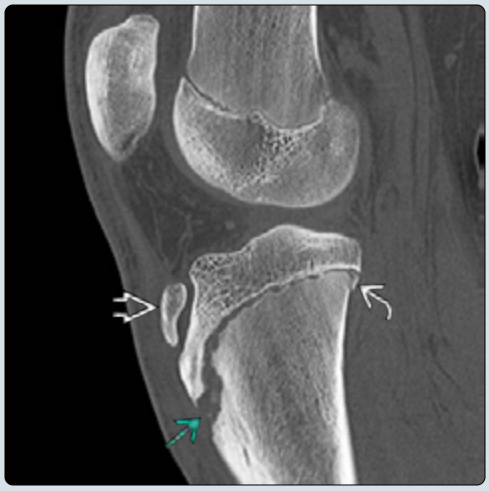
CLINICAL ISSUES

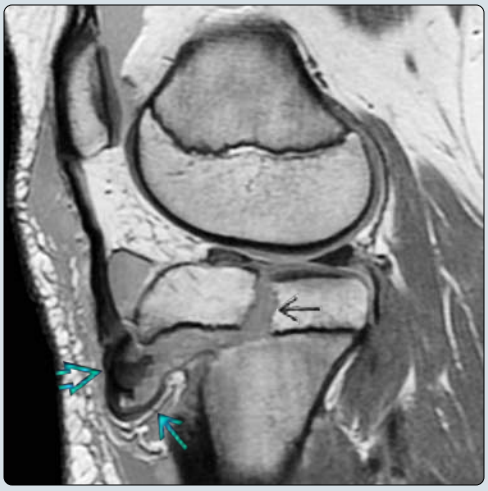
- Vast majority: Boys 13-17 years old
- Presentations include swelling, tenderness, knee held in mild flexion with inability to fully extend actively
- Nonoperative management for minimally displaced fractures limited to distal tibial tubercle
- Open reduction, internal fixation otherwise required to
  - Restore extensor mechanism alignment
  - Establish congruence of articular surface of tibia
- Complications in 25-30%: Bursitis, tubercle prominence > refracture, genu recurvatum, limb length discrepancy





(Left) *Lateral knee radiograph with mild internal rotation in a 17-year-old boy after a football injury shows moderate proximal displacement of a large tibial tubercle fragment with widening of the growth plate. A moderate joint effusion is noted.* **(Right)** *Lateral knee radiograph in a 15-year-old boy after a fall shows mild widening & irregularity of the growth plate deep to the tibial tubercle with posterior extension to the proximal tibial physis & metaphysis as a Salter-Harris II fracture.*





(Left) *Sagittal CT in a 14-year-old boy after a basketball injury shows mild widening & irregularity of the growth plate deep to the tibial tubercle with posterior extension to the metaphysis. A well-defined proximal fragment is likely chronic.* **(Right)** *Sagittal PD MR in a 15-year-old boy after a fall shows avulsion of a tibial tubercle fragment by the patellar tendon. The fracture extends proximally with a Salter-Harris III configuration. Stripped distal periosteum is entrapped in the fracture.*

Triplane Fracture

KEY FACTS

IMAGING

- Distal tibial fracture in 3 planes; may be true Salter-Harris IV (SH IV) or combinations of SH II & III
- Classic triplane fracture pattern
  - Coronal fracture plane through distal tibial metaphysis & diaphysis
  - Transverse fracture plane through physis (growth plate)
  - Sagittal fracture plane through epiphysis
- Intramalleolar extension may be intra- or extraarticular
- Oblique coronal plane of metaphysis/diaphysis may be hidden on lateral radiograph if nondisplaced & overlapping tibial-fibular interface
- CT more accurate for determining course & comminution of fracture, amount of displacement, & articular surface involvement

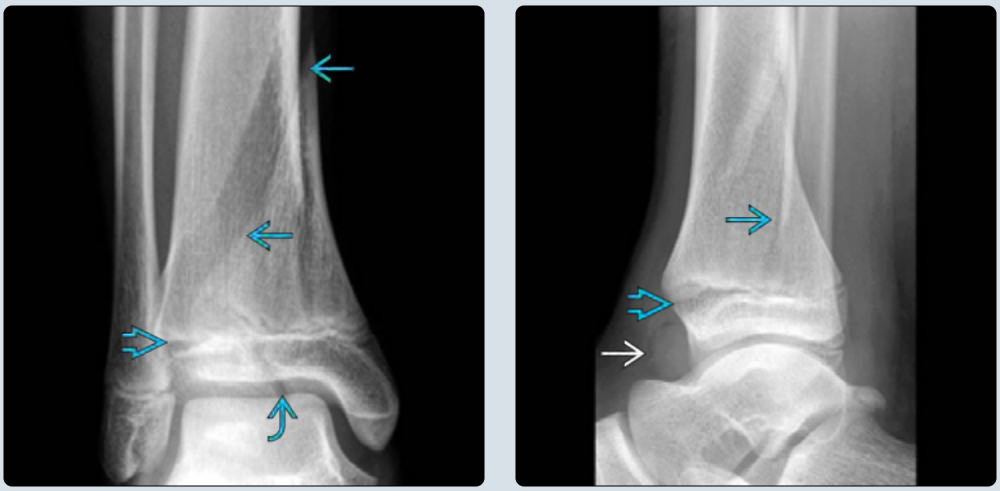
PATHOLOGY

- Fracture typically occurs around time of earliest physeal closure, which affects planes of fracture extension
  - Fusion begins in central tibial physis at Kump bump
  - Extends posteromedial then anterolateral
- Mechanism: External rotation
- Rapariz triplane fracture classification: Variations of 2-, 3-, or 4-part fractures for 6 possible subtypes

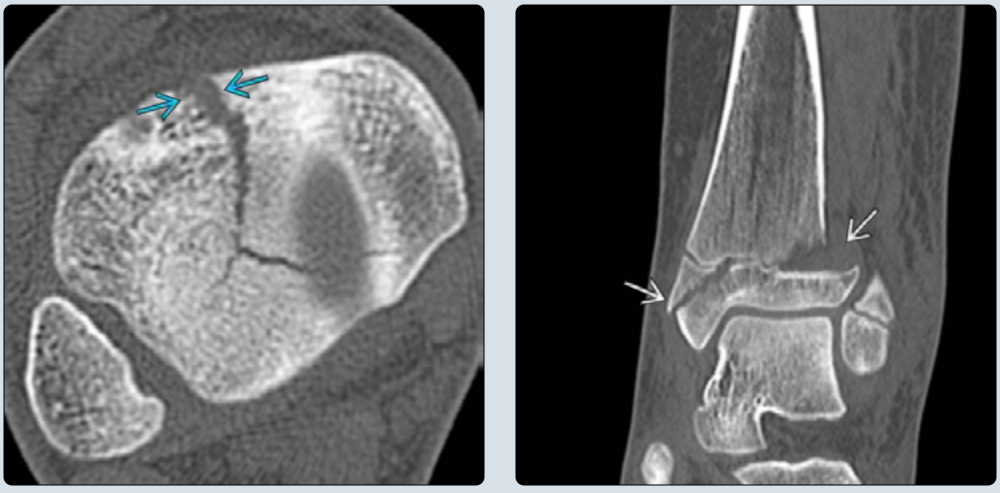
CLINICAL ISSUES

- 5-10% of pediatric intraarticular ankle fractures
- Typically affects adolescents within 18 months of tibial growth plate closure (12-15 years old)
  - 2-part triplane younger than 3-part triplane fractures
- Nonoperative treatment for ≤ 2 mm of displacement & extraarticular fractures
- Operative treatment for > 2 mm of articular step-off

(Left) *AP radiograph in a 15-year-old boy after a fall shows a triplane fracture of the distal tibia with the following fracture planes: Coronal oblique of the metaphysis & diaphysis, horizontal of the anterolateral physis, & sagittal of the epiphysis.* **(Right)** *Lateral radiograph of the same patient shows the coronal metadiaphyseal & horizontal anterior physeal components. Note the moderate tibiotalar joint effusion.*



(Left) *Axial bone CT in the same patient shows the predominantly sagittal fracture plane through the distal tibial epiphysis. In many cases, this plane allows the most accurate assessment of the articular surface distraction, which is a key determinant for treating with a closed vs. open reduction.* **(Right)** *Coronal bone CT in a 13 year old who sustained an ankle injury after falling from a scooter shows an extraarticular intramalleolar triplane fracture variant.*



TERMINOLOGY

Synonyms

- Adolescent triplane fracture, transitional injury

IMAGING

General Features

- Best diagnostic clue
  - Complex fracture of adolescent distal tibia involving metaphysis, physis, & epiphysis
    - Fracture components in transverse, sagittal, & coronal planes
- Location
  - Classic triplane fracture pattern
    - Coronal fracture plane through distal tibial metaphysis & diaphysis
    - Transverse fracture plane through physis (growth plate)
    - Sagittal fracture plane through epiphysis
      - Intramalleolar extension may be intra- or extraarticular
- Morphology
  - Distal tibial growth plate closes in predictable pattern
    - Begins in central tibial physis at Kump bump
    - Tibial physeal fusion posteromedial then anterolaterally
    - Fracture typically occurs around time of earliest physeal closure at Kump bump, which affects planes of fracture extension

Radiographic Findings

- Distal tibial fracture in 3 planes; may be true Salter-Harris IV (SH IV) or combinations of SH II & III
  - Oblique coronal plane of metaphysis/diaphysis may be hidden on lateral view if nondisplaced & overlapping tibial-fibular interface
- Soft tissue swelling & joint effusion

CT Findings

- Bone CT
  - Axial with sagittal & coronal reformations, ± 3D
  - More accurate over radiographs for determining course & comminution of fracture, amount of displacement, & articular surface involvement

PATHOLOGY

General Features

- Mechanism: External rotation

Staging, Grading, & Classification

- Rapariz triplane fracture classification: Variations of 2-, 3-, or 4-part fractures for 6 possible subtypes
  - 2-part fracture: Most common
    - True SH IV fracture
    - Either medial or lateral
  - 3-part fracture: Combination of fractures: SH II (AP view) & SH III (lateral view)
    - ± separation at Kump bump
  - 4-part fracture: Combinations of above
- 2- or 3-part intramalleolar variant
  - Depending on fracture line extension, fracture may be intra- or extraarticular
    - Type I: Intraarticular at junction of medial malleolus & tibial plafond
    - Type II: Intraarticular outside weight-bearing zone of plafond
    - Type III: Extraarticular

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain
  - Swelling
  - Inability to bear weight
- Other signs/symptoms
  - Bruising
  - Associated fibular fracture in 1/2 of patients

Demographics

- Age
  - Adolescent within 18 months of tibial growth plate closure
    - 12-15 years old
      - Males: 15-16 years old
      - Females: 13-14 years old
  - 2-part triplane younger than 3-part triplane fractures
- Gender
  - M > F
- Epidemiology
  - 5-10% of pediatric intraarticular ankle fractures

Treatment

- Nonoperative treatment
  - ≤ 2 mm of displacement & extraarticular fractures
    - Most 2-part fractures
  - Closed reduction with internal rotation of foot
    - Immobilization & long leg cast
- Operative treatment
  - > 2 mm of articular step-off
    - Most 3-part fractures
  - Internal fixation with screws, percutaneous K-wires

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Juvenile Tillaux Fracture

KEY FACTS

IMAGING

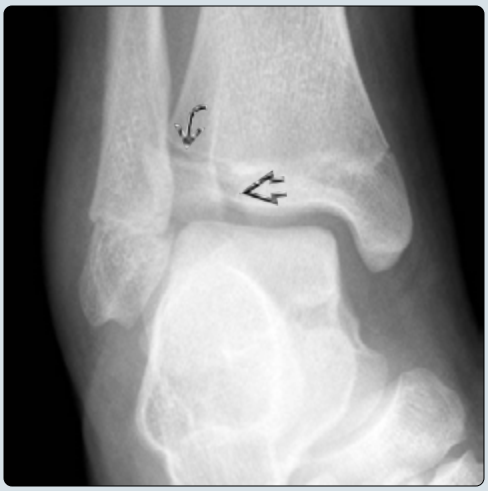
- Salter-Harris III fracture of anterolateral distal tibial physis & epiphysis
  - Widening of physis laterally
  - Contiguous sagittal fracture plane extending through epiphysis to tibiotalar joint
- Occurs after fusion of central & medial physis but before closure of anterolateral physis
  - Juvenile Tillaux fracture results from superimposed distraction of anterior inferior tibiofibular ligament during supination, external rotation
  - Ligament usually remains intact
- Radiographs typically diagnostic
- CT obtained to determine degree of articular surface distraction (> 2 mm necessitates operative fixation)
  - Reformatted images in 3 planes essential to fully characterize fracture

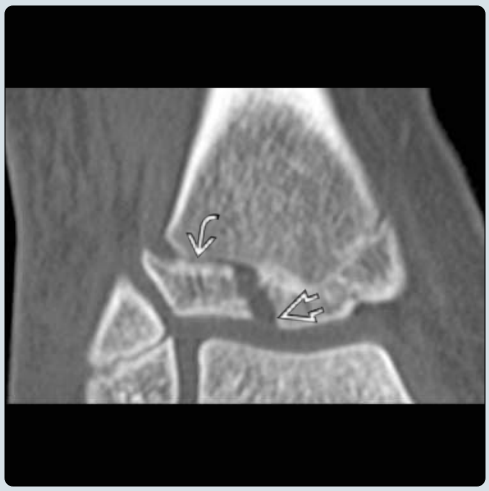
CLINICAL ISSUES

- 6% of pediatric ankle fractures
- Affects adolescents 0-18 months prior to distal tibial growth plate closure
  - Age range: 12-15 years old
  - Median age: 12 years in females, 14 years in males
- Nonoperative treatment (closed reduction + immobilization) with fracture displacement < 2 mm
- Internal fixation with screws & K-wires if articular surface fracture displacement > 2 mm
  - Smooth articular surface required to restore joint function, prevent premature degeneration
- Subsequent growth disturbance uncommon as fracture occurs after majority of physis has already closed

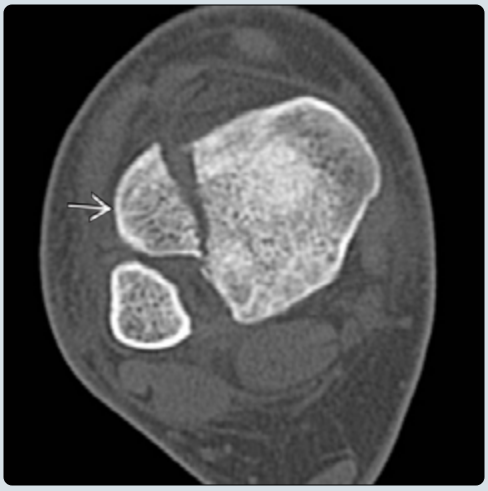
DIAGNOSTIC CHECKLIST

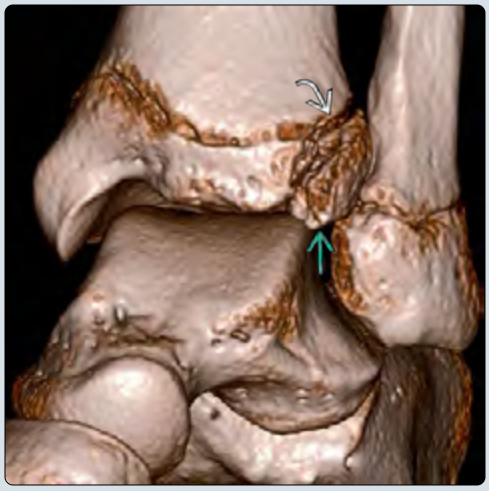
- Differentiate from triplane fracture
  - Salter-Harris IV fracture with coronal metadiaphyseal fracture plane





(Left) *AP radiograph of the right ankle in a 13-year-old patient after a trampoline injury shows a Salter-Harris III fracture of the distal tibia. The fracture extends horizontally through the lateral physis & vertically through the epiphysis to the articular surface, typical of a juvenile Tillaux fracture.* **(Right)** *Coronal bone CT in the same patient demonstrates the physeal & epiphyseal fracture planes. The tibial articular surface gap is > 2 mm, requiring surgical fixation.*





(Left) *Axial bone CT in the same patient shows distraction & rotation of the anterolateral epiphyseal fracture fragment.* **(Right)** *Coronal oblique 3D bone CT of the left ankle in a 15-year-old patient shows a displaced lateral tibial epiphyseal fragment with disruption of the subjacent physis, consistent with a tibial Salter-Harris III (or juvenile Tillaux) fracture. The epiphyseal fragment is rotated & distracted by ~ 6 mm at the articular surface.*

TERMINOLOGY

Synonyms

- Tillaux fracture

IMAGING

General Features

- Best diagnostic clue
  - Widening of distal tibial physis laterally with contiguous sagittal fracture plane extending through epiphysis to tibiotalar joint
- Location
  - Anterolateral aspect of distal tibial physis & epiphysis
- Morphology
  - Distal tibial growth plate closes in predictable pattern
    - Begins in anteromedial tibial physis at Kump bump (or Kump hump)
    - Anterolateral distal tibial growth plate closes last
  - Juvenile Tillaux fracture
    - Occurs after fusion of central & medial physis but before closure of anterolateral physis
    - Salter-Harris III fracture results from distraction of anterior inferior tibiofibular ligament
      - Ligament usually remains intact

Radiographic Findings

- Salter-Harris III fracture of anterolateral distal tibial epiphysis
- ± lateral displacement of fracture fragment
- Soft tissue swelling ± joint effusion

CT Findings

- Obtained to determine degree of articular surface distraction
  - > 2 mm necessitates operative fixation
- Reformatted images in 3 planes essential to fully characterize fracture
  - Width & location of greatest distraction
  - Degree of vertical step-off
  - Presence of loose intraarticular fragments
  - Associated soft tissue injury

MR Findings

- Generally not indicated
- Will show associated ligamentous injury or subsequent growth plate disturbance (uncommon)

Imaging Recommendations

- Best imaging tool
  - AP, lateral, & mortise/oblique radiographic views
    - Fracture may be obscured by fibula on AP view
  - Bone CT to determine articular surface distraction

PATHOLOGY

General Features

- Mechanism: Avulsion of anterolateral tibial epiphysis by anterior inferior tibiofibular ligament
  - Due to external rotation, supination superimposed on partially fused growth plate

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Anterior &/or lateral ankle pain, tenderness
  - Lateral swelling
  - Inability to bear weight
- Other signs/symptoms
  - Bruising, limp, external rotation of foot

Demographics

- Age
  - Affects adolescents 0-18 months prior to distal tibial growth plate closure
    - 12-15 years old
    - Median age: 12 years in females, 14 years in males
- Gender
  - More common in girls (F:M = 4:1)
- Epidemiology
  - 6% of pediatric ankle fractures

Natural History & Prognosis

- Subsequent growth disturbance uncommon as fracture occurs after majority of physis has already closed

Treatment

- Nonoperative treatment (closed reduction + immobilization) with fracture displacement < 2 mm
- If articular surface fracture displacement > 2 mm
  - Internal fixation with screws & K-wires
    - Smooth articular surface required to restore joint function, prevent premature degeneration
  - Alternatively, percutaneous pinning with arthroscopic assistance may be considered

DIAGNOSTIC CHECKLIST

Consider

- Differentiate from triplane fracture
  - Salter-Harris IV fracture with coronal metadiaphyseal fracture plane

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Osteomyelitis

KEY FACTS

IMAGING

- Long bone metaphyses 70% (femur > tibia > humerus), short bones 6%, pelvis 5%, spine 2%
  - Metaphysis or equivalent > epiphysis, diaphysis
  - Multifocal in 10% overall but 22% in neonates
- Absence of radiographic bone findings does not exclude early osteomyelitis
  - Earliest finding: Soft tissue swelling next to bone
  - Bone destruction, periosteal reaction by 7-14 days
- MR: Best advanced imaging choice if diagnosis unclear with localized symptoms or concern for complications
  - T1: Poorly defined metaphyseal marrow abnormality
  - T2 FS/STIR: Bright marrow, periosteal, & soft tissue edema; ± adjacent joint effusion
  - T1 C+ FS: Rim-enhancing abscesses (soft tissue, subperiosteal, intraosseous); ↓ enhancement of otherwise occult unossified epiphyseal cartilage lesions in infants

TOP DIFFERENTIAL DIAGNOSES

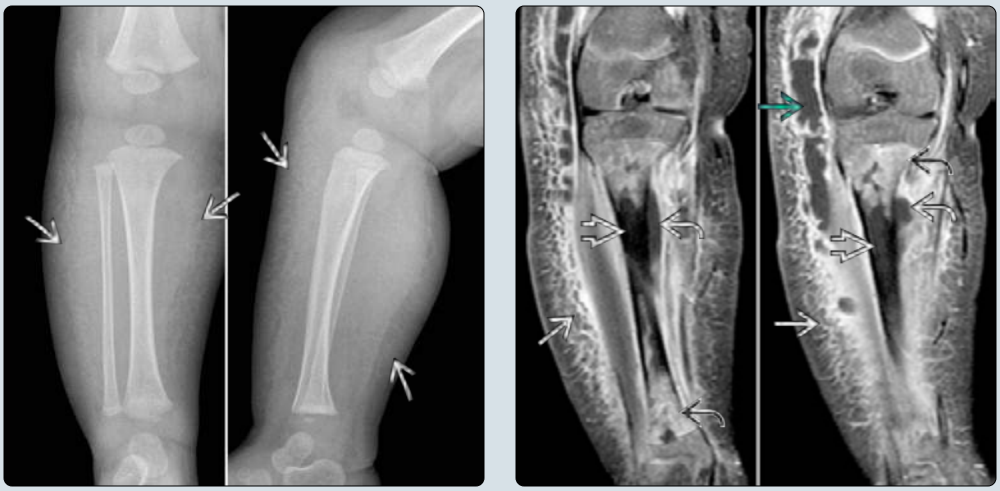
- Ewing sarcoma
- Neuroblastoma metastases
- Langerhans cell histiocytosis
- Septic arthritis
- Bone infarct
- Leukemia
- Chronic recurrent multifocal osteomyelitis

PATHOLOGY

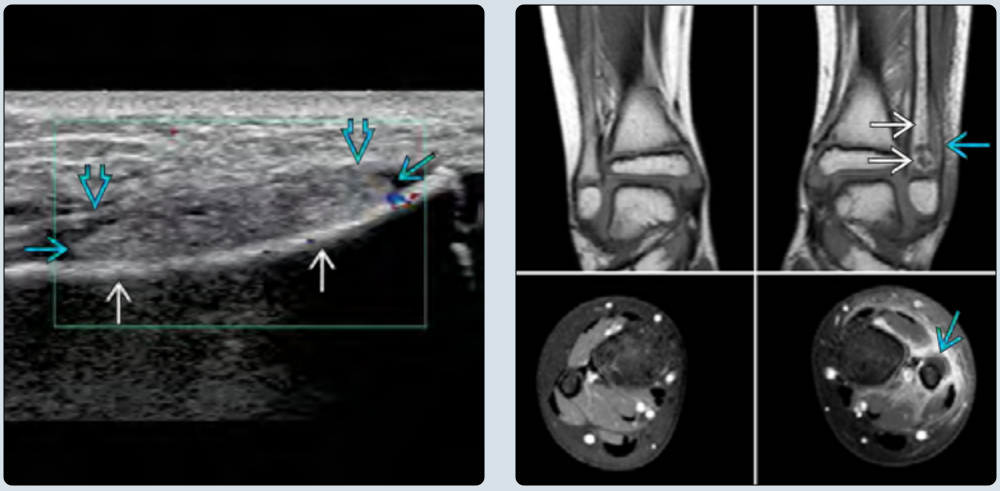
- Hematogenous source most common
- *Staphylococcus aureus* > 80-90% of cases
  - Identification of infectious agent often fails despite blood cultures & site sampling

CLINICAL ISSUES

- Fever, pain, limp, tenderness, swelling
- Presentation often nonspecific, may delay diagnosis
- Treat with IV then PO antibiotics + abscess drainage



(Left) *AP (left) & lateral (right) radiographs of a 9-month-old infant show extensive subcutaneous edema of the lower leg with blurring of soft tissue planes but no discrete bony abnormality.* **(Right)** *Coronal T1 C+ FS MR images show foci of decreased marrow enhancement from an intraosseous abscess with adjacent increased marrow enhancement from infected or reactive viable marrow. Subperiosteal & soft tissue fluid collections & overlying fat edema are noted in this patient with MRSA osteomyelitis.*



(Left) *Longitudinal color Doppler ultrasound in a 6 year old with fever & lower extremity swelling shows an echogenic avascular collection that is lifting the periosteum off of the cortex.* **(Right)** *Coronal T1 (top) & axial T1 C+ FS (bottom) MR images in the same patient show heterogeneous marrow of the distal left fibula with a rim-enhancing subperiosteal abscess. Surrounding soft tissue edema is also noted in this patient with MSSA osteomyelitis.*

TERMINOLOGY

Definitions

- Osteomyelitis: Bone infection, most commonly bacterial

IMAGING

General Features

- Best diagnostic clue
  - Radiographs: Nonspecific soft tissue edema in setting of limp, pain, swelling, &/or fever (< 7-10 days)
  - MR: Heterogeneous, poorly defined metaphyseal or metadiaphyseal marrow signal abnormalities + subperiosteal fluid & soft tissue edema
- Location
  - Long bone metaphyses 70% (femur > tibia > humerus), short bones 6%, pelvis 5%, spine 2%
    - Metaphysis or equivalent > epiphysis, diaphysis
      - Bone adjacent to growth cartilage (e.g., synchondrosis or apophysis) considered metaphyseal equivalent; typically affected in ages 6-16 years
  - Multifocal in 10% overall but 22% in neonates

Radiographic Findings

- Absence of findings does not exclude early osteomyelitis
- Earliest finding: Soft tissue swelling next to bone
  - Displacement or obliteration of fat planes
  - Reticulation of subcutaneous fat
- Lytic bone lesion in 7-14 days (or longer) after onset
  - Vague lucency → permeation → destruction
- Periosteal reaction seen by 7-14 days
- Healing changes on follow-up may look bizarre due to extensive bone involvement &/or superimposed pathologic fracture (due to ↑ activity on weak bone)
- Subacute: Lucent lesion + sclerotic rim (Brodie abscess)
- Chronic: Sclerosis or mixed sclerotic/lucent foci
  - ± cloaca (lucent drainage tract through cortex)
  - ± sequestrum (radiodense necrotic bone)
  - Garré sclerosing osteomyelitis (thick cortex)

Ultrasonographic Findings

- Sensitive for drainable fluid collections
  - Joint effusions; soft tissue &/or subperiosteal fluid
- Cortical bone changes rarely seen

MR Findings

- T1WI
  - Poorly defined, heterogeneous dark marrow (due to loss of normal fatty marrow signal)
  - Subperiosteal foci of bright fat (from cortical disruption & marrow lipocyte necrosis)
  - Penumbra sign (in subacute Brodie abscess): ↑ T1 signal rim of granulation tissue
- T1WI FS
  - Bright marrow not more specific for infarction than infection in setting of sickle cell disease
- T2WI FS/STIR
  - Bright marrow + soft tissue edema
    - Soft tissue changes usually moderate to marked
  - Discrete soft tissue & subperiosteal fluid collections
    - Elevated thin, dark periosteum
    - Disrupted dark cortex
  - ± adjacent joint effusion
    - Septic arthritis more likely than reactive fluid
      - Spread of infection to joint ↓ > 1-2 years of age
  - Chronic: Dark signal sclerosis, fibrosis
- T1WI C+ FS
  - ↑ marrow enhancement: Hyperemia/edema
  - ↓ marrow enhancement: Pressure necrosis vs. frank pus
  - Rim-enhancing soft tissue & subperiosteal abscesses
  - Brodie abscess: Central nonenhancement (pus), inner rim of ↑ enhancement (granulation tissue), outer low signal rim (sclerosis)

Nuclear Medicine Findings

- Bone scan: ↑ uptake in angiographic, blood pool, & delayed phases: 80-94% sensitive
  - Positive in 24-72 hours; can demonstrate multiple sites
  - Central photopenia with bone infarct or abscess
- PET: ↑ metabolic activity; 95% sensitive

Imaging Recommendations

- Best imaging tool
  - MR most sensitive & specific modality for early infection with localized symptoms
    - Confirms diagnosis or delineates alternative processes
    - Shows drainable abscesses & intraspinal extension
  - Bone scintigraphy helpful if site & diagnosis unclear
  - Whole-body MR helpful for localizing drainable collections in ill patient with known aggressive infection
- Protocol advice
  - T2 FS or STIR MR crucial for edema/fluid detection
  - T1 C+ FS MR delineates drainable fluid & otherwise occult foci of epiphyseal cartilage involvement in infancy
    - Contrast of questionable utility with otherwise normal study beyond 18 months of age

DIFFERENTIAL DIAGNOSIS

Ewing Sarcoma

- Aggressive lytic diaphyseal/metadiaphyseal lesion, most common in patients > 5 years old
- Sharply marginated interface of intramedullary tumor with normal fatty marrow on T1 MR; large heterogeneously enhancing soft tissue mass with contrast

Neuroblastoma Metastases

- Aggressive but frequently subtle lytic lesions, most common in patients < 5 years old
- Calcified primary suprarenal/paraspinal tumor

Langerhans Cell Histiocytosis

- Punched-out, well-defined lytic lesions + enhancing mass
- Flat bones & spine frequently involved

Septic Arthritis

- Effusion + enhancing mildly thickened synovium
- Adjacent marrow & soft tissue edema favor infected joint

Bone Infarct

- Acute: Focal poorly defined marrow edema with nonenhancement & mild adjacent soft tissue changes
- Chronic: Serpentine medullary sclerosis on radiographs; MR shows characteristic double line sign (low/high T2 signal)

Leukemia

- Metaphyseal lucent bands &/or permeative lytic lesions in patients < 10 years old
- Diffuse marrow replacement on T1 MR classic, not specific

Chronic Recurrent Multifocal Osteomyelitis

- Noninfectious episodic bone inflammation
- Mixed bubbly lucent & sclerotic metaphyseal or metadiaphyseal lesions of femur, tibia; spine, pelvis, clavicle, & mandible also typical

PATHOLOGY

General Features

- Pathophysiology
  - Hematogenous seeding > > penetrating injury or contiguous spread
    - After infancy, transphyseal vessels resolve
      - Avascular physis maintains relative barrier between epiphysis & metaphysis, preventing continuous epiphyseal & joint spread
      - Barrier nonexistent in infants or after skeletal maturity → ↑ rates of septic arthritis
    - Slow flow through looping metaphyseal venules
      - Primary sites for bacterial lodging
    - Intramedullary infection → edema, vascular congestion → ↑ pressure → transcortical spread
    - Periosteal attachment tight at physis, loose at metaphysis/diaphysis in children
      - Marked elevation by pus or tumor possible
- Organisms
  - Identified in only 35-66% of needle aspirates, 36-55% of blood cultures
  - *Staphylococcus aureus* > 80-90% of cases (> 55% due to methicillin-resistant strains)
    - Panton-Valentine leukocidin: Necrotizing toxin secreted by MRSA > MSSA → more invasive infections
      - Persistent bacteremia despite IV antibiotics
      - ↑ rates of shock, septic emboli, deep venous thrombosis, & fluid collections requiring drainage
  - Streptococcus in ~ 10%: Group A β hemolytic, *S. pyogenes*, *S. pneumoniae*
  - Children 6 months to 3 years of age: *Kingella kingae*
    - Relatively ↓ bone & soft tissue reaction & ↑ epiphyseal cartilage lesions vs. other organisms
    - PCR much more sensitive than culture
  - Neonates: Group B *Streptococcus*
  - Sickle cell disease: *Salmonella*
  - Immunocompromised: *Streptococcus pneumoniae*, tuberculosis
  - Penetrating foot trauma: *Pseudomonas aeruginosa*
  - Purpuric rash, shock: Meningococcemia (severe infarctions → amputation; less severe → growth arrest)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, ↓ range of motion, ↓ weight bearing, tenderness, swelling, fever
  - Minor trauma history in 1/3 of patients
- Labs: ↑ ESR, CRP > ↑ WBC

Demographics

- Age
  - Primarily disease of infants & young children
    - 1/2 of cases occur before 5 years of age
- Epidemiology
  - 1/5,000 < 13 years old in USA; 1/800-10,000 elsewhere

Natural History & Prognosis

- Complications include septic arthritis, deep venous thrombosis, fracture, septic emboli, multisystem failure, growth disturbance
  - More likely with treatment delay of > 4 days

Treatment

- Identify infectious agent: Image-guided needle aspiration or open surgical biopsy + blood culture
- Antibiotics (IV followed by PO), pain management
- Surgery/intervention: Abscess (intraosseous, subperiosteal, soft tissue) drainage, sequestrectomy, management of sinus tracts & pathologic fractures

DIAGNOSTIC CHECKLIST

Consider

- Imaging after treatment: Uncomplicated osteomyelitis may require 6 months for MR normalization
  - Nonspecific for ongoing bone infection vs. resolving inflammation/healing
  - Drainable collections can be sampled to guide therapy

Image Interpretation Pearls

- For aggressive bone masses, generally consider
  - Infection: Nonenhancing fluid > solid enhancing tissue; moderate to marked surrounding soft tissue edema
  - Tumor: Solid enhancing tissue > nonenhancing fluid; surrounding soft tissue edema typically mild

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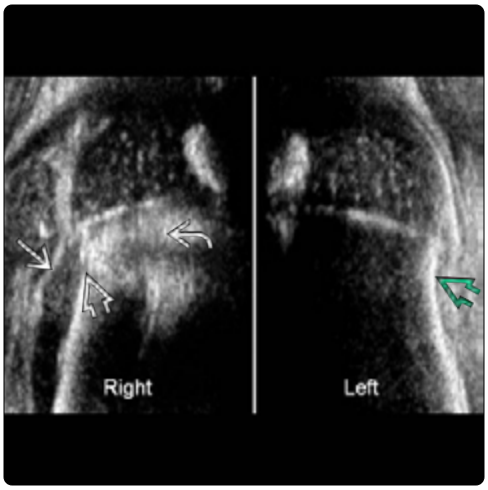
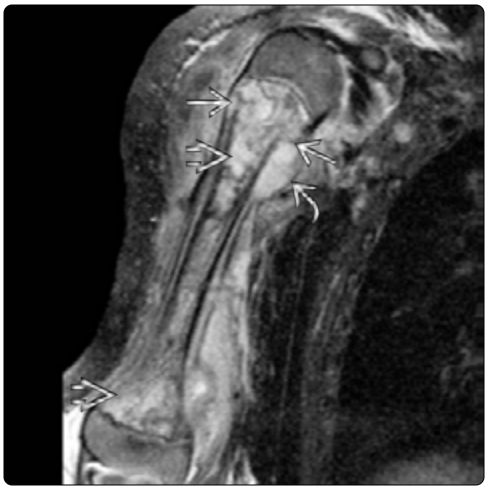
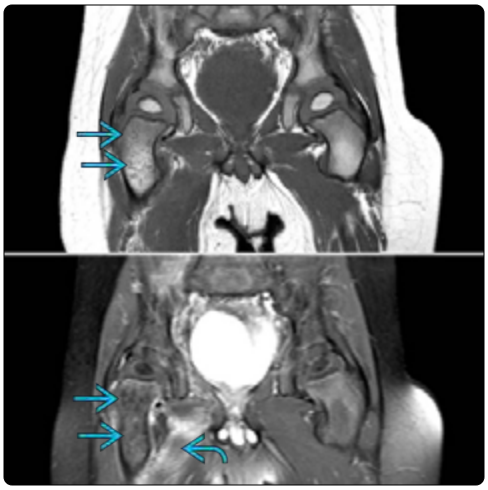
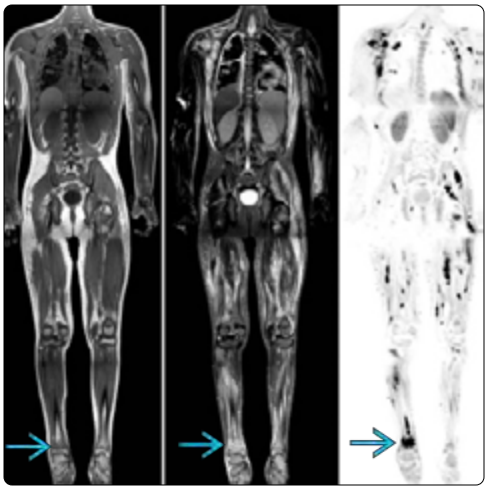


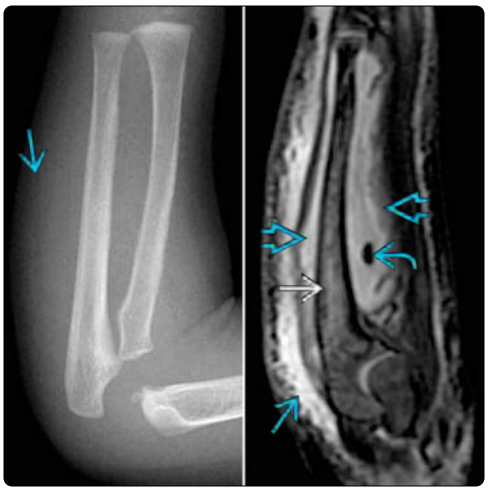


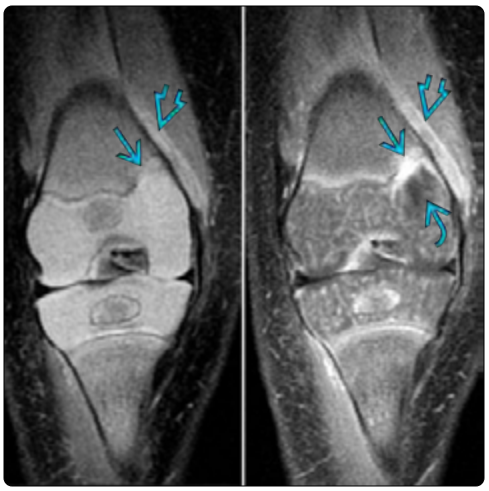
(Left) *Coronal ultrasound of the proximal right humerus in a 5-month-old boy shows a subperiosteal fluid collection with disruption of the metaphyseal cortex & disturbed underlying medullary echotexture. Compare the normal left humeral sonographic architecture.* **(Right)** *Coronal STIR MR of the same patient with osteomyelitis shows abnormal fluid signal throughout the right humerus plus a nondisplaced pathologic metaphyseal fracture. A subperiosteal fluid collection is noted.*





(Left) *Coronal T1 (top) & T1 C+ FS (bottom) MR images in a 1 year old with a fever & a limp show abnormal poorly defined marrow heterogeneity of the proximal right femur with surrounding soft tissue edema, typical of osteomyelitis.* **(Right)** *Coronal T1 (left), STIR (middle), & DWIBS (right) MR images in a 9 year old with MRSA bacteremia & septic emboli show right distal tibial osteomyelitis, pulmonary nodules, & numerous foci of myositis & fasciitis with muscular abscesses.*





(Left) *Lateral radiograph (left) & sagittal STIR MR (right) in an infant with ulnar osteomyelitis show soft tissue edema, a large subperiosteal abscess, & marrow heterogeneity. A small subperiosteal globule followed fat signal on all sequences, consistent with marrow lipocyte necrosis.* **(Right)** *Coronal PD FS (left) & T1 C+ FS (right) MR images in a neonate with femoral osteomyelitis show focal loss of normal low signal ZPC with poor enhancement of unossified epiphyseal cartilage & overlying soft tissue edema.*

Syphilis

KEY FACTS

TERMINOLOGY

- Congenital syphilis (CS): Transmission of *Treponema pallidum* to fetus from infected mother
  - Early-onset CS: Clinically manifests < 2 years of age
  - Late-onset CS: Clinically manifests > 2 years of age

IMAGING

- Usually widespread, symmetric
  - Can be asymmetric or solitary
- Periosteal reaction most common finding
- Metaphyseal lesions
  - Nonspecific lucent bands with intact ZPC typical
    - Can have serrated/sawtooth metaphyses
  - Wimberger corner sign: Destruction of proximal medial tibias with sparing of first few mm of new bone
- Saber shins: Anterior tibial cortical thickening & bowing (older children)
- Pathological fractures, usually metaphyseal

TOP DIFFERENTIAL DIAGNOSES

- Bacterial osteomyelitis
- Neuroblastoma or leukemia
- Physiologic periosteal reaction
- Infantile myofibromatosis
- Child abuse

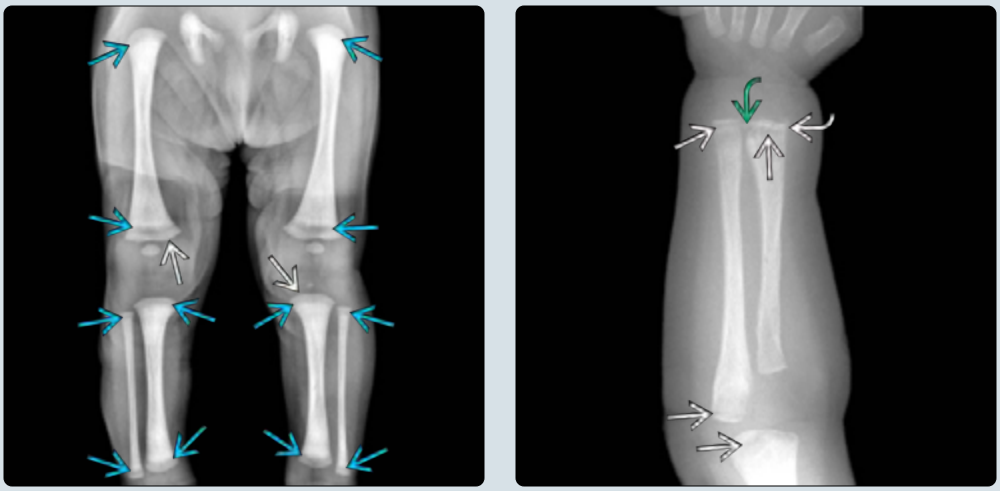
CLINICAL ISSUES

- Fetal/perinatal demise in 40% of infants with CS
- 2/3 of infants with CS asymptomatic at birth
- Symptoms: Hepatosplenomegaly, rhinitis ("snuffles"), rash
  - Parrot pseudoparalysis: Not moving painful extremity
- Prevention of CS: Treatment of seropositive mothers
- Treatment of CS: Intravenous penicillin for infants

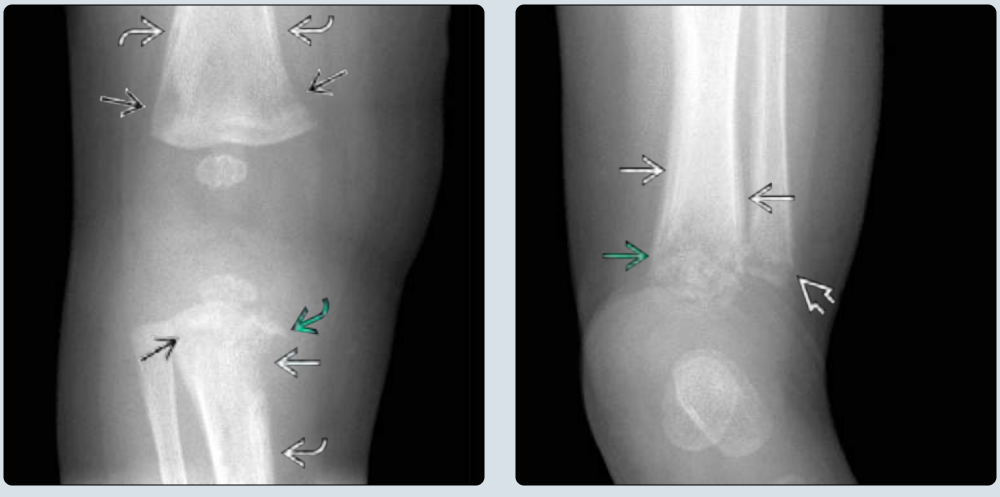
DIAGNOSTIC CHECKLIST

- Consider CS in infant with widespread polyostotic findings
- Infant with CS & multiple fractures may mimic abuse

(Left) *Frontal radiograph of the lower extremities in a 3-week-old girl with acute congenital syphilis (CS) shows metaphyseal lucent bands in all of the long bones, though ZPCs are intact. This finding is nonspecific, as systemic stress, neuroblastoma, & leukemia can cause a similar appearance.* **(Right)** *AP radiograph shows metaphyseal lucent bands in the radius, ulna, & humerus. Periosteal reaction & a destructive metaphyseal lesion are also seen in the distal radius.*



(Left) *AP knee radiograph of an infant with acute CS shows a destructive lesion in the medial tibial metaphysis, though the first few mm of newly formed bone are spared. When bilateral, this is the Wimberger corner sign, which is highly suggestive of CS. Other metaphyseal lucencies & periosteal reaction are also noted.* **(Right)** *Newborn ankle radiograph of CS shows a fracture through a lucent band in the distal fibula. Periostitis & permeation are seen in the distal tibia. Fractures in CS can be confused with abuse.*



TERMINOLOGY

Definitions

- Congenital syphilis (CS): Transmission of *Treponema pallidum* (*T. pallidum*) to fetus from infected mother
  - Early-onset CS: Clinically manifests < 2 years of age
  - Late-onset CS: Clinically manifests > 2 years of age
- "The great imitator" (coined by Sir William Osler)

IMAGING

General Features

- Best diagnostic clue
  - Infant with widespread metaphyseal findings
  - Wimberger corner sign: Bilateral destruction of proximal medial tibias
    - Spares first few mm of newly formed bone
    - Highly suggestive but not specific for CS
- Location
  - Often widespread but can involve single bone

Radiographic Findings

- Periosteal reaction most common finding
  - Typically symmetric
- Long bones
  - Metaphyses: Typically first area of involvement
    - Nonspecific horizontal lucent bands, classically with sparing of thin radiodense periphyseal zones of provisional calcification (ZPC)
      - Can see metaphyseal irregularity (serrated, sawtooth)
    - Destructive lucent lesion
      - Tibia > femur > humerus
      - Wimberger corner sign: Upper medial tibias
  - Diaphyses
    - Cortical thickening ± destructive lesions
      - Saber shin: Anterior tibial cortical thickening/bowing (late childhood)
- Pathological fractures, frequently multiple
  - Most common at metaphysis
- Skull: Multiple lytic calvarial lesions
- Chest: Diffuse pulmonary opacities

DIFFERENTIAL DIAGNOSIS

Bacterial Osteomyelitis

- Usually monostotic

Leukemia

- Symmetric metaphyseal lucent bands

Neuroblastoma

- Diffuse bone metastases (asymmetric)

Systemic Stress

- May cause metaphyseal lucent bands with intact ZPC

Rickets

- Metaphyseal cupping, fraying, & splaying
- Loss of ZPC

Physiologic Periosteal Reaction

- Symmetric & smooth, of entire diaphysis

Infantile Myofibromatosis

- Relatively symmetric bubbly metaphyseal lucent lesions of infant, often with ≥ 1 soft tissue myofibroma

Child Abuse

- Healing classic metaphyseal lesion can mimic CS

PATHOLOGY

General Features

- Typically due to transplacental transmission

Microscopic Features

- Metaphyseal lucent bands may be secondary to
  - Stress response from systemic disease
  - Syphilitic granulation tissue
- Focal erosions due to syphilitic granulation tissue

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 2/3 of infants with CS asymptomatic at birth
    - Untreated CS typically shows symptoms by 5 weeks
  - Hepatosplenomegaly, rhinitis ("snuffles"), rash
- Other signs/symptoms
  - Early-onset (0-2 years) CS
    - Parrot pseudoparalysis: Immobile limb 2° to pain
      - Radiograph of limb shows destructive lesion
  - Late onset (2 to ~ 30 years) CS
    - Bone changes: Frontal bossing, saddle nose, maxillary hypoplasia, saber tibia, gummatous periostitis

Demographics

- Epidemiology
  - Rates of CS ↑ in developed countries
    - Follows ↑ rates of acquired syphilis

Natural History & Prognosis

- Fetal/perinatal demise in 40% of infants with CS
- Early CS bone lesions usually resolve with treatment
  - Typically subsequent normal growth

Treatment

- Prevention of CS: Treatment of seropositive mothers
- Intravenous penicillin for infected infants

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Consider CS in infant with widespread polyostotic findings
- Infant with CS & multiple fractures may mimic abuse

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Septic Arthritis

KEY FACTS

TERMINOLOGY

- Septic arthritis: Microbial (typically bacterial) invasion of joint leading to inflammation & purulence

IMAGING

- Best clue: Joint effusion in non-weight-bearing child with fever > 38.5°C + ↑ serum inflammatory markers
- Radiographs: Displacement of fat pads ± widened joint
- US: Highly sensitive for fluid distending joint capsule
  - Complexity & volume do not predict/exclude infection
- MR: Nonspecific joint fluid with synovial thickening & enhancement
  - Findings favoring septic arthritis over transient synovitis
    - Presence of marrow &/or soft tissue edema
    - ↓ enhancement/perfusion of articular epiphysis

TOP DIFFERENTIAL DIAGNOSES

- Transient synovitis
- Juvenile idiopathic arthritis
- Trauma
- Reactive effusion due to adjacent bone pathology

PATHOLOGY

- Mechanism: Hematogenous spread vs. direct spread from adjacent osteomyelitis or puncture wound
- Most common organisms overall: *Staphylococcus aureus* > streptococcal species
  - Neonates: Group B streptococci, gram-negative rods, *Neisseria gonorrhoeae*
  - < 4 years of age: *Kingella kingae* most common

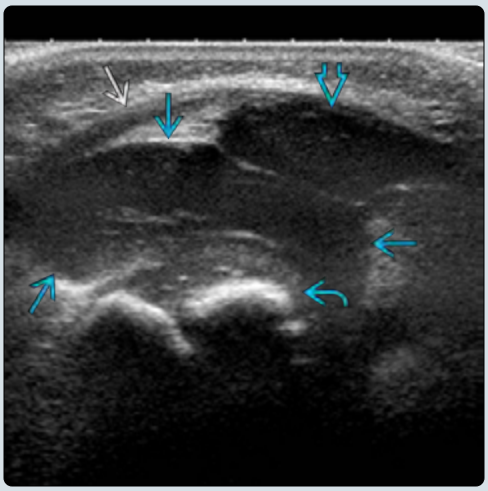
CLINICAL ISSUES

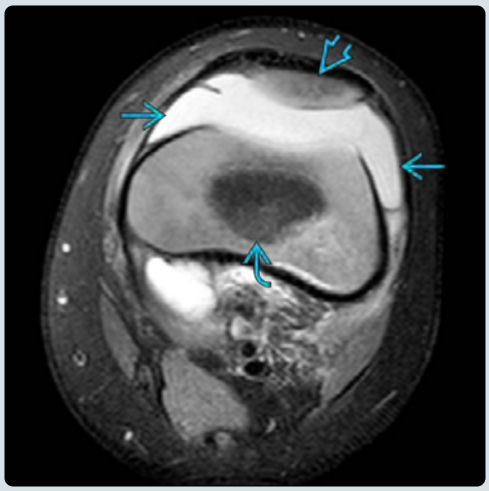
- Treatment: Emergent arthroscopy/arthrotomy with joint irrigation + IV antibiotics to prevent long-term sequelae

DIAGNOSTIC CHECKLIST

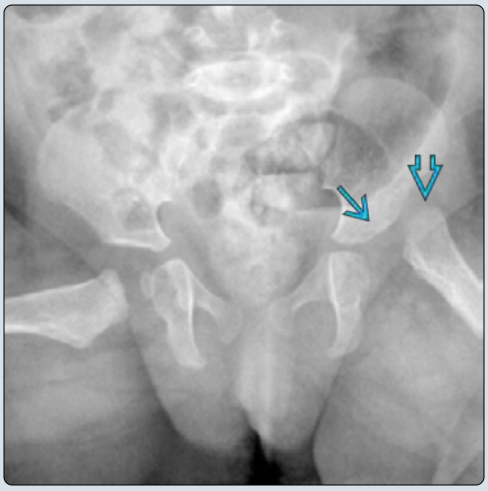
- Synovitis has many etiologies; always consider infection
  - Imaging alone cannot exclude infection of joint fluid
  - Joint aspiration required for 100% confidence

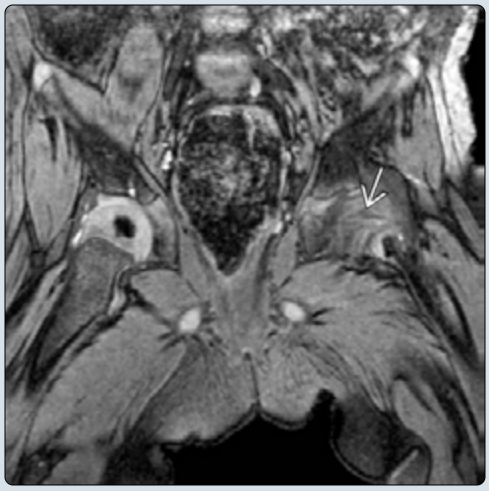
(Left) *Longitudinal US of the anterior midline knee in a 1 year old with swelling shows a large hypoechoic joint effusion deep to the quadriceps tendon & unossified patella. The partially ossified distal femoral epiphysis lies deep to the effusion.* **(Right)** *Axial T2 FS MR of the same patient shows the effusion lifting the unossified patellar cartilage off of the partially ossified distal femoral epiphysis. Septic arthritis was confirmed upon joint drainage.*





(Left) *Frog-leg lateral radiograph of the pelvis in a 1 year old with a history of left hip neonatal septic arthritis shows sequelae of delayed treatment: The left acetabulum is dysplastic with dislocation of the femur & no visible femoral head ossification center. The right hip is normal.* **(Right)** *Coronal T2* GRE MR of the same patient shows complete destruction of the left femoral head due to prior septic arthritis. This is one of the most feared consequences of septic arthritis, leading to lifelong morbidity.*





TERMINOLOGY

Definitions

- Septic arthritis: Microbial (typically bacterial) invasion of joint leading to inflammation & purulence

IMAGING

General Features

- Location
  - Affects lower extremity joints in ≥ 75% of cases
  - Multiple joints in ~ 10-15%

Radiographic Findings

- Sensitivity for fluid distending joint capsule varies on location: Knee, ankle, elbow >> hip, shoulder
  - Displacement of fat pads ± widening of joint space, soft tissue edema with blurred fat-muscle interfaces

Ultrasonographic Findings

- Anechoic to complex fluid distending joint capsule; complexity & volume do not predict sterile vs. infected fluid
- Synovial thickening, ± hyperemia

MR Findings

- Nonspecific synovitis (as with many etiologies)
  - Homogeneously bright T2/STIR fluid in joint
  - Synovial thickening & enhancement
- Findings favoring septic arthritis over transient synovitis
  - Presence of marrow &/or soft tissue edema
  - ↓ enhancement/perfusion of articular epiphysis, especially in tight joint (e.g., femoral head at hip)
- Findings favoring septic arthritis over reactive fluid secondary to adjacent metaphyseal osteomyelitis
  - Epiphyseal marrow edema, surrounding soft tissue edema, epiphyseal nonenhancement

Imaging Recommendations

- Best imaging tool
  - US: Highly sensitive for joint fluid; ~ 5% false-negative rate in first 24 hours
  - MR ± contrast
    - Best tool for detecting adjacent bone & soft tissue infections/collections
    - Can detect other diagnoses

DIFFERENTIAL DIAGNOSIS

Transient Synovitis

- Typically without fever or ↑ serum inflammatory markers
- Nonspecific MR appearance of synovitis
  - Joint fluid + mild synovial thickening & enhancement
  - Lacks surrounding marrow or soft tissue edema

Juvenile Idiopathic Arthritis

- ± "rice bodies": Numerous, tiny low T2 signal intensity bodies of uniform size throughout joint fluid

Trauma

- ± marrow & soft tissue edema, ligament & cartilage injuries

Reactive Effusion Due to Adjacent Bone Pathology

- Osteomyelitis, bone tumor, Legg-Calvé-Perthes

PATHOLOGY

General Features

- Mechanisms of joint seeding
  - Hematogenous spread: Vascularized synovium lacking basement membrane allow bacterial entry
  - Direct spread: Adjacent osteomyelitis, puncture wound
- Causative organisms
  - *Staphylococcus aureus* > streptococcal species overall
  - Neonates: Group B streptococci, gram-negative rods, *Neisseria gonorrhoeae*
  - < 4 years of age: Gram-negative bacteria, *Kingella kingae*
    - *K. kingae* often lacks systemic markers of inflammation
- Osteocartilaginous destruction caused by
  - Neutrophil & bacterial proteolytic enzymes → articular & unossified epiphyseal cartilage damage
  - ↑ joint pressure by pus → ↓ perfusion of articular epiphysis → ischemic injury

CLINICAL ISSUES

Presentation

- Fever, pain, swelling, ↓ motion, non-weight-bearing
- Kocher criteria: ↑ likelihood for hip septic arthritis over transient synovitis with ↑ number of predictors
  - Fever > 38.5°C, non-weight-bearing, WBC > 12 x 10^9 cells/L, ESR ≥ 40 mm/hour; later addition to original criteria: CRP > 20 mg/L
    - With all present, specificity for septic arthritis ~ 60-99%

Demographics

- ~ 6.5% of pediatric arthritis; peak age: ~ 2-3 years

Natural History & Prognosis

- Long-term sequelae in up to 40%
  - Limited motion, dislocation, degeneration, ankylosis, limb length discrepancy, avascular necrosis

Treatment

- Emergent arthroscopy or arthrotomy + joint irrigation
- IV antibiotics for 2-4 days followed by oral antibiotics for 10 days; longer course for concomitant osteomyelitis

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Transient Synovitis

KEY FACTS

TERMINOLOGY

- Idiopathic self-limited inflammation of pediatric hip
- Synonyms: Toxic synovitis

IMAGING

- Radiographs: Low sensitivity for hip joint effusion; look for convex gluteal fat pad & medial joint space widening
- US: Highly sensitive for joint fluid
  - Distention of joint capsule by anechoic, hypoechoic, or complex fluid
  - ± synovial thickening, hyperemia
- MR: Nonspecific fluid + synovial enhancement
  - Findings favoring transient synovitis over septic arthritis
    - Absence of adjacent marrow or soft tissue edema
    - Normal enhancement/perfusion of femoral head

TOP DIFFERENTIAL DIAGNOSES

- Septic arthritis
- Juvenile idiopathic arthritis
- Trauma
- Reactive effusion due to adjacent bone pathology (e.g., Legg-Calvé-Perthes)

PATHOLOGY

- Viral etiology considered most likely

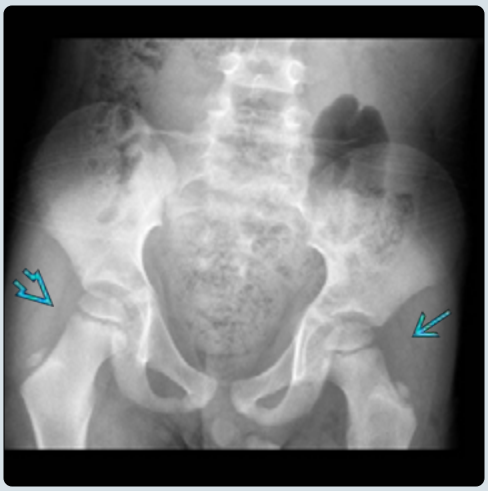
CLINICAL ISSUES

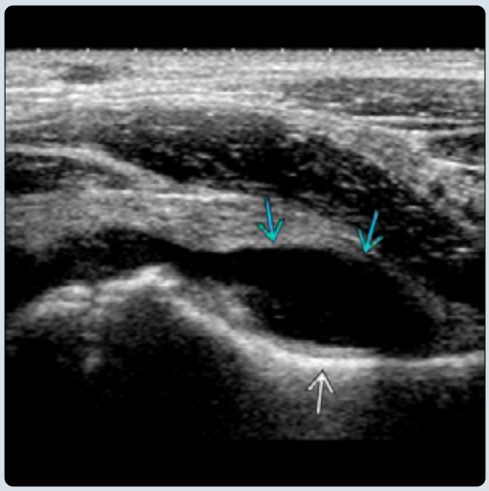
- 3-8 years of age; mean: 4.7-5.5 years
- Limping ± pain; typically afebrile (> 90%)
- Kocher criteria: ↑ likelihood for septic arthritis over transient synovitis with ↑ number of positive parameters
  - Fever, nonweight-bearing, ↑ WBC, ↑ ESR
- Transient synovitis self-limited, lasting 7-10 days
- Conservative management with bed rest + NSAIDs
  - Hip aspiration expedites clinical improvement

DIAGNOSTIC CHECKLIST

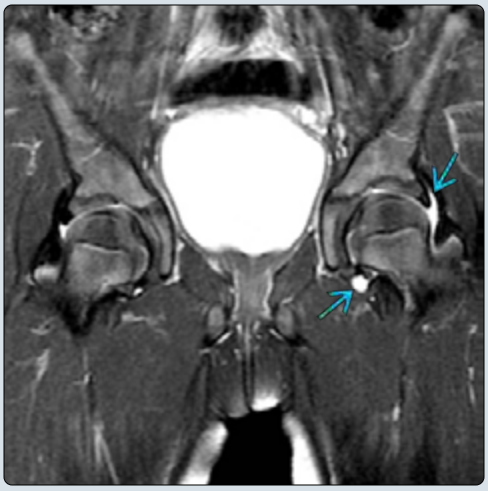
- Imaging alone cannot exclude infection of detected fluid
  - Joint aspiration required for 100% confidence

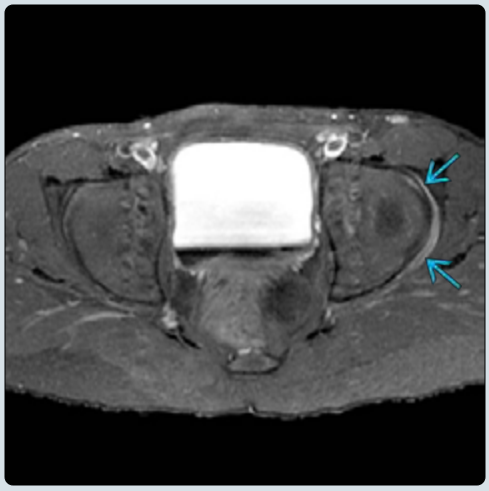
(Left) *AP radiograph of the pelvis in an afebrile 5 year old with a new onset of limp & left hip pain shows bowing of the left gluteal fat pad as compared to the normal right side, suggesting a left hip joint effusion.* **(Right)** *Sagittal ultrasound of the left hip in the same patient shows convex bowing of the joint capsule by hypoechoic fluid that distends the joint at the femoral neck. The patient had normal laboratory markers & was successfully treated conservatively for toxic synovitis.*





(Left) *Coronal T2 FS MR in a 4 year old with several days of left thigh pain & limping shows a small left hip joint effusion with no adjacent marrow or soft tissue edema.* **(Right)** *Axial T1 C+ FS MR in the same patient shows mildly increased synovial enhancement of the left hip joint as compared to the right. The patient was successfully treated conservatively for toxic synovitis given the combination of clinical & imaging features.*





TERMINOLOGY

Synonyms

- Toxic synovitis

Definitions

- Idiopathic self-limited inflammation of pediatric hip

IMAGING

General Features

- Best diagnostic clue
  - Hip effusion & limp in child without fever or elevated serum markers of inflammation
- Location
  - Bilateral in ~ 5%

Radiographic Findings

- Low sensitivity for hip joint effusion
- ± convex gluteal fat pad & medial joint space widening

Ultrasonographic Findings

- Distention of joint capsule by anechoic, hypoechoic, or complex fluid
  - Anterior joint capsule demonstrates convex bulge (rather than normal concavity) over femoral neck
- ± synovial thickening, hyperemia

MR Findings

- Features of nonspecific synovitis (with many etiologies)
  - Homogeneously bright T2/STIR fluid in hip joint
  - Mild synovial thickening & enhancement
- Findings favoring transient synovitis over septic arthritis
  - Absence of adjacent marrow or soft tissue edema
  - Normal enhancement/perfusion of femoral head

Imaging Recommendations

- Best imaging tool
  - US: Highly sensitive for hip joint fluid
    - US not specific for sterile vs. infected fluid (regardless of size or complexity)
- Protocol advice
  - US: Patient supine with hip in neutral position
    - Anterior parasagittal approach by transducer along femoral neck long axis
    - Comparison images of contralateral hip
  - MR: Coronal plane key for comparison of hips
    - T1, T2 FS/STIR, T1 C+ FS

DIFFERENTIAL DIAGNOSIS

Septic Arthritis

- Bacterial infection leads to joint destruction without emergent drainage & IV antibiotics
- Favored over transient synovitis with
  - High number of clinical/laboratory Kocher criteria
  - MR demonstrating
    - Adjacent marrow & soft tissue edema
    - ↓ enhancement/perfusion of femoral head

Juvenile Idiopathic Arthritis

- ± "rice bodies": Numerous, tiny, low T2 signal intensity bodies of uniform size throughout joint fluid

Trauma

- ± marrow & soft tissue edema, ligament & cartilage injuries

Reactive Effusion Due to Adjacent Bone Pathology

- Osteomyelitis, bone tumor, Legg-Calvé-Perthes

PATHOLOGY

General Features

- Etiology
  - Viral more likely than trauma or allergy

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute pain + limping; afebrile (> 90%)
    - Pain at groin, thigh, or medial knee
- Clinical profile
  - Kocher criteria: Fever > 38.5°C, nonweight-bearing, WBC > 12 x 10^9 cells/L, ESR ≥ 40 mm/hour
    - ↑ likelihood for septic arthritis with ↑ number of positive parameters
      - CRP > 20 mg/L also suggests septic joint

Demographics

- Typically 3-8 years of age; mean: 4.7-5.5 years
- M:F = 2-3:1

Natural History & Prognosis

- Self-limited; lasts 7-10 days

Treatment

- Conservative management with bed rest + NSAIDs
- Hip aspiration expedites clinical improvement
  - Also allows confident exclusion of septic joint

DIAGNOSTIC CHECKLIST

Consider

- Primary concern: Missing septic joint that needs emergent drainage & IV antibiotics to prevent long-term damage
- Imaging alone cannot exclude infection of detected fluid
  - Joint aspiration required for 100% confidence

Image Interpretation Pearls

- Synovitis has many etiologies; always consider infection

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1. Nouri A et al: Transient synovitis of the hip: a comprehensive review. J Pediatr Orthop B. 23(1):32-6, 2014
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Soft Tissue Abscess

KEY FACTS

TERMINOLOGY

- Abscess: Walled-off liquefied collection of necrotic tissue, inflammatory cells, & bacteria

IMAGING

- Most commonly affects single site: Expected lymph node location vs. other subcutaneous or intramuscular focus
  - Septic emboli can cause multifocal collections
- Ultrasound: Excellent for detecting superficial collections & defining drainability
  - Thick-walled centrally avascular collection with surrounding edema ± hyperemia
  - Swirling internal debris upon compression
- MR: Clearly defines deep extent, evaluates adjacent bone/cartilage/joint, & helps exclude other diagnoses
  - Centrally nonenhancing fluid collection with thick, enhancing wall & peripheral poorly defined edema
  - Restricted diffusion centrally

TOP DIFFERENTIAL DIAGNOSES

- Soft tissue sarcoma
- Lymphatic malformation
- Hematoma
- Myositis ossificans
- Rhabdomyolysis/infarction

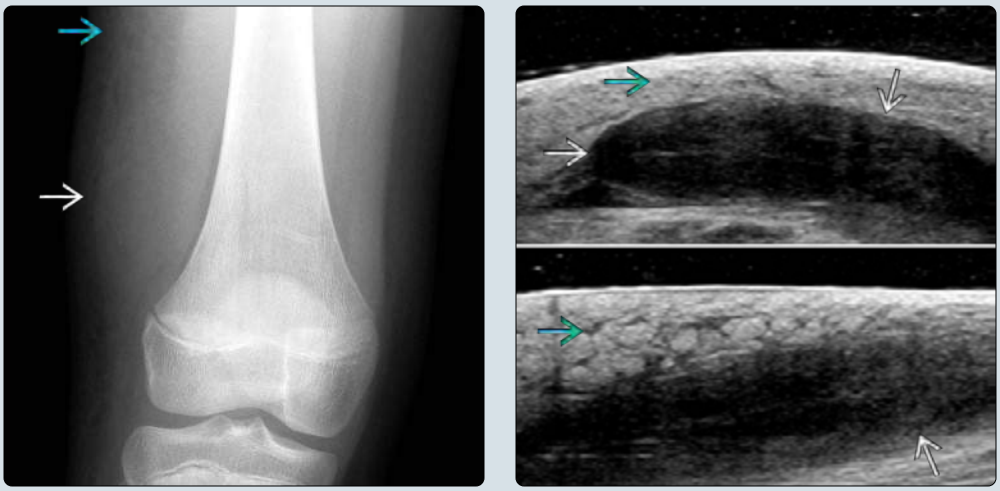
PATHOLOGY

- *Staphylococcus aureus* > > streptococcal species
- *Bartonella henselae*: Regional lymphadenitis ± suppuration in cat scratch disease

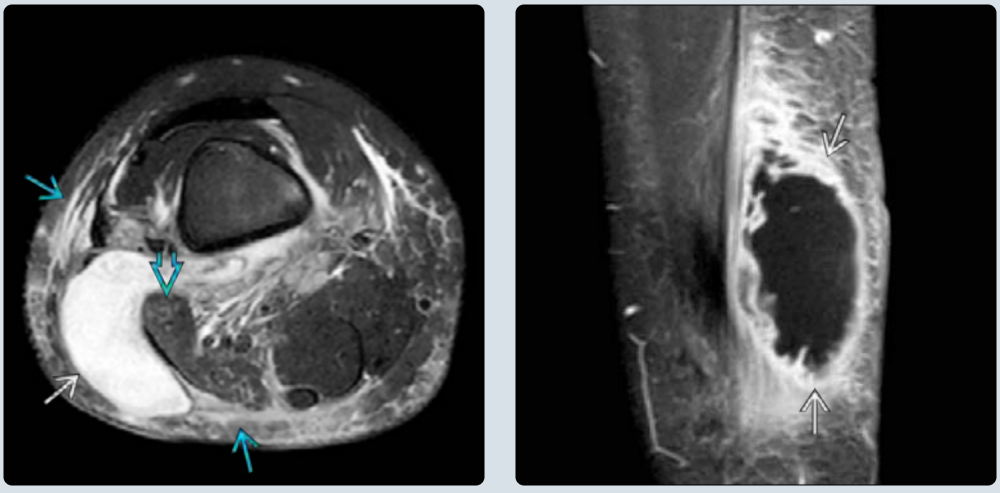
CLINICAL ISSUES

- Presentation: Swelling, erythema, tenderness, limited motion, fever; ± fluctuance, sepsis
- Treatment: Drainage procedure + IV antibiotics
  - Whole-body MR may be useful to screen large territories for drainable collections in setting of systemic infection

(Left) *AP radiograph of the knee in an 11-year-old patient with lateral swelling & tenderness of the distal thigh shows blurring of the normally sharp interfaces between subcutaneous fat & muscle, typical of edema. There is focal bulging of the soft tissues distally.* **(Right)** *Longitudinal ultrasound of the same patient shows thickening & reticular edema of the subcutaneous fat overlying a heterogeneously hypoechoic ovoid collection.*



(Left) *Axial T2 FS MR of the same patient shows a well-circumscribed, crescentic, hyperintense collection overlying the distal biceps femoris muscle. There is surrounding soft tissue edema.* **(Right)** *Sagittal T1 C+ FS MR in the same patient shows thick, irregular rim enhancement of the fluid collection, typical of an abscess. Pus was encountered upon surgical drainage.*



TERMINOLOGY

Synonyms

- Pyomyositis, suppurative lymphadenitis

Definitions

- Abscess: Walled-off liquefied collection of necrotic tissue, inflammatory cells, & bacteria
- Phlegmon: Poorly defined focus of necrotic & inflammatory tissue ± components of liquefaction
- Cellulitis: Poorly defined inflammatory changes of superficial soft tissue infections
- Suppurative or purulent: Pus forming/containing

IMAGING

General Features

- Best diagnostic clue
  - US: Thick-walled avascular collection with swirling internal echogenicities & overlying edema
  - MR: Centrally nonenhancing fluid collection with thick, enhancing wall & peripheral poorly defined edema
- Location
  - Most commonly affects single site; possibilities include
    - Expected sites of lymph nodes
    - Isolated collection in muscle > tendon sheath, bursa
    - Spread from deep (adjacent osteomyelitis & subperiosteal abscess) or superficial (cellulitis, penetrating wound) source
  - Septic emboli can cause multifocal collections

Radiographic Findings

- Edema: Subcutaneous reticulation with blurring of normally sharp fat-muscle interfaces
- ± focal bulge at site of collection

Ultrasonographic Findings

- Grayscale ultrasound
  - Well-defined collection with thick, irregular wall
  - Contents range from anechoic to isoechoic
    - Internal debris swirls with dynamic compression
- Color Doppler
  - No internal vascularity; ± ↑ peripheral vascularity

MR Findings

- T2 FS/STIR
  - Collection of hyperintense fluid centrally
  - Wall may show hypointense inner rim (minerals, hemorrhage, fibrous tissue) & hyperintense outer rim (granulation tissue)
  - Surrounding edema
- Mildly thickened, irregular, enhancing rim after contrast without substantial nodularity
- Restricted diffusion centrally

Imaging Recommendations

- Best imaging tool
  - US excellent for detecting superficial collections & defining drainability
  - MR more clearly defines collection deep extent & relationships to vital structures, evaluates adjacent bone & cartilage for infection, helps exclude other diagnoses

DIFFERENTIAL DIAGNOSIS

Soft Tissue Sarcoma

- Typically solid, well-circumscribed mass with little (if any) surrounding edema
- Variable enhancement ± foci of necrosis, cysts

Lymphatic Malformation

- Multicystic mass crossing soft tissue compartments
- Fluid-fluid levels due to internal hemorrhage
- Rim & septal enhancement only

Hematoma

- Heterogeneous intramuscular collection after trauma

Myositis Ossificans

- Heterogeneous intramuscular mass with marked surrounding edema on T2 FS/STIR MR
- Peripheral Ca^{2+} within weeks diagnostic
- Occurs after injury but trauma history lacking in 40%

Rhabdomyolysis/Infarction

- Large regions of necrotic muscle after compression/trauma
- Very ↑ serum markers of muscle breakdown

PATHOLOGY

General Features

- Most common organisms
  - *Staphylococcus aureus* > > streptococcal species
  - *Bartonella henselae*: Regional lymphadenitis ± suppuration in cat scratch disease
    - Axillary, epitrochlear > head/neck or inguinal nodes
  - Mycobacterial infections may result in large muscular abscesses (classically psoas with rim Ca^{2+})
- Mechanisms
  - Cellulitis spreading to deeper soft tissues or regional lymph nodes
  - Extension from adjacent deep infection
  - Hematogenous seeding of muscle
  - Puncture wound

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Swelling, erythema, tenderness, limited motion, fever

Treatment

- Drainage procedure + IV antibiotics
  - Drainage may not be necessary if collection < 2 cm in noncritical location

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KEY FACTS

TERMINOLOGY

- Widespread misuse of "hemangioma" nomenclature in medical literature
  - True hemangioma: Benign vascular neoplasm
- Infantile hemangioma (IH)
  - Most common soft tissue tumor of childhood
  - Predictable life cycle: Usually absent at birth, rapid growth over first few weeks/months of life (proliferating phase), spontaneous regression over subsequent months to years (involuting phase)

IMAGING

- Elongated, lobulated, well-defined, highly vascular, solitary or multifocal, superficial soft tissue mass/masses appearing in first few weeks of life
  - Common cutaneous IH not usually imaged due to typical timeline & appearance
  - Deeper subcutaneous IH without typical skin findings likely to be imaged, as diagnosis less clear
- Doppler should clearly document characteristic internal vascularity throughout proliferating IH
- Imaging features change with involution

TOP DIFFERENTIAL DIAGNOSES

- Venous malformation
- Lymphatic malformation
- Arteriovenous malformation
- Congenital hemangioma
- Kaposiform hemangioendothelioma
- Soft tissue sarcoma

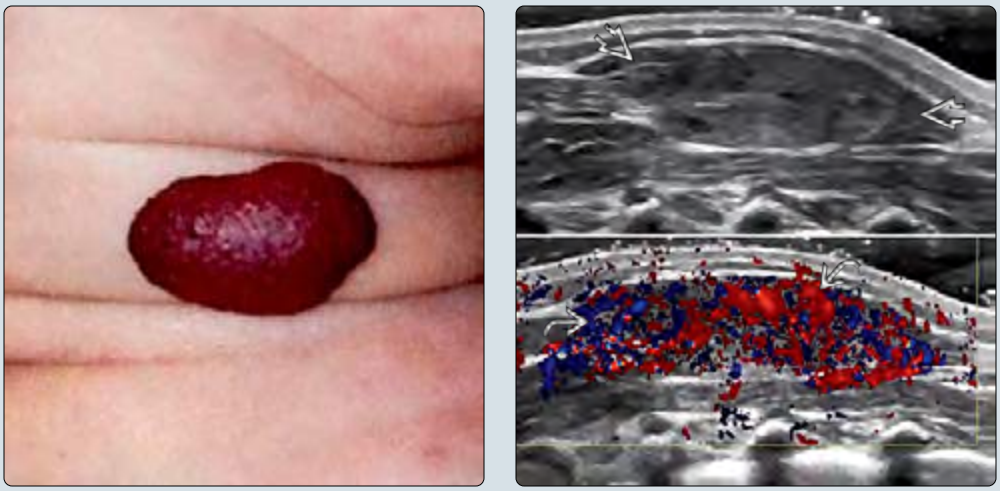
CLINICAL ISSUES

- Most follow benign course without therapy required
- Complications &/or associated anomalies more likely if IH large, segmental, facial, or multifocal

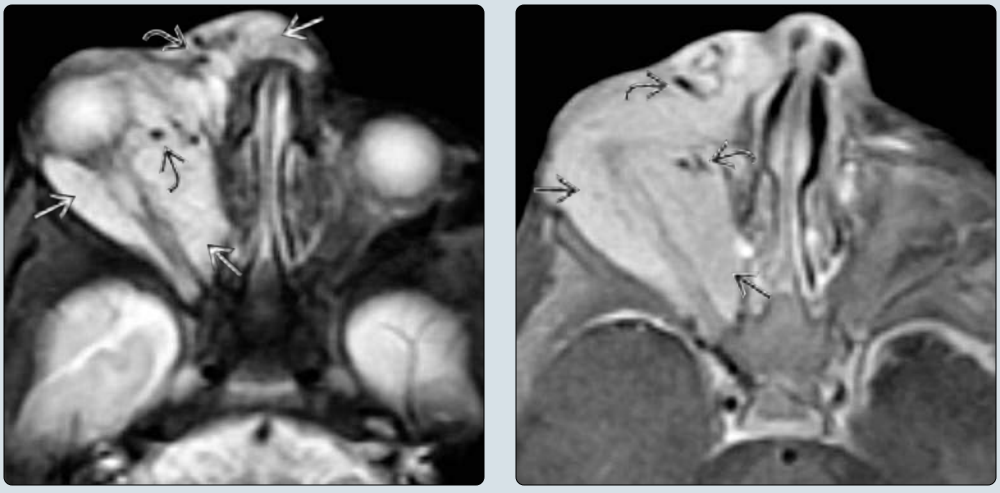
DIAGNOSTIC CHECKLIST

- If imaging appearance, timeline, or physical exam findings atypical for IH, exclude other lesions by biopsy

(Left) *Photograph of a 4-month-old patient shows a well-defined, raised "strawberry" lesion on the neck that appeared after birth, typical of a proliferating infantile hemangioma (IH).* **(Right)** *Longitudinal ultrasound images at the site of a palpable back mass in the same patient show a well-defined lentiform subcutaneous lesion with mildly heterogeneous internal echogenicity. The mass demonstrates a high vessel density on color Doppler imaging, typical of a proliferating deep IH.*



(Left) *Axial T2 FS MR in a 2-month-old girl with a periorbital IH & proptosis shows diffusely high signal intensity throughout the lobular nasal, periorbital, & retrobulbar components of the mass. Several low signal intensity flow voids (due to high/fast flow vessels) are visualized in the mass.* **(Right)** *Axial T1 C+ FS MR in the same patient shows diffuse homogeneous enhancement of the mass (other than scattered internal flow voids). These are typical MR features of a proliferating IH.*



TERMINOLOGY

Synonyms

- Capillary hemangioma, strawberry hemangioma
- Not cavernous hemangioma (actually venous malformation)
- Not synovial hemangioma (actually venous malformation)
- Not hemangioendothelioma (actually higher grade vascular tumor)

Definitions

- 2014 revised classification by the International Society for the Study of Vascular Anomalies (ISSVA) retains 2 main categories
  - Vascular tumors (true neoplasms with cellular proliferation; generally grow out of proportion to patient)
  - Vascular malformations (congenital errors of vessel development; generally grow commensurate with patient)
- Hemangioma: Benign vascular neoplasm with predictable life cycle
  - Infantile hemangioma (IH)
    - Most common soft tissue tumor of childhood
    - Usually absent at birth with rapid growth over first few weeks/months of life (proliferating phase) followed by spontaneous regression over subsequent months to years (involuting phase)
  - Congenital hemangioma (CH)
    - Fully developed at birth
    - Rapidly involuting (RICH) vs. noninvoluting (NICH)
      - RICH mostly involuted by 14 months

IMAGING

General Features

- Best diagnostic clue
  - Lobular, well-defined, highly vascular solitary or multifocal superficial soft tissue masses appearing in first few weeks of life
    - Common cutaneous lesions not usually imaged due to typical appearance & timeline
    - Deeper subcutaneous lesions without typical skin findings likely to be imaged, as diagnosis less clear
- Location
  - Head & neck (60%)
  - Trunk (25%)
  - Extremities (15%)
- Other imaging characteristics
  - Depends on timing of imaging
    - Proliferating phase: Homogeneous & diffuse enhancement; high-flow vessels within/adjacent to mass
    - Involuting phase: Heterogeneous with varying fatty infiltration; ↓ flow/enhancement of mass

Radiographic Findings

- Radiography
  - Soft tissue mass without Ca^{2+}
    - Phleboliths found in venous malformations, not IH

MR Findings

- Proliferating phase
  - Largely homogeneous except for occasional flow voids
  - T1: Intermediate signal intensity
  - T2 FS/STIR: High signal intensity
    - Usually not as bright as fluid
  - T1 C+ FS: Diffuse early, homogeneous enhancement
- Involuting phase
  - Varying degrees of fatty infiltration with ↓ flow/enhancement

Ultrasonographic Findings

- Grayscale ultrasound
  - Lobular mass with variable echogenicity & few macroscopic vessels
- Color Doppler
  - "Lights up" during proliferating phase
    - High vessel density on color Doppler (> 5 vessels/cm^2)
    - High systolic Doppler shift (> 2 kHz): Many arterial tracings with low resistive index but no arterialized veins to suggest shunting
  - With involution/treatment: ↓ vessel density, ↑ resistive index

Imaging Recommendations

- Best imaging tool
  - Cutaneous IH (majority) can be diagnosed by physical appearance & temporal growth history without imaging
  - Imaging used for diagnosing deeper lesions or when location/number of cutaneous lesions implicate associated anomalies &/or complications
    - IH over lower midline back: Spine anomalies
    - Perineal IH: PELVIS/LUMBAR syndrome
    - Segmental or bearded facial IH: PHACE(S), airway involvement
    - Periocular IH: ± intraorbital components, various ophthalmologic complications
    - ≥ 5 cutaneous IHs: ↑ likelihood of visceral involvement, especially liver
      - Numerous liver lesions can lead to hepatic & heart failure, abdominal compartment syndrome, hypothyroidism
  - For deeper lesions, ultrasound with Doppler can be highly suggestive of IH in right age range
- Protocol advice
  - Doppler should clearly document characteristic flow throughout lesion
  - MR imaging should include flow-sensitive & fluid-sensitive sequences as well as T1 C+ FS

DIFFERENTIAL DIAGNOSIS

Venous Malformation

- Multiple serpentine channels &/or lobular soft tissue mass/masses infiltrating various compartments
- Fluid signal intensity mass ± fluid-fluid levels
- Puddling of contrast in mass with gradual complete enhancement (except for thrombi)
- Phleboliths virtually diagnostic in children

Lymphatic Malformation

- Multicystic mass crossing soft tissue planes

- May contain fluid-fluid levels, debris
- Rim/septal enhancement
  - Microcystic components may appear more solid

Arteriovenous Malformation

- Tangle of high-flow vessels with shunting
- ± other soft tissue components

Congenital Hemangioma

- Solid, heterogeneous, less well-defined mass ± Ca^{2+}, hemorrhage, necrosis
- Fully developed at birth; different cutaneous features vs. IH

Kaposiform Hemangioendothelioma

- Usually poorly defined, infiltrating, solid enhancing mass presenting in infant with Kasabach-Merritt phenomenon
- ± marked surrounding edema: Difficult to discern from lesion margins

Soft Tissue Sarcomas

- More likely to be firm, solid, round ball with heterogeneous enhancement & intermediate to low vascularity
- Relatively uncommon in first few months of life

PATHOLOGY

Microscopic Features

- Proliferating phase: Masses of plump endothelial cells forming small vascular channels; high expression of angiogenic factors
- Involuting phase: Flat endothelial cells + fibrofatty replacement

Immunohistochemical Features

- Glucose transporter 1 (GLUT1) positive during all phases
  - CH: GLUT1 negative

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually presents during infancy as asymptomatic soft mass
    - With cutaneous involvement (most common): Lobular bright red or pink lesion
      - Localized: Nodule, papule
      - Segmental: Plaque-like over region
    - With deeper subcutaneous lesions: Bluish hue to skin
- Other signs/symptoms
  - During proliferation, high flow may cause bruit, warmth
  - May present with complication
    - Compression of vital structures, liver failure, etc.

Demographics

- Age
  - At birth: Typically absent
    - 30% have precursor lesion (pallor, ecchymosis, telangiectasia)
  - 0-2 months: Almost all double in size
  - 3-5 months: 80% of maximum size
  - 9-12 months: Peak size reached in almost all lesions
  - 12-48 months: Most rapid phase of involution
  - By 84-108 months: Involution complete
    - 20-50% with cutaneous residua
- Gender
  - Female predominance (F:M = 1.5-4.0:1.0)
- Ethnicity
  - Caucasians > > African Americans, Hispanics, Asians
- Epidemiology
  - Multiple lesions in 15-30% of patients
  - Up to 10% of premature infants affected vs. 5% overall

Natural History & Prognosis

- Most follow benign course
- Complications more likely if IH large, segmental, multifocal, or on face
  - Ulceration (most likely with intertriginous & perioral lesions), bleeding
  - Compression of vital structures (airway, orbit)
  - Heart failure, liver failure, hypothyroidism, &/or compartment syndrome with high liver lesion burden
  - Psychological issues (especially with facial lesions)
  - Coagulopathy is not associated with IH

Treatment

- Most IHs require no imaging or therapy
- Therapy reserved for large or complicated lesions or for significant residua following involution
- 1st-line treatment is medical
  - Propranolol (β-blocker) has replaced oral steroids as primary therapy due to low side effect profile
- Other therapies: Intralesional steroids, surgical removal, pulsed-dye laser

DIAGNOSTIC CHECKLIST

Consider

- If imaging appearance, timeline, or physical exam findings are atypical for IH, exclude other lesions by biopsy
- IH patients with cutaneous segmental lesions, certain facial lesions, or ≥ 5 lesions otherwise require further directed imaging due to associated anomalies/complications

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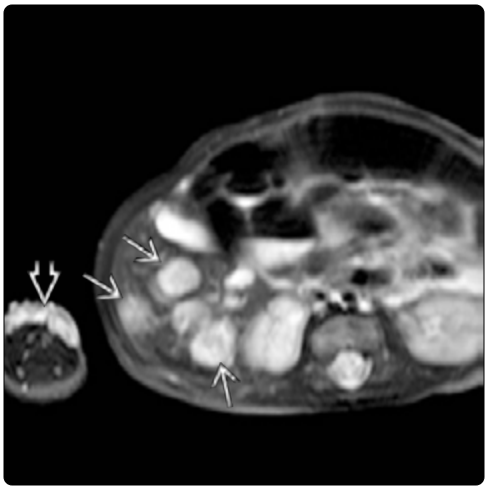
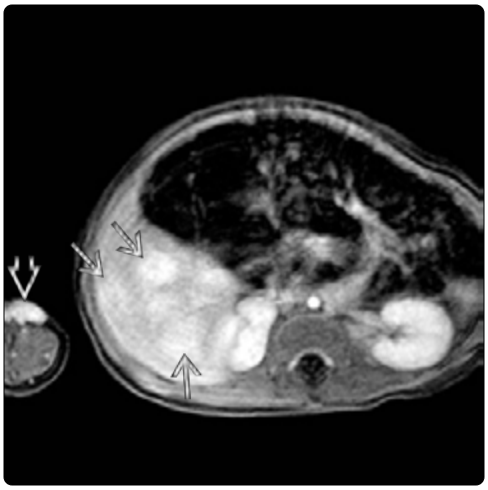
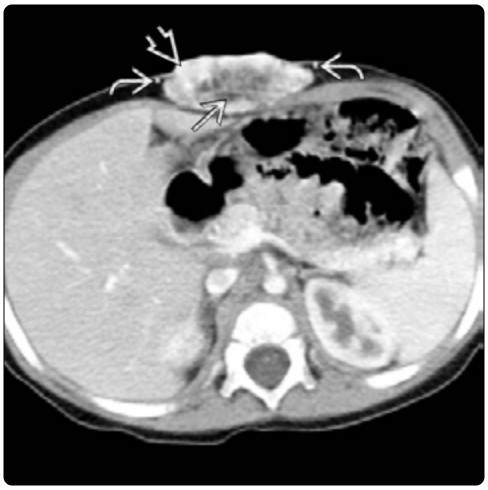
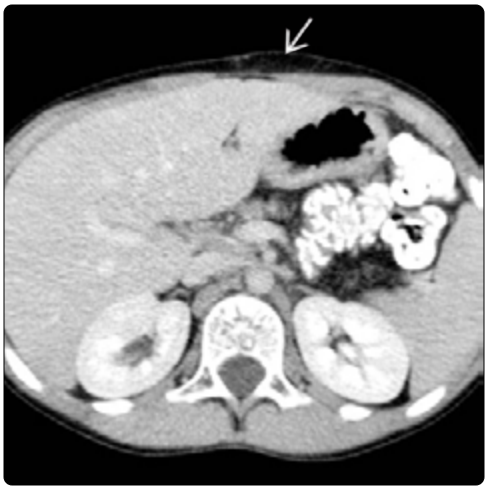


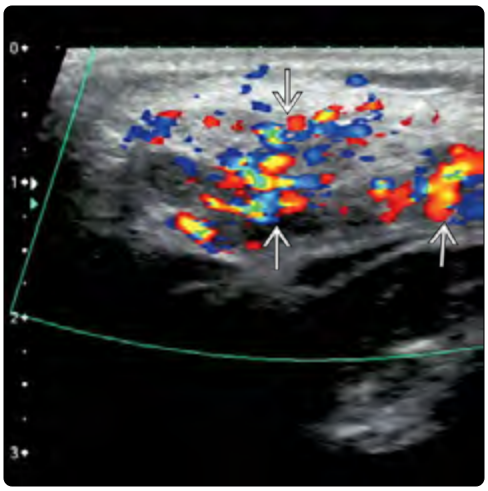


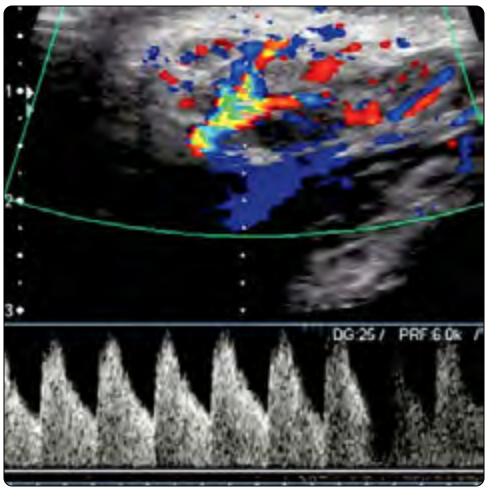
(Left) *Axial T2 FS MR through the abdomen of a 3 month old with a known forearm IH shows multiple round, hyperintense hepatic lesions. The lobular superficial hyperintense right forearm mass is partially visualized as well.* **(Right)** *A 45-second delayed axial T1 C+ FS MR in the same patient shows nearly homogeneous enhancement of the hepatic & forearm lesions, typical of multifocal IHs.*





(Left) *Axial abdominal CECT in a 12-month-old girl with a known superficial IH shows an elongated, well-defined, lobular mass with peripheral enhancement & central fatty infiltration. Note the prominent vessels along the lesion margins. These findings are typical of an IH during early involution.* **(Right)** *Axial CECT in the same patient 7 years later shows minimal residual prominence of fat at the site of the previously visualized lesion, consistent with an involuted IH.*





(Left) *Color Doppler ultrasound in a 2 month old at the level of a growing soft tissue mass of the back (which was not present at birth) shows a lobular heterogeneous lesion of the subcutaneous tissues with high vessel density, typical of a proliferating IH.* **(Right)** *Spectral tracing through the same lesion on a transverse color Doppler ultrasound shows low-resistance arterial waveforms, typical of IH.*

Kaposiform Hemangioendothelioma

KEY FACTS

TERMINOLOGY

- Aggressive, locally invasive vascular neoplasm of spindled endothelial cells & abnormal lymphatics

IMAGING

- Best clue: Poorly defined, diffusely enhancing soft tissue mass infiltrating multiple tissue planes/compartments in infant with coagulopathy
- ~ 80-90% have continuous cutaneous lesions & deep components
- Most common locations: Extremity > trunk > head & neck; regional lymph node involvement common

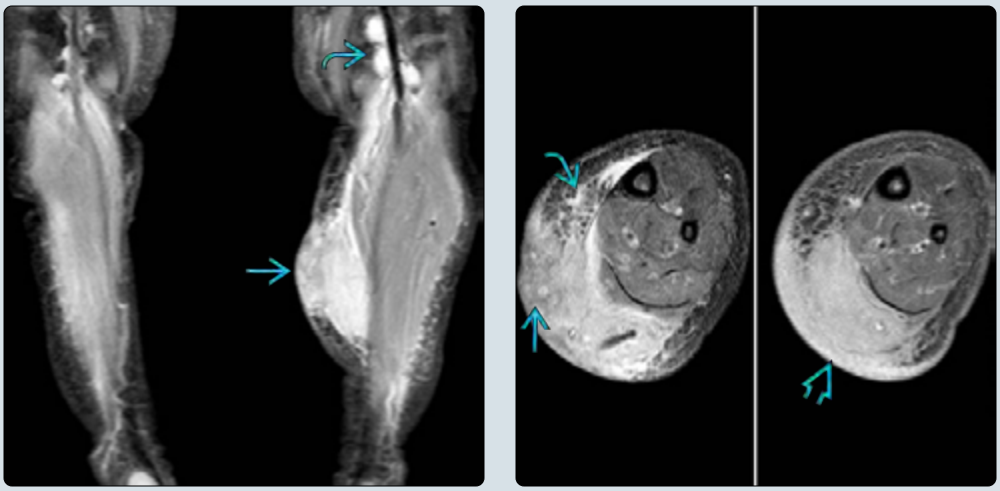
CLINICAL ISSUES

- Characteristic cutaneous vascular lesion ~ 90%; indurated, ill-defined, red-purple mass or plaque
- Profound, sustained consumptive coagulopathy [true Kasabach-Merritt phenomenon (KMP)] ~ 70%
  - ↓ platelets, fibrinogen
  - KHE lesions at risk for KMP include those with
    - Deeper > superficial components (78% vs. 36%)
    - Retroperitoneal & thoracic involvement; cutaneous lesion > 8 cm
- > 90% present in infancy; median age: 2 months
- Mortality ranges from 12-30%
- Treated lesions do not fully regress & may recur
- Treatment in setting of KMP includes steroids plus vincristine &/or sirolimus
  - Limited role for platelet transfusions
  - Complete surgical excision is curative but rarely an option due to infiltrative nature of KHE with extension into vital regions (in addition to associated coagulopathy)

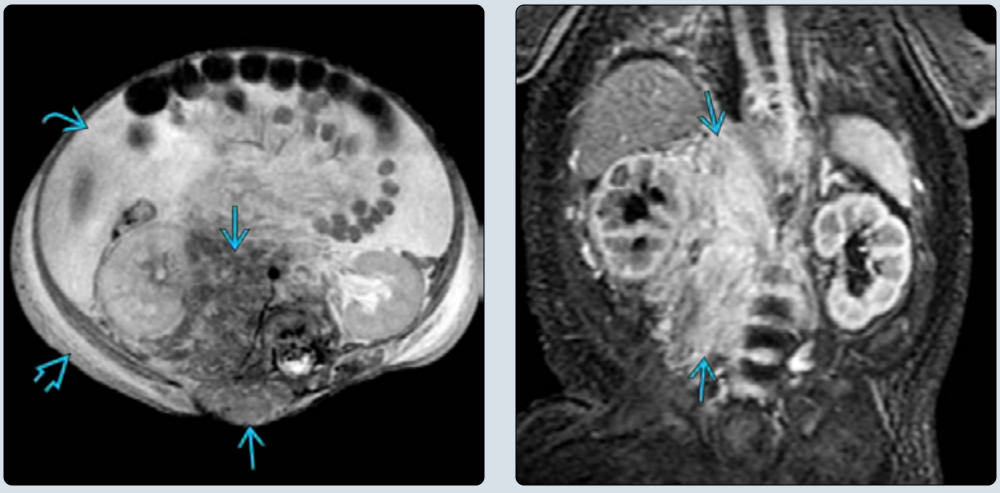
DIAGNOSTIC CHECKLIST

- Enhancing poorly defined soft tissue mass in infant with thrombocytopenia should raise concern for KHE
- In conjunction with characteristic cutaneous lesion, these features may obviate biopsy

(Left) *Coronal STIR MR in a 15 day old with a lobular cutaneous/subcutaneous mass shows diffuse high signal intensity in the lesion. Note the ipsilateral popliteal adenopathy.* **(Right)** *Axial T2 FS MR (left) & T1 C+ FS MR (right) images show foci of intermediate T2 signal intensity intermixed with a poorly circumscribed, infiltrative high signal intensity mass. Note the mild edema surrounding the lesion. The lesion enhances diffusely after contrast administration. KHE was confirmed by biopsy.*



(Left) *Axial FS T2 MR in a 3 week old with consumptive coagulopathy & abdominal distention shows a large, poorly defined intermediate T2 signal intensity mass extending from the skin into the retroperitoneum. There is diffuse body wall edema & a large volume of ascites.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows early, diffuse enhancement of the infiltrative mass, confirmed to be a KHE.*



TERMINOLOGY

Definitions

- Kaposiform hemangioendothelioma (KHE): Aggressive, locally invasive vascular neoplasm of spindled endothelial cells & abnormal lymphatics; **not** equivalent to hemangioma

IMAGING

General Features

- Best diagnostic clue
  - Poorly defined, diffusely enhancing soft tissue mass infiltrating multiple tissue planes/compartments in infant with coagulopathy
- Location
  - Extremity > trunk > head & neck
    - ~ 26% have continuous extension into > 1 region
  - Most have superficial & deep involvement, extending from skin through subcutaneous fat into muscle
    - ~ 80-90% have continuous cutaneous lesions & deep components
    - Visceral & bony involvement infrequent
  - Regional lymph node involvement common
  - Very rare reports of multifocality vs. distant metastases
- CT/MR/US
  - Infiltrating, poorly defined mass
    - Thickening/expansion of involved soft tissues
    - Variable degree of reticular foci or stranding extending from mass into surrounding tissues
      - Difficult to distinguish surrounding edema from true tumor margins
  - May have internal foci of hemorrhage, fibrosis
    - Intermediate to dark T2 components amidst otherwise T2 bright lesion on MR
  - Vigorous, diffuse enhancement
  - Moderately increased vascularity, particularly along deep margins of lesion (rather than internal)
    - ± prominent feeding/draining vessels
    - No large tangles of vessels or significant shunting

DIFFERENTIAL DIAGNOSIS

Soft Tissue Infection (Cellulitis/Fasciitis/Myositis)

- Redness, swelling, pain, ↑ inflammatory markers

Microcystic Lymphatic Malformation

- Longstanding tissue thickening ± cutaneous blebs

Hemangioma, Infantile (IH) vs. Congenital (CH)

- Typically well-defined, high-flow cutaneous/subcutaneous mass of infancy without muscular involvement

Infantile Myofibroma

- Solid soft tissue mass of infants with variable heterogeneity, Ca^{2+}, tissue infiltration, bone involvement

Rhabdomyosarcoma

- Well-defined, solid soft tissue mass of children (not neonates) with only mild internal vascularity

PATHOLOGY

General Features

- Histologic & clinical overlap with tufted angioma (TA)
  - Now largely considered spectrum of same disease

Microscopic Features

- Coalescing nodules of spindled endothelial cells surrounded by abnormal lymphatic channels
  - Intermixed platelet microthrombi, extravasated red blood cells, hemosiderin

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Characteristic cutaneous vascular lesion ~ 90%
    - Indurated, ill-defined, red-purple mass or plaque
  - Profound, sustained consumptive coagulopathy [true Kasabach-Merritt phenomenon (KMP)] ~ 70%
    - ↓ platelets, fibrinogen
    - KHE lesions at risk for KMP include those with
      - Deeper > superficial components (78% vs. 36%)
      - Retroperitoneal & thoracic involvement
      - Cutaneous lesion > 8 cm
    - ~ 10% will develop KMP after presentation for other symptoms: Median delay of 6.5 weeks

Demographics

- > 90% present in infancy; median age: 2 months

Natural History & Prognosis

- Mortality ranges from 12-30%
- Treated lesions do not fully regress & may recur
- Long-term complications depend on original site of involvement; may include pain, edema, organ dysfunction

Treatment

- KHE with KMP
  - Steroids plus vincristine &/or sirolimus
  - Limited role for platelet transfusions (only prior to surgery or for active bleeding)
    - Platelets rapidly consumed by lesion, causing enlargement & pain
  - Complete surgical excision curative but rarely option due to infiltrative nature of KHE with extension into vital regions (in addition to associated coagulopathy)
  - Embolization can be temporizing
- KHE without KMP
  - Oral steroids alone may be used for growing lesions

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KEY FACTS

TERMINOLOGY

- Subtype of congenital slow- or low-flow vascular malformation due to error in vein formation; not neoplastic
  - Not hemangioma (benign vascular neoplasm of capillaries) or arteriovenous malformation (high flow)

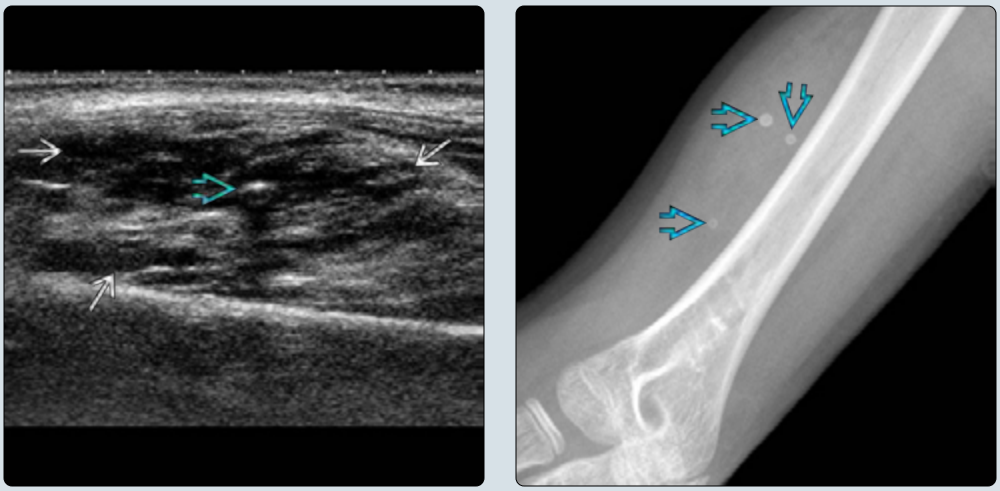
IMAGING

- Locations
  - Most commonly subcutaneous &/or intramuscular but may involve bone, synovium, viscera
  - Focal, multifocal, or diffuse throughout region
- Morphology
  - Well-circumscribed lobulated mass vs. extensive confluent, infiltrative lesion
  - ± discrete serpentine venous channels of abnormal number, size, shape, & location
- Radiography: Phleboliths in mass essentially diagnostic
- MR: High fluid signal intensity ± layering fluid-fluid levels
  - Phleboliths typically small, round, & dark
  - Diffuse or patchy enhancement of venous malformation (VM); discrete venous channels will diffusely enhance
- US: Mass of heterogeneous echotexture
  - Hypoechoic/anechoic tubular channels in clusters
    - Compressible; will slowly refill
  - Phleboliths: Round echogenic foci with posterior acoustic shadowing & twinkle artifact
  - Detectable venous waveforms often sparse

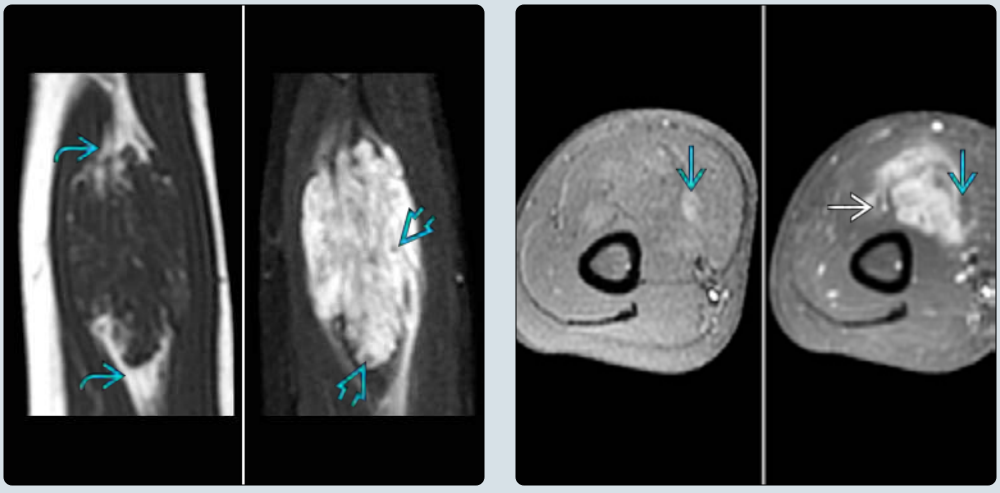
CLINICAL ISSUES

- Soft, compressible mass without thrill
  - Enlarges with Valsalva/crying/dependent positioning
  - Bluish skin discoloration with superficial lesions
  - Episodic pain &/or swelling
- Grow proportional to child but may enlarge suddenly due to hemorrhage, thrombosis, or hormonal changes
- Treatments include conservative therapy (compression garments, antiinflammatory medications), percutaneous procedures (sclerotherapy, laser), surgical resection

(Left) *Longitudinal US of the upper arm in a 9-year-old girl with new pain & swelling shows a multilobulated collection of hypoechoic channels within the biceps muscle. There is an echogenic focus in the mass with posterior acoustic shadowing.* **(Right)** *AP radiograph of the humerus in the same patient confirms numerous round intralesional Ca^{2+} with lucent centers, typical of phleboliths in a venous malformation (VM).*



(Left) *Coronal T1 (left) & STIR (right) MRs of the same VM show the lobulated soft tissue mass following fluid signal intensity except for the thin septations & phleboliths. Note the fat along the margins & septations.* **(Right)** *Axial T1 FS pre- (left) & postcontrast (right) MRs through the same mass show that the lesion is largely isointense to muscle before contrast followed by heterogeneous enhancement. A focal thrombus shows T1 shortening before contrast & no enhancement after contrast.*



TERMINOLOGY

Synonyms

- Venous anomaly, common venous malformation (VM)
- Not hemangioma (benign vascular neoplasm of capillaries) or arteriovenous malformation (high-flow lesion)

Definitions

- Subtype of congenital slow- or low-flow vascular malformation due to error in vein formation
- Not neoplastic

IMAGING

General Features

- Location
  - Head/neck (40%), extremities (40%), trunk (20%)
  - Focal, multifocal, or diffuse throughout region
  - Most commonly subcutaneous &/or intramuscular but may involve bone, synovium, viscera
    - Often cross soft tissue planes/compartments
- Morphology
  - Well-circumscribed lobulated mass vs. extensive confluent, infiltrative lesion
  - ± discrete serpentine venous channels of abnormal number, size, shape, & location

Radiographic Findings

- Lobulated soft tissue mass ± calcified phleboliths (30%)

MR Findings

- T1WI
  - Largely isointense to muscle
    - Hemorrhage or proteinaceous debris may have mildly ↑ signal intensity relative to muscle
    - Phleboliths typically small, round, & dark
    - Noncalcified internal thrombi may be bright
    - Bright fat often interspersed within/around lesion
- T2WI FS
  - Serpentine venous channels generally very bright
    - Tightly clustered masses of abnormal channels show thin, dark, intervening septations
    - Large varicosities may show low signal internally due to disturbed flow; can mimic clot
  - Layering fluid-fluid levels often present
    - Due to hemorrhage or proteinaceous debris
  - Absence of flow voids in VM
  - Phleboliths typically small, round, & dark
- T2* GRE
  - Useful to show chronic joint complications of hemarthroses from synovial VM
    - "Blooming" (exaggerated signal loss) of synovium from hemosiderin deposits
    - Focal or diffuse cartilage loss
- T1WI C+ FS
  - Diffuse or patchy enhancement of VM; discrete venous channels will diffusely enhance
    - Thrombi, intermixed lymphatic components will not enhance
  - Dynamic imaging shows gradual delayed contrast filling + absence of large arteries or arteriovenous shunting
- MRV
  - Useful to exclude intralesional high-flow vessels & confirm patency of deep venous system & varicosities
    - Slow or low flow can lose signal, mimic clot

Ultrasonographic Findings

- Grayscale ultrasound
  - Mass of heterogeneous echotexture
    - Hypoechoic/anechoic tubular channels in clusters
      - Compressible; will slowly refill
    - Intermixed & peripheral patchy foci of ↑ echogenicity due to fat
    - Phleboliths: Round echogenic foci with posterior acoustic shadowing & twinkle artifact
  - ± discrete venous channels of abnormal number, size, shape, & location in surrounding tissues
- Pulsed Doppler
  - Lack of arterial waveforms or arterialized draining veins
    - However, 1-2 normal arteries can be encased by VM
  - Detectable venous waveforms often sparse
    - Random velocity spikes may be due to application of pressure to nearby tissues; can mimic arterial flow
- Color Doppler
  - Majority show little internal vascular flow due to slow flow through dysplastic venous channels
    - Valsalva or compression/release will ↑ flow

Imaging Recommendations

- Best imaging tool
  - US (& potentially radiographs) can be diagnostic of VM
  - MR will show confirmatory features & clarify extent
- Protocol advice
  - US: Assess lesion response to compression
  - MR: Rapid postcontrast 3D GRE sequence (FSPGR or Dixon) can confirm deep venous system or varicosity patency (as MRV confounded by slow or low flow)
    - Blood pool contrast agent helpful if covering multiple large territories in extensive VM setting

DIFFERENTIAL DIAGNOSIS

Infantile Hemangioma

- Characteristic life cycle: Small or absent at birth, rapid growth during early infancy, gradual involution over years
- Solid ovoid vs. lobular, elongated soft tissue mass, typically in cutaneous &/or subcutaneous tissues
- Variable sonographic echogenicity due to mix of proliferating & involuting components; no phleboliths
- Highly vascular by Doppler US; many arterial waveforms
- Bright (but not fluid) T2 signal intensity with diffuse early enhancement on MR

Arteriovenous Malformation

- Tangle of enlarged tortuous arteries & veins with variable soft tissue components
- High flow with shunting by ultrasound, MRA/MRV

Lymphatic Malformation

- Another common slow- or low-flow lesion
- Compressible fluid-filled macrocystic soft tissue mass
  - Thin enhancing internal septations, internal debris/fluid-fluid levels, no internal vascularity
- Microcystic lesion more solid-appearing, infiltrative

Soft Tissue Sarcoma

- Typically firm, well-defined, round or ovoid hypovascular solid soft tissue mass with variable enhancement
- ± cystic components but solid components typically visible

Plexiform Neurofibroma

- Elongated lobular masses along courses of nerves
  - In multiple adjacent nerves: "Bag of worms" appearance
- Cross section of lesion shows target sign on MR
  - Bright peripherally, dark centrally on T2/STIR
  - Dark peripherally, bright centrally on T1 C+ FS

Spindle Cell Hemangioma

- Uncommon benign neoplastic vs. reactive vascular lesion
- May contain phleboliths
- Typical soft tissue lesion in Maffucci syndrome

Fibroadipose Vascular Anomaly

- Uncommon solid lesion of extremity musculature
- Contains abnormal veins + dense fibrous & fatty tissue
- Constant pain + contracture

PATHOLOGY

General Features

- Genetics
  - *TEK* mutation in many VM (50% of sporadic VM)
- Associated abnormalities
  - Klippel-Trenaunay syndrome
    - Capillary-lymphatic-venous malformation of extremity (usually lower) + lipomatous & osseous overgrowth
    - Port-wine capillary stain on lateral extremity with lymphatic vesicles
    - Varicosities of superficial veins
      - Marginal venous system may dominate over diminutive or absent deep venous system
  - Maffucci syndrome
    - Vascular soft tissue masses + enchondromatosis
      - Soft tissue masses likely represent spindle cell hemangiomas rather than true VM

Staging, Grading, & Classification

- 2014 revised classification of VM by International Society for Study of Vascular Anomalies
  - Common VM (94%)
  - Familial VM cutaneomucosal
    - Multifocal lesions of lips, tongue
    - Dilated neck, upper extremity veins
  - Blue rubber bleb nevus syndrome
    - Multifocal cutaneous, muscular, gastrointestinal VM
      - Intestinal VM leads to chronic bleeding, anemia
  - Glomuvenous malformation
    - Presence of glomus cells in VM
    - Darker, less compressible superficial lesions
  - Cerebral cavernous malformation

Microscopic Features

- Irregular variably sized channels of flattened endothelium with variable smooth muscle & no internal elastic lamina
- Intraluminal thrombi common
- May contain intermixed lymphatic components

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Soft, compressible mass without thrill
    - ± small internal firm thrombi/hard phleboliths
    - Enlarges with Valsalva/crying/dependent positioning
  - Bluish skin discoloration with superficial lesions
  - Episodic pain &/or swelling, which may be due to
    - Engorgement from stasis
    - Intralesional thrombosis
    - Hemorrhage into adjacent tissues, joints
    - Local compression of adjacent tissues
    - Local muscular dysfunction
- Other signs/symptoms
  - Grow proportional to child but may enlarge suddenly due to hemorrhage, thrombosis, or hormonal changes
  - Large varicosities can lead to thromboembolism

Natural History & Prognosis

- Prognosis depends on lesion size, extent, & location
- Larger, more extensive lesions may cause lifelong morbidity

Treatment

- Conservative therapy
  - Compression garments
  - Antiinflammatory medications
  - Low molecular weight heparin if thrombosis risk ↑
- Percutaneous procedures
  - Direct injection with sclerosing agent under fluoroscopic/ultrasound guidance
    - Multiple procedures may be required
  - Laser ablation (Nd:YAG laser) of superficial VM
  - Endovenous ablation of varicosities
    - Radiofrequency vs. laser
- Surgical resection of focal lesions
  - ± percutaneous sclerosis for more extensive VM

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Multiple fluid-fluid levels in pediatric soft tissue mass strongly suggest slow- or low-flow vascular malformation
- Phleboliths in soft tissue mass virtually diagnostic of VM

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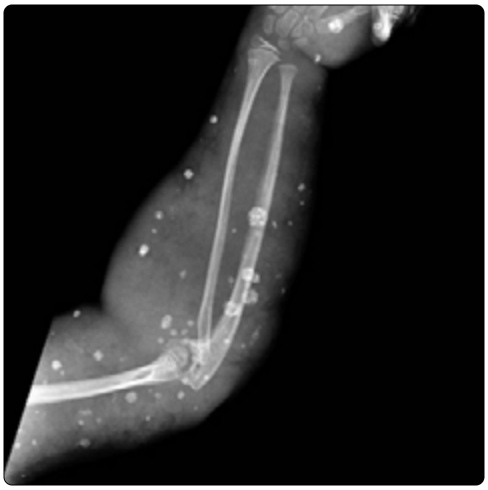
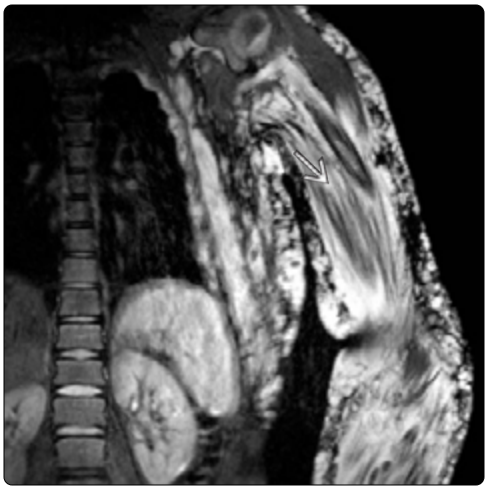
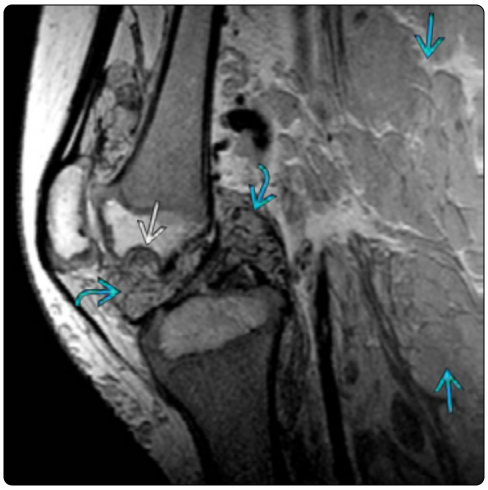
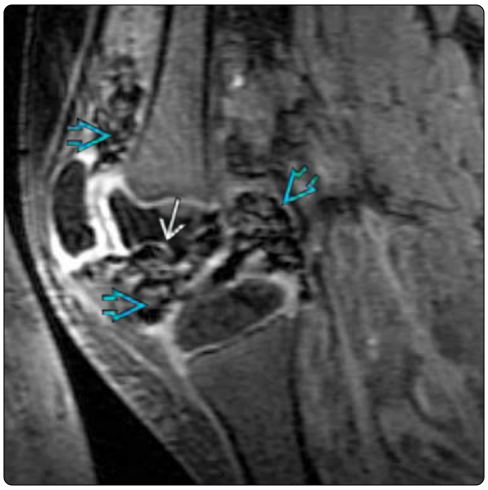


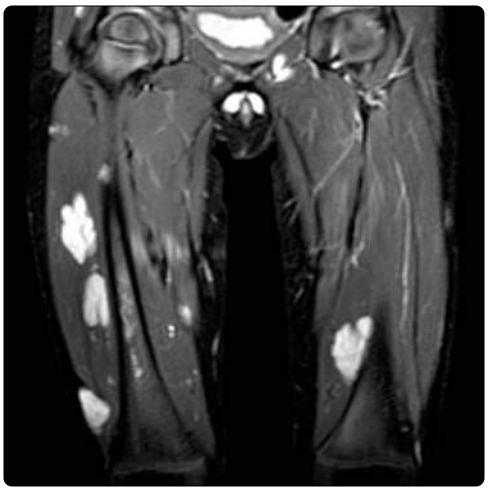


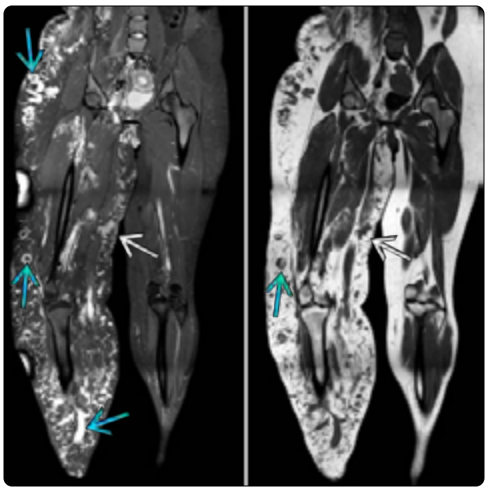
(Left) *Oblique radiograph in a patient with an extensive VM shows numerous lobular soft tissues masses containing phleboliths throughout the upper extremity. The bones have a gracile appearance, & there is longstanding radial head dislocation.* **(Right)** *Coronal T2 FS MR in the same patient shows extensive lobular & serpentine foci of fluid signal intensity throughout the subcutaneous fat of the upper extremity & chest wall. Note that the intramuscular components tend to follow the muscle's long axis.*





(Left) *Sagittal PD MR in a 9 year old with an extensive lower extremity VM shows the masses infiltrating all visible muscles & extending into the knee joint. There is marked cartilage loss with a large distal femoral erosion.* **(Right)** *Sagittal T2* GRE MR in the same patient shows "blooming" of hemosiderin within the joint due to recurrent hemarthrosis from the synovial VM. Severe cartilage loss & a large femoral erosion are again noted.*





(Left) *Coronal STIR MR in a 3-year-old patient with blue rubber bleb nevus syndrome shows multiple lobulated intramuscular fluid signal intensity masses, typical of VMs.* **(Right)** *Coronal STIR (left) & T1 (right) MRs in a teenager with Klippel-Trenaunay syndrome show fatty & bony overgrowth of the right lower extremity with extensive varicosities. Postcontrast images (not shown) confirmed that the suggested venous filling defects were due to slow flow. Note the reticular microcystic lymphatic disease.*

Lymphatic Malformation, Musculoskeletal

KEY FACTS

TERMINOLOGY

- Subtype of congenital slow/low flow vascular malformation due to error in lymphatic vessel formation
- Results in well-defined cyst-like (macrocystic) &/or infiltrative, solid-appearing (microcystic) mass of abnormal lymphatic channels
  - Individual cyst size: Macrocyst > 1 cm, microcyst < 1 cm

IMAGING

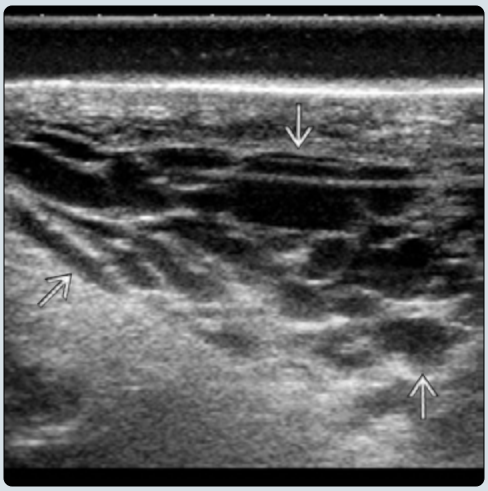
- Macrocystic LM: Lobulated, well-defined cystic lesion with numerous thin internal septations
  - ± multiple fluid-fluid levels, typically due to hemorrhage
  - Soft & compressible by US with internal swirling debris
  - Protein, blood products, chyle/fat may cause bright T1 signal intensity on MR
  - Enhancement limited to rim, septations
- Microcystic LM: More poorly defined & solid appearing
  - ± diffuse enhancement
- Locations: Face, neck, chest, axilla > > abdomen, pelvis, extremities
  - Frequently extend across tissue planes/compartments

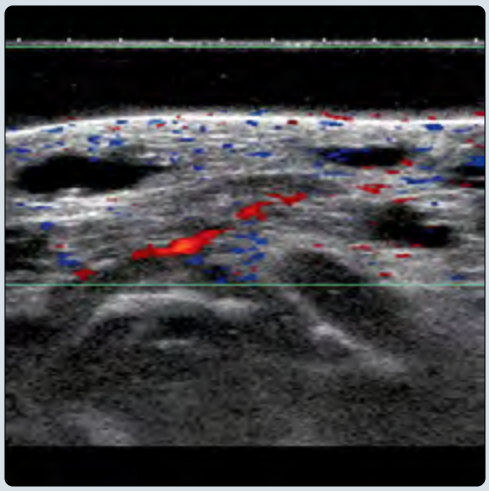
TOP DIFFERENTIAL DIAGNOSES

- Infantile hemangioma
- Venous malformation
- Arteriovenous malformation
- Soft tissue sarcoma
- Soft tissue infection

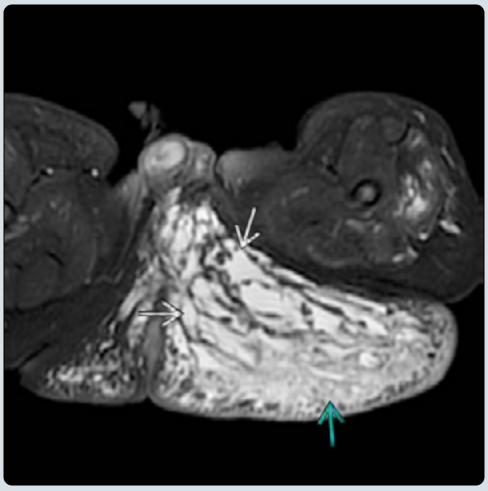
CLINICAL ISSUES

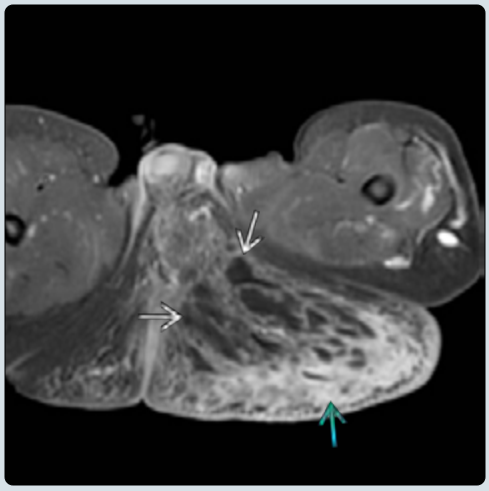
- Soft & pliable mass, often apparent at birth
  - Can compress airway or other vital structures
  - May present later with pain & rapid enlargement due to hemorrhage, inflammation, or hormonal stimulation
- Primary treatment: Surgical resection &/or percutaneous sclerotherapy
  - Reports of successful medical therapy with sirolimus





(Left) *Transverse ultrasound of the left buttock in a 2 day old with a clinically apparent capillary-veno-lymphatic malformation shows a predominantly anechoic mass with thin septations in the subcutaneous soft tissues, typical of a macrocystic lymphatic malformation.* **(Right)** *Longitudinal color Doppler ultrasound in the same patient shows no significant internal vascular flow within the cysts.*





(Left) *Axial T2 FS MR in the same patient shows contiguous cystic foci with internal septations, typical of macrocystic components. The lesion is poorly defined & infiltrative superficially with slightly lower signal intensity, typical of microcystic components. The mildly thickened septations were likely due to interval infection.* **(Right)** *Axial T1 C+ FS MR in the same patient shows mild septal enhancement of the macrocystic components with more confluent enhancement of the microcystic components.*

TERMINOLOGY

Abbreviations

- Lymphatic malformation (LM)

Synonyms

- Common (cystic) LM, lymphatic anomaly
- Avoid incorrect & confusing terminology
  - Cystic hygroma reserved for abnormal midline posterior nuchal translucency on early fetal ultrasound
    - Different than anterolateral neck LM, which does not implicate chromosomal anomalies
  - Lymphangioma implies neoplasm
    - LM not neoplastic

Definitions

- Subtype of congenital slow/low flow vascular malformation due to error in lymphatic vessel formation
  - Results in well-defined cyst-like (macrocystic) &/or infiltrative, solid-appearing (microcystic) mass of abnormal lymphatic channels
    - Individual cyst size: Macrocyst > 1 cm, microcyst < 1 cm
  - Mass lacks communication with normal lymphatics

IMAGING

General Features

- Location
  - Face, neck, chest, axilla > > abdomen, pelvis, extremities
  - Focal > > multifocal
  - Soft tissue > > bone
  - Frequently extend across tissue planes/compartments
- Size
  - Highly variable
  - May enlarge rapidly with hemorrhage, infection, or hormonal stimulation (puberty, pregnancy)
- Morphology
  - Macrocystic: Lobulated, well-defined cystic lesion with numerous thin internal septations
  - Microcystic: More poorly defined, infiltrative, & solid appearing
  - Components of both may be present

MR Findings

- **T2 FS/STIR**: Largely bright fluid signal intensity mass
  - Intervening hypointense septa
  - Fluid-fluid levels due to hemorrhage within some cysts
    - Less frequently seen on other sequences
- **T1**: Hemorrhage, protein, fat/chyle within cysts may cause bright signal intensity
- **T1 C+ FS**
  - Macrocystic LM: Thin rim/septal enhancement of cysts
    - Septations may be thicker from prior inflammation or hemorrhage
  - Microcystic LM: ± confluent enhancement of infiltrating tissue
- **MRA/MRV**: No high flow vessels intrinsic to lesion
  - Nearby normal vessels may be encased by infiltrating LM

Ultrasonographic Findings

- Grayscale ultrasound
  - Macrocystic LM: Anechoic cystic mass, usually with thin internal septations
    - ± ↑ echogenicity related to hemorrhage or proteinaceous fluid
    - Soft, compressible with swirling debris internally
  - Microcystic LM: Poorly defined region of subcutaneous soft tissue thickening
    - Mildly hypo- or hyperechoic
- Color Doppler
  - No vascular flow identified in cysts
  - Flow may be identified in septations, typically from encased normal vessels

Imaging Recommendations

- Best imaging tool
  - US often diagnostic for macrocystic LM, though deep extent & relationship to vital structures may be unclear
  - MR will typically confirm diagnosis & extent of disease
- Protocol advice
  - US: For any soft tissue mass include
    - Compression cine to evaluate internal contents
    - Doppler assessment for presence & types of vascularity
  - MR: For any soft tissue mass include
    - Subtracted (pre- from post-) contrast-enhanced FS T1
      - Particularly relevant in LM as intrinsic T1 shortening from hemorrhage/protein may confound postcontrast images

DIFFERENTIAL DIAGNOSIS

Infantile Hemangioma

- True vascular neoplasm (benign)
- Characteristic life cycle: Small or absent at birth, rapid growth during early infancy, gradual involution over years
- Solid ovoid or lobular, elongated soft tissue mass, typically in cutaneous/subcutaneous tissues
- Variable sonographic echogenicity due to mix of proliferating & involuting components
- Highly vascular by Doppler US
- Bright (but not fluid) T2 signal intensity with diffuse early enhancement on MR

Venous Malformation

- Focal mass vs. conglomeration of abnormal tubular slow flow channels
- Bright (fluid) T2 signal intensity ± fluid-fluid levels on MR
- May have prominent fat along margins or septa
- Phleboliths in soft tissue mass essentially pathognomonic

Arteriovenous Malformation

- Tangle of enlarged tortuous arteries & veins with variable soft tissue components
- High flow with shunting by ultrasound, MRA/MRV

Soft Tissue Sarcoma

- Typically firm, well-defined round or ovoid hypovascular solid soft tissue mass with variable enhancement
- May have cystic components but solid components usually visible

Soft Tissue Infection

- Cellulitis: Poorly defined soft tissue thickening with irregular serpentine pockets of fluid
- Abscess: Well-defined hypoechoic collection with mobile debris, thick wall

PATHOLOGY

General Features

- Associated abnormalities
  - Klippel-Trenaunay syndrome
    - Capillary-lymphatic-venous malformation of extremity (usually lower) + lipomatous & osseous overgrowth
    - Port-wine capillary stain on lateral extremity with lymphatic vesicles
    - Varicosities of superficial veins
      - Marginal venous system may dominate over diminutive or absent deep venous system
  - Congenital lipomatous overgrowth with vascular malformations, epidermal nevi, & skeletal anomalies
    - Truncal lipomatous masses ± LM
  - LM may be associated with trisomies 13, 18, 21, or Turner syndrome

Staging, Grading, & Classification

- 2014 revised classification by International Society for Study of Vascular Anomalies
  - Common (cystic) LM: Macrocystic, microcystic, mixed
  - Generalized lymphatic anomaly (GLA)
    - Multifocal lesions commonly involving pleura, spleen, bones (axial & appendicular skeleton)
      - Progressive cortical destruction not seen in GLA
    - Macrocystic LM often present
  - Gorham-Stout disease ("disappearing bone disease")
    - Overlap with GLA but with less extensive disease
    - Favors axial skeleton
    - Progressive regional osteolysis (hallmark of disease) due to focal microcystic LM
  - Channel-type (or "central conducting-type") LM
    - May see dilated central lymphatics &/or secondary manifestations
      - Intestinal lymphangiectasia with bowel wall thickening & abnormal mesentery causing protein-losing enteropathy
      - Chylous pleural effusions & ascites
      - Extremity edema
  - Primary lymphedema

Microscopic Features

- Variably sized channels lined by flattened endothelium
- Larger channel walls show variable amount of fibrous tissue & smooth muscle
- May be mixed with venous &/or capillary components

Immunohistochemical Features

- Endothelium stains positive for lymphatic markers
  - PROX-1 & VEGFR-3 most sensitive, specific
  - D2-40 & LYVE-1 less sensitive in large channel LM

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Soft & pliable mass without pain
- Other signs/symptoms
  - Pain &/or rapid enlargement due to hemorrhage, inflammation, or hormonal stimulation
  - Cutaneous vesicles suggest LM, though overlying skin often normal
  - Compression of airway or other vital structures
  - Diffuse limb enlargement

Demographics

- Age
  - Identified at birth or prenatally in majority of cases

Natural History & Prognosis

- LM generally grow commensurate with patient
  - May enlarge rapidly due to hemorrhage, inflammation, or hormonal stimulation
- Prognosis better for small focal lesions
  - Difficult to treat entirety of large infiltrating lesion with surgery or sclerotherapy, leading to recurrence

Treatment

- Surgical resection &/or percutaneous sclerotherapy
  - Combination often required for larger infiltrating lesions
  - Sclerotherapy primarily utilized for macrocystic disease
    - Direct injection of sclerosing agent into LM under sonographic/fluoroscopic guidance
      - Doxycycline, bleomycin, OK-432, ethanol
      - Utility of bleomycin also reported in microcystic LM
    - Major complications (skin necrosis, nerve damage, extremity swelling, muscle atrophy, disseminated intravascular coagulation) uncommon
    - Full results of sclerosis can take months to manifest
      - May take multiple staged procedures to treat lesion
- Medical therapy with sirolimus gaining favor
- Compression garments

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Multicystic pediatric soft tissue mass with fluid-fluid levels strongly suggests slow/low flow vascular malformation

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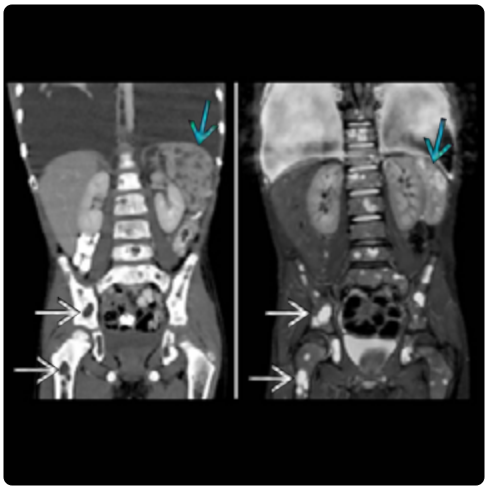
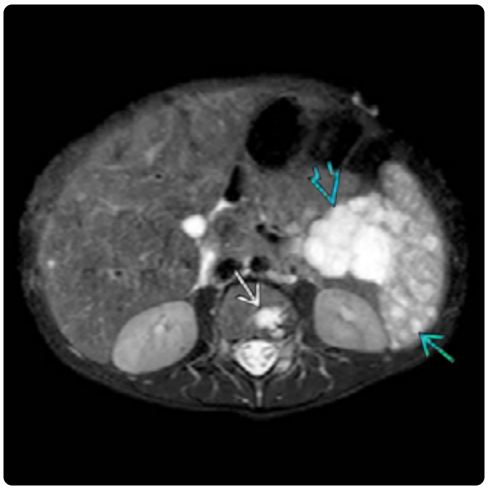
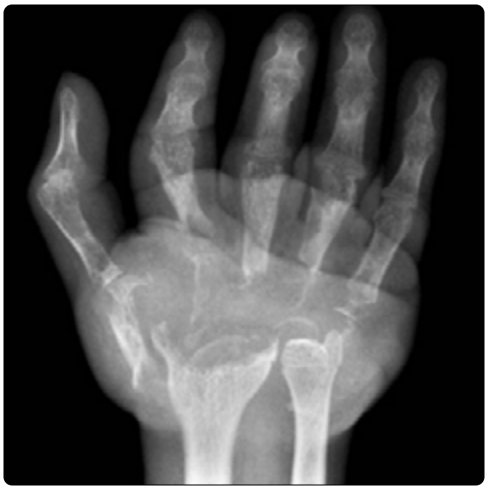
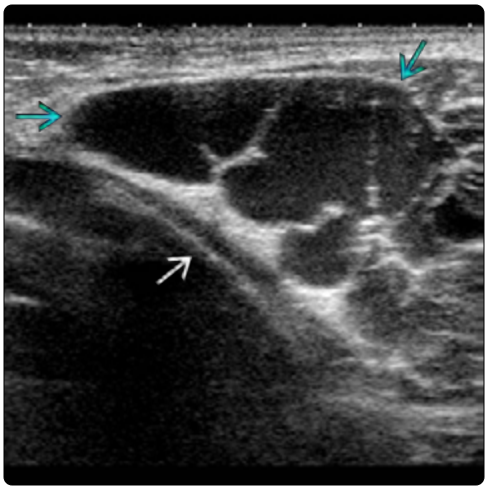


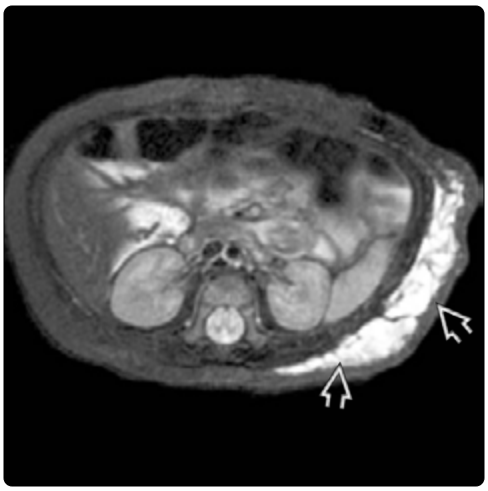


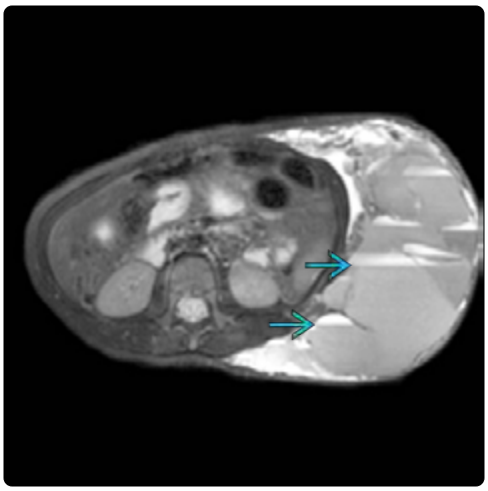
(Left) *Coronal CECT (left) & STIR MR (right) images of the abdomen in a 5-year-old patient with shortness of breath show large bilateral pleural effusions, numerous well-defined lucent (CT) & fluid signal intensity (MR) bone lesions without cortical destruction, & multiple splenic lesions.* **(Right)** *Axial T2 FS MR in the same patient shows the bone & splenic lesions as well as a lobulated macrocystic mass in the splenic hilum. The constellation of findings in this case is typical of generalized lymphatic anomaly.*





(Left) *PA radiograph in a 14-year-old patient with years of pain shows severe osteolysis of the wrist & hand, typical of Gorham-Stout disease. The bone resorption had progressed severely over 3 years compared to a prior radiograph (not shown).* **(Right)** *Ultrasound of the axillary soft tissues in a 2-year-old patient with a soft pliable mass shows a well-defined, lobulated, multicystic lesion with thin internal septations, typical of a macrocystic lymphatic malformation. Note the intact underlying rib.*





(Left) *Axial T2 FS MR shows an elongated, septated fluid signal intensity mass of the subcutaneous left flank, typical of a lymphatic malformation.* **(Right)** *Axial T2 FS MR obtained 1 month later in the same patient shows marked interval enlargement of the mass. Numerous fluid-fluid levels are now seen in the mass, typical of layering blood products due to interval hemorrhage.*

KEY FACTS

TERMINOLOGY

- Congenital high-flow vascular lesion with abnormal direct connections between arteries & veins (with no intervening capillary bed)

IMAGING

- Tangle of abnormal high-flow vessels with variable (often minimal) soft tissue mass
- Draining veins enlarged > tortuous feeding arteries
- Grayscale US: Clustered anechoic tubular channels
- Color Doppler US: Channels all fill with color; artifactual flow in surrounding soft tissues due to vibration
- Pulsed Doppler US: Numerous low-resistance (high diastolic flow) arterial waveforms & arterialized venous waveforms; spectral broadening
- MR spin-echo: Tangle of flow voids in lesion (before & after contrast); phase-encoding pulsation artifact at nidus; ± surrounding edema or soft tissue components
- CTA/MRA: Rapid contrast enhancement of nidus; early enhancement of draining veins due to shunting

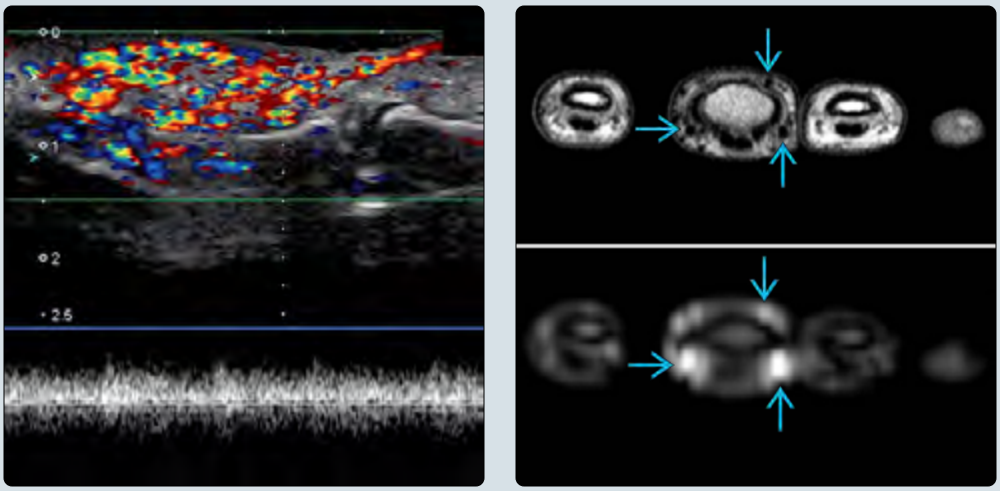
TOP DIFFERENTIAL DIAGNOSES

- Infantile hemangioma
- Venous malformation
- Arteriovenous fistula
- Soft tissue sarcoma
- *PTEN* hamartoma

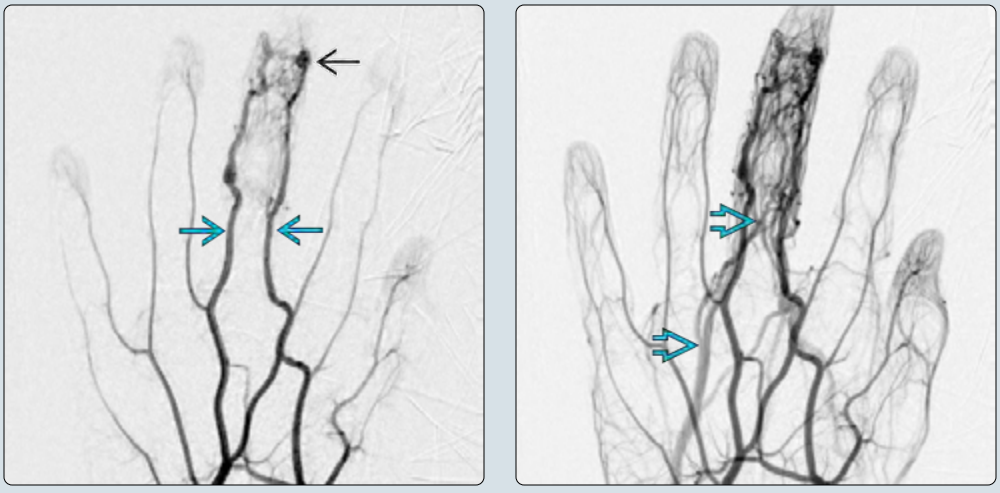
CLINICAL ISSUES

- Presentation: Warm, pulsatile mass with thrill/bruit, pain, congestive heart failure (< 2%), steal phenomenon, skin discoloration, ulceration, prolonged bleeding
- May enlarge rapidly due to hemorrhage, thrombosis, or periods of hormonal stimulation
- Treatment generally consists of transarterial embolization without, or in conjunction with, surgical resection

(Left) *Pulsed Doppler ultrasound in a 9 year old with a painful blue finger demonstrates a poorly defined but highly vascular mass along the volar aspect of the middle finger. The spectral tracing shows a very low-resistance arterial waveform with spectral broadening, typical of a shunting high-flow lesion.* **(Right)** *Axial PD (top) & 2D TOF (bottom) MRs in the same patient show numerous enlarged vessels (flow voids on PD & flow-enhancement on TOF) throughout the visualized digit.*



(Left) *Arterial phase digital subtraction angiogram of the hand in the same patient shows a nidus of abnormal tangled arteries in the distal middle finger (3rd digit) with enlargement of the supplying digital arteries.* **(Right)** *DSA in the same patient, slightly later in the same run, shows early visualization of enlarged draining veins, consistent with shunting. Due to the patient's symptoms, the digit was ultimately amputated with an arteriovenous malformation confirmed on pathologic examination.*



TERMINOLOGY

Abbreviations

- Arteriovenous malformation (AVM)

Definitions

- Congenital high-flow vascular lesion with abnormal direct connections between arteries & veins (i.e., no intervening capillary bed)

IMAGING

General Features

- Morphology: Cluster of abnormal high-flow vessels; variable (but often minimal) associated soft tissue mass; draining veins enlarged > tortuous feeding arteries
- Location: Head & neck (~ 47%), extremities (~ 28%)
- **Radiograph**: ± mass-like density of soft tissues without phleboliths; rare destructive bone involvement; chest radiograph may show cardiomegaly from heart failure
- **Ultrasound**: Often used for initial investigation of mass
  - Grayscale: Clustered anechoic tubular channels (high vessel density) ± adjacent soft tissue abnormalities
  - Color Doppler: Channels all fill with color flow; artifactual flow in surrounding soft tissues due to vibration
  - Pulsed Doppler: Numerous low-resistance (high diastolic flow) arterial waveforms with spectral broadening; arterialized venous waveforms
- **MR**
  - Spin-echo: Tangle of flow voids in lesion (before & after contrast) due to high flow; phase-encoding pulsation artifact of nidus; ± surrounding edema or soft tissue components (in up to 50%, especially multicompartmental AVMs)
  - Gradient-echo: High-flow vessels typically bright before & after contrast
- **CTA/MRA**: Useful for characterizing mass, determining involvement of adjacent tissues, & planning embolization or surgery through measurement & mapping of vessels
  - Rapid contrast enhancement of nidus
    - Time-resolved/dynamic MRA: "Contrast rise time" < 20 seconds highly specific for high-flow lesions
  - Early enhancement of draining veins due to shunting

DIFFERENTIAL DIAGNOSIS

Infantile Hemangioma

- Discrete, heterogeneous, typically subcutaneous, high-flow, benign soft tissue neoplasm in infant
- Ultrasound shows patchy regions of ↑ & ↓ echogenicity
  - High vessel density by color Doppler (not grayscale)
  - Low-resistance arterial waveforms during proliferation (but no shunting)

Venous Malformation

- Discrete or infiltrating low-flow mass, often intramuscular
- Phleboliths in pediatric mass virtually diagnostic
- Multiple high-signal serpentine channels on T2 FS MR

Arteriovenous Fistula

- Direct high-flow communication between artery & vein without surrounding tangle of abnormal vessels

Soft Tissue Sarcomas

- Typically well-circumscribed, solid mass
- Most have low to intermediate levels of vascularity

PTEN Hamartoma of Soft Tissue

- Heterogeneous mass with variable soft tissue components, including myxomatous & fatty elements
- High-flow vessels typical
- Phenotype of *PTEN* mutation: Macrocrania, skin lesions

PATHOLOGY

General Features

- Genetics
  - *RASA1* (angiogenic gene) mutations
    - Capillary malformation-arteriovenous malformation
    - Some forms of Parkes Weber syndrome
  - Mutations of *ENG*, *ACVRL1*, *SMAD4* genes
    - Hereditary hemorrhagic telangiectasia

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Warm, pulsatile mass with thrill/bruit, pain
- Other signs/symptoms
  - Congestive heart failure (< 2%), steal phenomenon, skin discoloration, ulceration, prolonged bleeding
  - May enlarge rapidly related to hemorrhage, thrombosis, or hormonal stimulation
  - Limb length discrepancy: Bony overgrowth

Demographics

- ~ 80% of superficial AVMs present in childhood
  - Large shunting lesions may present soon after birth
  - Hormonal stimulation of puberty or pregnancy → ↑ AVM size & symptoms in 2nd/3rd decades of life

Natural History & Prognosis

- Schobinger classification: Outlines progressive clinical course of untreated AVMs
  - Stage 1 (quiescence)
  - Stage 2 (expansion): ↑ pulse & thrill
  - Stage 3 (destruction): Local destruction with pain, ischemia, necrosis
  - Stage 4 (decompensation): High-output cardiac failure

Treatment

- Cure rarely achievable; main goal: Clinical improvement
- Conservative management: Compression garments
- Transarterial embolization
  - 75% volume devascularization rates reported in 47-94%
- Surgical excision
  - Resection best < 24 hours after embolization
- Recurrence rate: > 80%

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Fibromatosis

KEY FACTS

TERMINOLOGY

- Synonyms: Aggressive fibromatosis, extraabdominal desmoid tumor, musculoaponeurotic fibromatosis
- Group of benign disorders with fibrous growth & tendency to slowly infiltrate adjacent tissues

IMAGING

- Homogeneous or heterogeneous with low T1 & T2 signal
- May be well defined or infiltrative
- Superficial
  - Slow growing
  - Children: Calcifying aponeurotic fibroma, lipofibromatosis, inclusion body fibromatosis
  - Adult: Plantar, palmar fibromatosis
- Deep
  - Large, can be more rapidly enlarging
  - Children: Fibromatosis colli, infantile myofibroma, & myofibromatosis
  - Teenage/adult: Desmoid-type aggressive fibromatosis
    - Typically intermuscular location: Split fat sign
    - Extends along fascial planes: Fascial tail sign, may extend distant from lesion
- Features helpful in diagnosis
  - Neonatal sternocleidomastoid muscle: Fibromatosis colli
  - Multiple in young children: Infantile myofibromatosis
  - Adipose tissue: Lipofibromatosis

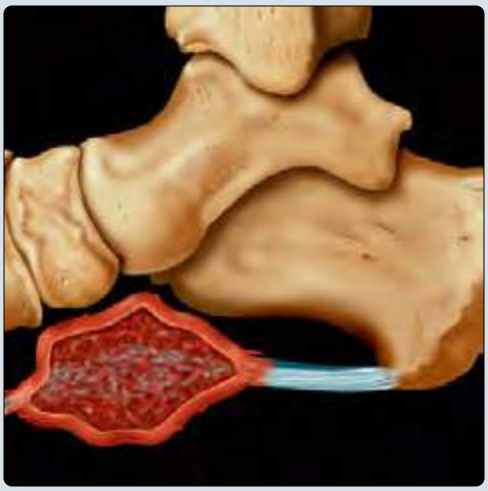
CLINICAL ISSUES

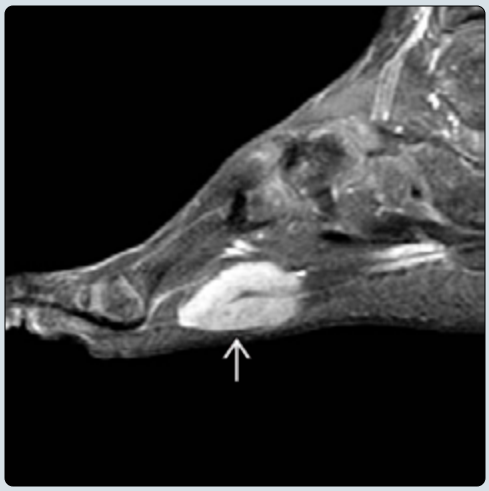
- Wait & see in some cases: May regress or stop growing
- Surgical excision with wide margins
  - Complete excision often difficult due to infiltration
- Frequent local recurrence: 30-65%
- Lacks metastatic potential

DIAGNOSTIC CHECKLIST

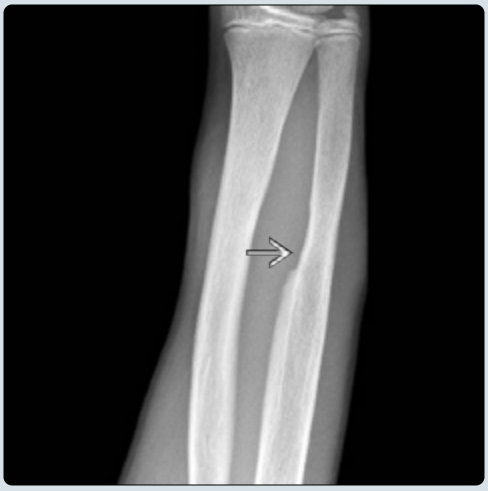
- Can mimic soft tissue sarcomas
- Due to slow growth, comparison to multiple prior exams recommended to appreciate interval change

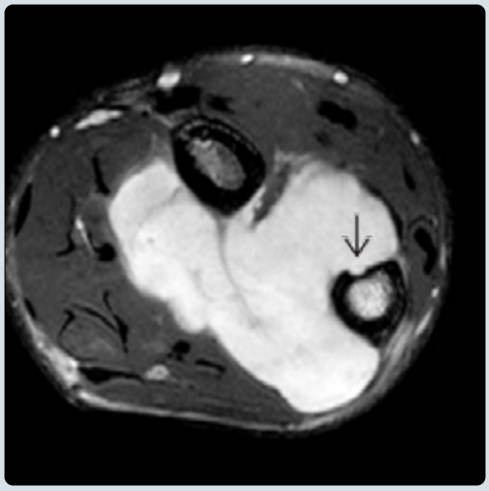
(Left) *Sagittal graphic shows a soft tissue mass of plantar fibromatosis (red) along the plantar aspect of the foot.* **(Right)** *Sagittal T1 C+ FS MR shows a foot mass in a 9-year-old child who presents after an injury on rocks. The mass shows focal, intense contrast enhancement ⇨ along the plantar aponeurosis. This lesion was isointense to muscle on T1 & T2 images (not shown). Biopsy confirmed plantar fibromatosis, which may appear as a well- or ill-defined soft tissue mass on MR.*





(Left) *AP radiograph in a 14-year-old boy who injured his forearm 2 weeks prior shows remodeling of the ulna with cortical scalloping of the lateral margin ⇨.* **(Right)** *Axial T2 FS MR in the same teenager shows a hyperintense mass in the region of the interosseous membrane between the radius & ulna. Notice the cortical remodeling of the volar margin of the ulna ⇨ by this desmoid-type fibromatosis.*





TERMINOLOGY

Synonyms

- Aggressive fibromatosis, extraabdominal desmoid tumor, musculoaponeurotic fibromatosis

Definitions

- Group of benign disorders with fibrous growth & tendency to slowly infiltrate adjacent tissues
- Frequently recurs but lacks metastatic disease

IMAGING

General Features

- Location
  - Superficial
    - Slow growing
    - Children: Calcifying aponeurotic fibroma, lipofibromatosis (previously known as infantile fibromatosis of nondesmoid type), inclusion body fibromatosis (dorsal digits of hands & feet)
    - Adult: Plantar, palmar fibromatosis
  - Deep
    - Large & more rapidly enlarging
    - Children: Fibromatosis colli, infantile myofibroma, or myofibromatosis
    - Adult: Desmoid-type fibromatosis (aggressive fibromatosis) of abdominal wall or other sites
- Size
  - Superficial type up to 5 cm
  - Deep type usually < 10 cm but can be quite large; may be up to 20 cm (especially in Gardner syndrome)
- **Palmar fibromatosis**
  - Dupuytren contracture
  - Most commonly > 65 years old
- **Plantar fibromatosis**
  - Most commonly 30-50 years old
  - Well- or ill-defined soft tissue masses along deep plantar aponeurosis
  - Treatment: Conservative with surgery used for larger &/or infiltrative lesions that fail conservative treatment
    - 20-40% recurrence, typically during 1st postop year
- **Desmoid-type fibromatosis (aggressive fibromatosis)**
  - Infantile fibromatosis, extraabdominal desmoid tumor, musculoaponeurotic fibromatosis
  - 2nd to 4th decades (peak: 25-35 years old)
  - F > M in younger
  - Shoulder/upper arm most common location
  - Erosions, scalloping, or bowing of bones
  - Nodular (more common in adults) or infiltrative (children) patterns
  - Higher potential for recurrence, locally aggressive
  - May spontaneously regress
- **Abdominal wall fibromatosis**
  - Most commonly rectus abdominis muscle & adjacent fascia
  - Often related to recent pregnancy (in past year)
- **Myofibromatosis**
  - Most common in infancy but may occur in adults
  - ↑ in size & number to 1 year of age, then can regress
  - 3 types: Solitary, multicentric ± visceral involvement
  - Bones, muscle, subcutaneous tissue, viscera
  - ± Ca^{2+}
  - May have target appearance on MR or US (center mildly hyperintense on T1 & nonenhancing)
  - Sharply defined bubbly lucent lesions (metaphyseal > diaphyseal), often relatively symmetric
- **Fibromatosis colli**
  - 2-4 weeks old at presentation, often with history of breech presentation, instrumentation, & primiparous birth
  - Enlargement of sternocleidomastoid muscle, variably heterogeneous
  - Diagnosis most commonly made upon physical exam, sometimes on US
  - Self-limiting, spontaneously resolves, no recurrence; physical therapy may help
- **Fibrous hamartoma of infancy**
  - Neonates → young children (typically first 2 years of life, 1/4 congenital)
  - Subcutaneous or reticular dermis, 0.5 → 4.0 cm
  - Axilla, shoulders, inguinal region, & chest wall most common locations
  - Excision usually curative, excellent prognosis
  - MR: Varying amounts of fibrous & fatty tissue
    - May have streaks of fat

Radiographic Findings

- Soft tissue mass
- ± periosteal reaction, cortical destruction, scalloping, or erosions

MR Findings

- T1WI
  - Typically hypointense to muscle; can be isointense
  - Frequently centered in intermuscular location with rim of surrounding fat (split fat sign)
- T2WI
  - Hyperintense or heterogeneous with bands of hypointense signal (fibrous components)
    - Depends on cellular compared to collagen components
  - Often insinuates slowly along fascial planes (fascial tail sign); may extend distance from main lesion
- DWI
  - Mean ADC value higher than malignant soft tissue tumors
- T1WI C+
  - Intense enhancement

Ultrasonographic Findings

- Nonspecific soft tissue mass with variable echogenicity; may be well circumscribed or infiltrative & poorly defined

DIFFERENTIAL DIAGNOSIS

Congenital Infantile Fibrosarcoma

- < 5 years old, typically presents < 2 years of age, commonly neonatal
- Histologic diagnosis difficult
- Can erode adjacent bone
- Heterogeneous enhancement, may be cystic
- ± metastatic disease (truncal > extremity)

- Better prognosis than adults

Adult Fibrosarcoma

- Teenagers → adults
- Can contain Ca^{2+} or ossification
- Metastases
- Higher cellularity, more mitoses with nuclear pleomorphism
- ± necrotic or hemorrhagic foci

Synovial Sarcoma

- In close proximity to joint
- Amorphous Ca^{2+}
- May be small & cystic appearing before contrast

Rhabdomyosarcoma

- ± necrotic or hemorrhagic foci
- Intermediate to high T2 signal, variable enhancement
- Metastases

Venous Malformation

- Low-flow lesion with phleboliths &/or thrombi, fluid-fluid levels, & patchy or diffuse enhancement on MR

PATHOLOGY

General Features

- Etiology
  - Activation of β-catenin pathway
- Genetics
  - Most sporadic
  - Wnt/β-catenin pathway gene mutation
  - *APC* gene mutation (long arm of chromosome 5q21-22)
- Associated abnormalities
  - Familial adenomatosis fibromatosis (FAP)
    - 1,000x greater risk of developing fibromatosis due to *APC* gene
    - Fibromatosis in 10-20% FAP patients
  - Trisomies 7, 8, 14, 20
  - Gardner syndrome

Gross Pathologic & Surgical Features

- Fibrous tissue proliferation with varying amounts of collagen
- Locally aggressive infiltrative behavior & recurrences

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Palpable mass
  - Firm, poorly circumscribed, slowly growing mass
  - Tends to extend beyond palpable limits
  - Flexion contractures
  - ± tenderness (related to nerve compression or infiltration)

Demographics

- Age
  - Puberty to 40s (peak age: 23 years)
  - Infantile fibromatosis typically < 2 years, rarely > 5 years old
- Ethnicity
  - More common in Caucasians
- Epidemiology
  - 2-4/1 million per year

Natural History & Prognosis

- Recurrence: 30-65% locally
  - Larger lesions, < 30 years old, female predominance, location
  - Controversy over positive resection margin

Treatment

- With tissue confirmation: Wait & see; some lesions regress
- Surgical excision with wide margins
  - For abdominal wall, symptomatic lesions, & progressive disease
  - Often wide excision is not feasible due to loss of function
- Antiestrogen (tamoxifen) therapy, NSAIDs
- Anthracyclines
- Radiation or adjuvant chemotherapy
- Amputations (occasional) for palliative repeated recurrences & continued progression

DIAGNOSTIC CHECKLIST

Consider

- Can mimic soft tissue sarcomas (fibrosarcomas)

Image Interpretation Pearls

- Often contains regions of ↓ signal on T2 FS & T1 C+ FS MR
  - May represent inactive fibrous rather than active cellular components
- Due to slow growth, comparison to multiple prior exams recommended to appreciate interval change

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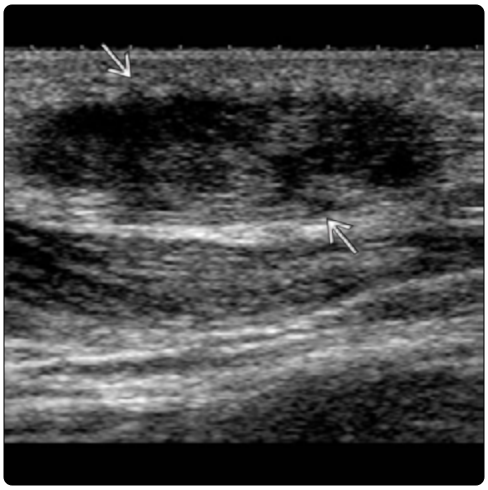
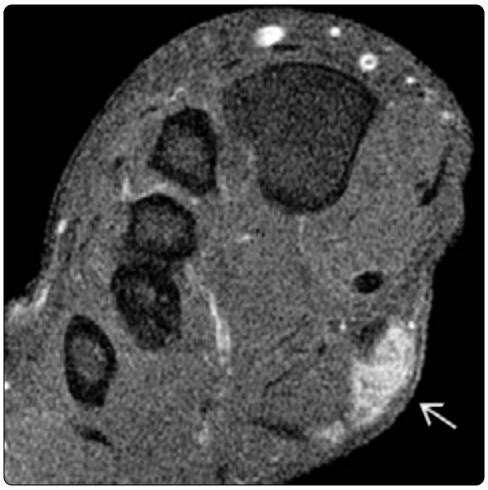
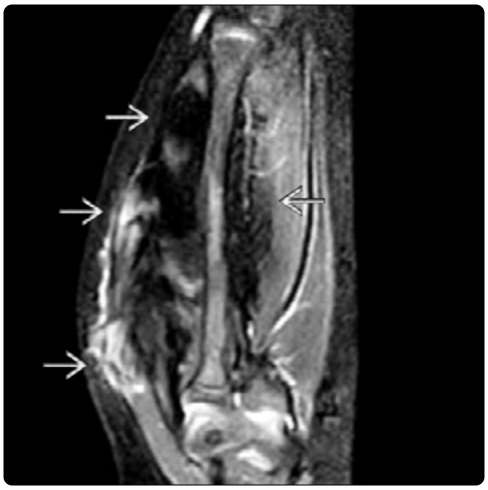
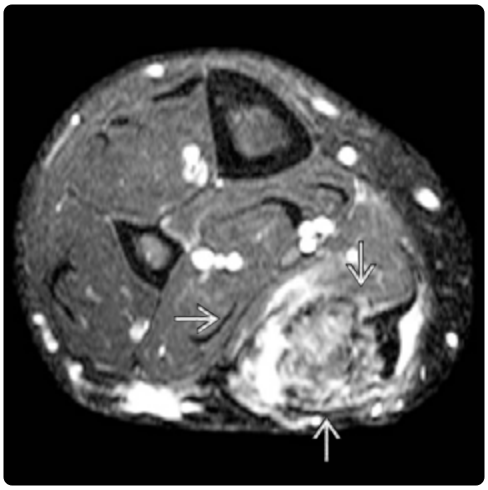


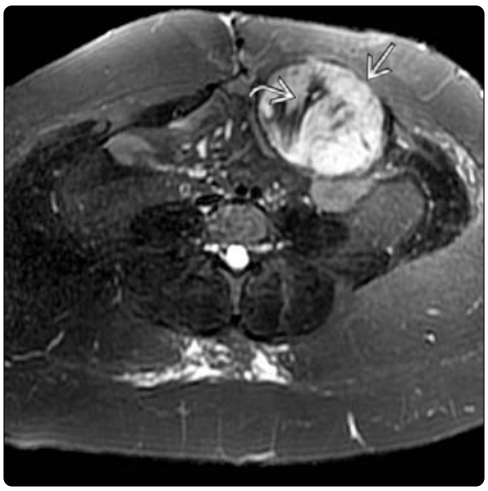


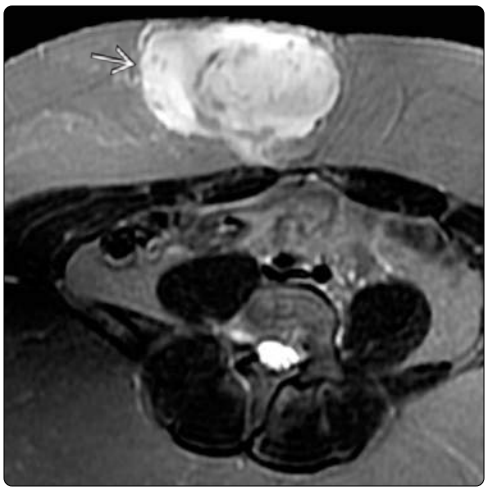
(Left) *Longitudinal US in a teenager shows a well-circumscribed, predominantly hypoechoic mass in the plantar foot. US of fibromatosis is often nonspecific.* **(Right)** *Axial T2 FS MR in the same patient 3 years later shows a hyperintense lesion inseparable from the plantar aponeurosis due to recurrent plantar fibromatosis.*





(Left) *Coronal STIR MR of the forearm in a child just under 2 years of age (presenting with a mass) shows a heterogeneous, predominantly hypointense, ill-defined, & infiltrative soft tissue mass. This was biopsied & proven to be desmoid-type fibromatosis.* **(Right)** *Axial T2 FS MR shows a heterogeneous, ill-defined soft tissue mass in the posterior right calf of a 16-year-old patient who had recurrent desmoid-type fibromatosis.*





(Left) *Axial T2 FS MR shows a heterogeneous mass within the left rectus abdominis muscle of a 25 year old with Down syndrome & a history of multiple abdominal surgeries. Notice the hypointense bands within the mass.* **(Right)** *Axial T2 FS MR shows a hyperintense subcutaneous mass extending to the anterior margin of the right rectus abdominis muscle in a 17-year-old boy. The mass was intensely enhancing (not shown) & proved to be a desmoid fibromatosis.*

Infantile Myofibroma/Myofibromatosis

KEY FACTS

TERMINOLOGY

- Infantile myofibroma: Solitary benign fibrous tumor of young children with high rate of spontaneous regression
- Myofibromatosis: Multicentric disease with soft tissue & bone lesions ± visceral involvement
  - Poorer prognosis with visceral lesions

IMAGING

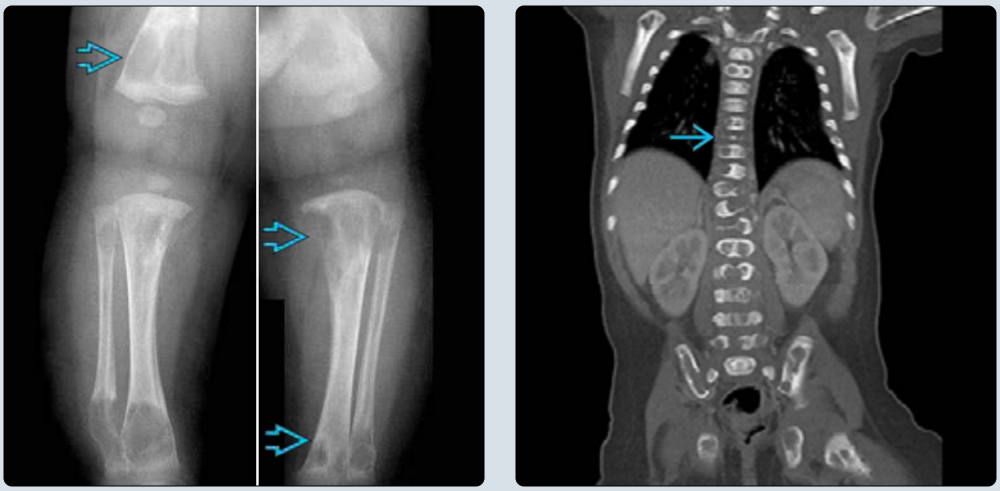
- Myofibroma: Solitary, solid soft tissue mass in infant
  - ± internal Ca^{2+}, bizarre adjacent bone remodeling
  - Hypovascular internally by Doppler
  - T2 FS MR: Variable internal signal intensity
  - T1 C+ FS MR: Peripheral/rim enhancement ("target")
  - Typical locations
    - Cutaneous, subcutaneous, muscular: 86%
    - Head & neck (50%) > trunk > extremities
- Myofibromatosis (25-55% of myofibroma cases)
  - Soft tissue masses
  - Well-defined bony metaphyseal/metadiaphyseal lucent lesions ± remodeling, cortical loss
  - Visceral involvement in 25-37% of multicentric cases
    - Lungs > gastrointestinal tract > heart > liver

PATHOLOGY

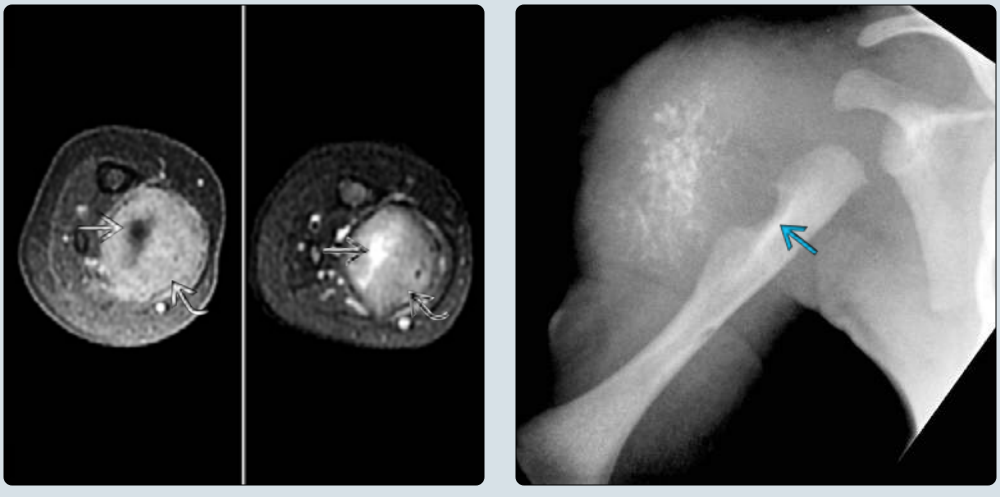
- Peripheral clusters of spindled myofibroblastic cells
- Central cellularity & vascularity similar to hemangiopericytoma

CLINICAL ISSUES

- Presentations: Soft tissue mass ± red-purple skin nodules
  - Perinatal in 50%; 90% diagnosed by age 2
- Solitary/multicentric without visceral involvement: High rate of spontaneous regression over 1-2 years; 1% mortality
  - Observation vs. surgical resection
- Multicentric with visceral involvement: 48-75% mortality historically due to obstruction, organ failure
  - Favorable outcomes with low-dose chemotherapy



(Left) *Radiographs in a 2 month old with fussiness show numerous well-circumscribed, lucent lesions of the lower extremities bilaterally. The distribution is relatively symmetric in the metaphyses/metadiaphyses. Some lesions show remodeling, while others show cortical loss. Several of the ovoid eccentric lesions have a "side-by-side" configuration. **(Right)** Coronal CT (same infant) with myofibromatosis shows lucent lesions of most of the visualized bones with 1 level of complete vertebra plana.*



(Left) *Axial T1 C+ FS (left) & axial T2 FS (right) MR images in a 4 month old with multiple soft tissue masses show peripheral enhancement & T2 hypointensity with central nonenhancement & T2 hyperintensity of an infantile myofibroma.* **(Right)** *AP radiograph in a newborn with a large shoulder mass shows clustered speckled Ca^{2+} in the central necrotic portion of the mass. There is unusual remodeling of the adjacent humerus with expansion & focal surface scalloping. Myofibroma was confirmed on biopsy.*

TERMINOLOGY

Definitions

- Infantile myofibroma: Solitary benign fibrous tumor of young children with high rate of spontaneous regression
- Myofibromatosis: Multifocal disease
  - Multicentric without visceral involvement: Soft tissue &/or bone lesions
  - Multicentric with visceral involvement (or generalized): Additional organ systems involved, poorer prognosis

IMAGING

General Features

- Best diagnostic clue
  - Myofibroma: Solitary, solid, centrally nonenhancing soft tissue mass in newborn
  - Myofibromatosis: Numerous, relatively symmetric, well-defined metaphyseal/metadiaphyseal lucent lesions ± remodeling, cortical loss
- Location
  - Cutaneous, subcutaneous, muscular: 86%
  - Solitary lesions: Head & neck (50%) > trunk > extremities
  - Multicentric disease in 25-55%
    - Visceral involvement in 25-37% of multicentric cases
      - Lungs > gastrointestinal tract > heart > liver
    - Uncommon forms
      - Large, extensive retroperitoneal/paraspinal lesions
      - Intracranial involvement, intra- or extraaxial

Radiographic Findings

- Soft tissue mass ± internal Ca^{2+}, bizarre adjacent bone remodeling
- Multifocal extensive, well-circumscribed, lucent metaphyseal, & metadiaphyseal (> central diaphyseal) lesions of bone
  - Relatively symmetric distribution
  - ± cortical loss, expansile remodeling
  - Periosteal reaction usually absent or solid
  - Can cause vertebra plana
  - Sclerosis may represent early healing

CT Findings

- Enhancing rim with necrotic center ± central Ca^{2+}
- Bone lesions follow radiographic appearance

MR Findings

- T2WI FS: Variable internal signal intensity
- T1WI C+ FS: Peripheral/rim enhancement ("target")
  - Diffuse enhancement less common

Ultrasonographic Findings

- Heterogeneous, may be hypo- or isoechoic
- Hypovascular internally by Doppler

DIFFERENTIAL DIAGNOSIS

Congenital/Neonatal Soft Tissue Mass

- Vascular anomaly
  - Neoplasm
    - Hemangioma, infantile or congenital
    - Kaposiform hemangioendothelioma
  - Malformation
    - Venous
    - Lymphatic
- Other fibrous lesions
  - Fibrosarcoma
  - Fibrous hamartoma of infancy
- Teratoma
- Neuroblastoma

Multifocal Infantile Lesions

- Langerhans cell histiocytosis
- Metastatic neuroblastoma
- Multifocal vascular anomalies
- Disseminated infection

PATHOLOGY

Microscopic Features

- Peripheral clusters of spindled myofibroblastic cells
- Central cellularity & vascularity similar to hemangiopericytoma

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Soft tissue mass ± red-purple skin nodules

Demographics

- Age
  - Perinatal in 50%; 90% diagnosed by age 2

Natural History & Prognosis

- Solitary/multicentric without visceral involvement: High rate of spontaneous regression over 1-2 years
  - May increase in size & number prior to regression
  - Overall 1% mortality
- Multicentric with visceral involvement: 48-75% mortality historically due to obstruction, organ failure

Treatment

- Solitary: Observation or surgical resection
  - 10% recurrence
- Multicentric with visceral involvement: Favorable outcomes with low-dose chemotherapy

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Plexiform Neurofibroma

KEY FACTS

TERMINOLOGY

- Neurofibroma: Type of benign peripheral nerve sheath tumor with 3 subtypes (localized, diffuse, plexiform)
  - Plexiform subtype occurs almost exclusively in neurofibromatosis type 1
  - Extensive plexiform neurofibromas → massive enlargement of body part (elephantiasis neuromatosa)
- Malignant peripheral nerve sheath tumor (MPNST)
  - Plexiform neurofibromas at ↑ risk → MPNST

IMAGING

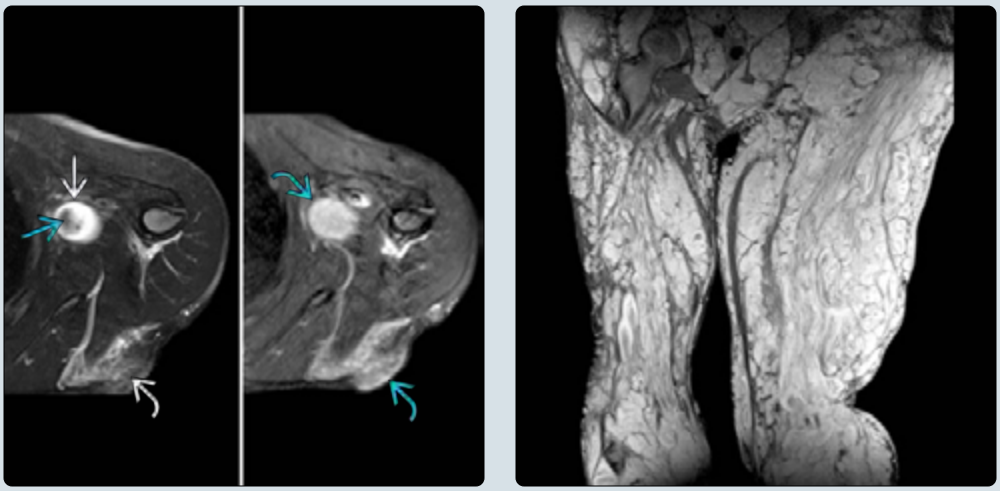
- Plexiform neurofibroma: Tortuous, lobulated expansion of long segments of nerves & branches ("bag of worms")
- Radiographs: Nonspecific soft tissue mass without Ca^{2+}; bony remodeling/erosions or mass effect on vital structures may occur from numerous adjacent neurofibromas
- T1 MR: Hypo- to isointense to skeletal muscle
  - Split fat sign: Rim of fat surrounding surrounding mass
- T2 MR: Predominately hyperintense lobules along nerve
  - Target sign: ↑ peripheral & ↓ central signal intensity
- T1 C+ MR: Heterogeneous or central enhancement
- MR features suggesting MPNST: ↑ size, cysts, peripheral enhancement, perilesional edema, heterogeneity on T1 (specific to NF1)
- FDG PET: SUVmax ≥ 3.5 sensitive but not specific for MPNST

TOP DIFFERENTIAL DIAGNOSES

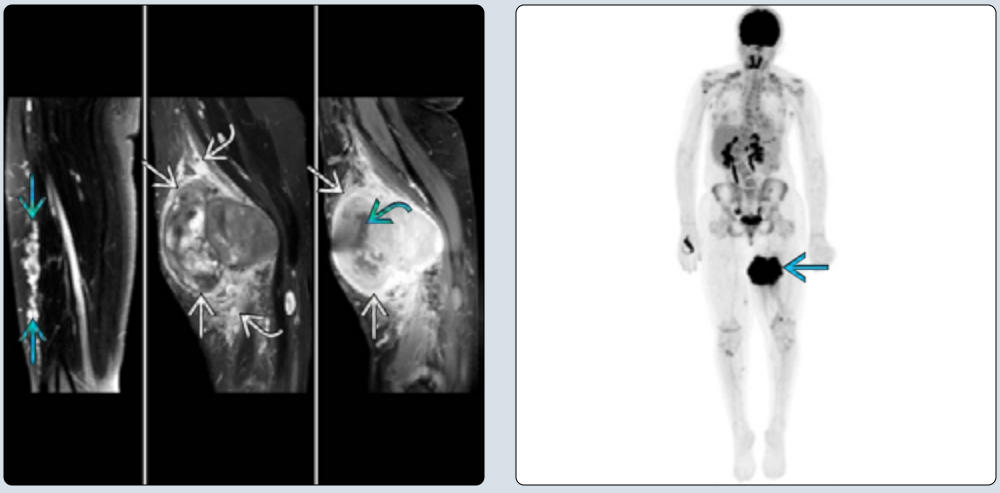
- Venous malformation, soft tissue sarcoma, MPNST, polyneuropathy

CLINICAL ISSUES

- Symptoms include pain, neurologic deficit, mass effect
- Treatment of plexiform neurofibroma typically conservative due to inseparability of lesion & involved nerve
  - Total resection of lesion → extensive neurologic deficit
  - Surgery only as needed for debulking symptomatic lesions or foci concerning for MPNST



(Left) *Axial T2 FS MR (left) in a 14 year old with neurofibromatosis type 1 (NF1) shows a localized neurofibroma in the left axilla with a target sign (high peripheral ➡ & low central ⇨ signal). There is a poorly defined diffuse neurofibroma posteriorly ➡. Axial T1 C+ FS MR (right) shows enhancement of the lesions ⇨.* **(Right)** *Coronal STIR MR in a 15 year old with NF1 shows extensive plexiform neurofibromas with massive enlargement of both thighs (elephantiasis neuromatosa).*



(Left) *Initial sagittal T2 FS MR (left) in an NF1 patient shows a thin elongated plexiform neurofibroma in the anterior thigh ⇨. Seven years later, this mass had rapidly enlarged with sagittal T2 FS (middle) & T1 C+ FS (right) MRs showing marked enlargement & heterogeneity of the mass ➡ with central nonenhancement ⇨ from necrosis. Perilesional edema ➡ is also present.* **(Right)** *Coronal FDG PET in the same patient shows increased uptake ⇨ (SUVmax of 13.7) within the mass. Pathology confirmed an MPNST.*

TERMINOLOGY

Abbreviations

- Neurofibromatosis type 1 (NF1)
- Benign peripheral nerve sheath tumor (BPNST)
- Malignant peripheral nerve sheath tumor (MPNST)

Definitions

- Neurofibroma: Type of BPNST
  - 3 neurofibroma subtypes: Localized, diffuse, plexiform
- MPNST: Spindle cell sarcoma arising from nerve or BPNST
  - Plexiform neurofibromas at ↑ risk of MPNST

IMAGING

General Features

- Morphology
  - Plexiform type: Tortuous expansion of nerve(s) & branches by elongated lobules of varying size
    - May be relatively limited to courses of nerves vs. massive lobular proliferations engulfing & compressing adjacent tissues
      - Bag of worms appearance
      - Massive disfiguring enlargement of body part (elephantiasis neuromatosa)
  - MPNST: ↑ heterogeneity with indistinct margins

Radiographic Findings

- Nonspecific soft tissue swelling without Ca^{2+}
- Large & numerous plexiform neurofibromas → bony remodeling/pressure erosions, mass effect on vital structures

Ultrasonographic Findings

- Predominantly hypoechoic lobular mass(es) along nerve(s)
  - May show ↑ echogenicity centrally

MR Findings

- T1WI
  - Hypo- to isointense to skeletal muscle
  - Split fat sign: Rim of fat surrounding tumor
- T2WI FS or STIR
  - Predominately ↑ signal intensity
  - Target sign: ↑ peripheral, ↓ central signal intensity when individual lobule viewed in cross section
- T1WI C+
  - Variable: Often heterogeneous & diffuse
  - Lesions with target sign on T2WI may show only central enhancement

Nuclear Medicine Findings

- FDG PET/CT
  - SUVmax ≥ 3.5: Concerning for MPNST

Imaging Recommendations

- Best imaging tool
  - MR to characterize neurofibromas & evaluate depth & relationship to vital structures

DIFFERENTIAL DIAGNOSIS

Venous Malformation

- Numerous thin low signal septa in fluid-filled mass
  - ± fluid-fluid levels of stagnant blood
- Gradual patchy enhancement (often peripheral > central)
- May have fat along margins & septa
- Round lucent-centered Ca^{2+} (phleboliths) confirmatory

Soft Tissue Sarcoma

- Well-defined solid, firm mass without entering/exiting nerve; no target sign or split fat sign
- Typically intermediate to high signal on T2/STIR MR; variable enhancement

Malignant Peripheral Nerve Sheath Tumor

- MR features favoring MPNST over plexiform neurofibroma (≥ 2: Specificity 90%, sensitivity 61%)
  - Largest dimension > 5 cm, cystic foci, peripheral enhancement, perilesional edema, heterogeneity on T1 (in NF1)
  - Also consider with preferential growth or loss of target sign (if previously present)

Polyneuropathy

- Various acute & chronic polyneuropathies cause diffuse enlargement of multiple adjacent nerves ± ↑ enhancement
- Nerve architecture maintained without "target" lobules

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Plexiform type: Symptoms depend on location/extent
    - Pain & neurologic deficit vary with nerve involved
    - Symptoms from compression of adjacent structures (e.g., airway, spinal cord)
  - MPNST: ↑ pain, ↑ neurologic defect, & rapid growth

Natural History & Prognosis

- Neurofibromas (of all types) typically grow slowly
- Transformation of neurofibroma to MPNST
  - Lifetime risk of MPNST in patients with NF1: 8-13%
  - MPNST has poor prognosis
    - Recurrence: ~ 40-65%; distant metastases: ~ 40-68%
    - 5-year survival: 23-44%

Treatment

- Plexiform neurofibromas
  - Typically conservative management (as total resection → extensive neurologic deficit)
  - Surgery reserved for debulking of symptomatic lesions &/or masses otherwise concerning for MPNST
- MPNST: Wide surgical excision + chemotherapy & radiation

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Rhabdomyosarcoma, Musculoskeletal

KEY FACTS

TERMINOLOGY

- Mesenchymal sarcoma arising from rhabdomyoblasts (primitive muscle cells); lacks normal differentiation into skeletal muscle
  - Embryonal RMS (60-70% of childhood RMS)
    - Most common type in patients < 15 years old
    - Most commonly genitourinary, head & neck, retroperitoneum
  - Alveolar RMS (20% of RMS)
    - Average age: 15 years old
    - Most commonly extremity, trunk, & perianal/perirectal
  - Undifferentiated RMS
    - 30-50 year olds, rarely in children

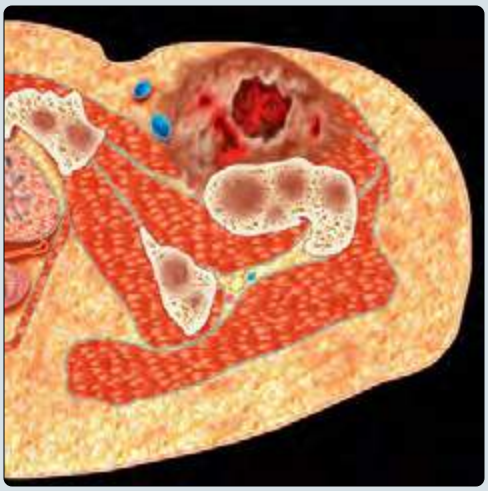
IMAGING

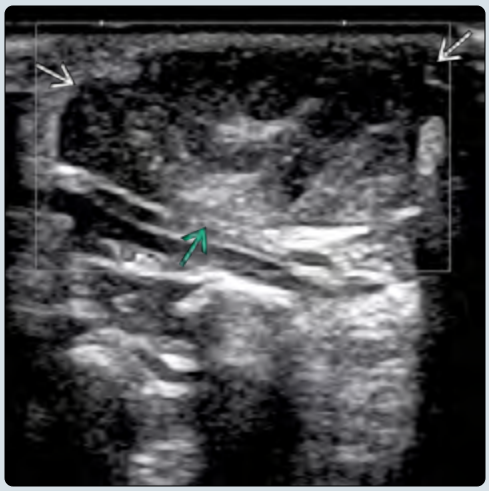
- Best clue: Solid, mildly heterogeneous intramuscular mass without significant surrounding soft tissue edema
- Usually round with well-circumscribed, lobular margins
- May have tail of tumor extending proximal/distal
- Radiographs: Ca^{2+} not typical
- US: Variable amounts of internal vascularity; not compressible
- MR: Moderately to markedly hyperintense (T2 FS/STIR) to skeletal muscle, ± heterogeneity (necrosis & hemorrhage)
- MR: Variable enhancement, ranging from minimal to diffuse, heterogeneous to homogeneous
- US & MR features can strongly suggest sarcoma
- MR better at defining deep extent & relationship to critical structures (e.g., neurovascular bundle, joints, etc.)
- PET best for staging

CLINICAL ISSUES

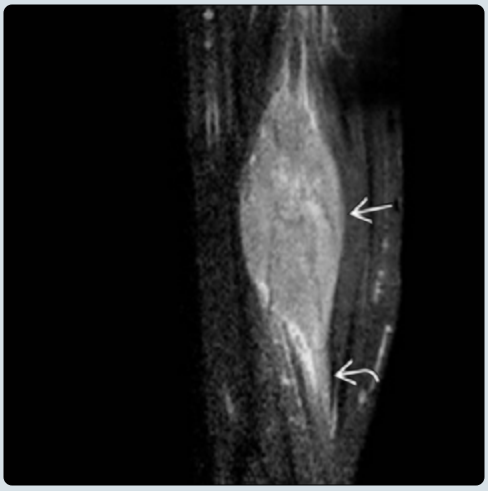
- Most common soft tissue sarcoma in children
- Typically presents as enlarging, firm, painless mass
- 2 age peaks: 2-6 years, 14-18 years
- Prognosis worse with alveolar subtype, tumor > 5 cm, metastatic disease at presentation (lung, bone marrow, lymph nodes most common)

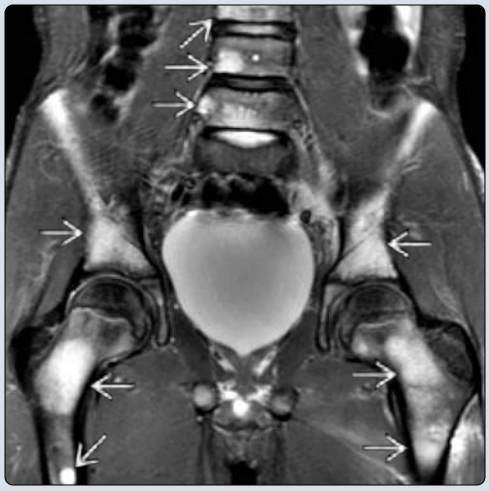
(Left) *Axial graphic shows a round, heterogeneous soft tissue mass anterior to the left hip with foci of hemorrhage (red). Note the multifocal osseous metastatic disease (brown).* **(Right)** *Color Doppler US in a 6 month old with a firm hand mass shows a nonspecific, mildly heterogeneous palmar soft tissue lesion. Features that lead away from considering an infantile hemangioma include the lack of significant internal vascularity, the presence of deep extension, & the report of firmness. Biopsy confirmed a RMS.*





(Left) *Sagittal STIR MR shows a well-circumscribed, intermediate signal intensity soft tissue mass in the posterior medial right forearm. Note the tail of tumor extending distally & the lack of significant surrounding edema. This mass was a biopsy-proven alveolar RMS.* **(Right)** *Coronal STIR MR in a 10 year old with an intramuscular gluteal mass (not shown) demonstrates numerous hyperintense osseous metastatic foci within the lower lumbar spine, pelvis, & femurs. Biopsy of the mass showed alveolar RMS.*





TERMINOLOGY

Definitions

- Mesenchymal sarcoma arising from rhabdomyoblasts (primitive muscle cells); lacks normal differentiation into skeletal muscle
  - Embryonal rhabdomyosarcoma (RMS) (60-70% of childhood RMS)
    - Resembles skeletal muscles in 6- to 8-week fetus
    - Most common type in patients < 15 years old
    - Most commonly genitourinary, head & neck, retroperitoneum
    - Botryoid subtype (sarcoma botryoides) (10% of RMS)
      - Grape-like polyploid masses or clusters protruding into lumen of vagina, bladder, biliary tree, & nasopharynx
  - Alveolar RMS (20% of RMS)
    - Resembles skeletal muscle in 10-week fetus
    - Average age: 15 years
    - Most commonly extremity, trunk, & perianal/perirectal
  - Undifferentiated RMS
    - 30-50 year olds; rarely in children

IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed, solid, mildly heterogeneous intramuscular mass without significant surrounding soft tissue edema
- Location
  - Head & neck (28-40%)
    - Nasopharynx, sinuses, orbit, soft tissues (7%)
  - Genitourinary (20%)
    - Prostate, bladder, vagina, paratesticular
  - Extremities (15-20%)
    - Typically intramuscular
  - Truncal/retroperitoneal (11-17%)
- Size
  - Quite variable
- Morphology
  - Usually round with well-circumscribed, lobular margins
  - May have tail of tumor extending proximal/distal

Radiographic Findings

- Soft tissue fullness similar to muscle on radiographs
- Rarely with adjacent bone involvement
- Ca^{2+} not typical

CT Findings

- Not primarily used for mass investigation
- Lung CT for metastatic work-up

Ultrasonographic Findings

- Solid, mildly heterogeneous soft tissue mass with variable amounts of internal vascularity
- Often round & well-circumscribed, though deeper infiltration possible

MR Findings

- T1WI
  - Similar signal to skeletal muscle
- T2WI FS/STIR
  - Moderately to markedly hyperintense to skeletal muscle
    - Not typically cystic
  - Can be heterogeneous with necrosis & hemorrhage
  - Typically minimal (if any) surrounding edema
- T1WI C+ FS
  - Variable enhancement, ranging from minimal to diffuse, heterogeneous to homogeneous
- DWI
  - Typically restricts diffusion; can ↑ conspicuity of primary & metastatic lesions
- MRA/MRV
  - May be helpful to determine relationship of mass to neurovascular bundle

Nuclear Medicine Findings

- PET
  - Better than conventional imaging in staging disease (except for very small lung lesions)
  - Intense FDG uptake by soft tissue tumor
- Bone scan traditionally performed for skeletal metastases

Imaging Recommendations

- Best imaging tool
  - US & MR features can strongly suggest sarcoma
  - MR better at defining deep extent & relationship to critical structures (e.g., neurovascular bundle, joints, etc.)
- Protocol advice
  - Consider subtraction MR images of pre- from postcontrast T1 FS
    - Avoids pseudoenhancement postcontrast due to preexisting hemorrhage, protein, etc.

DIFFERENTIAL DIAGNOSIS

Other Soft Tissue Sarcomas

- Synovial sarcoma
  - Ca^{2+} in 1/3, may be small & cystic-appearing
  - Usually not intraarticular but near joint; often abuts bone
- Extraosseous Ewing sarcoma
  - Similar in appearance to RMS, usually 2nd-3rd decades
- Fibrosarcoma
  - Typically infantile in children
  - May be highly cystic or solid & highly vascular
  - Frequently infiltrative

Neurofibroma

- Most common in neurofibromatosis type 1 (NF1)
- Numerous well-defined lobules of plexiform neurofibroma
- Classic target appearance on T2/STIR MR
- Loss of target appearance & disproportionate growth suggest malignant degeneration

Venous Malformation

- Patchy enhancement, fluid-fluid levels, ± phleboliths

Infantile Hemangioma

- Solid but soft mass of infants with mild lobulations, variable echogenicity
- Highly vascular on color Doppler US with > 5 vessels per cm^2
  - Low-resistance arterial waveforms during proliferation

Myositis Ossificans

- Heterogeneous intramuscular mass in older children with prior acute trauma (2/3) or microtrauma history
- Marked inflammatory reaction of surrounding muscle
- Calcifies peripheral to central over weeks/months

Abscess

- Heterogeneous collection with thick, irregular wall & septations, swirling internal debris with compression
- Peripheral hyperemia/enhancement
- Moderate to marked surrounding edema

PATHOLOGY

General Features

- Genetics
  - Embryonal: Loss of chromosome 11 genomic material
  - Alveolar: Translocation of chromosomes 1 or 2 & 13
    - Translocation t(2;13) (q35;q14), results in *PAX3-FKHR* fusion gene (55%)
    - Translocation t(1;13) (p36;q14), results in *PAX7-FKHR* fusion gene (22%)
    - Better prognosis in *PAX7-FKHR* than *PAX3-FKHR* (more invasive)
- Associated abnormalities
  - NF1, Li-Fraumeni syndrome, Costello syndrome, Beckwith-Wiedemann

Staging, Grading, & Classification

- TNM
  - Tx: Primary tumor cannot be assessed
  - T0: No evidence of primary tumor
  - T1: Tumor ≤ 5 cm in greatest dimension
    - T1a: Superficial tumor
    - T1b: Deep tumor
  - T2: Tumor > 5 cm in dimension
    - T2a: Superficial tumor
    - T2b: Deep tumor
  - Nx: Regional tumor cannot be assessed
  - N0: No regional lymph node metastasis
  - N1: Regional lymph node metastasis
  - M0: No distant metastases
  - M1: Distant metastases

Microscopic Features

- Embryonal RMS: Primitive with dense spindle-shaped cells with hyperchromatic nuclei & cytoplasmic processes, strap cell appearance
- Alveolar RMS: Large oval cells separated into nests of "alveoli" separated by fibrous septa
- Central cells necrotic & loosely arranged with peripheral cells arranged into well-organized picket fence appearance
- Immunohistochemical staining: MyoD1, myoglobin, myogenin, actin & desmin

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Enlarging, firm soft tissue mass
- Other signs/symptoms
  - < 1/2 experience pain; symptoms depend on location

Demographics

- Age
  - 65% present < 6 years old
  - 2 age peaks: 2-6 & 14-18 years of age
    - Embryonal: < 15 years old; head & neck, genitourinary
      - 46% occur in patients < 5 years old
    - Alveolar: Typically adolescents & young adults (extremity, paratesticular, & truncal)
      - Occurs at all ages
- Gender
  - Overall M > F (1.5:1), extremity: M < F (0.8:1), genitourinary: M > F (3:1)
- Epidemiology
  - 5-10% of childhood malignant solid tumors overall
    - 4-6 cases/1 million per year
  - Most common soft tissue sarcoma in children

Natural History & Prognosis

- Prognosis depends on
  - Site of tumor
    - Orbit & nonparameningeal more favorable
    - Extremity: Poorer prognosis, ↑ incidence of alveolar RMS, often lymph node (+), ↑ metastases at presentation
    - Chest poor prognosis: 45% 5-year survival, tends to be alveolar RMS, ↑ metastases at presentation, location difficult for resection & radiation
  - Size: < 5 cm more favorable
  - Age: Alveolar ↓ prognosis < 1 year old & > 10 years old
  - Histiologic type: Embryonal better than alveolar RMS
  - DNA component: Hyperdiploid (embryonal RMS) better than diploid or tetraploid (alveolar RMS)
  - Tumor expression of p-glycoprotein gene: More likely multidrug resistant
  - Lymph node involvement
  - Metastatic disease at presentation has much poorer prognosis: Lung, bone marrow, lymph nodes most common

Treatment

- Neoadjuvant & adjuvant chemotherapy, surgery, radiation

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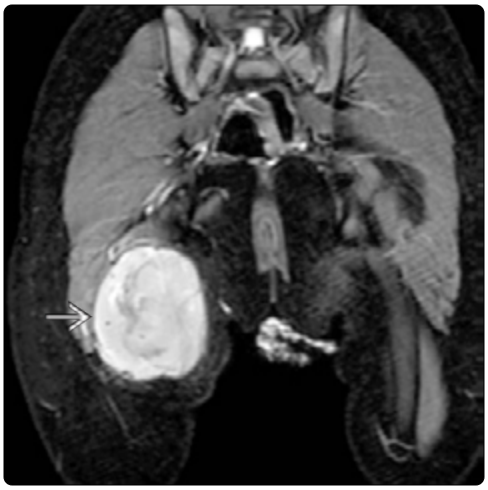
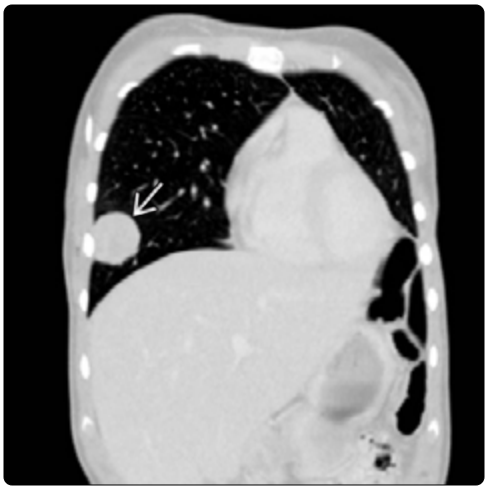
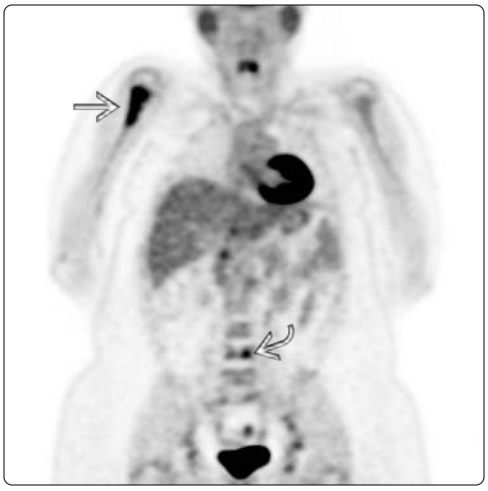
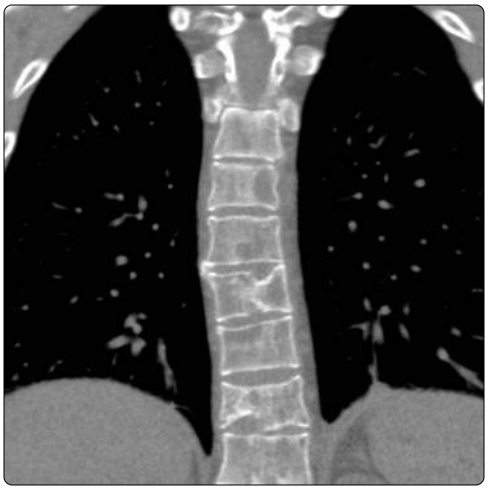


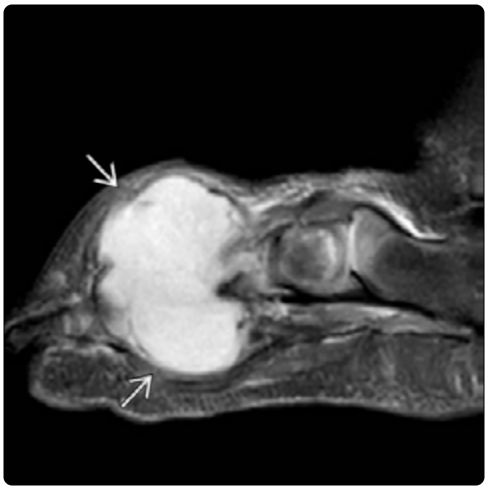


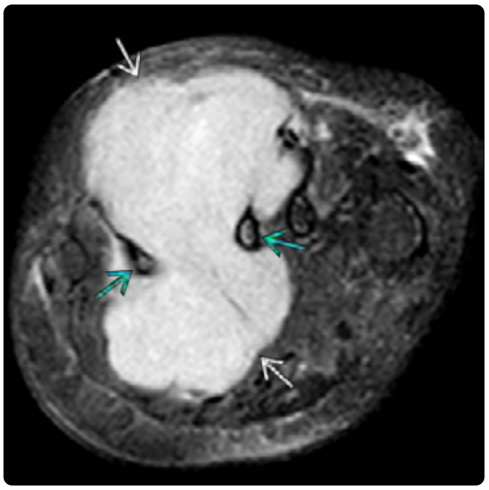
(Left) *Coronal STIR MR in a 21 month old who presented with a posterior right thigh mass shows a moderately hyperintense (but not fluid intensity), well-circumscribed mass without surrounding edema. The appearance is concerning for a sarcoma, & biopsy showed an embryonal RMS.* **(Right)** *Coronal CECT in a patient with a known relapsed alveolar RMS (first diagnosed 5 years previously) shows a large right lower lobe metastatic lesion. Multiple metastatic foci were also present in the left lower lobe (not included).*





(Left) *Coronal PET in a 24 year old with a relapsed alveolar RMS shows FDG uptake in a right proximal humeral metastasis with a maximum SUV of 8.5. A lumbar spine metastasis was among many others noted.* **(Right)** *Coronal reformatted image from a NECT of the chest in the same patient 1 year prior shows diffuse lytic thoracic spine metastases with mild pathologic compression deformities at a few levels.*





(Left) *Sagittal T2 FS MR in an 8 month old shows a large, round, well-circumscribed, moderately hyperintense (but not fluid intensity) mass between the metatarsals.* **(Right)** *Axial T2 FS MR in the same patient shows the dumbbell-shaped mass splaying & partially encasing the 3rd & 4th metatarsals. Note the lack of surrounding edema. RMS was confirmed at biopsy.*

KEY FACTS

TERMINOLOGY

- Heterogeneous group of malignant mesenchymal tumors
- Most common pediatric soft tissue sarcomas: Rhabdomyosarcoma (~ 19-50%), extraskeletal Ewing sarcoma/PNET, synovial sarcoma

IMAGING

- Best clue: Well-circumscribed round or ovoid predominantly solid (firm) soft tissue mass without surrounding edema
  - Overlaps many benign pediatric soft tissue masses
- Variable internal heterogeneity
  - Solid tumor components show variable enhancement
  - Intermixed nonenhancing foci: Hemorrhage, necrosis, Ca^{2+}
    - Some sarcomas may appear partially or entirely cystic until contrast administered
- MR with IV contrast: Best study to characterize mass, determine local extent, & follow local therapy response

TOP DIFFERENTIAL DIAGNOSES

- Vascular anomalies
- Periarticular cysts or cyst-like lesions
- Myositis ossificans
- Benign fibrous or neurogenic tumors
- Abscess

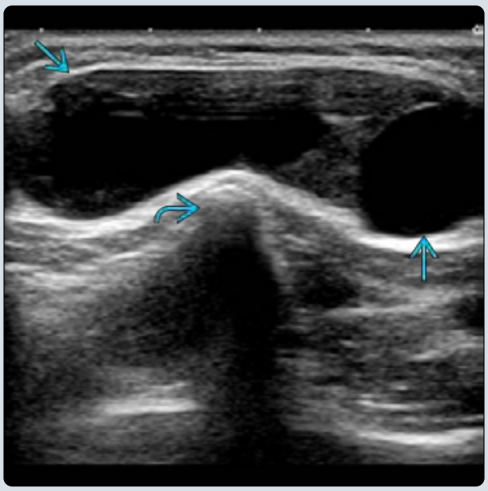
CLINICAL ISSUES

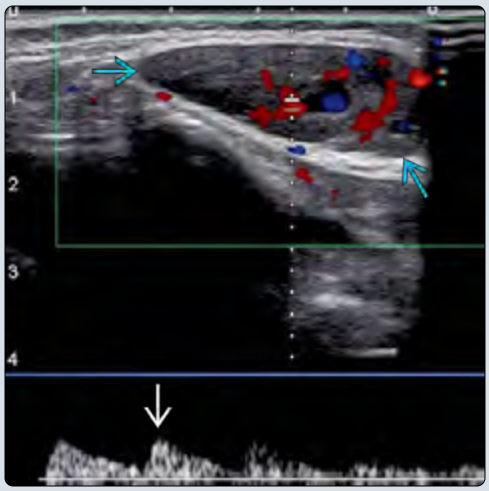
- Most common signs/symptoms: Palpable firm mass

DIAGNOSTIC CHECKLIST

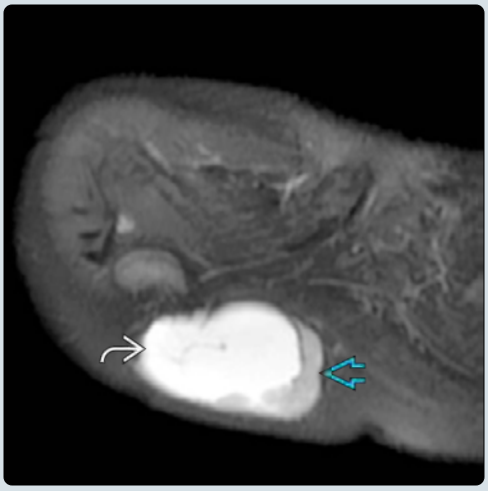
- Benign & malignant soft tissue masses overlap in appearances
- Without specific clinical & imaging features of benign process → biopsy required
- If specific features strongly suggest benign self-limited or medically treatable process → close clinical & imaging follow-up
- Smaller focal lesions frequently excised regardless

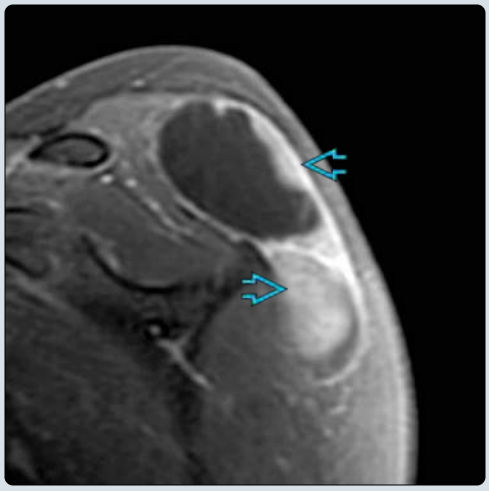
(Left) *Longitudinal ultrasound though a firm mass in the posterior right shoulder of a 5 year old shows a mixed cystic & solid lesion overlying the scapula.* **(Right)** *Transverse pulsed Doppler ultrasound in the same patient shows a moderate amount of internal vascularity throughout the solid portions of the lesion, including several arterial waveforms. Spectral tracings (at several locations) are critical for confirming real flow within a lesion.*





(Left) *Axial T2 FS MR in the same patient shows nodular intermediate signal intensity components along the wall of the mass. Some of the cystic portions show thin internal septations.* **(Right)** *Sagittal T1 C+ FS MR in the same patient shows enhancement of the solid components of the mass. While some of the cystic features could be seen in a lymphatic malformation, the enhancing solid components with internal vascularity are concerning for malignancy. A soft tissue Ewing sarcoma was confirmed with biopsy.*





TERMINOLOGY

Definitions

- Heterogeneous group of malignant mesenchymal tumors
  - Most common pediatric soft tissue sarcoma: Rhabdomyosarcoma (~ 19-50%)
  - Extraskeletal Ewing sarcoma/primitive neuroectodermal tumor & synovial sarcoma next most common

IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed round or ovoid predominantly solid (firm) soft tissue mass without surrounding edema
    - Overlaps many benign pediatric soft tissue masses

MR Findings

- T1: Typically similar to muscle signal intensity ± bright foci of hemorrhage
- T2 FS/STIR: Relatively uniform intermediate to bright signal intensity of solid tissue
  - ± cystic/necrotic foci of very bright signal; purely cystic appearance uncommon (but still requires contrast)
  - Typically lacks significant surrounding muscle edema
- T1 C+ FS: Wide range of enhancement patterns
  - Often intermediate to low-level enhancement
    - ± nonenhancing foci of necrosis or hemorrhage
- DWI: Highly cellular tumors show restricted diffusion

Ultrasonographic Findings

- Grayscale ultrasound
  - Firm mass of variable internal echogenicity
    - Typically of intermediate or low-level echoes
  - Rarely anechoic with posterior acoustic enhancement & compressibility → favors benign fluid-filled process
    - Must look closely for solid nodular components
- Color Doppler
  - Variable internal vascularity
    - Frequently only mild to intermediate
    - Check for spectral waveforms to confirm true flow

Imaging Recommendations

- Best imaging tool
  - MR ± IV contrast: Best study to characterize mass, determine local extent, & follow local response during/after therapy
- Protocol advice
  - MR technique
    - T1 sequence in at least 1 plane
      - Axial images show relationship of mass to neurovascular bundle (normally encased by fat)
      - Additional FS T1 sequence may help evaluate etiologies of bright signal in mass & determine true enhancement after contrast administration
    - Multiplanar fluid-sensitive T2 FS or STIR sequences to highlight most pathologies
    - Contrast critical for showing solid vs. necrotic foci
      - Subtraction of precontrast images most accurate

DIFFERENTIAL DIAGNOSIS

Vascular Anomalies

- Infantile hemangioma
- Venous malformation
- Lymphatic malformation

Benign Fibrous/Fibrohistiocytic Tumors

- Fibrous hamartoma of infancy
- Myofibroma/myofibromatosis
- Nodular fasciitis
- Fibromatosis

Neurogenic Tumors

- Plexiform, localized, or diffuse neurofibroma
- Schwannoma

Fat-Containing Neoplasms

- Lipoma
- Lipoblastoma
- Hibernoma

Periarticular Cysts

- Ganglion, synovial, or parameniscal cysts

Infectious/Inflammatory Masses

- Granuloma annulare
- Abscess

Posttraumatic Lesions

- Myositis ossificans
- Fat trauma/necrosis
- Hematoma

CLINICAL ISSUES

Presentation

- Most common signs/symptoms: Palpable firm mass

Natural History & Prognosis

- Factors relevant to prognosis: Tumor, node, metastasis staging & tumor size, site, invasiveness/resectability, histologic grade, patient age

Treatment

- Combination of surgery, chemotherapy, &/or radiation
  - Use & order depends on many factors

DIAGNOSTIC CHECKLIST

Consider

- Benign & malignant soft tissue masses overlap in appearances; biopsy often required
- Correlation with clinical history important
  - Inflammatory & posttraumatic etiologies should be considered when pain & surrounding edema present
  - Firm painless or mildly tender mass more concerning
  - Indolent growth does not exclude malignancy

Image Interpretation Pearls

- Soft tissue sarcomas often show well-defined margins
- Significant surrounding edema favors (but does not confirm) infectious/inflammatory or posttraumatic etiology; consider close follow-up before biopsy

Pediatric Soft Tissue Malignancies

Tumor	Characteristic Imaging Features	Clinical Features	Prognosis
Rhabdomyosarcoma	Typical soft tissue sarcoma without specific features: Well-circumscribed round/ovoid solid mass of intermediate/high T2 MR signal intensity & variable enhancement	Peak incidence overall: 2-6 years old; head/neck & GU sites more common than extremities; metastases in 15-20% at presentation	5-year survival: 75%
Synovial sarcoma	30% show Ca^{2+}; often cystic-appearing; enhances with IV contrast; T2 MR triple sign (of intensities) not specific; contiguity with bone more common than remodeling or invasion	30% < 20 years old; lower extremity most common; often near joint (rarely in joint); slow growth common	5-year survival: 27-61%; late recurrence common
Extraskeletal Ewing sarcoma/PNET	No specific features overall; aggressive chest wall mass (Askin tumor) with pleural effusion & rib destruction very suggestive	10-30 years old; most common in Caucasians; extraskeletal less common than skeletal	5-year survival: 56-70%
Infantile fibrosarcoma	Frequently infiltrative; often contains prominent cystic spaces; may be highly vascular	0-2 years old; 30-80% detected by/at birth; rapidly growing; skin involvement mimics vascular lesion; distal extremities most common	5-year survival: 80% (better than adolescent/adult fibrosarcoma)
Malignant rhabdoid tumor	No specific features overall	< 1 year old; ± concomitant CNS or renal rhabdoid tumors	5-year survival: < 50%
Malignant peripheral nerve sheath tumor	Enlarging mass (> 5 cm) amidst stable PNs; ↑ activity on PET helpful to distinguish from PN	~ 50% in neurofibromatosis type I (8-13% lifetime risk of malignant peripheral nerve sheath tumor)	5-year survival: 23-51%
Dermatofibrosarcoma protuberans	Nodular superficial mass with cutaneous elongation ± satellite nodules	Slow-growing discolored skin mass	5-year survival: > 99%
Granulocytic/myeloid sarcoma (chloroma)	Background of diffuse marrow abnormalities	Acute myelogenous leukemia	5-year survival: 20-25%
Neuroblastoma	Typically small cutaneous & subcutaneous masses; ± discrete paraspinal primary mass; ± extensive marrow abnormalities	Wide range of clinical presentations; soft tissue metastases most common < 18 months of age	Excellent in infant with metastases limited to liver, skin, & bone marrow; others variable
Alveolar soft part sarcoma	May be brighter than muscle on T1 MR; often shows prominent peripheral vascularity with vigorous enhancement	Head/neck most common in children	5-year survival: 56-70%
Epithelioid sarcoma	Ca^{2+} in 20-30%; may have moderate surrounding edema with extension along fascia & tendon sheaths (uncommon for other sarcomas)	75% between 10-39 years old Distal upper extremity most common; slow growth frequent; proximal type more aggressive	5-year survival: Up to 80%
Angiosarcoma	Evidence of prior hemorrhage; tangles of vessels uncommon in superficial lesions	Typically de novo; rarely arise from preexisting vascular malformation	5-year survival: As low as 15%
Liposarcoma	Fat-poor myxoid subtype in 2nd decade of life	Extremely uncommon < 8-10 years old	Better than adult types

PN = plexiform neurofibromas.
Other less common pediatric soft tissue sarcomas: Leiomyosarcoma, low-grade fibromyxoid sarcoma, clear cell sarcoma, malignant fibrous histiocytoma, hemangiopericytoma.

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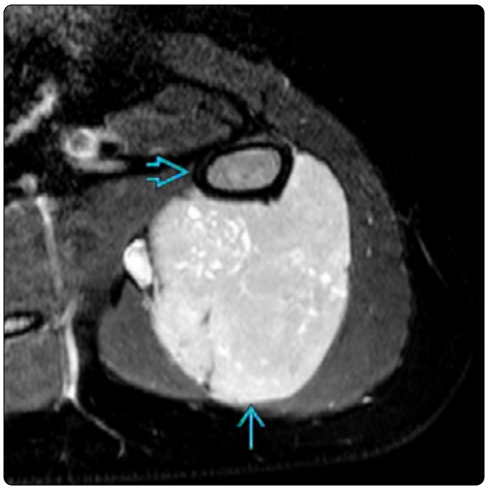
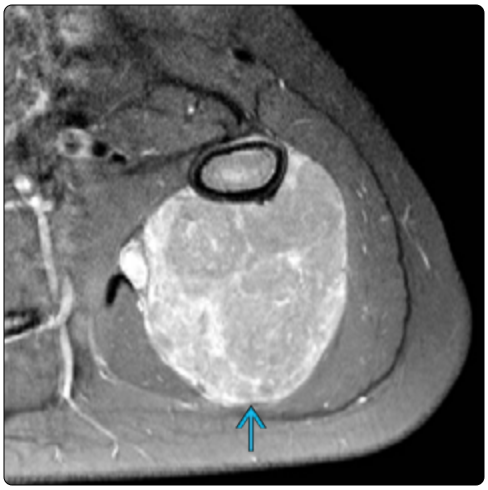
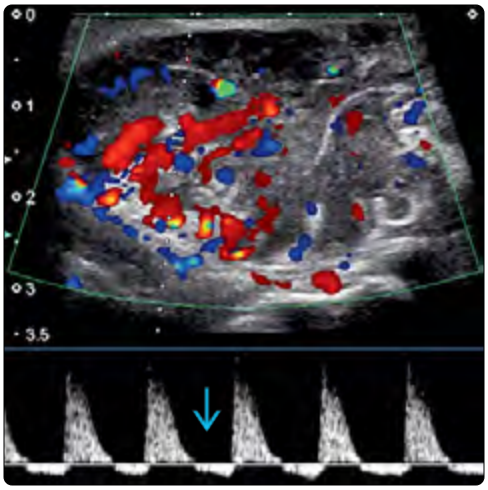
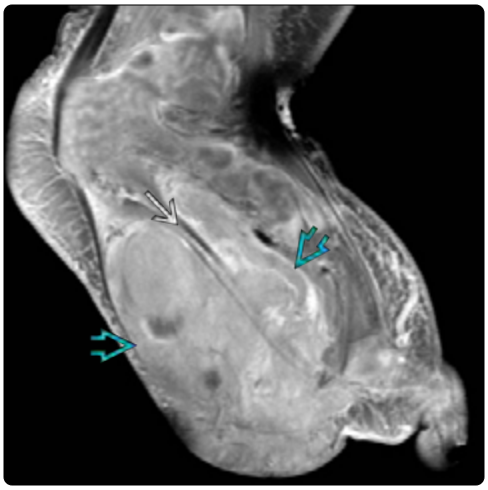


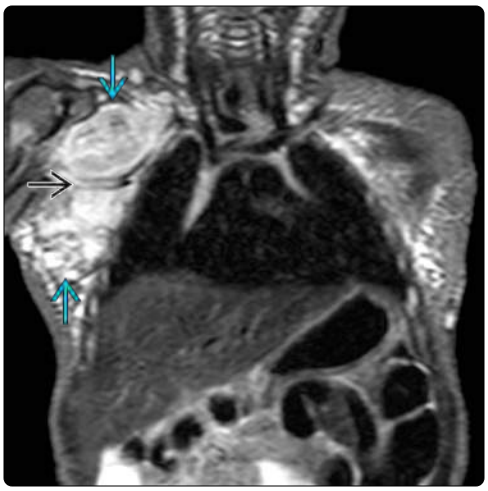


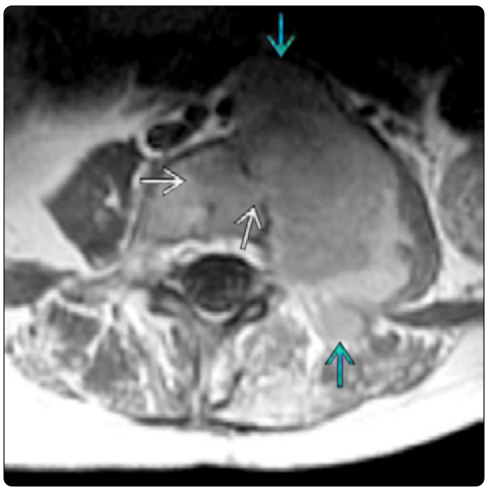
(Left) *Axial T2 FS MR in a 4 year old with a firm mass shows a well-circumscribed, round, mildly heterogeneous intermediate to bright intramuscular mass directly abutting the posterior left humerus. Note the lack of surrounding edema.* **(Right)** *Axial T1 C+ FS MR in the same patient shows moderate heterogeneous enhancement throughout the mass. The overall appearance as a "solid soft tissue ball" is highly suggestive of a soft tissue malignancy. Biopsy confirmed a synovial sarcoma.*





(Left) *Transverse color Doppler ultrasound through a growing plantar soft tissue mass of the foot in a young infant shows a heterogeneous & highly vascularized lesion. The high resistance waveforms (with reversed diastolic flow) & deep extent would be atypical for an infantile hemangioma.* **(Right)** *Sagittal T1 C+ FS MR in the same patient shows heterogeneous enhancement of the deep & infiltrative mass that encases the flexor tendons. An infantile fibrosarcoma was found upon biopsy.*





(Left) *Coronal STIR MR in a 6 month old shows a heterogeneous soft tissue lesion of the chest wall encasing the neurovascular bundle. A malignant rhabdoid tumor was found at surgery. No renal or CNS rhabdoid lesions were identified.* **(Right)** *Axial T1 C+ MR in a neurofibromatosis type I patient shows interval growth of a paraspinal soft tissue mass with invasion of the adjacent lumbar vertebral body. A malignant peripheral nerve sheath tumor was confirmed at biopsy.*

Granuloma Annulare

KEY FACTS

TERMINOLOGY

- Benign, inflammatory lesion of superficial soft tissues
- Cutaneous form very familiar to dermatologists
- Subcutaneous form most commonly encountered by radiologists due to
  - Deeper location
  - Nonspecific clinical appearance

IMAGING

- Location: Subcutaneous lesion(s) of extensor surfaces of lower leg (pretibial), foot, & forearm/elbow or scalp
- Morphology: Elongated, nodular, & poorly defined
- Radiography: Swelling without Ca^{2+} or bone involvement
- Ultrasound: Hypoechoic, mildly heterogeneous, infiltrating superficial lesion
- T2 FS MR: Heterogeneous with regions of ↓ & ↑ signal
  - Indistinct margins with surrounding edema common
- T1 C+ FS MR: Homogeneous to mildly heterogeneous

TOP DIFFERENTIAL DIAGNOSES

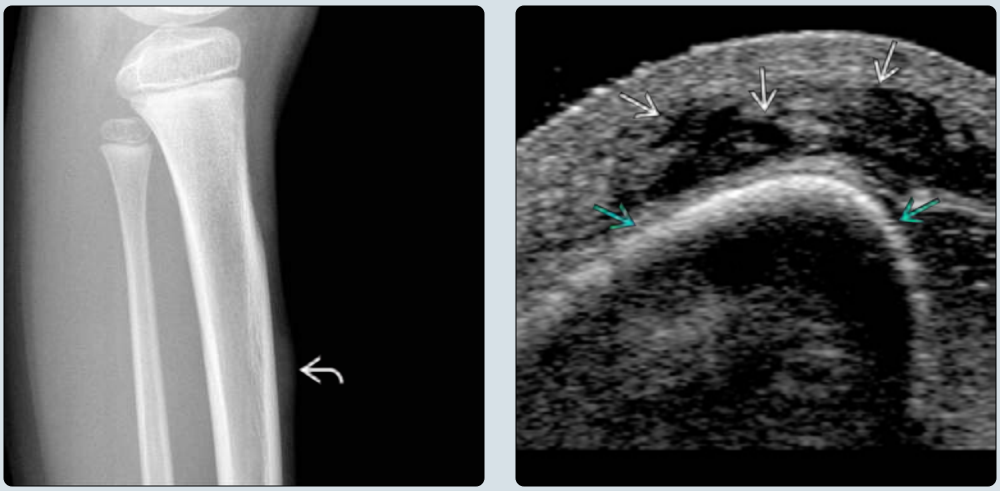
- Trauma (contusion or fat necrosis)
- Chronic foreign body
- Microcystic lymphatic malformation
- Cellulitis
- Fibromatosis

PATHOLOGY

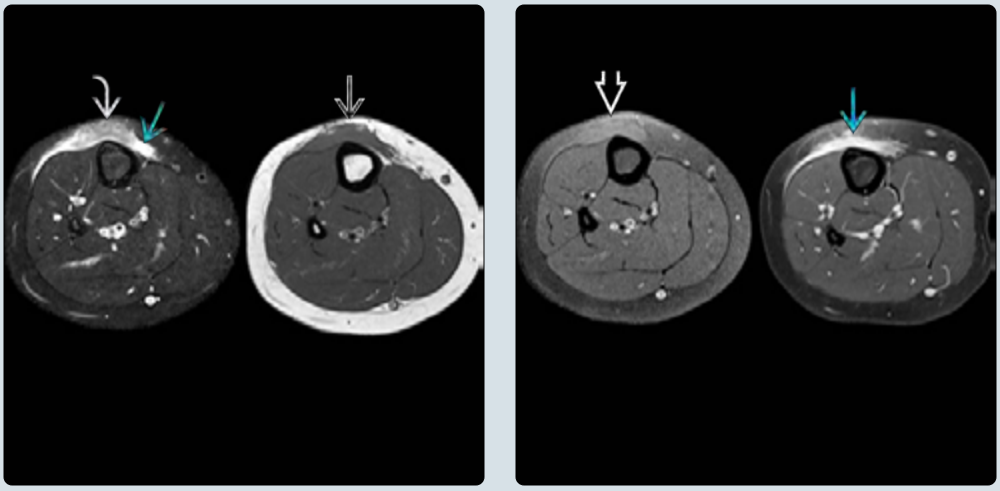
- Fine-needle aspiration not adequate for diagnosis
  - May lead to mistaken diagnosis of malignancy
- Mucin staining characteristic

CLINICAL ISSUES

- Typical presentation: Firm, painless, subcutaneous nodule in otherwise healthy young child
- Natural history: Spontaneous involution
- Lesions with characteristic history, location, & imaging: Consider observation vs. incisional biopsy for diagnosis
- Recurrence can occur after incisional or excisional biopsy



(Left) *Lateral radiograph of a 6-year-old girl presenting with a painless, firm bump at the anterior tibia shows a soft tissue mass without Ca^{2+} or underlying bony abnormality. A similar but smaller mass was present on the contralateral side (not shown).* **(Right)** *Transverse ultrasound in the same patient shows a hypoechoic, mildly heterogeneous, elongated, & lobulated mass with indistinct margins anterior to the tibial diaphysis. The mass proved to be subcutaneous granuloma annulare on biopsy.*



(Left) *Axial T2 FS MR (L) in a 4-year-old girl with subcutaneous granuloma annulare shows a heterogeneous, elongated pretibial mass with foci of intermediate & high signal intensity tracking over the superficial fascia. Axial T1 MR (R) shows the mass to be isointense to muscle.* **(Right)** *Axial T1 FS MR (L) in the same patient shows the lesion to be isointense to muscle with homogeneous enhancement on the axial T1 FS C+ MR image (R).*

TERMINOLOGY

Abbreviations

- Subcutaneous granuloma annulare (SGA)

Synonyms

- Benign rheumatoid nodule, pseudorheumatoid nodule, deep granuloma annulare, subcutaneous palisading granuloma, palisading granuloma nodosum, isolated subcutaneous nodule, isolated subcutaneous granuloma, necrobiotic granuloma

Definitions

- Benign inflammatory skin lesion of unknown etiology
- 4 clinically distinct subtypes
  - Localized
  - Generalized
  - Perforating
  - Subcutaneous: Subtype of concern to radiologists

IMAGING

General Features

- Location
  - Extension up to (but not deep to) investing fascia
    - Abnormal muscle signal highly atypical
  - Distributed within feet, lower legs, fingers, hands, forearms, & scalp; can be multifocal & bilateral
    - Primarily extensor surfaces of extremities
    - Classic location: Pretibial (19-65%)
  - Penile SGA rare but reported

Radiographic Findings

- Superficial soft tissue mass
  - No Ca^{2+} or bone involvement

Ultrasonographic Findings

- Hypoechoic, mildly heterogeneous nodular lesion

MR Findings

- T1WI
  - Iso- to slightly hyperintense to skeletal muscle
- T2WI FS
  - Heterogeneous with areas of ↓ & ↑ signal intensity
  - Reticular ↑ signal in surrounding subcutaneous fat
- T1WI C+ FS
  - Homogeneous to mildly heterogeneous enhancement

DIFFERENTIAL DIAGNOSIS

Trauma (Contusion or Fat Necrosis)

- Focally edematous soft tissue after trauma, often overlying bony protuberance
- May ultimately lead to focal fat thinning

Chronic Foreign Body

- Hyperechoic foreign body surrounded by poorly defined hypoechoic tissue

Microcystic Lymphatic Malformation

- Longstanding, poorly defined, infiltrative lesion
- ± macrocysts (> 1 cm)
- Often has characteristic cutaneous blebs

Cellulitis

- Poorly defined, serpentine foci of fluid tracking through indurated fat
- Erythema & tenderness typical

Nodular Fasciitis

- Benign rapidly growing lesion, often painful
- Frequently at subcutaneous fat-fascial interface

Muscle Hernia

- Superficial fascial defect with dynamic muscle protrusion

Fibromatosis

- Typically deeper, infiltrative, benign, but locally aggressive, lesion
- Often has foci of ↓ & ↑ T2 signal intensity

PATHOLOGY

Microscopic Features

- Degenerating collagen with peripheral palisading histiocytes & surrounding reactive inflammatory cells
- Mucin staining characteristic
- Fine-needle aspiration suboptimal
  - May cause lesion to look aggressive like malignancy

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless, firm subcutaneous nodules in extremities or scalp

Demographics

- Age
  - Subcutaneous form almost exclusively seen in children
  - Mean age: 4.3 years

Natural History & Prognosis

- Spontaneous involution typical (over months to years)
- Recurrence in 19-75% of cases
  - May occur following incisional or excisional biopsy

Treatment

- Observation with typical clinical & imaging findings
- No proven efficacious medical treatment
- Incisional or excisional biopsy diagnostic but not necessarily curative

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Myositis Ossificans

KEY FACTS

TERMINOLOGY

- Myositis ossificans (MO): Benign reactive ossifying soft tissue mass; clear history of trauma in 60-75%

IMAGING

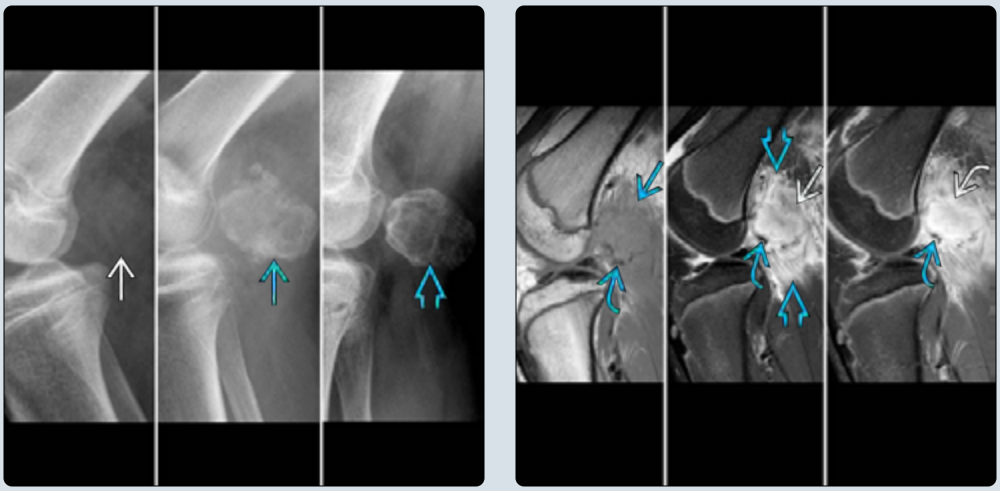
- Best diagnostic clue: Heterogeneous intramuscular mass occurring after trauma with evolving imaging appearance
- Radiographic findings
  - 0-2 weeks: Nonspecific soft tissue swelling/mass
  - 2-6 weeks: Faint but increasing peripheral Ca^{2+}
  - 6-8 weeks: Sharply circumscribed osseous mass
  - 5-6 months: Ossified mass with ↑ maturity, ↓ size
- CT: Rim of mineralization by 4 weeks; cortical & trabecular bone ± fatty marrow within months
- MR appearance also varies with age of lesion
  - Early: T2 heterogeneity internally with mild peripheral mineralization + marked surrounding edema
    - Active areas show intense enhancement
  - Late (mature): Well-circumscribed mass with rim of low signal cortex & little surrounding edema
    - Internal foci of fatty yellow marrow on all sequences
  - GRE: Sensitive for mineralization ("blooming" artifact)

TOP DIFFERENTIAL DIAGNOSES

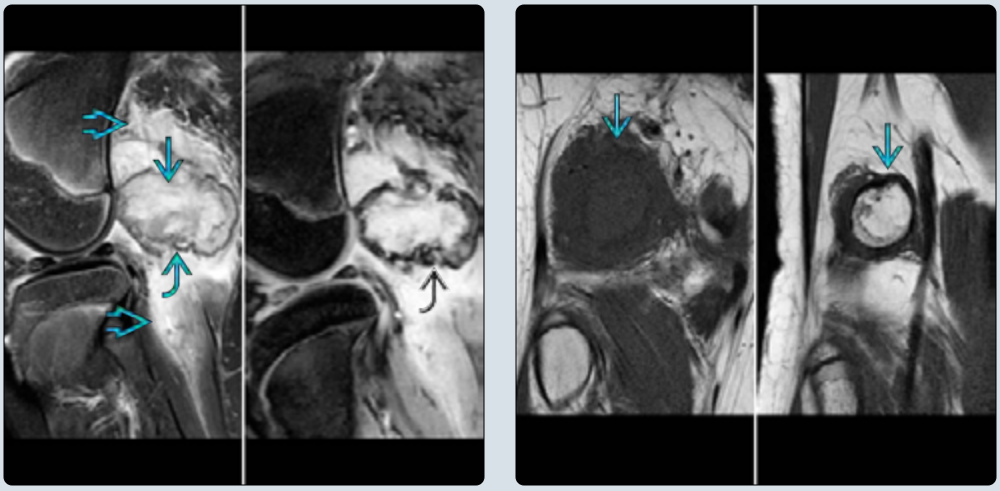
- Surface osteosarcoma
- Soft tissue sarcoma
- Hematoma
- Abscess

DIAGNOSTIC CHECKLIST

- Short-term radiographic follow-up or CT to demonstrate zonal ossification
- MR of early MO can mimic malignant soft tissue tumors
  - Marked surrounding edema is atypical in sarcomas
- Indeterminate lesions should undergo adequate biopsy
  - Pathologist to be aware if MO being considered as early MO may mimic malignancy under microscope



(Left) *Serial lateral radiographs in a 14-year-old dancer with a popliteal mass show an evolving lesion with only soft tissue edema present initially (left). A 6-week follow-up (middle) shows peripheral ossification with progressive maturation at 6 months (right).* **(Right)** *Initial sagittal MR in the same patient shows a mass which is isointense on PD (left), hyperintense on T2 FS (middle), & diffusely enhancing on T1 FS C+. Small peripheral hypointense foci represent early ossification. Adjacent edema is also present.*



(Left) *Six-week follow-up sagittal MR in the same patient shows heterogeneous internal signal in the mass on the T2 FS image (left) with a continuous peripheral hypointense rim & continued surrounding edema. Note the "blooming" artifact on the GRE sequence (right).* **(Right)** *Coronal T1 MR in the same patient at 6 weeks (left) & 1 year (right) of follow-up shows progressive maturation of the mass to an ossified lesion of fatty yellow marrow with no surrounding edema. The mass was then resected.*

TERMINOLOGY

Definitions

- Myositis ossificans (MO): Benign ossifying soft tissue mass; reactive lesion typically occurring after muscle insult
  - Clear history of trauma in 60-75% of cases
  - Others: Repetitive microtrauma, inflammation, ischemia

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous intramuscular mass with peripheral mineralization developing within weeks
    - Appearance changes over time as lesion matures

Radiographic Findings

- 0-2 weeks: Nonspecific soft tissue swelling/mass
- 2-6 weeks: Faint Ca^{2+}, largely peripheral
- 6-8 weeks: Sharply circumscribed peripheral Ca^{2+}
- 5-6 months: More mature ossified mass with ↓ size

Ultrasonographic Findings

- Early: Heterogeneous lesion with ↓ central & ↑ peripheral echogenic zones
- Late: Peripheral zone shows posterior shadowing

CT Findings

- NECT
  - Low attenuation mass without mineralization early
  - Rim of mineralization usually seen by 4 weeks
  - Gradual maturing ossification demonstrates cortical & trabecular bone ± fat attenuation of yellow marrow

MR Findings

- Early: Round, elongated, or poorly defined intramuscular mass + intense surrounding edema
  - T2WI: Heterogeneous signal intensity
  - T1WI: Isointense to skeletal muscle
  - ± small areas of ↓ peripheral signal (mineralization)
- Intermediate: ↓ peripheral signal more apparent
  - ± central areas of irregular low signal
  - ± fluid/fluid levels from hemorrhage
- Late (mature): Well-defined mass of variable heterogeneity with minimal if any surrounding edema
  - Foci isointense to fat (yellow marrow) on all sequences
  - Confluent peripheral rim of low signal intensity
- GRE: Most sensitive for mineralization
  - Exaggerated signal loss or "blooming" artifact
- Active lesions: Heterogeneous to intense enhancement after contrast administration

Imaging Recommendations

- Best imaging tool
  - Short interval follow-up radiographs in 3-4 weeks (when MO suspected): New peripheral mineralization
  - CT: Earliest detection of zonal pattern of mineralization

DIFFERENTIAL DIAGNOSIS

Surface Osteosarcoma

- Underlying bone shows cortical scalloping, periosteal reaction

Soft Tissue Sarcoma

- Typically well circumscribed without surrounding edema
- May contain Ca^{2+} (typically dystrophic, not ossification)

Hematoma

- Heterogeneous mass after trauma or with coagulopathy
- Subacute/chronic hematomas may develop Ca^{2+}

Venous Malformation

- Large phleboliths may occur in extensive lesions

Soft Tissue Abscess

- Clinical symptoms typically suggestive of infection
- Mass with thick, irregular enhancing wall & central nonenhancing fluid

PATHOLOGY

Microscopic Features

- Early lesions: Immature, highly cellular fibroblastic lesion
  - Differentiation from sarcoma may be difficult
- Late (mature) lesions: Distinct zonal pattern of ossification
  - Peripheral: Rim of mature lamellar bone

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Firm/hard soft tissue mass ± pain, tenderness
    - Clear history of traumatic event absent in up to 40%
    - ± history of repetitive microtrauma (athlete), inflammation, or ischemia
- Other signs/symptoms
  - Mechanical limitation of adjacent joint

Treatment

- Surgical excision of symptomatic lesions
  - Recurrence less likely if lesion fully mature

DIAGNOSTIC CHECKLIST

Consider

- Heterogeneous solid intramuscular mass in teenager/young adult with marked surrounding muscle edema: Strongly consider MO, especially after trauma
  - Recommend short-term follow-up radiographs or CT rather than immediate biopsy
- If biopsy required for indeterminate lesion: Obtain core, incisional, or excisional biopsy
  - Fine-needle aspiration may be inconclusive

Image Interpretation Pearls

- Earliest MR findings of MO: Mass with small peripheral areas of ↓ signal + surrounding edema

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Ewing Sarcoma

KEY FACTS

TERMINOLOGY

- Ewing sarcoma family of tumors: Ewing sarcoma, primitive neuroectodermal tumor, Askin tumor, extraosseous Ewing sarcoma
- Aggressive, small, round, blue cell tumor that typically arises in bone

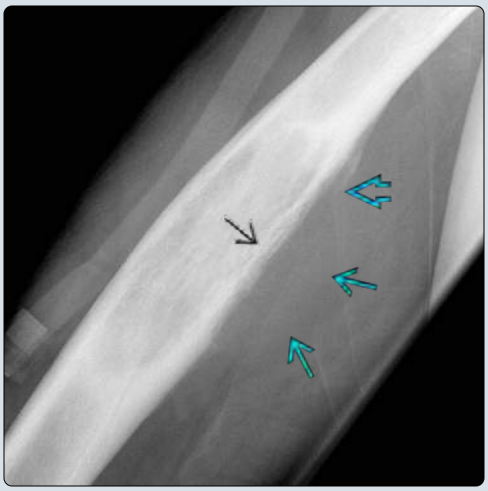
IMAGING

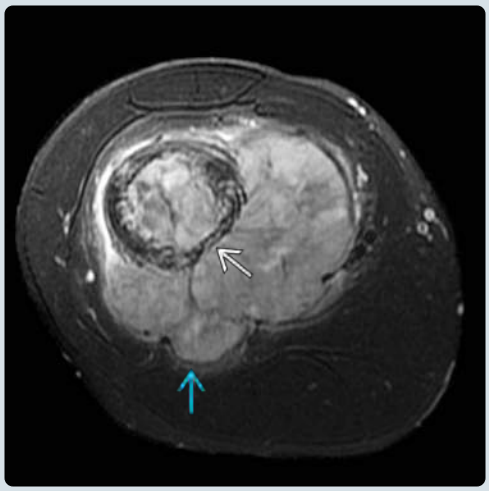
- Highly aggressive appearance
  - Lucent ill-defined intramedullary lesion, poorly marginated; can show mild expansile remodeling
    - Sclerosis in up to 25% of cases (not outside bone)
  - Permeative or moth-eaten cortical destruction
  - Aggressive periosteal reaction: Spiculated, sunburst, lamellated onion skin appearance, &/or Codman triangle
  - Associated soft tissue mass
    - Disproportionately larger than amount of bone destruction
- Greater propensity for flat bones (scapula, pelvis) than other primary bone malignancies
- Diaphyseal involvement more common than with other bone malignancies
- MR for local evaluation: Intraosseous & soft tissue extent, relationship to joint & neurovascular bundle
  - Coronal/sagittal T1 MR best shows true tumor margin in bone: Sharp demarcation vs. adjacent fatty marrow
  - Early joint-to joint marrow sequence (coronal T1 &/or STIR) to look for intraosseous skip metastases before smaller FOV high-detail assessment
- Chest CT, PET/CT for staging

CLINICAL ISSUES

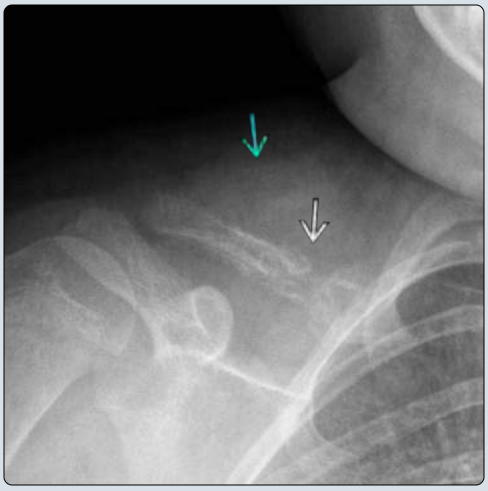
- 2nd most common primary bone malignancy in children after osteosarcoma
- Most common during 2nd decade of life

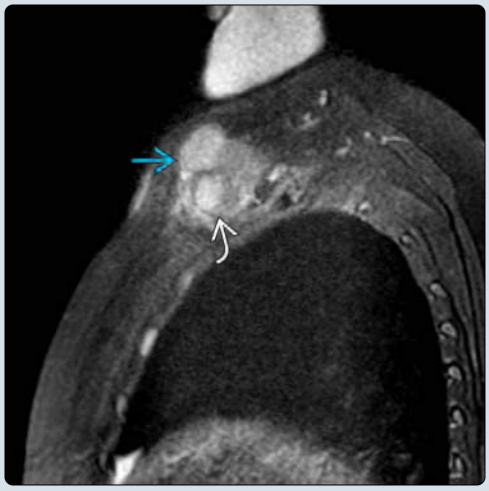
(Left) *Lateral radiograph in a 17 year old with pain shows mild expansile remodeling of the mid femoral diaphysis. There is an underlying lucent lesion with mixed margins. The overtly aggressive features include frank cortical permeation, elevated & interrupted periosteal reaction, & a soft tissue mass.* **(Right)** *Axial T2 FS MR in the same patient shows an intramedullary mass permeating the cortex nearly circumferentially with an overlying solid soft tissue mass. These features are classic for Ewing sarcoma.*





(Left) *AP radiograph in a 7 year old who presented with soft tissue swelling about the clavicle shows a destructive lesion with aggressive periosteal reaction & a soft tissue mass.* **(Right)** *Sagittal T2 FS MR in the same child shows a hyperintense lesion within the clavicle, permeating the cortex, & extending into an overlying soft tissue mass. These features are typical for Ewing sarcoma.*





TERMINOLOGY

Synonyms

- Ewing sarcoma (EWS), malignant primary bone tumor
- EWS family of tumors: Ewing, primitive neuroectodermal tumor (PNET), Askin tumor, extraosseous Ewing

Definitions

- Aggressive, small, round, blue cell tumor that typically arises in bone
- Closely related to PNET of bone

IMAGING

General Features

- Best diagnostic clue
  - Central, diaphyseal, permeative lytic lesion with lamellated onion skin periosteal reaction
- Location
  - Can occur in any bone & soft tissue
    - Lower extremity: 41%
    - Pelvis: 26%
      - Greater propensity for flat bones (scapula, pelvis) than other primary bone malignancies
    - Chest wall: 16%
    - Metaphyseal & diaphyseal: 94%
      - Diaphyseal involvement more common than with other bone malignancies
      - In diaphyseal location, involvement tends to be central; in metaphyseal location, involvement tends to be eccentric
    - Extraosseous: Trunk most common
    - Metastatic disease
      - Most commonly lungs, bones
- Size
  - > 5 cm
- Morphology
  - Bone lesions classically have large soft tissue mass relative to degree of bone change
  - Extraosseous EWS arises from soft tissues, not bone
    - Appears as nonspecific, well-circumscribed soft tissue mass

Radiographic Findings

- Radiography
  - Highly aggressive appearance overall
    - May sometimes show expansile remodeling but not without other more aggressive features
  - Ill-defined (wide zone of transition) intramedullary lesion
  - Mixed lytic & sclerotic
  - Permeative or moth-eaten appearance of cortical destruction
    - Infiltration of tumor through haversian canals of cortical bone
  - Cortical thickening, violation, or rarely saucerization
  - Aggressive periosteal reaction, often interrupted
    - Spiculated
    - Lamellated onion skin appearance
    - Sunburst or hair standing on end patterns: Periosteal reaction laid down along Sharpey fibers (which attach periosteum to underlying cortical bone) in attempt to wall off tumor
    - Codman triangle: Elevation of new bone along margin of periosteal reaction
  - No ossified tumor matrix (i.e., no soft tissue osteoid) but can be sclerotic in flat bones
    - Bone necrosis, reactive sclerosis
  - Associated soft tissue mass, often disproportionately larger than amount of bone destruction
  - ± pathologic fracture

CT Findings

- NECT
  - Depicts aggressive periosteal reaction & bone destruction
  - May be helpful in complex anatomic areas (pelvis, spine, skull base)
  - Chest CT for pulmonary metastasis

MR Findings

- T1WI
  - Coronal/sagittal best shows true tumor margin in bone: Sharp demarcation vs. adjacent normal fatty marrow
  - Axial may be helpful for relationship of tumor to neurovascular bundle
- T2 FS/STIR
  - Heterogeneous mass of intermediate to high signal intensity
  - Surrounding marrow, periosteal, & soft tissue edema
- DWI
  - Typically restrict diffusion in cellular areas
  - ADC value may help in monitoring response to therapy
- T1WI C+ FS
  - Heterogeneous enhancement
  - Baseline for postchemotherapy MR in determining response to therapy

Nuclear Medicine Findings

- Bone scan
  - Intense uptake
  - Traditionally used for evaluation of metastatic bone disease
- PET
  - Lesions demonstrate 18F-FDG avidity
  - Useful in staging & monitoring response to therapy
  - Possible predictor of outcome

Imaging Recommendations

- Best imaging tool
  - MR for local evaluation: Intraosseous & soft tissue extent, relationship to joint & neurovascular bundle
- Protocol advice
  - Early joint-to-joint marrow sequence (coronal T1 &/or STIR) to look for intraosseous skip metastases
  - Then smaller FOV pre- & postcontrast for high detail

DIFFERENTIAL DIAGNOSIS

Osteomyelitis

- May be difficult to differentiate from EWS, as both can have very aggressive imaging appearances
- Typically has less enhancing soft tissue & more nonenhancing fluid pockets & surrounding inflammation than EWS
- More common in children < 5 years of age
- More rapid presentation of symptoms

Osteosarcoma

- Osteoid tumor matrix in 90%
- More commonly involves long bone metaphysis

Metastatic Neuroblastoma

- More common in children < 3 years of age
- Multifocal permeative lucent lesions
- Calcified abdominopelvic mass

Langerhans Cell Histiocytosis

- Lytic bone lesion, often with sharp punched-out margins
- Homogeneously enhancing soft tissue mass in bone defect
- ± T2 MR hypointense rim, surrounding marrow edema > soft tissue edema

PATHOLOGY

General Features

- Genetics
  - EWS family of tumors: t(11;22)(q24;q12) (85%)

Gross Pathologic & Surgical Features

- Tan-gray tumors with areas of necrosis, hemorrhage

Microscopic Features

- Highly cellular, sheets of cells, little stroma
- Small, round, blue cell tumor

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Presents with pain & swelling
  - Tenderness/palpable mass
- Other signs/symptoms
  - May be associated with systemic symptoms/signs: Leukocytosis, fever, anemia, elevated sedimentation rate
    - Mimics osteomyelitis
  - Pathologic fracture (up to 15%)
    - Not considered adverse prognostic sign

Demographics

- Age
  - 2nd most common primary bone malignancy in children after osteosarcoma
  - Median age: 15 years
  - 80% are < 20 years old
  - Rare before 5 years of age
- Gender
  - M > F (1.5-2.0:1.0)
- Ethnicity
  - Caucasians 9x more common than African Americans

Natural History & Prognosis

- 5-year survival rate related to stage at diagnosis
  - 60-70% when disease localized
  - 20-30% 2-year survival with metastatic disease
- Poorer prognosis in
  - Boys
  - Patients 15-18 years & older
  - Metastatic disease
  - Larger tumor volume
  - Pelvic location
  - Higher serum lactate dehydrogenase levels prior to treatment
  - Previous treatment of malignancy: EWS as 2nd malignancy
- Survival better for lesions of extremities than those of axial skeleton
  - Pelvic lesions tend to be larger at presentation
- ↑ incidence of developing other future solid neoplasms
  - Treatment-related acute myeloid leukemia & myelodysplastic syndrome in 1-2% of patients
- Overall survival rate: 41%

Treatment

- Neoadjuvant & adjuvant chemotherapy
  - Vincristine, cyclophosphamide, doxorubicin, or actinomycin D
- Radiation therapy
- Resection of primary tumor
- Limb salvage procedures
- IGF-1R antibody
- Investigational: High-dose therapy followed by stem cell transplant for metastatic disease at presentation & patients with poor response to initial chemotherapy

DIAGNOSTIC CHECKLIST

Consider

- 18F-FDG PET in determining active residual/recurrent tumor from therapeutic changes

Image Interpretation Pearls

- May mimic osteomyelitis clinically or in laboratory findings

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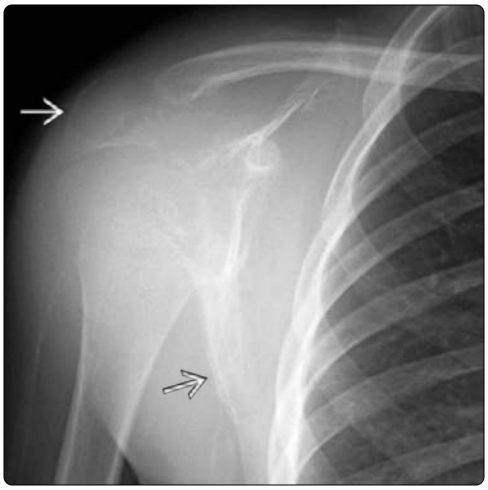
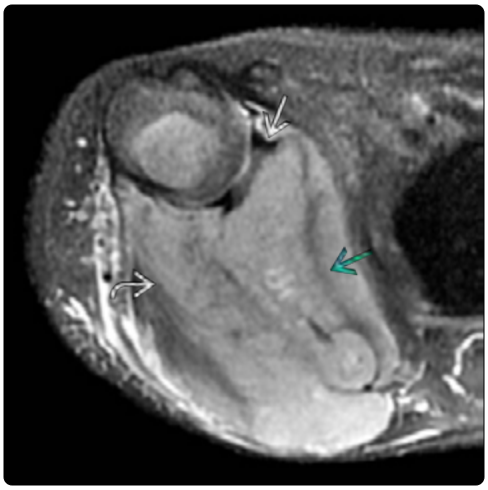
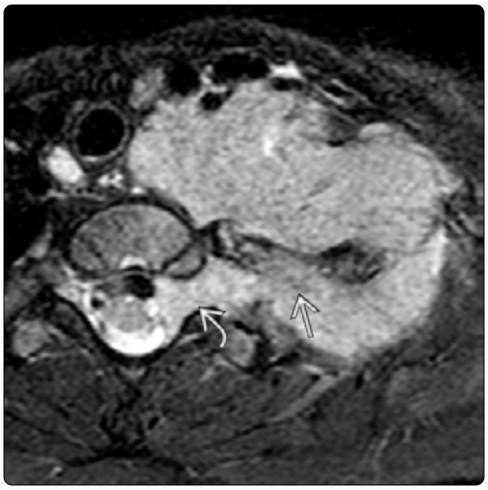
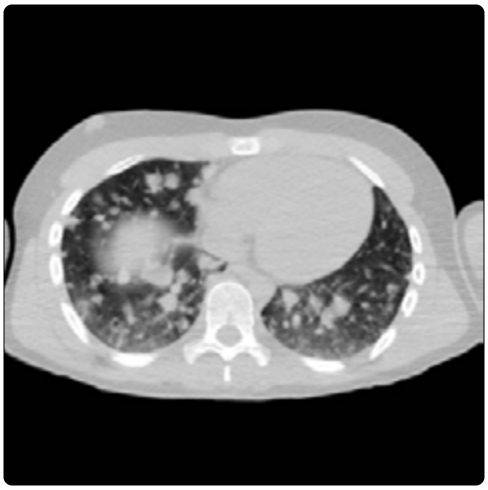


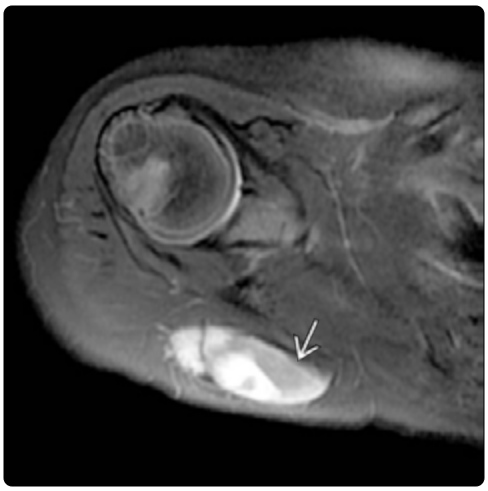


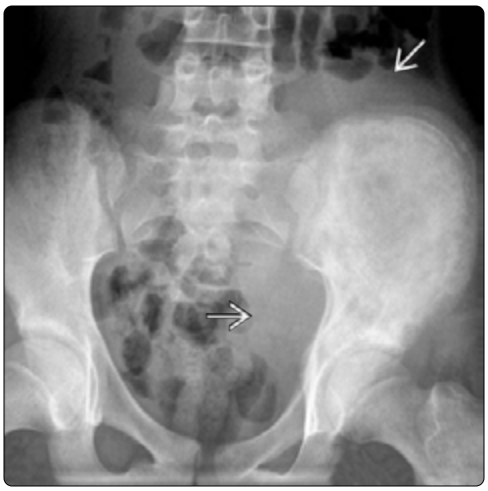
(Left) *AP radiograph in a 5 year old with shoulder pain shows abnormal lucency, permeation, & expansion of the entire right scapula.* **(Right)** *Axial T2 FS MR in the same patient shows that the entire scapula has been replaced by a solid mass that extends into the surrounding soft tissues. A thin low signal rim of expanded & permeated cortex remained. Note the residual glenoid articular cartilage & labrum. Biopsy of the mass confirmed Ewing sarcoma.*





(Left) *Axial STIR MR shows a destructive 1st rib lesion. There is a large surrounding soft tissue mass with neuroforaminal extension. Biopsy confirmed Ewing sarcoma.* **(Right)** *Axial NECT in a teenager with pelvic Ewing sarcoma shows innumerable lung nodules of metastatic disease.*





(Left) *Axial T2 FS MR in a 5 year old with a shoulder mass shows a heterogeneous mixed solid & cystic mass in the posterior right shoulder. The solid component enhanced with contrast (not shown) & had internal vascularity on a preceding Doppler US (not shown). Soft tissue Ewing sarcoma was confirmed at excision.* **(Right)** *AP radiograph shows a large soft tissue mass displacing bowel from a permeated, mottled, & expanded left iliac bone with overlying periosteal reaction. Ewing sarcoma was confirmed on biopsy.*

Osteosarcoma

KEY FACTS

TERMINOLOGY

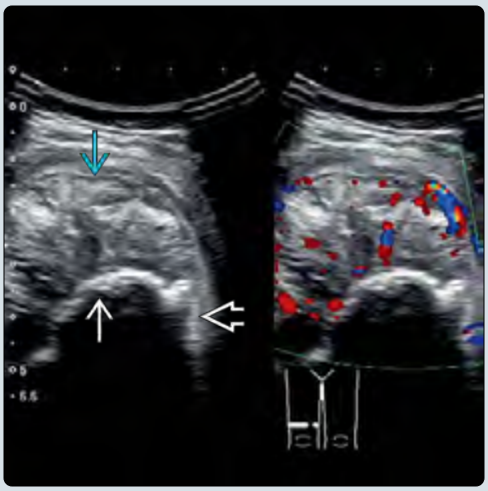
- Conventional high-grade intramedullary osteosarcoma (OS): 75-85%
- Less common OS subtypes (5% or less each): Telangiectatic, low-grade intramedullary, surface (low to high grade), multicentric, extraskeletal, secondary

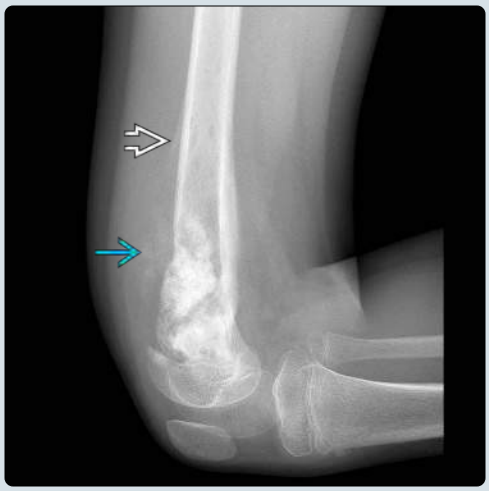
IMAGING

- Aggressive metaphyseal/metadiaphyseal lesion with variable mix of bone destruction & bone production (cloud-like osteoid)
  - 55-80% around knee; axial skeleton < 20%
- Radiographs often diagnostic or highly suggestive
- MR to characterize tumor + intra- & extraosseous extent
  - Large field of view joint-to-joint imaging
    - Look for skip metastases
    - T1 best sequence for intraosseous tumor margin
  - High-detail focused imaging with surface coils
    - Heterogeneous signal intensity on all sequences
      - Mineralized tumor: Dark T2
      - Nonmineralized tumor: Intermediate to bright T2
      - Necrosis: Bright T2; extensive fluid-fluid levels (layering blood products) in telangiectatic subtype
    - Evaluate relationship to physis, joint, neurovascular bundle
- PET for distant metastases; chest CT for small nodules

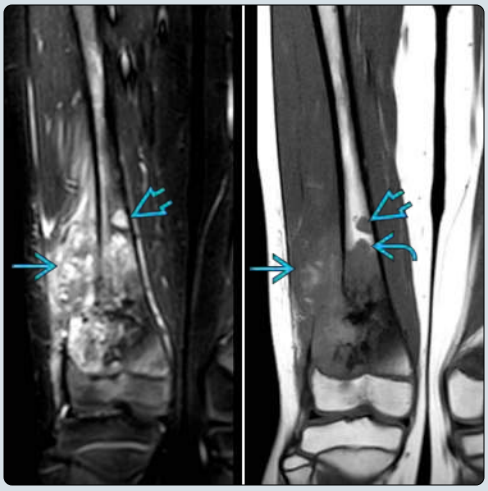
CLINICAL ISSUES

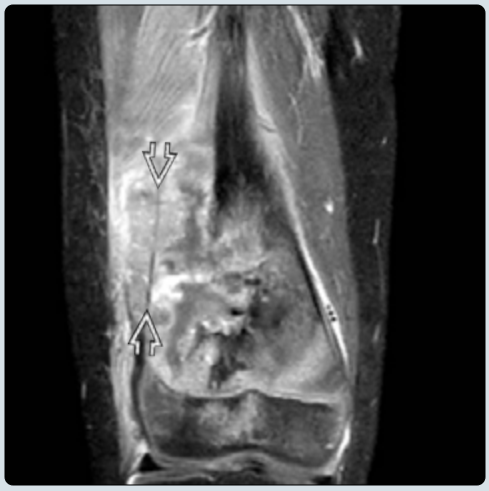
- Most common malignant primary bone tumor in children/young adults
- Bimodal age distribution: 10-30 years, > 60 years
- Presentations: Pain, mass, recent trauma
- Treatment
  - Neoadjuvant chemotherapy → surgical resection → adjuvant chemotherapy
    - Improved survival if necrosis > 90% at resection
  - Lung nodule metastasectomy (if low-volume disease)
    - 15-20% with metastases at diagnosis





(Left) *Grayscale (left) & color Doppler (right) US images of an 8 year old with a hard mass of the distal thigh show a heterogeneous mass arising from an underlying permeated femur. Note the periosteal elevation & increased vascularity of this aggressive lesion.* **(Right)** *Lateral radiograph in the same patient shows an aggressive mixed lytic & blastic lesion of the distal femoral metadiaphysis with cloud-like osteoid anteriorly. Note the elevated, interrupted periosteal reaction (Codman triangle) proximally.*





(Left) *Coronal STIR (left) & T1 (right) MR images of the same patient show heterogeneity of the metadiaphyseal mass with extraosseous extension of tumor. On the T1 image, the tumor has a sharp but nonsclerotic interface with the adjacent fatty yellow marrow, a typical feature of aggressive bone neoplasms. A small skip metastasis is noted.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows heterogeneous enhancement of this conventional osteosarcoma (OS). The mass elevates & extends beyond the periosteum.*

TERMINOLOGY

Synonyms

- Osteogenic sarcoma, conventional osteosarcoma

Definitions

- Osteosarcoma (OS): Malignant tumor with ability to produce osteoid directly from neoplastic cells

IMAGING

General Features

- Best diagnostic clue
  - Aggressive metaphyseal/metadiaphyseal lesion with mix of bone destruction & new bone formation
    - Sclerotic (cloud-like) osteoid density extending beyond margins of underlying bone

Radiographic Findings

- Radiography
  - Conventional OS (75-85%)
    - Poorly defined, intramedullary mass
    - Extends through cortex: Frank lysis &/or moth-eaten destruction
    - Aggressive periosteal reaction: Codman triangle, sunburst appearance
    - Soft tissue mass with cloud-like osteoid matrix (90%)
  - Telangiectatic OS (< 5%)
    - Purely lytic geographic lesion; blown-out appearance
    - Cystic cavities filled with blood/necrosis
      - Fluid-fluid levels in 90% (may mimic ABC)
    - Enhancing nodular components
    - Pathologic fracture in 25%
  - Parosteal OS (3%)
    - Low-grade surface OS; better prognosis than conventional OS
      - > 90% 5-year survival
    - Age: 20-50 years (older than conventional OS)
    - Classically at distal posterior femoral metaphysis
    - Invasion of marrow in 25%
  - Periosteal OS (1%)
    - Intermediate to high-grade surface OS
    - Attached to underlying cortex with thickening, scalloping, &/or saucerization of cortex
    - Usually diaphyseal with no medullary involvement
      - Medullary involvement may have poorer prognosis
    - Femur + tibia (85-95%); ulna + humerus (5-10%)
  - High-grade surface OS (1%)
    - Peak incidence in 2nd decade of life
    - Partially mineralized mass
    - Underlying cortex often partially destroyed
    - Periosteal new bone along margins of lesion
    - May have minimal medullary involvement
  - Multicentric OS (1%)
    - Synchronous osteoblastic osteosarcomas at multiple sites (usually symmetric)
    - Exclusively in children (5-10 years)
    - Extremely poor prognosis
  - Secondary OS (5%)
    - Association with preexisting bone lesion: Paget disease, prior radiation, bone infarct

MR Findings

- T1WI
  - Very low signal: Mineralized tumor
  - Low-intermediate signal: Solid, nonmineralized tumor
  - High signal: Hemorrhage
  - Best sequence for determining true tumor margin
- T2 WI FS/STIR
  - Very low signal: Mineralized tumor
  - Intermediate-high signal: Nonmineralized tumor
  - High signal: Necrosis
    - Fluid-fluid levels with hemorrhagic components, especially telangiectatic OS
  - Tumor margin may blend with surrounding edema
- DWI
  - Highly cellular components restrict diffusion
    - Hemorrhage may also restrict diffusion
  - ADC values may help evaluate treatment response
    - 95% ↑ in ADC values after neoadjuvant chemotherapy correlates with histologic necrosis of > 90% (improved prognosis)
- T1 WI C+ FS
  - Heterogeneous, variable enhancement
    - Subtracted post- minus precontrast images helpful to determine true enhancement vs. T1 shortening due to hemorrhage
  - Changes in dynamic enhancement parameters reflect therapy response better than tumor volume changes

Nuclear Medicine Findings

- Bone scan
  - Intense uptake; used to detect metastatic disease
    - Not reliable for skip lesions
      - Lesions in same bone or across joint; occur in 2-6%
- PET
  - Intense metabolic activity in viable tumor
    - Differentiates from necrosis/posttherapeutic change
    - May help predict/evaluate treatment response

Imaging Recommendations

- Best imaging tool
  - Radiograph: Primary investigative tool for bone pain
    - Often diagnostic in OS
  - MR to further characterize lesion including extent in marrow, physis, joint, & soft tissues [including relationship to neurovascular (NV) bundle]
  - CT of chest for pulmonary metastatic disease
  - Sensitivity for skeletal metastases
    - PET/CT > whole-body MR > bone scan

DIFFERENTIAL DIAGNOSIS

Ewing Sarcoma

- Aggressive long bone diaphyseal or flat bone lesion
- Often shows permeation, mild remodeled expansion, large soft tissue mass
- No osteoid production

Stress Fracture

- Linear sclerosis without bone destruction
  - Often perpendicular to bone long axis
- Subtle solid periosteal reaction or cortical thickening

Aneurysmal Bone Cyst

- Numerous fluid-fluid levels with septal enhancement
- No soft tissue mass, nodular enhancement, or sclerosis
- Bony expansion more frequent than cortical disruption

Osteomyelitis

- Acute (most common): Typically presents prior to development of much aggressive radiographic change
  - MR shows poorly defined marrow abnormalities ± subperiosteal & soft tissue fluid collections
- Chronic: Can show mixed sclerotic & lucent foci

Myositis Ossificans

- Reactive lesion, typically to muscle injury
- Marked edema often surrounds intramuscular mass
- Ca^{2+} first seen at periphery of lesion within weeks

Bone Infarction

- Acute: May show only periosteal reaction & marrow edema
- Chronic: Classic serpentine/geographic sclerotic foci

PATHOLOGY

General Features

- Genetics
  - Most cases sporadic
  - Predisposing syndromes
    - Li-Fraumeni (*TP53* mutation)
    - Hereditary retinoblastoma (*RB1* mutation)
    - Rothmund-Thomson (*RECQL4* mutation)
- Associated abnormalities
  - Prior radiation, Paget disease

Microscopic Features

- Highly pleomorphic, spindle-shaped tumor cells producing different forms of osteoid
- 3 main histologic subtypes depending on sarcomatous component: Osteoblastic (50%), chondroblastic (25%), fibroblastic (25%)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, soft tissue swelling/mass
- Other signs/symptoms
  - Recent trauma, fever
- Clinical profile
  - Pathologic fracture: 5-10% in conventional OS
  - Pulmonary metastases can cause pneumothorax (calcifying)
  - Less common metastases: Bone, lymph nodes, liver, brain

Demographics

- Age
  - Bimodal distribution: 10-30 years; over 60 years
- Epidemiology
  - Most common malignant primary bone tumor in children/young adults
  - Incidence: 5/1 million children ≤ 19 years old

Natural History & Prognosis

- Prognosis depends on
  - Age, gender, tumor volume, histology, location, & stage
    - Better prognosis for extremity vs. axial tumors
    - 15-20% have metastatic disease at diagnosis
      - 80-85% to lungs; bone 2nd most common
  - Best predictor: Degree of necrosis following neoadjuvant chemotherapy
    - Improved survival with > 90% necrosis
- OS has ↑ incidence of secondary malignancies

Treatment

- Neoadjuvant chemotherapy → surgical resection → adjuvant chemotherapy
- Limb salvage procedures (> 80% of cases)
- Pulmonary nodule metastasectomy in isolated or low volume metastatic disease (can be curative)
- 5-year survival
  - Without metastases: 70-80%
  - With metastases at presentation: 20-30%
    - Pulmonary more favorable than bone or other sites
  - With metastases > 2 years after chemotherapy: 40%

DIAGNOSTIC CHECKLIST

Consider

- Possible prognostic indicators during/after neoadjuvant chemotherapy: ↑ ADC values (MR), ↓ dynamic enhancement (MR), ↓ SUVmax values (PET)

Image Interpretation Pearls

- Bone MR
  - Do not miss distinct skip or other metastatic lesions
    - Requires joint-to-joint imaging
  - Evaluate relationship to physis, joint, & NV bundle
    - Requires focused high-detail imaging
- Chest CT: Calcified & noncalcified pulmonary nodules must be presumed as metastatic until proven otherwise
  - Even in geographic locations with endemic granulomatous diseases (such as histoplasmosis)

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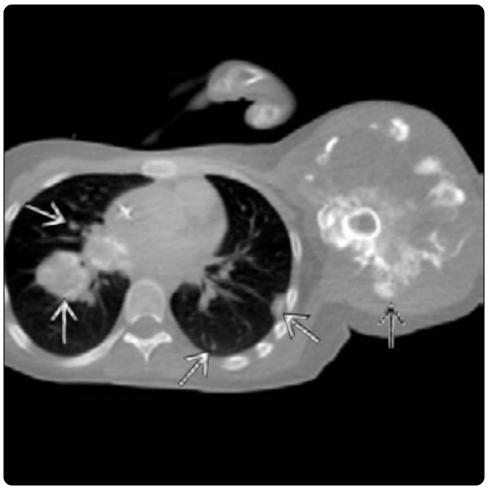
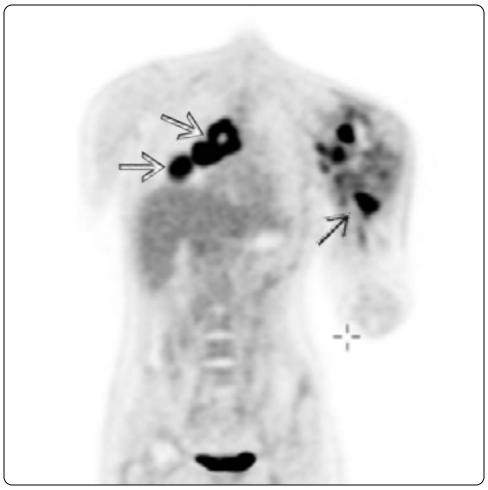
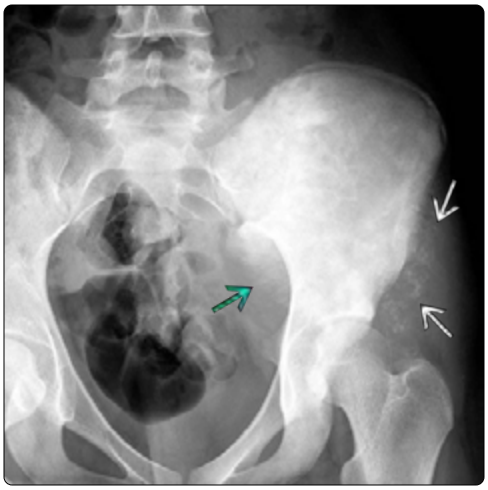
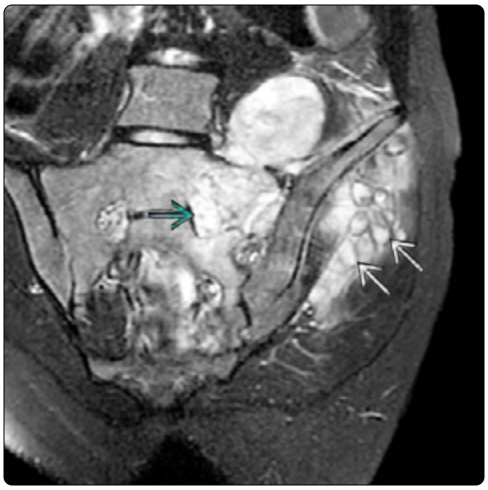


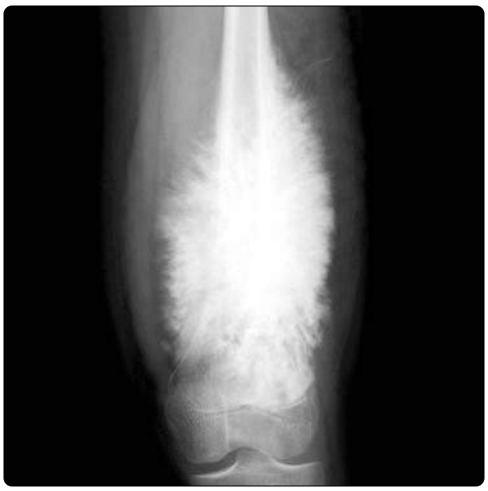


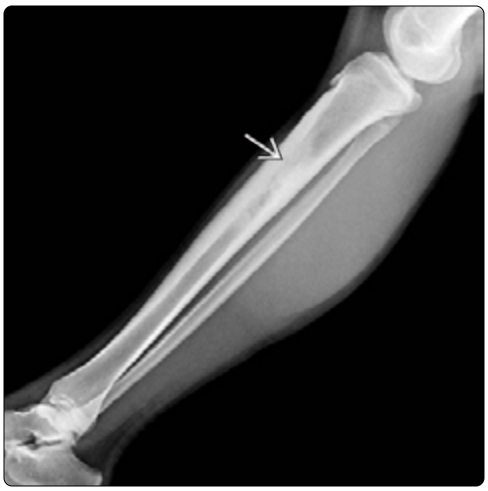
(Left) *Axial CT image in a teenager with osteoblastic OS shows a large, partially ossified left humeral mass ⇨ with multiple partially calcified lung metastases ➡.* **(Right)** *Coronal PET/CT image in the same patient shows increased metabolic activity in the shoulder mass ⇨ with an SUVmax of 8.7. Intense FDG accumulation is also seen within the right subcarinal/hilar & lung metastases ➡ with an SUVmax of 11.3. Metastatic lesions were also found in the brain & right adrenal gland (not shown).*





(Left) *AP radiograph in a 16 year old presenting with back & leg pain shows sclerosis of the left iliac wing with aggressive sunburst periosteal reaction ⇨. "Ring & arc" chondroid Ca^{2+} ➡ are seen within the lateral soft tissues.* **(Right)** *Coronal oblique T2 FS MR in the same patient shows heterogeneity of the sacrum & left ilium, both of which are involved by tumor. Soft tissue components of the mass extend into multiple left neural foramina ⇨. Note the multiple tumor cartilage lobules ➡ in this chondroblastic OS.*





(Left) *AP radiograph of a 12-year-old patient with leg swelling for 2 weeks shows an aggressive sunburst periosteal reaction within this distal femoral osteoblastic OS.* **(Right)** *Lateral radiograph shows a poorly defined intramedullary sclerotic lesion ➡ within the right tibial diaphysis. There is no visible periosteal reaction or soft tissue mass in this osteoblastic OS.*

Leukemia

KEY FACTS

TERMINOLOGY

- Leukemia: Malignancy of hematopoietic stem cells diffusely infiltrating or replacing normal bone marrow
- Granulocytic sarcoma or chloroma: Soft tissue mass of leukemic cells typically found with AML

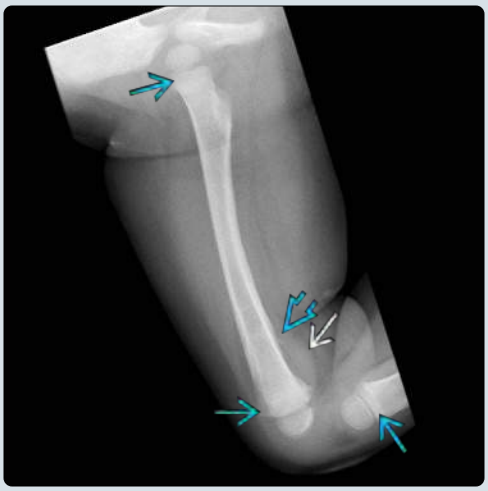
IMAGING

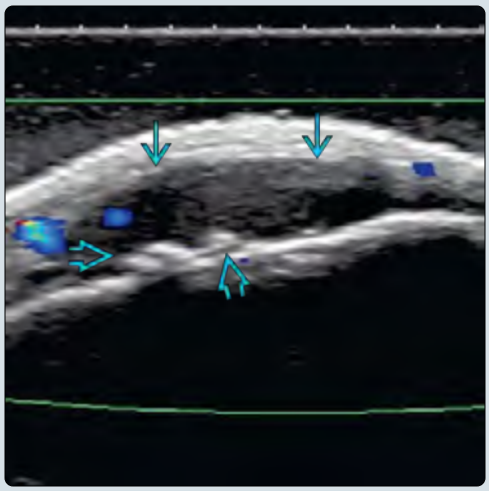
- Radiographs often normal or with subtle findings
  - Diffuse osteoporosis
  - "Leukemic lines"
    - Radiolucent metaphyseal bands; ZPC often intact
  - Focal bone destruction
    - Poorly defined metaphyseal osteolytic lesions with "moth-eaten" or permeative appearance
  - Aggressive periosteal reaction (lamellated, spiculated, interrupted) even if some components appear smooth
  - Pathologic fracture
  - Chloroma (granulocytic sarcoma)
  - ± chronic bone infarcts in treated patients
    - Avascular necrosis (AVN) with subchondral collapse
- MR: Leukemia may completely replace normal fatty marrow
  - Discordance of signal between abnormal infiltrated marrow & subcutaneous fat
  - ± superimposed acute infarction, fracture, or osteomyelitis causing focal symptoms

CLINICAL ISSUES

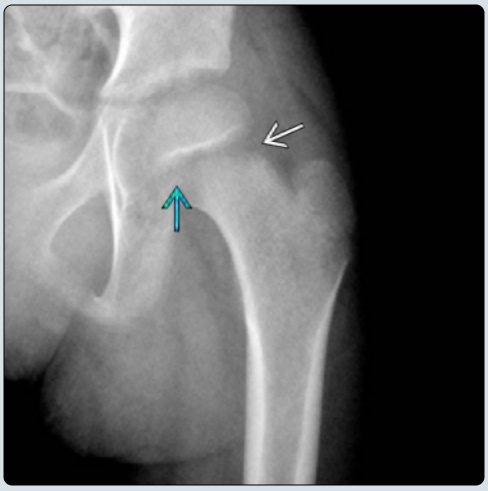
- Leukemia most common childhood malignancy
  - Subtypes: ALL > 75%, AML 15-20%, CML < 5%
- Most common presentations
  - Bone or joint pain, limp, swelling
  - Fatigue (anemia), fever ± infection, petechiae, bleeding
  - Hepatosplenomegaly, lymphadenopathy > 60%
- ALL: 5-year survival > 85%; 60-80% for others
- Treatment: Chemotherapy with steroids ± radiation, GCSF (→ ↑ red marrow content), stem cell transplant

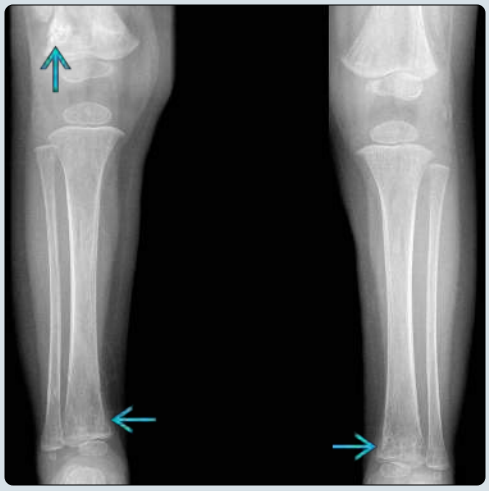
(Left) *Lateral radiograph in a 14 month old with anemia, fever, & refusal to bear weight shows subtle metaphyseal lucencies. There is a Codman triangle of aggressive periosteal reaction at the distal femur in association with a soft tissue mass.* **(Right)** *Transverse color Doppler US (same patient) shows a left temporal soft tissue mass with underlying calvarial permeation & irregular periosteal reaction. Patient was diagnosed with acute myelogenous leukemia (AML) & multifocal granulocytic sarcoma.*





(Left) *AP radiograph in an 8 year old with hip pain & acute lymphocytic (or lymphoblastic) leukemia (ALL) shows a lucent metaphyseal band within the proximal femur. The zone of provisional calcification is poorly visualized in this case.* **(Right)** *AP radiograph of each lower leg in an 11 month old shows pathologic fractures of the femoral, tibial, & fibular metaphyses at sites of underlying permeative osteolytic change. The patient was ultimately diagnosed with leukemia.*





TERMINOLOGY

Synonyms

- Acute lymphocytic (or lymphoblastic) leukemia (ALL), acute myelogenous leukemia (AML), myeloblastoma, chronic myelogenous leukemia (CML), chronic lymphocytic leukemia (CLL), juvenile chronic myeloid leukemia (JCML)

Definitions

- Leukemia: Malignancy of hematopoietic stem cells diffusely infiltrating or replacing normal bone marrow
- Granulocytic sarcoma or chloroma: Soft tissue mass of leukemic cells typically found with AML
- Extramedullary sanctuary sites: Sites of leukemia with barriers to systemic therapy (CNS & reproductive organs)

IMAGING

Radiographic Findings

- Radiography
  - Often normal or with very subtle metaphyseal findings
    - Only 40% have radiographic findings
  - Diffuse osteoporosis
    - Coarse trabeculae with abnormally well-visualized sclerotic rim [due to intact zone of provisional calcification (ZPC) or subchondral bone]
    - Multiple flattened, collapsed, or biconcave vertebrae
  - "Leukemic lines"
    - Radiolucent metaphyseal bands; ZPC often intact
      - Stress of disease or leukemic infiltration
    - ± dense metaphyseal bands post therapy
  - Focal bone destruction
    - Poorly defined osteolytic lesions with "moth-eaten" or permeative bone destruction
  - Periosteal reaction
    - Usually has some aggressive features (lamellated, spiculated, interrupted) even if some components appear smooth
      - Subperiosteal infiltration by malignant cells through haversian canals
      - Subperiosteal hemorrhage less common
  - Sclerotic foci
    - Typically in myelogenous leukemia
    - ± chronic bone infarcts in treated patients
  - Pathologic fracture
    - Usually metaphyseal
    - Can simulate nonaccidental trauma
  - Chloroma (granulocytic sarcoma)
    - Nonspecific soft tissue mass
    - Most commonly head & neck, soft tissues, GI system, lymph nodes, peritoneum, or bony mass
      - Can simulate meningioma or epidural hematoma
    - Poor prognosis
  - Late findings
    - Avascular necrosis (AVN) with subchondral collapse
    - Endochondral ossification rarely interrupted with persistence of unossified cartilage in metaphysis

CT Findings

- Bone CT
  - Permeative bone destruction, periosteal reaction
  - ± soft tissue mass of chloroma
  - Sclerotic foci of AVN

MR Findings

- **T1WI**: Confluent dark leukemic infiltrate replacing normal high-signal fatty yellow marrow
  - Marrow signal abnormally discordant from signal of bright subcutaneous fat
- **T2 FS/STIR**: Poorly defined patchy or diffusely bright marrow signal of visualized bones
  - Marrow signal abnormally discordant from signal of dark subcutaneous fat
- **T1 C+ FS**: Uniform intermediate to mildly bright enhancement of leukemic marrow diffusely
- **DWI**: May show restricted diffusion
- Special circumstances
  - Acute infarction or osteomyelitis
    - May present before, during, or after treatment
    - Focal regions of superimposed marrow heterogeneity with ↓ enhancement & overlying periosteal & soft tissue edema
  - Chloroma
    - Intermediate to bright T2 FS/STIR signal soft tissue mass with heterogenous or uniform enhancement
  - Recovery of normal marrow signal after treatment
    - Gradual process in setting of effective therapy
      - ↑ fat fraction, T1 shortening
  - Relapse after stem cell transplant
    - Numerous, round, well-circumscribed "dots" of leukemic infiltrate
    - T1 dark, T2 FS/STIR bright; enhance with contrast
    - May require targeted biopsy rather than blind iliac marrow aspiration to confirm recurrence

Nuclear Medicine Findings

- Bone scan
  - ↑ radiotracer uptake in tumor
    - May underestimate disease
- PET
  - Helpful to identify extramedullary disease

Imaging Recommendations

- Best imaging tool
  - MR: T1, T2 FS/STIR, T1 C+ FS
    - Musculoskeletal symptoms with normal radiographs
    - Whole-body MR can screen for osteonecrosis

DIFFERENTIAL DIAGNOSIS

Metastatic Neuroblastoma

- Bone involvement similar to leukemia
  - Metaphyseal lucent bands
  - "Moth-eaten" bone destruction
  - Circumscribed or diffuse marrow replacement
  - Spiculated periosteal reaction of skull

Sickle Cell Disease

- Hyperplastic red marrow replaces varying degrees of normal yellow fatty marrow
  - Drops signal on opposed-phase imaging
- Marrow infarctions, H-shaped vertebrae

Gaucher Disease

- Marrow accumulation of glucocerebroside ± infarctions
- Erlenmeyer flask deformities of undertubulated distal femurs

Langerhans Cell Histiocytosis

- Focal, punched-out lytic lesion
- Intense marrow edema + homogeneous soft tissue mass
- ± periosteal reaction, soft tissue edema

Osteomyelitis

- Symptoms similar to leukemia
- Extensive, poorly defined soft tissue abnormalities
- Poorly defined metaphyseal-centric marrow process
  - ± drainable fluid collections

Congenital Syphilis

- Metaphyseal lucent bands
- Hepatosplenomegaly, lymphadenopathy, anemia, skin rash
- Wimberger corner sign: Focal destruction of medial proximal tibial metaphysis

Lymphoma

- Typically solitary, often sclerotic focus
- Older age group

Ewing Sarcoma

- Diaphyseal lesion with aggressive periosteal reaction & permeative bone destruction
- Sharply circumscribed marrow interface on T1 MR
- Large soft tissue mass

Rickets

- Physeal lengthening/widening with loss of ZPC
- Metaphyseal fraying, cupping, & splaying

Physeal Stress Injuries

- Physeal lengthening/widening with loss of ZPC
- Isolated to painful extremity

PATHOLOGY

General Features

- Etiology
  - Arises from primitive stem cell either de novo or from preleukemic state
- Genetics
  - ↑ ALL risk in Down syndrome, Li-Fraumeni syndrome, Fanconi anemia, immunodeficiencies
  - CML translocation chromosome 9 & 22 classic (Philadelphia chromosome)
- Associated abnormalities
  - Myelodysplastic syndrome (1/3 develop AML)
  - Long-term risk from low medical levels of diagnostic imaging ionizing radiation debated
- Classified
  - Basis of cell maturity: Acute (blasts) or chronic (more mature cells)
  - Basis of cell type: Lymphocytic or myelogenous form

Microscopic Features

- Acute: Infiltration of bone marrow by poorly differentiated blast cells
  - ALL: Patternless sheets of small blue cells
  - AML: Wright stain or Giemsa preparation with lysosomal cytoplasmic structure (Auer rods)
- Chronic: Mature leukocyte infiltration
  - CML: Mature granulocyte with normal lymphocyte count, Philadelphia chromosome
  - CLL: Mature lymphocytes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Bone or joint pain, limp, swelling
    - Sharp, localized, recurrent arthralgias (75%)
  - Fatigue (anemia), fever ± infection, petechiae, bleeding
  - Hepatosplenomegaly, lymphadenopathy > 60%
- Other signs/symptoms
  - Large mediastinal mass
    - Superior vena cava syndrome, tachypnea, respiratory distress
  - ↑ ESR, thrombocytopenia, neutropenia

Demographics

- Age
  - ALL: Peak 2-10 years
  - AML: Peak > 65 years, but accounts for 15-20% of childhood leukemias
  - CML: Peak 30-50 years
  - CLL: Median 60 years
- Epidemiology
  - Most common pediatric malignancy
    - ALL most common form in children
      - ALL > 75%, AML 15-20%, CML < 5%

Natural History & Prognosis

- ALL: 5-year survival > 85%
- AML: 5-year survival ~ 60-70%
- CML: 5-year survival ~ 60-80%

Treatment

- Chemotherapy with steroids ± radiation
- Intrathecal chemotherapy for CNS disease
- Granulocyte colony-stimulating factor (GCSF)
- Stem cell transplant

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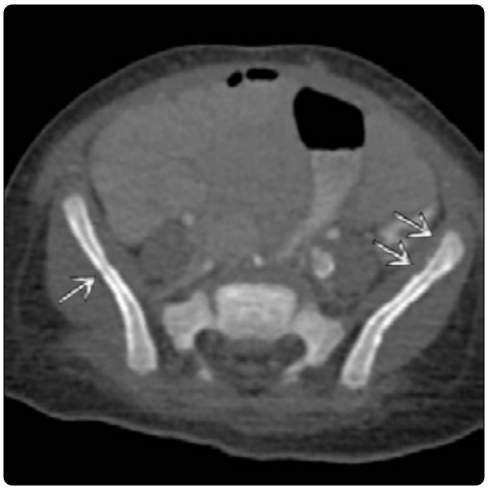
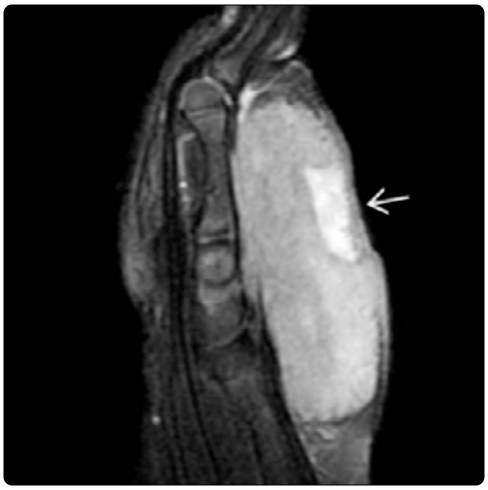
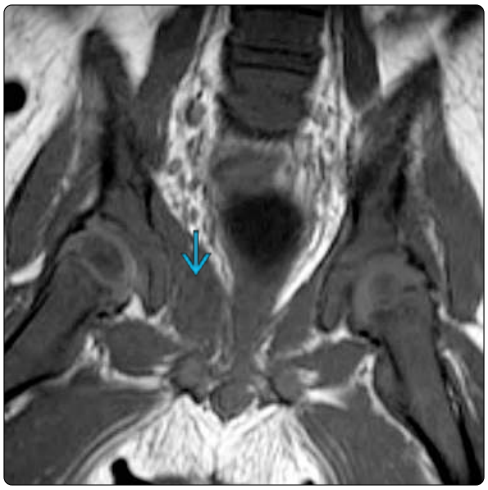
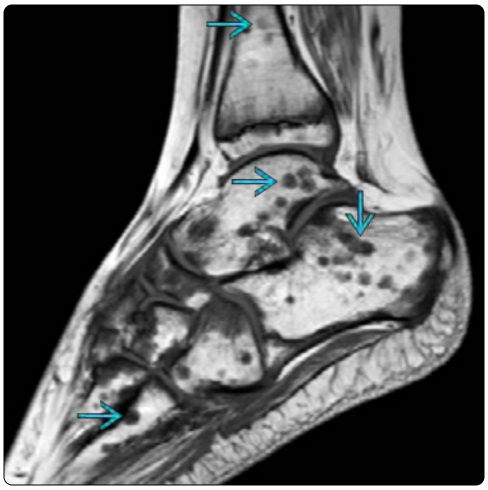


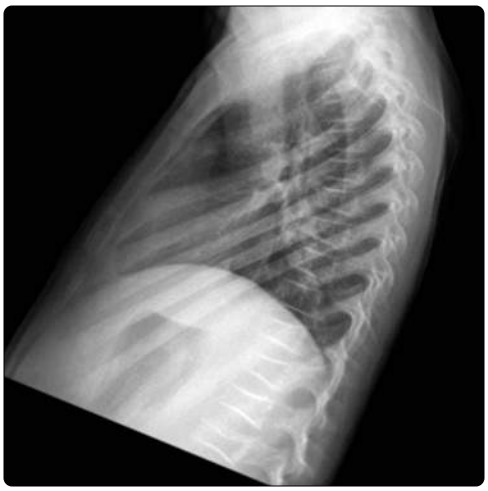


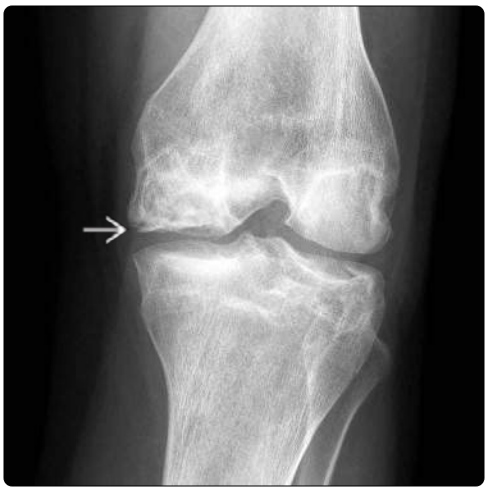
(Left) *Axial CECT (in bone windows) in a 14 month old with AML shows subtle cortical permeation & aggressive periosteal reaction along both iliac wings.* **(Right)** *Sagittal T2 FS MR in an 18 month old who presented with an enlarging dorsal wrist mass & multiple scalp lesions (not included) shows a solid-appearing, mildly hyperintense mass ultimately diagnosed as a granulocytic sarcoma (or chloroma). More focal hyperintense signal within the lesion is due to a recent biopsy.*





(Left) *Coronal T1 MR in a 21 month old with AML shows diffuse loss of normal fatty marrow signal throughout all visualized bones, typical of leukemic marrow. An associated intramuscular granulocytic sarcoma was more clearly seen on other sequences.* **(Right)** *Sagittal T1 MR in a 14 year old with a history of ankle pain 2 years after a bone marrow transplant for ALL shows numerous round lesions in a background of normal yellow marrow. Recurrent leukemia was confirmed by biopsy in this case.*





(Left) *Lateral chest radiograph in a 5 year old with newly diagnosed ALL who had lucent metaphyseal bands on hip radiographs (not included) shows osteoporosis & compression fractures throughout the spine.* **(Right)** *AP radiograph in a teenager with a history of treated ALL shows medullary sclerosis & subchondral collapse from steroid-induced osteonecrosis. Less pronounced osteonecrotic changes are also seen in the tibial plateau & lateral femoral condyle.*

Langerhans Cell Histiocytosis

KEY FACTS

TERMINOLOGY

- Langerhans cell histiocytosis (LCH): Spectrum of diseases caused by neoplastic clonal proliferations of CD1a, CD207, S100 protein-positive dendritic cells
- Single system (SS) (unifocal or multifocal) vs. multisystem (MS) disease
  - Bone (80-90%), skin (50%) most frequently involved
  - Risk organ (RO) involvement: Liver, spleen, marrow; confers worse prognosis (high risk)
  - Other organs: Lymph nodes, lung, pituitary, thymus, GI tract

IMAGING

- Best clue: Well-defined round or lobulated lytic punched-out skull lesion(s) without sclerotic rim
- Variable radiographic appearance of skeletal lesions depending on phase of disease ± therapy
- Monostotic vs. multifocal involvement: 50-75% vs. 10-20%
- Affected sites: Flat bones (50%) vs. long bones (30%)
  - Skull > ribs > femur > pelvis > spine
- FDG PET highly sensitive for active LCH (FDG avid); less sensitive for vertebral disease than whole-body STIR MR

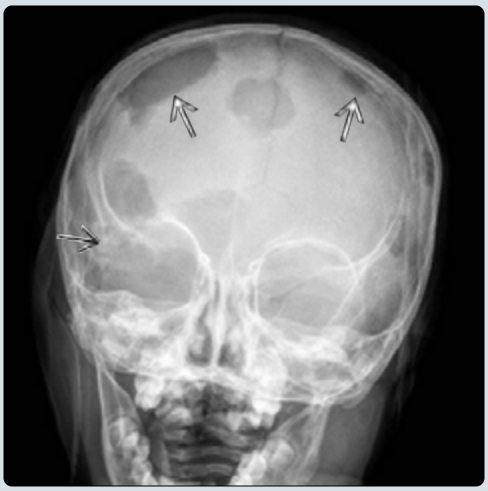
CLINICAL ISSUES

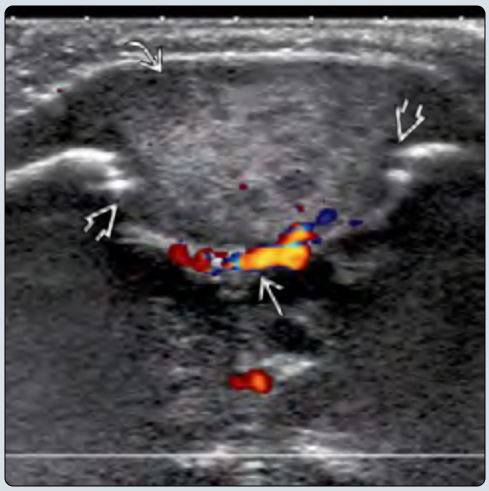
- Age: 90% of LCH cases < 15 years at presentation
  - SS, unifocal (70% of cases); peak age: 5-15 years
  - SS, multifocal (20% of cases); peak age: 1-5 years
  - MS RO+ disease (10% of cases); peak age: 0-2 years
- Mortality: SS or MS RO- disease < 5%; MS RO+ disease 10-50%
- Spontaneous regression of unifocal bone disease common
- Chemotherapy, steroids for multifocal SS or MS disease
- Disease reactivation in 25-75% (MS > SS)
- Long-term sequelae in 25-70% (MS > SS)

DIAGNOSTIC CHECKLIST

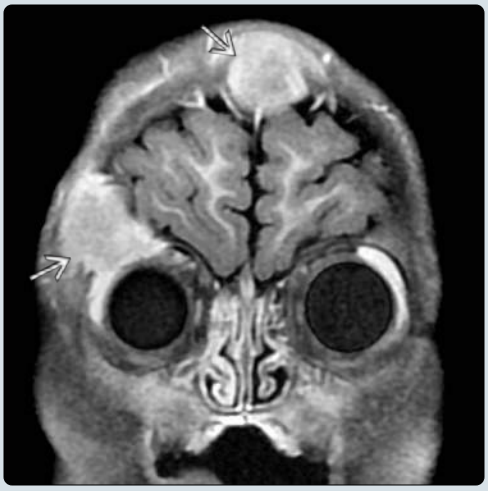
- LCH almost always reasonable diagnostic consideration for pediatric bone lesion due to variable appearances

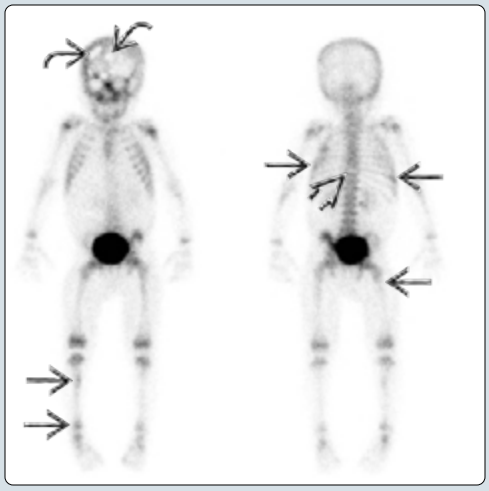
(Left) *AP radiograph of the skull in a 1-year-old girl with palpable masses shows multiple punched-out lytic lesions of the calvarium. The superior & lateral walls of the right orbit are destroyed.* **(Right)** *Coronal color Doppler ultrasound of the midline frontal calvarium in the same patient shows a discrete hypovascular soft tissue mass filling a site of complete osteolysis. The mass is compressing the adjacent superior sagittal sinus.*





(Left) *Coronal T1 C+ FS MR in the same patient shows enhancing soft tissue masses at the sites of the lytic bone lesions.* **(Right)** *Anterior & posterior Tc-99m MDP bone scan in the same patient show multiple foci of decreased radiotracer uptake within the skull. A focus of decreased uptake at T9 corresponded to a level of vertebra plana on radiographs (not shown). Several foci of increased skeletal uptake are also noted. Langerhans cell histiocytosis (LCH) was confirmed with biopsy.*





TERMINOLOGY

Synonyms

- Histiocytosis X, eosinophilic granuloma (EG), Hand-Schüller-Christian disease, Letterer-Siwe disease

Definitions

- Langerhans cell histiocytosis (LCH): Spectrum of disorders caused by neoplastic clonal proliferations of CD1a, CD207, S100 protein-positive dendritic cells
- Single system (SS) (unifocal or multifocal) vs. multisystem (MS) disease
  - Bone (80-90%), skin (50%) most frequently involved
    - Lung most common SS site in adults
  - Risk organ (RO) involvement: Liver, spleen, marrow; confers worse prognosis ("high risk")
    - Lung no longer considered RO
  - Other organs: Lymph nodes, pituitary, thymus, GI tract
  - CNS-risk lesion: Skull base & many facial lesions
    - At risk for diabetes insipidus & delayed neurodegenerative changes

IMAGING

General Features

- Best diagnostic clue
  - Well-defined round or lobulated lytic punched-out skull lesion(s) without sclerotic rim
- Location
  - Monostotic involvement (unifocal SS): 50-75%
  - Multifocal SS involvement: 10-20%
  - Predilection for flat bones (> 50% of cases): Calvarium, mandible, ribs, pelvis, scapula
  - Long bones affected ~ 30% of cases
    - Metaphysis, diaphysis > epiphysis
  - Decreasing order of frequency: Skull, ribs, femur, pelvis, spine
- Morphology
  - Variable radiographic appearance of skeletal lesions depending on phase of disease ± therapy
    - Lucent > sclerotic; geographic vs. permeative; well-defined sclerotic vs. poorly defined borders; aggressive vs. nonaggressive periosteal reaction

Radiographic Findings

- Radiography
  - Skull (50%)
    - Most common presentation: Solitary skull lesion
    - Well-defined lytic lesion without sclerotic rim
      - Sclerotic rim does occur during healing phase (which may occur before therapy)
    - "Beveled" edge: Asymmetric involvement of inner vs. outer table of skull
    - Coalescence of lesions: Geographic skull
    - Button sequestrum: Sclerotic focus within lytic lesion
    - Soft tissue mass overlying lytic lesion
    - Floating tooth sign: Lesion in alveolar mandible → loss of dense lamina dura around tooth
  - Appendicular skeleton
    - Variable appearance
    - Lesions respect joint space/growth plate
  - Spine
    - Vertebra plana: Complete collapse of vertebral body

MR Findings

- T1WI
  - Intermediate to low signal lesion replacing marrow fat
- T2WI FS
  - Intermediate to high signal within focal lesion
    - Surrounded by ill-defined hyperintense marrow > soft tissue edema
    - Hyperintense signal deep to or overlying periosteum
  - Purely medullary lesion vs. cortical destruction with soft tissue mass
  - ± hypointense lesion rim
  - Findings may change with phase of disease
- STIR
  - Similar findings to T2WI FS
  - Whole-body STIR to assess for multifocal disease
    - More sensitive for skeletal & extraskeletal LCH than radiographs or bone scan
    - Limited ability to distinguish active vs. residual disease
- T1WI C+ FS
  - Diffuse enhancement of lesion & adjacent edema typical

Nuclear Medicine Findings

- Bone scan
  - Most lesions show ↑ radiotracer uptake
  - Lesions may demonstrate ↓ tracer uptake, sometimes with surrounding halo of ↑ uptake
    - Particularly in skull lesions
  - Lesions may not be detected with scintigraphy alone
    - Lower sensitivity than radiographic survey
- PET/CT
  - FDG PET highly sensitive for active LCH (FDG avid)
    - Less sensitive than MR in vertebral disease
  - Also demonstrates healing before other modalities
  - Higher radiation dose than other modalities; radiation dose reduction can be achieved with
    - Low-dose lesion-selective CT technique
    - PET/MR fusion

Imaging Recommendations

- Best imaging tool
  - PET/CT vs. whole-body STIR MR

DIFFERENTIAL DIAGNOSIS

Osteomyelitis

- Moderate to marked soft tissue inflammation
- Cortical & marrow abnormalities less well defined
- Rim-enhancing fluid collections often present

Metastatic Neuroblastoma

- Lucent metaphyseal bands &/or lytic metaphyseal permeative lesions
- Radiating spicules of periosteal new bone formation typical in permeative skull lesions

Leukemia

- Lucent metaphyseal bands &/or lytic metaphyseal permeative lesions
- ± diffusely abnormal background marrow

Ewing Sarcoma

- Permeative bone destruction, wide zone of transition, aggressive periosteal reaction; often large soft tissue mass
- ± remodeling with expansion, saucerization
- May involve entire flat bone; calvarium uncommon

Congenital Syphilis

- Hepatosplenomegaly, lymphadenopathy, rash, lucent metaphyseal bands
- Wimberger sign: Lytic lesion of proximal medial tibia

Infantile Myofibromatosis

- Single or multifocal bone &/or soft tissue lesions
- Expansile bubbly lucent metaphyseal lesions
- Heterogeneous MR signal/enhancement

PATHOLOGY

General Features

- Historic categorization
  - Letterer-Siwe (acute disseminated form): 10%
  - Hand-Schüller-Christian (chronic disseminated form): 20%
  - EG (isolated bone or lung involvement): 70%

Microscopic Features

- Active lesion: Granuloma of dendritic Langerhans cells > inflammatory cells
- Later stages: Macrophages > Langerhans cells + fibrotic & xanthomatous changes
- Birbeck granule: Classic tennis racquet organelle found by electron microscopy in up to 40% of Langerhans cells
  - Diagnostic importance now diminished due to immunostaining of specific markers (CD1a, CD207, S100 protein)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Localized bone pain, tenderness
  - Soft tissue swelling or mass
- Other signs/symptoms
  - Rash, limping, exophthalmos, hepatosplenomegaly, lymphadenopathy, otitis externa, mastoiditis, gingivitis
- Clinical profile
  - Fever, ↑ erythrocyte sedimentation rate, leukocytosis

Demographics

- Age
  - 90% of cases < 15 years at presentation
  - SS, unifocal (70% of cases); peak age: 5-15 years
  - SS, multifocal (20% of cases); peak age: 1-5 years
  - MS RO+ (10% of cases); peak age: 0-2 years
- Gender
  - M:F = 1.2-3:1
- Epidemiology
  - Annual incidence: 5 per 1 million children < age 15 years

Natural History & Prognosis

- Mortality
  - SS or no RO (MS RO-) disease: < 5%
    - Spontaneous regression of unifocal bone disease common
  - MS disease + RO involvement (MS RO+): 10-50%
    - Progressive disease on multiagent chemotherapy at 6 weeks: 40-80% mortality
  - Lung disease in adults: 25% at 5 years
    - Regresses with smoking cessation
- Disease reactivation
  - Unpredictable timeframe, number of episodes
  - Up to 25% in multifocal SS (bone) disease
  - 50-75% in MS disease
  - Bone most frequent site of reactivation
- Long-term sequelae
  - Usually related to destructive/fibrotic changes at site(s) of initial disease
  - 70% in MS disease, 25% in SS disease
  - Diabetes insipidus, CNS degeneration (which may be paraneoplastic) most serious

Treatment

- SS disease
  - Unifocal bone disease: Watchful waiting vs. curettage & local steroid injection
  - Multifocal bone disease or CNS-risk lesion: Chemotherapy & steroids x 6-12 months
- MS disease (± RO): Multiagent chemotherapy x 12 months

DIAGNOSTIC CHECKLIST

Consider

- LCH should be considered in differential diagnosis for most pediatric bone lesions due to variable appearances
- If favoring LCH on initial targeted radiographs/MR, consider whole-body survey (radiographic vs. MR vs. PET) to help direct biopsy & determine disease extent

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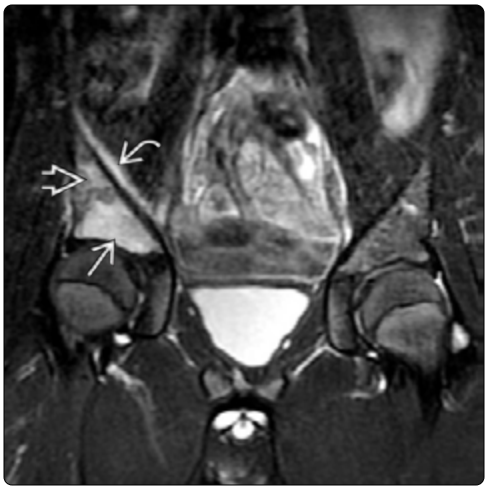
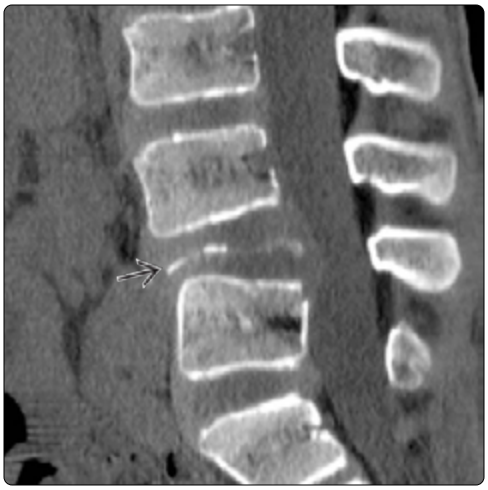
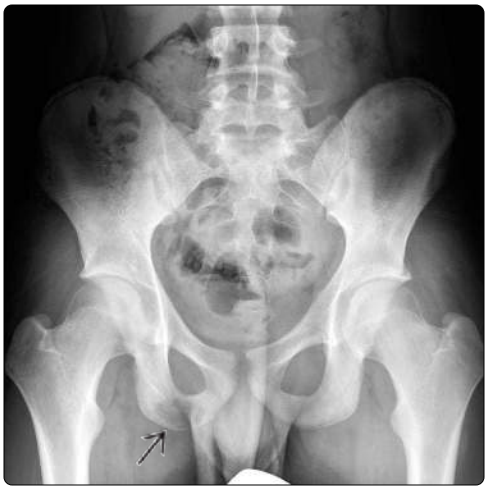
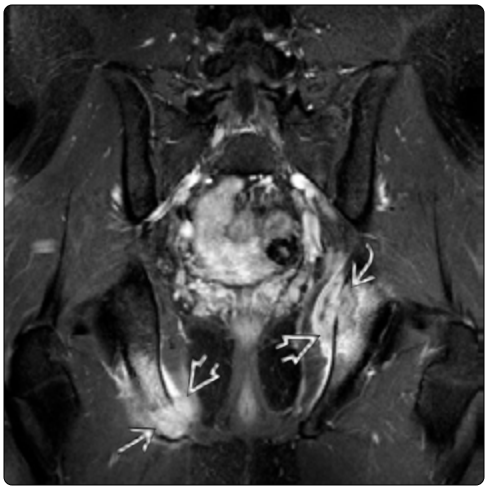


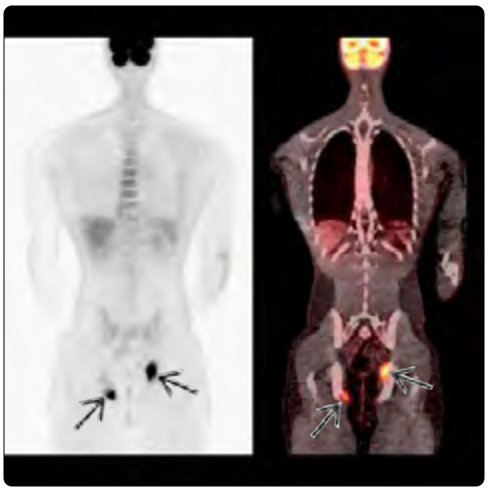


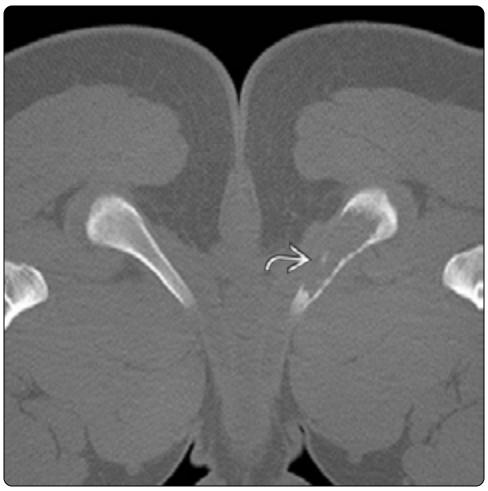
(Left) *Coronal T2 FS MR in a 10-year-old patient with right hip pain shows a homogeneously hyperintense lesion of the right iliac marrow without adjacent bone destruction. There is surrounding marrow & juxtacortical edema. LCH was confirmed with biopsy.* **(Right)** *Sagittal bone CT in a 12-year-old patient undergoing evaluation for a spinal deformity shows a wafer-like L4 vertebral body (vertebra plana), ultimately proven to be secondary to LCH.*





(Left) *AP radiograph in a 15-year-old boy with weeks of pelvic pain shows a lucent lesion with intermediate zone of transition in the right ischium.* **(Right)** *Coronal T1 C+ FS MR in the same patient shows enhancing bone lesions arising from the right ischial tuberosity & left posterior acetabular wall. Contiguous enhancing soft tissue masses overlie foci of cortical destruction, & there is adjacent juxtacortical & marrow edema at each site.*





(Left) *Coronal FDG PET/CT in the same patient shows FDG avidity (or increased metabolic activity) in the lesions, typical of LCH. No other lesions were identified.* **(Right)** *Prone axial bone CT of the same patient, obtained at the time of biopsy, shows the destructive lytic lesion of the right ischium. No residual posterior cortex remains, & the anterior cortex appears permeated. Biopsy confirmed LCH.*

Fibroxanthoma

KEY FACTS

TERMINOLOGY

- Very common benign fibrous lesion of pediatric bone
- Term fibroxanthoma includes
  - Nonossifying fibroma
    - > 2-cm length; encroachment on medullary cavity
  - Fibrous cortical defect
    - < 2-cm length
    - Essentially isolated to cortex

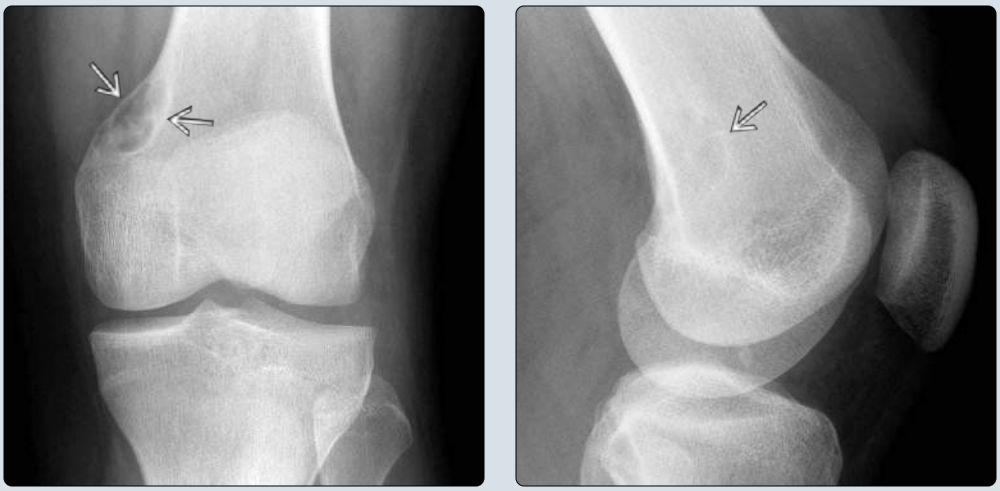
IMAGING

- Eccentric, elongated, bubbly, lucent lesion in long bone metaphysis/diaphysis with narrow zone of transition, lobular or smooth sclerotic margin, & no periosteal reaction
- Expected involution/healing → gradual sclerosis, resolution
- Typical locations
  - Metaphysis of long bone: Up to 93%
    - Distance from physis ↑ with age
  - Located around knee: 55-89%
- Radiographs usually diagnostic
  - Atypical features should suggest other lesion or superimposed complication (such as pathologic fracture)

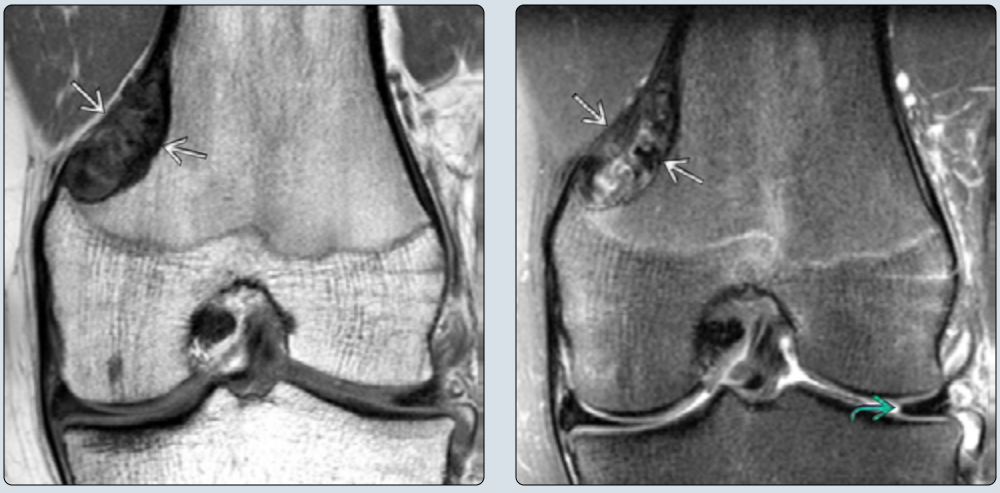
CLINICAL ISSUES

- Usually asymptomatic & incidentally discovered
- May develop symptoms (uncommon)
  - Acute pain with pathologic fracture
  - Gradual pain with stress fracture of adjacent bone (very rare)
  - Paraneoplastic rickets/osteomalacia (extremely rare)
- More likely developmental defect than neoplasm
- Occurs in up to 35% of children
  - Peak age: 10-15 years (75% in 2nd decade)
- Gradual healing, spontaneous regression of most cases in late adolescence
- Treatment only required if patient at high risk for pathologic fracture

(Left) *AP radiograph of the left knee in a 14-year-old boy with pain after an injury shows an incidental eccentric bubbly lucent lesion ➡ in the medial femoral metaphysis with a narrow zone of transition & sclerotic border, typical of a nonossifying fibroma (NOF) or fibroxanthoma. There is mild expansile remodeling & endosteal scalloping at this level without periosteal reaction.* **(Right)** *Lateral radiograph of the knee in the same patient again demonstrates the nonaggressive features of the NOF ➡.*



(Left) *Coronal T1 MR in the same patient shows heterogeneous, but largely hypointense, signal of the intralesional content ➡. The surrounding bone marrow & soft tissues are normal.* **(Right)** *Coronal T2 FS MR in the same patient shows heterogeneous, but largely hypointense, signal of the intralesional content ➡ with normal adjacent marrow & soft tissues. (The study was performed for, & confirmed, a suspected meniscal tear ⇨.)*



TERMINOLOGY

Synonyms

- Nonossifying fibroma (NOF), fibrous cortical defect (FCD)

Definitions

- Very common benign fibrous lesion of pediatric bone, usually asymptomatic
- Term fibroxanthoma includes
  - NOF
    - > 2-cm length
    - Encroachment on medullary cavity
  - FCD
    - < 2-cm length
    - Essentially isolated to cortex

IMAGING

General Features

- Best diagnostic clue
  - Eccentric, elongated, bubbly, lucent lesion in long bone metaphysis/diaphysis with narrow zone of transition, sclerotic margin, & no periosteal reaction
- Location
  - Metaphysis of long bone involved in up to 93%
    - Distance from physis ↑ with age
  - Tibia (43%), femur (38%), fibula (8%)
    - Around knee: 55-89%
  - Less common in upper extremity: 8%
    - Humerus: 5%
  - Usually solitary in absence of syndrome
- Size
  - 0.5-7.0 cm
- Morphology
  - Elongated, lobular, eccentric lesion

Radiographic Findings

- Radiography
  - Eccentric, cortically based, lucent lesion
  - Lesion long axis parallels bone long axis
  - Narrow zone of transition
  - Scalloped, lobular, or smooth deep sclerotic margin
  - ± mild expansile remodeling of overlying cortex &/or endosteal scalloping
    - Cortex thinned but usually intact (not always)
  - No matrix calcification
  - ± septation/trabeculation
  - No periosteal reaction without fracture or other complication
  - ↑ mineralization in healing stages

CT Findings

- Bone CT
  - Parallels radiographic findings
  - Attenuation of lesion matrix slightly > bone marrow

MR Findings

- T1WI
  - Low signal intensity centrally
  - Peripheral hypointense rim (reactive sclerosis)
- T2WI
  - Variable signal intensity centrally, most commonly low
    - T2-hypointense components due to fibrous tissue ± hemosiderin
  - Visible internal septa
  - Peripheral hypointense rim (reactive sclerosis)
  - No overlying soft tissue mass
  - No adjacent marrow, periosteal, or soft tissue edema without superimposed complication
- T1WI C+
  - Variable enhancement centrally, may be avid

Ultrasonographic Findings

- Grayscale ultrasound
  - Scalloped cortex filled with hypoechoic tissue
  - ↓ size, ↑ echogenic foci with healing
- Color Doppler
  - May show prominent internal blood flow

Nuclear Medicine Findings

- Bone scan
  - Almost all lesions show some degree of uptake
    - Mild to intense uptake in healing lesions
    - Faint to absent uptake in inactive or fully healed lesions
- PET/CT
  - Mildly to intensely ↑ metabolic activity (may mimic malignancy)
    - ↓ activity with healing, though FDG avidity of these lesions often differs from Tc-99m bone scan uptake
    - Correlation with CT appearance critical (or radiographs if PET performed without CT)

Imaging Recommendations

- Best imaging tool
  - Initial radiographs diagnostic in most cases
  - Usually no other imaging needed unless
    - Complications suspected by clinical or radiographic features
    - Larger lesion identified
      - May be less pathognomonic
      - Follow-up to evaluate growth & fracture risk
  - CT/MR helpful in confirming underlying fibroxanthoma in atypical or complicated lesion or with unusual clinical symptoms

DIFFERENTIAL DIAGNOSIS

Distal Femoral Metaphyseal Irregularity

- Cortical undulation/irregularity/interruption by lucent lesion with concave sclerotic deep margin
- Always at posterior, medial distal femur near medial gastrocnemius or adductor magnus attachments
- Overlaps fibroxanthoma imaging & histology despite poorly applied term cortical desmoid

Aneurysmal Bone Cyst

- Lucent metadiaphyseal lesion with marked expansile remodeling ± internal septations
- CT/MR: Fluid-fluid levels, septal enhancement

Unicameral Bone Cyst

- Centrally located metadiaphyseal lucent lesion without profound expansion or septations

- Fallen fragment sign in cases of fracture
- Classic location: Proximal humerus

Fibrous Dysplasia

- Mildly expansile medullary diaphyseal lesion
- Ground-glass (think frosted or smudged glass) appearance of matrix

Chondromyxoid Fibroma

- Eccentric bubbly lucent metadiaphyseal lesion
- ~ 90% expansile
- Much less common than fibroxanthoma

Enchondroma

- Central diaphyseal bubbly lucent lesion
- ± "ring & arc" chondroid matrix
- Most commonly found in hands & feet

PATHOLOGY

General Features

- Genetics
  - Vast majority of these lesions sporadic
  - Multiple lesions seen in
    - Neurofibromatosis type 1 (NF1)
    - Jaffe-Campanacci syndrome (may be NF1 subset)
      - Multifocal fibroxanthomas with extraskeletal manifestations in children
      - Café au lait spots, mental retardation, hypogonadism, cryptorchidism + cardiovascular, renal, & ocular abnormalities

Gross Pathologic & Surgical Features

- Fibrous, fleshy tissue with shades of gray & yellow
- Cystic changes, hemorrhage, &/or necrosis in larger lesion with pathologic fracture

Microscopic Features

- NOF/FCD histologically identical
- Bundles of spindle-shaped fibroblasts, scattered multinucleated giant cells, & foamy histiocytes
- Arranged in storiform pattern

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually asymptomatic & identified incidentally
- Other signs/symptoms
  - Acute pain with pathologic fracture
    - Risk greatest if lesion > 3.3 cm in size or causing loss of > 50% cortical thickness in weight-bearing bone
    - Quantitative CT methods better predictor of pathologic fracture risk
  - Adjacent stress fractures reported rarely with gradual pain onset
  - Rarely complicated by other processes (such as infection)
  - Oncogenic osteomalacia extremely rare
    - Paraneoplastic syndrome where benign bone tumor releases factors resulting in renal phosphate wasting with subsequent rickets/osteomalacia
    - Usually in adults (mean age: 35-40 years)

Demographics

- Age
  - 2-20 years; peak: 10-15 years
    - 75% in 2nd decade of life
    - Usually not seen after age 30
- Gender
  - M:F = 2:1
- Epidemiology
  - Most common fibrous lesion of bone
  - Occurs in up to 35% of children

Natural History & Prognosis

- Benign lesion; no malignant transformation risk
  - More likely developmental defect than neoplasm
- Presents during childhood, usually disappears in late adolescence
  - Involution over 2-4 years
- Rarely grows rather than involutes

Treatment

- Usually not required
- Curettage with bone grafting of larger lesions at risk for fracture
- Conventional casting or internal fixation if fracture occurs
  - Lesion may heal after fracture

DIAGNOSTIC CHECKLIST

Consider

- No need to biopsy or treat if appearance typical & fracture risk low
- MR to assess any atypical radiographic features

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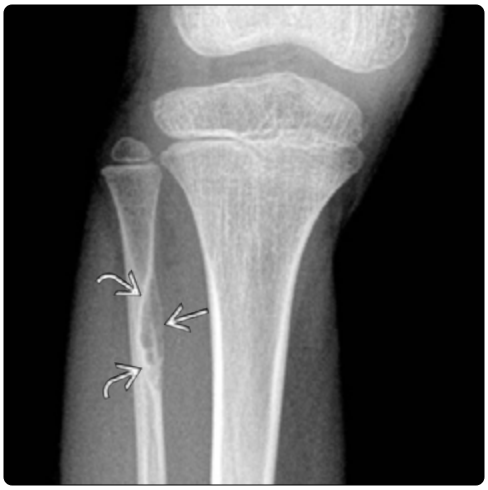
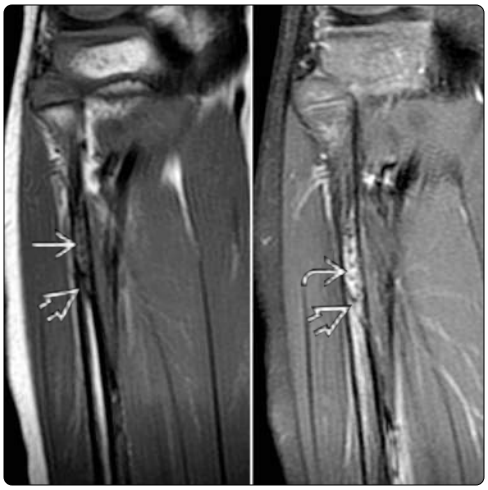
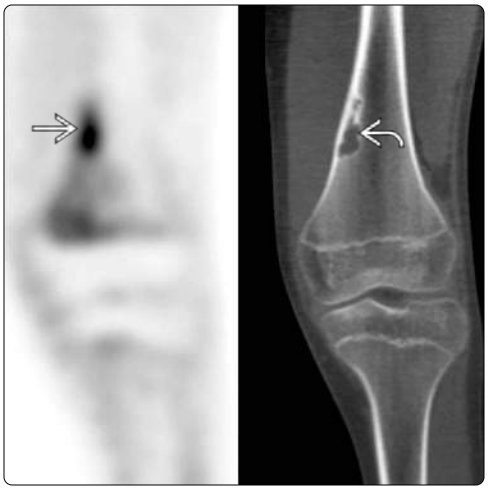
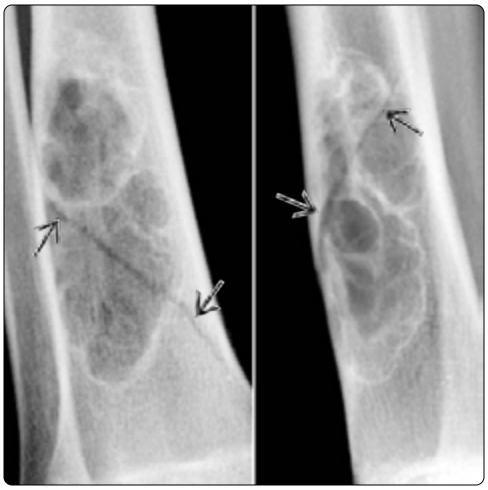


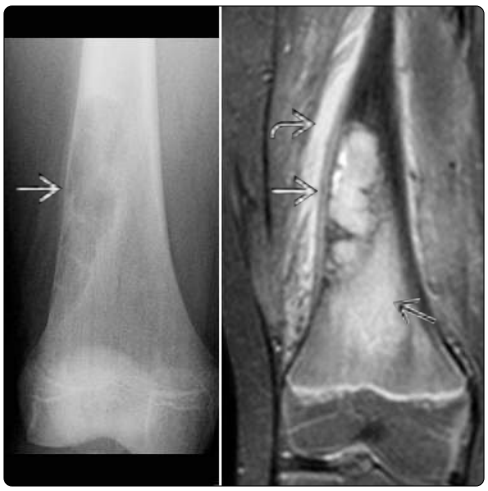


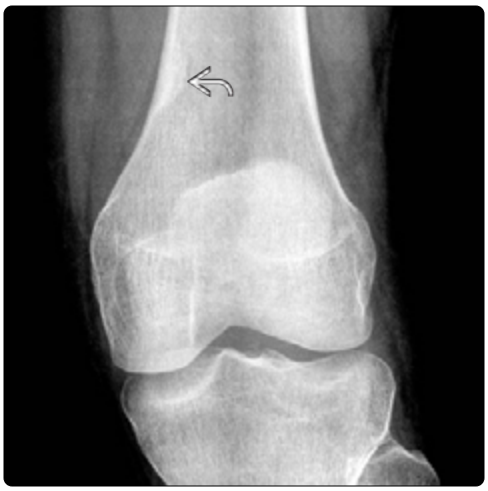
(Left) *AP radiograph shows a well-circumscribed, bubbly, lucent lesion eccentrically located in the proximal fibular diaphysis with a deep margin of sclerosis & mild expansile remodeling of the overlying cortex, typical of a nonossifying fibroma.* **(Right)** *Coronal T1 (left) & T1 C+ FS (right) MR images in the same patient show the low signal intensity rim of the lesion. The lesion shows intermediate signal intensity internally prior to contrast administration with heterogeneous enhancement after contrast.*





(Left) *Coronal FDG PET/CT from a teenager with nasopharyngeal carcinoma shows increased metabolic activity in an eccentric distal femoral lesion that has typical CT characteristics of an NOF.* **(Right)** *AP (left) & lateral (right) radiographs show an obliquely oriented pathologic fracture through a relatively large NOF in the distal tibia of this teenage patient who fell from a height of 12 feet.*





(Left) *AP radiograph (left) & coronal T2 FS MR (right) in a 10-year-old boy with knee pain show a large NOF with atypical periosteal reaction & edema of the surrounding soft tissues & marrow. No fracture was identified. Biopsy confirmed an NOF with superimposed osteomyelitis.* **(Right)** *AP radiograph of the knee in a 16-year-old girl shows a well-defined, crescentic, sclerotic lesion in the medial aspect of the distal femoral metadiaphysis, typical of a nearly healed fibroxanthoma/NOF.*

Osteoid Osteoma

KEY FACTS

TERMINOLOGY

- Benign osteoblastic lesion characterized by < 2 cm nidus of osteoid/woven bone in fibrovascular stroma

IMAGING

- Best clue: Well-defined small lucent lesion in tubular bone cortex with surrounding sclerosis & edema
- Locations
  - Cortical: 70-80%
    - Diaphyses > metaphyses of tubular bones
  - Medullary: 20-30%
    - Epiphyses or equivalents (intraarticular 10%)
    - Spine posterior elements
- Radiographs/CT
  - Small round or ovoid lucent nidus ± central Ca^{2+}
  - Surrounding cortical thickening, solid periosteal reaction
  - Prominent cortical vessels near nidus
- MR
  - Variable nidus signal on T2 FS MR; may be "target"
  - Surrounding marrow, periosteal, & soft tissue edema
    - Joint effusion/synovitis if osteoid osteoma intraarticular
  - Hyperenhancing nidus on early dynamic T1 C+ FS MR
- NM bone scan: Double density sign

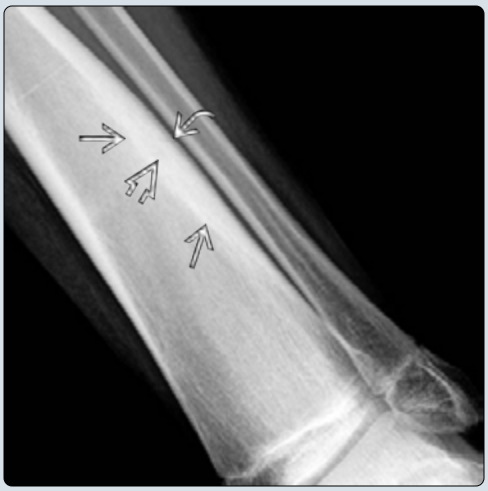
TOP DIFFERENTIAL DIAGNOSES

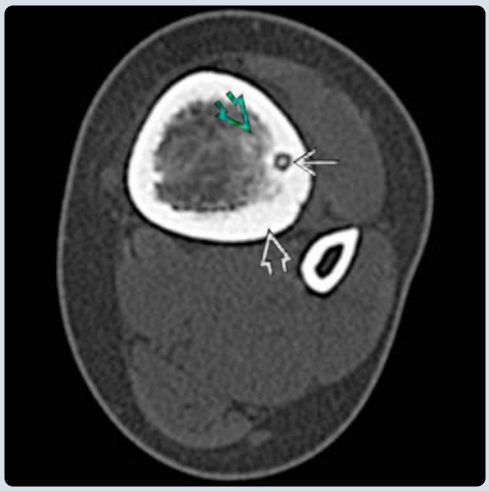
- Osteomyelitis, Langerhans cell histiocytosis, stress injury, osteoblastoma, osteosarcoma

CLINICAL ISSUES

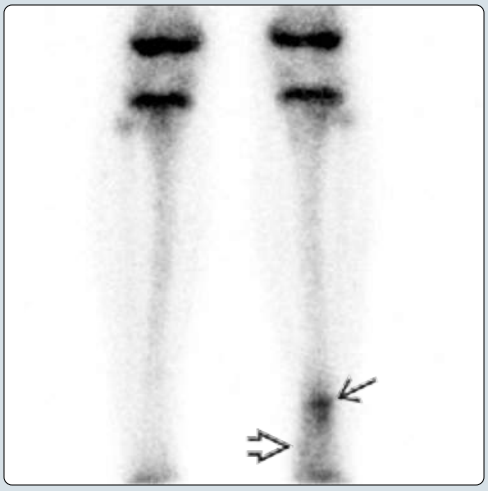
- Insidious pain, classically worse at night & relieved by NSAIDs
- Gradual spontaneous regression accelerated by NSAIDs
- Surgery curative if nidus completely resected
- Image-guided percutaneous interventions highly effective due to nidus visualization

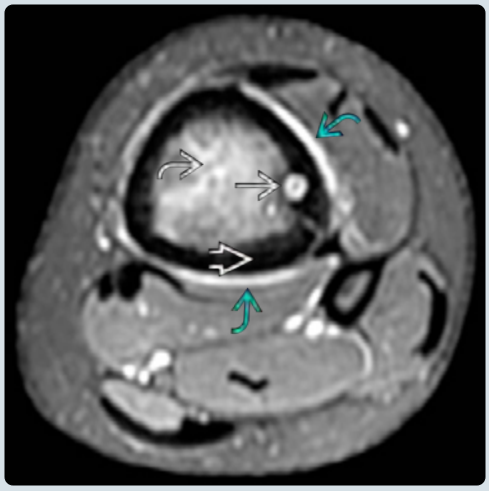
(Left) *AP radiograph of the left tibia in a 15-year-old girl with 1 year of lower leg pain shows a region of diaphyseal cortical thickening laterally with mild overlying solid periosteal reaction & a subtle lucent focus centrally.* **(Right)** *Axial bone CT of the same patient shows calcification within the hypoattenuating nidus of this osteoid osteoma (OO). The lesion is surrounded by cortical thickening & medullary sclerosis.*





(Left) *Tc-99m nuclear medicine bone scan of the same patient shows focally increased uptake at the level of the nidus. This "hot" focus is superimposed on a background of mildly increased radiotracer activity in the distal tibia (the double density sign, typical of OO).* **(Right)** *Axial T2 FS MR of the same patient shows a target appearance of the nidus with adjacent cortical thickening & surrounding marrow & periosteal edema.*





TERMINOLOGY

Abbreviations

- Osteoid osteoma (OO)

Definitions

- Benign osteoblastic lesion characterized by < 2-cm nidus of osteoid/woven bone in fibrovascular tissue

IMAGING

General Features

- Best diagnostic clue
  - Small well-defined, cortically based lucent lesion of tubular bone with surrounding sclerosis & edema
- Location
  - Distribution in body
    - Femur, tibia: 53-60%
    - Phalanges of hands & feet: 21%
    - Spine: 9%
      - Posterior elements: 90%
      - Vertebral body: 10%
      - Lumbar > cervical > thoracic > sacral
  - Distribution in bone
    - Cortical: 70-80%
      - Diaphyses > metaphyses of tubular bones
    - Medullary: 20-30%
      - Epiphyses or equivalents (10% intraarticular)
      - Posterior vertebral elements
    - Subperiosteal
- Size
  - Nidus: < 2 cm
- Morphology
  - Isolated ovoid or round lucent nidus with surrounding sclerosis, periosteal reaction, edema
    - Multicentric cases rarely reported

Radiographic Findings

- Radiolucent central nidus usually < 1 cm
  - Nidus often partly calcified
- Adjacent cortical thickening & medullary sclerosis common
  - Less frequent with medullary/intraarticular OO
- Periosteal reaction: Solid > lamellated
  - Often distant from intraarticular OO
- Focal cortical bulge or scalloping may be present
- Joint effusion, synovitis with intraarticular OO
- ± distal osteoporosis, muscle atrophy

CT Findings

- Small, well-defined, round or ovoid hypoattenuating nidus surrounded by thickened cortex & medullary sclerosis
- Central Ca^{2+} of nidus often present, may have target appearance
- Overlying nonaggressive periosteal reaction
- Prominent cortical vascular channels near nidus

MR Findings

- T1WI
  - Low to intermediate signal nidus
  - ± adjacent muscle atrophy
- T2WI FS/STIR
  - Unmineralized nidus shows intermediate to high signal; mineralized nidus shows low signal
  - ± target appearance of nidus
    - Low signal centrally from nidus Ca^{2+}
    - Surrounding high signal of unmineralized nidus
    - Outer low-signal rim of sclerosis
  - Adjacent abnormal fluid signal
    - Marrow, periosteal, & soft tissue edema
      - Extensive marrow edema can obscure small nidus
      - Half-moon sign (or half-circle) of marrow edema may be suggestive of femoral neck OO
      - "Pseudotumor" of surrounding soft tissue inflammation reported at phalangeal OO
    - Joint effusion/synovitis if OO intra/periarticular
- T1WI C+ FS
  - Dynamic imaging: Peak enhancement of nidus during early arterial phase with subsequent partial washout
    - Slower enhancement of adjacent marrow edema
  - Reactive synovitis with intra/periarticular OO

Ultrasonographic Findings

- Color Doppler
  - ↑ vascularity of superficial nidus
    - May be obscured by overlying thickened cortex
  - Can be used to localize lesion for biopsy

Nuclear Medicine Findings

- Bone scan
  - ↑ radiotracer uptake
    - Double density sign: Small focus of intensely ↑ activity (nidus) surrounded by larger area of less intensely ↑ activity (reactive sclerosis)
  - SPECT: ↑ sensitivity for subtle spine OO
    - Fusion with targeted CT or MR at site of abnormal uptake helpful

Imaging Recommendations

- Best imaging tool
  - SPECT-CT (if OO suspected)
- Protocol advice
  - CT: Limit coverage to region of abnormality based on preceding imaging studies
  - MR: Dynamic postcontrast sequence most helpful for localizing nidus

DIFFERENTIAL DIAGNOSIS

Osteomyelitis

- Chronic infection with Brodie abscess
  - Sclerosis & periosteal reaction ± lucent abscess cavity, necrotic bone sequestrum
    - Margins of abscess, sequestrum more likely irregular
      - Peripheral enhancement of abscess
      - Penumbra sign of chronic intraosseous abscess: High T1 signal rim (precontrast) of granulation tissue lining sclerotic margin
    - Cloaca drainage tract may be present
- Acute infection
  - Poorly defined marrow edema, periosteal reaction
    - Intramedullary OO may be confused for early infection by MR if nidus not clearly seen
  - More likely to have osteolysis & fluid collections than OO

Langerhans Cell Histiocytosis

- Lytic lesion with well-defined or permeative margins ± lamellated periosteal reaction
- Sclerotic rim with healing
- Calvarial lesion may have "button sequestrum"

Stress Injury

- Radiolucency & sclerosis more linear & perpendicular to cortex (rather than parallel)

Osteoblastoma

- Histologically similar but larger than OO (> 2-2.5 cm)
- More frequently involves spine
- Constellation of symptoms less classic than OO
- Reactive bone formation uncommon

Osteosarcoma

- Intramedullary location most common
- Combination of aggressive bony destructive features & cloud-like osteoid matrix

PATHOLOGY

General Features

- Benign tumor consisting of osteoblastic mass (nidus) surrounded by zone of reactive sclerosis
  - Zone of sclerosis not integral part of tumor: Represents reversible reactive change; not always present
  - Nidus has limited growth potential
- Prostaglandin E2 elevated 100-1,000x within nidus
  - May be cause of pain & vasodilation

Microscopic Features

- Nidus composed of osteoid tissue or mineralized, immature bone surrounded by fibrovascular stroma
  - Variable amounts of osteoblasts & giant cells resembling osteoclasts
  - Unmyelinated nerve fibers present
- Sclerosis surrounding nidus composed of dense bone

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Local pain, worse at night; ↓ by NSAIDs in < 30 minutes (75%)
    - Up to 5% of patients present without pain
- Other signs/symptoms
  - Local swelling, erythema, point tenderness
  - Symptoms may last weeks to years before presentation
  - Spinal involvement: Painful scoliosis with concavity of curvature toward side of lesion
    - Scoliosis improves/resolves if nidus is resected within 15 months of diagnosis
  - Intraarticular lesion
    - Pain, limp, ↓ range of motion, swelling
  - ↑ urinary excretion of major prostacyclin metabolite (6-keto-PGF1α)
    - Returns to normal after removal of nidus

Demographics

- Age
  - 5-35 years old; 50% between ages 10-20 years
- Gender
  - M:F = 2-4:1
- Epidemiology
  - 4% of primary bone tumors
  - 11-14% of benign bone tumors

Natural History & Prognosis

- No malignant potential
- No growth progression
- Without treatment, lesion ultimately regresses in 6-15 years with symptom resolution
- Long-term bone growth disturbances can result from hyperemia of OO
  - Overgrowth, angular deformities

Treatment

- Medical: NSAIDs
  - Can expedite lesion healing/regression to < 3 years
  - May not be acceptable option due to lifestyle or medical complications
- Surgical resection curative if nidus completely removed
  - Tetracycline with fluorescence or radionuclide labeling used historically for lesion localization at surgery
- CT-guided percutaneous interventions
  - Radiofrequency ablation, laser photocoagulation, cryoablation, trephine/drill excision
- MR-guided high intensity focused ultrasound

DIAGNOSTIC CHECKLIST

Consider

- Image-guided therapy often more successful than surgical resection due to nidus visualization

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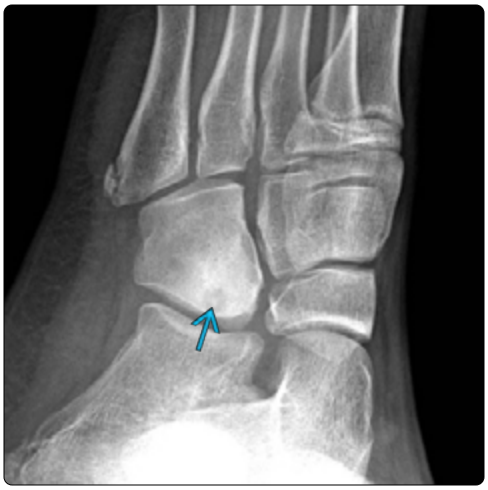
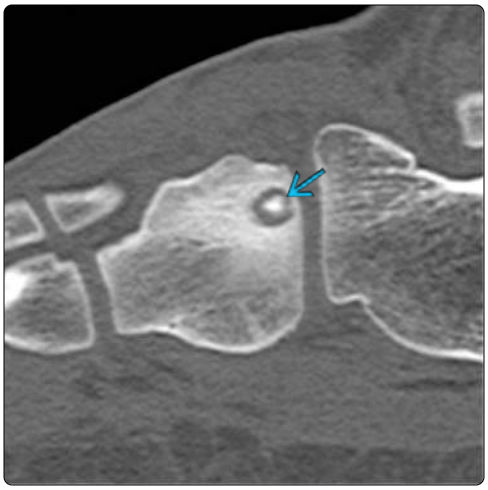
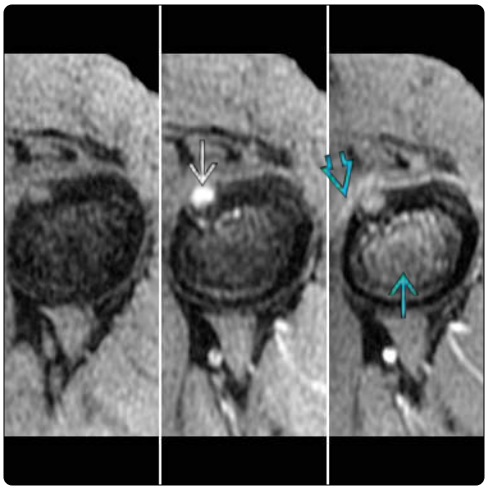
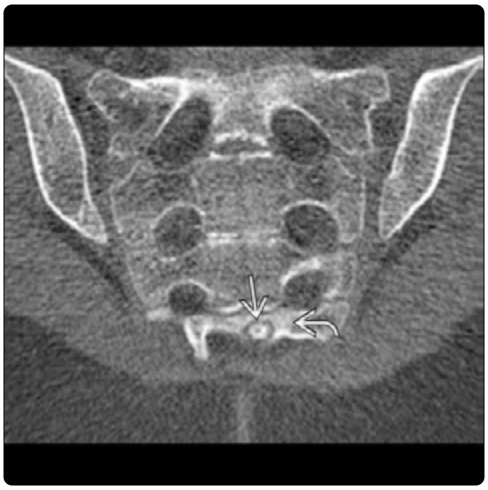


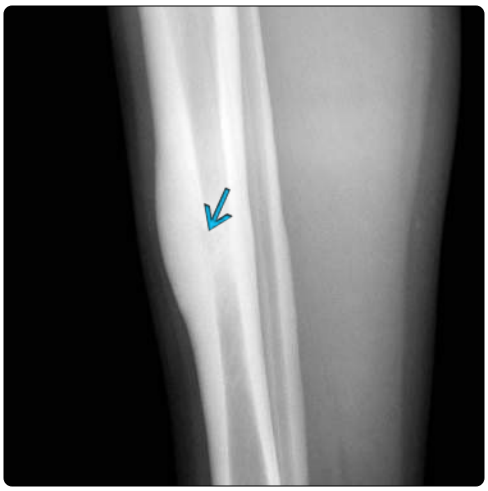


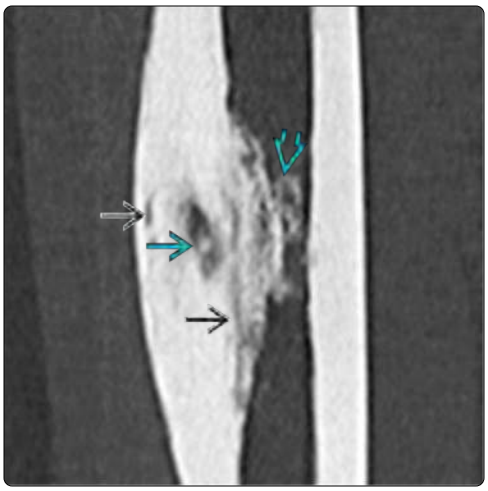
(Left) *Oblique radiograph in a 13-year-old boy with lateral foot pain shows a centrally calcified lucent focus in the posterior cuboid. Note the medullary sclerosis surrounding the lesion.* **(Right)** *Sagittal bone CT in the same patient shows the hypoattenuating round nidus of the cuboid OO with central calcification & surrounding sclerosis.*





(Left) *Axial dynamic T1 FS MR images before (left), 1 minute after (middle), & 8 minutes after (right) contrast administration in an 11-year-old patient with an OO of the humeral cortex show early intense enhancement of the nidus with gradual vague enhancement of the adjacent marrow & overlying tissues.* **(Right)** *Coronal oblique bone CT of the sacrum in an 8-year-old girl with pain shows a round hypoattenuating nidus with central calcification & adjacent sclerosis, consistent with an OO.*





(Left) *Lateral radiograph in a 16-year-old boy with lower leg pain shows moderate tibial cortical thickening & medullary sclerosis surrounding a subtle lucent focus, suggesting an OO.* **(Right)** *Coronal bone CT in the same patient shows the centrally calcified hypoattenuating nidus, typical of an OO. Note the prominent vascular channels amidst the cortical thickening, typical of this highly vascularized lesion. Adjacent medullary sclerosis is also noted.*

Chondroblastoma

KEY FACTS

TERMINOLOGY

- Benign bone tumor with immature cartilage cells (chondroblasts) in heterogeneous matrix

IMAGING

- Vast majority originate in epiphysis (85%), apophysis (12%), or other epiphyseal equivalent (tarsals, carpals, patella)
  - Up to 55% extend to adjacent metaphysis
  - Pure metaphyseal/diaphyseal location uncommon (2%)
- Tubular bones affected most frequently
  - Long bones in 75-80%
- Radiographs
  - Well-circumscribed, lucent epiphyseal lesion
  - Chondroid "ring & arc" Ca^{2+} in 30-50%
  - Solid periosteal reaction of adjacent metaphysis in 50-60%
- T2WI FS MR
  - Heterogeneous internally with foci of low or intermediate T2 signal intensity
    - Distinct from most other cartilage tumors
  - Low signal intensity rim
  - Surrounding marrow, periosteal, & soft tissue edema
  - Reactive joint effusion in up to 50%
  - Secondary aneurysmal bone cyst in 15-33%

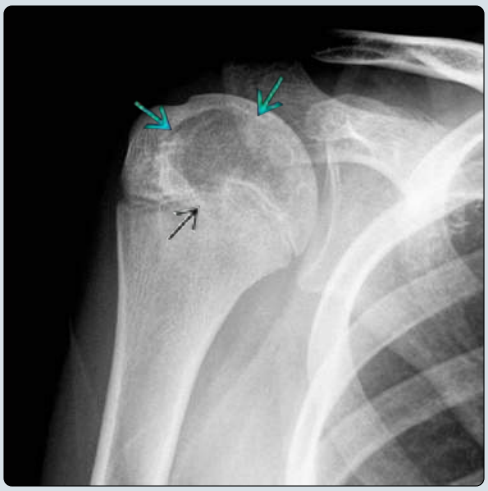
TOP DIFFERENTIAL DIAGNOSES

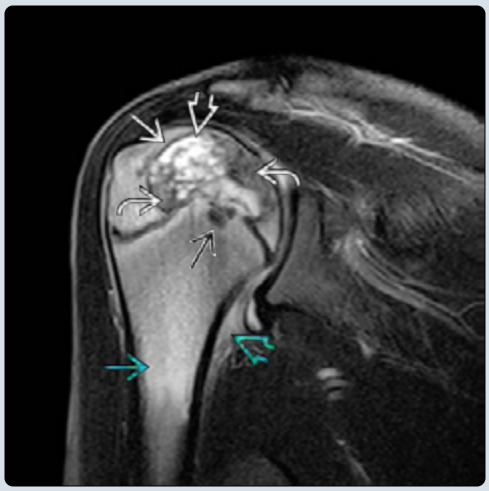
- Osteoid osteoma
- Osteomyelitis
- Langerhans cell histiocytosis
- Metastatic disease
- Giant cell tumor

CLINICAL ISSUES

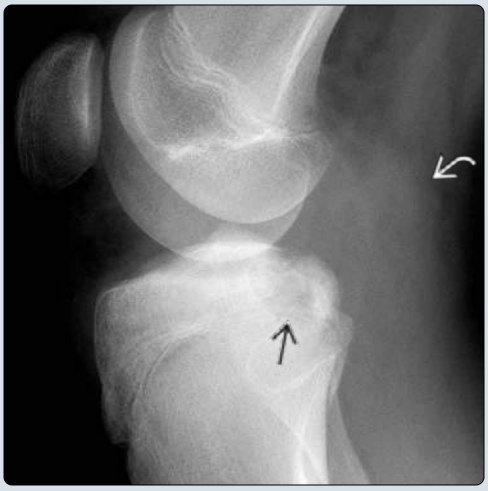
- Presentations: Pain, tenderness, stiffness, swelling, limping
- Peak age: 15-20 years
- Treatment: Surgical curettage ± bone grafting preferred over resection due to adjacent growth plate
  - Recurrence in 5-35%

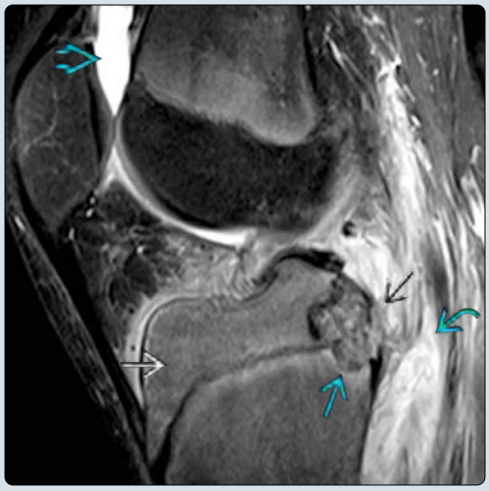
(Left) *AP radiograph of the right shoulder in external rotation shows a well-circumscribed, lucent lesion with a partially sclerotic rim originating in the proximal humeral epiphysis. A small focus of physeal breach is suggested.* **(Right)** *Coronal T2 FS MR in the same patient shows foci of increased & decreased T2 signal intensity in the epiphyseal lesion with a low signal intensity rim, typical of a chondroblastoma. There is a breach of the physis with edema of the adjacent marrow & soft tissues.*





(Left) *Lateral knee radiograph in a 13-year-old girl with 1 year of pain shows a lobular lucent lesion in the posterior tibial epiphysis. There is fullness & poor definition of the surrounding soft tissues, suggesting edema.* **(Right)** *Sagittal T2 FS MR in the same patient shows heterogeneous low signal intensity in the lesion with metaphyseal extension & cortical breach. There is surrounding marrow & soft tissue edema as well as a joint effusion. Chondroblastoma was confirmed at surgery.*





TERMINOLOGY

Definitions

- Benign bone tumor with immature cartilage cells (chondroblasts) in heterogeneous matrix

IMAGING

General Features

- Best diagnostic clue
  - Well-circumscribed, lucent epiphyseal lesion in teenager
- Location
  - Majority originate in epiphysis (85%), apophysis (12%), or other epiphyseal equivalent (tarsals, carpals, patella)
    - Up to 55% extend to metaphysis
    - Pure metaphyseal/diaphyseal location rare (2%)
  - Tubular bones affected most frequently
    - Long bones in 75-80%
      - Proximal tibia, distal & proximal femur; proximal humerus most frequent
    - Foot involved in 5-16%
      - Talus, calcaneus ~ 40% each
  - Rare cortical disruption with extraosseous or intraarticular extension

Radiographic Findings

- Radiography
  - Well-circumscribed, lucent epiphyseal lesion
  - Mildly lobulated with sclerotic rim
  - Chondroid ring & arc Ca^{2+} in 30-50%
  - Solid periosteal reaction of adjacent metaphysis in 50-60%
  - Bubbly expansile remodeling if aneurysmal bone cyst (ABC) develops

CT Findings

- Bone CT
  - ± chondroid Ca^{2+} (~ 44%)
  - Cortical disruption (19%)
  - Periosteal reaction of adjacent metaphysis

MR Findings

- T2WI FS
  - Heterogeneous internally with foci of low or intermediate T2 signal intensity
    - Ca^{2+} vs. hemosiderin vs. cellular regions
    - Distinct from most other high T2 signal intensity cartilage tumors
  - Low signal intensity rim
  - Surrounding marrow, periosteal, & soft tissue edema
  - Reactive joint effusion in up to 50%
  - Multiloculated component containing fluid-fluid levels due to secondary ABC (15-33%)
- T1WI C+ FS
  - Variable enhancement of lesion internally
  - Enhancement of edematous marrow & soft tissues

DIFFERENTIAL DIAGNOSIS

Osteoid Osteoma

- Cortex of metaphysis/diaphysis most common (but can be epiphyseal)
- Adjacent edema & cortical thickening
- Lucent nidus usually small

Osteomyelitis

- Metaphyseal origin most common
- Aggressive bony features, usually without well-defined lesion
- Fluid collections often present

Langerhans Cell Histiocytosis

- Cortical destruction with soft tissue mass
  - Often uniform high T2 signal intensity & enhancement
  - May have low T2 signal intensity rim
- Favors metadiaphysis or flat bones

Metastatic Disease

- Often multifocal with aggressive features & enhancement

Giant Cell Tumor

- Subchondral epiphyseal lesion without sclerotic rim
- Uncommon in skeletally immature patients

PATHOLOGY

General Features

- Closely packed immature cartilage cells (chondroblasts) with scattered mature cartilage & giant cells
- Intercellular Ca^{2+} (chicken wire) in 60%
- Secondary ABC in up to 33%

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain, tenderness, stiffness, swelling, limping

Demographics

- Age
  - Peak: 15-20 years; range: 3-85 years
- Gender
  - M:F = 1.6-2.7

Treatment

- Surgical curettage ± bone grafting preferred over resection due to adjacent growth plate
  - Recurrence in 5-35%
    - More likely with skeletal immaturity

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Osteochondroma

KEY FACTS

TERMINOLOGY

- Common benign, developmental bony surface lesion resulting from displaced growth cartilage; not neoplastic

IMAGING

- Lobulated sessile or pedunculated bony protuberance
  - Demonstrates flowing corticomedullary continuity with underlying ("parent") bone
  - Covered by cap of hyaline cartilage
- Radiographs usually diagnostic
- MR typically reserved for pain/complications or if diagnosis unclear
  - Fracture (± radiographic visibility) may occur at stalk
  - Mass effect may lead to
    - Friction/irritation of overlying soft tissues ± bursa formation
    - Compression neuritis leading to denervation
    - Vascular compression or rarely occlusion, pseudoaneurysm

TOP DIFFERENTIAL DIAGNOSES

- Osteosarcoma (parosteal)
- Myositis ossificans
- Periosteal chondroma

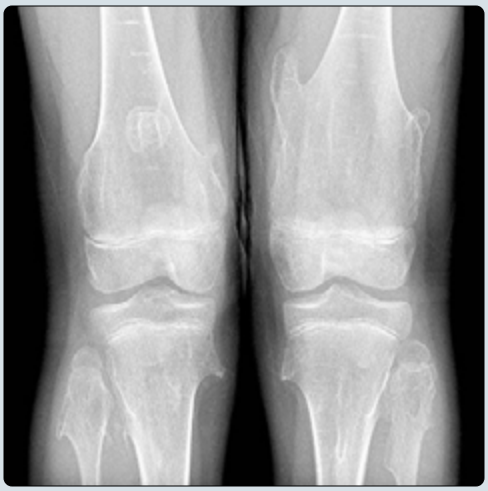
PATHOLOGY

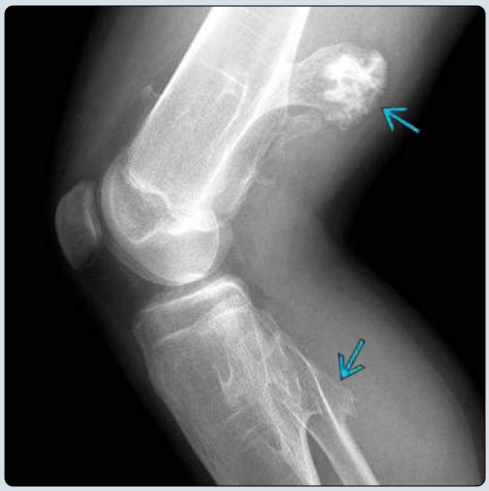
- Solitary > > multiple
  - Hereditary multiple exostoses (HME) patients
    - Develop characteristic growth disturbances
    - Have 1-5% lifetime risk of malignant degeneration vs. < 1% risk for solitary osteochondroma (OC)
      - OC cartilage cap thickness > 1-2 cm in skeletally mature patient more likely to harbor malignancy

CLINICAL ISSUES

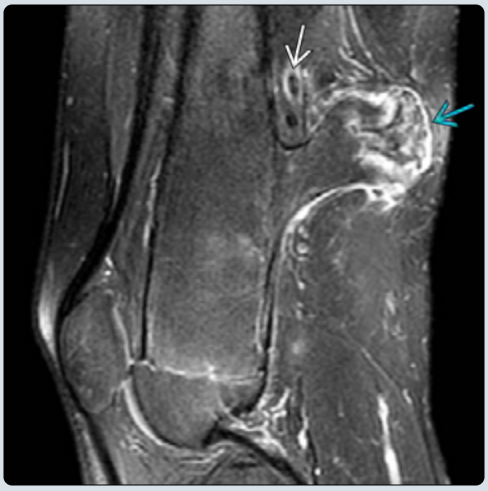
- Most commonly found in patients age 10-35 years
- Typically presents as painless hard mass
- Lesion growth should cease at skeletal maturity
  - Further ↑ suggests malignant degeneration

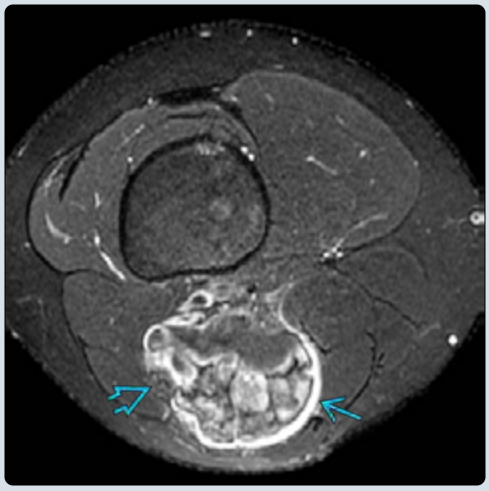
(Left) *AP radiograph of the lower extremities in a 13 year old with hereditary multiple exostoses (HME) shows numerous osteochondromas (OCs) about the knees, ranging in morphology from pedunculated to sessile. Note the broadened metadiaphyses typical of HME.* **(Right)** *Subsequent lateral radiograph of the right knee in the same patient shows that the pedunculated OCs ⇨ point away from the joint. Note the characteristic corticomedullary continuity of the OCs with the underlying bones.*





(Left) *Sagittal T2 FS MR in the same patient shows the pedunculated distal femoral OC ⇨ in close proximity to the popliteal vessels ➡. OCs may compress or injure adjacent vessels, leading to pseudoaneurysm.* **(Right)** *Axial T2 FS MR in the same patient shows a thin, bright cartilage cap ⇨ overlying the heterogeneously mineralized cartilage, typical of OCs. The sciatic nerve ⇨ is displaced by the OC.*





TERMINOLOGY

Abbreviations

- Osteochondroma (OC)

Synonyms

- Exostosis

Definitions

- Benign, developmental bony surface lesion arising from displaced growth cartilage
  - Not true neoplasm
- Maintains corticomedullary continuity with underlying bone
- Covered by hyaline cartilage cap

IMAGING

General Features

- Best diagnostic clue
  - Lobulated sessile or pedunculated bony protuberance demonstrating flowing corticomedullary continuity with underlying bone
- Location
  - Any bone that develops by endochondral ossification
    - Distal femur > proximal femur, proximal tibia, humerus
      - Metaphysis/metadiaphysis > diaphysis > epiphysis
    - Hands, feet, scapula, spine, pelvis, skull base < 10% each
  - > 80% solitary, < 20% multiple
- Morphology
  - Spectrum between extremes
    - Sessile: Broad base with underlying bone
    - Pedunculated: Narrow or bulbous mass with thin stalk of attachment to bone
      - Usually points away from joint due to traction

Radiographic Findings

- Sessile or pedunculated bony lesion arising from surface of bone
- Corticomedullary continuity with underlying bone
- Chondroid calcifications may be seen in cartilage cap
- U-shaped rim of sclerosis if viewed en face

CT Findings

- CTA
  - Useful for assessing vascular complications
    - Vessel compression, occlusion, arteriovenous fistula, or pseudoaneurysm may result from mass effect by adjacent OC

MR Findings

- T1WI
  - Continuity of dark cortex & bright fatty marrow signal between lesion & underlying bone
- T2WI FS
  - Marrow & cortex signal similar to "parent" bone
    - Marrow edema due to contusion or fracture
  - Bright cartilage cap overlying ossified lesion
    - Size of cartilage cap (with regard to malignant potential) essentially irrelevant in skeletally immature patient
      - Size conferring ↑ risk of malignancy in adults debated: > 1 cm vs. > 2 cm
  - Very sensitive for soft tissue complications
    - Friction/compression of overlying muscle & fat ± bursa formation
    - Nerve compression
    - Vascular compression & pseudoaneurysm formation
      - Pseudoaneurysm very heterogeneous due to turbulent internal flow & layers of thrombus; pulsation artifact in phase-encoding direction
    - Edema from fracture through stalk
- T1WI C+ FS
  - Enhancement of growth plate at osteocartilaginous junction
  - Variable enhancement of remaining cartilage cap
    - May show lobular septal & rim enhancement

Ultrasonographic Findings

- Can confirm cortical continuity, cartilage cap
- May be useful to evaluate surrounding soft tissues
  - Relationship of OC to nerve or vessel
    - Color Doppler to look for vascular complication
  - Bursa formation

Nuclear Medicine Findings

- Bone scan
  - ↑ activity at growth plate of cartilage cap
  - ↓ activity with skeletal maturity
  - ↑ activity beyond skeletal maturity: Concerning for malignancy

Imaging Recommendations

- Best imaging tool
  - Radiographs usually diagnostic
  - MR excellent for soft tissue complications
- Protocol advice
  - T2 FS/STIR most sensitive MR sequence for complications
  - Contrast-enhanced MR rarely needed

DIFFERENTIAL DIAGNOSIS

Osteosarcoma (Parosteal)

- Most common surface osteosarcoma
- Ossified soft tissue mass arising from outer periosteum
- May have broad base vs. cleavage plane with bone
  - Lacks "flowing" corticomedullary continuity of OC
- Most common at posterior distal femoral metaphysis
- Peak incidence: 20-40 years of age

Myositis Ossificans

- Initially, noncalcified soft tissue mass overlying bone
- Develops peripheral calcification within weeks, progressing to center
- Trauma history often (but not always) present

Periosteal/Juxtacortical Chondroma

- Uncommon cartilaginous surface lesion
- Partially interrupted, layered or scalloped cortex deep to cartilage focus (rather than corticomedullary continuity)

PATHOLOGY

General Features

- Etiology
  - Displaced physeal cartilage herniates through periosteal bone cuff
  - Endochondral ossification of this cartilage yields growing bony protuberance
  - Growth of OC expected to cease at skeletal maturity
    - Continued growth or pain concerning for malignant transformation
  - ↑ OC incidence with prior radiation (up to 24% develop exostoses)
    - Mean latency of 5-12 years
- Associated abnormalities
  - Hereditary multiple exostoses (HME)
    - Numerous OCs with resulting growth disturbances
      - Valgus tibiotalar tilt
      - Limb length discrepancy
      - Pseudo-Madelung deformity
      - Coxa valga
    - 1:50,000-1:100,000
    - Autosomal dominant with incomplete penetrance
      - Mutations of exostosin tumor suppressor genes on chromosomes 8 (*EXT1*), 11 (*EXT2*), & 19 (*EXT3*)
    - ↑ likelihood of malignant transformation to chondrosarcoma
      - 1-5%; typically after skeletal maturity
  - Dysplasia epiphysealis hemimelica (Trevor disease)
    - Epiphyseal (or equivalent) OCs
    - Multiple bones of single extremity often involved on either medial or lateral side
    - Lower extremity (particularly foot/ankle) much more commonly affected than upper
    - Mechanical symptoms, limb deformity common
  - Subungual exostosis
    - Lesion deep to nailbed
  - Bizarre parosteal osteochondromatous proliferation (BPOP or Nora lesion)
    - Phalanges involved in 70% of cases
    - Ossified pedunculated lesion attached to (but not flowing from) cortex

Gross Pathologic & Surgical Features

- Lobulated cartilage cap with shiny bluish-gray surface
  - Thickness typically on order of few mm but up to 2 cm acceptable
  - > 1-2 cm in skeletally mature patient more likely to harbor malignancy

Microscopic Features

- Perichondrium covering hyaline cartilage cap continuous with parent bone periosteum
- Cap organization similar to normal growth cartilage

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless mass with deformity
- Other signs/symptoms
  - Pain secondary to
    - Soft tissue friction or bursitis
    - Nerve or vessel compression
    - Fracture of exostosis stalk
    - Malignant transformation (uncommon)
      - Chondrosarcoma in vast majority
  - Spinal cord compression from vertebral lesion
  - Hemo-/pneumothorax from rib lesion

Demographics

- Age
  - 10-35 years of age for solitary lesion
  - Most HME patients diagnosed by age 10
- Epidemiology
  - Most common bone "tumor"
    - Solitary OC in 1-2% of population
    - Up to 15% of all bone tumors
    - Up to 50% of benign bone tumors

Natural History & Prognosis

- Lesion growth should cease at skeletal maturity
  - Spontaneous regression rarely reported
- Continued growth suggests malignant transformation
  - Extremely rare in skeletally immature
  - Solitary exostosis lifetime risk: < 1%
  - HME lifetime risk: 1-5%

Treatment

- Surgical resection for symptomatic lesions or lesions growing beyond skeletal maturity
  - Entire cartilage cap & perichondrium must be removed to prevent recurrence
- Additional procedures often required in HME to correct deformities

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Demonstration of corticomedullary continuity crucial

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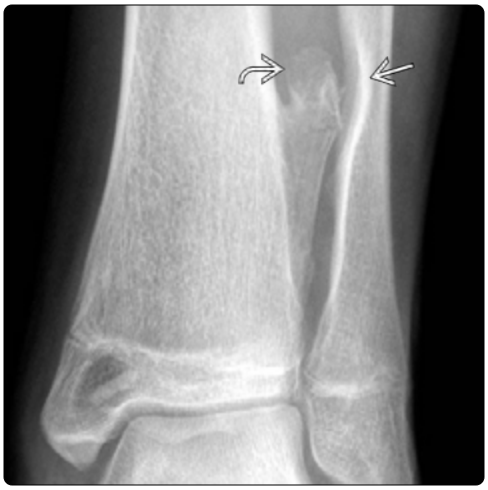
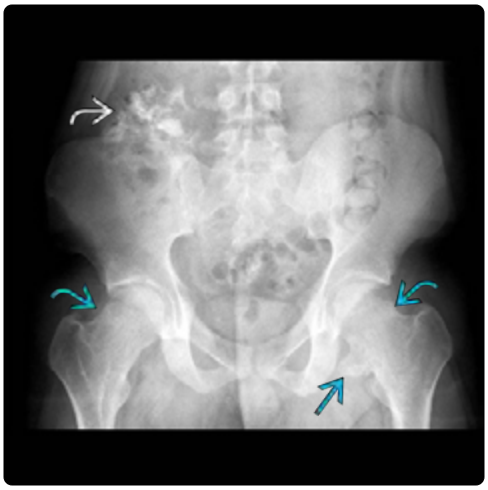
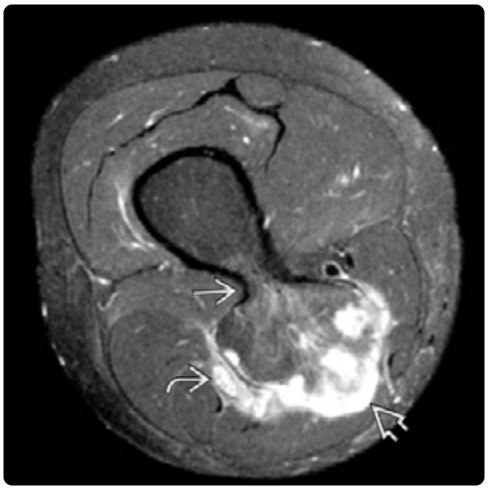
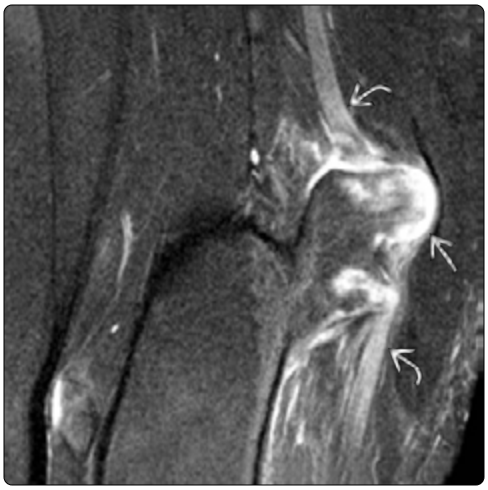


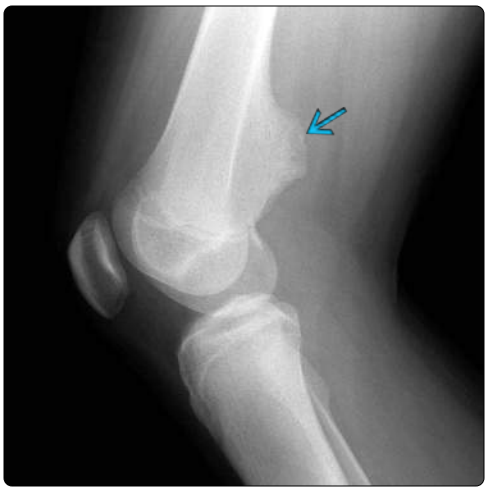


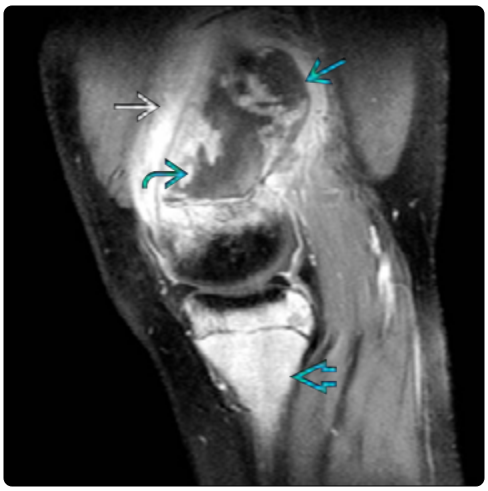
(Left) *Oblique radiograph of the left ankle in a teenager shows the local pressure effect of a tibial OC causing chronic bony remodeling of the adjacent fibular diaphysis.* **(Right)** *AP radiograph of the pelvis in a 17-year-old HME patient shows a large, partially mineralized, pedunculated OC arising from the right iliac crest. There is an additional left femoral neck OC. Note the broadened femoral necks typical of HME.*





(Left) *Axial T2 FS MR of the right thigh in a 14-year-old girl shows a pedunculated OC with a thin stalk arising from the posteromedial femur. A thin high signal cartilage cap is noted. The sciatic nerve is displaced laterally & shows abnormal high T2 signal internally.* **(Right)** *Sagittal T2 FS MR of the same patient shows displacement & abnormal fluid signal of the sciatic nerve due to mass effect from the adjacent OC.*





(Left) *Lateral radiograph of the right knee in a patient with recently diagnosed leukemia & subsequent onset of acute knee pain shows a sessile OC of the distal femur.* **(Right)** *Sagittal T1 C+ FS MR in the same patient shows homogeneous, diffusely abnormal marrow enhancement of the proximal tibia due to leukemic infiltration. Heterogeneous enhancement of the distal femoral marrow (with overlying soft tissue edema) was due to superimposed acute infarction with extension into the OC.*

KEY FACTS

TERMINOLOGY

- Limb salvage procedure: Strategic coordination of tumor resection & limb reconstruction to preserve function without compromising overall survival
- 2 main types of reconstruction: Endoprosthetic & biological
  - Growing endoprosthesis used in pediatric patients when > 30 mm of growth remains
- General complications include infection & tumor recurrence
  - Endoprosthesis complications: Hardware loosening or fracture, periprosthetic fracture
  - Biological complications: Nonunion

IMAGING

- Careful evaluation of evolving postoperative appearance (in conjunction with prior exams) for
  - Hardware or periprosthetic fracture
  - Altered alignment of hardware
  - Progressive radiolucency about hardware
  - Absence of progressive bony union
  - Increasing soft tissue swelling
- Understanding type of reconstruction & expected complications aids in accurate interpretation

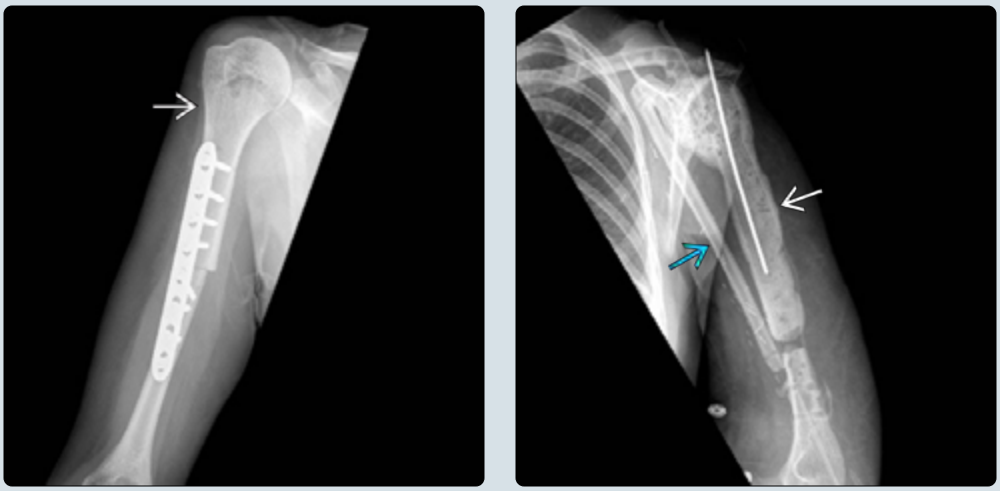
CLINICAL ISSUES

- Complications may be asymptomatic or present with prosthetic locking, extremity pain, joint instability, joint effusion & soft tissue swelling, warmth, erythema, or draining sinus tract
- Treatment differs with regard to acute or chronic infection of endoprosthesis

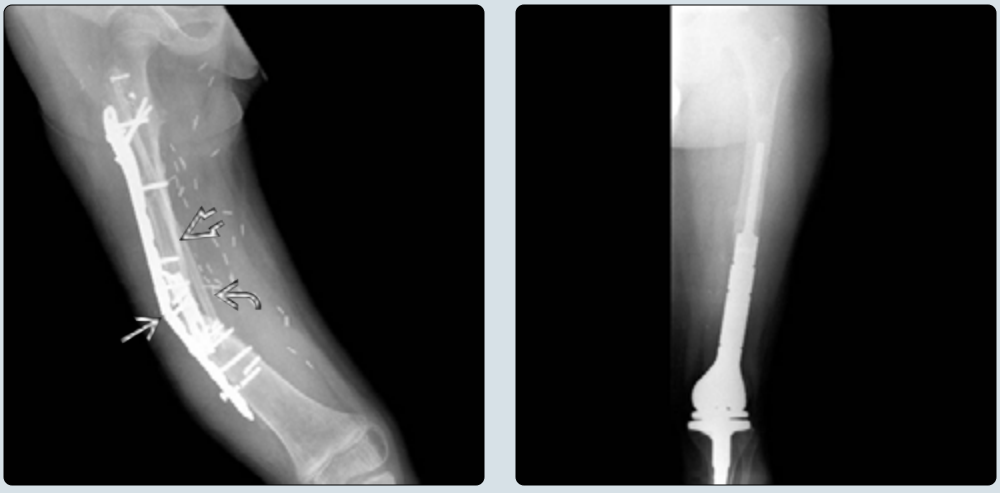
DIAGNOSTIC CHECKLIST

- CT can help evaluate degree of bone union in biologic reconstruction
- PET/CT &/or MR often performed in routine oncologic follow-up → may provide useful adjunct in interpreting limb radiographs
- Metal artifact reduction techniques advised for any postoperative CT/MR

(Left) *AP radiograph in a 24-year-old woman shows an osteoarticular allograft ➡ at the right proximal humerus with a compression plate & associated screws transfixing the osteotomy site between the proximal allograft & distal host bone.* **(Right)** *AP radiograph of the left humerus in a 15-year-old girl shows an antibiotic-impregnated cement spacer ➡ & associated K-wire put in place of an infected osteoarticular allograft. The fibular autograft ⇨ remains in place.*



(Left) *AP radiograph of a 7-year-old girl shows a biological reconstruction with a free vascularized fibular autograft ➡ alongside an intercalary allograft ➡ at the right femur. There is a fracture of the fixation plate ➡ at a site of nonunion distally at the host-allograft junction. The nonunion is not well seen due to the overlying hardware.* **(Right)** *AP radiograph of the left femur in a 16-year-old boy shows the typical appearance of a modular oncologic total knee arthroplasty with cement.*



TERMINOLOGY

Definitions

- Limb salvage surgery: Tumor resection & reconstruction techniques optimized to preserve adequate function of involved limb without compromising overall survival
- Oncologic hardware relates to 2 main types of reconstruction
  - Endoprosthetic reconstruction: Conventional vs. growing
    - Growing endoprosthesis used in pediatric patients when > 30 mm of growth remains
  - Biological reconstruction: Use of native or donor tissue

IMAGING

Radiographic Findings

- Endoprosthesis: Conventional vs. growing
- Biological
  - Intercalary or osteoarticular allograft
  - Allograft-prosthesis composite
  - ± vascularized fibula autograft
    - May be used within medullary canal of allograft
- Complications of endoprosthetic device
  - Implant fracture
  - Periprosthetic fracture
  - Aseptic loosening
- Complications of biological reconstruction
  - Osseous nonunion: Lack of new bone formation & ↓ lucency at host-allograft junction over time
- General complications: Infection or recurrent malignancy shows irregular bone destruction, aggressive periosteal reaction, soft tissue swelling/fluid collection/mass

CT Findings

- Bone CT
  - May occasionally be useful to determine degree of bone defects related to suspected hardware loosening
  - Used to assess degree of bone union in biological reconstruction
- Metal artifact reduction techniques include dual energy CT & iterative reconstruction

Ultrasonographic Findings

- Less artifact overlying metal (though metal prevents deep visualization)
- May be useful to evaluate postoperative fluid collections or tumor recurrence about hardware

Nuclear Medicine Findings

- No studies specifically aimed at oncologic prostheses regarding infection & aseptic loosening
- PET/CT: Useful to evaluate for tumor recurrence
  - Does not reliably distinguish aseptic loosening from infection in adult studies
- Bone scan: Useful for screening adult patients with problematic knee & hip prostheses
  - Nonspecific regarding differentiation of aseptic loosening & infection
- Combined labeled leukocyte/Tc-99m sulfur colloid scintigraphy: ↑ activity on leukocyte imaging with lack of uptake on marrow imaging suggests infection in adult patients with hip & knee prostheses

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Extremity pain, joint instability, joint effusion, prosthetic locking, warmth, erythema, draining sinus tract

Natural History & Prognosis

- Endoprosthetic reconstruction
  - Good medium- to long-term results regarding implant survival in 1 study
    - Failure rate of 34% at mean follow-up of 15 years due to mechanical causes (51%), infection (33%), local recurrence (16%)
    - Incidence of infection greater in children than adults (due to repeated lengthening procedures)
- Biological reconstruction
  - Combined failure rates due to nonunion, fracture, & infection: 15-34%
  - Osseous union may take months to years
    - ↑ nonunion rates with adjuvant chemotherapy
  - Free vascularized bone grafts or nonvascularized bone grafts can be used to address some complications
  - Vascularized fibula graft may be used within medullary canal of allograft during initial reconstruction with aim to ↓ complications

Treatment

- Infection
  - Acute infection of endoprosthesis
    - Debridement & intensive antibiotic administration: ~ 30% success rate
  - Chronic infection of endoprosthesis
    - Low rate of eradication with local treatment alone
      - i.e., surgical debridement, arthroscopic washout, antibiotics, or antibiotic-laden beads or cement
    - 2-stage revision standard of care for total joint arthroplasties
      - Remove old prosthesis & replace with antibiotic-impregnated spacer for minimum of 6 weeks
      - New endoprosthesis replaces spacer when laboratory findings indicate infection eradicated (i.e., negative culture of periprosthetic aspirate)
      - ~ 70% success rate; some patients still need amputation
- Mechanical failure: Revision

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KEY FACTS

TERMINOLOGY

- Developmental dysplasia of hip (DDH): Spectrum of hip abnormalities including dysplastic acetabulum & femoral head malpositioning
- Much more common in girls, breech positioning, oligohydramnios, Caucasians
- Ligamentous laxity contributes to DDH

IMAGING

- Ultrasound is modality of choice for infants 1-4 months old
- Radiographs necessary after 4-5 months
  - Proximal femoral epiphysis ossifies, blocks ultrasound beam, limits evaluation

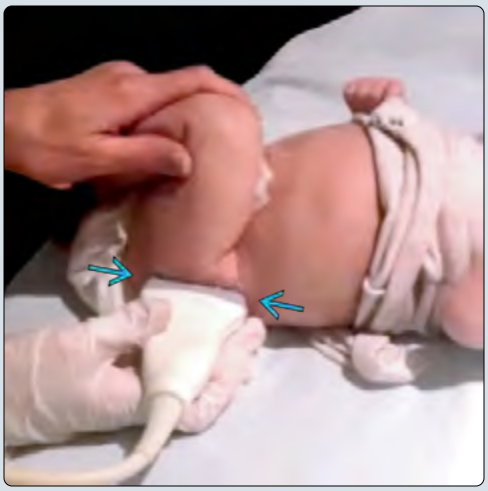
PATHOLOGY

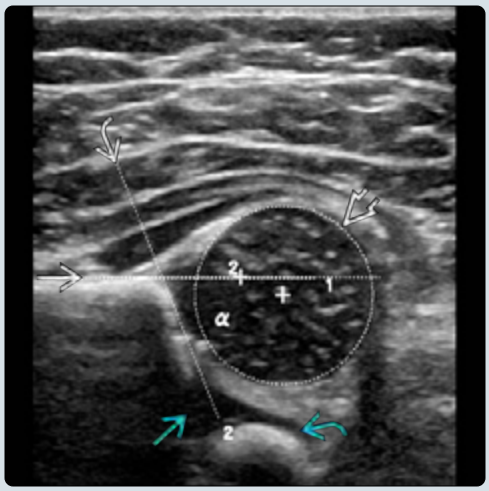
- Normal
  - α angle ≥ 60°, coverage ≥ 50%, no instability
- Immature hip (applies only to infants < 3 months of age)
  - α angle 50-59°, coverage 45-50%, no instability
  - Small risk of delayed DDH; follow-up recommended to confirm normal development
- Mild hip dysplasia
  - α angle 50-59°, coverage 40-50%
  - May observe, repeat US in 1 month, especially if ≤ 2 months; older infants usually treated with harness
- Moderate hip dysplasia
  - α angle ≤ 50°, coverage ≤ 40%, any instability
  - Treated with harness; repeat US q4 weeks until normal
- Severe hip dysplasia
  - Grossly dysplastic acetabulum, dislocated hip
  - No improvement in harness by 4 weeks → operative management

CLINICAL ISSUES

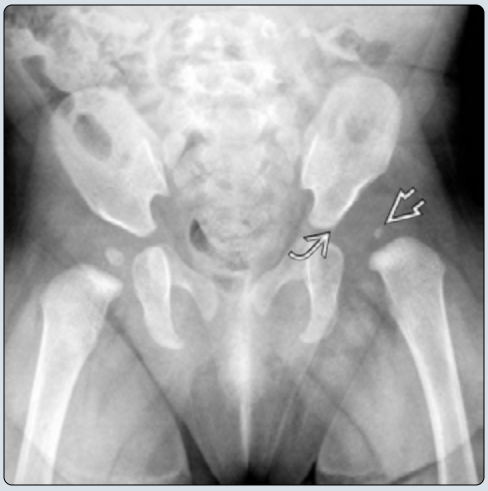
- Treat with Pavlik harness to flex, abduct, & externally rotate hips
- Frequently repeat US to evaluate progression
- Occasionally surgical hip reduction & casting are required

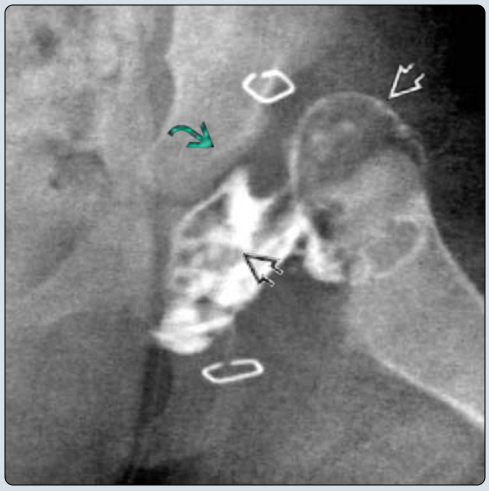
(Left) *The US transducer is placed over the lateral hip with slight posterior obliquity to obtain a coronal flexed image. Note the use of 2 hands (& the foot pedal to save images).* **(Right)** *Coronal flexed US in the same patient shows the unossified femoral head, the straight segment of the iliac bone, & a line drawn along the acetabular roof. The α angle is measured between these lines & should be ≥ 60°. The iliac line should cover the femoral head by ≥ 50%. Note the triradiate cartilage & ischium.*





(Left) *Frontal radiograph in a 6-month-old boy with a left hip clunk reveals severe left developmental dysplasia of hip. The acetabular roof is quite steep, & the ossified femoral head is small with superolateral dislocation.* **(Right)** *An intraoperative arthrogram shows contrast outlining a dysplastic, mostly unossified left femoral head. Note that the acetabular roof is quite steep. The femur is dislocated from the severely dysplastic & shallow joint space. This child required an osteotomy.*





TERMINOLOGY

Abbreviations

- Developmental dysplasia of hip (DDH)

Synonyms

- Congenital dislocation of hip (archaic)

Definitions

- Spectrum of progressive hip abnormalities developing during infancy & resulting in acetabular dysplasia with femoral head malpositioning
  - Ranges from mild subluxation → dislocatable hip → fixed hip dislocation

IMAGING

General Features

- Best diagnostic clue
  - Abnormal position & delayed ossification of femoral head in setting of abnormally steep & shallow acetabulum
- Location
  - Femur displaced superolaterally from acetabulum

Ultrasonographic Findings

- Grayscale ultrasound
  - Sonography directly visualizes cartilaginous & osseous components of hip
  - Static (anatomic) & stress (dynamic) imaging evaluates
    - Acetabular morphology
    - α angle (normal ≥ 60°)
    - Coverage of femoral head (normal ≥ 50%)
    - Dynamic subluxation during stress maneuvers
- Color Doppler
  - May be helpful in assessing femoral head perfusion, especially in patients abducted in Pavlik harness
  - Not generally part of standard hip ultrasound exam

Radiographic Findings

- Radiography
  - **Hilgenreiner (horizontal) line**
    - Drawn through bilateral triradiate cartilages
  - **Perkin (perpendicular) line**
    - Drawn from superolateral rim of acetabulum through line of Hilgenreiner
    - Should intersect medial femoral metaphysis, or femoral head should project in inferior medial quadrant
      - Femoral head usually ossifies by 2-3 months
  - **Acetabular angle**
    - Angle between line drawn along acetabular roof & Hilgenreiner line
      - Mathematically complementary to α angle of US
    - Decreases as hip matures
      - Normal < 30° in 1st year of life
      - Normal < 24° in 2nd year of life
  - **Shenton line**
    - Drawn along superior aspect of obturator foramen & medial of femoral neck
    - Normally represents contiguous arc
    - If noncontiguous, suggests hip subluxation (DDH)

CT Findings

- NECT
  - Limited CT scans sometimes performed to confirm hip position after open surgical reduction & spica casting

Nonvascular Interventions

- Arthrogram
  - Occasionally performed intraoperatively

Imaging Recommendations

- Best imaging tool
  - US modality of choice for infants 1-4 months of age
    - Screening hip US not recommended < 4 weeks of age due to presence of physiologic laxity
    - However, US should be performed if clinical exam suggests dislocation or significant instability
  - Radiographs necessary after 4-5 months
    - Proximal femoral epiphysis ossifies, blocks ultrasound beam, limits evaluation
- Protocol advice
  - Important to examine both hips
  - Careful US technique mandatory; use high-frequency transducer (7-9 MHz), MSK settings, foot pedal
  - **Coronal view**
    - Infant at rest, in supine or lateral decubitus position
    - Transducer parallel to lateral aspect of hip; may need to angle transducer 10-15° posteriorly into oblique coronal plane
    - Normal: α angle ≥ 60°, ≥ 50% femoral head coverage
    - Image should show
      - Straight segment of iliac wing
      - Center of femoral head (maximal diameter)
      - Deepest part of acetabulum (should see triradiate cartilage & ischium posteriorly)
  - **Transverse flexion view**
    - Infant's hip held in 90° flexion
    - Place transducer over posterolateral hip in anatomic transverse plane
    - Image should show
      - Femoral metaphysis, femoral head, ischium (posterior acetabulum)
      - Femoral head should rest on ischium
      - Unstable if head slips or pistons beyond ischium on stress maneuvers
  - **Posterior lip view**
    - Stay in coronal flexed position, slide transducer posteriorly, remove 10-15° obliquity
    - Image should show
      - Ilium, ischium, & intervening triradiate cartilage: Posterior acetabular lip
      - Should **not** see femoral head
    - If normal, gently push thigh posterior to check stability
    - If subluxed, gently pull to determine if head can be easily reduced

DIFFERENTIAL DIAGNOSIS

Cerebral Palsy, Congenital Coxa Valga, Neuromuscular Disease

- Abnormal muscular tension causes subluxation/abnormal alignment

- Other radiographic findings of neuromuscular disease often present
  - Gracile long bones, muscle wasting, coxa valga, "windswept pelvis"

Septic Arthritis

- Acute: Joint fluid may displace femoral head
  - Clinical/laboratory findings of infection present
- Chronic: Abnormally small & misshapen femoral head & acetabulum

Proximal Focal Femoral Deficiency

- Rare congenital anomaly characterized by varying degrees of proximal femoral hypoplasia/aplasia; acetabulum may be normal

PATHOLOGY

General Features

- Etiology
  - Cartilaginous components on both sides of hip joint must be closely apposed to develop properly
  - DDH likely multifactorial, includes ligamentous laxity
    - Effect of maternal hormones (especially relaxin)
    - Intrinsic acetabular/femoral head deficiency
    - Reduced in utero space
      - Breech position (extreme hip flexion, knee extension)
      - Oligohydramnios
      - 1st-born child

Staging, Grading, & Classification

- Graf staging based on single, static coronal image
- Most recommendations now include description of femoral head coverage & require stress imaging
- Simpler US classification includes normal, immature, & abnormal (with mild, moderate, severe modifier)
- **Normal**
  - Sometimes called Graf type I
  - α angle ≥ 60°, coverage ≥ 50%
  - Requires no treatment
- **Immature hip**
  - Sometimes called Graf type IIa
  - Applies only to infants < 3 months of age
  - α angle 50-59°, coverage 45-50%
  - Small risk of delayed DDH
    - Recommend follow-up to confirm normal development
    - Requires no treatment
- **Mild hip dysplasia**
  - α angle 50-59°, coverage 40-50%
  - May be observed by orthopedists with repeat US in 1 month, especially if < 2 months of age
  - Older infants usually treated with harness
- **Moderate hip dysplasia**
  - α angle ≤ 50°, coverage ≤ 40%
  - Unstable on stress imaging or clinical exam
  - Treated with Pavlik harness with repeat US q4 weeks until normal
- **Severe hip dysplasia**
  - Grossly dysplastic acetabulum with dislocated hip
  - Can treat with Pavlik harness for no more than 4 weeks
    - If no improvement in Pavlik harness by 4 weeks, continue to operative management

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymmetric skin or gluteal folds
  - Leg length discrepancy
  - Palpable click or clunk during stress maneuvers: Ortolani & Barlow
- Other signs/symptoms
  - Delayed ambulation or limp in toddlers

Demographics

- Gender
  - M:F = 1:5-8
- Epidemiology
  - Incidence: 2-20 per 1,000 births

Natural History & Prognosis

- Mild hip dysplasia may resolve spontaneously & never cause clinical problems
  - 60-80% of abnormalities found by clinical exam resolve spontaneously
  - 90% of abnormalities found by US resolve spontaneously
- Moderate or severe dysplasia can cause long-term disability, limb shortening, decreased range of motion, degenerative change, avascular necrosis
  - Some patients may eventually require hip replacement
- Excellent prognosis when diagnosed & treated early with Pavlik harness
- Delayed diagnosis or treatment can result in irreversible dysplasia requiring iliac osteotomy/shelving procedure

Treatment

- Pavlik harness to flex, abduct, & externally rotate hips
  - Increases femoral head engagement with acetabulum
  - Overabduction can cause femoral head ischemia
- Occasionally surgical hip reduction & casting required
  - Salter osteotomy, Steele triple osteotomy, Pemberton or Chiari procedure, femoral osteotomy

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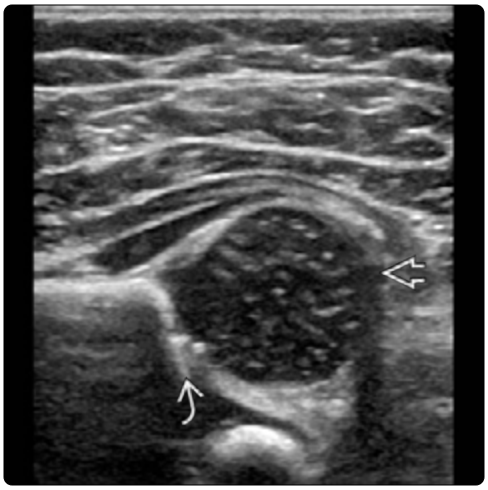
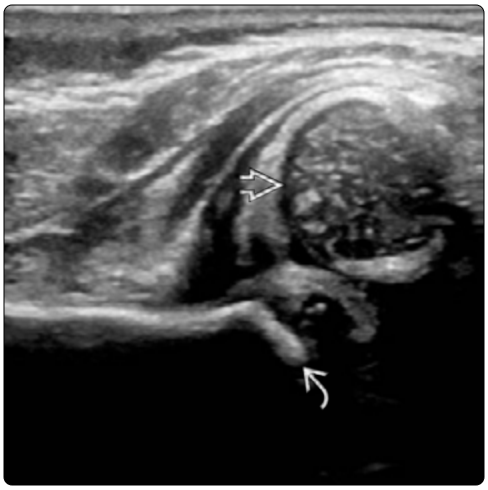
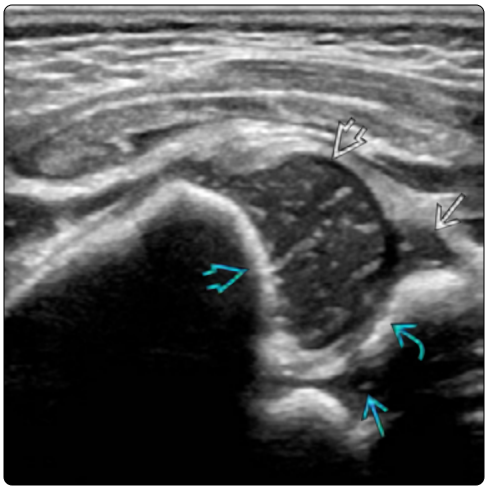
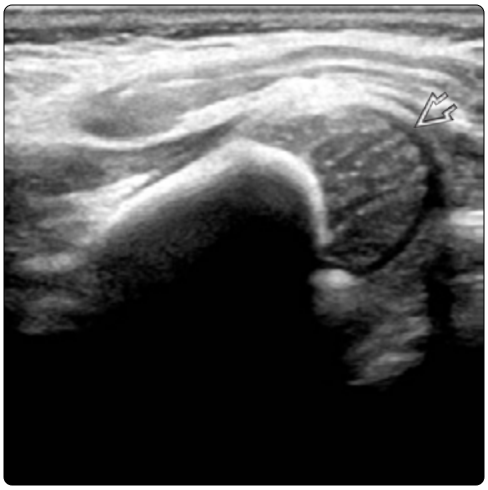


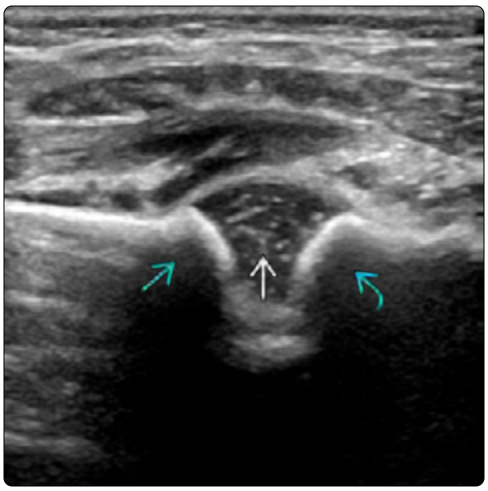


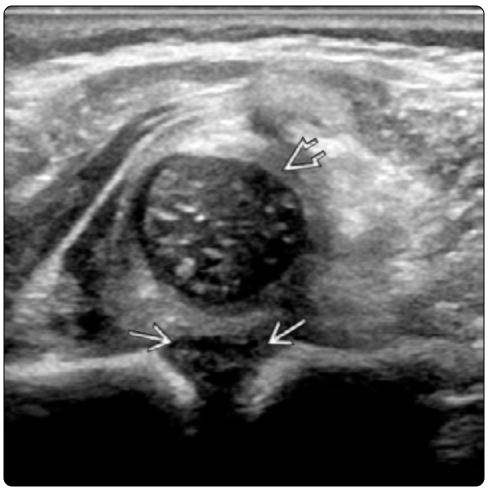
(Left) *Coronal flexed US of a normal right hip shows ≥ 50% coverage of the unossified femoral head. The acetabular roof is normal with an a angle ≥ 60° (measurement not shown).* **(Right)** *Similar image of the left hip in the same patient shows dislocation of the femoral head & a steep acetabular roof (with an a angle of 40°). As is customary, the image is rotated 90° counterclockwise from the anatomic position, so a radiographically steep acetabular roof appears as a "shallow" angle on US.*





(Left) *Transverse flexed stress view shows a normal, stable right hip. Note how the femoral metaphysis, triradiate cartilage, & ischium form a U-shape concavity in which the unossified femoral head rests. Even with stress, a stable hip will remain resting against the ischium & posterior labrum.* **(Right)** *In contradistinction, the unstable left hip (in the same patient) is dislocated. The femoral head lies posterior & lateral, no longer resting in a U-shaped concavity.*





(Left) *The posterior lip US view is useful to determine stability but is often misunderstood. The unossified cartilage between the iliac bone & ischium is actually the posterior lip of the acetabulum, not the femoral head. If a hip is unstable, the femoral head will be seen rising over the convex margin of the acetabular cartilage with stress.* **(Right)** *In this already dislocated hip, the femoral head is seen just above (posterior & lateral) the posterior lip of the acetabulum.*

Proximal Focal Femoral Deficiency

KEY FACTS

TERMINOLOGY

- Proximal femoral focal deficiency: Malformation in which complete growth & development of upper femur fails to occur

IMAGING

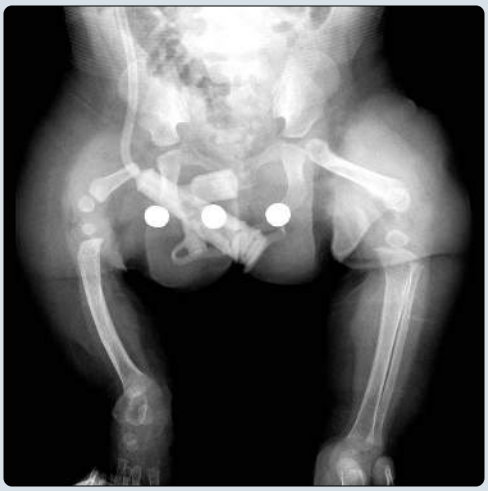
- Often difficult to classify with radiographs at early age (due to delayed ossification)
- Can classify earlier with MR

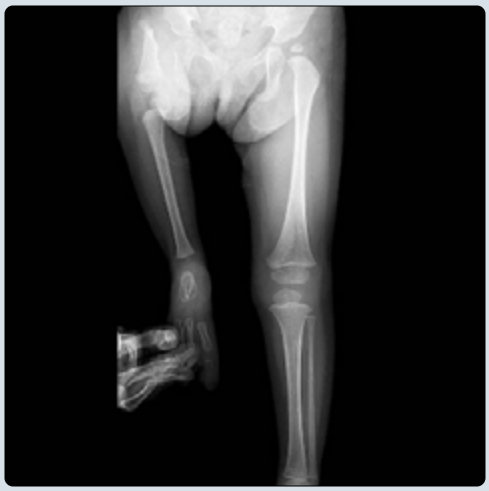
PATHOLOGY

- Aitken classification
  - Class A (38%)
    - Femoral head present & acetabulum normal
    - All parts of femur connected by bone
    - Subtrochanteric varus common
  - Class B (32%)
    - Femoral head present within acetabulum & acetabulum adequate or moderately dysplastic
    - Bone does not connect femoral head & shaft
    - Pseudoarthrosis is often present but does not heal at skeletal maturity
  - Class C (17%)
    - Femoral head absent or represented by ossicle
    - Femur tapers proximally
    - Acetabulum severely dysplastic
  - Class D (13%)
    - Both femoral head & acetabulum absent
    - Distal femoral segment shortened & deformed
    - Obturator foramen of pelvis enlarged
- Associations: Ipsilateral fibular hemimelia in ~ 50%, unstable knee, tarsal coalition, ↓ number of foot rays; abnormal contralateral extremity in 26%

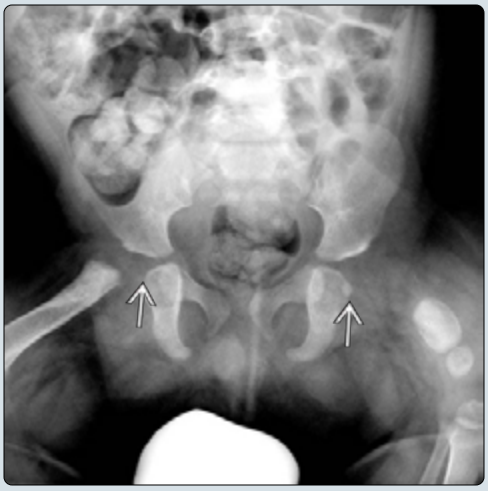
CLINICAL ISSUES

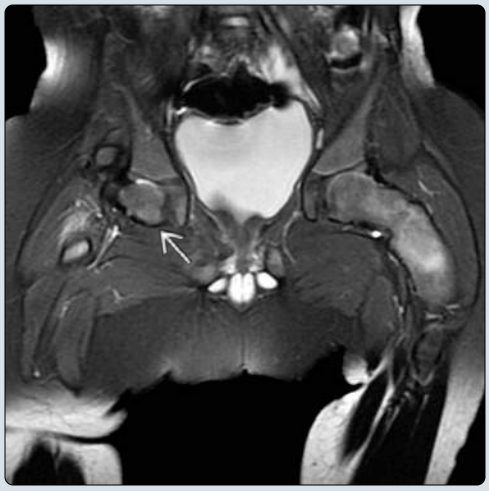
- 4 major biomechanical problems
  - Hip instability, malrotation of thigh with flexed knee, deficient proximal musculature, leg length discrepancy





(Left) *AP radiograph in a 6 month old shows bilateral (right worse than left) proximal femoral focal deficiency (PFFD). Notice the right-sided fibular hemimelia, anterior tibial bowing, 4 rays of the right foot, & severely dysplastic acetabulum.* **(Right)** *AP radiograph in a 2 year old shows a short right femur with no right femoral head ossification. The right acetabulum appears dysplastic. This is best classified as Aitken class B or C. There is absence of the right fibula & only 4 rays of the right foot.*





(Left) *AP radiograph in a 4 month old shows bilateral tiny femoral head ossification centers with bilateral PFFD changes.* **(Right)** *Coronal T2 FS MR in the same child at 20 months shows no connection of the right femoral head to the remainder of the femur. There is continuity of the left femoral head & neck, which eventually developed pseudoarthrosis 1 year later (not shown). This is best classified as bilateral Aitken class B.*

TERMINOLOGY

Abbreviations

- Proximal femoral focal deficiency (PFFD)

Definitions

- Malformation in which complete growth & development of upper femur fails to occur
- Spectrum ranging from mere mild shortening & varus deformity of otherwise normal femur to severe handicap of absent femur except for distal fragment
- Accompanied by varying degrees of acetabular aplasia, thigh muscular hypoplasia & dysplasia, & shortened lower extremity length

IMAGING

Radiographic Findings

- Radiography
  - Pelvis
    - Obturator foramen: Enlarged
    - Acetabulum: Supraacetabular bump of Court, horizontal or small/shallow/dysplastic roof
    - When detached femoral head fixed in acetabulum, concurrence of supraacetabular bump, enlarged obturator foramen, horizontal acetabular roof, & pencil-pointing of upper end of detached distal femur common
  - Femur
    - Short femur; delayed appearance or nonappearance of femoral capital ossification center
      - Average age of appearance in PFFD is 25 months vs. 3-6 months in normal patients
    - Misshapen femoral head & neck, coxa vara
    - Upper end of disconnected distal femur: Either bulbous or pencil-pointed

MR Findings

- More accurate than radiographic evaluation for classification of PFFD
  - Radiographs tend to overestimate degree of deficiency prior to complete ossification of cartilaginous femur
- Shows cartilage structure of acetabulum & upper femur in infants to assist prognosis & treatment planning
- Presence or absence of unossified femoral head & neck & their degree of connection to shaft can be identified
- Hip joint poorly forms when femoral head fixed in acetabulum
- Sartorius muscle is enlarged, which may explain flexion, abduction, & external rotation of hip

Ultrasonographic Findings

- Assist in Aitken classification
- Prenatal diagnosis may be difficult

PATHOLOGY

General Features

- Etiology
  - Embryology
    - Developmental insult: PFFD occurs at 4-6 weeks during limb-bud formation, growth, & differentiation
    - Acetabulum & proximal femur develop from common anlage in embryo
- Associated abnormalities
  - Ipsilateral fibular hemimelia (absence) in ~ 50% (suspect when lateral malleolus absent)
  - Knee often unstable & may dislocate
  - Absent or hypoplastic cruciate ligaments & menisci
  - Ball-&-socket ankle, clubfoot, tarsal coalition, ↓ number of foot rays
  - Abnormal contralateral extremity in 26%

Staging, Grading, & Classification

- At least 8 classification schemes
- Goals of classification: Predict limb function & treatment planning
- Aitken classification (1968)
  - Class A (38%): Femoral head present & acetabulum normal; short femur, all parts of femur connected by bone; subtrochanteric varus common
  - Class B (32%): Femoral head present within acetabulum & acetabulum adequate or moderately dysplastic; bone does not connect femoral head & shaft
  - Class C (17%): Femoral head absent or represented by ossicle; acetabulum severely dysplastic
  - Class D (13%): Both femoral head & acetabulum absent; distal femoral segment shortened & deformed; obturator foramen of pelvis enlarged

CLINICAL ISSUES

Treatment

- 4 major biomechanical problems
  - Hip instability, malrotation of thigh with flexed knee, poor development of proximal musculature, leg length discrepancy
- Treatment is highly individualized; options include (when PFFD unilateral) prosthesis, limb lengthening, hip reconstruction, & Syme amputation
- Bilateral PFFD: Most ambulate well on shortened legs
  - Usually not treated surgically unless severe foot deformities
  - Children ambulate at home without prostheses; prostheses used in social settings to achieve stature closer to that of peers

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- If radiographs show normal acetabulum at birth, normal cartilaginous femoral head is likely

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Legg-Calvé-Perthes Disease

KEY FACTS

TERMINOLOGY

- Symptomatic growth disturbance of capital femoral epiphysis due to idiopathic osteonecrosis

IMAGING

- Radiographic flattening & fragmentation of sclerotic capital femoral epiphysis ± "cystic" metaphyseal changes
- ↓ perfusion of femoral head acutely
  - Photopenia on nuclear medicine bone scan
  - ↓ enhancement on MR: Sagittal plane detects early anterior involvement
- Gradual coxa magna, coxa plana, & coxa brevis with femoral head extrusion, femoroacetabular impingement, labral tears, joint degeneration

TOP DIFFERENTIAL DIAGNOSES

- Septic arthritis
- Transient synovitis
- Juvenile idiopathic arthritis
- Slipped capital femoral epiphysis
- Epiphyseal dysplasias

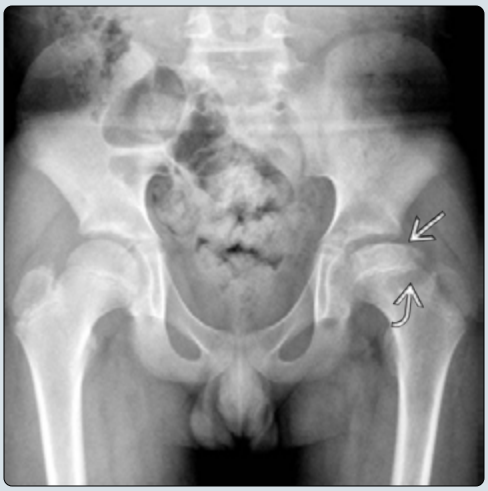
CLINICAL ISSUES

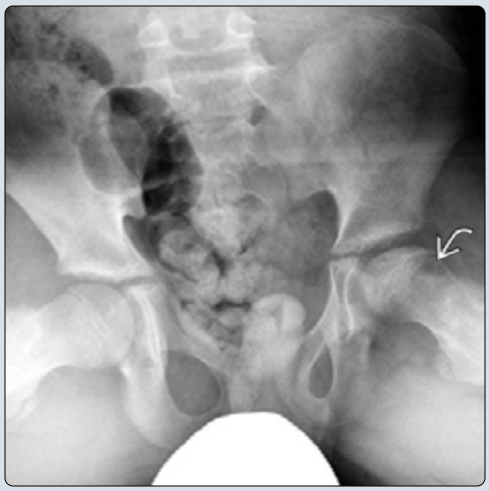
- Limp due to groin, thigh, or referred knee pain
- Age: 4-12 years old; peak: 5-7 years old
- 10-20% bilateral, usually asynchronous
- Worse prognosis: > 8 years of age at onset, greater femoral head + lateral pillar involvement, subchondral fracture, metaphyseal changes, physeal arrest, aspherical femoral head with joint incongruence
- Surgical interventions: Femoral head containment with joint congruence key to maintaining femoral head shape & preventing accelerated joint degeneration

DIAGNOSTIC CHECKLIST

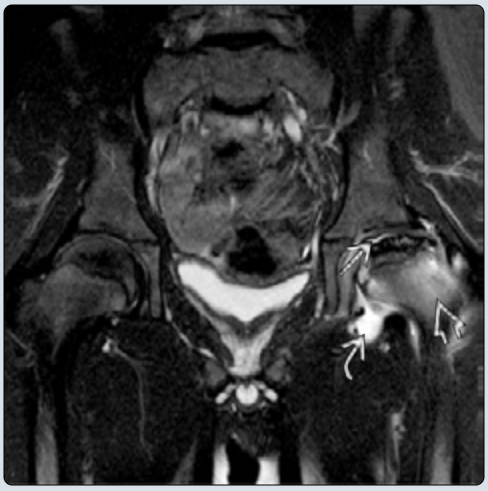
- Anterior femoral head most frequently affected: Include sagittal MR plane
- Subtracted pre- from postcontrast T1 FS: Most sensitive MR sequence for acute ischemia, revascularization

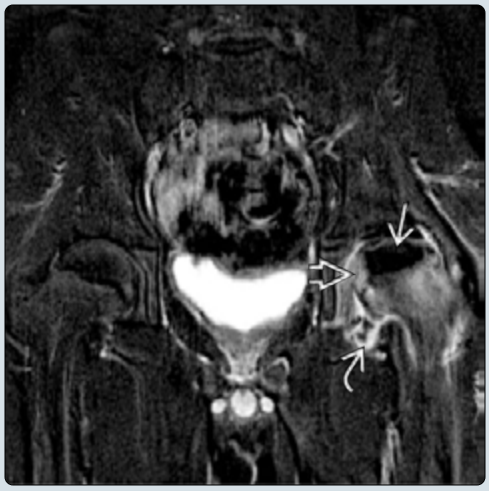
(Left) *AP radiograph in a 10-year-old boy with left hip pain shows flattening, broadening, & sclerosis of the left femoral head. The left hip joint is widened, & the acetabular roof is flattened. The femoral neck is short & broad with a lateral metaphyseal lucency.* **(Right)** *Frog leg lateral radiograph in the same patient shows similar left hip findings with better definition of the cystic-appearing femoral neck lucency. The constellation of findings is typical for Legg-Calvé-Perthes (LCP) disease.*





(Left) *Coronal T2 FS MR in the same patient shows fluid within a subchondral fracture of the flattened & hypointense left femoral head. There is hyperintense marrow edema in the femoral neck with an adjacent joint effusion.* **(Right)** *Subtracted T1 C+ FS MR in the same patient shows no enhancement of ~ 80% of the left femoral head (as compared to the normal right) with the medial ~ 20% showing hyperenhancement. Adjacent reactive synovitis is noted.*





TERMINOLOGY

Abbreviations

- Legg-Calvé-Perthes (LCP) disease

Synonyms

- Legg-Perthes or Perthes disease
- Juvenile idiopathic osteonecrosis

Definitions

- Idiopathic avascular necrosis of femoral head in children

IMAGING

General Features

- Best diagnostic clue
  - Sclerosis, fragmentation, & flattening of capital femoral epiphysis in child 5-7 years of age
  - ↓ perfusion of femoral head (earliest stage) in otherwise healthy child with pain
    - Photopenia on Tc-99m nuclear medicine bone scan
    - ↓ enhancement on MR
- Morphology
  - Proximal femoral configuration depends on extent & severity of involvement & timing of imaging

Radiographic Findings

- Radiography
  - Does not depict earliest stages of insult or repair
  - Active early: Sclerosis, subchondral fracture (lucency), early fragmentation of femoral head
  - Active late: Late fragmentation (± reossification) with flattening (asphericity), extrusion of femoral head, metaphyseal irregularity or "cystic" change
    - Hinge abduction: Medial joint space widening with abnormal lateral femoral head rotating on superolateral acetabular rim
  - Healed: Reossification completed with absence of sclerotic avascular bone
    - Coxa magna (enlarged femoral head), coxa plana (flattened), coxa irregularis (irregular), coxa brevis (short neck, greater trochanter overgrowth), hip joint incongruence
  - Waldenström classification: Radiographic reflection of LCP natural evolution; developed in 1922
    - Initial/necrotic stage (lasts 6 months to 1 year)
    - Fragmentation stage (lasts 2-3 years)
    - Reparative/healing/reossification stage (lasts 1-2 years)
    - Growing/remodeling stage (changes up to skeletal maturity)
    - Definite stage (final shape with congruent or incongruent joint)

MR Findings

- T1WI
  - Patchy hypointensity replacing fatty marrow signal due to necrosis
  - Returning fatty marrow signal intensity with revascularization
- T2WI FS
  - Patchy femoral head ↑ signal (edema) or ↓ signal (sclerosis)
  - Hyperintense joint effusion & synovitis
- T2* GRE
  - Cartilage disturbances: Epiphyseal thickening, metaphyseal abnormalities, physeal bony bridge, osteochondritis dissecans, articular surface degeneration
- DWI
  - ↑ ADC values
- T1WI C+ FS
  - Acute ischemia: ↓ enhancement
    - Subtraction of unenhanced T1 FS from T1 C+ FS (perfusion): Earliest detection/extent assessment
    - Sagittal images show early anterior involvement
  - Adjacent marrow edema, synovitis: ↑ enhancement
  - Revascularized bone: ↑ enhancement
    - Improved prognosis with lateral pillar perfusion; may predict radiographic maintenance of lateral pillar
- MR Arthrogram
  - Long-term sequelae include joint degeneration & femoroacetabular impingement with labral tears
- Advanced techniques
  - Delayed gadolinium enhancement, T1rho, & T2 mapping detect cartilage degeneration prior to conventional sequences

Nuclear Medicine Findings

- Bone scintigraphy
  - Photopenia from ischemia
    - In correct clinical setting, associated joint effusion must be sampled to exclude septic arthritis
  - ↑ uptake with revascularization

Imaging Recommendations

- Best imaging tool
  - MR or NM bone scan for early changes of ischemia & revascularization
    - MR better delineates location of involvement
- Protocol advice
  - MR: FS postcontrast images key
    - Subtraction most helpful (perfusion)
    - Sagittal plane most sensitive for early changes
    - Specific thresholds of ↓ enhancement & ↑ ADC values may predict worse outcomes
  - NM bone scan: Pinhole hip images

DIFFERENTIAL DIAGNOSIS

Septic Arthritis

- Fever, leukocytosis, elevated erythrocyte sedimentation rate
- Hip held in flexion, abduction, external rotation
- Joint effusion ± marrow edema
  - Reduced perfusion of femoral head due to large effusion
- May yield long-term ossific abnormalities or joint destruction

Transient Synovitis

- Self-limited acute synovitis in ages 3-10 years
- Significant effusion & capsular distension
- No long-term femoral head changes

Juvenile Idiopathic Arthritis

- Synovitis predominates over bony findings early

- ± rice bodies in joint fluid

Slipped Capital Femoral Epiphysis

- Posterior, medial displacement of femoral head
- Older patients (age 10-15 years), often obese

Osteoid Osteoma

- Local pain worse at night, decreased by salicylates
- Uncommonly epiphyseal
- May have sclerosis, marrow/soft tissue edema, effusion

Juvenile Osteonecrosis

- Avascular necrosis due to known cause: Sickle cell anemia, steroids, Gaucher, after hip dislocation

Epiphyseal Dysplasias

- Multifocal: Spondyloepiphyseal & multiple epiphyseal dysplasias
- Limited to hips: Meyer dysplasia
  - Age 2-4 years, bilateral in 60%, asymptomatic

PATHOLOGY

Staging, Grading, & Classification

- Prognostic staging during radiographic evolution
  - Catterall classification (originated in 1971, 1st widely used system): Based on extent of epiphyseal involvement
  - Salter-Thompson scheme (originated in 1984): Based on extent of subchondral fracture
  - Herring system (originated in 1992): Based on lateral pillar (lateral 1/3 of femoral head) involvement by AP view
    - Most reliable, reproducible
- Prognostic staging of final morphology at maturity
  - Stulberg classification based on shape, congruency of femoral head
- Radiographic systems may miss window of opportunity for intervention while waiting to predict outcomes at (or beyond) midfragmentation stage; perfusion MR allows assessment during initial stage

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Limping with groin, thigh, or referred knee pain
- Clinical profile
  - No specific history of trauma
  - ↓ range of motion with loss of abduction & internal rotation

Demographics

- Age
  - 4-12 years; peak: 5-7 years
- Gender
  - M:F = 4-5:1
- Epidemiology
  - Affects 3.8-15.6/100,000 children
  - 10-20% bilateral, usually asynchronous

Natural History & Prognosis

- Worse prognosis
  - > 8 years old at onset
  - Greater extent of femoral head involvement
  - Necrosis/height loss of lateral pillar
  - Subchondral fracture
  - Metaphyseal changes
  - Premature physeal closure
  - Aspherical femoral head
  - Lateral subluxation with joint incongruence
- 21-77% with premature physeal closure at hip
  - 3.5 years earlier than unaffected side
  - High percentage develop limb length discrepancy > 1 cm

Treatment

- Conservative
  - Bed rest, abduction stretching, bracing
  - Best nonoperative outcomes in patients < 6 years of age
- Surgical
  - Femoral/pelvic osteotomies, shelf acetabuloplasty
    - Femoral head containment with joint congruence key to maintaining femoral head shape & preventing accelerated joint degeneration
  - Greater trochanter epiphysiodesis to prevent overgrowth with abnormal gait & instability

DIAGNOSTIC CHECKLIST

Consider

- MR & NM bone scan detect early ischemia & revascularization
- Anterior femoral head most frequently affected → include sagittal MR plane

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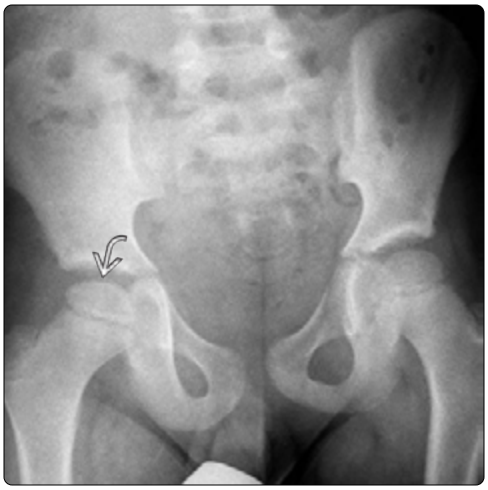
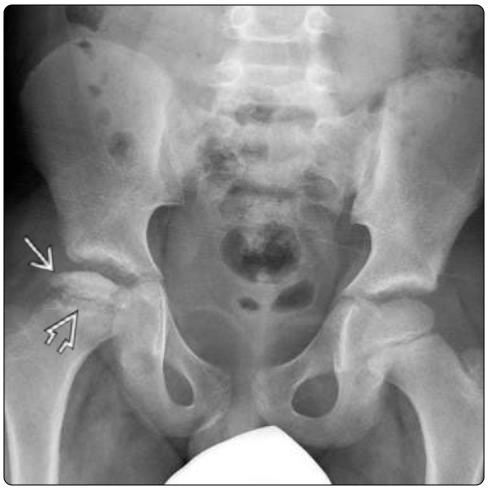
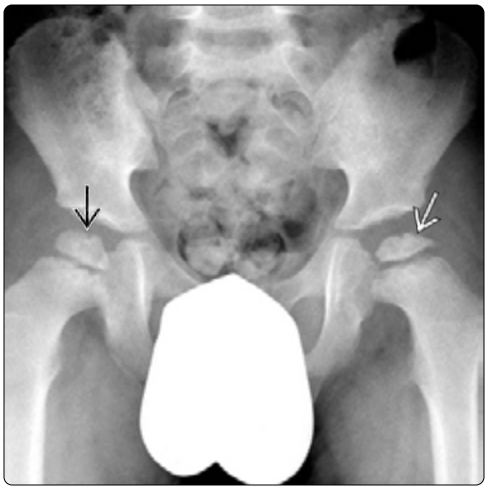
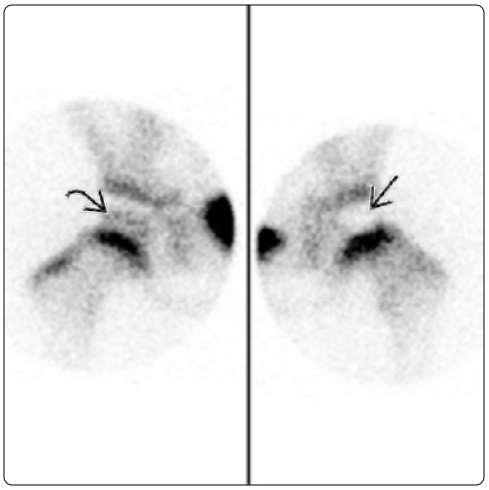


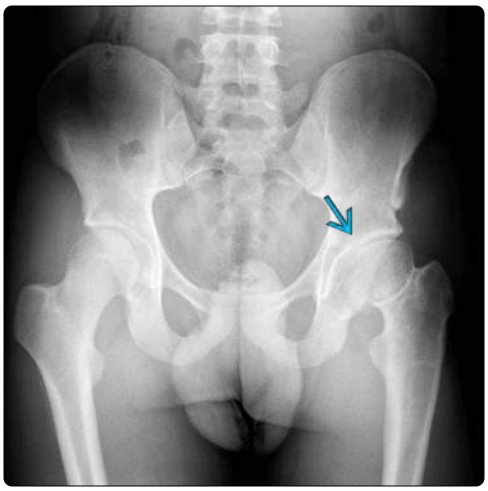


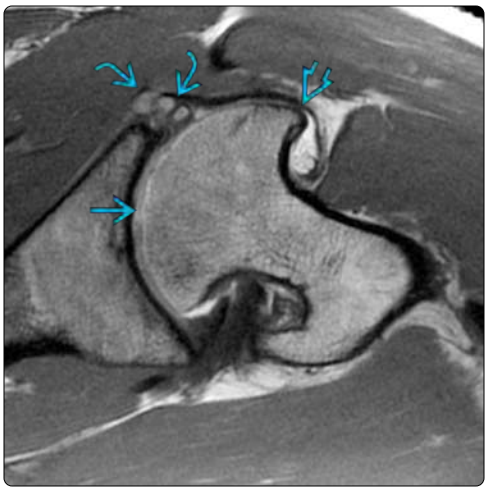
(Left) *AP radiograph in an 8-year-old boy with right hip pain for 3 weeks shows mild sclerosis of the right femoral head with a superimposed subchondral lucency, consistent with a fracture in the setting of avascular necrosis of LCP.* **(Right)** *AP radiograph in the same patient 6 months later shows progressive collapse, sclerosis, broadening, & lateral uncovering of the right femoral head. There are increased lucent metaphyseal irregularities suggesting a degree of impaired endochondral ossification.*





(Left) *AP radiograph in a 4-year-old boy with left hip pain shows left femoral head flattening & sclerosis, consistent with LCP. Note the central femoral notch in the otherwise normal right femoral head, a normal variant.* **(Right)** *Pinhole images from a Tc-99m MDP bone scan show decreased radiotracer uptake in the left femoral head as compared to the normal right side, consistent with ischemia.*





(Left) *AP radiograph in an 18-year-old with a history of left hip LCP & chronic pain shows a dysplastic left acetabulum with coxa magna, plana, & breva of the proximal femur.* **(Right)** *Axial oblique PD MR arthrogram in the same patient shows chronic LCP findings of coxa magna, plana, & breva with an anterior femoral head protuberance that is causing femoroacetabular impingement. The anterior superior labrum is torn & replaced by para- & intralabral cysts. Articular cartilage degeneration is noted.*

Slipped Capital Femoral Epiphysis

KEY FACTS

TERMINOLOGY

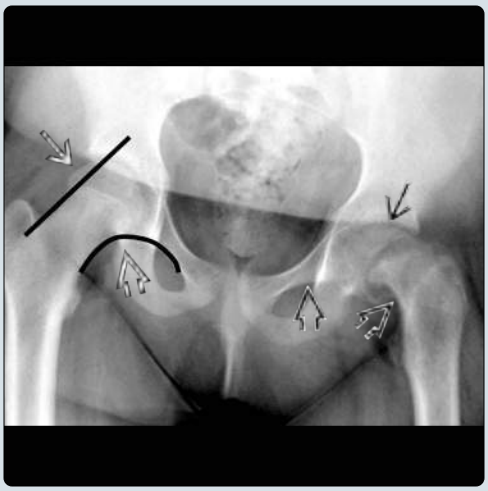
- SCFE: Salter-Harris I fracture of subcapital femoral physis due to chronic stress of weight bearing
  - Femoral head slips posterior & medial to metaphysis

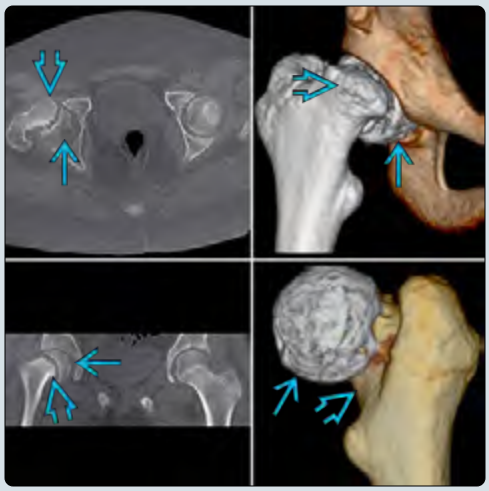
IMAGING

- AP view: Subcapital femoral physis abnormally smooth, lucent, & elongated ("wide")
  - Visible before medial femoral head displacement
- Frogleg lateral view (essential for diagnosis): Posterior displacement of femoral head relative to metaphysis
  - Femoral head-neck angle for severity assessment
- CT/MR more accurately determine severity of slip
- MR more sensitive than radiographs for diagnosis & complications
  - "Preslip" physeal elongation ("widening") on T1
  - ± marrow edema, synovitis with slip on T2 FS/STIR
  - Long-term: Femoroacetabular impingement, labral tear, articular cartilage damage, avascular necrosis (AVN)
- Incidence of bilateral SCFE varies widely: 18-80%
  - At initial presentation: 9-22%
  - Contralateral slip usually within 18 months

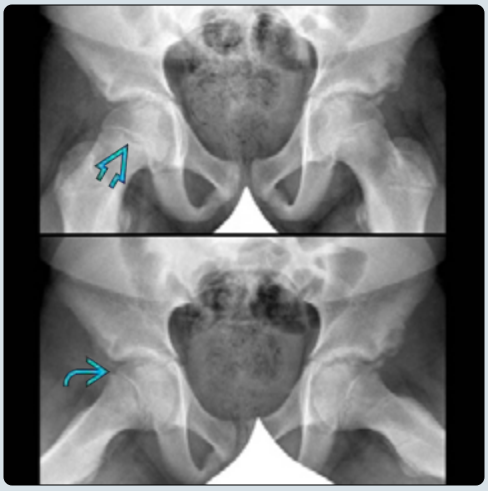
CLINICAL ISSUES

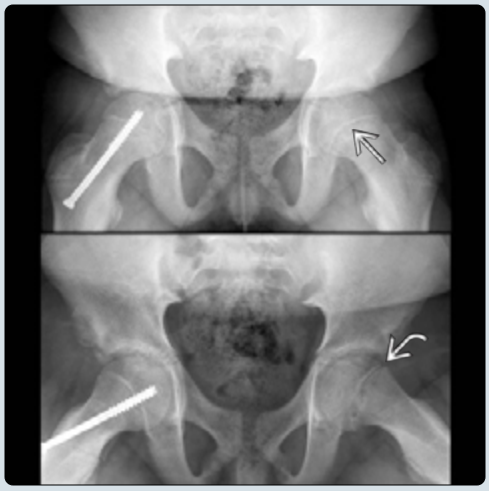
- Limp, pain, limited motion; symptoms often mild for weeks with acute worsening
  - Pain in hip, groin, or proximal thigh in 85%
  - Distal thigh or knee pain in 15%
- Girls average 11-12 years, boys average 13-14 years
- Major predisposing factor: Obesity
- Prognosis poorer for unstable (unable to bear weight) SCFE: ↑ AVN risk
- Most common treatments
  - Percutaneous single screw in situ fixation without femoral head manipulation (stable & mild unstable SCFE)
  - Open surgical hip dislocation with capital realignment (increasing use in moderate & severe unstable SCFE)
  - Prophylactic contralateral fixation controversial





(Left) *AP radiograph in an obese 10-year-old girl with left hip pain shows intersection of the lateral right femoral head ➡ by the line of Klein (normal). Such a line would not intersect the slipped left femoral head ⇨. A normal continuous Shenton arc can be drawn on the right ➡ but not the left ⇨.* **(Right)** *Axial (top left) & coronal (bottom left) 2D & posterior (bottom right) & anterior (top right) 3D surface-rendered CT images in a 12 year old show a chronic SCFE with a posteromedial slip of the right femoral head ⇨ relative to the neck ⇨.*





(Left) *AP (top) & frogleg lateral (bottom) radiographs in an obese 12-year-old patient with right hip pain show an abnormally smooth & lucent right subcapital physis ⇨ with offset of the femoral head-neck junction ⇨, consistent with a SCFE.* **(Right)** *Follow-up radiographs 1 month later in the same patient status post right SCFE pinning demonstrate interval development of an abnormally smooth & lucent left subcapital physis ➡ with mild head-neck offset on the frogleg lateral view ➡, consistent with a left SCFE.*

TERMINOLOGY

Abbreviations

- Slipped capital femoral epiphysis (SCFE, pronounced "skiffie")

Definitions

- Salter-Harris I fracture of subcapital femoral physis due to chronic stress of weight bearing
  - SCFE term not used for acute trauma
- Capital femoral epiphysis = femoral head
- Subcapital physis = proximal femoral growth plate

IMAGING

General Features

- Best diagnostic clue
  - Posterior displacement of femoral capital epiphysis relative to metaphysis on frogleg lateral view
- Location
  - Rates of bilateral SCFE vary widely: 18-80%
    - At initial presentation: 9-22%
    - 88% of contralateral SCFE occur within 18 months
- Morphology
  - Femoral head slips posterior, inferior, & medial relative to metaphysis

Radiographic Findings

- Wide (elongated), smooth, & lucent subcapital femoral physis
  - May occur before visible femoral head displacement
- AP view: Medial displacement of femoral head relative to metaphysis
  - Klein line: Straight line drawn along lateral femoral neck normally intersects outer 15-20% of femoral head
    - Slipped femoral head lies medial to Klein line
    - ↑ sensitivity if > 2-mm difference in amount of femoral head lateral to line vs. contralateral side
  - Shenton arc: Normally continuous smooth curve drawn along undersurface of superior pubic ramus & medial femoral neck
    - SCFE disrupts arc as slipped femoral head displaces femoral neck laterally & superiorly
  - Dense medial femoral metaphysis due to normal overlap with ischium
    - Lost in SCFE due to lateral & superior displacement of femoral neck
- Frogleg lateral view: Posterior displacement of femoral head relative to metaphysis
  - Appears medial relative to acetabulum, pelvis
  - More sensitive for slip than AP view
  - Staging on frogleg lateral view
    - Percentage of epiphyseal displacement: Mild 0-33%, moderate 34-49%, severe > 50%
    - Southwick slip angle: Mild 0-30°, moderate 31-50°, severe > 50°
- Metaphysis: Scalloping, irregularity, sclerosis, & posterior beaking if chronic
- Valgus SCFE (lateral & superior slip of epiphysis) uncommon
  - Typically has posterior component revealed on lateral view (as Klein line insensitive)

CT Findings

- Axial oblique & sagittal planes best reveal maximum displacement
- Osteoporosis, ↓ muscle bulk if chronic

MR Findings

- Physeal elongation ("widening") of "preslip" best seen on coronal T1
- ± marrow edema, synovitis with slip on T2 FS/STIR
- Severity of slip angle more accurate than radiographs
- Periosteal disruption confirms instability
- Chronicity suggested with remodeling, osteoporosis, ↓ muscle bulk
- Long-term: Femoroacetabular impingement (FAI), labral tear, & articular cartilage damage likely ↑ with greater degree of slip & residual head-neck offset
  - dGEMRIC may reveal early cartilage degeneration

Nuclear Medicine Findings

- Bone scan
  - Acute SCFE: ↑ uptake at physis (fracture)
  - Chondrolysis: ↑ uptake on acetabular & femoral sides of joint (synovitis)
  - Avascular necrosis (AVN): ↓ uptake in femoral epiphysis (infarct)

Imaging Recommendations

- Best imaging tool
  - AP & frogleg lateral radiographs of both hips
    - Frogleg view: Abducted, externally rotated femurs
    - Do not use force to secure frogleg lateral position (may worsen slip)
- Protocol advice
  - SCFE may be subtle on radiographs; consider urgent MR to include coronal T1 if any doubt exists

DIFFERENTIAL DIAGNOSIS

Legg-Calvé-Perthes Disease

- Idiopathic AVN of hip, classically 5-8 years old
- Marrow edema, synovitis/effusion, ↓ femoral head enhancement early on MR
- Subsequent radiographic sclerosis & collapse of femoral head ossification center, coxa magna

Juvenile Idiopathic Arthritis

- Nonspecific joint effusion & synovitis, often insidious
- ± multiple joints, characteristic rash, systemic symptoms

Renal Osteodystrophy

- Generalized bony sclerosis with "fuzzy" margins
- Subperiosteal & subsymphyseal resorption
- Multifocal physeal elongation ("widening"): ↑ SCFE risk

Traumatic Salter-Harris I Fracture

- Unequivocal history of acute trauma in adolescence
- Rarely in newborns with difficult delivery

Septic Arthritis

- ↑ WBC, ESR, fever, & failure to bear weight
- Marrow & soft tissue edema often surround effusion

Transient Synovitis

- Limp & pain without systemic findings
- Lack of marrow & soft tissue edema surrounding effusion

Idiopathic Chondrolysis

- Accelerated hip joint cartilage destruction in preadolescent without clear etiology
- Early: Geographic ↑ T2 signal & enhancement centrally in femoral head ± synovitis
- Late: Joint space loss & degenerative change

PATHOLOGY

General Features

- Associated abnormalities
  - AVN may be due to
    - Vessel injury acutely at time of slip
    - Vascular compression from operative reduction
    - ↑ intracapsular pressure by effusion reducing flow
  - Residual head-neck offset post therapy can result in cam-type FAI & premature joint degeneration

Microscopic Features

- Physeal architectural distortion before fracture
- Fracture occurs in zone of hypertrophic chondrocytes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Limp, pain, limited motion
    - Pain in hip, groin, or proximal thigh in 85%
    - Distal thigh or knee pain in 15%
- Other signs/symptoms
  - Clinical staging
    - Stable: Able to bear weight (even with crutches)
      - More likely to have delayed presentation
    - Unstable: Unable to bear weight
    - Timing of symptoms: Acute < 3 weeks, chronic > 3 weeks
      - Acute on chronic: Gradual onset, suddenly worse (most common)

Demographics

- Age
  - Girls: Range 8-15 years, average 11-12 years
  - Boys: Range 10-17 years, average 13-14 years
- Gender
  - M:F = 2.5:1:0
- Ethnicity
  - Nearly 4:1 = African Americans:Caucasians
- Epidemiology
  - Most common hip disorder in adolescents
  - Incidence: 0.7-10.8 per 100,000
- Predisposing factors
  - Obesity most significant factor
  - Adolescent growth spurt
  - Endocrine: Primary hypothyroidism, hypogonadism
  - Renal rickets, radiation therapy, chemotherapy

Natural History & Prognosis

- Prognosis poorer for unstable SCFE → 10-60% develop AVN
  - ↑ risk with slip severity, younger age, manual reduction, ↑ intracapsular pressure
    - Some advocate surgery within 24 hours to ↓ risk
- Chondrolysis from SCFE most frequently due to persistent (not transient) joint penetration by pin
- Joint degeneration secondary to incongruity, residual head-neck offset → cam-type FAI

Treatment

- Percutaneous pin/screw fixation of epiphysis to femoral neck through physis
  - In situ fixation (no attempted reduction due to ↑ risk of AVN with manual reduction)
    - Single cannulated screw typically used
    - Deformed anterior femoral-head neck junction remains
      - ↓ risk of AVN, ↑ risk of FAI & joint degeneration
- Surgical hip dislocation (open reduction + internal fixation)
  - Seeing ↑ use, particularly for unstable severe slip & chronic slip
    - Capsulotomy advocated to ↓ risk of AVN
    - Modified Dunn procedure with capital realignment ± osteochondroplasty
      - ↓ FAI & AVN
- Prophylactic contralateral fixation controversial; strongly considered in
  - Predisposing disease
  - Girls < 10 years old, boys < 12 years old

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Frogleg lateral view essential
- Beware comparison to contralateral side
  - Up to 22% with contralateral SCFE at presentation

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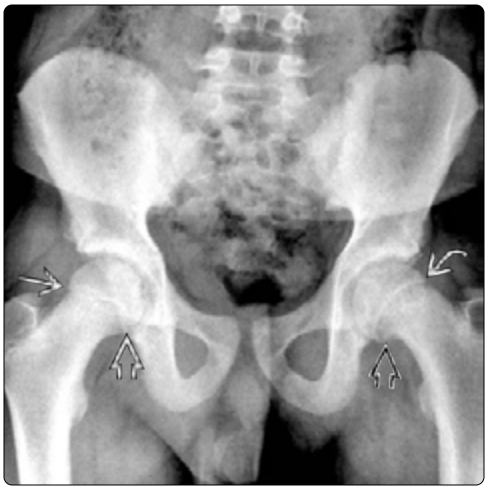
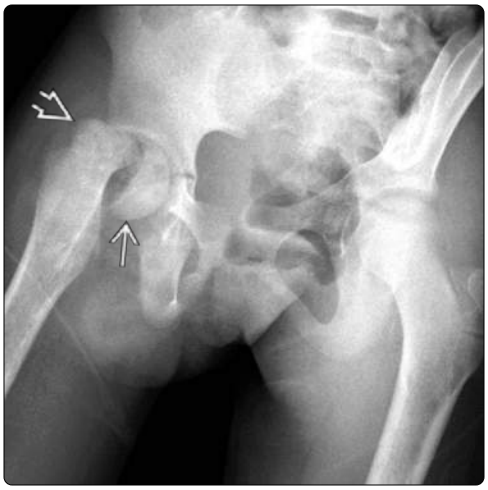
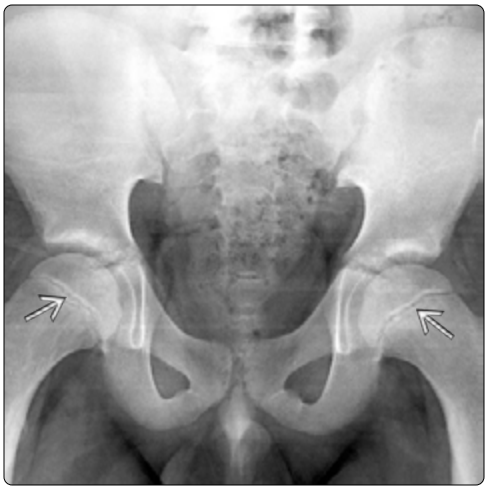
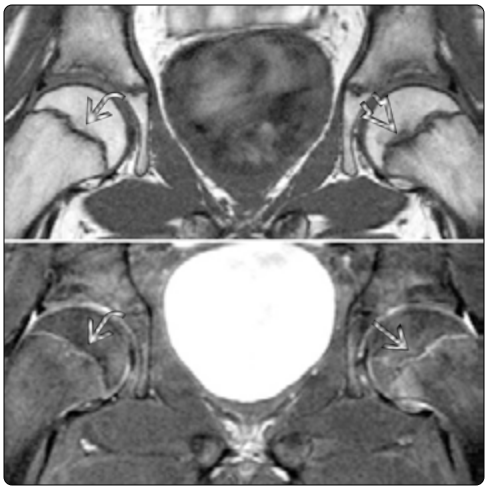


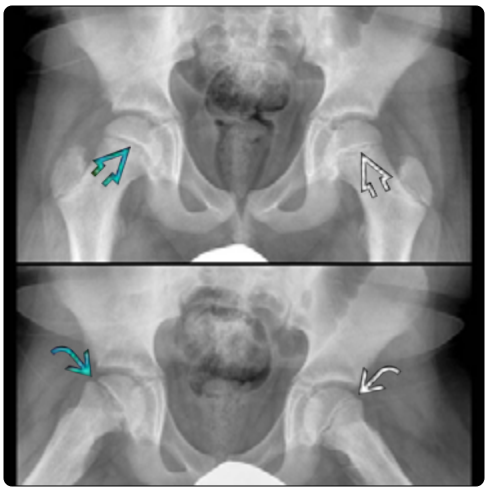


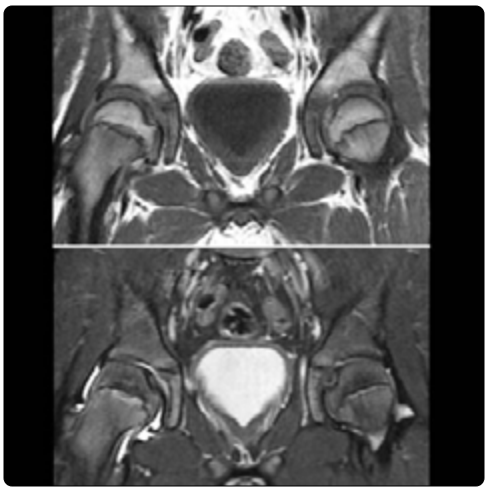
(Left) *AP radiograph in a 15-year-old boy shows slight posterior slippage of the left femoral head with an abnormally smooth, widened, & lucent physis. The left medial femoral metaphyseal corner does not overlap the ischium. Note the normal undulating right proximal femoral physis & normal metaphyseal relationship to the ischium.* **(Right)** *AP radiograph in a 9-year-old patient with prior right hip septic arthritis shows a severe medial slip of the femoral head with displacement of the femoral neck.*





(Left) *AP radiograph in a 14-year-old boy with left hip pain shows normal-appearing subcapital femoral physes.* **(Right)** *Coronal T1 MR (top) of the same patient shows periphyseal foci of fatty marrow signal loss with poor definition of the subcapital femoral physis, suggesting a "preslip". No physeal abnormality can be seen on the corresponding T2 FS MR (bottom). Note the normal right side.*





(Left) *AP (top) & frog-leg lateral (bottom) radiographs in an 11-year-old with hip pain show an abnormally smooth & wide right subcapital physis with femoral head-neck offset, consistent with SCFE. The left subcapital physis appears normal with questionable head-neck offset.* **(Right)** *Coronal T1 (top) & T2 FS (bottom) MR of the same patient shows an abnormal right subcapital physis & associated joint effusion from SCFE. However, the left subcapital physis is normal.*

Achondroplasia

KEY FACTS

TERMINOLOGY

- Most common nonlethal skeletal dysplasia

IMAGING

- Symmetric shortening of all long bones
  - Proximal most affected (rhizomelic)
- Lower extremity shortening
  - Femur barely longer than tibia
  - Ice cream scoop shape of proximal femurs in infants
  - Lower femoral & proximal tibial epiphyses/metaphyses with cone or chevron shape
  - Relatively elongated fibulas
- Upper extremity shortening
  - Humerus barely longer than ulna
  - Short metacarpals & phalanges with trident hand configuration: 3 forks = thumb, digits 2 & 3, digits 4 & 5
- Pelvis: Squared iliac wings, narrowed sacrosciatic notches, horizontal acetabular roofs, overall coupe-type champagne glass configuration
- Spine: Gibbus deformity of thoracolumbar junction, anterior vertebral body beaking or wedging (bullet-shaped), posterior vertebral body scalloping, progressive narrowing of interpediculate distances from L1-L5
- Skull base: Keyhole foramen magnum, narrow jugular foramina → venous hypertension → ventriculomegaly

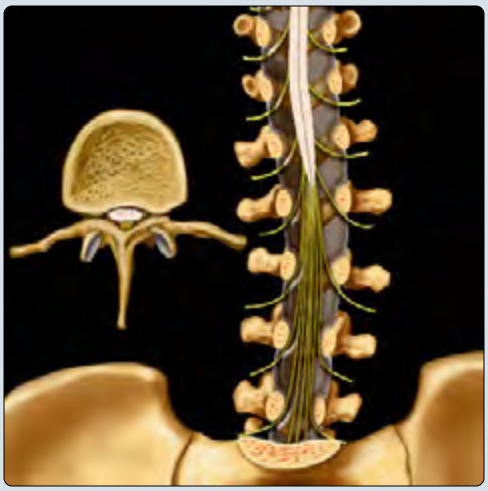
PATHOLOGY

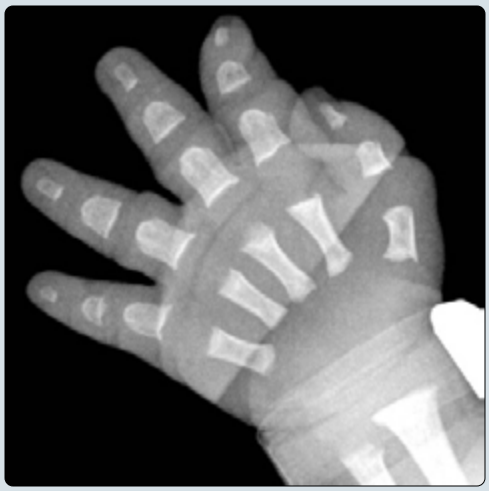
- Autosomal dominant, 80% sporadic
- Mutation at chromosome 4p16.3: Fibroblast growth factor receptor-3 gene (*FGFR3*)
  - Other mutations of same gene cause mild hypochondroplasia & lethal thanatophoric dysplasia
- Fetal cell-free DNA from maternal plasma replacing amniocentesis for prenatal diagnosis

CLINICAL ISSUES

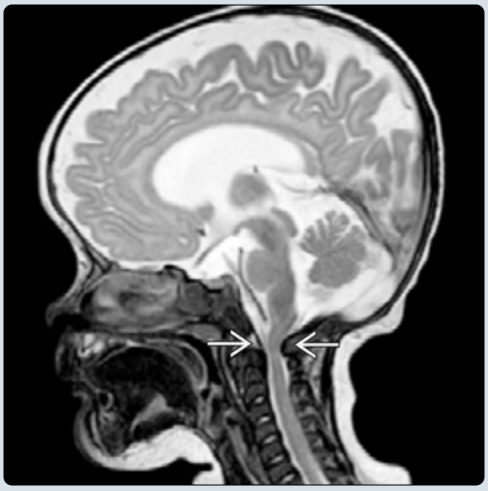
- Incidence: 1:10,000-40,000 live births
- 2-5% risk of sudden death from cervicomedullary compression

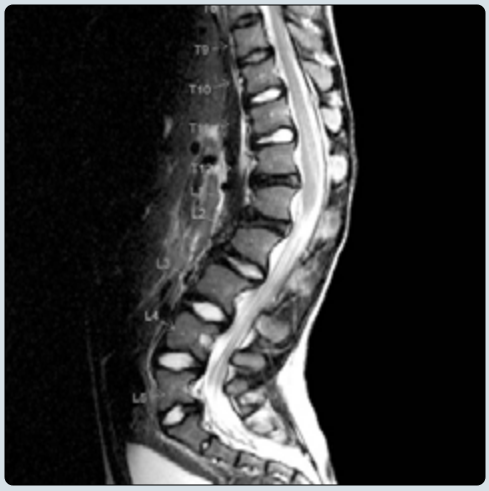
(Left) *Axial & coronal graphics show spinal canal stenosis with short pedicles & progressive narrowing of the interpediculate distances from L1-L5.* **(Right)** *PA radiograph shows a trident hand configuration in a newborn with achondroplasia: The fingers are of approximately equal length & diverge from one another in 2 pairs plus the thumb (i.e., there is splaying of the middle & ring fingers). The metacarpals & phalanges are short & broad.*





(Left) *Sagittal T2 MR in a 2 month old shows narrowing of the craniocervical junction with compression of the brainstem & upper cervical spinal cord. Note the enlarged lateral ventricles & subarachnoid spaces (due to jugular foramen stenosis with venous hypertension & impaired CSF resorption).* **(Right)** *Sagittal T2 MR in a 4 year old shows lumbar spinal canal stenosis with scalloping of the posterior vertebral bodies. A bullet configuration with anterior vertebral body wedging is seen at T12-L2.*





TERMINOLOGY

Definitions

- Mutations of fibroblast growth factor receptor-3 gene (*FGFR3*) at chromosome location 4p16.3 underlie achondroplasia & related dysplasias (FGFR3 group)
  - Achondroplasia
    - Most common nonlethal skeletal dysplasia
    - Large cranium; small face & chest; short limbs
  - Hypochondroplasia
    - Milder skeletal dysplasia affecting similar sites
      - Usually not recognized until child > 2 years old
      - Severe forms overlap with achondroplasia
    - Milder changes of hands & spine
    - Fibular overgrowth
  - Thanatophoric dysplasia
    - Most common lethal bone dysplasia
    - Lungs cannot ventilate due to short ribs; micromelia

IMAGING

Radiographic Findings

- Radiologic evaluation begins with skeletal survey
- Radiography
  - Skull, face, & brain
    - Calvaria enlarged with frontal bossing, megalencephaly
    - Flat nasal bridge
    - Skull base small with narrow foramen magnum
      - Concern for brainstem compression
    - Petrous pyramids closer to midline than normal
    - Basal angle low: 85-120°; steep clivus
    - Basilar impression
    - Narrow jugular foramina may cause ventriculomegaly via venous hypertension
    - Short petrous carotid canals
    - Mastoids underpneumatized
    - Midface hypoplasia, dental crowding
    - Choanal atresia: Occasional
  - Spine
    - Spinal canal stenosis seen as short pedicles on lateral view with ↓ interpediculate distances on AP view
      - Interpediculate distances become progressively smaller lower in lumbar spine (opposite of normal)
    - Vertebral bodies: Short, bullet-shaped in early life; concave posterior surfaces (scalloping)
      - Vertebral heights normal
    - Transverse processes short
    - Thoracolumbar gibbus or kyphosis in infancy
    - ↑ lumbar lordosis after infancy with horizontal sacrum
    - Cervical instability
  - Chest
    - Short ribs & sternum
    - Cup-shaped anterior rib ends
    - Clavicles: Musk ox horn shape, relatively long
  - Pelvis
    - Champagne glass shape (coupe type, not tulip or flute) of inner margin
      - Pelvic cavity broad & short
    - Small squared iliac wings with horizontal acetabular roofs & rounded iliac crests (tombstone or elephant ear appearance)
    - Narrowed sacrosciatic notches
  - Upper extremity
    - Rhizomelic shortening: Humerus barely longer than ulna
    - Outward lateral bulging of humerus at deltoid insertion
    - Concave medial distal radial metaphysis
    - Hand: Delayed bone age
    - Metacarpals & phalanges short, stubby
    - Trident hand with 3 forks: Thumb, digits 2 & 3, digits 4 & 5
      - Splaying of middle & ring fingers
  - Lower extremity
    - Rhizomelic shortening: Femur barely longer than tibia
    - Femoral necks short with coxa vara
    - Hemispheric femoral head
    - Ice cream scoop shape of proximal femurs in infants
    - Flared (widened) metaphyses capped by large epiphyses
    - Diaphyseal widths normal but appear broadened due to shortening
    - Distal femoral & proximal tibial metaphyses/epiphyses cone or chevron in shape
    - Bowlegs: Genu varum (> 90%)
    - Delayed ossification of tibial epiphysis
    - Fibula longer than tibia
    - Posteroinferior calcaneal pseudospurs before ossification of calcaneal apophysis
    - Joint laxity

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal studies may be normal on early scan with long bone shortening not detected until 22-24 weeks gestation
    - Foreshortening of limbs (< 3rd percentile)
    - ↑ biparietal diameter (> 95th percentile)
    - Flat nasal bridge
    - Collar hoop sign: Overgrowth of periosteum with small echogenic hook at proximal femoral metaphyseal-epiphyseal junction
    - Rounded metaphyseal-epiphyseal interface

Other Modality Findings

- MR: Herniated nucleus pulposus
- CT: Intracranial ventricular enlargement frequent

DIFFERENTIAL DIAGNOSIS

Hypochondroplasia

- Milder short-limbed dwarfism with similarly affected sites
  - Hands & feet broad, stubby
  - Midface hypoplasia
  - Foramen magnum small
  - Lumbar spine: ↓ interpediculate distance
  - Vertebral bodies: Posterior scalloping
- Often not clinically apparent until 2 years of age

Pseudoachondroplasia

- Normal skull; severe epiphyseal findings

Metatropic Dysplasia

- Dumbbell-shaped femurs
- Kyphoscoliosis, caudal appendage (tail)

Other Conditions With Endochondral Growth Slowing

- Homozygous achondroplasia (lethal), Ellis van Creveld syndrome, thanatophoric dysplasia (lethal), Morquio disease

PATHOLOGY

General Features

- Etiology
  - ↓ rate of endochondral ossification
    - Vertebral bodies & skull base: Impaired endochondral growth results in stenosis of foramen magnum & spinal canal
- Genetics
  - Defect on chromosome 4p16.3
    - Fibroblast growth factor receptor-3 (*FGFR3*) gene
      - > 98% G1138A (glycine to arginine substitution at nucleotide 1138)
    - Prenatal diagnosis by next generation sequencing of cell-free fetal DNA from maternal plasma
      - Traditional methods: Amniocentesis or chorionic villus sampling
  - Autosomal dominant
    - 80% of cases new (de novo) mutations
  - Heterozygous: Common
  - Homozygous: Rare, lethal

Microscopic Features

- Histology of epiphyseal & physeal growth cartilage normal

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Characteristic face with midface hypoplasia, saddle nose, & frontal bossing
  - Rhizomelic limb-shortening
  - Elbow extension limited
  - Trident hand
  - Thoracolumbar gibbus or kyphosis
  - Lumbar lordosis exaggerated
    - Buttocks prominent & abdomen protuberant after walking begins
  - Hypermobility of joints, especially knees
  - Delayed gross & fine motor milestones; hypotonia

Demographics

- Epidemiology
  - Heterozygous achondroplasia
  - Incidence: 1:10,000-40,000 live births

Natural History & Prognosis

- Normal intelligence; lifespan ~ 10 years less than unaffected population
  - Obesity common & disabling in older children
  - ↑ incidence of orthopedic & neurologic complications
    - Cervical instability in infancy
    - Basilar impression, Chiari 1, syringomyelia
    - Lumbosacral stenosis
    - Tibial bowing in 42%
      - Tibial osteotomy in 22%
  - 2-5% risk of sudden death due to cervicomedullary compression: Suspect with central hypopnea, ↓ arousal state, small foramen magnum, hypotonia, hyperreflexia
  - Upper airway obstruction ~ 5%
  - Otitis media in 90% during first 2 years
    - Ventilation tubes in 80% over lifespan
    - Conductive hearing loss in 40%

Treatment

- Available for most complications
  - Cervicomedullary decompression surgery in 17%
    - Potentially urgent to prevent sudden death
  - Cardiorespiratory & sleep dysfunction
    - Mild midface hypoplasia & relative adenotonsillar hypertrophy: Treat with tonsillectomy & adenoidectomy
    - Upper airway obstruction: Nocturnal continuous positive airway pressure
    - Upper airway obstruction associated with hypoglossal canal stenosis: Various options including foramen magnum decompression
    - Ventriculomegaly from jugular foramen stenosis: Treat with shunt as needed
- Limb lengthening: Controversial, high complication rate

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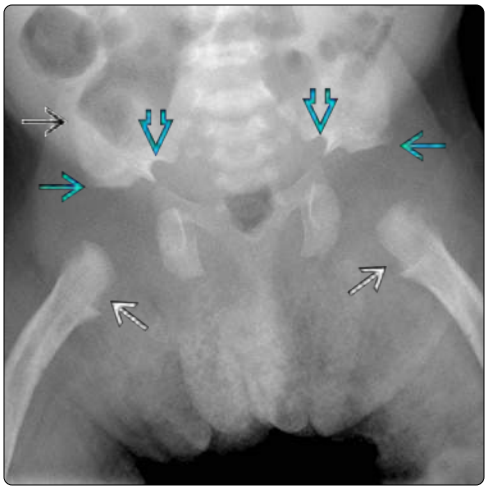
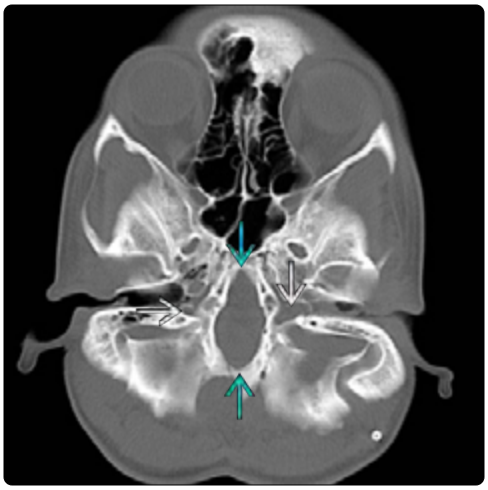
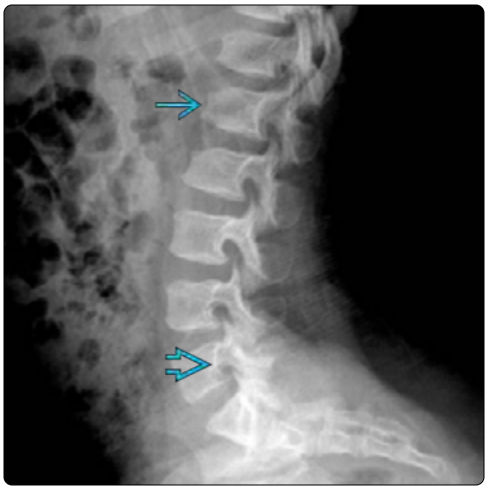
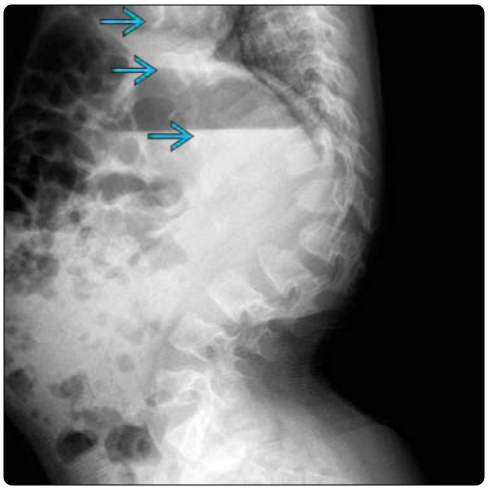


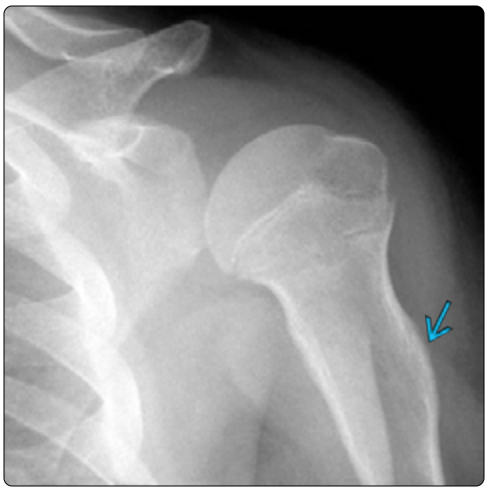


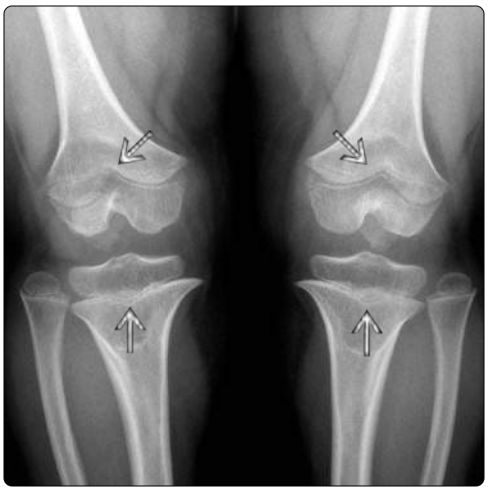
(Left) *AP radiograph in 2 day old with a prominent forehead & short stature shows multiple features of achondroplasia, including ice cream scoop configurations of the proximal femurs, horizontal acetabular roofs, narrowed sacrosciatic notches, tombstone iliac wings, & an overall coupe-type champagne glass appearance of the pelvis.* **(Right)** *Axial bone CT in a teenager with achondroplasia & shunted hydrocephalus shows a keyhole foramen magnum with jugular foramen hypoplasia.*





(Left) *Lateral radiograph in 4 year old with achondroplasia shows prominent posterior scalloping & anterior (bullet) wedging of the vertebral bodies. The pedicles are also short. Note the typical exaggerated lumbosacral lordosis with a horizontal sacrum.* **(Right)** *Lateral radiograph in a 5 year old shows thoracolumbar kyphosis (or gibbus deformity) with an exaggerated lumbar lordosis. There is L2 anterior beaking & increased concavity of the posterior vertebral bodies. The ribs are shortened with anterior flaring.*





(Left) *External rotation radiograph shows a prominent deltoid insertional irregularity of the proximal humerus.* **(Right)** *AP radiograph in a 7 year old with knee pain & achondroplasia shows metaphyseal flaring with chevron or upside-down chevron deformities of the distal femoral & proximal tibial epiphyses bilaterally. The fibulas are elongated.*

Mucopolysaccharidoses

KEY FACTS

TERMINOLOGY

- Heterogeneous group of lysosomal storage diseases due to deficiency of enzymes that degrade glycosaminoglycans (GAGs)
- Dysostosis multiplex: Constellation of bone dysplasia features seen variably in mucopolysaccharidoses (MPS)

IMAGING

- Thoracolumbar gibbus (sharply angled kyphosis) with anteriorly beaked vertebral bodies
- Oar-shaped ribs (thin proximally, wide distally)
- Short, thick clavicles
- Pelvis: Ilia small & tapered inferiorly, steep acetabular roofs, hip subluxation, coxa valga, ± femoral head avascular necrosis
- Pointed proximal 2nd-5th metacarpals
- Diaphyseal widening of tubular bones
- Odontoid hypoplasia & other C1/C2 abnormalities → atlantoaxial subluxation → cord compression

TOP DIFFERENTIAL DIAGNOSES

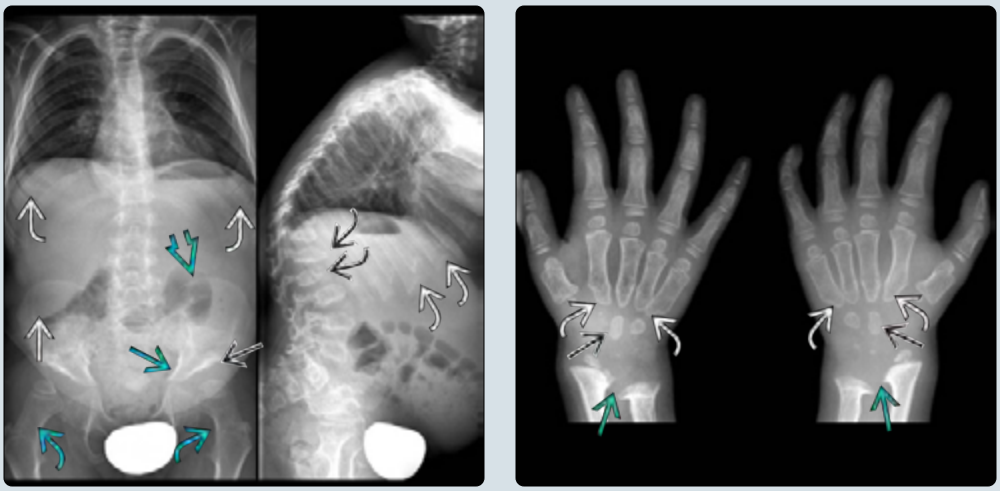
- Legg-Calvé-Perthes disease
- Sickle cell disease
- Gaucher disease
- Spondyloepiphyseal dysplasia
- Multiple epiphyseal dysplasia
- Spondylometaphyseal dysplasia

PATHOLOGY

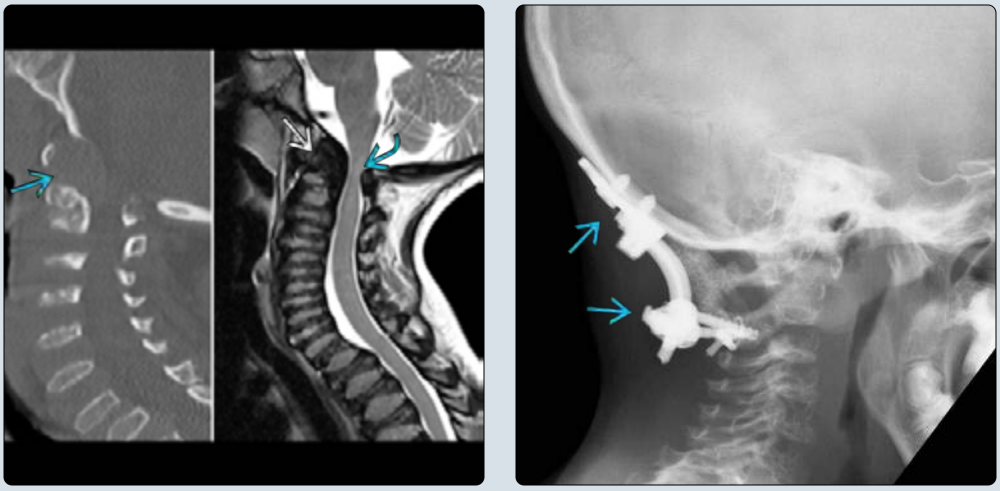
- GAGs ubiquitous in connective tissues throughout body
- GAGs accumulate in lysosomes in bone marrow & multiple viscera → dysfunction

CLINICAL ISSUES

- Stem cell transplantation by unrelated donor umbilical cord blood: MPS I-H, VI
  - Best done when < 2 years old, no CNS disease
- Enzyme replacement therapy: MPS I, II, VI
  - Enzymes do not cross blood-brain barrier



(Left) *Frontal (left) & lateral (right) radiographs in a 6-year-old girl with MPS IV show thoracolumbar kyphosis, small irregular vertebrae, beaked vertebrae, wide ribs, hepatomegaly, coxa valga deformities, steep acetabula, & rounded iliac wings with inferiorly tapered ilia.* **(Right)** *Frontal radiographs of the hands in the same child show short, wide 2nd-5th metacarpals with pointed proximal ends, small irregular carpal bones, & irregular centrally sloped metaphyses of the distal radius & ulna.*



(Left) *Sagittal CT (left) & T2 MR (right) show the C-spine in the same child. The odontoid is hypoplastic with soft tissue filling the expected location of the odontoid. There is narrowing of the spinal canal at the craniocervical junction with cord compression.* **(Right)** *Lateral radiograph in the same child after craniocervical junction decompression & fusion from the occiput to the C2 level is shown. Cord compression in MPS can lead to cervical/brainstem myelopathy & respiratory failure.*

TERMINOLOGY

Abbreviations

- Mucopolysaccharidosis/mucopolysaccharidoses: MPS
- MPS I-H: Hurler syndrome
- MPS I-S: Scheie syndrome
- MPS I-H/S: Hurler-Scheie syndrome
- MPS II: Hunter syndrome
- MPS III: Sanfilippo syndrome
- MPS IV: Morquio syndrome
- MPS V: Nonexistent, now classified as MPS I-S
- MPS VI: Maroteaux-Lamy syndrome
- MPS VII: Sly syndrome or β-glucuronidase deficiency
- MPS VIII: Nonexistent
- MPS IX: Hyaluronidase deficiency

Definitions

- Heterogeneous group of lysosomal storage diseases due to deficiency of glycosaminoglycan (GAG) degrading enzymes
  - GAGs formerly called mucopolysaccharides
- Dysostosis multiplex (DM): Constellation of bone dysplasia features seen variably in MPS
- DM group: Includes all storage diseases that lead to skeletal dysplasia
  - MPS, mucolipidoses, others (Gaucher, Niemann-Pick, gangliosidosis, fucosidosis, mannosidosis, sialidosis)

IMAGING

Radiographic Findings

- Key features
  - Thoracolumbar gibbus (sharply angled kyphosis) with anteriorly beaked vertebral bodies
  - Oar-shaped ribs (thin proximally, wide distally)
  - Short, thick clavicles
  - Pelvis: Ilia small & tapered inferiorly, steep, poorly formed acetabular roof, hip subluxation, coxa valga, femoral head deformities ± avascular necrosis (AVN)
  - Pointed proximal 2nd-5th metacarpals
  - Diaphyseal widening of small tubular bones

Other Modality Findings

- MPS I-H typifies DM features seen in other MPS types
- MPS I-H, Hurler syndrome: Severe DM
  - Brain: Abnormal white matter; delayed myelination; ↑ size of perivascular spaces, sulci, ventricles
  - Neurocranium: Enlarged; early closure of sagittal & lambdoid sutures; thickened skull base; J-shaped sella
  - Face: Mandibular condyles flat/concave ± TMJ ankylosis; underpneumatized mastoids
  - Craniocervical junction: Spinal cord compression
  - Spine: Hypoplastic dens with C1/C2 subluxation; C3/C4 subluxation; thoracolumbar gibbus
    - May lead to spinal canal stenosis ± syringohydromyelia
  - Chest & shoulders: Trachea narrow; ribs wide & oar-shaped; clavicles short & thick; scapulae elevated with dysmorphic glenoid fossae
  - Pelvis & hips: Ilia small & taper inferiorly; steep, poorly formed acetabula; femoral head subluxation & developmental deformity ± AVN; coxa valga
  - Knees: Genu valgum
  - Arms: Humeral neck varus (hatchet-shaped humerus); wide humeral midshaft; distal radial & ulnar physes tilt toward each other
  - Wrists: Carpals small & irregular
  - Hands: Wide & short metacarpals with proximal pointed ends at 2-5; wide proximal & middle phalanges; synovitis & claw deformity, trigger finger
  - Cardiovascular: Cardiomyopathy; arterial narrowing; mitral/aortic stenosis
- MPS I-S: Scheie syndrome
  - Mild DM
- MPS II: Hunter syndrome
  - Mild to severe DM
- MPS I-H/S: Hurler-Scheie syndrome
  - Mild to moderate DM
- MPS IIIA-D: Sanfilippo syndrome
  - Mild or absent DM
  - Attenuated (mild) form: Mental retardation, no DM
- MPS IVA-B: Morquio syndrome
  - Type A: Severe DM
    - Cord compression at C1/C2 level due to ≥ 1 of following
      - Transverse ligament laxity
      - Dural thickening from deposition of GAGs
      - Odontoid hypoplasia ± anterior soft tissue mass of unossified fibrocartilage & reactive changes
      - Indentation of posterior arch of C1
      - Chronic subluxation at C1/C2 → ligamentous hypertrophy → further narrowing at craniocervical junction → additional cord compression
      - Cervical cord compression can lead to myelopathy & respiratory failure
    - Cord compression at thoracic/thoracolumbar level due to gibbous formation due to vertebral malformations
    - Narrow, soft trachea collapses during neck flexion
    - Pectus carinatum
    - AVN of femoral heads
    - Ribs wide but not oar-shaped
  - Type B: Moderate DM
- MPS VI: Maroteaux-Lamy syndrome
  - Mild to severe DM
  - Severe form may have cervical & thoracic cord compression similar to type IV
- MPS VII: Sly syndrome
  - Mild to severe DM
    - Femoral head: AVN
- MPS IX: Hyaluronidase deficiency
  - Mild DM: Extremely rare

Imaging Recommendations

- Best imaging tool
  - Most findings made on radiographs
    - Brain & spinal MR may be needed

DIFFERENTIAL DIAGNOSIS

Legg-Calvé-Perthes Disease

- Idiopathic AVN of femoral head
- Isolated to hip without other MPS/DM features

Sickle Cell Disease

- Predominately African American hemoglobinopathy
- Widespread marrow hyperplasia & osteonecrosis

Gaucher Disease

- Storage disorder (not MPS) under DM group
- Osteoporosis with medullary expansion, undertubulation
- Osteonecrosis, H-shaped vertebral bodies

Spondyloepiphyseal Dysplasia

- No clinical or lab features of MPS
- Platyspondyly & abnormal epiphyses

Multiple Epiphyseal Dysplasia

- No clinical or lab features of MPS
- Findings at multiple epiphyses

Spondylometaphyseal Dysplasia

- No clinical or lab features of MPS
- Findings in spine & metaphyses

PATHOLOGY

General Features

- Etiology
  - Enzymatic deficiencies prevent GAG degradation
  - GAGs accumulate in lysosomes in bone marrow & multiple viscera → dysfunction
- Genetics
  - All MPS have autosomal recessive genetic abnormality (except MPS II: X-linked recessive)
    - MPS I-H, I-S, & I-H/S have same biochemical defect with spectrum of phenotypic severity
  - MPS I-H/S: > 110 gene mutations reported

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Spectrum of phenotypes (mild to severe)
    - Most common: Organomegaly, DM, mental retardation/developmental delay
  - 45% of parents of MPS I children report change in facial features (1st clue to child's disease)
  - Brain: Regression of speech & learning skills → mental retardation (not in MPS IV & VI)
  - Head/face: Large head, coarse hair & facial features, proptosis, corneal opacification, recurrent otitis media, flared nostrils, protruding tongue, ↓ hearing
  - Neck: Adenotonsillar enlargement, snoring, sleep apnea, tracheobronchomalacia
  - Spine & chest: Thoracolumbar gibbus (age 6-14 months in MPS I-H), scoliosis, spondylolisthesis in adults with MPS III, pectus carinatum
  - Respiratory: Frequent pneumonia
  - Cardiovascular: Valvular thickening, stenosis, insufficiency; cardiomyopathy, heart failure
  - Abdomen: Protuberant due to hepatosplenomegaly, umbilical/inguinal hernia, intestinal pseudoobstruction, idiopathic diarrhea
  - General: Short (except MPS I-S), claw hand, trigger finger, thick skin, carpal tunnel syndrome, ↓ joint mobility
  - Hydrops fetalis: MPS IV, VII
  - General anesthesia: ↑ risk due to redundant supraglottic tissue, ↓ airway size & stability, unstable C1/C2 joints in MPS I, II, IV, VI, & VII

Demographics

- Age
  - Clinical onset usually < 6 years old
  - Radiographic onset: May be at birth
- Epidemiology
  - Overall incidence of all types of MPS ~ 1 in 20,000 live births

Natural History & Prognosis

- Varies depending on type & severity
- Life span without treatment: MPS I-H < 10 years, MPS II roughly 15 years
- Near-normal lifespan: MPS I-S

Treatment

- Prevention of CNS damage: Perinatal screening → earliest possible treatment
- Stem cell transplantation using unrelated donor umbilical cord blood
  - Replacing conventional bone marrow transplant
  - Most primitive stem cells in cord blood have proliferative advantage & ↓ frequent & severe GVHD
  - Early transplant beneficial: MPS I-H (but does not arrest bone dysplasia), MPS VI
    - Best done when < 2 years old & no CNS disease
  - Transplant not beneficial: MPS II, MPS III
    - Fails to arrest encephalopathy
- Enzyme replacement therapy: MPS I, II, VI
  - Enzymes do not cross blood-brain barrier
- Surgery
  - Hydrocephalus: Ventriculoperitoneal shunt (fewer complications prior to stem cell transplant)
  - Corneal opacity: Corneal transplant
  - Spine: C1/C2 stabilization, craniocervical junction decompression, fusion of progressive kyphosis
  - Airway obstruction or eustachian tube obstruction: Tonsillectomy & adenoidectomy
  - Valvular heart disease: Valve replacement
  - Carpal tunnel decompression
  - Hernia repair

DIAGNOSTIC CHECKLIST

Reporting Tips

- Bony features of DM: Get metabolic correlation

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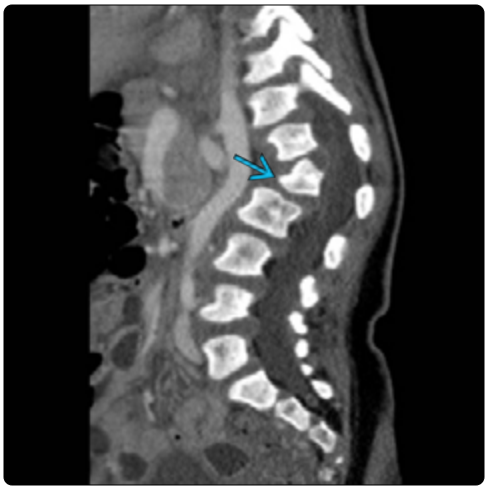
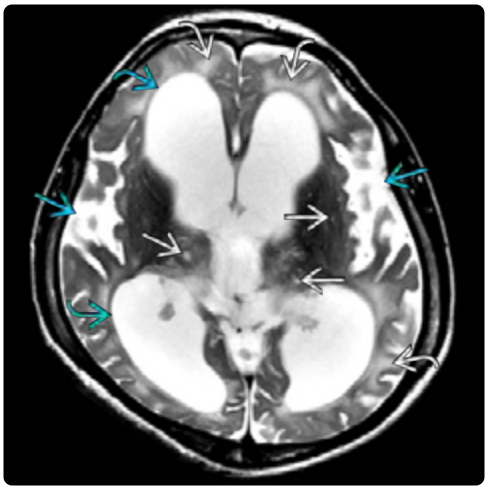
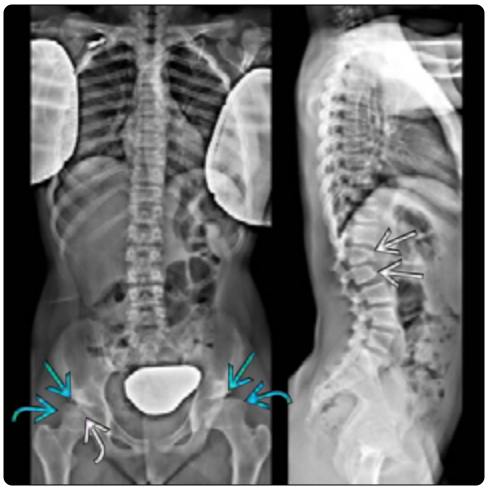
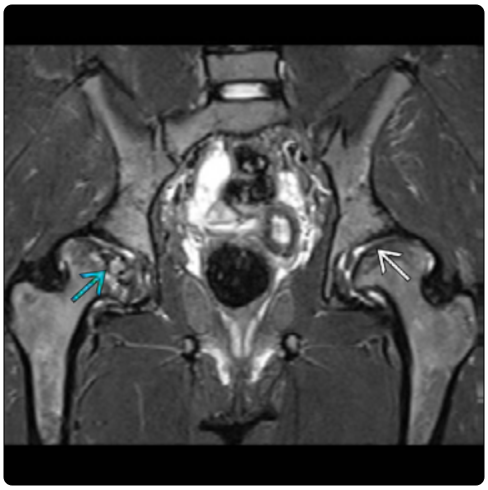


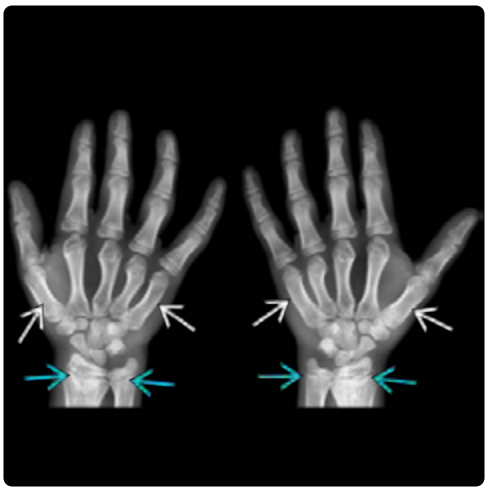


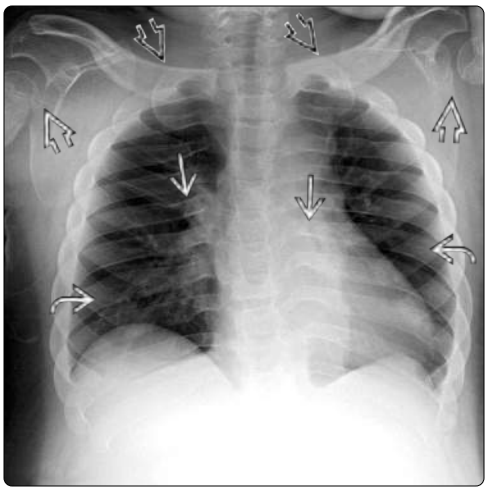
(Left) *Sagittal NECT shows the thoracolumbar spine in a 9-year-old girl with MPS II (Hunter syndrome). There is gibbus deformity with anterior beaking of the L1 vertebral body. (Right) Axial T2 brain MR in the same patient shows enlarged ventricles & sulci. There is also increased signal within the periventricular white matter with a few enlarged perivascular spaces.*





(Left) *Frontal (left) & lateral (right) radiographs of the spine in a 15-year-old girl with MPS VI show focal kyphosis with anterior beaking of the L1 & L2 vertebrae. There is underdevelopment of the acetabula. The femoral heads are broad with lateral uncovering. Lucency & sclerosis in the right femoral head suggests avascular necrosis (AVN). (Right) Coronal T2 FS MR of the pelvis in the same patient confirms AVN & collapse of the right femoral head. Less extensive AVN is also present in the left femoral head.*





(Left) *Bilateral frontal radiographs of the hands in the same girl with MPS VI show short, wide metacarpals. There is also irregularity of the metaphyses of the distal radius & ulna & slight irregularity of the distal radial epiphyses. (Right) AP radiograph in a 10-year-old girl with MPS II (Hunter syndrome) shows thickened clavicles bilaterally. The ribs have a characteristic oar shape with narrowing medially but progressive broadening anterolaterally. The glenoid fossae are poorly formed bilaterally.*

Osteogenesis Imperfecta

KEY FACTS

TERMINOLOGY

- Group of clinically heterogeneous genetic disorders caused by type I collagen abnormalities
- ↑ bone fragility → frequent fractures → malunion & bowing
  - ↑ likelihood of subsequent fractures

IMAGING

- Numerous in utero or perinatal fractures of short, poorly mineralized bones (type II)
- Other types: Multiple fractures in thin, overtubulated long bones + vertebral fractures + osteoporosis
- Radiographs generally sufficient to suggest diagnosis
- CT or MR for axial skeleton complications

PATHOLOGY

- Modified versions of original (1979) Sillence classification generally used
- Original types I-IV based on clinical & radiologic manifestations & inheritance
- Types V-IX subsequently proposed due to mutations outside type I collagen genes ± different phenotype, inheritance
- Associations: Hearing loss (usually as adults), thin skin with subcutaneous hemorrhages, cardiac disease, hernias

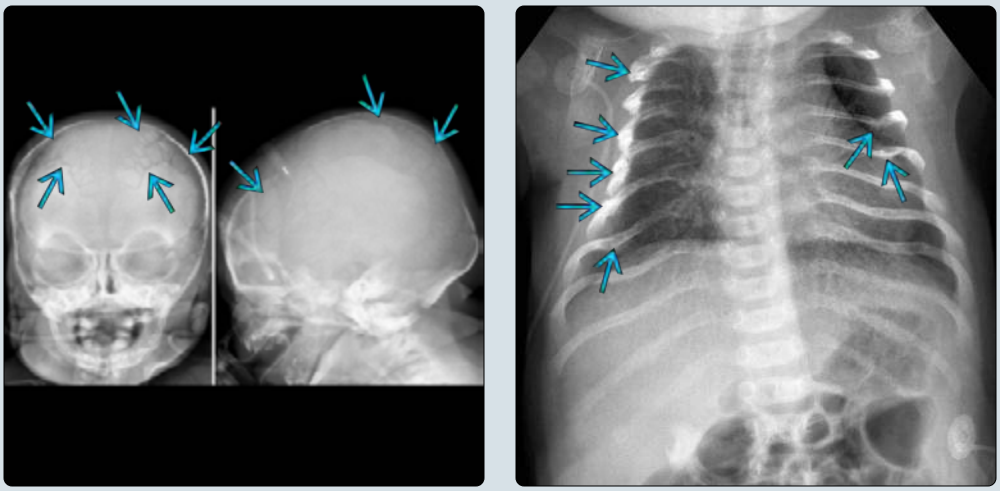
CLINICAL ISSUES

- Severity of osteogenesis imperfecta (OI) (mild → severe)
  - Type I < IV < VI < VII < III < II
- Type II: Lethal
- Type III: Premature death
- Treatment: Intravenous bisphosphonates, surgery

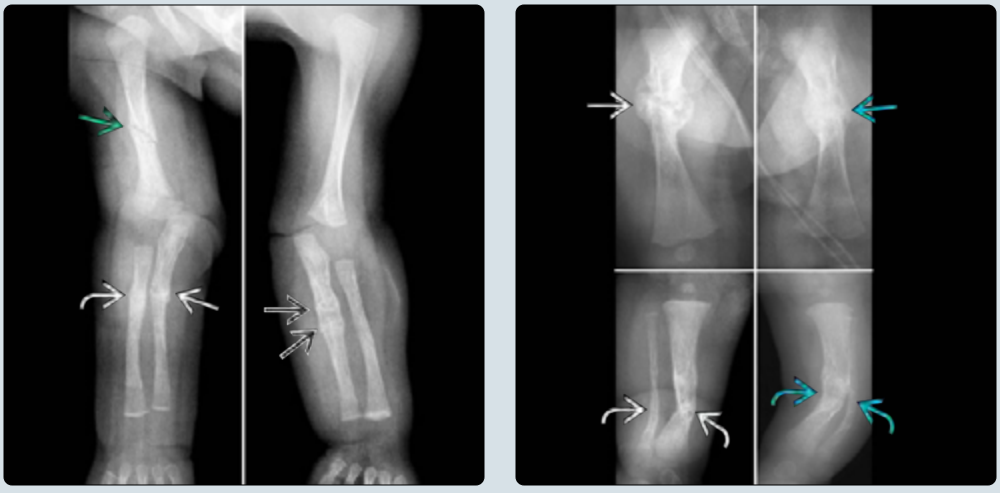
DIAGNOSTIC CHECKLIST

- Wide range of radiographic features across OI types
- Radiographic appearance alone does not classify OI
  - Family history, clinical features, histology, & genetic screening all contribute

(Left) *Frontal & lateral radiographs of the skull in a 1-day-old girl with multiple fractures seen on fetal ultrasound shows multiple (> 10) wormian bones throughout the skull. The patient was subsequently diagnosed with type III osteogenesis imperfecta (OI).* **(Right)** *Frontal radiograph of the chest in the same patient shows deformities & healing fractures of multiple bilateral ribs. Intrauterine fractures can be seen in OI, particularly in types II & III.*



(Left) *Frontal radiographs show the right & left upper extremities in the same patient. There are healing fractures of the right humerus, right radius, & right ulna, as well as multiple healing fractures of the left ulna.* **(Right)** *Frontal radiographs show the right & left lower extremities in the same patient with OI type III. There are healing fractures of the right & left femurs. Bowing deformities of both the right & left tibias/fibulas (secondary to healing fractures) are also noted.*



TERMINOLOGY

Abbreviations

- Osteogenesis imperfecta (OI)

Synonyms

- Van der Hoeve-de Kleyn syndrome, fragilitas ossium, osteopsathyrosis, Lobstein disease, Vrolik disease

Definitions

- Group of clinically heterogeneous genetic disorders caused by type I collagen abnormalities
- ↑ bone fragility → frequent fractures → malunion & bowing
  - Further ↑ likelihood of subsequent fractures
- Dentinogenesis imperfecta: Soft, cracked, discolored teeth

IMAGING

General Features

- Best diagnostic clue
  - Multiple in utero, perinatal fractures of short, poorly mineralized bones (type II)
  - Other types: Multiple fractures in thin, overtubulated long bones + vertebral fractures + osteoporosis

Radiographic Findings

- Radiography
  - Skull
    - Macrocranium
    - Frontal bossing, wide fontanelles, & sutures
    - Multiple wormian bones (≥ 10)
    - Platybasia & basilar impression/invagination
  - Spine
    - Kyphoscoliosis, biconcave or flattened vertebral bodies (platyspondyly), compression fractures
      - High rates of scoliosis progression in types III & IV
      - Lower rate of progression in type I
    - Spondylolisthesis due to pedicle elongation
  - Chest
    - Thorax deformity → respiratory insufficiency
    - Sternum-manubrium bowing (convex outward)
    - Cardiac anomalies
  - Pelvis & hips
    - Coxa vara, protrusio acetabuli
  - Long bones
    - Diaphyseal overtubulation (thin, gracile)
    - Thin cortex
    - Bowing
    - "Popcorn" epiphyses, metaphyses
      - Especially knee & ankle
      - Resolve in adolescence (type III > types I, IV)
    - Hyperplastic callus formation + radioulnar interosseous membrane Ca^{2+} → radial head dislocation in type V
    - Zebra sign with bisphosphonate therapy
      - Sclerotic metaphyseal bands paralleling growth plates
      - Number of bands = number of treatments
  - Feet
    - Flatfoot & skewfoot (forefoot adduction, heel valgus, navicular abduction on talus)

Ultrasonographic Findings

- Fetal US may detect more severe types (II & III)
  - ↓ skull echogenicity of spine & long bones
    - Unusually well-visualized brain
  - Small thorax ± rib fractures
  - Long bones: Short, bowed, multiple fractures ± callus

CT Findings

- Bone CT
  - Can help evaluate complications in axial skeleton
    - Cervical spine: Basilar invagination
    - Temporal bone: Otic capsule abnormalities resembling otospongiosis; stapes crura fractures & footplate fixation
    - Lumbar spine: Pedicle elongation + spondylolisthesis
  - Fetal CT done at some institutions if type II OI suspected
    - ↓ skull ossification, ↓ thorax size, bowed long bones & multiple fractures
- CTA
  - Rare: Aortic, carotid, vertebral artery dissection

MR Findings

- T2WI
  - Assess brainstem, spinal cord in basilar invagination

Nuclear Medicine Findings

- Bone scan
  - ↑ uptake with fractures

Imaging Recommendations

- Best imaging tool
  - Radiographs generally sufficient to suggest diagnosis
  - CT or MR for axial skeleton complications

DIFFERENTIAL DIAGNOSIS

Diseases That Cause Bone Bowing

- Neurofibromatosis type 1
- Fibrous dysplasia
- Hyperparathyroidism
- Rickets
- Many bone dysplasias

Diseases With Fragile Bones

- Bruck syndrome
  - Fragile bones with congenital joint contractures
- Osteoporosis-pseudoglioma syndrome
  - Fragile bones with congenital blindness
- Cole-Carpenter syndrome
  - Fragile bones with cranial synostosis & ocular proptosis
- Idiopathic juvenile osteoporosis
  - No extraskeletal abnormalities

Hyperplastic Bone Formation

- Stress injury
- Myositis ossificans/heterotopic ossification
- Osteosarcoma
- Chronic osteomyelitis

Increased Wormian Bones

- Cleidocranial dysplasia
- Hypophosphatasia

- Hypothyroidism
- Pyknodysostosis
- Menkes syndrome

Child Abuse

- Fractures, retinal hemorrhages, intracranial injury, bruises
- No wormian bones, osteoporosis, or blue sclera

PATHOLOGY

General Features

- Genetics
  - Type I collagen composed of triple helix
    - 2 α-1 chains (*COL1A1* gene)
    - 1 α-2 chain (*COL1A2* gene)
    - Helix structure possible by repeating glycine
    - Additional proteins involved in collagen folding
      - Cartilage-associated protein (*CRTAP* gene)
      - Prolyl-3-hydroxylase-1 (*LEPRE1* gene)
      - Cyclophilin B (*PPIB* gene)
  - > 800 type I collagen mutations known
    - Quantitative disorders: Milder OI
    - Qualitative disorders: More severe OI
  - OI type I: Premature stop codon in *COL1A1*
  - OI types II-IV: Glycine substitutions in *COL1A1*/*COL12*
  - OI types V-VI: Unknown
  - OI type VII-VIII: *LEPRE1* or *CRTAP* mutation
  - OI type IX: *PPIB* mutation
  - Screening for OI biochemical abnormalities
    - DNA sequencing most sensitive
    - Cultured skin fibroblasts test rarely used now
- Associated abnormalities
  - Hearing loss (usually as adults), thin skin with subcutaneous hemorrhages, cardiac disease, hernias

Staging, Grading, & Classification

- Modified versions of original (1979) Sillence classification
  - Original types I-IV based on clinical, radiologic manifestations, & inheritance
  - Types V-IX subsequently proposed due to mutations outside type I collagen genes ± different phenotypes, inheritance
- OI type I: Mild, not deforming
  - Sclerae: Blue in most
  - Fractures: Spectrum of none (in 10%) → numerous; less common after puberty
  - Stature: Often normal, 20% with mild kyphoscoliosis in adults
  - Dentinogenesis imperfecta: Usually not present
- OI type II (subtypes A-C): Perinatal lethal
  - Sclerae: Blue
  - Beaded ribs, "accordion" femurs
  - Myriad of prenatal fractures
  - Poor to no ossification of skull
  - Intracranial hemorrhage, lung hypoplasia → death
- OI type III: Deformation severe
  - Triangular face due to underdeveloped facial bones
  - Sclerae: Gray
  - Kyphoscoliosis, chest deformity, bowed bones
  - Stature: Short
  - Usually wheelchair bound
- OI type IV: Deformation moderate
  - Sclerae: White
  - Bowing of long bones, vertebral fractures
  - Dentinogenesis imperfecta: In some but not all
  - Stature: Short
  - Walk with braces or crutches
- OI type V: Deformation moderate
  - Sclerae: White
  - Stature: Short
  - Dentinogenesis imperfecta: Absent
  - Hyperplastic callus formation
  - Calcified radioulnar interosseous membrane → bilateral anterior radial head dislocation
- OI type VI: Deformation moderate to severe
  - Sclerae: White or faintly blue
  - Stature: Moderately short
  - Vertebral compression fractures
  - Distinct histological features
- OI type VII: Deformation moderate
- OI type VIII: Deformation severe to perinatal lethal

CLINICAL ISSUES

Demographics

- Epidemiology
  - OI type I: Incidence 1:30,000
  - OI type II: Incidence 1:60,000
  - Other types rarer

Natural History & Prognosis

- Severity of OI (mild → severe)
  - Type I < IV < VI < VII < III < II
- Type II: Lethal
- Type III: Premature death

Treatment

- Intravenous bisphosphonates
  - Inhibit osteoclasts, ↑ bone mass/mineralization
  - ↓ bone pain & fractures
  - ↓ rate of scoliosis progression in type III if started < 6 years
  - Jaw osteonecrosis not reported in this population
- Physical therapy, orthopedic surgery

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Wide range of radiographic features across OI types
- Radiographic appearance alone does not classify OI
  - Family history, clinical features, histology, & genetic screening all contribute

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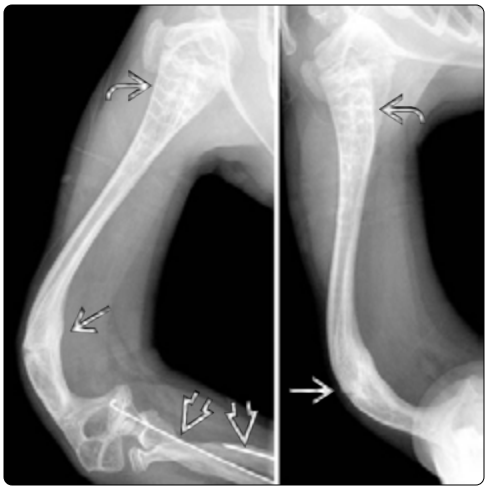
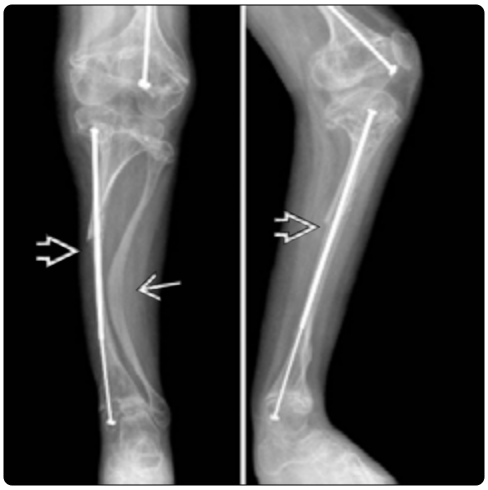
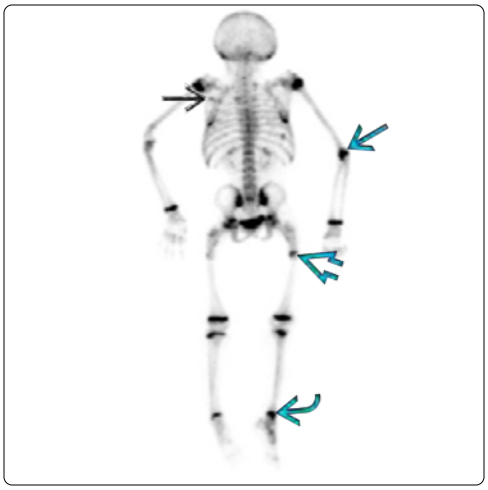
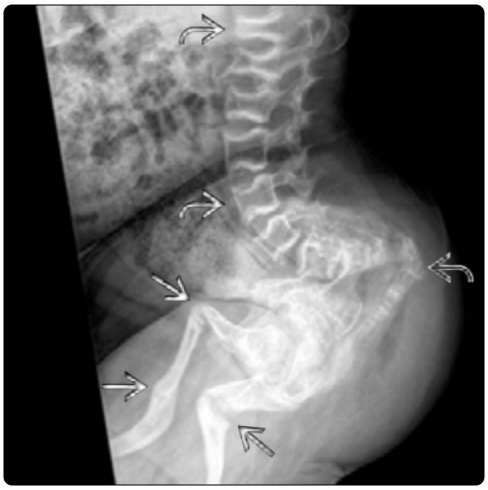


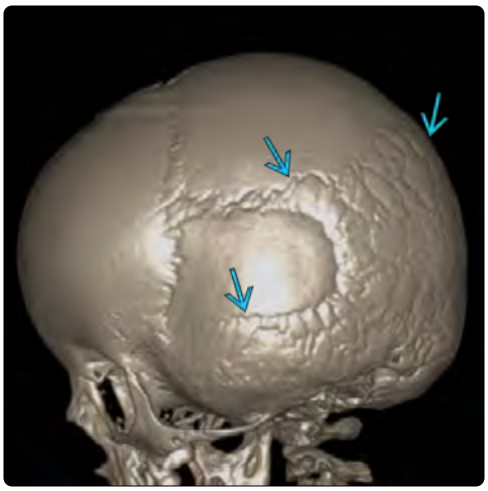


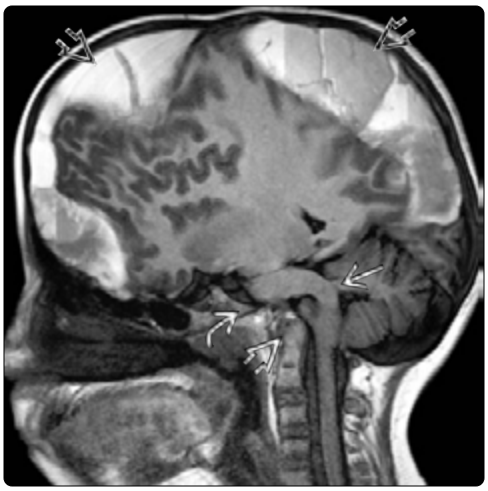
(Left) *AP (left) & lateral (right) radiographs in an 11-year-old OI patient show a gracile, bowed appearance of the right humerus with the apex at the site of a prior fracture. Sclerotic metaphyseal lines are seen in the proximal humerus secondary to episodes of bisphosphonate therapy. Rods are seen in the radius & ulna.* **(Right)** *AP (left) & lateral (right) radiographs of the lower leg in the same patient show osteoporotic, gracile, & bowed bones with a prior fracture & hardware.*





(Left) *Posterior projection bone scan in a 12-year-old girl with OI shows increased uptake in the olecranon, proximal femur, & distal tibia on the right, which were known fractures. Increased uptake in the left scapula was from a fracture that was occult on radiographs.* **(Right)** *Lateral lumbar radiograph in a 16 year old with OI shows biconcave/flattened vertebral bodies, severe sacral kyphosis, bilateral femur fractures with malunion & bowing, & diffusely demineralized.*





(Left) *3D surface-rendered NECT in a boy with OI shows numerous wormian bones throughout the posterior skull. 3D reformats can help differentiate wormian bones from fractures in children with OI.* **(Right)** *Sagittal T1 brain MR in a child with OI shows the dens protruding into the foramen magnum with a horizontal clivus & brainstem angulation. Large, heterogeneous, subdural hematomas are present. Patients with OI are predisposed to intracranial hemorrhages after minor trauma.*

Osteopetrosis

KEY FACTS

TERMINOLOGY

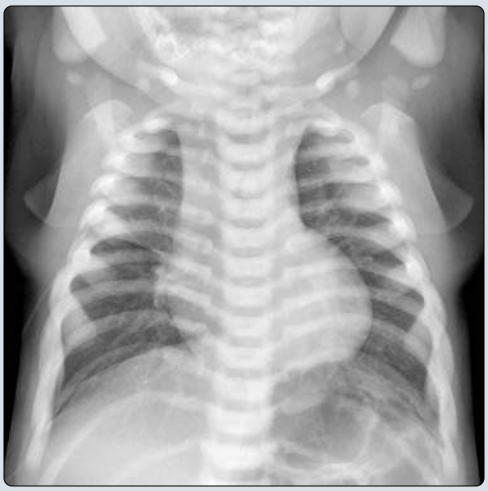
- Heterogeneous group of genetic disorders with ↑ bone density due to impaired bone resorption by osteoclasts
  - Osteopetrosis, autosomal dominant type 1
    - Not true osteopetrosis (poorly understood)
  - Osteopetrosis, autosomal dominant type 2
    - Most common form of osteopetrosis
    - More likely to present incidentally or due to pathologic fracture later in life
  - Osteopetrosis, autosomal recessive (ARO)
    - > 50% due to mutation of *TCIRG1* → infantile malignant osteopetrosis
      - Typically present in 1st year of life with macrocephaly, progressive blindness > deafness, hepatosplenomegaly, recurrent infections, severe anemia, fractures
      - 70% mortality by 6 years without stem cell transplant
    - Other forms of ARO range from mild to severe

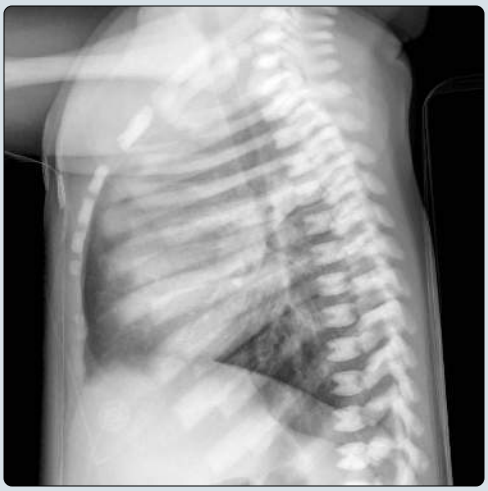
IMAGING

- Generalized osteosclerosis ± alternating radiolucent bands in metaphyses
  - Loss of corticomedullary definition
  - Undertubulated Erlenmeyer flask deformity of metaphyses
  - Bone in bone appearance of flat or small bones
  - ± pathologic fractures
- Thick, dense skull base &/or calvarium
- Vertebrae: Well-defined sclerotic borders at endplates ("sandwich" or "rugger jersey" vertebrae)

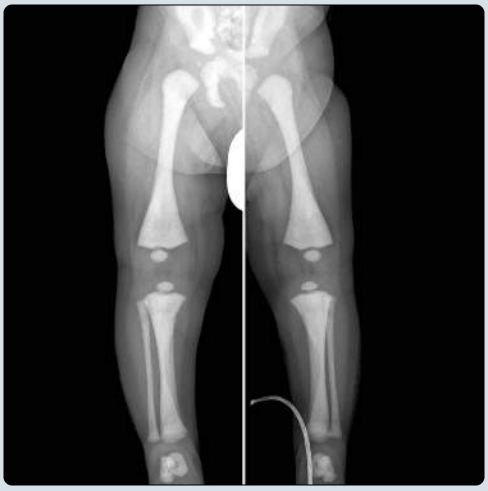
PATHOLOGY

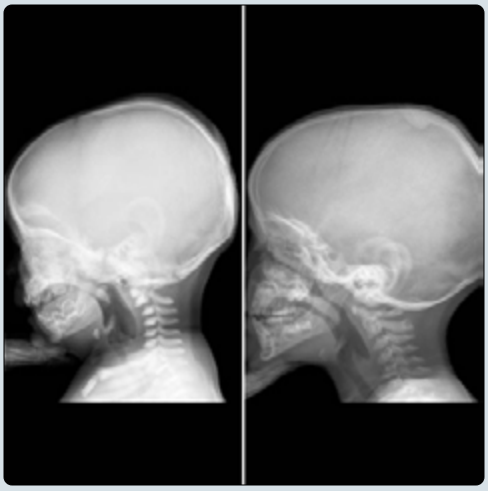
- Defective bone remodeling results in effacement of marrow cavity & abnormal bone structure, impairing
  - Hematopoiesis → anemia, recurrent infections
  - Foraminal development → cranial nerve compression
  - Bone strength → pathologic fractures





(Left) *AP chest radiograph in a 6 month old with fever shows diffusely increased sclerosis of all visualized bones plus thickening of the ribs.* **(Right)** *Lateral radiograph in the same patient again shows the diffuse bony sclerosis & rib expansion. These findings ultimately led to a diagnosis of osteopetrosis.*





(Left) *Bilateral lower extremity radiographs in the same patient show increased sclerosis of all visualized bones. The distal femurs show undertubulation (Erlenmeyer flask deformity) with loss of normal corticomedullary interfaces.* **(Right)** *Lateral skull radiographs in an osteopetrosis patient taken at diagnosis at 6 months of age (left) & 2 years after bone marrow transplant (right) show interval restoration of normal bony mineralization with corticomedullary distinction.*

TERMINOLOGY

Definitions

- Heterogeneous group of genetic disorders characterized by ↑ bone density due to impaired bone resorption by osteoclasts
  - Osteopetrosis, autosomal dominant type 1 (ADO1)
    - Not true osteopetrosis (mechanism poorly understood)
      - *LRP5* mutation → high bone mass
  - Osteopetrosis, autosomal dominant type 2 (ADO2)
    - Albers-Schönberg disease, osteoclast rich
    - Most common form of osteopetrosis
    - *CLCN7* mutation in most → ↓ of chloride channel 7
  - Osteopetrosis, autosomal recessive (ARO)
    - > 50% due to mutation of *TCIRG1* → infantile malignant osteopetrosis (IMO)
    - Other forms of ARO range from mild to severe

IMAGING

General Features

- Location
  - Skull, brain, face
    - Thick, dense skull base &/or calvarium
    - Mandible: Frequent site of osteomyelitis
  - Axial skeleton
    - Vertebrae: Well-defined sclerotic borders at endplates ("sandwich" or "rugger jersey" vertebrae)
    - Pelvis: Bone within bone appearance of flat bones
  - Appendicular skeleton
    - Dense skeleton: Generalized vs. alternating radiolucent bands in metaphyses
      - Loss of corticomedullary definition
      - Bone within bone (endobone) appearance
    - Metaphyseal widening/flaring caused by defective tubular remodeling
      - Erlenmeyer flask deformity of distal femur, proximal humerus & tibia
    - Pathologic fractures
    - Coxa vara
    - ± osteopetrorickets
      - Metaphyseal widening, cupping, & fraying superimposed over findings of osteopetrosis

Nuclear Medicine Findings

- Bone scan
  - ↑ radiotracer at sites of sclerosis
  - ↓ renal activity ("superscan")

DIFFERENTIAL DIAGNOSIS

Bisphosphonate-Induced Osteopetrosis (Acquired Osteopetrosis)

- Metaphyses: Undertubulated with dense transverse bands
- Marginal sclerosis: Epiphyses & vertebral bodies

Chronic Renal Failure

- Vertebral bodies: Sclerosis of endplates (rugger jersey) but less dense & less well defined

PATHOLOGY

General Features

- Genetics
  - Genetic mutations involving osteoclasts
    - 70% of cases accounted for by 12 genes
    - > 50% of IMO due to mutation of *TCIRG1*

Microscopic Features

- Primary spongiosa persists & fills medullary cavity instead of being removed during normal growth
  - Leaves no room for hematopoietic marrow & prevents development of normal mature trabeculae, which normally provide strength to bone

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - ARO/IMO: Impaired hematopoiesis leads to thrombocytopenia, anemia, & infection; cranial nerve compression leads to impaired vision, sensorineural hearing loss
  - ADO: Nontraumatic fractures, cranial nerve palsy, osteoarthritis of hip, mandibular osteomyelitis

Demographics

- Age
  - ARO/IMO typically present < 1 year of age
  - ADO2 typically in late childhood or adolescence

Natural History & Prognosis

- IMO: 70% mortality by 6 years if untreated
- ADO can be completely asymptomatic

Treatment

- Hematopoietic stem cell transplantation: Only cure
  - Addresses bone marrow failure & underlying bone resorption abnormality
    - Up to 88% 5-year survival (depending on transplant type)
    - Resolution of IMO radiographic findings in 6-12 months

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KEY FACTS

TERMINOLOGY

- Synovial inflammation of unknown cause
- Arthritis begins < 16 years of age
- ≥ 6 weeks of symptoms
- International League of Associations for Rheumatology (ILAR) classification of JIA
  - Systemic arthritis: Arthritis of ≥ 1 joint + daily spiking fever for ≥ 3 consecutive days
    - Accompanied by ≥ 1 of following: Evanescent rash, hepatomegaly &/or splenomegaly, serositis
  - Oligoarticular arthritis: Arthritis of < 5 joints in first 6 months of disease
  - Polyarticular arthritis: Arthritis of ≥ 5 joints in first 6 months of disease
    - Rheumatoid factor positive or negative
  - Psoriatic arthritis
  - Enthesitis-related arthritis
  - Undifferentiated or unclassified arthritis

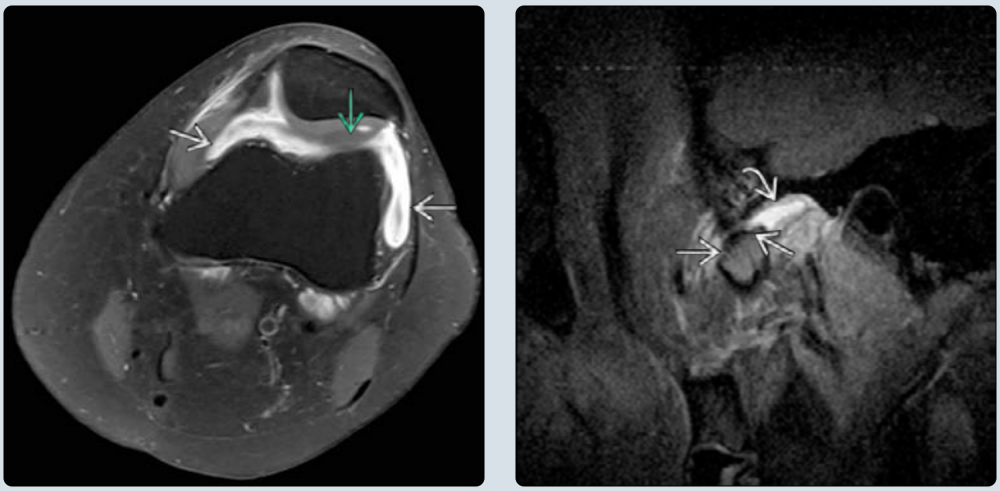
IMAGING

- Radiographs: Characteristic findings seen late in disease
  - Early to intermediate: Osteoporosis, periarticular soft tissue swelling, joint capsule distention (by effusion &/or thickened synovium/pannus), marginal erosions
  - Late: Joint space loss with gradual ankylosis, growth disturbances (e.g., hypoplasia vs. overgrowth)
- MR: Joint effusion with synovial thickening & enhancement, bone marrow edema, cartilage loss ± bone erosions
  - Hypointense rice bodies: Detached fragments of necrotic synovium layering in joint effusion
- US: Compressible hypoechoic joint fluid vs. noncompressible synovial pannus
  - Color/power Doppler for active vs. inactive synovitis

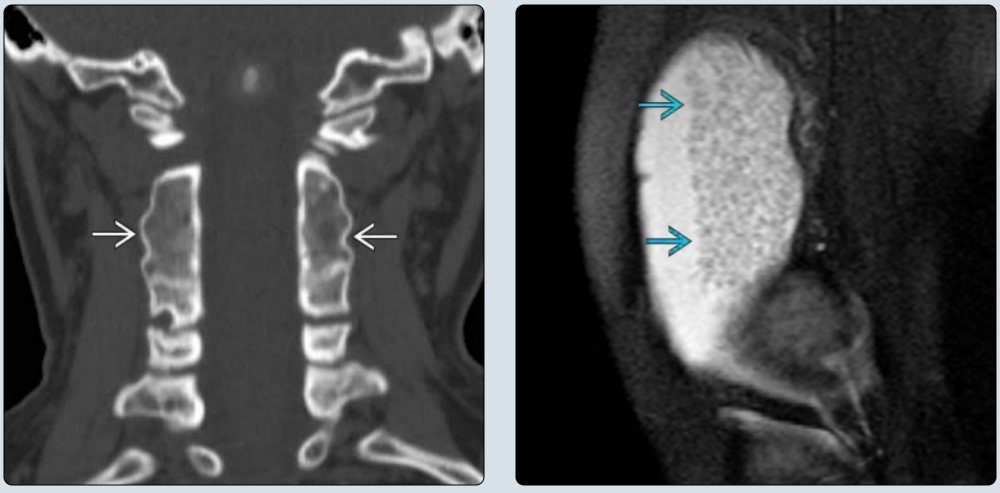
CLINICAL ISSUES

- Presents with joint swelling/effusion, stiffness, pain & tenderness, ↑ warmth
- 1-3 years of age (largest peak); 8-10 years (smaller peak)

(Left) *Axial T1 C+ FS MR in an 18 year old with a history of juvenile idiopathic arthritis (JIA) shows diffuse synovial thickening & enhancement (synovitis) of the knee joint. A small joint effusion reveals the normal absence of synovium overlying the patellar articular cartilage.* **(Right)** *Open mouth sagittal T1 C+ FS MR of the TMJ shows erosions & flattening of the mandibular condyle with synovial thickening & enhancement. The normally visible articular disc is unrecognizable, likely degenerated & torn.*



(Left) *Coronal NECT reformatted image in a 13-year-old JIA patient with neck pain & stiffness shows osseous ankylosis of the apophyseal/facet joints from C2 to C5.* **(Right)** *Sagittal T2 FS MR shows innumerable uniform low-signal nodules (rice bodies) layering dependently within a large knee joint effusion.*



TERMINOLOGY

Abbreviations

- Juvenile idiopathic arthritis (JIA)

Synonyms

- Juvenile rheumatoid arthritis (JRA), juvenile chronic arthritis

Definitions

- Synovial inflammation of unknown cause
- ≥ 6 weeks of joint symptoms
- Begins < 16 years of age
- Most widely used classification developed by International League of Associations for Rheumatology (ILAR)
  - Systemic arthritis (Still disease): 10-20%
    - Arthritis of ≥ 1 joint + daily spiking fever for ≥ 3 consecutive days
    - Accompanied by 1 or more of following: Evanescent rash, hepatomegaly &/or splenomegaly, serositis (pericarditis, pleuritis, peritonitis), generalized lymphadenopathy
    - Can be associated with interstitial lung disease, macrophage activation syndrome, amyloidosis
  - Oligoarticular arthritis: Most common form of JIA (50%)
    - Arthritis of < 5 joints in first 6 months of disease
      - Persistent: < 5 joints affected during entire disease
      - Extended: ≥ 5 joints affected after first 6 months
    - Young girls
    - Large joints: Knee, ankle, elbow
    - Iridocyclitis in up to 30%
  - Polyarticular rheumatoid factor (RF)-positive arthritis
    - Arthritis of ≥ 5 joints in first 6 months of disease
    - ≥ 2 positive tests for RF at least 3 months apart within first 6 months of disease
    - More commonly adolescent girls
    - Typical: Symmetric small joints of hands
  - Polyarticular RF-negative arthritis
    - Arthritis of ≥ 5 joints in first 6 months of disease
    - No characteristic pattern
    - Any age throughout childhood
  - Psoriatic arthritis (< 15%)
    - Arthritis & psoriasis or arthritis & ≥ 2 of following: Dactylitis ("sausage digit"), nail pitting or onycholysis, psoriasis in 1st-degree relative
  - Enthesitis-related arthritis (< 7%)
    - Arthritis & enthesitis, or
    - Arthritis or enthesitis with ≥ 2 of following
      - Sacroiliac joint tenderness &/or inflammatory lumbosacral pain
      - Positive HLA-B27 antigen test
      - Onset of arthritis in male ≥ 6 years old
      - Symptomatic anterior uveitis
      - Presence of 1st-degree relative with ankylosing spondylitis, enthesitis-related arthritis, inflammatory bowel disease with sacroiliitis, Reiter syndrome, or acute anterior uveitis
    - Enthesitis most commonly at Achilles, plantar fascia
  - Undifferentiated arthritis
    - Does not fulfill criteria for any category or fulfills criteria for multiple categories

IMAGING

General Features

- Best diagnostic clue
  - Joint effusion with synovial thickening & enhancement in patient with symptoms suggestive of JIA
- Location
  - Any joint; large joints most common
    - Polyarticular also involves small joints (hands & feet)
    - Cervical spine apophyseal joints rare at presentation but > 1/2 eventually develop involvement

Radiographic Findings

- Most characteristic findings seen late in disease
- General
  - Early to intermediate: Osteoporosis, periarticular soft tissue swelling, joint capsule distention (by effusion & or pannus/thickened synovium), marginal erosions
  - Late: Joint space loss with gradual ankylosis, subluxation, adjacent periosteal reaction, growth disturbances (including shortening/hypoplasia due to premature growth plate closure, enlarged overgrown epiphyses, limb length discrepancy)
- Mandible
  - Micrognathia or unilateral hypoplasia
  - Antegonial notching of mandible (concave undersurface)
- Wrist
  - Squared or angular carpal bones
  - Carpal erosions (which overlap normal indentations)
  - Accelerated maturation
- Hip
  - Coxa valga, coxa magna, protrusio acetabuli
  - Overgrowth of femoral capital epiphysis
  - Abnormal growth of femoral neck
- Knee
  - Squared inferior margin of patella
  - Widened intercondylar notch
- Cervical spine
  - Atlantoaxial subluxation
  - ↓ disc space with narrowed vertebral body height

MR Findings

- Only modality for marrow edema (preerosive finding)
- Most sensitive technique for detecting active inflammation (synovitis & marrow edema)
- T2 FS/STIR
  - Intermediate to hyperintense pannus & hyperintense joint effusion
    - Hypointense rice bodies: Detached fragments of necrotic synovium
  - Patchy hyperintense marrow edema
  - Cartilage loss ± bone erosions
  - Hyperintense tenosynovitis: Most common at extensor tendons of hand & feet, peroneal & posterior tibialis tendons of ankle
  - Hypoplastic menisci & cruciate ligaments
  - Regional lymphadenopathy
- T1 C+ FS
  - Early scanning after contrast administration (< 5 minutes) most sensitive & specific for abnormal synovium
    - Contrast gradually leaks into joint fluid

- Actively inflamed enhancing synovium largely outlines (except where synovium routinely lacking) nonenhancing joint effusion &/or fibrotic/necrotic pannus
  - Early dynamic synovial enhancement correlates with active disease & better reflects treatment response than synovial volumes
- T2* GRE
  - Marginal erosions
- Advanced techniques
  - T2 mapping, T1rho, dGEMRIC: May reveal microstructural cartilage changes prior to conventional sequences

Ultrasonographic Findings

- More sensitive for synovitis than clinical exam
- Distinguishes articular vs. tenosynovial disease
- Compressible hypoechoic joint fluid vs. noncompressible synovial pannus
- Color/power Doppler helpful for active vs. inactive synovitis
- Can visualize some erosions, evaluate cartilage thickness
- Guides injections & biopsies

DIFFERENTIAL DIAGNOSIS

Septic Arthritis

- Majority monoarticular, often from adjacent osteomyelitis
  - *Staphylococcus aureus* most common
- Rapid presentation with adjacent marrow & soft tissue edema favor infection
- Kocher criteria: If specific thresholds met for fever, non-weight-bearing, leukocytosis, ESR, & CRP → strongly favor infected joint
  - Urgent washout required to prevent long-term damage

Transient Synovitis

- Self-limited, typically at hip of 3-6 year olds
- Diagnosis of exclusion: Absence of Kocher criteria

Posttraumatic

- Nonspecific effusion ± synovitis with trauma history

Bone Infarction

- Acute or chronic avascular necrosis may cause synovitis

Synovial Venous Malformation

- Septated fluid-filled channels/cysts infiltrating synovium → hemarthrosis → eventual joint degeneration
- ± phleboliths, fluid-fluid levels, variable enhancement

Hemophilic Arthropathy

- Relevant clinical history usually apparent
- Repetitive hemorrhage into joints with hemosiderin deposition & eventual joint degeneration

Pigmented Villonodular Synovitis

- Monoarticular with hemosiderin deposition

Synovial Chondromatosis/Osteochondromatosis

- Monoarticular disease with multiple intraarticular bodies of variable size; may be calcified or ossified

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Arthritis (1 or more joints): Swelling/effusion, stiffness, pain & tenderness, ↑ warmth
- Other signs/symptoms
  - Fatigue, fevers, weight loss, rash, growth failure
  - TMJs often involved but often asymptomatic

Demographics

- Age
  - 1-3 years (largest peak); 8-10 years (smaller peak)
- Gender
  - M < F overall; systemic M = F
- Epidemiology
  - Most common cause of chronic arthritis in children
  - 16-150 cases/100,000 children

Treatment

- NSAIDs, systemic/local corticosteroids, disease-modifying agents (e.g., methotrexate), biologic agents (monoclonal antibodies or soluble receptors)
- Stem cell transplant in very rare cases

DIAGNOSTIC CHECKLIST

Consider

- Whole-body MR may have prognostic & therapeutic implications as active synovitis may be detected in clinically asymptomatic joints

Image Interpretation Pearls

- Synovitis has many causes; use clinical & imaging clues to narrow differential diagnosis
  - Do not overlook septic arthritis
- In longstanding JIA, difficult to distinguish ongoing active inflammation vs. reactive & degenerative changes

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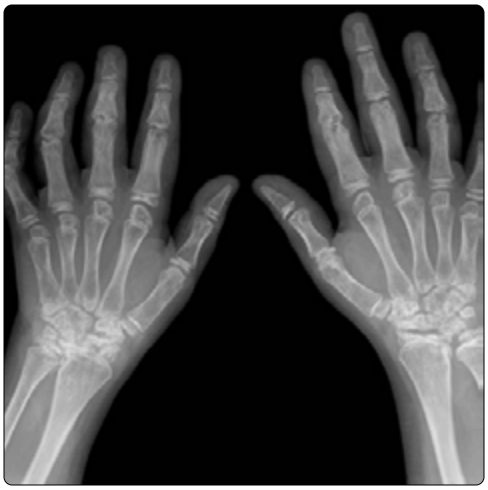
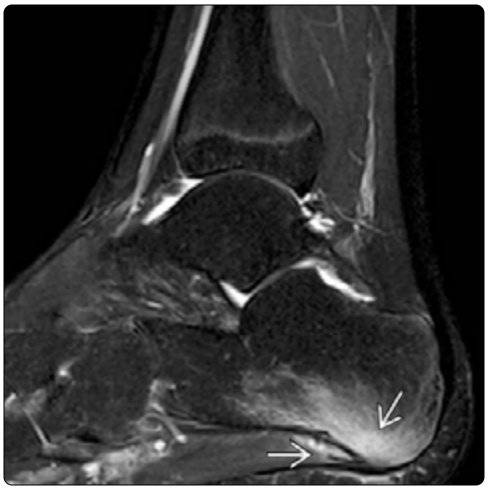
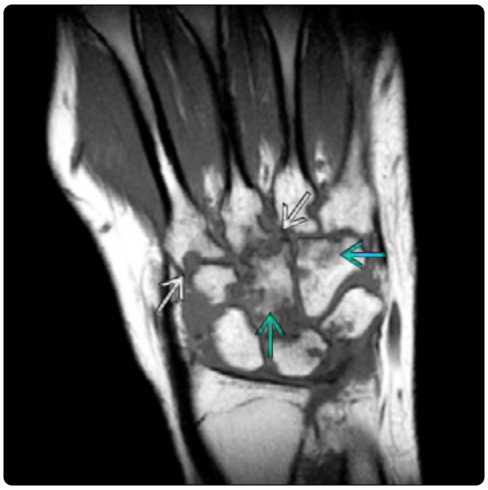
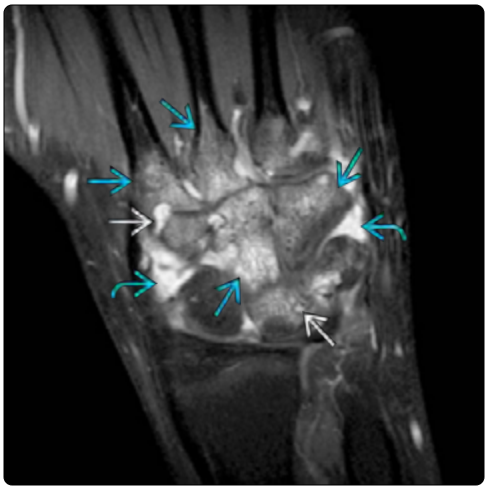


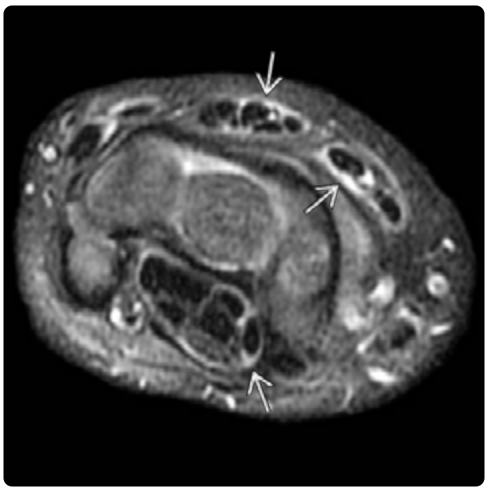


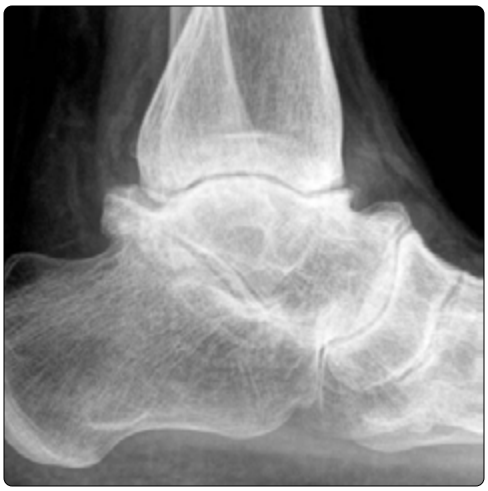
(Left) *PA radiograph in an 11 year old with polyarticular rheumatoid factor-positive JIA shows extensive erosions of the hands & wrists in a fairly symmetric distribution. The erosive changes are greatest at the carpal bones & PIP joints.* **(Right)** *Sagittal T2 FS MR in a 17 year old with a history of JIA shows plantar fascial enthesitis with edema ➡ of the calcaneal bone marrow & adjacent soft tissues at & surrounding the attachment of the plantar fascia.*





(Left) *Coronal T1 MR in a 13 year old with JIA & increased swelling shows extensive erosions ➡ of the carpals & metacarpals. Note the carpometacarpal joint space loss & patchy marrow edema ⇨. (Right) Coronal T1 C+ FS MR in the same teenager shows multiple wrist erosions ➡ & moderate synovial enhancement (or synovitis) ⇨. The marrow edema ⇨ is much more extensive than seen on the precontrast T1 sequence.*





(Left) *Axial T2 FS MR in an 8 year old with JIA & wrist stiffness shows hyperintense signal surrounding the flexor & extensor tendons ➡ of the wrist. There was also mild enhancement at these levels after contrast administration (not shown), consistent with tenosynovitis.* **(Right)** *Lateral radiograph shows degenerative arthritis changes from longstanding JIA, including diffuse joint space narrowing, large spurs, & flattening of the talar dome.*

Dermatomyositis

KEY FACTS

TERMINOLOGY

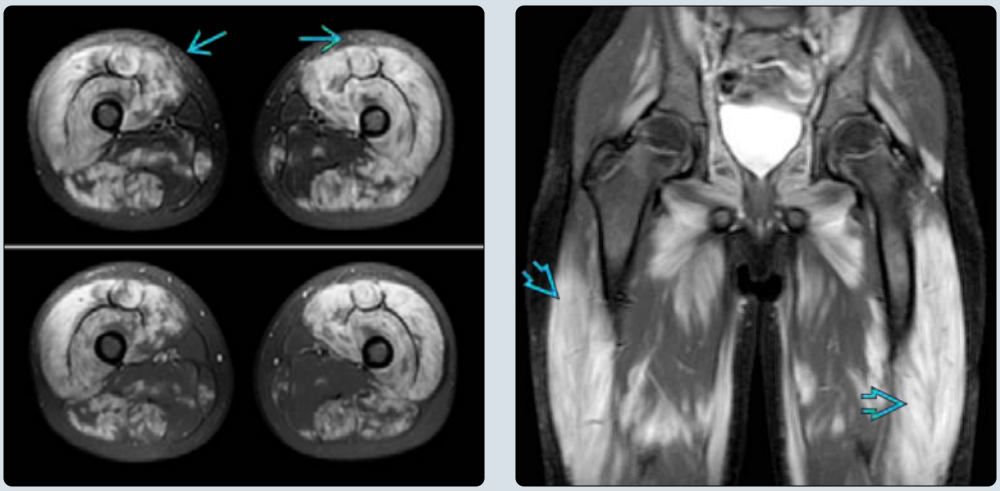
- Juvenile dermatomyositis (JDM): Diffuse nonsuppurative inflammation of striated muscle, subcutaneous fat, & skin
- Most common (85%) juvenile idiopathic inflammatory myopathy

IMAGING

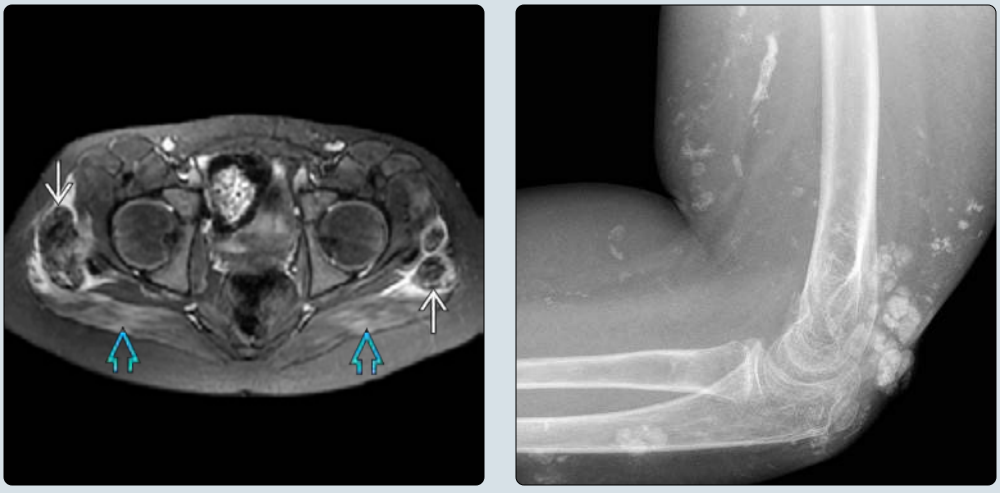
- Patchy to diffusely infiltrative ↑ fluid signal intensity of muscles on STIR or T2 FS MR
  - Symmetric involvement of proximal musculature
    - Thighs > pelvis > shoulders
      - Vastus lateralis & vastus intermedius most common
  - ± fatty muscle infiltration & atrophy chronically; not prominent feature early
- Subcutaneous fat involvement: High specificity (but low sensitivity) for predicting progression to chronic forms
- Soft tissue Ca^{2+} (30-70%)
  - Develops months-years after disease onset, usually periarticular

CLINICAL ISSUES

- Median age of onset of JDM: 7-11 yr
- Most common symptoms
  - Proximal muscle weakness ± tenderness, easily fatigued
  - Rash (heliotrope eyelid rash & Gottron papules classic)
- Other manifestations: Arthritis, pericarditis, pulmonary fibrosis, gastrointestinal symptoms including dysphagia & ulceration, fever, weight loss
- JDM diagnosis classically made by characteristic skin rash + 3 of the following
  - Symmetric proximal muscle weakness
  - ↑ muscle enzymes in serum
  - Characteristic electromyography
  - Characteristic changes on muscle biopsy
- Diagnosis confirmation now more commonly employs characteristic MR findings & autoantibody profiles
- Variable disease course: Up to 73% with active disease > 10 yr after diagnosis; 3% mortality



(Left) *Axial T2 FS (top) & T1 C+ FS (bottom) MR images of the thighs in a 4-year-old girl with proximal weakness & a rash show extensive, symmetric, infiltrative high T2 signal intensity & enhancement throughout the anterior > posterior compartment muscles with relative sparing medially. Abnormal reticular foci are also seen in the subcutaneous fat. **(Right)** Coronal STIR MR in the same patient shows the extent of symmetric muscular edema/inflammation throughout the pelvis & thighs, typical of JDM.*



(Left) *Axial T2 FS MR in the same patient 4 years later shows the interval development of multiple ovoid, predominantly low signal intensity intramuscular masses laterally, consistent with calcinosis. Patchy inflammation remains in the gluteus maximus muscles. **(Right)** Lateral radiograph in a teenager with JDM & limited elbow motion shows innumerable soft tissue Ca^{2+}, predominantly in a periarticular location. Note the abundance of Ca^{2+} along the extensor surface of the joint.*

TERMINOLOGY

Definitions

- Juvenile dermatomyositis (JDM): Diffuse nonsuppurative inflammation of striated muscle, subcutaneous fat, & skin
- Most common (85%) juvenile idiopathic inflammatory myopathy

IMAGING

General Features

- Best diagnostic clue
  - Patchy to diffusely infiltrative ↑ fluid signal intensity of muscles on MR, symmetric in anterior thighs & pelvis
- Location
  - Proximal musculature: Thighs > pelvis > shoulders
    - Most common: Vastus lateralis & vastus intermedius
  - ± pharyngeal striated muscles → dysphagia

Radiographic Findings

- Soft tissue Ca^{2+} (30-70% of JDM patients)
  - Develops from months to years after disease onset
  - Amorphous, globular, or sheet-like; usually periarticular

CT Findings

- High-resolution NECT of lung
  - Interstitial lung disease, chest wall Ca^{2+}, airway disease
  - ≥ 50% of JDM patients have abnormal pulmonary function tests

MR Findings

- Streaky or infiltrative ↑ fluid signal intensity (edema) of affected muscles, most conspicuous on T2 FS or STIR
- ± elongated, crescentic, & reticular foci of fluid signal intensity along fascial planes & in subcutaneous fat
  - Subcutaneous fat involvement: High specificity (but low sensitivity) for predicting progression to chronic forms
    - May lead to calcinosis or lipodystrophy (focal or generalized subcutaneous fat loss)
- ± chronic fatty muscle atrophy (not predominant feature)
- Whole-body STIR MR: Potential screening for disease extent &/or therapy-induced avascular necrosis (AVN)

Imaging Recommendations

- Best imaging tool
  - MR: Axial T2 FS or STIR

DIFFERENTIAL DIAGNOSIS

Symmetric Muscle Abnormalities

- Delayed onset of muscle soreness
  - Preceding history of intense exercise
- Other idiopathic inflammatory myopathies
  - Juvenile connective tissue disease myositis
  - Juvenile polymyositis
- Muscular dystrophies (MD)
  - Duchenne MD (most common; occurs in male patients)
    - Progressive fatty infiltration of proximal musculature
- Longstanding static or progressive neurologic processes
  - Cerebral palsy, myelomeningocele, polio, etc.
  - Often lead to muscle atrophy & bony abnormalities

Asymmetric or Unilateral Muscle Abnormalities

- Infection
  - Primary pyogenic or viral myositis
  - Secondary to other sources of infection (adjacent osteomyelitis, septic emboli, etc.)
- Injury
  - Focal tear, hematoma, myositis ossificans
  - Rhabdomyolysis from crush injury, compartment syndrome, medication, or primary infarction
  - Denervation
  - Radiation
- Eosinophilic fasciitis
- Infiltrating vascular anomaly

PATHOLOGY

Staging, Grading, & Classification

- Classifications of JDM disease patterns include
  - Limited, chronic nonulcerative, chronic ulcerative (involves skin & gastrointestinal tract)
  - Monocyclic, polycyclic, chronic continuous

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Symmetric proximal muscle weakness ± tenderness; easily fatigued
- Other signs/symptoms
  - Heliotrope rash & Gottron papules classic; may occur prior to, during, or after muscle weakness symptoms
  - Other manifestations: Arthritis, pericarditis, pulmonary fibrosis, gastrointestinal symptoms including dysphagia & ulceration, fever, weight loss
- JDM diagnosis classically made by characteristic skin rash + 3 of the following
  - Symmetric proximal muscle weakness
  - ↑ muscle enzymes in serum
  - Characteristic electromyography
  - Characteristic changes on muscle biopsy
- Diagnosis confirmation now more commonly employs combination of characteristic MR findings & autoantibody profiles in correct clinical setting

Natural History & Prognosis

- Variable course of disease
  - Up to 73% with active disease > 10 yr after diagnosis
- Complications: Calcinosis, contractures, AVN (steroids)
- Mortality up to 3%

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Chronic Recurrent Multifocal Osteomyelitis

KEY FACTS

TERMINOLOGY

- Idiopathic disorder with multifocal nonpyogenic inflammatory bone lesions & relapsing/remitting course
- Associated with other inflammatory conditions (e.g., psoriasis, inflammatory bowel disease)

IMAGING

- Most commonly at periphyseal long bone metaphyses or metaphyseal-equivalent regions
  - Tibia, femur, spine, pelvis most commonly involved
  - Most common disease to involve medial clavicle
- Radiographs: Features may be mixed at presentation
  - Acute: Lytic lesion (usually in metaphysis near physis)
  - Late: Progressive adjacent sclerosis
  - Recurrence: New lytic areas or periosteal reaction
- MR with contrast
  - Edema & enhancement of lesion & surrounding marrow
  - ± soft tissue & periosteal inflammation but no abscess
  - ± reactive adjacent joint effusion/synovitis

TOP DIFFERENTIAL DIAGNOSES

- Bacterial osteomyelitis
- Leukemia
- Ewing sarcoma
- Langerhans cell histiocytosis

PATHOLOGY

- Nonspecific inflammatory changes
- Cultures negative for organisms

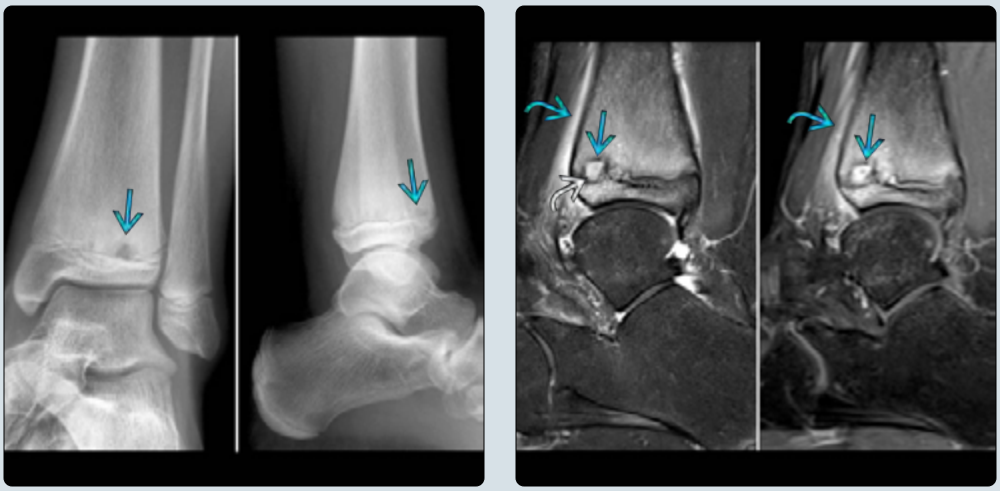
CLINICAL ISSUES

- Most common: 9-14 years
- Typically presents with symptoms at 1 site
  - Most (not all) develop other symptomatic sites

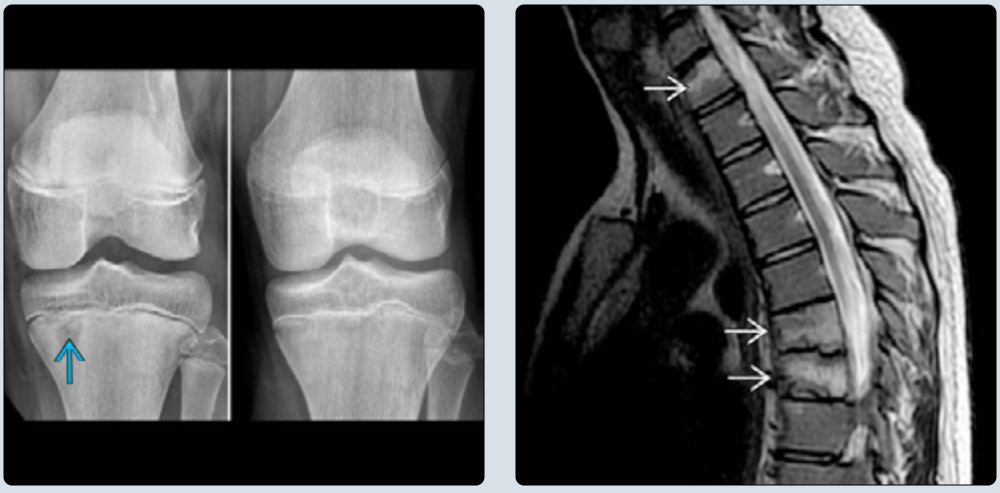
DIAGNOSTIC CHECKLIST

- Whole-body MR or bone scan to screen asymptomatic sites
- Report physeal involvement (due to risk of premature physeal closure)

(Left) *Frontal (left) & lateral (right) ankle radiographs in a 12-year-old girl with left ankle & knee pain show a lytic lesion in the distal tibial metaphysis with mild adjacent sclerosis.* **(Right)** *Sagittal T2 FS (left) & T1 C+ FS (right) MRs in the same girl show a focal metaphyseal lesion with increased T2 signal & enhancement. There is physeal extension with adjacent metaphyseal, epiphyseal, & periosteal edema.*



(Left) *AP knee radiograph in the same girl at presentation (left) shows a focal lytic lesion in the proximal left tibial metaphysis with mild adjacent sclerosis. Biopsy with culture grew no organisms. After treatment with a TNF-a antagonist, her symptoms & tibial lesion resolved (right), typical of CRMO.* **(Right)** *Sagittal T2 MR in a patient with Crohn disease & rashes shows increased signal within the T1, T6, & T7 vertebral bodies with loss of height at T6 & T7. The patient was ultimately diagnosed with CRMO.*



TERMINOLOGY

Abbreviations

- Chronic recurrent multifocal osteomyelitis (CRMO)

Definitions

- Idiopathic disorder with multifocal nonpyogenic inflammatory bone lesions & relapsing/remitting course

Associated Syndromes

- May be in spectrum of diseases with SAPHO (**s**ynovitis, **a**cne, **p**ustulosis, **h**yperostosis, **o**steitis)
- Associated with other inflammatory conditions [e.g., psoriasis, inflammatory bowel disease (IBD)]

IMAGING

General Features

- Best diagnostic clue
  - Multifocal lytic & sclerotic lesions of periphyseal metaphyses/metaphyseal equivalents or medial clavicle
- Location
  - Multiple sites of involvement over course of disease: ~ 80%
    - Can be bilateral with relatively symmetric distribution
  - Most common: Long bone metaphyses (periphyseal)
    - Lower extremity > upper extremity (3:1)
    - Tibia, femur, spine, pelvis most commonly involved
  - Additional locations
    - Metaphyseal equivalent bones (next to growth cartilage)
    - Clavicle (especially medial), fibula, ribs, mandible, sternum, scapula, hands, & feet

Radiographic Findings

- Early: Lytic lesion (usually metaphyseal)
- Late: Progressive adjacent sclerosis
  - Chronic lesions may be mixed lytic/sclerotic or purely sclerotic
  - Hyperostosis with cortical thickening
- Recurrence
  - New lytic area &/or periosteal reaction

MR Findings

- T1WI
  - ↓ signal within metaphysis adjacent to growth plate
- T2WI FS/STIR
  - Focal well-circumscribed periphyseal metaphyseal lesion of ↑ signal
    - ± breach of physis (which can lead to subsequent growth arrest)
  - Less pronounced poorly defined ↑ signal of bone marrow in adjacent metadiaphysis & epiphysis
    - Epiphyseal signal may be out of proportion to physeal involvement
  - ↑ signal along periosteum
  - ↑ signal in overlying soft tissues
  - ↑ signal within adjacent joints
    - Reactive effusion &/or synovitis
- T1WI C+ FS
  - Diffuse enhancement of bone & soft tissue abnormalities
    - No drainable soft tissue or subperiosteal collection
  - ± small foci of peripheral enhancement in bone (sterile intraosseous abscess or necrosis)
  - ± synovial thickening & enhancement in adjacent joints
- Whole-body MR
  - Techniques differ but typically include coronal STIR ± T1, DWI; ± sagittal STIR for vertebral involvement
  - Detects clinically asymptomatic lesions
  - At long-term follow-up (≥ 10 years), clinically asymptomatic patients may still have lesions

Nuclear Medicine Findings

- Bone scan
  - Tc-99m whole-body bone scan can also evaluate for asymptomatic lesions: ↑ radiotracer uptake

Imaging Recommendations

- Best imaging tool
  - Imaging work-up should begin with radiographs
  - Targeted MR helps confirm diagnosis & define extent of symptomatic disease
  - Whole-body imaging (bone scan or MR) to evaluate for additional asymptomatic sites
    - May play role in long term follow-up

DIFFERENTIAL DIAGNOSIS

Osteomyelitis

- Single site most common; medial clavicle rare
- Typically presents before lytic lesions occur
- Soft tissue &/or subperiosteal abscesses common

Ewing Sarcoma

- Permeative bone tumor with aggressive periosteal reaction
- Sharp interface between tumor & normal marrow on T1 MR
- Relatively large soft tissue mass common

Langerhans Cell Histiocytosis

- "Punched-out" lytic lesion classic
- Homogeneously enhancing soft tissue mass emanating from bone
- Marked surrounding marrow edema with less pronounced soft tissue findings
- Peak age: 5-10 years

Leukemia

- Osteoporosis, lucent metaphyseal bands ± subtle permeative lytic lesions & aggressive periosteal reaction
- Diffuse replacement of fatty marrow on T1 MR
- Peak age: 2-10 years

PATHOLOGY

General Features

- Etiology
  - Unknown, genetic & autoinflammatory causes suggested
  - Diagnosis of exclusion
- Genetics
  - Susceptibility locus identified at 18q21.3-22
  - Genetic etiology supported by patients with Majeed syndrome
    - Triad: CRMO, congenital dyserythropoietic anemia, inflammatory dermatosis

- Mutation of *LPIN2* gene
- Autosomal recessive

- Associated abnormalities
  - Dermatologic disorders
    - Palmoplantar pustulosis
    - Psoriasis
  - Autoinflammatory disorders
    - Takayasu arteritis
    - Wegener granulomatosis
  - Gastrointestinal disorders
    - IBD: Crohn disease & ulcerative colitis
  - Genetic disorders
    - Majeed syndrome
  - Others
    - Enthesis-related arthritis
    - SAPHO syndrome
      - Thought to be adult equivalent of CRMO but with some distinct differences
      - Etiology unknown (genetic, immunologic & bacterial mechanisms proposed)
      - Mean age: 28 years
      - Chest wall & pelvic disease predominates (especially sternoclavicular & 1st sternocostal joint)
      - Arthritis of peripheral joints
      - Ossification of ligaments & entheses
      - Associated dermatologic disorders more common

Gross Pathologic & Surgical Features

- Microscopic features
  - Nonspecific inflammatory changes with granulocytic infiltration
  - Acute: Polymorphonuclear leukocytes, osteoclastic bone resorption, ± multinucleated giant cells
  - Chronic: Lymphocytes, plasma cells, histiocytes with occasional granulomas, ↑ osteoblastic activity
  - Acute to chronic findings may be found in single lesion
  - Appearance of biopsy specimen may not correlate with clinical time course of disease
- Cultures negative for organisms

Laboratory Tests

- Nonspecific laboratory markers
  - Mildly elevated C-reactive protein (CRP) & erythrocyte sedimentation rate
  - Normal white blood cell count

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Typically starts with 1 symptomatic site: Pain, tenderness, swelling, limited range of motion
    - Most (not all) develop other symptomatic sites
  - May be symptomatic days to years prior to presentation
    - Often insidious with vague, nonspecific symptoms
- Other signs/symptoms
  - Systemic signs may be present but uncommon
    - Fever, weight loss, lethargy
  - Associated with dermatologic disorders & IBD

Demographics

- Age
  - Most commonly 9-14 years old
  - Reported as young as 6 months & as old as 55 years
- Gender
  - Female predominance
    - M:F ratio = 1:2.1

Natural History & Prognosis

- Most patients undergo spontaneous resolution over months to years
  - Symptoms up to 25 years following initial diagnosis

Treatment

- 1st-line treatment: NSAIDs
- Alternatives: Bisphosphonates, TNF-α antagonists, methotrexate

DIAGNOSTIC CHECKLIST

Consider

- Lack of improvement on antibiotics & presence of associated inflammatory disorder lend support to imaging findings suggestive of CRMO
- Bone scan or whole-body MR to evaluate multifocality
- Medial clavicular involvement strongly suggestive of CRMO

Image Interpretation Pearls

- Carefully inspect metaphyses on radiographs in patients with extremity pain
- ± soft tissue inflammation & joint effusion/synovitis
- ± small intraosseous fluid collections or necrotic bone
  - Soft tissue abscess, sequestra, or presence of fistula suggests bacterial osteomyelitis

Reporting Tips

- Suggest diagnosis along with appropriate differential considerations if imaging appearance characteristic
- Include presence/absence of transphyseal involvement
  - ↑ risk of growth disturbance

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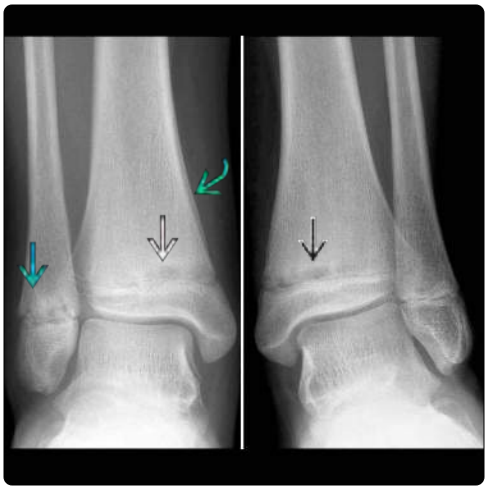
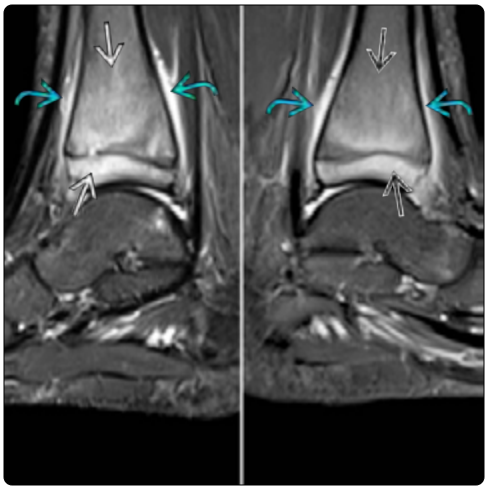
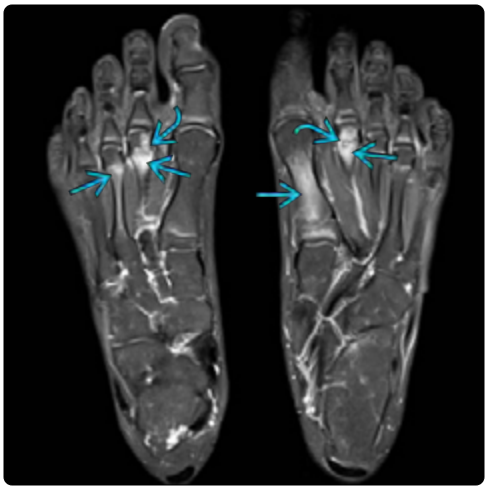
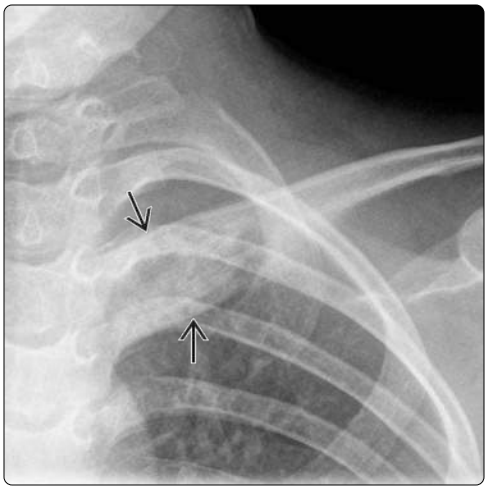


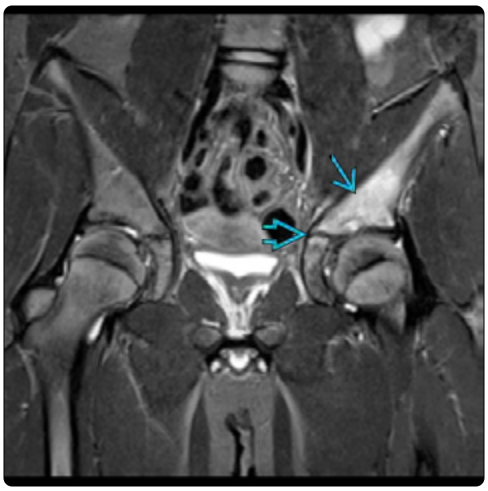


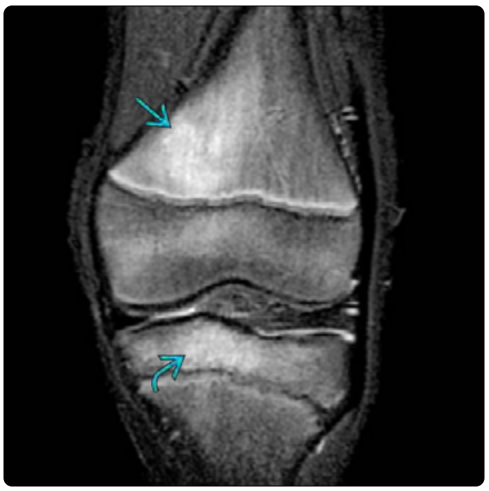
(Left) *Frontal radiographs of the ankles in a 13-year-old girl with bilateral ankle & foot pain show lucent lesions with mild adjacent sclerosis in the distal right & left tibial & right fibular metaphyses. Mild periosteal reaction is present along the distal right tibia.* **(Right)** *Sagittal T2 FS MRs of the ankles in the same girl show extensive marrow abnormalities within the right & left distal tibias. Periosteal edema is also seen bilaterally.*





(Left) *Long axis T2 FS MRs of the feet in the same girl show edema of several metatarsals. Note the transphyseal extension in both 2nd metatarsals. Biopsy & culture of the distal tibias revealed no organism. The lesions & history are typical of CRMO.* **(Right)** *AP radiograph of the left clavicle in an 8-year-old girl shows a mixed lytic & sclerotic lesion expanding the medial clavicle. CRMO is the most common disorder of the medial clavicle.*





(Left) *Coronal T2 FS MR of the pelvis in a 12-year-old boy with left hip & knee pain shows poorly defined hyperintense signal within right iliac bone. This region of the iliac bone is considered a metaphyseal equivalent due to its proximity to the triradiate growth cartilage.* **(Right)** *Coronal T2 FS MR of the left knee in the same boy shows poorly defined signal abnormalities within the distal femoral metaphysis & proximal tibial epiphysis. The patient was ultimately diagnosed with CRMO.*

Rickets

KEY FACTS

TERMINOLOGY

- Failure to mineralize cartilage & osteoid at growth plates (physes) of immature skeleton in setting of low ion concentrations (calcium or phosphorous)
  - Lack of phosphate (ultimate problem in all rickets) → failure of chondrocyte apoptosis → disruption of endochondral ossification
- Most common cause: Nutritional vitamin D deficiency

IMAGING

- Loss of normally thin, dense, well-defined zone of provisional Ca^{2+} at interface of physis & metaphysis
- Metaphyseal cupping, splaying, & fraying with lengthening ("widening") of adjacent radiolucent physis
- All sites of endochondral bone formation affected
  - Most pronounced at long bone metaphyses with greatest linear growth
- Sites of membranous bone growth less affected

TOP DIFFERENTIAL DIAGNOSES

- Newborn stress demineralization
- Leukemia or metastatic neuroblastoma
- Congenital syphilis
- Metaphyseal fracture of child abuse
- Physeal stress injury
- Physeal fracture
- Metaphyseal chondrodysplasias
- Hypophosphatasia

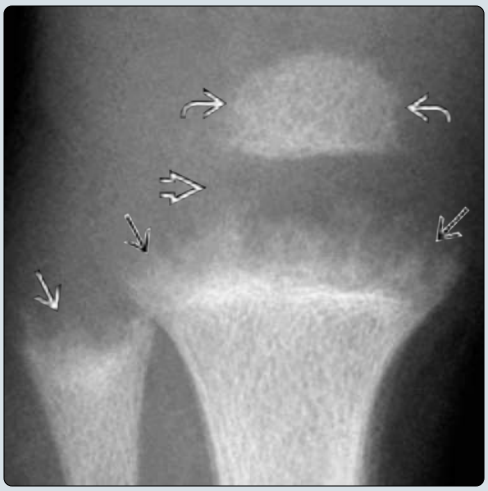
CLINICAL ISSUES

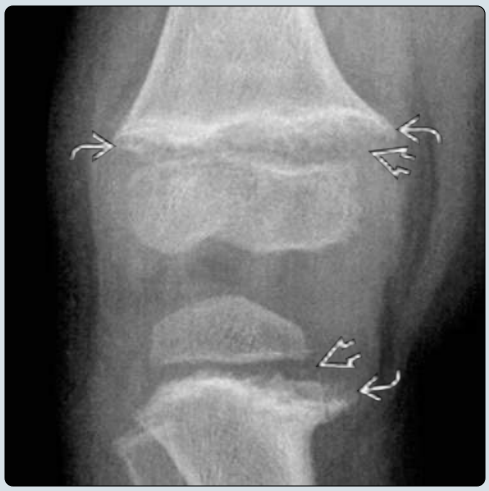
- Peak age for dietary rickets: 3 months to 2 years
- Most rickets responds to vitamin D therapy ± calcium

DIAGNOSTIC CHECKLIST

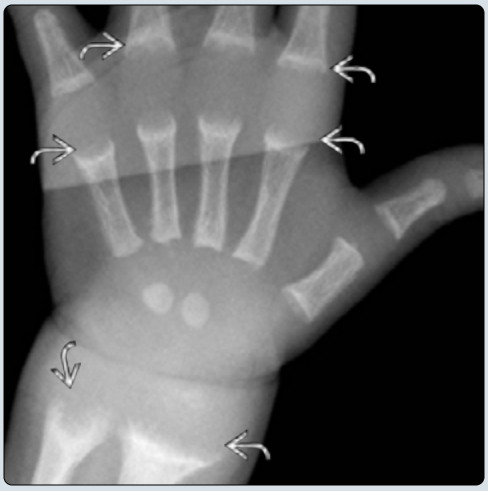
- Unexplained physeal widening at one site (i.e., without acute or chronic trauma) merits survey at other sites of rapidly growing long bones (knees, wrists)
  - Multisite physeal widening → get metabolic work-up

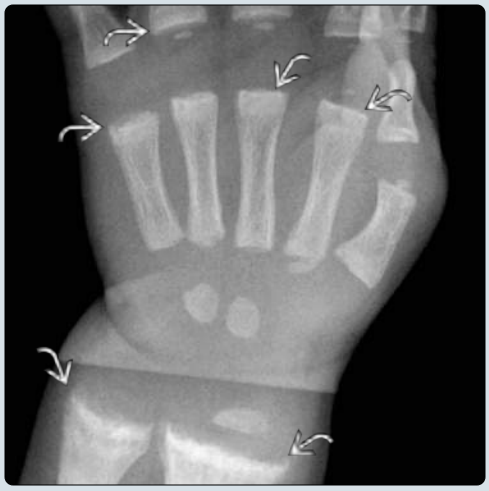
(Left) *AP radiograph in a young child with rickets shows fraying, cupping, & splaying of the proximal tibial ⇨ & fibular ⇨ metaphyses. There is loss of the normally dense, well-defined zones of provisional calcification (ZPC) that should be seen at the metaphyseal-physeal junctions. The radiolucent tibial physis is lengthened ⇨. The epiphyseal ZPC is also lost ⇨ with poor bony margin definition.* **(Right)** *AP knee radiograph in a 3 year old with rickets shows typical metaphyseal fraying, cupping, & splaying ⇨, as well as physeal lengthening ⇨.*





(Left) *PA wrist radiograph in this young child with rickets shows fraying, cupping, & splaying of every visualized metaphysis ⇨. The normal ZPCs are lost.* **(Right)** *PA radiograph in the same child 9 weeks after initiation of therapy shows that endochondral ossification has resumed with improved bone formation at all the growth centers. The visualized metaphyses ⇨ are normalizing in shape & density as compared to the prior study.*





TERMINOLOGY

Definitions

- Failure to mineralize cartilage & osteoid at growth plates (physes) of immature skeleton in setting of low ion concentrations
  - Initial deficiency may be considered as calcipenic or phosphopenic
  - Lack of phosphate becomes ultimate problem
    - Underlying factor in all rickets: Failed apoptosis of hypertrophic chondrocytes
      - Key enzyme (caspase-9) dependent on phosphorylation
      - Failure of apoptosis disrupts endochondral ossification
- Most common cause: Nutritional vitamin D deficiency
  - ↓ dietary vitamin D &/or ↓ exposure to sunlight
  - Primary calcium deficiency uncommon
- Many other causes, including
  - Prematurity
    - < 28 weeks gestational age, birth weight < 1,000 g
    - 80% of fetal bone mineralization occurs in 3rd trimester
      - Typical neonatal nutrition not sufficient
      - Exacerbated by medications used for chronic lung disease of prematurity, including steroids & calcium-excreting diuretics
  - Prenatal factors
    - Maternal medical problems resulting in severe vitamin D deficiency
  - Liver disease: ↓ formation of 25-hydroxy vitamin D; unconjugated bilirubin interferes with osteoblasts
    - Chronic liver disease
    - Anticonvulsant therapy
  - Malabsorption: Binding with malabsorbed fatty acids → ↓ calcium & vitamin D absorption
  - Hereditary hypophosphatemia: ↑ FGF-23 signaling
  - Renal tubular disorders: Various impairments of renal tubular resorption of phosphate & 1-α-hydroxylation of 25-hydroxy vitamin D
    - Vitamin D-resistant rickets
    - Fanconi syndrome
    - Lowe (oculocerebrorenal) syndrome
    - Chemotherapy: Ifosfamide renal-tubule toxicity
  - Oncogenic rickets: Rare paraneoplastic syndrome
    - Nonossifying fibroma
    - Hemangiopericytoma
    - Osteoblastoma
    - Linear sebaceous nevus syndrome
  - Chronic renal disease: Renal osteodystrophy with secondary hyperparathyroidism
    - ↓ glomerular function → phosphorus retention → hypocalcemia
    - Tubular dysfunction → ↓ synthesis of 1,25-dihydroxy vitamin D → hypocalcemia
    - Hypocalcemia → hyperparathyroidism

IMAGING

General Features

- Best diagnostic clue
  - Loss of normally thin, dense, well-defined zone of provisional Ca^{2+}(ZPC) at interface of physis & metaphysis
  - Metaphyseal cupping, splaying, & fraying with lengthening ("widening") of adjacent physis
- Location
  - All sites of endochondral bone formation affected
    - Most pronounced at long bone metaphyses with greatest linear growth
      - Distal femur, proximal tibia, proximal humerus, distal radius
  - Sites of membranous bone growth less affected

Radiographic Findings

- Axial skeleton
  - Costochondral junctions: Cupping, widening of anterior rib ends ("rachitic rosary")
  - Spine: Scoliosis, biconcave vertebral bodies
  - Pelvis: Inward migration of sacrum, acetabula → triradiate appearance; causes dystocia in adulthood
- Appendicular skeleton
  - Epiphyses: Loss of dense outline (ZPC) of secondary ossification centers resulting in irregular margins
  - Physes: Lengthening (longitudinal "widening") of radiolucent growth plates (due ↑ unossified cartilage)
  - Metaphyses: Fraying, splaying/transverse widening, & cupping with loss of normal dense ZPC
    - Loss of metaphyseal collar of Laval-Jeantet
  - Diaphyses: Bowing, osteoporosis, tunneling, fractures
    - Looser zones: Transversely oriented lucencies of insufficiency fractures in severe cases
  - Hip: Coxa vara, protrusio acetabuli
  - Knees: Genu valgum or varum
  - Periosteal reaction: Occult fracture or subperiosteal accumulation of unmineralized osteoid
- Skull, face (largely membranous growth except skull base)
  - Delayed sutural & fontanel closure
  - Postural molding, frontal bossing, platybasia
  - Delayed eruption of teeth, enamel hypoplasia
- Renal osteodystrophy with secondary hyperparathyroidism
  - Subperiosteal bone resorption
    - Distal clavicle, distal radius + ulna, middle phalanges of hands, medial femoral neck, upper medial proximal tibia
    - Lamina dura of teeth
  - Resorption at phalangeal tufts, pubic symphysis
  - Subchondral, subtendinous, subligamentous resorption
  - Endosteal bone resorption → lacy pattern of inner cortex (cortical tunneling)
  - Widened physes (subphyseal resorption, fibrous tissue deposition)
    - ↑ risk for slipped capital femoral epiphysis
  - Bone margins unsharp, hazy
  - Sclerosis with "smudged" trabeculae
  - Soft tissue Ca^{2+}
- Healing rickets after vitamin D therapy
  - ↑ density at ZPC after treatment in 2-3 weeks with nutritional rickets, 2-3 months with renal rickets

MR Findings

- Elongated cartilaginous physes with ↑ PD/T2 signal
- Absence of normal thin, low signal intensity ZPC

DIFFERENTIAL DIAGNOSIS

Newborn Stress Demineralization

- Metaphyseal lucent bands with intact ZPC in 1st 4-6 weeks after delivery

Leukemia or Metastatic Neuroblastoma

- Metaphyseal lucent bands with intact ZPC
- ± osteoporosis, permeative lesions, periosteal reaction

Congenital Syphilis

- Serrated lucent metaphyses
- Focal destruction of upper medial tibial metaphyseal cortex (Wimberger corner sign)

Metaphyseal Fracture of Child Abuse

- Classic metaphyseal corner injury (or bucket-handle fracture) due to fracture through primary spongiosa
- Only at sites of injury (not diffuse)
- ZPC & bony mineralization normal

Physeal Stress Injury

- Repetitive microtrauma to growth plate causes focal or diffuse longitudinal physeal widening & irregularity without transverse metaphyseal widening or cupping
  - Likely due to chronic metaphyseal vasculature injury impairing endochondral ossification in high-level athletes
- Single site usually affected (e.g., distal radius of "gymnast wrist" or proximal humerus of "Little Leaguer shoulder")
- Non-weight-bearing long bones (e.g., fibula, ulna) typically spared

Physeal Fracture

- Acute growth plate widening in Salter-Harris injuries
  - ± displacement of epiphysis + metaphyseal fragment
- Overlying soft tissue swelling
- Clear history of trauma usually present

Metaphyseal Chondrodysplasias

- Widespread abnormal metaphyseal configurations
- Varying etiologies & associated anomalies

Hypophosphatasia

- Wide spectrum of disease severity; due to inactive enzyme despite normal mineral concentrations
- Bowdler spurs characteristic

PATHOLOGY

General Features

- Etiology
  - Normal bone development requires adequate calcium + phosphorus (facilitated by vitamin D)
    - Essential for formation of hydroxyapatite crystals
    - Rickets due to ↓ availability of these substances
      - Also need adequate blood supply & cartilage for endochondral ossification
  - Vitamin D
    - ↑ absorption of calcium from intestine
    - ↑ calcium resorption in renal proximal tubules
    - Regulates apoptosis of chondrocytes, cartilage matrix mineralization, & metaphyseal angiogenesis
- Associated abnormalities
  - Fractures due to vitamin D deficiency may occur in mobile patients with radiologic manifestations of rickets (not isolated biochemical abnormalities)
  - Findings **not** attributable to rickets, which must suggest child abuse
    - Fractures in nonmobile infants
    - Fractures otherwise typical of child abuse (e.g., classic metaphyseal lesion or corner fracture, etc.)
    - Subdural hematoma or retinal hemorrhage

Microscopic Features

- Normal physis consists of 4 zones of cartilage
  - Germinal/resting cartilage cells (near epiphysis)
  - Proliferating cartilage cells
  - Hypertrophic columns of cartilage cells
  - Provisional Ca^{2+} of cartilage (ZPC)
    - 1st 3 zones: Radiolucent
    - ZPC: Thin, dense, undulating line at metaphyseal edge
- Metaphyseal spongiosa adjacent to ZPC: Cartilage matrix replaced by osteoid with remodeling & Ca^{2+} leading to spongy bone occupying marrow cavity
  - Rickets: Ca^{2+} of cartilage & osteoid does not occur → physeal/metaphyseal abnormalities

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Upper & lower extremities: Short stature, genu varum > genu valgum, metaphyseal swelling, pain, fractures
  - Skull: Craniotabes (easily indented soft bones), frontal bossing, delayed closure of anterior fontanelle
  - Torso: Scoliosis, accentuated lumbar lordosis, rachitic rosary (palpable expanded anterior ribs)

Demographics

- Age
  - Peak for dietary rickets: 3 months to 2 years

Treatment

- Most rickets responds to vitamin D therapy ± calcium

DIAGNOSTIC CHECKLIST

Consider

- Unexplained physeal widening at one site (i.e., without acute or chronic trauma) merits survey at other sites of rapidly growing long bones (knees, wrists)
  - If widening present at multiple sites, including non-weight-bearing long bones, suspect metabolic disease

Image Interpretation Pearls

- Be sure to look at growth centers of proximal humeri & femurs on neonatal chest & abdominal radiographs

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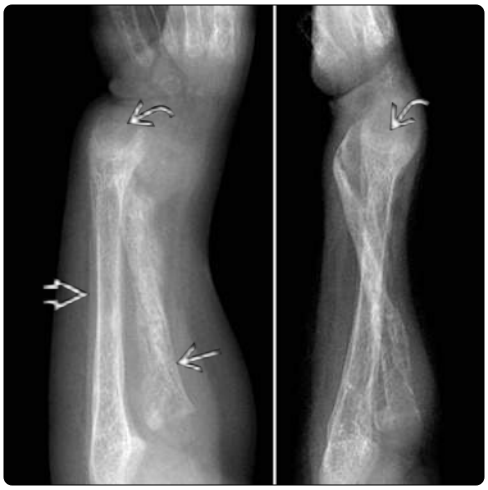
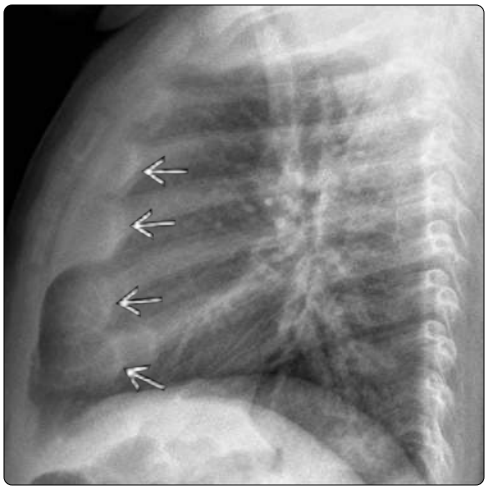
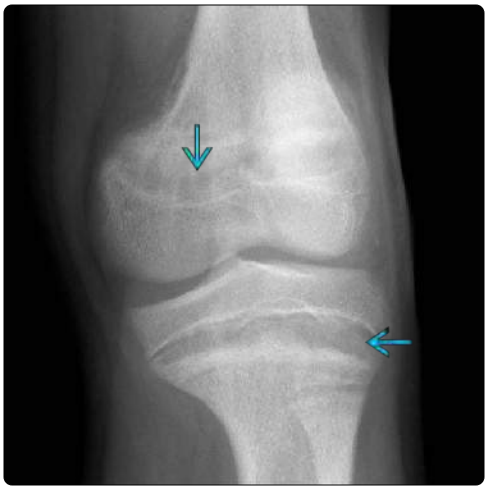
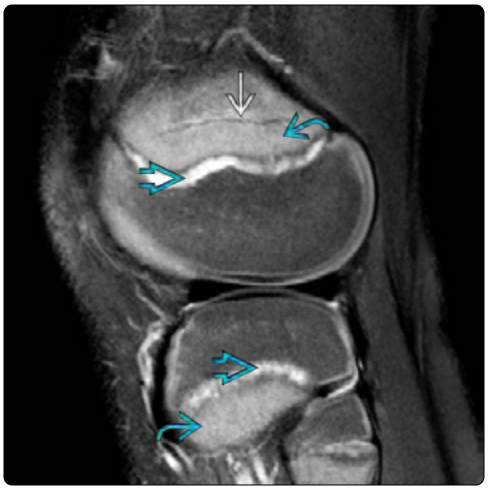


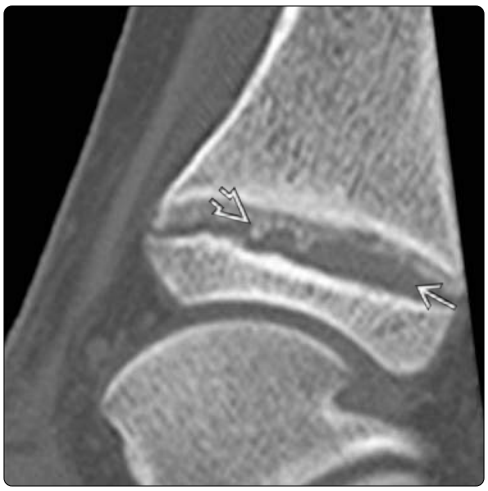


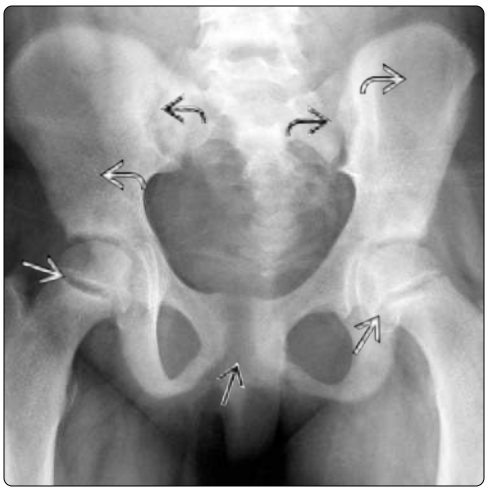
(Left) *Lateral radiographs of the forearm in a 2-year-old child with linear nevus sebaceous syndrome & associated paraneoplastic hypophosphatemic rickets show severe demineralization with periosteal reaction, cortical tunneling, & cupped, frayed metaphyses.* **(Right)** *Lateral radiograph in a 7-month-old boy with failure to thrive shows the widened & cupped anterior ribs typical of the "rachitic rosary."*





(Left) *AP radiograph of a 15 year old with knee pain shows loss of the normal dense ZPCs with partial mineralization of hypertrophied physeal cartilage. The metaphyses appear mildly frayed & cupped.* **(Right)** *Sagittal T2 FS MR in the same patient shows mild physeal lengthening ("widening") with abnormally hyperintense metaphyses that are partially mineralized. The thin, low-signal line represents an aberrant ZPC in this patient with nutritional rickets.*





(Left) *Sagittal CT reconstruction in a 12-year-old boy with ankle pain shows a lengthened ("widened") distal tibial physis with metaphyseal cupping & irregularity. This finding was present bilaterally at the tibias & fibulas.* **(Right)** *AP radiograph in the same patient, who was found to be in renal failure, shows typical changes of renal osteodystrophy & renal rickets with proximal femoral physeal widening, widened & irregular pubic symphysis & sacroiliac joints, & diffuse "smudging" of trabeculae.*

Sickle Cell Disease

KEY FACTS

IMAGING

- Skull: Widened diploic space
- Vertebrae
  - Infarctions: Central endplate depressions (H-shape or Lincoln log morphology)
  - Bone softening: Biconcave endplates (fish mouth)
- Ribs: Infarction part of acute chest syndrome (chest pain, dyspnea, cough + pulmonary consolidation)
- Long bone infarctions
  - Acute/subacute: Poorly defined marrow, soft tissue, & periosteal edema ± fluid collections
    - Not easily distinguished from osteomyelitis
    - Dactylitis (hand-foot syndrome): Hand & foot small tubular bone infarcts, usually at age 6-24 months
  - Chronic: Lucent or sclerotic medullary cavity
    - Diaphysis, metaphysis
    - Epiphyseal infarction most common in humeral & femoral heads: Sclerosis, subchondral collapse

PATHOLOGY

- Bony manifestations result from
  - Chronic anemia → red marrow hyperplasia
  - Immunocompromise & altered blood flow → osteomyelitis
  - Vasoocclusion → medullary infarction

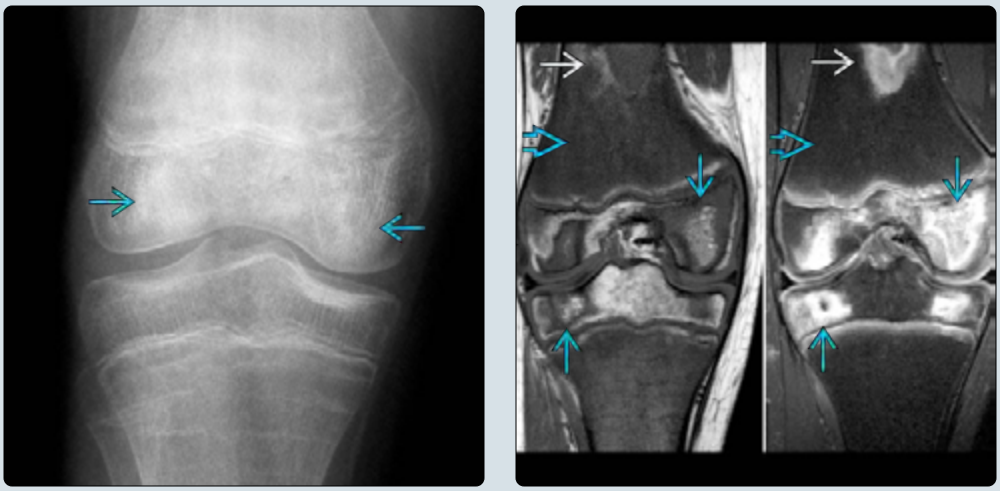
CLINICAL ISSUES

- Typical presentation: Pain due to vasoocclusive crisis involving any organ, most commonly bone
- Patients of African descent most commonly affected
- Mean survival: 42 years

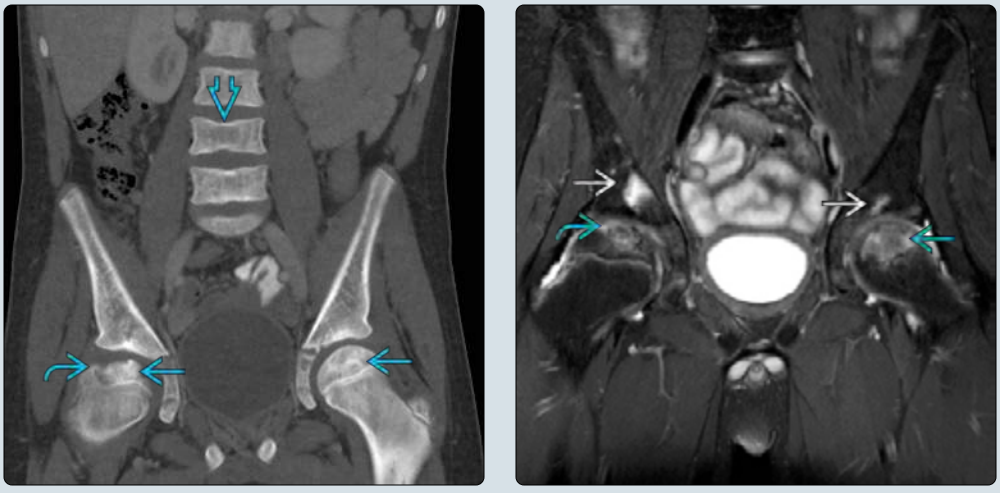
DIAGNOSTIC CHECKLIST

- Differentiation of acute bone infarct vs. osteomyelitis in SCD very difficult
  - Episodes of vasoocclusion > > osteomyelitis (50:1) in SCD
  - Cortical disruption & fluid collections (especially larger) favor osteomyelitis

(Left) *AP radiograph of the right knee in a patient with sickle cell disease (SCD) & pain shows patchy sclerosis of the femoral condyles, suggesting bone infarcts.* **(Right)** *Coronal T1 (L) & T2 FS (R) MR images in the same patient show heterogeneous geographic foci with serpiginous margins in the femoral & tibial epiphyses & femoral diaphysis, typical of bone infarcts. The abnormally low T1 & T2 signal intensity of the visualized metadiaphyses is due to increased red marrow & iron overload.*



(Left) *Coronal CECT in the same patient shows heterogeneous sclerosis of the femoral heads, consistent with avascular necrosis (AVN). There is subchondral collapse & fragmentation on the right with associated acetabular dysplasia. Central depressions are seen at several vertebral body endplates.* **(Right)** *Coronal T2 FS MR in the same patient shows articular cartilage loss on the right at the site of fragmentation. Infarcts are also seen in the left femoral head & bilateral iliac bones.*



TERMINOLOGY

Abbreviations

- Sickle cell disease (SCD); hemoglobin SS (Hb SS, Hgb SS)

Definitions

- SCD: Hb SS (homozygous) → severe anemia
- Sickle cell-hemoglobin C disease: Hb SC → mild anemia
- Sickle cell-α thalassemia: Hb S-α thal → severe anemia
- Sickle cell-β thalassemia: Hb S-β thal → mild-severe anemia
- Sickle cell trait: Hb SA (heterozygous gene carrier)
- Normal: Hb AA

IMAGING

General Features

- Best diagnostic clue
  - H-shaped vertebrae
  - Bone marrow infarctions with sclerosis
- Location
  - Percentage of all bone infarcts found in individual bones: Femur 16%, tibia 15%, humerus 13%, spine 11%, radius 10%, ulna 8%, pelvis 8%, others 19%

Radiographic Findings

- Skull
  - Expanded diploic space; hair on end appearance rare
  - Extramedullary hematopoiesis (EMH): Middle ear, paranasal sinuses
  - Mandible: Coarse trabeculae; condyle infarction
  - Orbital wall infarction → subperiosteal hemorrhage → proptosis (compression syndrome)
- Spine
  - Vertebral infarctions
    - Central endplate depression (superior + inferior endplate → H-shape or Lincoln log morphology)
    - ± compensatory enlargement of adjacent vertebra (tower vertebra)
    - ↓ height → kyphosis, lordosis
  - Smooth endplate concavities from bone softening (fish mouth vertebra)
  - EMH: Paraspinal/intraspinal masses
- Chest
  - Rib & sternal infarction: Part of acute chest syndrome (chest pain, dyspnea, cough + pulmonary consolidation)
    - Bone infarctions → lucent or sclerotic foci
  - EMH: Posterior mediastinal masses
  - Soft tissue findings provide useful clues
    - Enlarged heart (chronic anemia)
    - Absent splenic shadow (infarction)
    - Cholecystectomy clips (pigment gallstones)
- Pelvis
  - Osteomyelitis, protrusio acetabuli, infarction
- Extremities
  - Diametaphyseal infarction: Lucent or sclerotic serpiginous/geographic foci with healing (months)
  - Epiphyseal infarctions (avascular necrosis) most common in humeral & femoral heads
    - Sclerosis, subchondral fracture, coxa magna
  - Dactylitis (hand-foot syndrome): Hand & foot small tubular bone infarcts, usually at age 6-24 months
    - Soft tissue edema, solid periosteal reaction, cortical mottling, sclerosis
  - Infarctions up to physis → growth disturbances
    - Cone-shaped epiphyses of metacarpals, phalanges
    - Premature physeal fusion
  - Osteomyelitis: Femur, tibia, humerus most common
  - Marrow hyperplasia: Osteoporosis, thinned cortex, widened medullary spaces, coarsened trabeculae

MR Findings

- T1WI
  - Marrow hyperplasia: Bright yellow marrow replaced by darker red marrow (↓ fat, ↑ water content)
  - Variable appearance of infarctions
    - Serpiginous low signal intensity foci if chronic
- T1WI FS
  - Bright, patchy foci can favor infarction but not reliable for differentiating from infection
- T2WI FS
  - Poorly defined bright marrow, periosteal, & soft tissue edema from acute infarction or osteomyelitis
  - Soft tissue or subperiosteal collections or joint fluid seen in either process
    - Larger collections favor infection
  - Interrupted dark signal cortex favors infection
  - Double line sign: Alternating bright + dark T2 serpiginous foci of chronic infarctions throughout medullary cavities & epiphyses
  - ↓ marrow signal of iron overload from numerous transfusions
- T1WI C+ FS
  - Enhancement in infarction & infection variable
    - ↓ marrow enhancement (± fluid) in regions of acute infarction or ↑ pressure from infection/abscess
      - Thin rim enhancement favors infarct
      - Thick rim enhancement favors infection

Ultrasonographic Findings

- Fluid collections (especially larger) favor osteomyelitis
- Imaging guidance useful for sampling fluid for culture

Nuclear Medicine Findings

- Bone scan
  - Symmetric expansion of hematopoietic marrow involving femur, calvaria, small bones of hands/feet
  - Bone marrow infarction: Photopenic defect initially, may show ↑ activity with healing & revascularization
  - Bone infarction vs. osteomyelitis
    - Infarction: Tc-99m MDP bone scan shows ↑ activity; Tc-99m sulfur colloid marrow scan shows ↓ activity
    - Osteomyelitis: Bone scan shows ↑ activity; sulfur colloid marrow scan shows normal activity
- Tc-99m sulfur colloid
  - Can confirm masses as EMH

DIFFERENTIAL DIAGNOSIS

Langerhans Cell Histiocytosis

- Vertebra plana (flat vertebral body)
- "Punched-out" lytic bone lesions

Leukemia

- Osteoporosis, metaphyseal lucent bands, pathologic fractures
- May have more aggressive bone lesions & periosteal reaction

Thalassemia

- Changes of bone marrow expansion similar to SCD but exaggerated
  - H-shaped vertebra
  - "Hair on end" skull diploë
  - Avascular necrosis less common than in SCD
  - Paravertebral masses: EMH

Hereditary Spherocytosis

- Slight diploic widening, gallstones

Pyruvate Kinase Deficiency

- Wide diploic space, gallstones

Glucose-6-Phosphate Dehydrogenase Deficiency

- Bone changes absent

PATHOLOGY

General Features

- Etiology
  - Normal human hemoglobin A contains 4 globin chains: 2 α + 2 β chains
    - Hb SS: Abnormal β chains twist (polymerization) → RBC (red blood cell) distortion
    - RBC distortion exaggerated with deoxygenation → irreversible sickling (banana-shaped RBCs)
      - Sickled RBCs occlude vessels → infarction
      - Sickled RBCs quickly removed → anemia
  - Bony manifestations result from
    - Chronic anemia → red marrow hyperplasia → medullary cavity expansion, bone softening
    - Immunocompromise + abnormal blood flow → osteomyelitis
      - Osteomyelitis 2:1 *Salmonella*:*Staphylococcus*
    - Vasoocclusion → medullary infarction
- Genetics
  - Abnormal β globin gene on short arm of chromosome 11 → valine substituted for glutamic acid at β globin position 6 → Hb S

Microscopic Features

- Normally, red/cellular marrow of appendicular skeleton converts to yellow/fatty marrow during childhood
  - Begins in epiphyses & central diaphyses, gradually spreading toward metaphyses
  - Red marrow persists in normal adult axial skeleton
- Expansion of red marrow throughout skeleton in SCD

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pain due to vasoocclusive crisis involving any organ, most commonly bone
    - Infarction dactylitis often 1st manifestation
    - Painful chest/abdominal crises begin at age 2-3 years
  - Skeletal pain due to marrow infarction > > osteomyelitis
    - Fever, pain, swelling, ↓ motion, leukocytosis, ↑ inflammatory markers in either diagnosis
    - Osteomyelitis 50x less common than bone infarction
  - Splenomegaly initially, then splenic atrophy
    - Autosplenectomy by progressive infarction
  - Hemolytic anemia: Jaundice, gallstones
    - 50-70% have gallstones by adulthood
  - Marrow, liver, pancreas hemosiderosis (transfusions)
  - Stroke in 11% before 20 years old

Demographics

- Age
  - Typically detected on newborn screening
  - Painful crisis of bone in 50% by age 5
    - Most common cause of hospitalization
      - Acute chest syndrome 2nd most common
- Ethnicity
  - African Americans > Spanish, Mediterranean, Turkish, Arabian, Indian descent
- Epidemiology
  - Incidence: 1 in 375-650 African Americans have SCD
    - 8% of African Americans carry HbS gene
      - Up to 40% in some African tribes
  - Incidence: 1 in 2,000 Hispanics coming from Caribbean, Central America, South America

Natural History & Prognosis

- Mean survival: 42 years
- Acute chest syndrome: Most common cause of death

Treatment

- Sickle cell crisis: Oxygen, hydration, pain management, blood transfusion
- Prophylactic penicillin + pneumococcal & *Haemophilus influenzae* vaccines to prevent infection

DIAGNOSTIC CHECKLIST

Consider

- Differentiation of acute bone infarcts vs. osteomyelitis in SCD very difficult clinically & radiologically
- Episodes of vasoocclusion > > osteomyelitis (up to 50:1)

Image Interpretation Pearls

- Cortical disruption &/or fluid collections favor osteomyelitis

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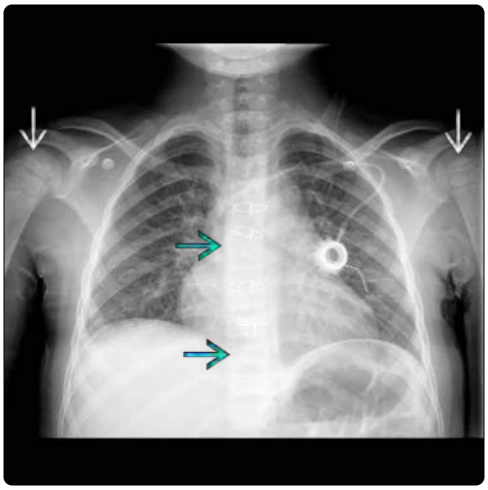
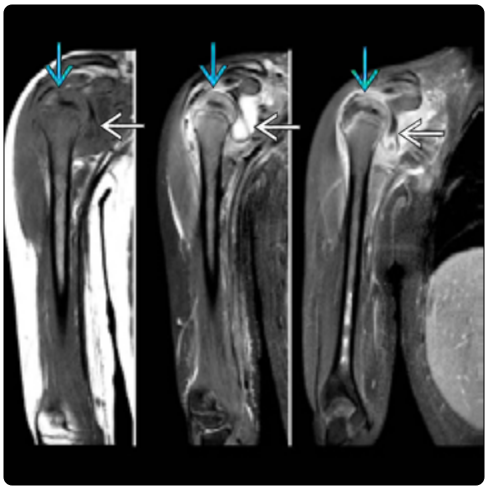
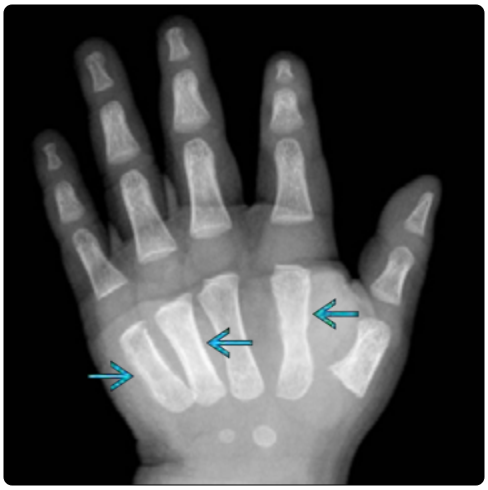
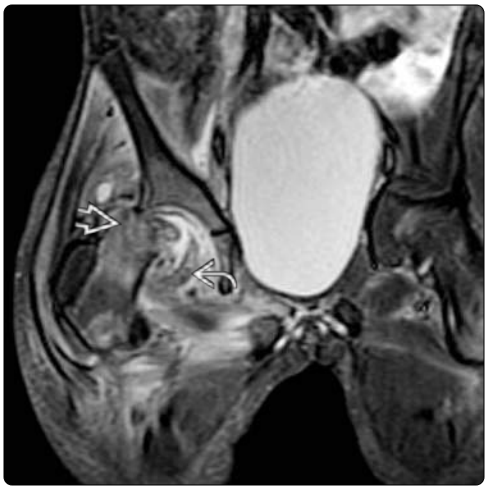


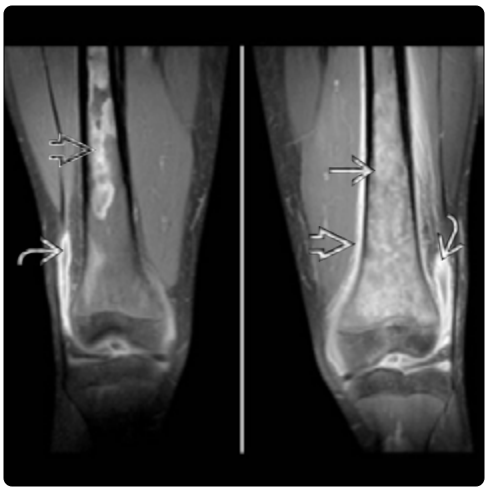


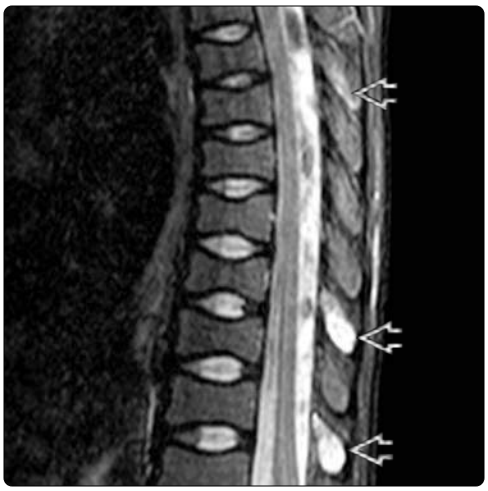
(Left) *AP chest radiograph in a 7 year old with SCD shows sclerosis of the humeral heads, consistent with AVN. Vertebral endplate concavities & central depressions are noted at multiple levels.* **(Right)** *Coronal T1 (L), STIR (M), & T1 C+ FS (R) MR images in the same patient show heterogeneous epiphyseal marrow, a moderate joint effusion, & surrounding soft tissue edema. These findings can be seen with acute bone infarction or osteomyelitis in SCD.*





(Left) *PA radiograph of the hand in an 11-month-old SCD patient with swelling shows medullary sclerosis & periosteal reaction of the 2nd, 4th, & 5th metacarpals, typical of dactylitis.* **(Right)** *Coronal T1 C+ FS MR in an SCD patient with recently diagnosed pelvic osteomyelitis, septic arthritis, & soft tissue abscesses shows an unusual complication: Complete slip of the capital femoral epiphysis from the femoral neck.*





(Left) *Coronal T1 C+ FS MR in a 14-year-old boy with SCD shows heterogeneous enhancement of the distal left femur with periosteal edema (due to infection vs. acute infarction). The bilateral knee synovitis could be reactive from either process or from septic arthritis. Subacute infarction is seen in the right femoral shaft.* **(Right)** *Sagittal STIR MR shows biconcave vertebral bodies at each level, typical of bone softening. There is also high signal intensity in multiple spinous processes from recent infarctions.*

Scoliosis

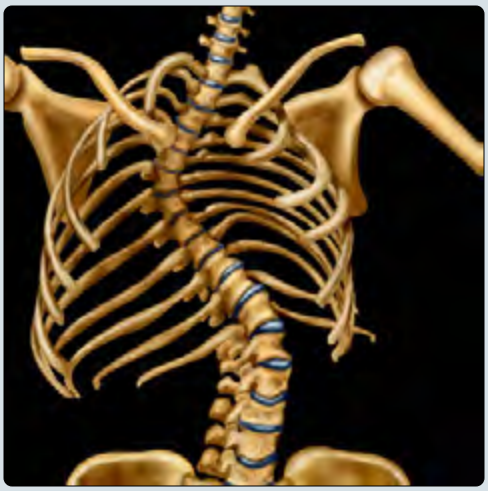
KEY FACTS

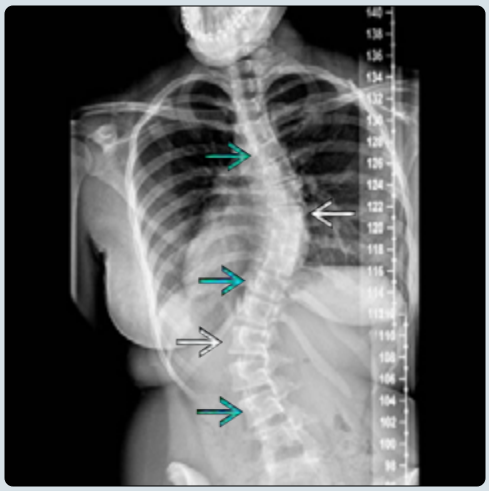
TERMINOLOGY

- Lateral curvature(s) of spine with Cobb angle of ≥ 10°
- Flexible: Nonstructural; corrects with ipsilateral bending
  - Usually does not progress; need not be included in fusion
- Rigid: Structural; does not correct with ipsilateral bending
  - Cobb angle remains ≥ 25° on ipsilateral bending
  - Included in spinal fusion
- **Curve etiologies**
  - Some diagnoses span categories
  - Idiopathic: Most common; 70-85% of all scoliosis
    - Classified according to time of onset
    - Infantile: < 3 years of age
      - Mostly develops during first 6 months of life
      - Typical: Convex left thoracic curve (70%)
      - M > F
    - Juvenile: 4-9 years
      - Typical: Convex right thoracic curve
      - Most likely to progress
    - Adolescent: > 10 years
      - Most common type: Convex right thoracic curve
      - M < < F
  - Congenital: 10%
    - Osteogenic: Segmentation anomaly
    - Neuropathic: Syrinx, tethered cord, diastematomyelia
  - Neuromuscular (neuropathic or myopathic)
    - Single long curve
    - Neuropathic: Spina bifida, cerebral palsy
    - Myopathic: Muscular dystrophies, spinal muscular atrophy
  - Developmental (skeletal dysplasia or dysostosis)
  - Tumor associated
    - Osteoid osteoma, cord neoplasm, neurofibroma

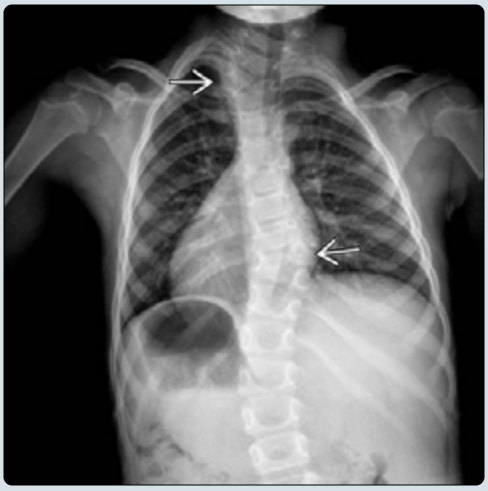
CLINICAL ISSUES

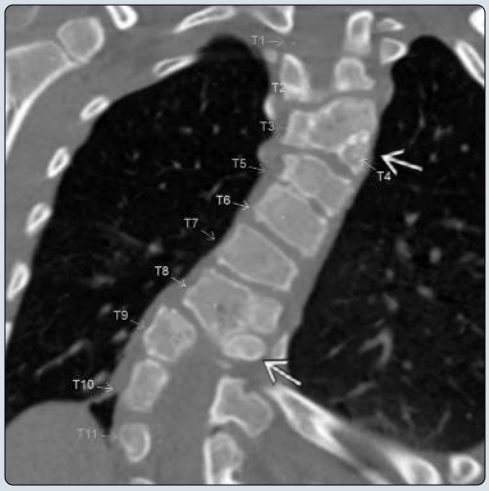
- Younger patients presenting with severe curves more likely to progress than older children with less severe curves
- Progression most likely during adolescent growth spurt





(Left) *Graphic shows a rotary S-shaped idiopathic scoliotic curve.* **(Right)** *PA radiograph of a 13 year old during follow-up for idiopathic scoliosis shows 47° convex right thoracic & 47° convex left thoracolumbar scoliotic curvatures. The terminal ⇨ & apical ➡ vertebrae for each curve are denoted. Note that the image is displayed from the perspective of the examining/operating orthopedist.*





(Left) *Frontal radiograph in 4 year old with congenital scoliosis shows hemivertebrae ➡ within the thoracic spine.* **(Right)** *Coronal reformatted NECT in a 7 year old with congenital scoliosis shows hemivertebrae ➡ at T4 & T8. CT can be very helpful in assessing congenital curves due to vertebral anomalies & for planning surgery in otherwise complex cases.*

TERMINOLOGY

Definitions

- Presence of lateral curvature(s) of spine with Cobb angle of ≥ 10°, often associated with vertebral rotation
- 2 types
  - Flexible: Nonstructural, corrects with ipsilateral bending, usually does not progress
  - Structural: Rigid, does not demonstrate correction with ipsilateral bending
    - Cobb angle of ≥ 25° on ipsilateral bending

IMAGING

General Features

- Morphology
  - Idiopathic
    - Most common: 70-85% of all scoliosis
    - Classified according to time of onset
    - Infantile: < 3 years of age
      - Most develop during first 6 months of life; M > F
      - 1/4 associated with hip dysplasia
      - Typical: Convex left thoracic curve (70%)
    - Juvenile: 4-10 years
      - Typical: Convex right thoracic curve, progressive
    - Adolescent: > 10 years
      - Most common type: Convex right S-shaped thoracic
      - Compensatory left convex lumbar, ↑ rapidly with growth spurts
      - M < < F
    - Adult: Curve begins after maturation
    - Idiopathic scoliosis is not associated with an underlying cause
      - However, underlying anomalies can cause similar curves: Syringohydromyelia, Chiari 1, cord neoplasm, disc disease
  - Congenital: 10%
    - Result of vertebral anomalies: Hemivertebrae most common (45%); occult spinal abnormality (15-40%)
    - Typical: Thoracic or thoracolumbar curve
    - Progressive scoliosis (3/4)
    - Image with MR
      - Progressive curve or surgery planned
    - Associated with
      - Spinal dysraphism: Lipoma, diastematomyelia, syringohydromyelia, tethered cord
      - Genitourinary anomalies (6%), cardiac anomalies (15%), rib anomalies
  - Neuromuscular (neuropathic or myopathic)
    - Cerebral palsy, poliomyelitis, muscular dystrophy, spinal muscular atrophy
  - Developmental (skeletal dysplasia or dysostosis)
    - Neurofibromatosis, achondroplasia, osteogenesis imperfecta
  - Tumor associated: Osteoid osteoma, cord neoplasm, neurofibroma

Radiographic Findings

- Imaging evaluation depends on cause of scoliosis
  - Initial erect anteroposterior view from chin to greater trochanter
    - Posteroanterior on follow-up (less breast radiation)
  - Lateral if clinical concern of excessive kyphosis or lordosis
  - Cobb method of measuring scoliosis angle
    - Parallel lines to superior terminal vertebra superior endplate & inferior terminal vertebra inferior endplate
      - If endplates not seen, use pedicles
    - Perpendicular intersecting lines then yield Cobb angle
    - Same vertebral bodies used for follow-up measurements
    - Curve progression present when > 5° between studies
- Lateral bending films to assess flexibility prior to surgery
- Left wrist/hand bone age film to assess amount of potential growth remaining in patient (& determine timing of surgery)
- Iliac crests on scoliosis study also used for skeletal maturation
  - Iliac crest divided into 4 quadrants; Risser grade according to ossification of iliac apophysis
  - Risser 0: No ossification; Risser I: Only lateral 1/4 ossified; Risser IV: All 4 quadrants ossified; Risser V: Fused iliac apophysis
  - At skeletal maturity, scoliosis unlikely to progress if < 30°
- Assessment of balance
  - Central sacral vertical line (CSVL): Vertical line drawn on frontal view perpendicular to imaginary tangential line across top of iliac crests, bisects sacrum
  - Plumb line: Center of C7 parallel to radiographic edges extending distally
  - Coronal balance on standing frontal image: Distance between CSVL & plumb line: > 2 cm abnormal
  - Sagittal balance: Distance between posterosuperior aspect of S1 vertebral body & plumb line: > 2 cm abnormal
- Vertebral rotation
  - Nash-Moe method: Percentage of pedicle on convex side of curve displaced in respect to vertebral body width
- **Findings by curve types**
  - Idiopathic
    - Prevalence of typical curvature: Convex right thoracic curve > right thoracic & left lumbar > right thoracolumbar > right lumbar
    - Vertebral rotation, L5 spondylolysis may be present
  - Congenital
    - Failure of vertebral formation (wedge vertebra, hemivertebra)
    - Failure of segmentation (pedicle bar, block vertebra)
  - Tumor associated
  - Neurofibromatosis
    - Most lack distinctive diagnostic features
    - Classic: High thoracic acute curvature, kyphosis, rib anomalies, posterior vertebral scalloping
  - Neuromuscular
    - Tethered cord, syrinx, Chiari
    - Single long curve
- EOS system: Biplanar x-ray imaging
  - Several times ↓ radiation dose compared to conventional radiographs
  - Standing ± bending images

CT Findings

- NECT

- Coronal, sagittal, & 3D reconstruction for surgical planning
- Evaluates congenital vertebral anomalies
- Assess for pseudoarthrosis following spinal fusion

MR Findings

- Indications for MR
  - Congenital scoliosis
  - Neuropathic cause
  - Juvenile onset: 4-10 years
  - Idiopathic
    - Rapid progression of curve
    - Unusual curves: Convex left thoracic, long right thoracolumbar curve, double thoracic
    - Widened spinal canal or foramina, osseous lesion, foot deformity
    - Pain, headache, neck pain, neurologic deterioration
    - Debatable MR imaging of typical curves without other indications

Ultrasonographic Findings

- 3D imaging for measuring vertebral rotation
- Measuring lengthening in magnetically controlled rods

Nuclear Medicine Findings

- Bone scan
  - SPECT imaging for pseudoarthrosis following spinal fusion surgery

DIFFERENTIAL DIAGNOSIS

Other Causes for Scoliosis

- Differentiated by clinical history, radiographic findings, & supplemented by MR
  - Osteoid osteoma
  - Inflammation, infection, or tumor
    - Many etiologies
  - Limb length discrepancy

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually asymptomatic
    - Idiopathic scoliosis usually detected during physical exam
  - Pain from progressive curvature or degenerative disc & facet disease

Demographics

- Age
  - Idiopathic
    - Infantile: < 3 years; juvenile: 4-10 years; adolescent: 10 years to skeletal maturity
- Gender
  - Idiopathic: Female predilection
  - Girls tend to progress more than boys in idiopathic scoliosis
- Epidemiology
  - 0.2-0.5% of population in USA

Natural History & Prognosis

- Idiopathic curves < 25-30° will not progress when skeletally mature
- Worsening curve in 10-25% of cases
  - During adolescent growth spurts
    - Younger patients with more severe curves at presentation much more likely to progress than older children with less severe curves
  - Curves > 40-50° after skeletal maturity
  - Cardiopulmonary complications from severe scoliosis

Treatment

- Mehta casting for infantile scoliosis
- Idiopathic
  - Observe < 20° in adolescent or < 30° in skeletally mature at presentation
  - Brace (orthotics)
    - 10 years or older
    - 25-45° at presentation (Risser 0-2)
    - > 25° with progression
  - Surgical
    - Curves > 45°
    - Progressive curves that fail bracing
- Congenital
  - Observation
  - Surgical: With progressive curve
    - In situ fusion, anterior & posterior epiphysiodesis, hemivertebrae resection, reconstructive osteotomies
- Neuromuscular
  - Typical: Anterior & posterior spinal fusion
- Complications of spinal fusion
  - Rod or wire breakage, slippage of hook, infection, spondylolysis, superior mesenteric artery syndrome, pseudoarthrosis

DIAGNOSTIC CHECKLIST

Consider

- Atypical/painful scoliosis: MR to exclude spinal pathology

Image Interpretation Pearls

- Offset at site of image stitching can create artifacts
- Radiographs may be displayed from examining/operating orthopedist's perspective rather than conventional radiologic perspective

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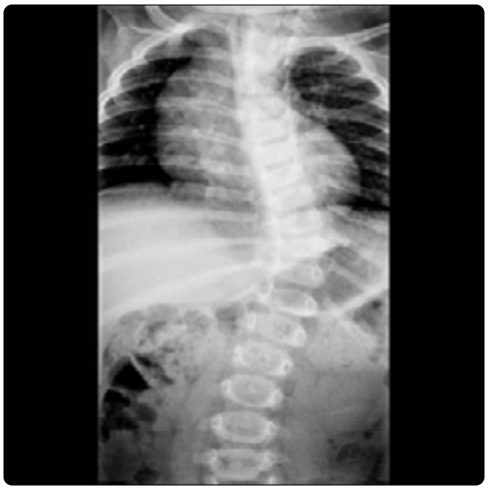
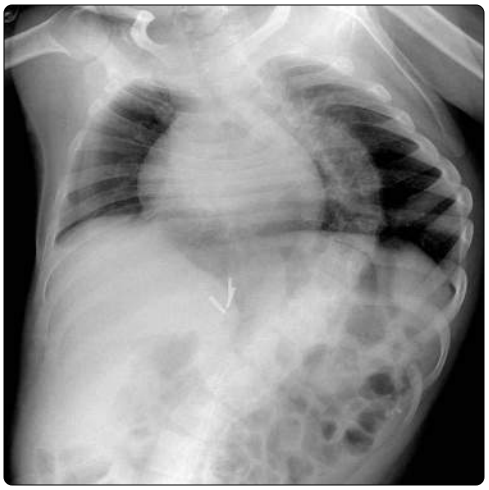
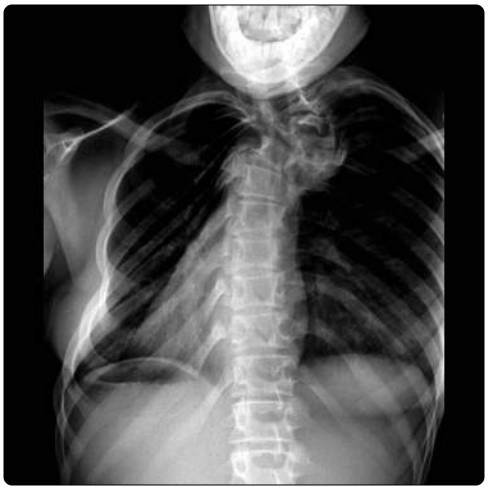
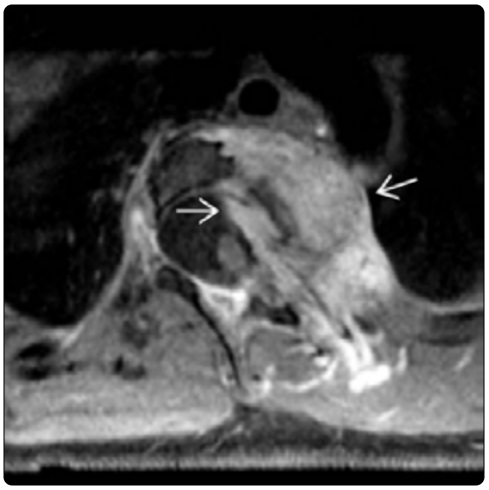


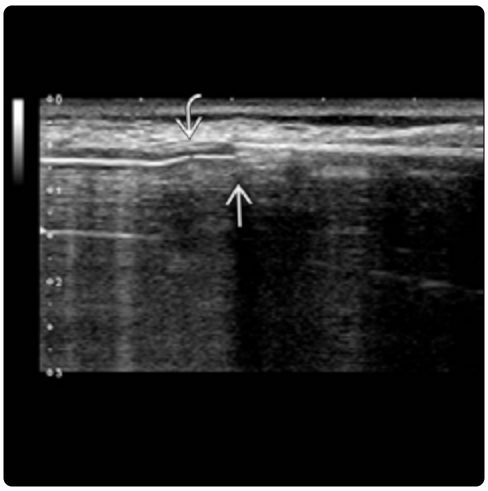


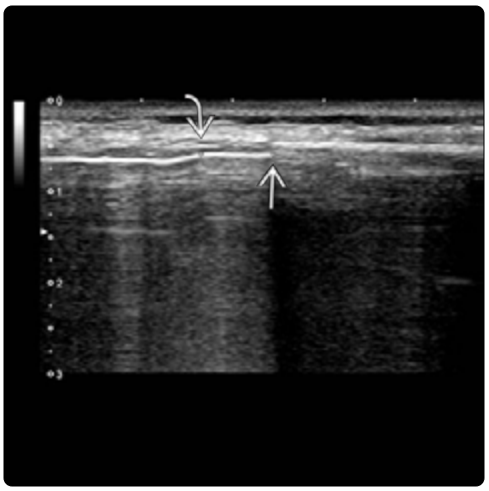
(Left) *Frontal sitting radiograph shows an 8 month old with a 40° convex left curvature of the thoracolumbar spine. This was classified as an infantile idiopathic scoliosis. This infant was treated with Mehta casting & eventually placement of magnetic growing rods.* **(Right)** *Frontal radiograph in a cerebral palsy patient shows a long, convex, left thoracolumbar C-shaped scoliotic curvature. This is an example of a characteristic neuromuscular scoliotic curvature.*





(Left) *Frontal radiograph as part of a scoliosis series in a 12 year old with back pain, café au lait spots, & neurofibromatosis type 1 shows a short-segment, convex, right upper thoracic curvature.* **(Right)** *Axial T1 C+ FS MR in the same patient shows lobular, enhancing paraspinal & intraspinal infiltrative lesions ➡ of plexiform neurofibromas.*





(Left) *Longitudinal US in a 4 year old with infantile idiopathic scoliosis and magnetically controlled growing rod shows a 5-mm distance between the housing unit ➡ & the neck of the extended part ➡. This was performed in ultrasound prior to lengthening.* **(Right)** *Longitudinal US in the same child performed 5 minutes later (status post 3 mm of lengthening) shows that the distance between the housing ➡ & neck ➡ of the extending part now measures 8 mm.*

Tarsal Coalition

KEY FACTS

TERMINOLOGY

- Congenital or acquired abnormal fusion of 2 tarsal bones; fusion may be osseous, cartilaginous, or fibrous

IMAGING

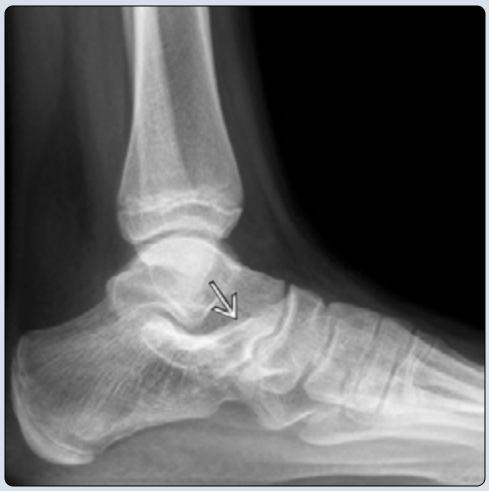
- Tarsal coalitions
  - Calcaneonavicular (CN)
  - Talocalcaneal (TC)
  - Uncommon: Talonavicular, calcaneocuboid, cubonavicular
- CN coalition
  - 45° internal oblique radiograph view (Slomann view)
  - Anteater nose sign = elongation of anterosuperior calcaneus on lateral view
  - ± hypoplastic talar head
- TC coalition
  - Most common: Middle facet; less common involvement of anterior or posterior facets
  - Harris-Beath (axial) & lateral views
  - Difficult to see coalition, CT often needed
  - Talar beak
  - C sign: Continuous uninterrupted line formed posteriorly by talar dome & sustentaculum tali on lateral view
  - Rounding of lateral talar process
  - Narrowing of posterior subtalar joint
  - "Ball-&-socket" ankle joint

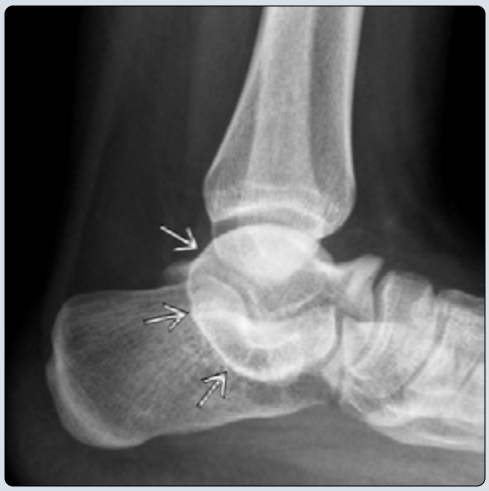
PATHOLOGY

- Congenital
- Acquired: Trauma, infection, arthritis, or surgery

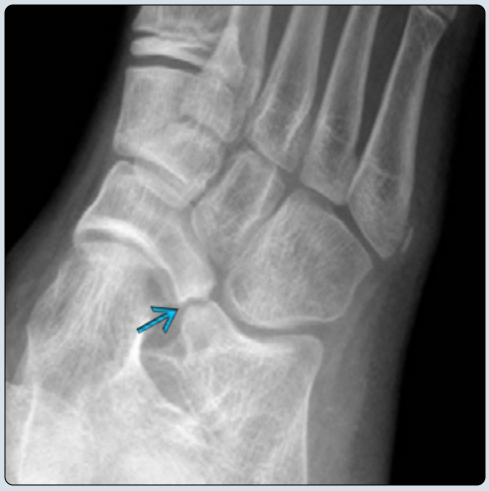
CLINICAL ISSUES

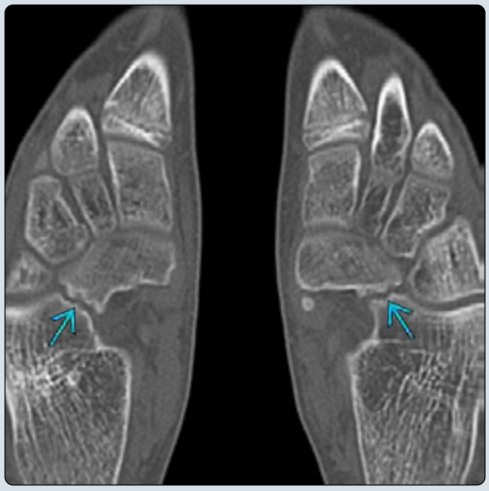
- Recurrent sprains, midfoot pain, limited subtalar motion, peroneal spastic flatfoot
- Symptoms occur as coalition ossifies
- 90% of all coalitions: TC or CN
- Up to 50% bilateral
- Look for > 1 coalition in same foot (rare)





(Left) *Lateral radiograph in a 10-year-old girl with foot pain for 1 year shows elongation of the anterior process of the calcaneus (anteater nose sign) ➡. The oblique view (not shown) confirmed a calcaneonavicular (CN) coalition.* **(Right)** *Lateral radiograph in a teenager who presented with recurrent ankle pain & swelling shows a continuous C sign ➡ of a talocalcaneal (TC) coalition. The C-shaped line is formed superiorly by the medial talar dome with continuity to the sustentaculum tali inferiorly.*





(Left) *Oblique foot radiograph in a 10 year old shows narrowing of the CN articulation ⇨ with sclerosis & irregularity of the apposed navicular & calcaneus consistent with a nonosseous coalition.* **(Right)** *Coronal oblique bone CT images in the same patient confirm bilateral nonosseous CN coalitions with narrowing, irregularity, & sclerosis at the CN articulations ⇨.*

TERMINOLOGY

Synonyms

- Tarsal fusion

Definitions

- Congenital or acquired abnormal fusion of 2 or more tarsal bones
  - This union may be osseous, cartilaginous, or fibrous

IMAGING

General Features

- Best diagnostic clue
  - Visualized close apposition or fusion of middle facet of talocalcaneal (TC) joint or anterodorsal calcaneus to navicular
- Location
  - Calcaneonavicular (CN)
  - TC
  - Uncommon: Talonavicular, calcaneocuboid, or cubonavicular
- Morphology
  - Synostosis = ossific bar
  - Synchondrosis = cartilaginous bar
  - Syndesmosis = fibrous union

Radiographic Findings

- Radiography
  - CN coalition
    - 45° internal oblique view (Slomann view)
      - Ossific bar connecting anterior process of calcaneus to dorsolateral navicular
      - Irregularity, sclerosis, or narrowing of CN space (fibrous or cartilaginous coalition)
    - Anteater nose sign = elongation of anterosuperior calcaneus on lateral view
    - Broadening of medial aspect of anterosuperior calcaneus in close apposition to navicular
    - ± hypoplastic talar head
    - Radiography typically diagnostic; other imaging may not be needed
  - TC coalition
    - Harris-Beath (axial) & lateral views
    - Difficult to see; CT often needed
    - Most commonly at middle facet; less common involvement of anterior or posterior facets
    - Talar beak: Impaired subtalar motion → elevation stress at insertion of TC ligament → cycles of osseous repair → beak immediately adjacent to dorsal aspect of talonavicular joint (not specific for coalition)
    - Rounding or blunting of lateral talar process
    - C sign: Continuous uninterrupted line formed by medial talar dome & sustentaculum tali on lateral view (50%) (sign of flat feet)
      - False positive in some due to pes planus & abnormal positioning
    - Absent middle facet sign: Middle facet inapparent on lateral view
    - "Ball-&-socket" ankle joint: Concave surfaces of tibia & fibula, domed talus (convex proximal margin); uncommon & nonspecific
    - Narrowing of posterior subtalar joint
    - Normal middle & posterior facets are parallel to each other on Harris-Beath view; middle facet slants inferomedially in fibrous or cartilaginous coalition (bar in ossific coalition)
- CT Findings
  - Axial, coronal, & sagittal reformats
  - CN coalition
    - Joint space narrowing or reactive sclerosis
    - Widening of medial aspect of anterosuperior calcaneus
    - ± bony bridge
    - Best seen on axial or sagittal planes
  - TC coalition
    - Ossific bar (best on coronal images)
    - Downward (may be horizontal) orientation/sloping of sustentaculum along middle facet (normally slopes upward medially)
    - Reactive subchondral sclerosis & narrowing with cystic & hypertrophic changes of TC joint (especially middle facet)
    - ± broadening or hypoplasia of sustentaculum

MR Findings

- T1WI
  - CN coalition
    - Sagittal & axial images best for detecting coalition
    - Hypointense reactive changes
  - TC coalition
    - Most commonly middle facet
    - Coronal image best for detecting coalition
    - Osseous: Bar with marrow signal connection
    - Fibrous & cartilaginous: Hypointense signal connection with hypointense subchondral marrow changes
- T2WI
  - Ossific: Bone marrow contiguity across either TC or CN coalition
  - Cartilaginous: Hyperintense fluid signal connection with reactive hyperintense subchondral bone marrow edema
  - Fibrous: Intermediate signal connection with reactive hyperintense subchondral bone marrow edema
  - Cartilaginous & fibrous: ↓ joint space

Imaging Recommendations

- Best imaging tool
  - Bone CT or 3-plane MR: TC coalition
  - 45° oblique radiograph: CN coalition

DIFFERENTIAL DIAGNOSIS

Subtalar Fractures

- Talar fractures
- Calcaneal fractures

Osteomyelitis

- Poorly defined marrow hyperintensity, ± erosion or sinus tract
- Pain, fever, erythema; ↑ C-reactive protein, sedimentation rate, leukocytosis

Osteochondritis Dissecans

- Osteocartilaginous change at medial talar dome; ± cysts, edema, sclerosis, fissuring, detached fragment
- Mostly adolescents, M > F

Juvenile Idiopathic Arthritis

- Synovitis with effusions, ± cartilage or osseous erosions
- Reactive marrow edema
- ± tenosynovitis

Tumor

- Chondroblastoma
- Bone cyst

PATHOLOGY

General Features

- Etiology
  - Congenital failure of segmentation & differentiation of primitive mesenchyme
  - Acquired: Trauma, infection, arthritis, or surgery
- Genetics
  - Autosomal dominance with high penetrance
- Associated abnormalities
  - Apert
  - Hand-foot-uterus syndrome
  - Proximal focal femoral deficiency
  - Hereditary symphalangism

Staging, Grading, & Classification

- TC coalition classification scheme based on 3D CT reconstruction (Rozansky et al.)
  - Type I: Linear coalition
  - Type II: Linear coalition with posterior hook
  - Type III: Shingled coalition
  - Type IV: Complete osseous coalition
  - Type V: Posterior coalition

Gross Pathologic & Surgical Features

- Osseous, fibrous, or cartilaginous connection

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Usually asymptomatic in early life
  - Recurrent sprains, chronic midfoot pain, limited subtalar motion
- Other signs/symptoms
  - Commonly asymptomatic, discovered after imaging for trauma
  - Pes planus + heel valgus
  - Flattening of medial arch
  - Pain with activity
  - Peroneal spastic flatfoot
    - Rigid valgus deformity, pain, & peroneal muscle spasm
    - Tarsal coalition most common cause
    - Other causes: Fracture, arthritis, & some tumors
  - TC: Limited subtalar motion & prominence inferior to medial malleolus (double medial malleolus sign)

Demographics

- Age
  - Symptoms occur as coalition ossifies
    - Talonavicular (3-5 years)
    - CN (8-12 years)
    - TC (12-16 years)
  - Fibrous coalition at birth, ossifies 2nd decade
- Gender
  - M > F
- Epidemiology
  - 1% incidence
  - 90% of all coalitions are TC or CN
  - Bilateral: 25-50%
  - Other tarsal coalitions uncommon

Natural History & Prognosis

- Coalitions at birth may be fibrous or cartilaginous & later ossify
- Symptoms more severe when coalition ossifies
- ↓ hindfoot motion makes child prone to ankle sprains
- Commonly discovered after imaging for ankle injuries

Treatment

- Conservative: 1st
  - Nonsteroidal antiinflammatory medication, arch supports, steroids, trial of casting, orthotics, & physical therapy
- Surgical or arthroscopic resection
  - CN: Resection of bony bridge or arthrodesis
    - Fat autograft interposition, extensor digitorum brevis interposition
  - TC: Resection of middle facet bony bridge with fat interposition
  - If excision fails or severe degenerative disease: Fusion or triple arthrodesis

DIAGNOSTIC CHECKLIST

Consider

- TC coalition: Coronal NECT or MR
- CN coalition: 45° internal oblique radiograph

Reporting Tips

- CT: Location & size of coalition, other coalitions (bilateral or in same foot), degenerative changes

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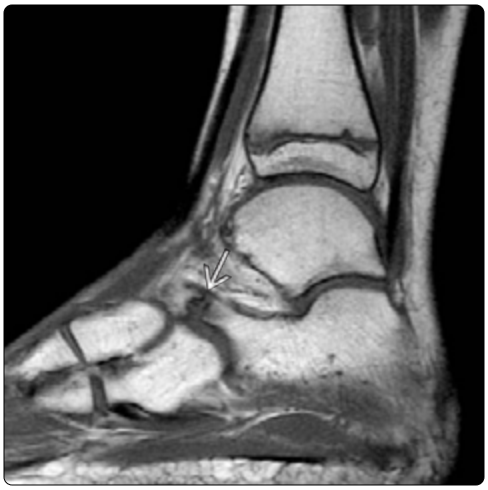
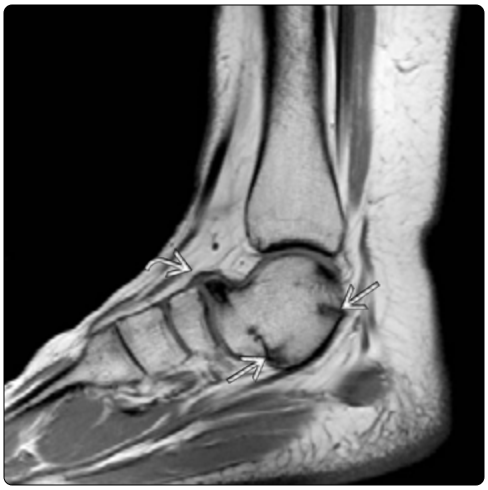
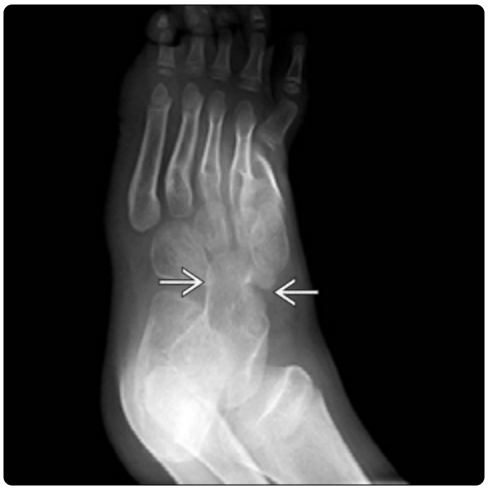
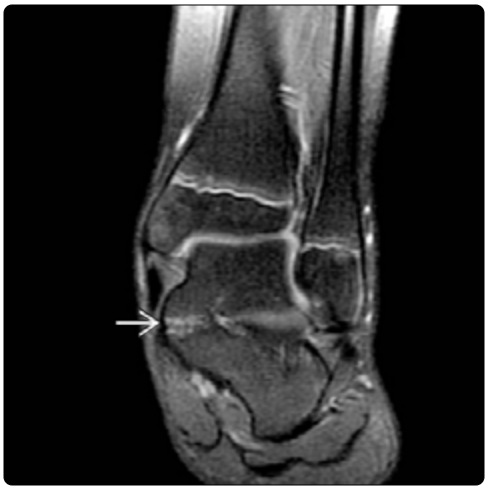


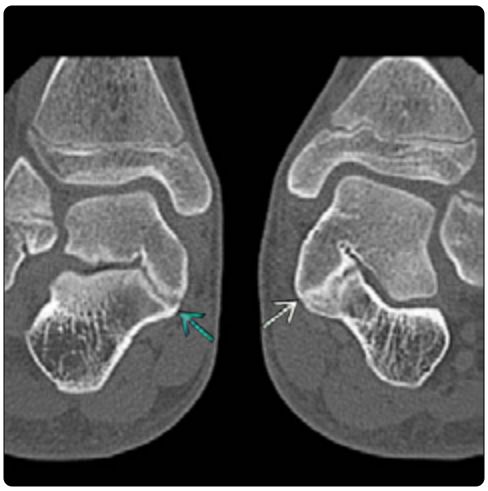


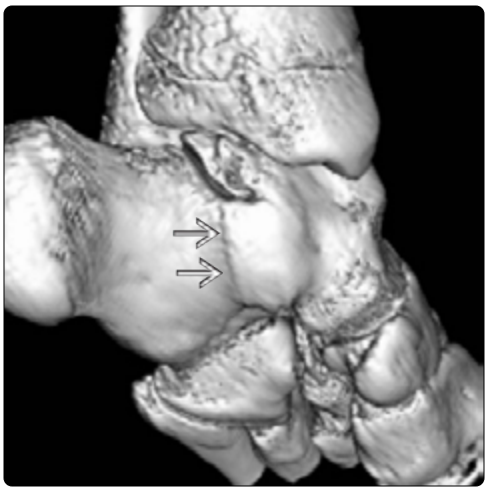
(Left) *Sagittal T1 MR in an 11 year old with acute on chronic ankle pain shows irregularity & narrowing of the CN joint ➡, indicating a nonosseous coalition. No other coalitions were found.* **(Right)** *Sagittal T1 MR in a 16 year old with ankle pain shows a large osseous TC coalition ➡ of the middle facet. Notice the talar beaking ➡, a common secondary finding that is the result of impaired talar motion.*





(Left) *Oblique radiograph in a 7 year old with dysmorphic craniofacial features shows a less common osseous coalition ➡ between the talus & navicular.* **(Right)** *Coronal T2 FS MR in an 11-year-old girl with painful pes planus shows irregularity, narrowing, & hyperintense T2 marrow signal ➡ of the TC joint indicating a nonosseous coalition at the middle facet.*





(Left) *Coronal bone CT in a 12 year old with bilateral ankle pain shows abnormally oriented downsloping middle TC facets with partial osseous bridging on the left ➡ & close apposition on the right ⇨.* **(Right)** *Oblique 3D reformation of the bone CT in the same patient shows the plantar aspect of the left TC coalition ➡.*

Brachial Plexopathy

KEY FACTS

TERMINOLOGY

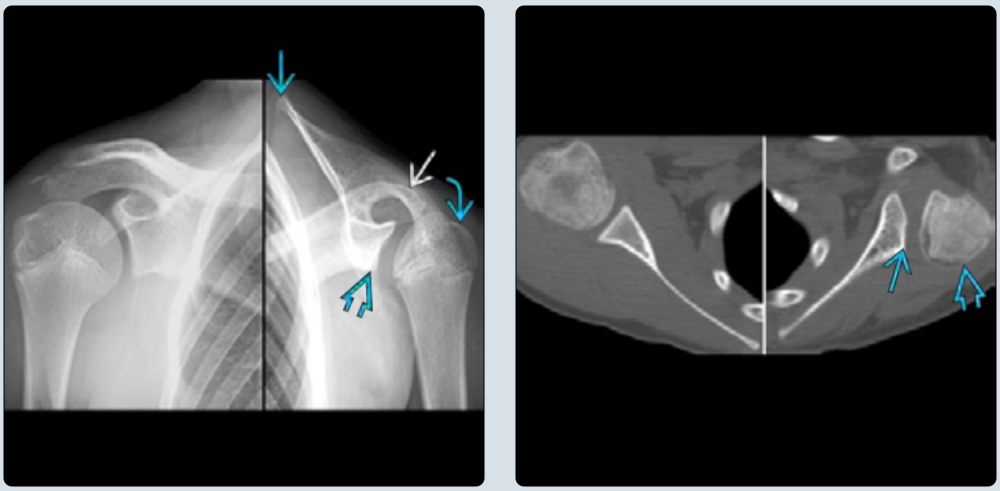
- Brachial plexopathy: Injury to ≥ 1 brachial plexus nerve roots, trunks, or cords → upper extremity contracture
- Glenohumeral dysplasia: Sequelae of brachial plexopathy on developing glenoid & humeral head

IMAGING

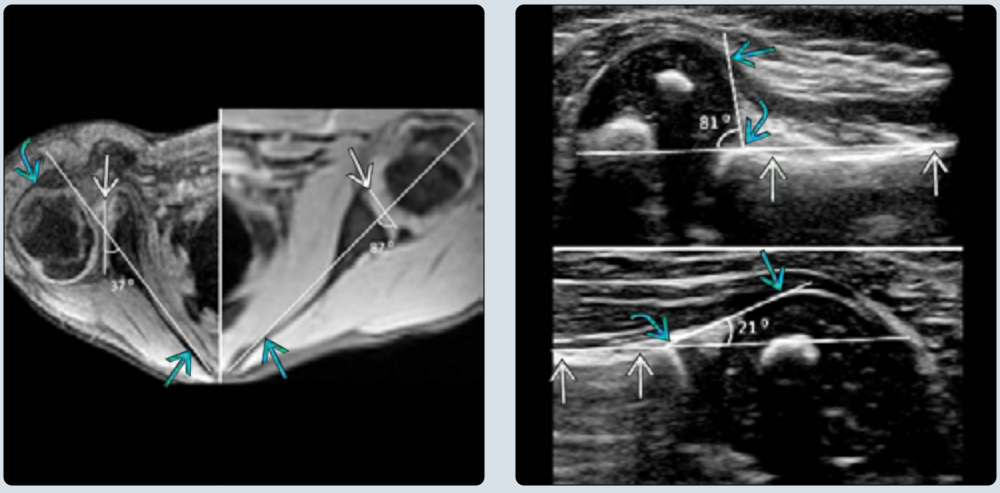
- Affected & unaffected shoulders imaged for comparison
- Radiographs
  - Dysplastic glenoid, winged scapula, hooked coracoid
  - Humeral head small & ovoid
- Ultrasound
  - Uses posterior axial approach to glenohumeral joint
  - Can assess humeral head prior to ossification
  - α angle = angle between posterior scapular margin & tangent to humeral head from posterior edge of glenoid
    - Normal ≤ 30°
  - % humeral head posterior to scapular margin
    - Humeral head ossification center should lie anterior to posterior scapular margin
- MR
  - Cartilage-sensitive GRE & PD FS sequences show unossified & ossified portions of glenoid & humeral head
    - Angle of glenoid version
      - ↑ retroversion with ↑ glenohumeral dysplasia
      - > 5° difference to unaffected side abnormal
    - % humeral head anterior to scapular line (PHHA)
      - ~ 50% normally
  - T1 shows fatty atrophy of shoulder musculature

CLINICAL ISSUES

- Shoulder traction at delivery → nerve injury → muscle imbalance → unopposed internal rotation → posterior glenoid/anterior humeral head cartilage loading → glenohumeral dysplasia → posterior subluxation
- Most deficits recover by 3 months; 20-30% have permanent deficit; 8-10% develop posterior subluxation



(Left) *Frontal radiographs of the normal right shoulder (L image) & abnormal left shoulder (R image) in a 12-year-old with glenohumeral dysplasia secondary to a left brachial plexopathy show a small, ovoid left humeral head, winging of the left scapula, a hooked coracoid, & a dysplastic left glenoid.* **(Right)** *Axial NECT images of the right (L image) & left (R image) shoulders in the same girl show that the small left humeral head is posteriorly displaced, articulating with a false glenoid.*



(Left) *Axial GRE MR of abnormal right (L image) & normal left (R image) shoulders shows a low right glenoscapular angle (between the scapular & glenoid lines) with the humeral head posterior to the scapular line.* **(Right)** *Posterior axial US of the left shoulder (top) in an infant with left brachial plexopathy shows a high a angle (between the posterior scapular margin & a tangent to the humeral head from the posterior glenoid), indicating posterior subluxation. The right a angle (bottom) is normal.*

TERMINOLOGY

Synonyms

- Neonatal/perinatal brachial plexus palsy, birth palsy, brachial plexus birth injury (BPBI), shoulder contracture

Definitions

- Brachial plexopathy: Injury to ≥ 1 brachial plexus nerve roots, trunks, or cords → upper extremity contracture
- Glenohumeral dysplasia: Sequelae of brachial plexopathy on developing glenoid & humeral head

IMAGING

General Features

- Best diagnostic clue
  - Retroverted glenoid + posteriorly displaced humeral head

Radiographic Findings

- Small & ovoid humeral head
- Dysplastic glenoid, winged scapula, hooked coracoid

Ultrasonographic Findings

- Assesses humeral head prior to ossification
- Uses posterior axial approach to glenohumeral joint
- Measurements made with neutral, internal, & external rotation of humerus
- α angle: Angle between posterior scapular margin & tangent to humeral head from posterior edge of glenoid
  - Normal ≤ 30°
- % humeral head posterior to posterior scapular margin
  - Humeral head ossification center normally lies anterior to posterior scapular margin
    - Posterior position indicates posterior subluxation

MR Findings

- GRE & PD FS: Cartilage-sensitive sequences show unossified & ossified portions of glenoid & humeral head
- T1: Shows fatty atrophy of shoulder musculature
- Measurements on axial images
  - Glenoscapular angle (GSA): Posteromedial angle between scapular & glenoid lines
    - Scapular line: Line connecting medial most scapula & midglenoid cartilage
    - Glenoid line: Line parallels glenoid articular cartilage
  - Glenoid version angle = GSA minus 90°
    - Negative number = Glenoid retroversion
    - > 5° more retroversion vs. unaffected side indicates glenoid dysplasia
  - % humeral head anterior to scapular line (PHHAC)
    - Normal ~ 50%

Imaging Recommendations

- Best imaging tool
  - Ultrasound allows early detection & dynamic assessment of glenoid dysplasia without radiation or sedation
  - MR provides best overall articular & soft tissue evaluation
- Protocol advice
  - Compare affected vs. unaffected shoulders

DIFFERENTIAL DIAGNOSIS

Pseudoparesis of Upper Extremity

- Due to birth trauma → clavicular or humeral fracture

Arthrogryposis

- Infant born with ≥ 2 joint contractures

Cervical Spinal Cord Injury

- Affects bilateral extremities + bladder & bowel function

PATHOLOGY

General Features

- Etiology
  - Shoulder traction at delivery → nerve injury → muscle imbalance → internal rotation → posterior glenoid/anterior humeral head cartilage loading → glenohumeral dysplasia → posterior subluxation
    - Subluxation develops gradually in 1st year of life in infants with more severe birth injuries

Staging, Grading, & Classification

- Birch glenoid classification: Concave-flat, convex, biconcave
- Waters classification system: Glenoid version, humeral head coverage/position, ± pseudoglenoid

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depends on level of injured nerves (C5-T1)

Demographics

- Epidemiology
  - 3/1,000 live births

Natural History & Prognosis

- Most BPBI patients have temporary injuries & recover by 1-3 months of age; persistence depends on
  - Type of nerve injury
  - Number of roots involved
- 20-30% have residual neurologic deficits at 3 years
  - Posterior humeral head subluxation in up to 33% of permanent BPBI

Treatment

- Conservative: Physical therapy, Botox injections, & closed reduction
- Surgical: Anterior soft tissue release ± tendon transfers

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Hemophilia

KEY FACTS

TERMINOLOGY

- X-linked recessive bleeding disorder resulting from clotting factor deficiencies
  - Hemophilia A (> 80% of cases): Factor VIII deficiency
  - Hemophilia B (< 15% of cases): Factor IX deficiency
- Hemophilic arthropathy: Progressive joint destruction due to recurrent hemarthrosis, synovitis, & erosions

IMAGING

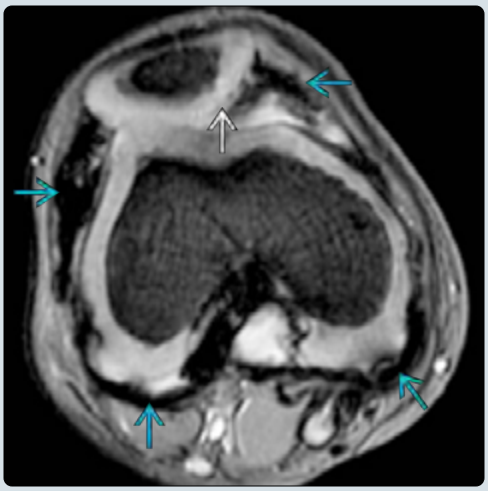
- Location of arthropathy: Knee > elbow > ankle > shoulder
- Radiograph: Hemophilic arthropathy
  - Distention of joint capsule by effusion & synovitis with eventual cartilage damage, erosions, & subchondral cysts
- MR: Hemophilic arthropathy
  - Variable signal intensity & heterogeneity of effusions due to blood products of different ages
  - Chronic hemosiderin deposits line synovium
    - Low signal intensity on all sequences with characteristic "blooming" of signal loss on T2* GRE
  - Eventual development of osteocartilaginous erosions
- US: Hemophilic arthropathy
  - Effusion of variable echogenicity/complexity
  - Synovial thickening with hyperemia by Doppler
  - Thinning, irregularity of hypoechoic cartilage

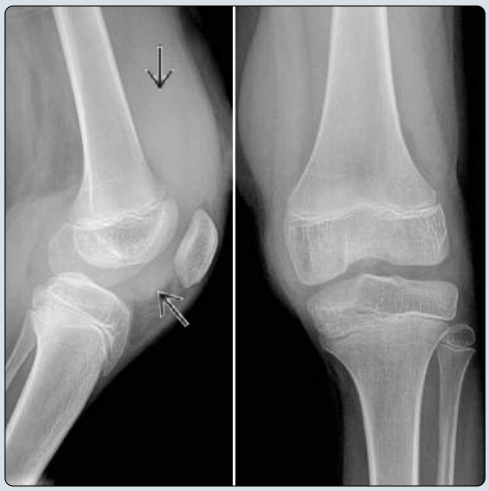
TOP DIFFERENTIAL DIAGNOSES

- Synovial venous malformation
- Pigmented villonodular synovitis
- Juvenile idiopathic arthritis

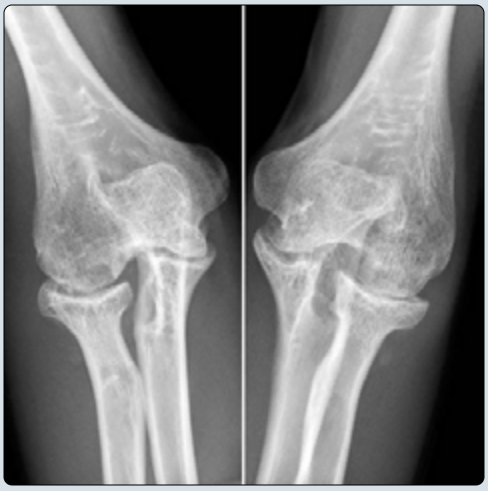
CLINICAL ISSUES

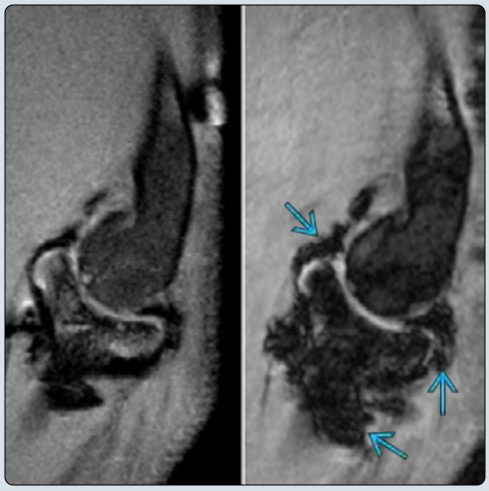
- Prevention: Aggressive prophylaxis with clotting factors
  - Significantly ↓ joint bleeds & chronic degeneration
- Acute hemarthrosis: Factor replacement + joint aspiration/irrigation
- Chronic hemarthrosis: Synovectomy (surgical or radiosynovectomy); arthroplasty





(Left) *Axial T2* GRE MR of the knee in a 7-year-old with hemophilia A shows extensive "blooming" (exaggerated signal loss) along the joint capsule due to prior episodes of hemarthrosis. Early cartilage irregularity is seen at the patellar medial facet.* **(Right)** *Lateral & AP knee radiographs in the same patient 6 years later at a time of swelling show a large dense joint effusion. The epiphyses are mildly large & irregular due to repeated hemarthroses & synovitis causing local hyperemia & cartilage damage.*





(Left) *AP radiographs of the bilateral elbows in the same patient 5 years later show the long-standing effects of recurrent hemarthroses, including irregular, misshapen, & overgrown epiphyses with joint space narrowing. These findings are due to synovitis with hyperemia (particularly during skeletal growth) & osteochondral injury.* **(Right)** *Sagittal PD FS (L) & T2* GRE (R) MRs from the same patient show "blooming" of synovial hemosiderin. There is irregularity & thinning of the articular cartilage with small erosions.*

TERMINOLOGY

Synonyms

- Hemophilia A: Factor VIII deficiency
- Hemophilia B: Factor IX deficiency, Christmas disease

Definitions

- Hemophilia: X-linked recessive bleeding disorder due to clotting factor deficiencies
- Hemophilic pseudotumor: Nonneoplastic mass from focally recurrent intraosseous, subperiosteal, or soft tissue bleeds

IMAGING

Radiographic Findings

- Hemophilic arthropathy (very common)
  - Distention of joint capsule by effusion & synovitis
  - Hyperemia leads to
    - Overgrowth (ballooning) of epiphyses
    - Premature physeal fusion → limb shortening
  - Inflammatory synovitis → cartilage destruction, erosions, subchondral cysts

Ultrasonographic Findings

- Effusion (hemorrhage) showing variable complexity
- Synovial thickening with hyperemia by Doppler
  - ↑ power Doppler signal of synovium → ↑ bleeding risk
- Thinning, irregularity of hypoechoic cartilage

MR Findings

- Hemophilic arthropathy
  - Variable signal intensity & heterogeneity of effusions due to blood products of different ages
  - Chronic hemosiderin deposits line synovium
    - Low signal intensity on all sequences with characteristic "blooming" of signal loss on T2* GRE
    - Hypertrophied enhancing synovium
  - Eventual development of osteocartilaginous erosions

Imaging Recommendations

- Best imaging tool
  - Ultrasound for acute hemarthrosis
  - MR provides superior assessment of entire joint

DIFFERENTIAL DIAGNOSIS

Hemophilic Arthropathy

- Synovial venous malformation
  - Blood-filled intraarticular mass leads to hemarthrosis, synovitis, erosions, degeneration
  - Infiltrating or discrete lobular &/or tubular fluid signal intensity masses with septations
    - ± fluid-fluid levels, thrombi
- Pigmented villonodular synovitis
  - Diffuse intraarticular form: Intermixed hemosiderin deposits & enhancing proliferative synovium often have radiating frond-like appearance
- Juvenile idiopathic arthritis
  - Joint effusion ± rice bodies; blood products uncommon
  - Synovitis leads to erosions & growth disturbances
    - Similar chronic radiographic appearance to hemophilia

PATHOLOGY

General Features

- Etiology
  - Sites of bleeds depend on clotting factor levels
    - Musculoskeletal system (exposed to repetitive mild trauma) accounts for up to 90% of sites
      - Joints: 70-80%; muscles: 10-20%

Staging, Grading, & Classification

- 2012 International Prophylaxis Study Group (IPSG) MRI arthropathy scale

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Acute hemarthrosis: Tense, swollen, red, painful joint occurring spontaneously or with minor trauma
  - Subacute or chronic hemarthrosis: Degenerative arthrosis, contractures

Demographics

- Age
  - 1st episode of joint hemorrhage by 2-3 years of age
- Gender
  - Males only (x-linked recessive)
- Epidemiology
  - Hemophilia A (> 80% of cases): 1:5,000 male patients worldwide
  - Hemophilia B: 1:30,000 male patients worldwide

Natural History & Prognosis

- Hemarthrosis classically in 70-90%
  - Substantial reduction by
    - Early initiation (< 2 years of age) of aggressive prophylactic replacement regimen of synthetic clotting factors (rather than isolated expectant administration)
      - Some patients will develop antibodies (inhibitors) to replacement factors
    - Joint aspiration ± irrigation upon bleed

Treatment

- Hemophilic arthropathy
  - Acute bleed: Clotting factor, joint aspiration/irrigation
  - Chronic: Synovectomy (surgical vs. radiosynovectomy); if end stage → arthroplasty

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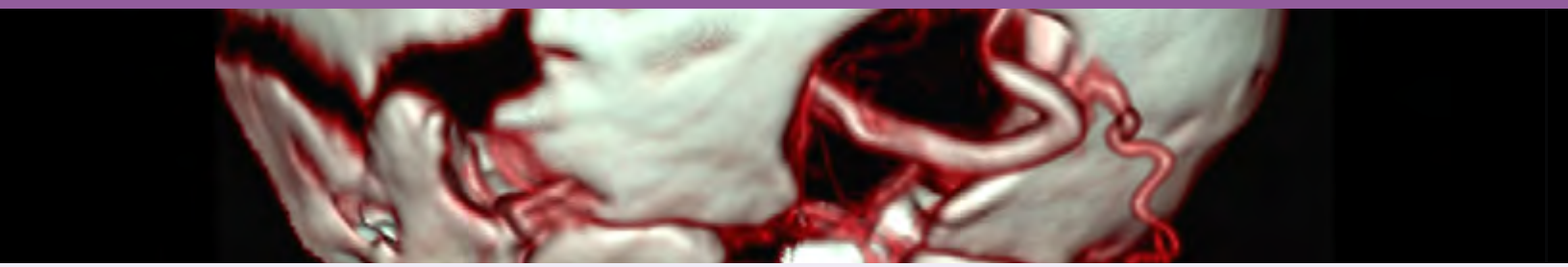
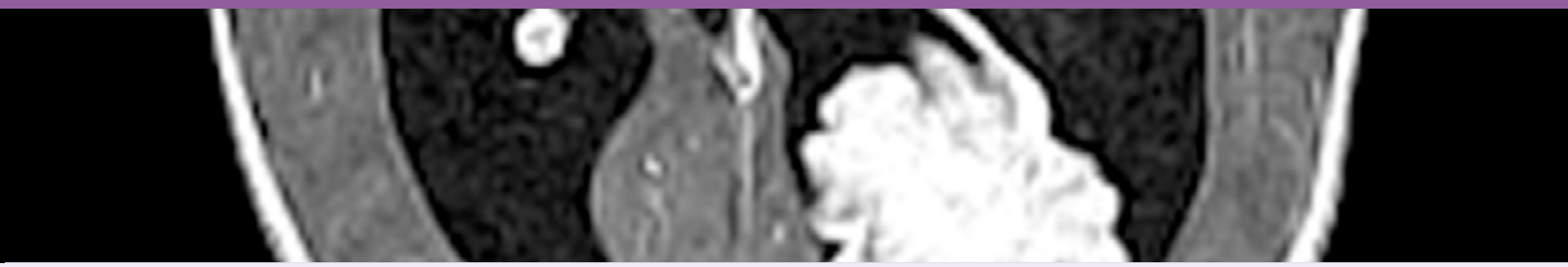
SECTION 7

Brain





Traumatic and Vascular Lesions

Metabolic, Infectious, and Inflammatory Disorders

Overview

This 3rd edition of Diagnostic Imaging: Pediatrics presents another great opportunity to expand & update the neuroradiology sections. As in the 2nd edition, we continue to add new diagnoses to the book. Length constraints mean that many diagnoses are only 1 or 2 pages long. We hope readers will see the increased number of diagnoses as outweighing the decrease in length & will explore the additional material in the Elsevier ebook, Expert Consult, where they will find expanded descriptions, case examples, & selected references to support the printed text.

The most difficult task in organizing the Brain section is deciding what to include & what to leave out. The brain section is divided into 6 subsections: Normal Developmental Variation, Congenital Malformations, Phakomatoses, Cysts & Neoplasms, Traumatic & Vascular Lesions, & Metabolic, Infectious, & Inflammatory Disorders. The included diagnoses are not meant to be all-encompassing but were selected to be the most helpful to the pediatric radiology practitioner. Some topics, such as Pineal Cyst & Enlarged Perivascular Spaces, are included because they are so common. Conversely, Vein of Galen Aneurysmal Malformations are rarely encountered but are a complex & classic pediatric entity that may be overwhelming to approach without a resource to provide direction. Some pathologies that were combined in the 2nd edition, such as Gray Matter Heterotopia, Polymicrogyria, & Focal Cortical Dysplasia, now more appropriately stand alone, but several combined topics, such as Demyelinating Diseases, still exist.

There has been an increase in the number of CNS neoplasm diagnoses in this edition, & most neoplasm sections have undergone extensive updates addressing our rapidly evolving understanding of the molecular & genetic underpinnings of pediatric CNS tumors. In contrast to the 2007 WHO CNS tumor classification, which relied heavily on the histologic characteristics of tumors, the new 2016 WHO CNS tumor classification relies heavily on molecular & genetic tumor markers to provide greater insight into the prognosis & optimal therapy for these neoplasms going forward. A number of important imaging markers that correlate well with molecular subtypes have been identified in pediatric CNS tumors, including both medulloblastoma & ependymoma. It is expected that radiogenomics, or the correlation of imaging & molecular data, will be a major area of research & clinical interest in the coming years.

Similar to past editions, this section contains chapters on leukodystrophies & mitochondrial encephalopathies. Both of these groups contain vast numbers of specific conditions, the majority of which are not discussed. It is hoped that the more common & characteristic entities presented will give the reader a better understanding of these complex conditions as a whole. Likewise, the Metabolic Diseases chapter attempts to introduce some of the other major groups of inborn errors of metabolism that the pediatric imager may encounter.

Terminology

There is a constant process in science & medicine of renaming entities to more accurately reflect new knowledge gained. This can make understanding, teaching, & researching these disease processes difficult. When a disease has many names, a comprehensive literature search becomes significantly more difficult to confidently perform. For this reason, we have made every effort to be comprehensive in listing the synonyms used both in the past & present to describe certain disease processes. We have also updated the titles of certain diagnoses (e.g., White Matter Injury of Prematurity rather than Periventricular Leukomalacia & Hypoxic-Ischemic Encephalopathy rather than Perinatal Asphyxia) to reflect evolving descriptions found in recent medical literature.

Normal Development

One of the great challenges in pediatric neuroimaging is understanding the dynamic development of the human brain from the fetus to the adult. Understanding this normal development allows for accurate identification of normal vs. abnormal & gives the imaging professional a conceptual framework to understand many CNS diagnoses, especially the congenital malformations. For instance, when evaluating a fetal MR of the brain, it is critical to know the expected sulcation pattern for a given gestational age in order to identify possible malformations of cortical development. A delayed sulcation pattern may suggest a malformation of cortical development on the spectrum of lissencephaly or gyral simplification, whereas the presence of too many sulci for gestational age may suggest polymicrogyria.

During the first 2 years of life, an understanding of the normal myelination pattern allows for accurate identification of conditions associated with delayed myelination (e.g., Pelizaeus-Merzbacher disease). Because understanding normal myelination is so critical, this edition includes a 4-page chapter dedicated to the topic. Another important point regarding myelination is that certain diseases have very different imaging characteristics during the 1st year of life compared with later years. A prime example of this can be seen in Tuberous Sclerosis in which cortical tubers & radial migration lines are T1 hyperintense & T2 isointense prior to myelination & exactly the opposite (T1 hypointense & T2 hyperintense) following myelination. Tuberous sclerosis is just 1 of many diagnoses with such dynamic imaging characteristics over time.

One final example of the importance of understanding normal CNS development can be found in the pituitary, which undergoes both age- & sex-specific changes in size & signal characteristics with time. The anterior pituitary in the newborn is very T1 hyperintense, often equal to that of the posterior pituitary. This is normal & should not be mistaken for hemorrhage or other pathology. This hyperintensity is presumed to be related to maternal hormones, as the same finding is not seen in early premature infants when imaged at term equivalent age. The anterior pituitary slowly decreases in T1 signal intensity & becomes isointense to the pons after the 1st few months of life. Regarding pituitary size, the normal pituitary often has a convex superior margin at birth, slowly becoming flat or concave over the ensuing weeks, an appearance that is typically maintained in male patients throughout life & in female patients until puberty. The imaging professional should not be alarmed to see an enlarged pituitary with a convex superior border in a teenage girl. This is a normal appearance in this age, & sex should not in & of itself raise suspicion for a pituitary lesion.

Hydrocephalus

Hydrocephalus is a distressingly common entity in pediatrics, the consequence of a multitude of primary pathologies that result in alterations in the production, distribution, & resorption of cerebrospinal fluid (CSF). In order to understand hydrocephalus, it is important to recognize that the CNS relies

on the ventricles, subarachnoid space (SAS), arachnoid granulations, pia, arachnoid, & dural sinuses to manage the flow & distribution of interstitial fluid, just as lymphatics perform this function in the liver, lungs, & other large organs. Thus, obstruction within the ventricular system will prevent egress of intraventricular CSF into the SAS, & increasing pressure within the ventricles will cause interstitial fluid to accumulate in the periventricular white matter (transependymal edema). Elevated pressure in the dural sinuses will prevent the transmission of CSF from the SAS into the sinuses, as will alteration in arachnoid granulation function caused by inflammation or metabolic disorders (communicating hydrocephalus). The obstruction of CSF flow through the foramen magnum & obex caused by the Chiari 1 malformation will prevent CSF in the spinal canal from reaching the sinuses, leading to accumulation of fluid in the central canal of the cord, or syringohydromyelia. A clear understanding of the normal pathways of CSF flow will allow the neuroradiologist to be confident in diagnosing alterations of these pathways that lead to hydrocephalus.

Cerebral Edema

A common & important neuroimaging finding is the development of brain edema, of which 4 basic types are recognized. Vasogenic edema reflects expansion of the interstitial fluid compartment in response to an irritating stimulus such as tumor or inflammation. Cytotoxic edema is due to expansion of the intracellular fluid compartment secondary to failure of the sodium-potassium pump regulation of the cell membrane. In transependymal edema, increased hydrostatic pressure in the ventricular system prevents the normal progression of interstitial fluid into the ventricles & causes congestion in the periventricular white matter. Perhaps the least understood is posttraumatic edema, in which diffuse brain swelling is caused by a combination of cytotoxic & vasogenic edema resulting from a cascade of events that include sodium-potassium pump failure, diminished cerebral perfusion & autoregulation, & loss of integrity of the blood-brain barrier. Posttraumatic cerebral edema typically sets in several hours after the initial injury & can lead to rapid increases in intracranial pressure & death. Clinical management of traumatic brain injury usually includes imaging reassessment within the first 12 hours after presentation to look for progression of initially identified injuries, such as contusions & intracranial hemorrhage. The radiologist must be diligent in looking for signs of increased brain swelling at this time also; such signs include effacement of previously identifiable sulci at the vertex & decrease in size of basal cisterns. The presence of these signs should lead to more aggressive monitoring & management of intracranial pressure. Waiting until clinical signs develop is too late to effectively combat the cycle of reduced cerebral perfusion & progressive cellular injury. Diffuse cerebral edema is the leading cause of death from cases of child abuse, & the traumatic nature of the insult may not be readily apparent at first presentation. Nontraumatic insults, such as acute encephalitis & status epilepticus, can also lead to this potentially fatal complication.

Imaging Protocols

The development & use of high-quality indication-specific imaging protocols are essential to the practice of pediatric neuroradiology. For CT imaging in the age of the "Image Gently" campaign, much of the focus has been appropriately on the reduction of radiation exposure to the patient. When an indicated CT exam is performed, it is imperative that we extract the maximum information from this study. One of the best ways to do so is to perform multiplanar reconstructions (MPR) in coronal & sagittal planes. MPRs have been shown to increase the detection of small intracranial hemorrhages & reduce the number of false positives by accurately characterizing some findings as artifactual. The same can be said for the detection of skull fractures, which is improved with MPR as well as 3D surface rendering.

Brain MR protocols for the neonate should be adjusted to confidently identify perinatal injuries, congenital malformations, & metabolic derangements. For the infant younger than 3 years, a focus on myelin maturation is key, with T1WI included in all protocols. Conversely, FLAIR sequences are typically avoided in the child under 24 months, as these may cause confusion in the incompletely myelinated brain. For the child over 24 months, adult-type sequences can be employed but should be specifically tailored to answer the clinical question at hand.

Volumetric MR is becoming increasingly important in pediatric imaging, with most weightings (e.g., T1, T2, PD, FLAIR, & SSFP) now available on most platforms. Volumetric imaging is optimal for tumor follow-up, giving the imaging professional confidence in tumor size measurements from 1 exam to the next. Volumetric FLAIR has become a critical sequence in seizure protocols to increase sensitivity for the often subtle findings of focal cortical dysplasia. 3D SSFP (e.g., FIESTA, CISS) images remove CSF flow artifact & provide high spatial resolution for the optimal evaluation of masses with a CSF interface. Finally, volumetric imaging is helpful because it allows reconstruction in multiple planes with no penalty in exam length.

Pediatric radiologists should be eager to employ advanced MR techniques, such as diffusion tensor imaging (DTI), susceptibility weighted imaging (SWI), & arterial spin labeling (ASL). DTI provides high-quality DWI & ADC derivatives, & the ability to perform tractography is very helpful in preoperative tumor & epilepsy planning. SWI provides highly sensitive detection of hemorrhagic foci in the brain parenchyma & important insight into the vascular supply to tumors & malformations. ASL provides the opportunity to acquire brain perfusion information without the need for contrast administration, thereby dramatically increasing the number of patients who can benefit from the added information perfusion analysis can provide.

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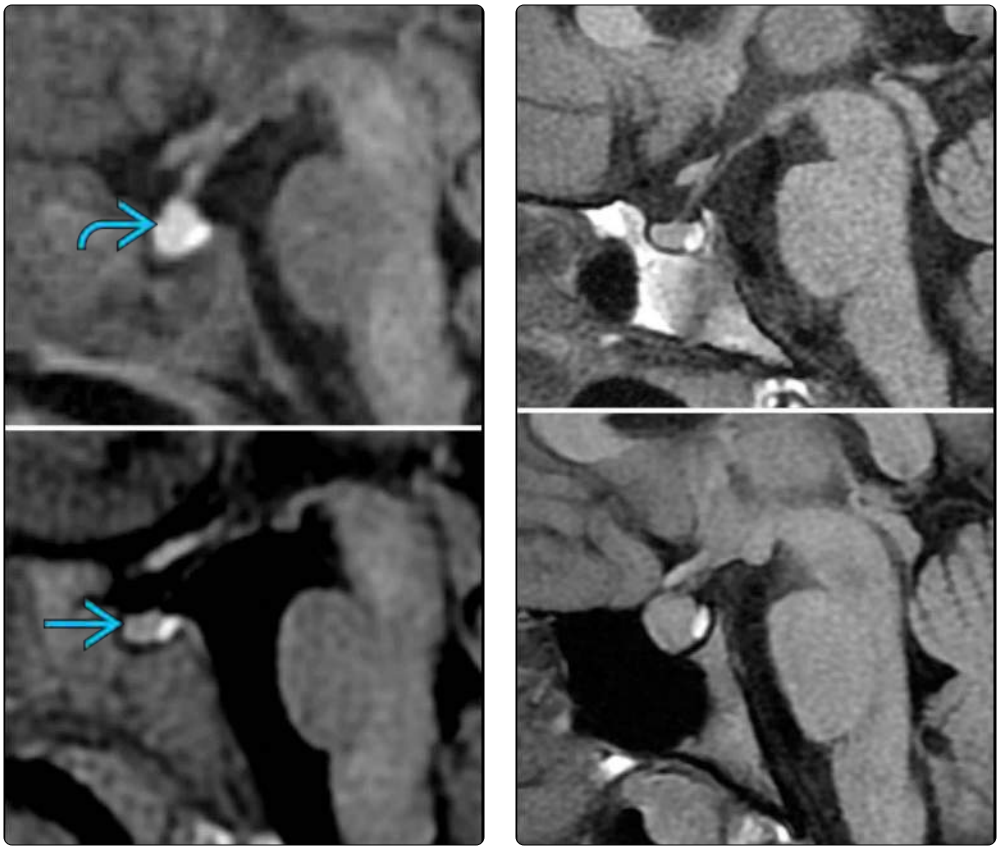
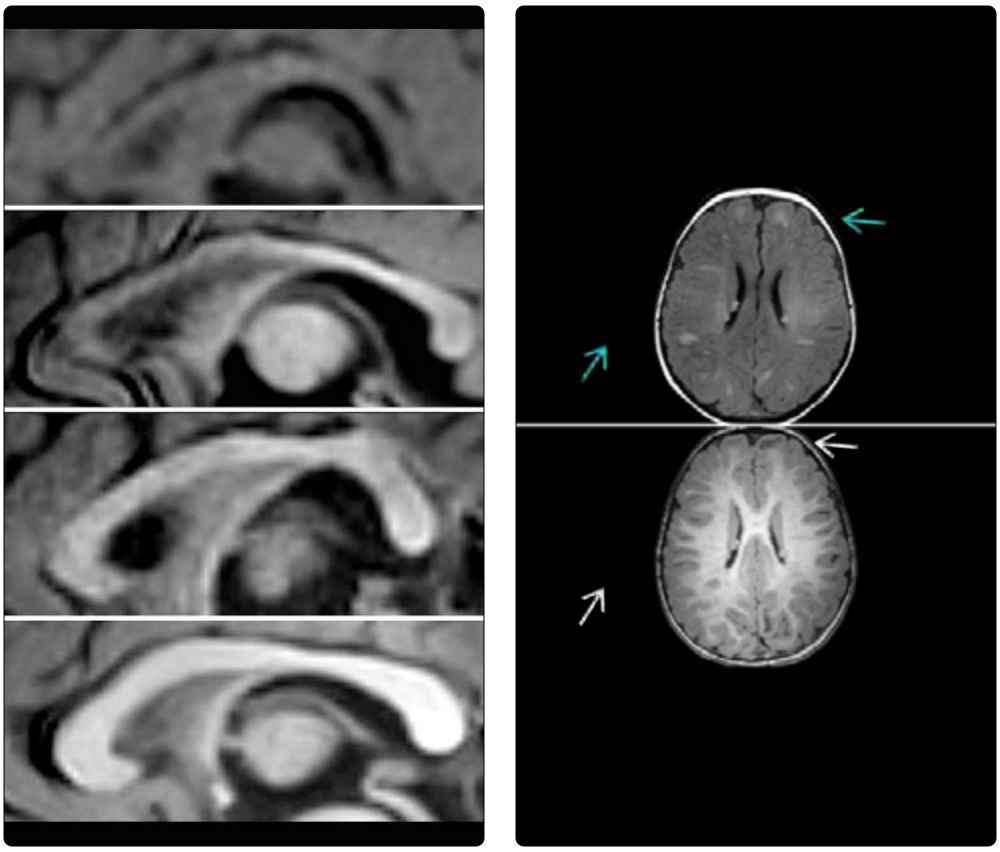
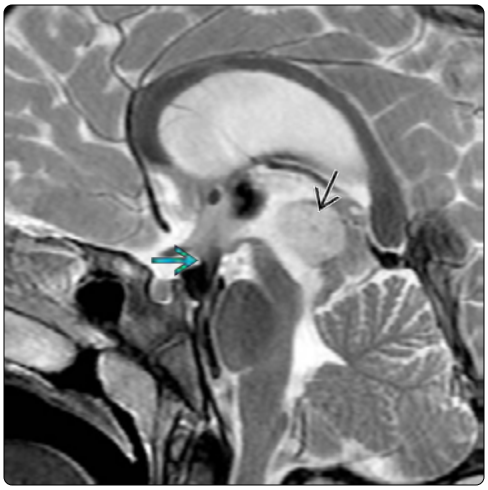
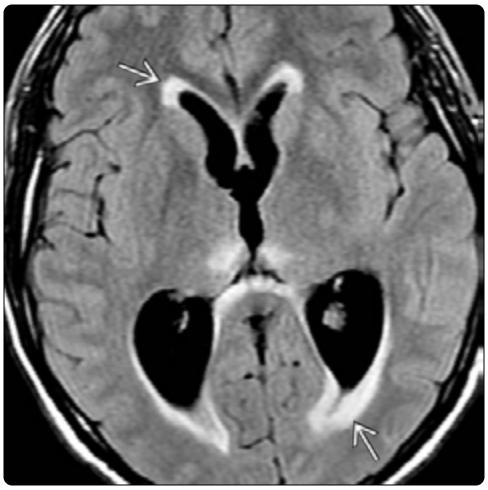
(Left) *Sagittal T1 MRs in a 7-day-old infant (top) & a 2-month-old infant (bottom) show the typical imaging evolution of the neonatal anterior pituitary, which is often very hyperintense (sometimes equal to the posterior pituitary) early with a superior convex border. This signal & morphology are presumably related to maternal hormones, as early premature infants imaged at term equivalent ages do not have this appearance. Over the 1st few weeks of life, the anterior pituitary becomes isointense to the pons with a flat or concave superior border.* **(Right)** *Sagittal T1 MRs in a 6-year-old girl (top) & a 17-year-old girl (bottom) show typical appearances of the anterior pituitary at these ages. An enlarged (up to 9 mm vertical) anterior pituitary with a convex superior border in a teenage girl should not raise concern.*



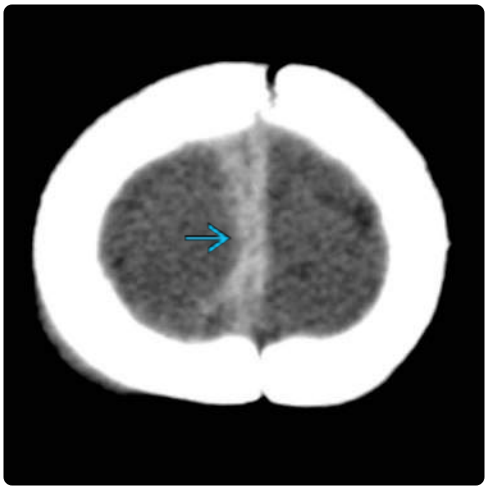
(Left) *Sagittal T1 MRs at 2, 4, 7, & 10 months (top to bottom, respectively) show development of the corpus callosum during the 1st year of life. Progressive myelination & thickening of the corpus callosum begin with the posterior body & splenium. The posterior 1/2 of the corpus callosum continues to thicken as the anterior body, genu, & rostrum begin to myelinate at 4 months. By the end of the 1st year of life, the corpus callosum is completely myelinated with uniform signal intensity.* **(Right)** *Axial T1 MRs a patient with tuberous sclerosis complex at 3 months (top) & 10 months (bottom) show that, prior to significant myelination, the cortical tubers & radial migration lines are hyperintense. Following myelination, these lesions show varying degrees of hypointensity compared with adjacent white matter.*

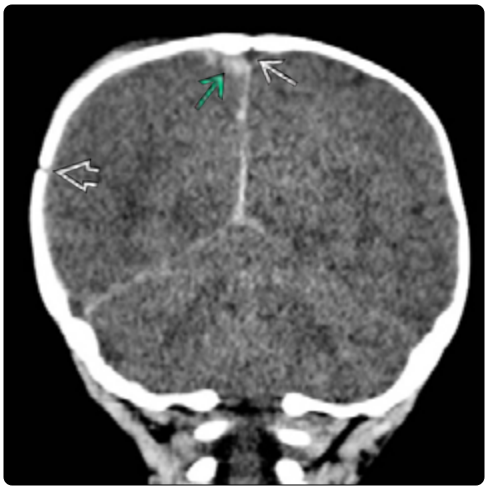




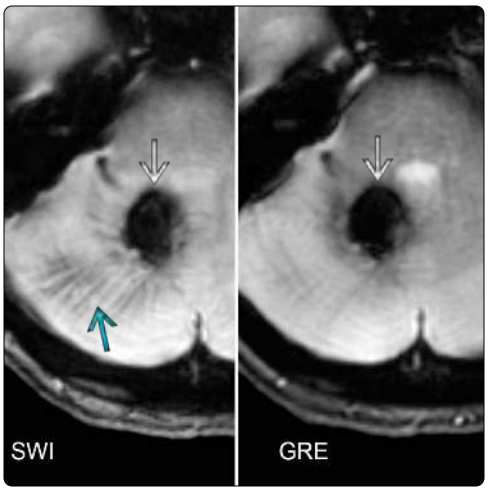


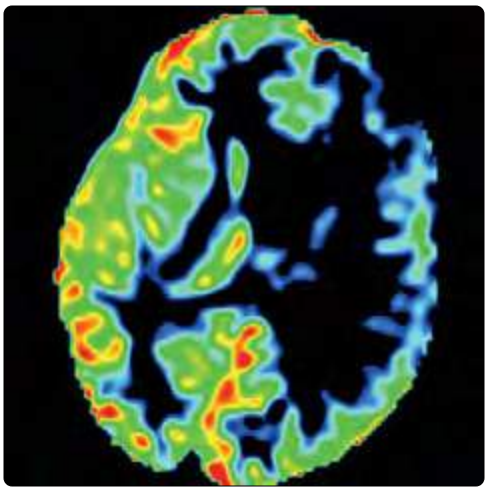
(Left) *Midline sagittal T2 MR in a 6-year-old child with a tectal mass shows obstruction of the cerebral aqueduct with mass effect on the midbrain. Treatment is an endoscopic 3rd ventriculostomy, which appears patent as evidenced by the dark flow artifact across the floor of the 3rd ventricle.* **(Right)** *Axial FLAIR MR in a 19-year-old man with a pineal germinoma shows ventriculomegaly with transependymal edema in the frontal & occipital regions (where hydrostatic pressure of interstitial fluid is highest).*





(Left) *Axial NECT in a 6-month-old infant with nonaccidental trauma shows hyperdensity in the region of the superior sagittal sinus. With axial images alone, it can be difficult to distinguish subdural hemorrhage from venous sinus thrombosis.* **(Right)** *Coronal NECT in the same patient clearly delineates the superior sagittal sinus from the adjacent subdural hemorrhage. Also note the axially oriented right parietal bone fracture, which was difficult to see on axial images.*





(Left) *Axial SWI (L) & T2* GRE (R) MRs in a patient with a cavernous malformation show the increased conspicuity of the associated developmental venous anomaly on SWI compared with GRE.* **(Right)** *Axial ASL in a 10-year-old boy with Sturge-Weber syndrome shows substantially decreased perfusion within the left frontal lobe (which was severely affected with pial angiomatosis & parenchymal atrophy). ASL is a perfusion technique that does not require contrast administration.*

Normal Myelination

KEY FACTS

IMAGING

- In general, myelin maturation proceeds from caudal to rostral, central to peripheral, dorsal to ventral
- Gray-white differentiation accentuated on NECT in neonate due to high water content of white matter (WM)
- T1WI & T2WI necessary to assess myelination on MR
  - T1WI: Key sequence in assessing myelination < 1 year
    - Bright signal reflects presence of proteolipid protein
  - T2WI: Key sequence in assessing myelination at 1-2 years
    - As myelin sheaths thicken, surrounding interstitial (extraaxonal) water displaced
    - Drop in T2 signal lags behind development of bright T1 signal
      - Myelin sheath has to thicken considerably before enough water displaced to reduce signal
    - Conventional double spin-echo sequences preferred for T2WI
      - PD/Intermediate echo images especially valuable < 24 months: Helps detect pathology
- Diffusion tractography (DTI) MR can elucidate fiber tracts as they become myelinated

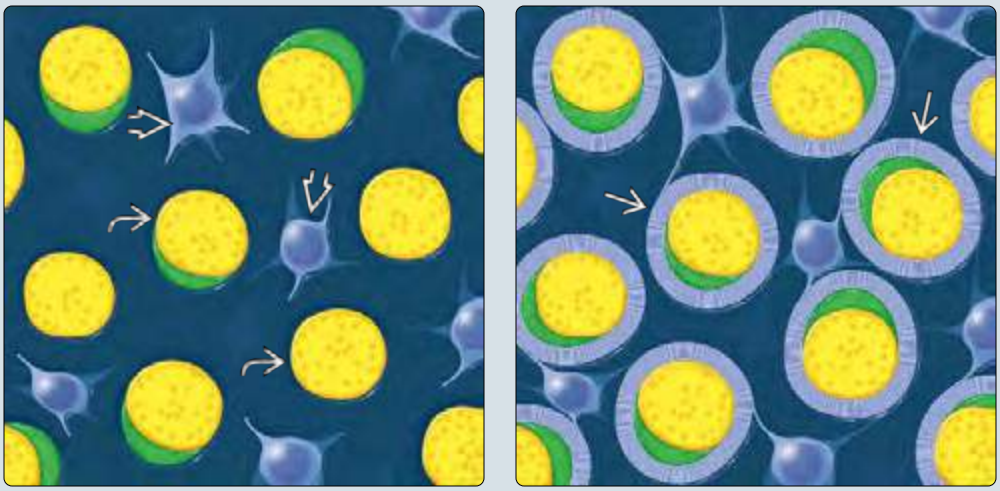
CLINICAL ISSUES

- Assessment of myelination essential to interpreting MR in children
- MR demonstration of normal myelination closely parallels developmental functional milestones
- All children should achieve adult appearance of WM on all MR sequences by 36-40 months

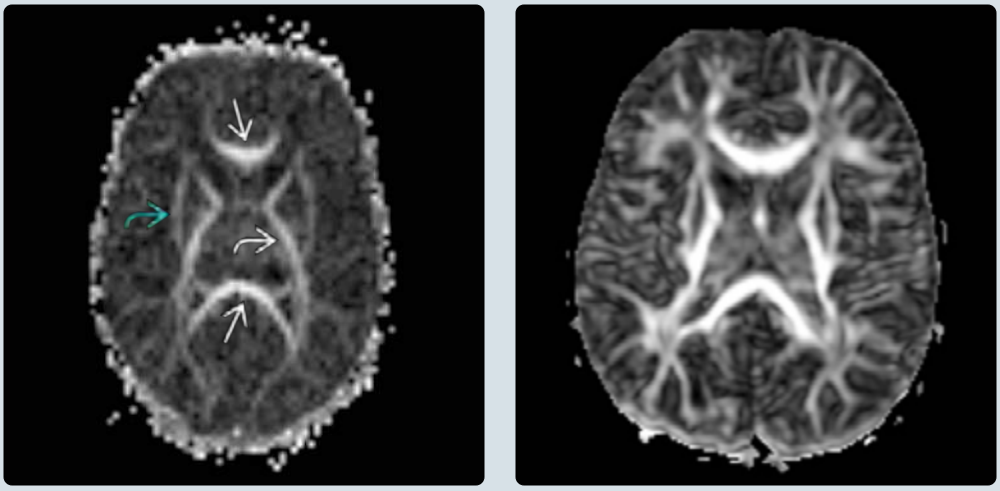
DIAGNOSTIC CHECKLIST

- Know gestational age before assigning myelination stage
  - Normal myelination milestones based upon postconception age, regardless of degree of maturity at delivery

(Left) *Oligodendrocytes lie in proximity to axons after migration. The presence of proteolipid protein in the oligodendrocytes results in T1 shortening (T1 bright signal) on MR, but the large amount of interstitial water (dark blue background) results in T2 prolongation (T2 bright signal) on MR.* **(Right)** *With time, the oligodendrocytes lay down multiple layers of myelin around each axon. As it thickens, the hydrophobic myelin drives out the interstitial water, causing reduced T2 signal (or T2 shortening) on MR.*



(Left) *Axial fractional anisotropy (FA) map in a newborn shows bright signal in the corpus callosum & internal & external capsules, reflecting the relatively dense axonal population & early myelination in these regions. FA maps can demonstrate white matter tracts prior to their delineation on conventional MR sequences.* **(Right)** *Axial FA map in a 9 month old shows extension of bright signal throughout the brain, reflecting the greater structural organization resulting from myelination.*



TERMINOLOGY

Definitions

- Process of myelin sheath development around axons throughout central nervous system
  - Begins in 5th fetal month & continues throughout life

IMAGING

General Features

- Best diagnostic clue
  - Appearance of myelination on imaging reflects presence of proteolipid & its effect on interstitial water
  - Myelin development correlates with functional milestones
- Location
  - In general, myelin maturation proceeds caudal to rostral, central to peripheral, dorsal to ventral
- Size
  - White matter (WM) tracts, especially corpus callosum, ↑ in size with myelin formation

CT Findings

- NECT
  - Gray-white differentiation accentuated in neonate due to high water content of WM
    - Unmyelinated WM is hypodense relative to gray matter & to myelinated WM
      - Reflects high ratio of interstitial water to axons
  - Density ↑ with myelination is relatively subtle, making CT insensitive to detecting delays in myelination

MR Findings

- T1WI
  - Key sequence in assessing myelination < 1 year
  - Bright signal reflects presence of proteolipid protein (PLP)
    - PLP expressed by mature (myelinating) oligodendrocytes as they start laying down myelin sheath
    - Extent of bright signal on T1WI reflects distribution of myelinating oligodendrocytes in infant brain
  - Detection of myelination (represented by hyperintense signal) progresses in predictable fashion
    - By term
      - Dorsal brainstem
      - Dentate nucleus
      - Optic tracts
      - Anterior commissure
      - Posterior limb of internal capsule
      - Rolandic & perirolandic gyri
      - Pyramidal tracts
    - By 2 months
      - Splenium of corpus callosum
      - Anterior limb internal capsule
      - Early optic radiations
    - By 4 months
      - Genu of corpus callosum
      - Optic radiations become more apparent
      - Peripheral rami in pyramidal tracts (perirolandic gyri)
    - By 6 months
      - Genu & splenium are equally hyperintense
      - Peripheral rami in parietal & occipital lobes become hyperintense
    - By 8 months
      - All but most peripheral rami of frontal gyri are hyperintense
    - By 10-12 months
      - Adult appearance of myelin achieved on T1WI
- T2WI
  - Key sequence in assessing myelination in children 1-2 years of age
  - As myelin sheaths thicken, surrounding interstitial (extraaxonal) water displaced
    - PLP is hydrophobic
    - Less interstitial water → darker signal on T2WI
  - Drop in T2 signal lags considerably behind presence of bright T1 signal
    - Process starts immediately after oligodendrocytes begin laying down myelin sheath
    - Sheath has to thicken considerably before enough water is displaced to reduce signal
  - Detection of myelination (represented by hypointense signal) progresses in predictable fashion
    - By term
      - Dorsal brainstem
      - Part of posterior limb of internal capsule
      - Perirolandic gyri
    - By 4 months
      - More hypointense signal in rolandic & perirolandic gyri
      - Splenium of corpus callosum
      - More anterior extension in internal capsule
    - By 8 months
      - Genu & splenium of corpus callosum
      - Anterior limb of internal capsule
      - Decreasing signal in centrum semiovale & optic radiations
      - Decreasing signal in basal ganglia & thalamus
    - By 12 months
      - External capsule hypointense signal becomes apparent
      - Expansion of centrum semiovale hypointensity
      - Clearly defined peripheral rami around central sulcus & in occipital poles
    - By 16 months
      - Better definition of deep nuclei in brainstem & basal ganglia
      - Peripheral rami in parietal lobes become hypointense
    - By 18 months
      - All but most peripheral frontal WM rami are now hypointense
      - Some residual hyperintense signal around trigones of lateral ventricles ("terminal zones") due to greater hydrostatic pressure in this region from confluence of WM tracts
    - By 36 months
      - Adult appearance of myelin achieved on T2WI
- PD/intermediate

- Very helpful in distinguishing gliosis from lack of myelination
  - Gliosis brighter than "terminal zones"
- FLAIR
  - Relatively "flat" images in immature brains
  - Signal changes associated with myelination (hyperintense to hypointense) similar to T2WI
  - Difficult to distinguish pathology from normal in infant
  - Tend to occur 2-3 months after changes visible on T2WI
    - Smaller amounts of interaxonal water may exert greater influence on FLAIR sequences
- DWI
  - ADC values predate T1/T2WI signal changes
  - Presence of myelin has significant effect on ability of water to diffuse
    - Fractional anisotropy (FA) ↑ with brain maturation
    - Diffusion perpendicular to myelin sheaths restricted with ↓ in extraaxonal water
    - Diffusivity along axon ↑
  - Diffusion tractography (DTI) can elucidate fiber tracts as they become myelinated
    - Correlates with functional milestones
    - May allow more specific identification of developing functional tracts
    - Mean diffusivity & FA measurements can be used to assess integrity of tracts
- MRS
  - Changes in relative metabolite concentrations in 1st 2 years of life may reflect myelination
  - Myoinositol & choline high in neonate
    - Choline ↓ with myelination
  - NAA ↑ with myelination in 1st year of life

Imaging Recommendations

- Best imaging tool
  - Use both T1WI & T2WI to assess myelination
    - T1WI under 12 months
    - T2WI from 12-24 months
- Protocol advice
  - IR may ↑ sensitivity to T1 shortening
  - FSE sequences may minimize appearance of abnormal hyperintensity
  - Conventional double spin-echo sequences preferred for T2WI
    - PD/intermediate echo images especially valuable < 24 months

PATHOLOGY

General Features

- Etiology
  - Oligodendrocyte precursors proliferate in germinal matrix
  - Immature oligodendrocytes (prooligodendrocytes) migrate throughout brain
    - Follow distribution of neurons
    - Mature to myelinating oligodendrocytes after reaching destination
- Genetics
  - 2 major structural proteins of myelin: Myelin basic protein (MBP) & PLP
    - *MBP* gene encoded on chromosome 18q
    - *PDXP* gene encoded on chromosome Xq21-q22

Staging, Grading, & Classification

- Myelination assessed as "appropriate" for age or delayed
- Delay in myelination should prompt investigation for possible causes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - MR demonstration of normal myelination closely parallels developmental functional milestones
  - Assessment of myelination essential aspect of interpreting MR in children
    - Analogous to documentation of developmental milestones by pediatrician

Demographics

- Age
  - All children should achieve adult appearance of WM by 36-40 months
- Gender
  - No significant male/female difference

Natural History & Prognosis

- Myelination progresses throughout life

Treatment

- Acquired disorders causing myelin delay can sometimes be treated

DIAGNOSTIC CHECKLIST

Consider

- Know gestational age before assigning myelination stage
  - Normal myelination milestones based upon postconception age, regardless of degree of maturity at delivery

Image Interpretation Pearls

- Use IR for T1WI < 10 months
- Use conventional double spin-echo sequences for T2WI

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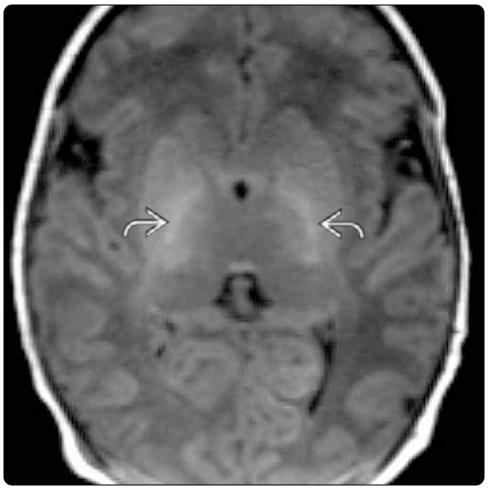
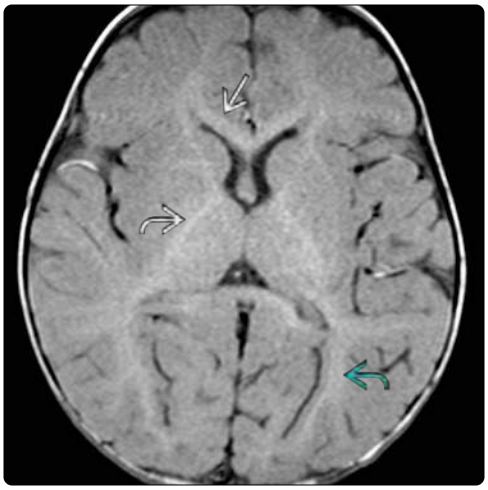
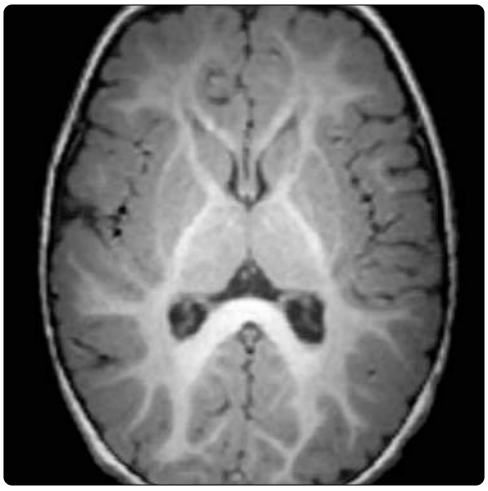
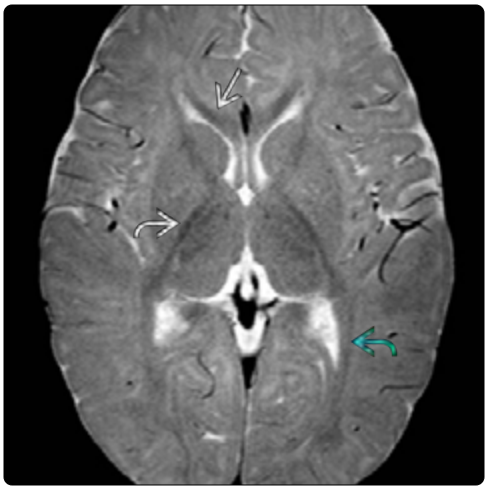


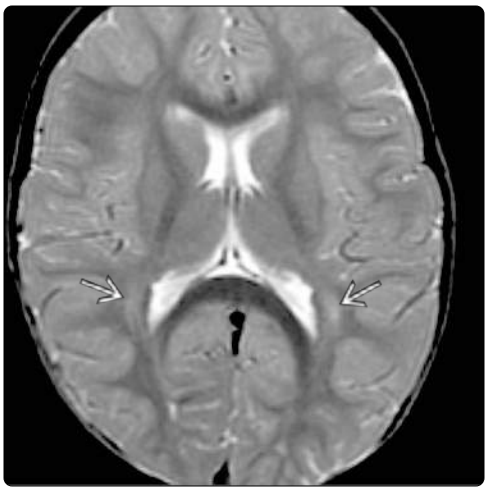


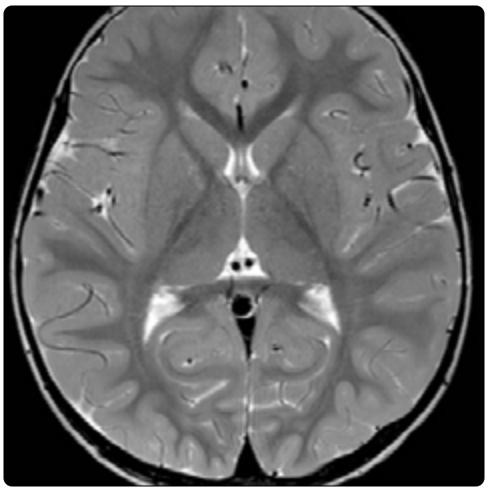
(Left) *Axial T1 MR in a term newborn shows faint bright signal in the posterior limbs of the internal capsules, indicating the presence of mature oligodendrocytes that are producing myelin in these regions.* **(Right)** *Axial T1 MR in a normal 6 month old shows faint bright signal in the internal capsules, corpus callosum, & optic radiations well before any hypointense signal will be obvious in these structures on T2WI.*





(Left) *Axial T1 MR in a 9 month old shows bright signal throughout the white matter. Myelination is far from complete at this stage, but the T1WI confirms the presence of mature oligodendrocytes throughout the brain.* **(Right)** *Axial T2 MR in another 9 month old shows hypointensity in the internal capsules, corpus callosum, & optic radiations areas that appear fully mature on T1WI. The displacement of interstitial water by the thickening myelin sheath lags far behind the signal changes on T1WI.*





(Left) *Axial T2 MR at 18 months of age shows some residual bright signal in the periventricular white matter of the parietal lobes. Displacement of interstitial water may lag in these regions due to the large volume of parenchyma contributing to centripetal CSF flow.* **(Right)** *Axial T2 MR in a 2 year old shows an adult pattern of myelination. Progressive myelination beyond this stage is not detectable by standard MR techniques.*

Enlarged Subarachnoid Spaces

KEY FACTS

TERMINOLOGY

- Idiopathic enlargement of subarachnoid spaces (SAS) during infancy

IMAGING

- Primary imaging modality: US
  - CT/MR used if fontanelle closing or to further investigate atypical clinical/US findings
- Best clue: Enlarged SAS & ↑ head circumference (> 95th percentile)
  - Ventricles may be mildly enlarged
- Symmetric bifrontal & bitemporal SAS
- All modalities show veins coursing through SAS
- SAS follow CSF appearance on all modalities
- No compression of veins or gyri
- No inward displacement of arachnoid membrane by subdural fluid
  - Small nonhemorrhagic subdural collections seen in ~ 4% of patients with enlarged SAS

TOP DIFFERENTIAL DIAGNOSES

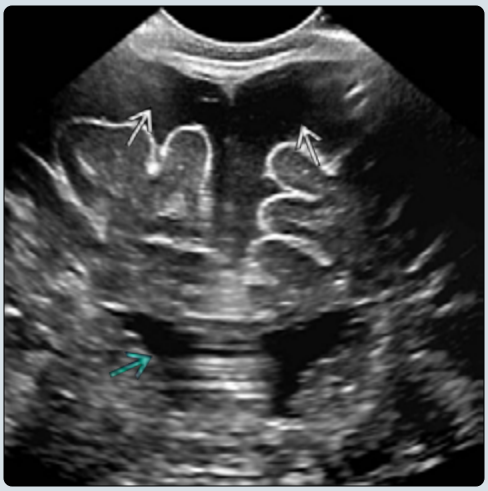
- Atrophy
- Acquired progressive communicating hydrocephalus
- Nonaccidental trauma (NAT)

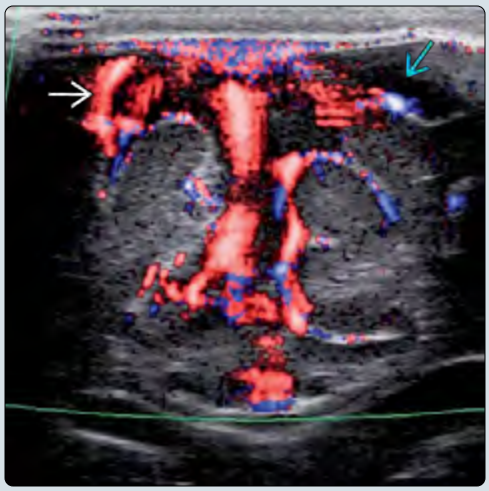
PATHOLOGY

- Etiology uncertain: Immature CSF drainage pathways likely
- Family history of macrocephaly > 80%

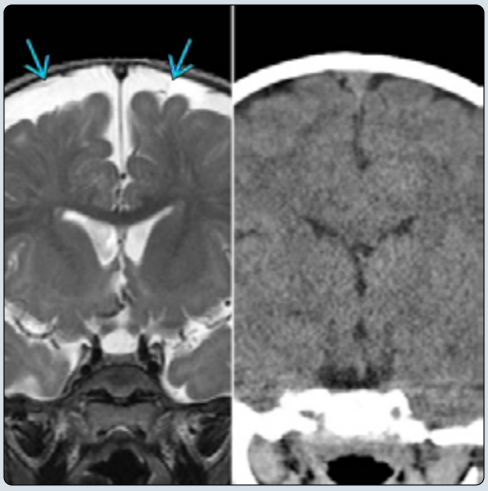
CLINICAL ISSUES

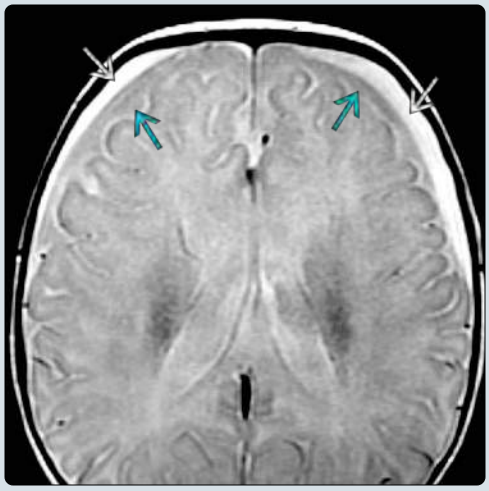
- Mild developmental delay alone should not prompt further imaging or subspecialty evaluation
  - Further evaluation required only in setting of focal neurologic signs &/or developmental regression
- Consider NAT if enlarged extraaxial spaces atypical
- SAS enlargement & developmental delay typically resolve without therapy by 2 years of age
- No treatment necessary





(Left) *Coronal US in a 7-month-old boy with macrocrania shows enlarged subarachnoid spaces (SAS) & normal ventricular size. Note the normal size of the sulci. This is a typical clinical history & imaging appearance for benign enlargement of the SAS.* **(Right)** *Coronal color Doppler US in a 4-month-old girl shows vessels traversing the enlarged SAS. Doppler US can be helpful to exclude subdural collections by demonstrating normal veins in the SAS.*





(Left) *Coronal T2 MR at 13 months (left) & NECT at 5 years (right) of age show expected resolution of the enlarged SAS over a 4-year period. Enlarged SAS typically resolve by 24 months of age.* **(Right)** *Axial PD MR in a 4-month-old girl with macrocrania shows enlarged SAS, which are isointense to the brain. Also note the small bilateral hyperintense subdural fluid collections, which can be seen in approximately 4% of patients with enlarged SAS.*

TERMINOLOGY

Abbreviations

- Subarachnoid spaces (SAS)
- Head circumference (HC)

Definitions

- Enlarged SAS in patient < 1 year of age with macrocrania (HC > 95%)

IMAGING

General Features

- Enlarged SAS in infant with macrocrania
  - Symmetric at bifrontal & bitemporal SAS
  - Normal SAS values differ significantly between studies
    - Interhemispheric width: 95th percentile: ~ 8 mm
    - Craniocortical width: 95th percentile: ~ 10 mm
    - Sinocortical width: 95th percentile: ~ 7 mm
- Ultrasound: Primary modality used whenever possible
  - Cortical veins seen within singular fluid space by grayscale & Doppler
    - No mass effect displacing veins against pia
    - No inward displacement of arachnoid membrane by subdural fluid
      - Subdural collections lack traversing veins
  - ± mild ventricular enlargement
- CT/MR: Used for screening when no acoustic window available for US (due to fontanelle closure) or if other neurologic signs/symptoms present
  - NECT: Enlarged SAS with normal sulci; no hemorrhage
    - Enlarged cisterns (especially suprasellar/chiasmatic)
  - CECT: Easily demonstrates veins traversing SAS
    - No abnormal meningeal enhancement
  - MR: Normal brain parenchyma without edema
    - SAS fluid follows CSF signal on all sequences
    - Small nonhemorrhagic subdural collections ~ 4%

Imaging Recommendations

- Protocol advice
  - After diagnosis, best follow-up: Clinical monitoring of HC & development of any neurologic findings
  - Follow-up with MR/CT typically not necessary, unless
    - Focal neurologic signs/symptoms
    - Suspicion for subdural collection on US

DIFFERENTIAL DIAGNOSIS

Atrophy

- Small HC; sulcal prominence out of proportion

Incidental Bilateral Subdural Fluid Collections

- Subdural fluid not normally visualized
  - Small nonhemorrhagic subdural collections seen in 4% of benign macrocrania patients
  - Characterized by crescentic fluid collection separating dura from arachnoid
    - No cortical veins traversing subdural space
    - Discrete arachnoid membrane displaced towards cortex, may be compressing SAS veins
    - May have different signal intensity on PD & other MR sequences compared to CSF
  - Discuss need for further work-up with referring clinician
    - Close clinical follow-up at minimum; work-up for nonaccidental trauma (NAT) at discretion of clinician

Nonaccidental Trauma

- Moderate/large or hemorrhagic subdurals or unusual clinical findings should raise concern

Glutaric Aciduria Type 1

- Enlarged sylvian fissures with delayed myelination
- Subdural collections may be present
- T2-hyperintense basal ganglia

Elevated Venous Pressures

- Causes: Cardiac disease, internal jugular vein sacrifice for ECMO, arteriovenous fistula, or sinus venous thrombosis

Communicating Hydrocephalus

- Often post hemorrhagic/post inflammatory/neoplastic

PATHOLOGY

General Features

- Etiology
  - Immature CSF drainage pathways: Most accepted theory
  - Family history of macrocephaly > 80%
- Associated abnormalities
  - Predisposition to bleed with minor trauma: Controversial
    - Possibility of ↑ risk for bridging vein injury & subdural collection/hematoma in absence of major trauma

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Macrocrania: HC > 95th percentile
  - Danger signs
    - Persistent or rapid deviation of HC from normal curve
    - Developmental regression, focal neurologic signs, vomiting, bruising
- Other signs/symptoms
  - Mild developmental delay common (20-50%) & usually resolves over time

Natural History & Prognosis

- Self-limited; resolves without therapy by 12-24 months

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Crucial to know HC
- Further evaluation with brain MR or CT if US atypical
  - Moderate/large/complex subdural collection → NAT work-up
  - Even small/simple subdural collections should be discussed with referring clinician to identify any concerns for NAT that merit further work-up

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Dandy-Walker Continuum

KEY FACTS

TERMINOLOGY

- Dandy-Walker continuum is clinically & radiologically heterogeneous group of PF malformations
  - Classic Dandy-Walker malformation (DWM)
  - Vermian hypoplasia (VH)
  - Blake pouch cyst (BPC)
  - Mega cisterna magna (MCM)

IMAGING

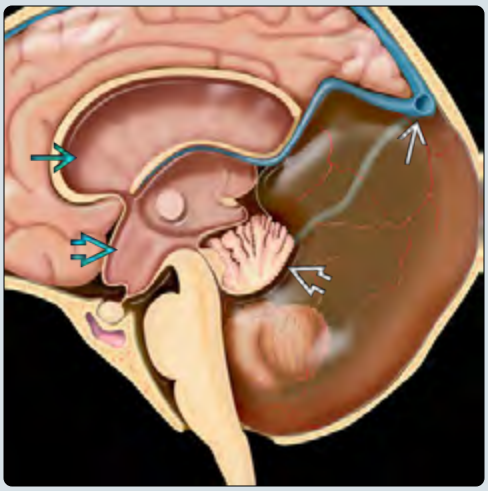
- Most severe to mildest: Classic DWM → VH → BPC → MCM
- Classic DWM
  - Triad of vermian agenesis/hypogenesis with 4th ventricle cystic dilatation with enlarged posterior fossa with elevation of torcular Herophili (torcular-lambdoid inversion)
  - ± hydrocephalus
- VH
  - Variable vermian hypoplasia ± superior rotation/elevation of vermian tissue
  - No PF enlargement or torcular-lambdoid inversion
- BPC
  - Elevated tegmento-vermian angle: Open 4th ventricle
  - Normal size & morphology of vermis
  - Cyst wall often not seen by imaging
- MCM
  - Enlarged retrocerebellar CSF cistern (> 10 mm)
- Occipital bone may appear scalloped/remodeled with all DW continuum types (including MCM)

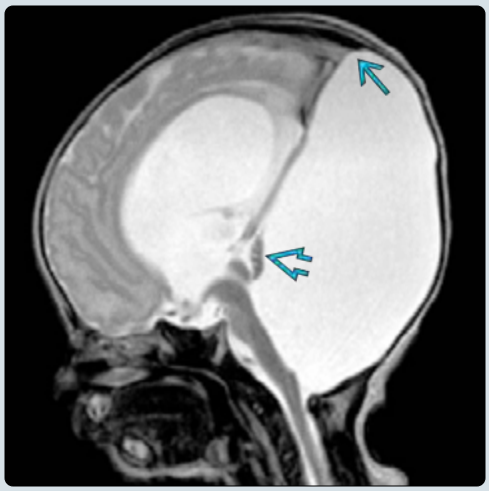
TOP DIFFERENTIAL DIAGNOSES

- PF arachnoid cyst
- Joubert syndrome & related disorders
- Congenital muscular dystrophy
- Cerebellar hypoplasia

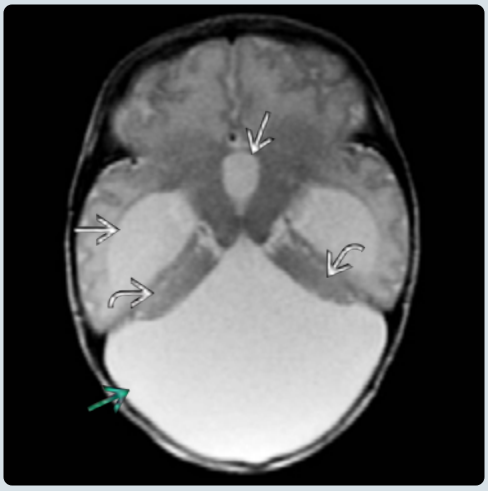
CLINICAL ISSUES

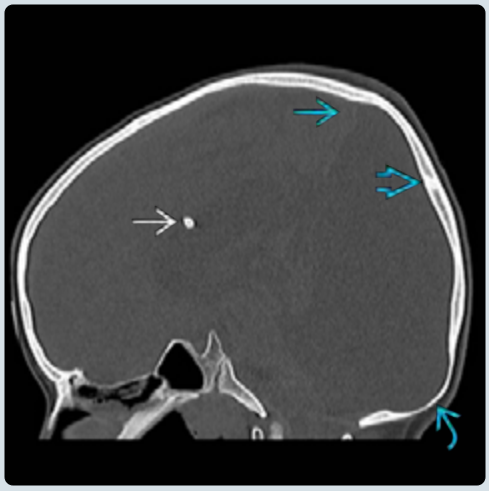
- Multiple associated syndromes & genetic mutations
- Wide range of clinical severity





(Left) *Graphic of the classic DWM shows an enlarged posterior fossa, elevated torcular Herophili, superior rotation of a hypoplastic cerebellar vermis, & posterior cystic expansion of the 4th ventricle. Lateral & 3rd ventriculomegaly is shown.* **(Right)** *Sagittal T2 MR shows the classic DWM in a newborn. The torcular Herophili is elevated with upward sloping tentorium. The vermis is extremely hypoplastic & rotated. The 4th ventricle communicates with a large posterior fossa cyst.*





(Left) *Axial T2 MR in the same patient with classic DWM shows hypoplastic cerebellar hemispheres splayed by the large posterior fossa cyst, which communicates with the 4th ventricle. Note the ventriculomegaly.* **(Right)** *Sagittal CT in bone windows of the same patient with a classic DWM at age 7 (after ventricular shunting) is shown. The torcular Herophili lies above the lambdoid suture (torcular-lambdoid inversion). There is remodeling of the occipital bone with scalloping & expansion.*

TERMINOLOGY

Abbreviations

- Classic Dandy-Walker malformation (DWM)

Synonyms

- Terms DW spectrum, DW variant, & DW complex have muddled clinical & imaging implications in literature

Definitions

- DW continuum represents clinically & radiologically heterogeneous group of posterior fossa (PF) malformations
- Referred to as continuum by some based on belief that DWM, vermian hypoplasia (VH), Blake pouch cyst (BPC), & mega cisterna magna (MCM) are spectrum of developmental anomalies of rhombencephalic vesicle roof
  - Terminology controversial & not uniformly accepted
  - Clinical severity ranges widely in this group

IMAGING

General Features

- Best diagnostic clue
  - Classic DWM triad: Vermian agenesis/hypogenesis with 4th ventricle cystic dilatation with enlarged PF with elevation of torcular Herophili
  - VH, BPC: Failed closure of 4th ventricle
- Morphology
  - DW continuum (from most to least severe)
    - DWM classic triad
      - Complete or partial agenesis of vermis
      - Cystic dilatation of 4th ventricle → rotation of hypoplastic vermis
      - Enlarged PF with upward displacement of tentorium & torcular Herophili (torcular-lambdoid inversion)
    - VH
      - Variable degree of VH ± rotation
      - No PF enlargement, no torcular-lambdoid inversion
    - BPC
      - Elevated tegmento-vermian angle: Open 4th ventricle
      - Normal size & morphology of vermis
    - MCM
      - Enlarged retrocerebellar cistern (> 10 mm) without mass effect on hindbrain
      - Normal vermis/4th ventricle/tegmento-vermian angle
      - In isolation, incidental finding

Radiographic Findings

- Radiography
  - Enlarged calvaria, particularly PF
  - DWM: Torcular-lambdoid inversion (transverse sinus grooves elevated above lambda)

CT Findings

- NECT
  - DWM: Large PF
    - Torcular-lambdoid inversion (torcular above lambdoid sutures)
  - Scalloped occipital bone, remodeled in all types of DW continuum

MR Findings

- T1WI, T2WI, FLAIR
  - DWM
    - Vermian agenesis/hypogenesis, hypoplastic vermis with superior rotation/elevation
    - Variable cerebellar hemisphere & brainstem hypoplasia &/or compression
    - Enlarged 4th ventricle communicating with retrocerebellar cyst
    - Elevated torcular Herophili with steep tentorium
    - Hydrocephalus common, though not originally described as part of DWM
  - VH
    - Varying degrees of vermian tissue present
    - Vertical height of vermis does not reach level of obex
    - ± rotation of vermis
  - BPC
    - Rotated but normal-appearing vermis
    - 4th ventricle communicates inferiorly with retrocerebellar CSF cyst
    - Cyst wall usually imperceptible on MR, often difficult to distinguish from VH & MCM
    - Choroid plexus may be seen along inferior surface of vermis (superior margin of cyst wall)
      - Can help distinguish from VH
  - MCM
    - Enlarged retrocerebellar CSF space
    - Normal vermis (not rotated or hypoplastic)
  - ± callosal anomalies, polymicrogyria, gray matter heterotopia, occipital cephalocele, myelination delay
- MRV
  - Elevated torcular Herophili (DWM)

Ultrasonographic Findings

- Pre- & postnatal US will visualize similar findings to MR

Imaging Recommendations

- Best imaging tool
  - MR best characterizes severity, associated anomalies
- Protocol advice
  - Routine MR with sagittal SSFP to look for cyst wall

DIFFERENTIAL DIAGNOSIS

Posterior Fossa Arachnoid Cyst

- True cyst that does not communicate with 4th ventricle
- ± mass effect on adjacent structures
- Included in DW continuum by some authors

Joubert Syndrome & Related Disorders

- Hypoplastic/dysplastic vermis, "bat wing" 4th ventricle, thickened & horizontal superior cerebellar peduncles with midbrain cleft → molar tooth appearance on axial images

Congenital Muscular Dystrophy

- VH with cobblestone cortex & Z-shaped brainstem

Cerebellar Hypoplasia

- ± brainstem hypoplasia (pontocerebellar hypoplasia)

PATHOLOGY

General Features

- Etiology
  - Vermis not formed by fusion of cerebellar hemispheres
  - Rhombencephalic vesicle roof divides into cranial [anterior membranous area (AMA)] & caudal [posterior membranous area (PMA)] segments
    - AMA invaded by neural cells → cerebellum
    - PMA expands then disappears to form outlet foramina of 4th ventricle
  - Hindbrain development arrested
    - Defective AMA & PMA → DWM & VH
    - Defective PMA only → BPC (incomplete/nonperforation of foramen of Magendie) & MCM (delayed perforation)
- Genetics
  - Multiple causative genes identified
    - *FOXC1* (chromosome 6p25.3)
    - Deletion of 3q24 (includes *ZIC1* & *ZIC4* genes)
- Associated abnormalities
  - 2/3 of DWM & related disorders have associated CNS &/or extracranial anomalies
    - Midline anomalies, neurocutaneous melanosis, PHACES
    - Craniofacial, cardiac, urinary tract, & orthopedic anomalies
    - Trisomy 18 > other trisomies
- Embryology
  - Common association of DWM/VH with facial, cardiovascular anomalies suggests onset between formation, migration of neural crest cells
    - 3rd-4th postovulatory week

Staging, Grading, & Classification

- Spectrum based on imaging findings: Classic DWM (most severe) → VH → BPC → MCM (mildest)

Gross Pathologic & Surgical Features

- DWM: Large PF with cystic dilatation of 4th ventricle
  - Roof of 4th ventricle in continuity with cyst wall that attaches to hypoplastic vermis anteriorly, cerebellar hemispheres laterally, & medulla caudally
  - 4th ventricle choroid plexus absent or displaced into lateral recesses

Microscopic Features

- DWM: Cyst wall has 3 layers
  - Outer: Continuous with leptomeninges
  - Intermediate: Stretched neuroglial layer, continuous with vermis
  - Inner: Glial tissue lined with ependyma/ependymal nests

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - DWM: Macrocephaly, bulging fontanel (obstructive hydrocephalus)
  - MCM: Incidental finding
- Clinical profile
  - Marked heterogeneity in clinical findings, even in families with same genetic mutations
  - Motor developmental delay, spastic paraplegia, seizures, variable intellectual disability

Demographics

- Age
  - DWM: 80% diagnosed by 1 year of age
    - Age of diagnosis depends on degree of hydrocephalus, supratentorial anomalies, & cerebellar dysfunction
- Epidemiology
  - 1:25,000-30,000 births
  - Accounts for 1-4% of all hydrocephalus cases

Natural History & Prognosis

- Cognitive outcome dependent upon associated syndromes, supratentorial anomalies, & hydrocephalus
- Classic DWM: Poor prognosis overall
  - Though developmental delay previously reported in 40-60%, recent literature suggests much higher incidence in true DWM
  - 80% with hydrocephalus
  - Seizures, hearing &/or visual difficulties, systemic abnormalities
- VH: Highly variable
  - Cognitive abnormalities in 40%-50%, though normal neurodevelopment has been reported in isolated VH
- BPC: Favorable outcome in isolation; however, significant proportion associated with other anomalies
- MCM: Normal neurodevelopmental outcome in isolation

Treatment

- CSF diversion if hydrocephalus: Ventriculoperitoneal shunt ± cyst shunt or marsupialization

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Thin, sagittal views crucial for delineation, diagnosis

Reporting Tips

- Terms in report, such as DW spectrum, variant, continuum, can be confusing
  - Use clear description & best categorization of abnormal PF findings
  - Note hydrocephalus & supratentorial anomalies

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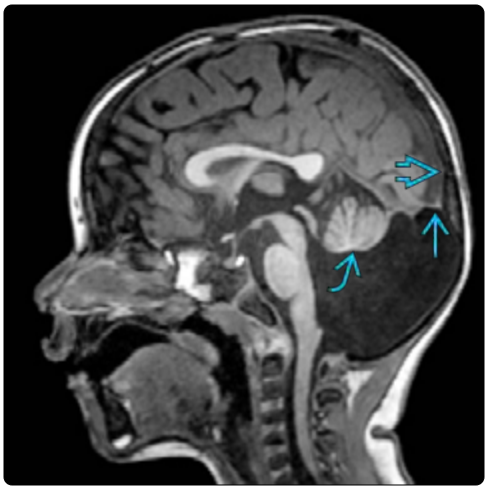
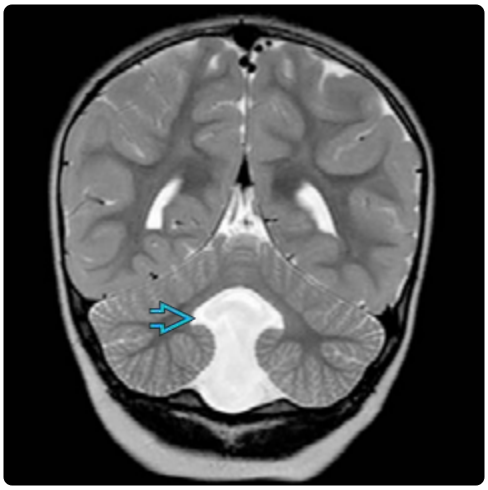
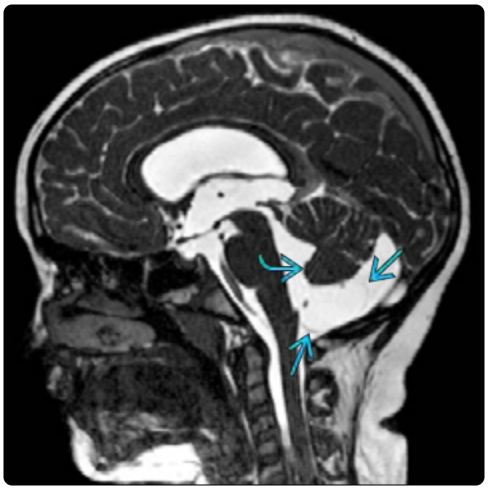
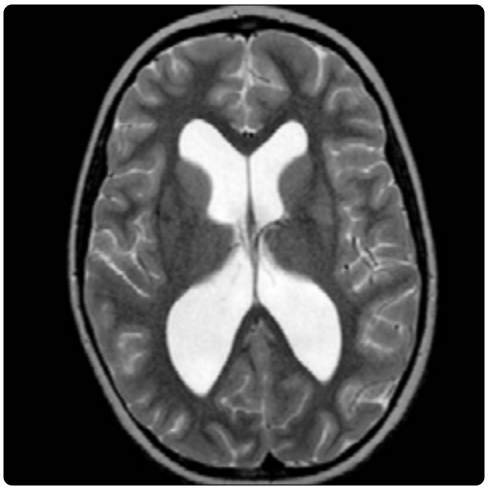


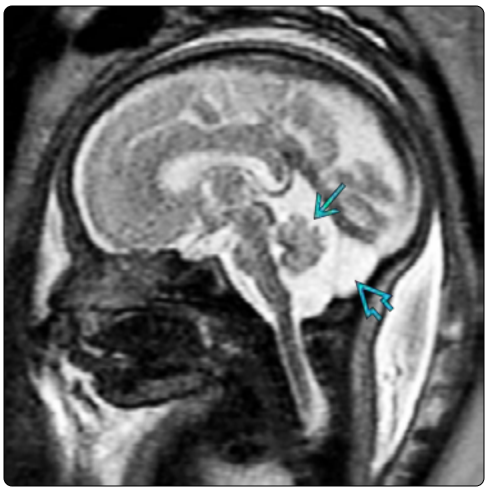


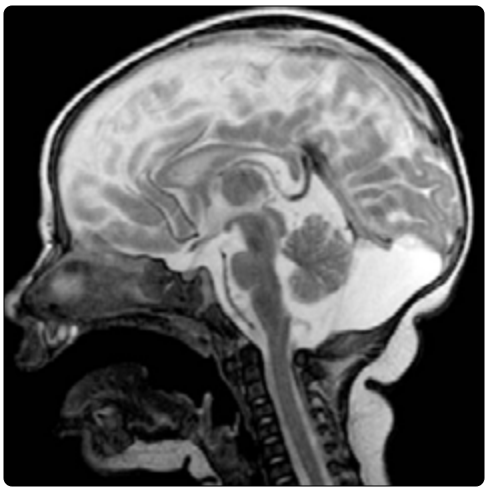
(Left) *Sagittal T1 MR shows vermian hypoplasia with rotation in a 3-year-old patient. The torcular Herophili is inferior to the lambdoid suture. The hypoplastic vermis is rotated.* **(Right)** *Coronal T2 MR of vermian hypoplasia in the same patient shows relatively normal cerebellar hemisphere development, creating a keyhole appearance of the 4th ventricle.*





(Left) *Sagittal SSFP MR of a Blake pouch cyst in an 8-year-old child shows elevation & mild rotation of an otherwise intact vermis. The cyst, which communicates with the 4th ventricle, has thin walls.* **(Right)** *Axial MR T2 in the same patient demonstrates ventriculomegaly. Depending on the degree of fenestration, a Blake pouch cyst can cause some degree of obstruction to normal CSF flow.*





(Left) *Sagittal SSFSE fetal MR at 33-weeks gestation shows a mega cisterna magna. In this patient, the cisterna magna is enlarged, measuring 15 mm in AP dimension. The vermis is normal in size, morphology, & orientation.* **(Right)** *Postnatal sagittal T2 MR of the same patient at 6 weeks of age. The cisterna magna remains enlarged with a normal vermis. In isolation, this is an incidental finding.*

KEY FACTS

TERMINOLOGY

- Rhombencephalosynapsis (RS)
- Lhermitte-Duclos disease (LDD) (dysplastic cerebellar gangliocytoma, Cowden syndrome)
- Joubert syndrome & related disorders (JSRD) (molar tooth malformation)

IMAGING

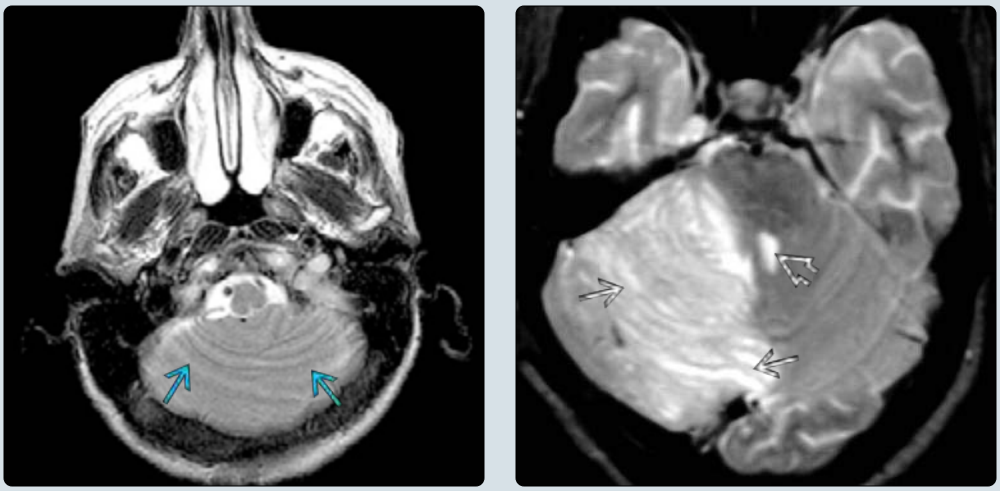
- RS: Incomplete separation of cerebellar hemispheres
  - Partial or complete absence of cerebellar vermis with folia continuous across midline
  - ~ 65% have coexisting aqueductal stenosis & hydrocephalus
- LDD: Well-circumscribed ↑ T2, ↓ T1 expansile lesion with enlarged & dysplastic cerebellar folia causing characteristic tiger stripe or corduroy striated pattern
- JSRD: Molar tooth configuration of brainstem marked by absent decussation of superior cerebellar peduncles
  - Thickened & horizontally oriented superior cerebellar peduncles, deep & broad interpeduncular fossa, deformed 4th ventricle, hypoplastic vermis

TOP DIFFERENTIAL DIAGNOSES

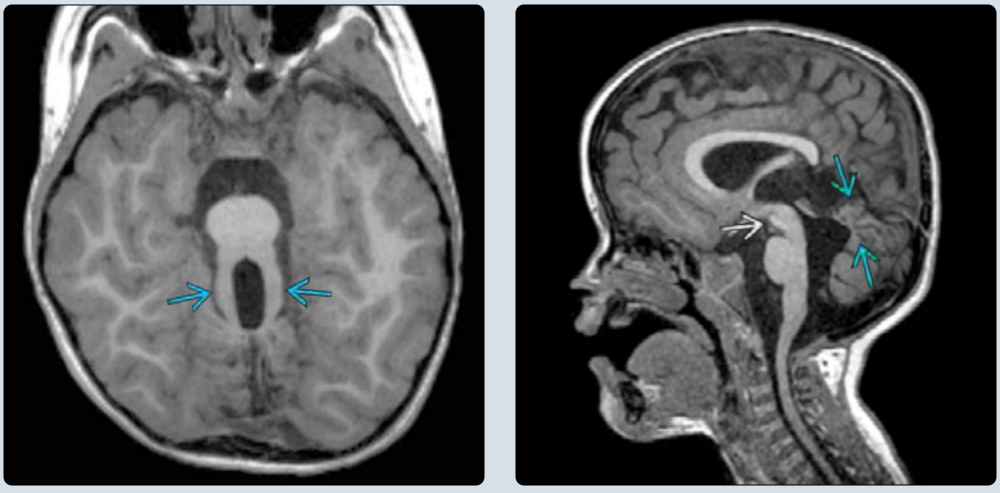
- Dandy-Walker continuum
- Congenital muscular dystrophy
- Isolated cerebellar hypoplasia
- Pontocerebellar hypoplasia

CLINICAL ISSUES

- RS: Widely varied depending on degree of associated anomalies; isolated cases may have subclinical symptomatology
- LDD: Longstanding history of poorly localized neurological symptoms
- JSRD: Clinically heterogeneous, often with hyperpnea + central apnea in neonatal period, abnormal eye movements



(Left) *Axial T2 MR in a patient with rhombencephalosynapsis shows lack of separation of the cerebellar hemispheres with transversely oriented folia that are continuous across the midline. No discrete vermis is seen.* **(Right)** *Axial T2 MR shows the expansile, striated, hyperintense lesion characteristic of Lhermitte-Duclos disease. Note the 4th ventricular compression.*



(Left) *Axial T1 MR in a 4-year-old patient shows a classic molar tooth configuration of the midbrain, consistent with Joubert syndrome & related disorders (JSRD). There is characteristic thickening & elongation of the superior cerebellar peduncles.* **(Right)** *Sagittal T1 MR in the same patient with JSRD demonstrates marked cerebellar vermian hypoplasia & dysplasia with flattening of the fastigial recess. There is also narrowing of the midbrain at the pontomesencephalic junction.*

TERMINOLOGY

Abbreviations

- Rhombencephalosynapsis (RS)
- Lhermitte-Duclos disease (LDD)
- Joubert syndrome & related disorders (JSRD)

Synonyms

- LDD: Cowden syndrome, dysplastic cerebellar gangliocytoma, diffuse hypertrophy of cerebellar cortex
- JSRD: Molar tooth malformation

Definitions

- RS: Incomplete separation of cerebellar hemispheres with absence or hypoplasia of vermis
- LDD: Focal enlargement (probably hamartomatous) of cerebellar cortex
- JSRD: Absence of decussation of superior cerebellar peduncles leading to molar tooth sign

IMAGING

General Features

- Best diagnostic clue
  - RS: Midline continuation of cerebellar hemispheres (a.k.a. "fusion") with at least partial absence of vermis
  - LDD: Sharply marginated region of enlarged & dysplastic cerebellar cortex with characteristic striated folial pattern on MR
  - JSRD: Molar tooth sign of midbrain with absence of decussation of superior cerebellar peduncles on DTI + small malformed vermis

MR Findings

- RS
  - Incomplete separation of cerebellar hemispheres: Diamond-shaped 4th ventricle on axial images, flattened fastigial recess, transversely oriented continuous folia, flat-based cerebellum
  - ± aqueductal stenosis with ventriculomegaly (~ 65%)
  - ± supratentorial anomalies of midline development: Deficiency/absence of septum pellucidum, corpus callosum hypogenesis/dysgenesis, absent anterior commissure, united fornices, incompletely separated thalami, holoprosencephaly, pituitary abnormalities
- LDD
  - Well-circumscribed, nonenhancing ↓ T1, ↑ T2 expansile cerebellar lesion
  - Enlargement of dysplastic cerebellar folia creating striated pattern
  - Mass effect can cause obstructive hydrocephalus
- JSRD
  - Molar tooth sign: Deep & broad posterior interpeduncular fossa, thick elongated superior cerebellar peduncles
  - Hypoplastic & dysplastic cerebellar vermis, high position of fastigium, superior midline vermian cleft on coronal images, enlarged posterior fossa
  - Deformed 4th ventricle: Triangular-shaped in midportion & batwing-shaped in superior portion on axial images
  - Hypoplasia of middle cerebellar peduncles, hypoplastic brainstem at pontomesencephalic junction, hypoplastic/dysplastic cerebellar hemispheres
  - ± supratentorial anomalies: Ventriculomegaly, ↓ cerebral volume, hypomyelination, corpus callosum dysgenesis

DIFFERENTIAL DIAGNOSIS

Dandy-Walker Continuum

- Absence of vermis with complete separation of cerebellar hemispheres, communication of 4th ventricle with posterior cyst, enlargement of posterior fossa

Congenital Muscular Dystrophy

- Cerebellar hypoplasia, z-shaped brainstem
- Extensive cortical abnormalities, eye anomalies

Isolated Cerebellar Hypoplasia

- TORCH infections, trisomy 21, many other causes

Pontocerebellar Hypoplasia

- Numerous syndromic forms

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - RS: Widely varied depending on degree of associated anomalies; isolated cases may have subclinical symptomatology
  - LDD: Longstanding history of poorly localized neurological symptoms
  - JSRD: Clinically heterogeneous, including neonatal hyperpnea intermixed with central apnea, abnormal eye movements

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- RS: Can be partial or complete; isolated partial can be difficult to diagnose → midline sagittal image necessary to identify all parts of vermis
- LDD: Classic striated pattern; should be followed to exclude neoplasm
- JSRD: Molar tooth sign pathognomonic, associated anomalies can help identify specific syndrome

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Cephalocele

KEY FACTS

TERMINOLOGY

- Extracranial extension of intracranial contents through defects of skull & dura
  - Contents
    - Meningocele: Meninges & CSF
    - Encephalocele: Brain tissue, meninges, CSF
  - Locations
    - Occipital (most common)
    - Sincipital/frontoethmoidal
    - Parietal (usually atretic)
    - Basal/nasopharyngeal
    - Remaining cranial vault, including calvarium, mastoid & petrous temporal bones, cavum trigeminale

IMAGING

- Meninges & CSF ± brain tissue protruding through defect
  - Herniated brain tissue may be dysplastic
- MR + MRV to assess cephalocele contents & abnormal venous sinuses; CT to assess bone defect

TOP DIFFERENTIAL DIAGNOSES

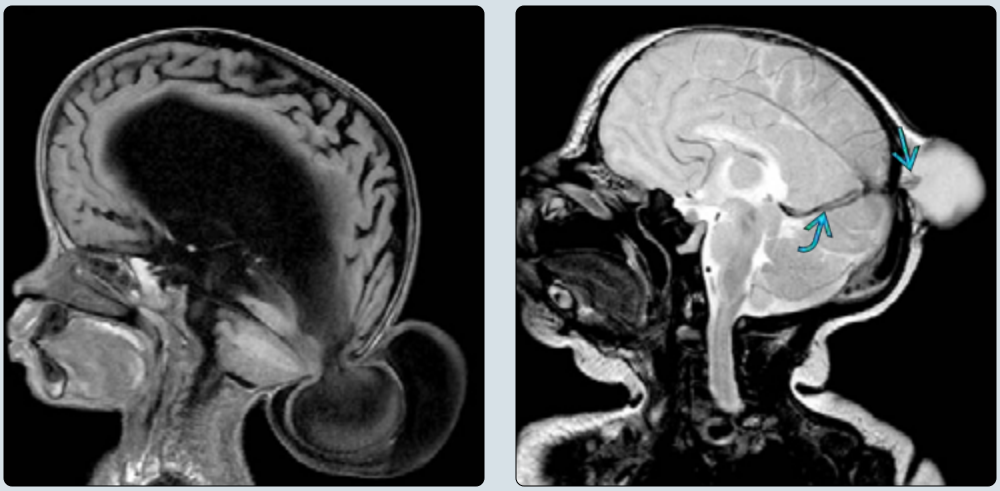
- Dermoid/epidermoid cyst
- Nasal glial heterotopia (nasal glioma)
- Sinus pericranii
- Assorted vascular anomalies (neoplasms & malformations)
- Cutis aplasia congenita
- Giant parietal foramina

CLINICAL ISSUES

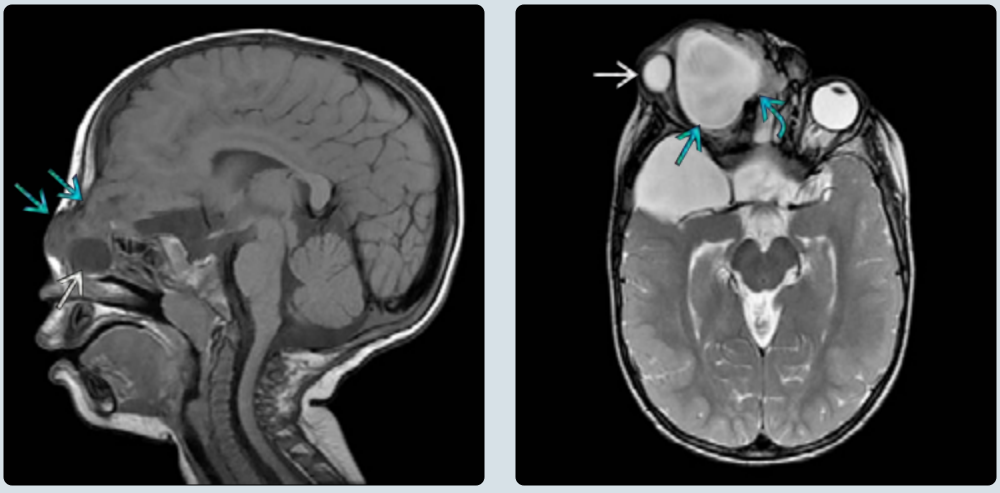
- Congenital more common than acquired
- Most cephaloceles (occipital) clinically obvious at birth
- Basal/nasopharyngeal cephaloceles may not manifest until end of 1st decade
- Prognosis & surgical options depend on cephalocele size, location, & contents + associated anomalies

DIAGNOSTIC CHECKLIST

- Normal cartilaginous nasofrontal region in infants can be problematic on CT; correlate with MR



(Left) *Sagittal T1 MR in a 20-month-old boy shows an occipital cephalocele containing meninges, CSF, brain, & lateral ventricle (meningoencephalocystocele). Note the increased craniofacial ratio (macrocephaly).* **(Right)** *Sagittal T2 MR of a 5-day-old boy with an atretic parietal cephalocele containing CSF & fibrous tissue. Note the persistent falcine sinus.*



(Left) *Sagittal T1 MR of a 2-year-old boy shows a nasoorbital encephalocele displacing & distorting the right globe. Note the brain tissue protruding through defects of the right frontal bone, nasal bone, & cribriform plate.* **(Right)** *Axial T2 MR in the same patient demonstrates a defect in the right anterior medial orbital wall with a large encephalocele filling the right orbit & anterior ethmoid sinuses. Note the mass effect on the right globe.*

TERMINOLOGY

Definitions

- Extracranial extension of intracranial contents through defects of skull & dura; categorized by contents & location
  - Congenital more common than acquired
- Contents
  - Meningocele: Meninges & CSF only
  - Encephalocele: Brain, meninges, CSF
  - Atretic cephalocele: Dermal, meningeal, & glial elements with fibrous tract extending intracranially
- Locations
  - Occipital: Most common location, up to 80% of cases
    - Supra- &/or infratentorial structures in cephalocele, including tentorium cerebelli & dural venous sinuses
    - Occipito-cervical (Chiari III): Very rare
  - Sincipital/Frontoethmoidal: 15% (↑ in southeast Asia)
  - Parietal: 10% (↑ in Japan at 38%)
    - Atretic parietal cephaloceles most common
      - Majority have benign clinical course
  - Basal/Nasopharyngeal: Up to 10% of cases
  - Remaining calvarium: Rare
  - Mastoid &/or petrous temporal bone: Rare
  - Cavum trigeminale: Rare

IMAGING

General Features

- Best diagnostic clue
  - Meninges & CSF ± brain tissue protruding through skull defect

CT Findings

- Excellent delineation of osseous defect

MR Findings

- T1WI, T2WI, FLAIR
  - Heterogeneous signal intensity of cephalocele contents reflecting brain parenchyma & CSF
    - Neural tissue may be dysplastic/gliotic
- MRV
  - Essential to characterize venous relationships to cephaloceles, particularly occipital
  - Venous anomalies in most atretic parietal cephaloceles
    - Persistent falcine sinus (± absent vein of Galen, straight sinus)
    - Superior sagittal sinus fenestration

Ultrasonographic Findings

- Cephaloceles (mainly occipital) can be detected in utero
- Postnatally, small superficial bumps over calvarium may receive ultrasound request first (particularly for small nasofrontal or atretic parietal cephaloceles)
  - May visualize calvarial defect, altered venous anatomy
  - MR should be strongly considered for palpable abnormalities over midline neuraxis due to frequency of underlying CNS anomalies

DIFFERENTIAL DIAGNOSIS

Dermoid/Epidermoid Cyst

- ± T1 hyperintense fat, diffusion restriction
- Commonly seen near suture lines (scalp, orbit)
- ± nasal pit in association with nasal dermal sinus

Nasal Glial Heterotopia

- Dysplastic neural tissue (not true nasal glioma)
- No communication with subarachnoid space

Sinus Pericranii

- Anomalous connection of intracranial & extracranial veins
- Compressible; engorge with Valsalva/crying

Vascular Anomaly

- Variety of solid & cystic lesions found in neonatal period
- Very rarely have communication through calvarium

Cutis Aplasia Congenita

- Skin defect ± calvarial defect, typically off midline

Giant Parietal Foramina

- Symmetric round/ovoid calvarial defects of paramidline parietal bones
- Can be single midline defect early

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Majority (occipital) clinically obvious
  - Smaller lesions: Soft, bluish (skin covered) or moist red (nonskin covered) discoloration over soft tissue mass
- Other signs/symptoms
  - Occipital: Microcephaly, hydrocephalus
  - Basal/Nasopharyngeal: Occult mass in oro-/nasopharynx → change in size with Valsalva, upper airway obstruction, nasal stuffiness/mouth breathing
  - Frontoethmoidal: Hypertelorism, broad nasal bridge, CSF rhinorrhea → meningitis

Demographics

- Ethnicity
  - Occipital most common in European & North American Caucasians
  - Frontoethmoidal most common in Southeast Asia & Latin America

Treatment

- Depends on cephalocele location, contents, & size + associated anomalies
- Goals of surgery: Prevent CSF leak & meningitis; improve functional & cosmetic deformities

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KEY FACTS

TERMINOLOGY

- Spectrum of congenital forebrain malformations characterized by failure of differentiation & midline cleavage of prosencephalon
- Traditionally, classic holoprosencephaly (HPE) divided (from most to least severe) into alobar, semilobar, & lobar subtypes
- Middle interhemispheric variant (MIH) or syntelencephaly generally accepted as subtype of HPE

IMAGING

- Abnormal communications of gray & white matter across midline
- Categories represent continuum of forebrain malformations without clear distinction between subtypes
- Alobar HPE: Complete absence of cleavage with "pancake" of anterior cerebral tissue, crescent-shaped monoventricle communicating with large dorsal cyst, "fused" thalami
- Semilobar HPE: Interhemispheric fissure & falx cerebri formed posteriorly with absence of frontal lobe separation & varying degrees of failed diencephalon cleavage
- Lobar HPE: 3rd ventricle fully formed (diencephalon separation), interhemispheric fissure & falx mostly formed with absent cleavage in inferior frontal lobes
- MIH: Lack of separation in posterior frontal & parietal regions with cleavage of anterior frontal & occipital lobes
- Absent cavum septum pellucidum in all forms

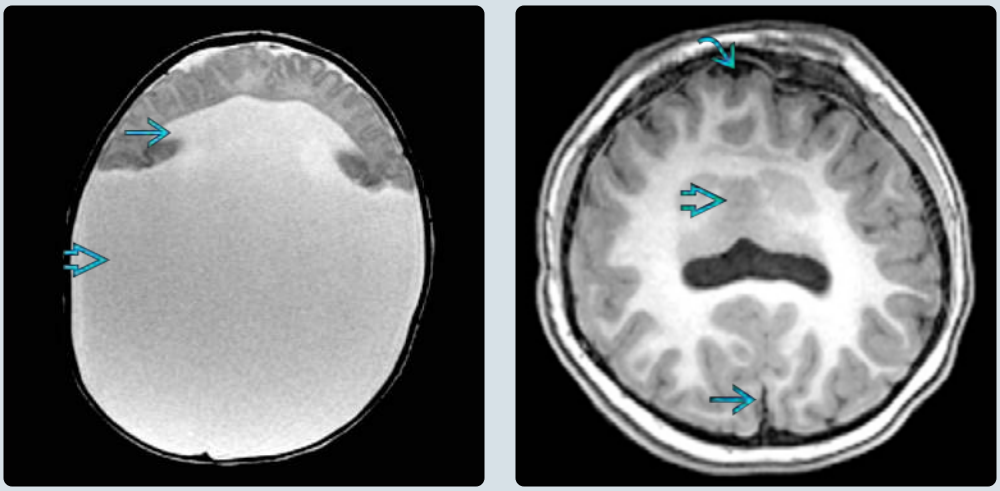
TOP DIFFERENTIAL DIAGNOSES

- Aqueductal stenosis
- Hydranencephaly
- Callosal agenesis with interhemispheric cyst
- Open-lip schizencephaly
- Septooptic dysplasia

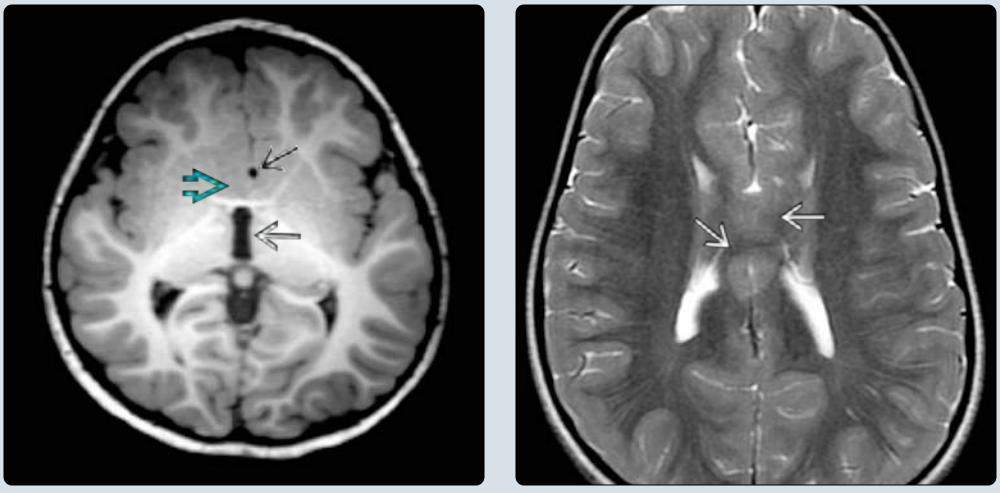
CLINICAL ISSUES

- Presentation varies widely depending on severity of malformation

(Left) *Axial T2 MR in a newborn with alobar holoprosencephaly (HPE) shows an anterior "pancake" of cerebral tissue continuous across the midline. There is a crescent-shaped monoventricle ⇨ communicating with a large dorsal cyst ⇨.* **(Right)** *Axial T1 MR of a 6-year-old child with semilobar HPE shows nonseparation of the frontal lobes ⇨ & caudate heads ⇨. However, there is separation of the parietal & occipital lobes with the falx cerebri ⇨ identified posteriorly.*



(Left) *Axial T1 MR in a 2-year-old child with lobar HPE shows failed separation of the anterior inferior frontal lobes with a small amount of gray matter ⇨ crossing the midline. Note that there is a characteristic azygous anterior cerebral artery ⇨. The 3rd ventricle ➡ is not hypoplastic.* **(Right)** *Axial T2 MR in a child with the HPE middle interhemispheric variant shows an abnormal communication of gray & white matter focally across the midline ➡ at the expected level of the midbody of the corpus callosum.*



TERMINOLOGY

Abbreviations

- Holoprosencephaly (HPE)
- Middle interhemispheric variant (MIH)

Definitions

- Spectrum of congenital forebrain malformations characterized by failure of differentiation & midline cleavage of prosencephalon
- Traditionally, classic HPE divided into alobar, semilobar, & lobar subtypes (due to failure of ventral induction)
- MIH or syntelencephaly generally accepted as subtype of HPE (due to failure of dorsal induction)
- Represents continuum of forebrain malformations without clear distinction between different subtypes

IMAGING

General Features

- Best diagnostic clue
  - Alobar HPE: Complete absence of cleavage with "pancake" of anterior cerebral tissue, crescent-shaped monoventricle communicating with large dorsal cyst, "fused" thalami
  - Semilobar HPE: Absent frontal lobe cleavage with parietooccipital lobe separation
  - Lobar HPE: Interhemispheric fissure & falx mostly formed with partial nonseparation of frontal lobes
  - MIH: Hemispheres not separated in posterior frontal & parietal regions

MR Findings

- T1WI, T2WI, FLAIR
  - Alobar: Monoventricle ± communication with dorsal cyst
    - Cerebral tissue flattened anteriorly like "pancake"
    - Absent corpus callosum, falx, interhemispheric fissure
  - Semilobar: Frontal lobes largely continuous across midline
    - Varying degrees of diencephalon separation (absent/small 3rd ventricle)
    - Posterior corpus callosum (splenium) present
  - Lobar: Interhemispheric fissure present along nearly entire midline
    - Rudimentary frontal horns typical, inferior portions of frontal lobes uncleaved
    - 3rd ventricle fully formed
    - Absence of genu & rostrum of corpus callosum
  - MIH: Lack of midline separation in posterior frontal & parietal regions with normal cleavage of polar areas of cerebrum
    - Genu & splenium present with absent body of corpus callosum
    - Normal basal ganglia
  - Absent septum pellucidum in all forms of HPE
  - No precise distinction between lobar & semilobar HPE
- Variable findings
  - Brainstem & cerebellar anomalies, such as rhombencephalosynapsis
  - Gray matter heterotopia: MIH > classic HPE
  - Azygous anterior cerebral artery

DIFFERENTIAL DIAGNOSIS

Aqueductal Stenosis

- Macrocephalic with moderate to severe enlargement of ventricles; small rind of cerebral tissue in maximal cases
- Preservation of falx

Hydranencephaly

- Absence of anterior circulation cerebrum with fluid-filled cranial vault
- Preservation of falx

Callosal Agenesis With Interhemispheric Cyst

- Cyst displaces cerebral tissue laterally, not anteriorly

Open-Lip Schizencephaly

- Separation of anterior & posterior cerebral tissue by CSF-filled cleft lined by polymicrogyric gray matter
  - Unilateral or bilateral
- Falx preserved

Septooptic Dysplasia

- Lack of septum pellucidum ± optic nerve hypoplasia, pituitary abnormalities, schizencephaly

PATHOLOGY

General Features

- Etiology
  - Teratogen exposure
    - EtOH, diabetes, cigarette smoking, retinoic acid
  - Many genetic/syndromic etiologies
    - Smith-Lemli-Opitz, Pallister-Hall
- Genetics
  - Standard karyotyping abnormal in 24-45% of HPE
    - Classically trisomy 13 (Patau syndrome)
  - At least 10% have microdeletions/duplications
    - > 12 known HPE-associated genes
- Associated abnormalities
  - Facial anomalies → hypotelorism/cyclopia, ethmocephaly (proboscis), cebocephaly (single naris), arrhinia, central incisor, midline or bilateral cleft lip/palate

Staging, Grading, & Classification

- Alobar, semilobar, lobar classification of DeMyer
- MIH described as HPE variant by Barkovich in 1993
- Other abnormalities of midline development include agenesis/dysgenesis of corpus callosum, septooptic dysplasia, & isolated absence of cavum septum pellucidum

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- HPE represents continuum of forebrain malformations
  - No clear distinction amongst different categories → provide clear anatomic descriptions in reports
  - Lack of separation described at many different locations in telencephalon & considered as midline anomalies vs. forme fruste of HPE

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Callosal Anomalies

KEY FACTS

TERMINOLOGY

- Agenesis (absence) of corpus callosum (ACC)
- Overlapping terms for other CC anomalies
  - Hypogenesis, dysgenesis may encompass all variations

IMAGING

- Axial: Distortion of lateral ventricles best clue to diagnosis
  - Parallel lateral ventricles with colpocephaly
    - Dilation of trigones + occipital & temporal horns
  - Probst bundles: Densely packed heterotopic white matter (WM) tracts parallel to interhemispheric fissure
- Sagittal: Best delineates callosal abnormalities
  - Cerebral sulci radiate to high-riding 3rd ventricle
  - Absent cingulate sulcus, hypoplastic underrotated cingulate gyrus
- Coronal images
  - No thick band of WM bridging cerebral hemispheres over lateral ventricles
  - Upturned anterior horns of lateral ventricles
    - Viking helmet, Texas Longhorn, trident-shaped
- ± interhemispheric cyst or lipoma
  - Cyst: Communicating (type I) or noncommunicating (type II) with ventricle
- Anterior cerebral arteries & branches do not conform to expected normal CC shape

TOP DIFFERENTIAL DIAGNOSES

- Destruction, attenuation, or immaturity of corpus callosum
- Failure of forebrain cleavage

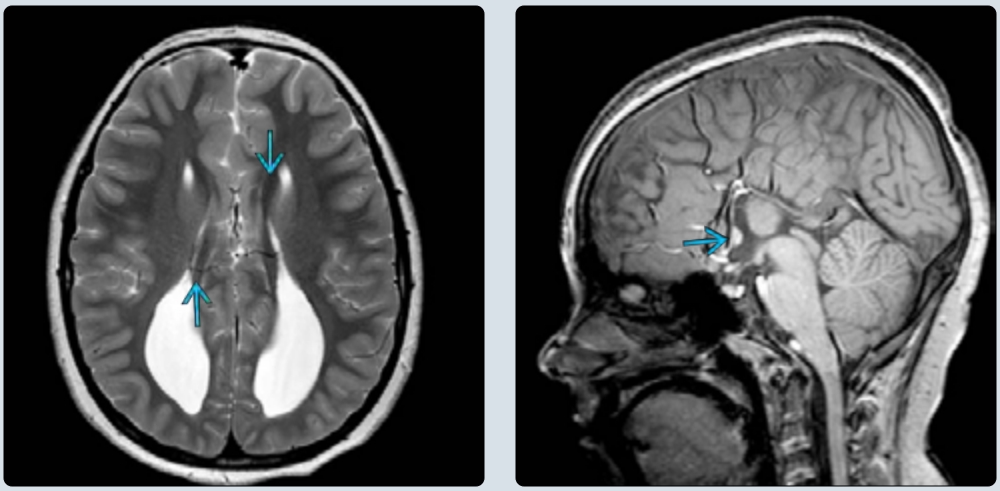
PATHOLOGY

- Many potential etiologies; associated with > 200 syndromes
  - Other CNS malformations often present

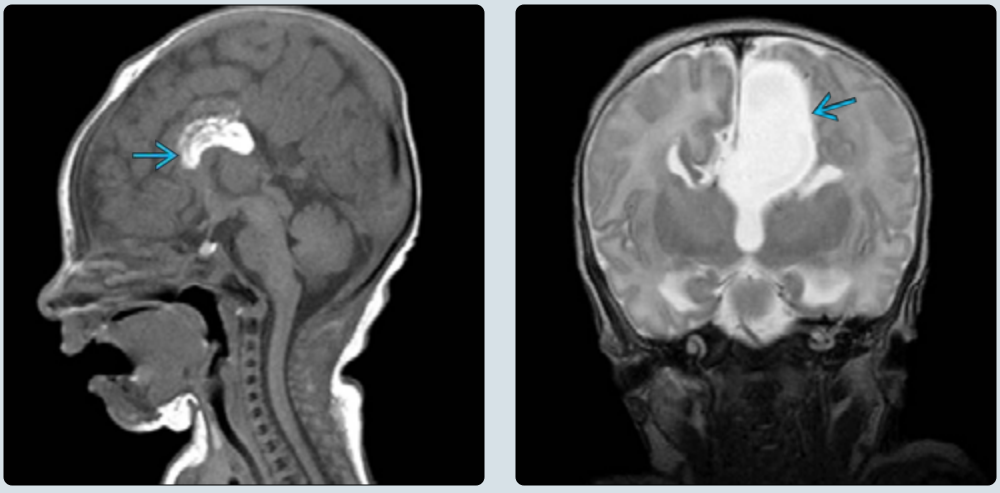
CLINICAL ISSUES

- Typical manifestations include seizures, developmental delay, anomalies of hypothalamic-pituitary axis
- Severity varies with type of CC anomaly, associated CNS malformations, genetic syndromes

(Left) *Axial T2WI MR in an 11 year old with agenesis of the corpus callosum (ACC) demonstrates a classic parallel configuration of the lateral ventricles with posterior dilation (colpocephaly). T2-hypointense Probst bundles ⇨ are seen along the medial margins of the lateral ventricles.* **(Right)** *Sagittal T1WI MR of the same patient demonstrates sulci radiating to the superior margin of the 3rd ventricle, which is elevated. There is absence of the cingulate sulcus. The anterior commissure ⇨ is thickened.*



(Left) *Sagittal T1WI MR of a newborn with ACC demonstrates an associated T1-hyperintense interhemispheric/pericallosal lipoma ⇨.* **(Right)** *Coronal T2WI MR of a different neonate with ACC demonstrates an associated interhemispheric cyst ⇨ that communicates with the ventricular system (type I). Also noted is the classic trident shape of the upturned lateral ventricle frontal horns.*



TERMINOLOGY

Abbreviations

- Agenesis of corpus callosum (ACC)
- Hypogenesis of corpus callosum (HCC)

Definitions

- Heterogeneous condition of congenital hypoplasia/dysplasia or absence of corpus callosum (CC)
- Various & overlapping terms used for CC anomalies outside of complete agenesis/absence
  - Hypoplasia implies thin CC with normal shape
  - Partial agenesis, dysplasia imply truncation or abnormal morphology otherwise
  - Hypogenesis, dysgenesis may encompass all variations

IMAGING

General Features

- Best diagnostic clue
  - Axial: Parallel lateral ventricles with colpocephaly
  - Sagittal: Sulci radiating to high-riding 3rd ventricle
  - Coronal: No thick band of white matter (WM) bridging cerebral hemispheres over lateral ventricles
- Associated interhemispheric lipoma or cyst
  - Interhemispheric cyst of ACC may be communicating (type I) or noncommunicating (type II) with ventricle
- Anterior cerebral arteries & branches do not conform to expected normal CC shape

MR Findings

- T1, T2, FLAIR
  - Sagittal midline
    - Elevated (high-riding) 3rd ventricle
    - Cerebral sulci radiate to 3rd ventricular roof
      - Absent cingulate sulcus
      - Hypoplastic, underrotated cingulate gyrus
    - ± interhemispheric lipoma or cyst
  - Coronal
    - Absence of bridging WM tracts over lateral ventricles
    - Absent or abnormal septum pellucidum
    - Upturned anterior horns of lateral ventricles, shaped like Viking helmet, Texas Longhorn, or trident
    - Underrotated, globular &/or hypoplastic hippocampi
  - Axial
    - Parallel orientation of lateral ventricles with colpocephaly (posterior dilation of trigones plus occipital & temporal horns)
    - Probst bundles: Densely packed heterotopic WM tracts running parallel to interhemispheric fissure
    - Decreased WM volume

Imaging Recommendations

- Protocol advice
  - Volume acquisition sequences with multiplanar reconstruction
  - Midline sagittal image best delineates CC abnormalities

DIFFERENTIAL DIAGNOSIS

Destruction of Corpus Callosum

- Surgery (callosotomy), trauma
- WM injury of prematurity
  - a.k.a. periventricular leukomalacia (PVL)

Attenuation of Corpus Callosum

- Hydrocephalus stretches/deforms CC, flattens fiber tracts

Immaturity of Corpus Callosum

- CC relatively thin & unmyelinated in neonates, even thinner in premature neonates

Failure of Forebrain Cleavage

- Varying degrees of CC absence depending on type of holoprosencephaly

PATHOLOGY

General Features

- Associated abnormalities
  - Additional CNS malformations typically present
    - Callosal anomalies found in 50% of patients with other brain malformations
  - Associated with > 200 named syndromes
- Embryology
  - Cells destined for CC begin forming at 6 weeks gestation
  - Axons begin crossing midline anteriorly at 13-14 weeks
  - Bidirectional growth with ultimate CC shape by 20 weeks
    - Previous literature describing "front to back" development has been largely replaced

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Seizures, developmental delay
  - Anomalies of hypothalamic-pituitary axis
- Other signs/symptoms
  - Widely variable depending on type of CC anomaly, associated CNS malformations, genetic syndromes
  - Isolated ACC rarely reported as incidental
    - Up to 75% with near-normal to normal intelligence
      - ± subtle learning, social, & neurological issues

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Hemimegalencephaly

KEY FACTS

TERMINOLOGY

- Hemimegalencephaly (HME) = hamartomatous overgrowth of part vs. entire hemisphere
- Due to defects in neuronal proliferation, migration, & organization

IMAGING

- Large cerebral hemisphere, hemicranium
  - Posterior falx, occipital pole extend to contralateral side
  - Enlarged ipsilateral ventricle with abnormally shaped frontal horn (often pointed)
    - No other process causes simultaneous enlargement of parenchyma & ipsilateral ventricle
- Gray matter (GM) abnormalities include pachygyria, polymicrogyria, & heterotopias
- White matter (WM) abnormalities
  - Increased volume, abnormal T2 signal intensity
    - Often dark prior to typical myelination timeframe
  - Poor GM-WM differentiation
- Size & signal intensity of HME can change over time
  - May atrophy with chronic seizure activity

PATHOLOGY

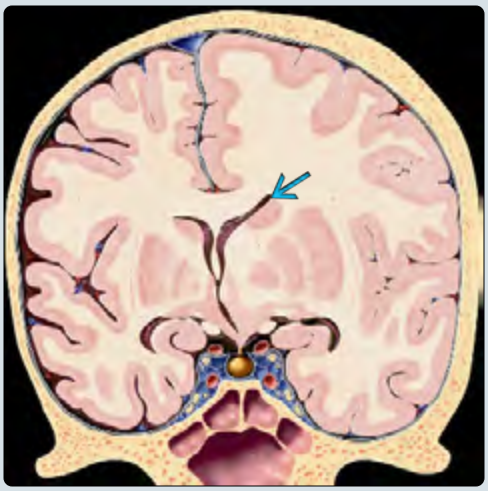
- Cortical GM & WM gliosis, dysmorphic neurons
- ± hypercellularity, balloon cells, immature cells, dystrophic Ca^{2+}

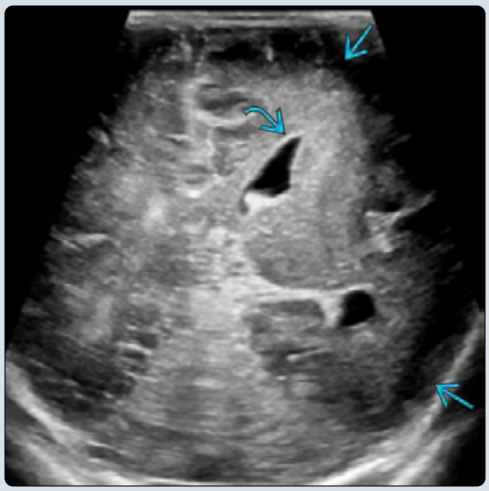
CLINICAL ISSUES

- Most common presentations: Seizures, hemiparesis, developmental delay
  - 50% with associated skin lesions, truncal/extremity overgrowth, &/or vascular malformations
- Anatomic or functional hemispherectomy for seizure control (as anticonvulsants usually ineffective)

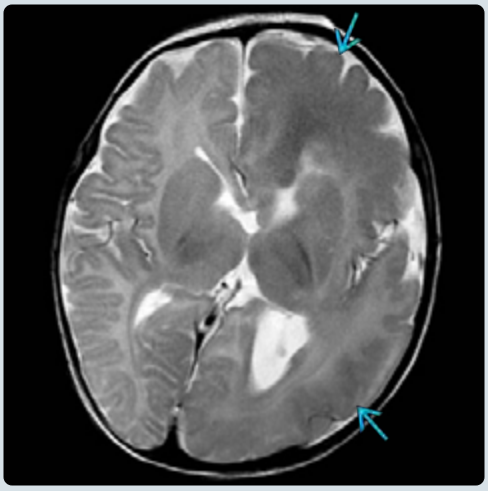
DIAGNOSTIC CHECKLIST

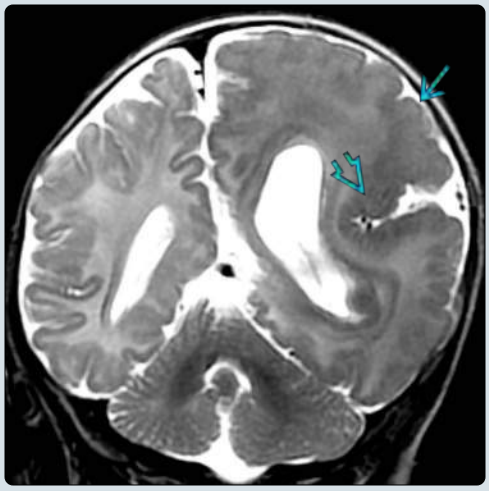
- Must exclude contralateral abnormalities for successful hemispherectomy





(Left) *Coronal graphic shows overgrowth of the left cerebral hemisphere. Note the shift of midline structures, excess of gray-white matter, flattened gyri, & abnormal lateral ventricle frontal horn.* **(Right)** *Coronal head ultrasound in a newborn with a large extremity vascular malformation shows overgrowth, abnormal sulcation, & increased echogenicity of the left cerebral hemisphere. The ipsilateral ventricle shows enlargement with an abnormal configuration.*





(Left) *Axial T2WI MR in the same patient at 3 months of age shows the enlargement of the left cerebrum with pachygyria & abnormal gray-white matter differentiation diffusely.* **(Right)** *Coronal T2WI MR in the same patient shows abnormal sulcation with pachygyria & polymicrogyria. Abnormal white matter signal intensity in the left cerebrum is due to neuronal dysplasia, accelerated myelination, &/or mineralization. The ipsilateral ventricle is enlarged, typical of hemimegalencephaly.*

TERMINOLOGY

Definitions

- Megalencephaly: Brain size & weight > 2 standard deviations above age-related normals
- Hemimegalencephaly (HME): Hamartomatous overgrowth of part vs. entire hemisphere
  - Classic HME (or unilateral megalencephaly): Holohemispheric involvement
  - Partial HME (or focal megalencephaly): Portion of hemisphere involved
  - Total HME: Involvement of unilateral cerebral hemisphere, cerebellum, & brainstem (rare)

IMAGING

General Features

- Best diagnostic clue
  - Enlarged dysplastic cerebral hemisphere (or portion of hemisphere) with enlarged ipsilateral lateral ventricle
    - Abnormal gray matter (GM) & white matter (WM)

MR Findings

- T1WI, T2WI, FLAIR
  - Abnormal GM: Pachygyria, polymicrogyria, heterotopia
  - Abnormal WM: ↑ volume, ↓ T2 signal intensity prior to expected myelination timeframe, variable T2 signal intensity after myelination
    - Abnormal anterior to posterior WM tracts
      - Aberrant midsagittal fibers: Deep to callosal body, between lateral ventricular anterior horns
      - Enlarged periventricular fibers: Along caudate nucleus near lateral ventricle
  - Blurred GM-WM junctions
  - Enlarged ipsilateral ventricle, pointed frontal horn
  - ± cerebellar, brainstem hemiovergrowth
  - Size & signal intensity of HME can change over time
    - May atrophy, change in T2 signal intensity
- T2* GRE
  - Signal loss from dystrophic Ca^{2+}
- In utero, 2nd trimester
  - Disruption of "transient" layers on DWI > SSFSE T2
  - Mass-like low T2 signal proliferation of germinal matrix/ventricular zone, mimics hemorrhage

DIFFERENTIAL DIAGNOSIS

Focal Cortical Dysplasia

- Small or large areas with blurring of GM-WM junction
- Can be identical in imaging & pathology to partial HME

Rasmussen Encephalitis

- Unilateral encephalitis with progressive atrophy

Tuberous Sclerosis Complex

- Heavy burden of cortical/subcortical tubers can mimic HME, but tuberous sclerosis complex bilateral

Gliomatosis Cerebri

- Diffusely infiltrating glioma effaces ipsilateral ventricle
- Rare in children, diagnosed much later than HME

Unilateral Cerebral Edema

- May be seen with trauma, infarction, or infection
- Morphology of underlying brain otherwise normal
- Effaces ipsilateral ventricle

PATHOLOGY

General Features

- Genetics
  - Abnormal cellular proliferation, migration, & differentiation
  - Due to dysregulation in PI3K/AKT/mTOR & Ras/MAPK molecular pathways
  - Associated anomalies include
    - Various overgrowth syndromes with extensive, predominantly slow flow vascular malformations
      - Klippel-Trenaunay
      - Proteus
    - Epidermal nevus syndrome
    - Phakomatoses: Neurofibromatosis type 1, tuberous sclerosis complex, hypomelanosis of Ito

Microscopic Features

- GM & WM gliosis, dysmorphic neurons
- ± hypercellularity, balloon cells, dystrophic Ca^{2+}

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Seizures, hemiparesis, developmental delay

Demographics

- Usually diagnosed during 1st year of life
- Of cortical dysplasias diagnosed by imaging: HME 3%

Treatment

- Anticonvulsants usually ineffective
- Anatomic or functional hemispherectomy
  - Earlier surgical intervention yields better outcomes
  - Must 1st confirm normal contralateral hemisphere

DIAGNOSTIC CHECKLIST

Consider

- May initially come to attention in utero as unilateral ventriculomegaly

Reporting Tips

- Define extent of HME + contralateral abnormalities

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Lissencephaly

KEY FACTS

TERMINOLOGY

- Extensive bilateral cortical malformations caused by arrested neuronal migration; results in thick cortex & smooth brain surface
- Many clinical & imaging phenotypes due to numerous underlying genetic mutations
  - Classical: Complete/incomplete agyria &/or pachygyria
    - Mildest form: Isolated band heterotopia (BH)
  - Syndromic: Various forms & associated anomalies

IMAGING

- Smooth cortical surface
  - Spectrum of ↓ number & depth of sulci + broad gyri
  - Hourglass or figure 8 cerebral hemispheres in agyria
- Thick abnormal 4-layer cortex (deeper 3 often seen on MR)
  - Superficial molecular layer
  - Thin outer cortical layer
  - Intervening cell-sparse white matter (WM) layer
  - Thick inner cellular layer, overlapping appearance of BH
- Deep WM: Volume loss, ↓ arborization
- Ventriculomegaly

TOP DIFFERENTIAL DIAGNOSES

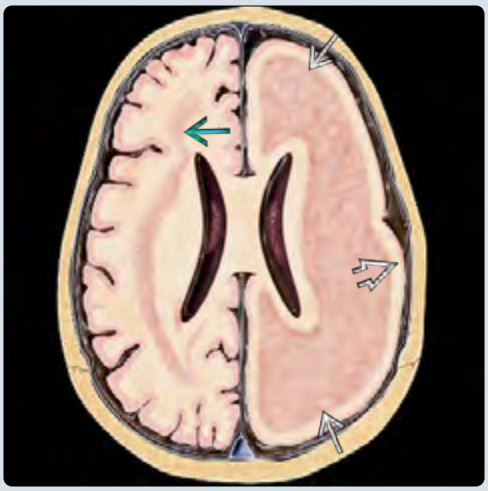
- Microcephaly with simplified gyral pattern
- Congenital muscular dystrophies
- Normal immature sulcation pattern of prematurity

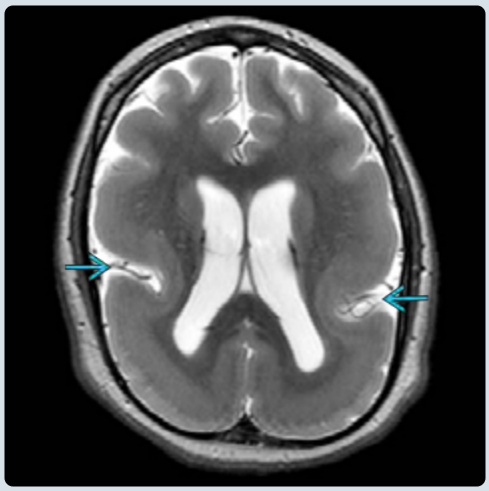
PATHOLOGY

- Many genetic mutations → spectrum of phenotypes
  - *LIS1* gene at 17p13.3: Autosomal dominant
    - Up to 60% of lissencephaly cases
    - Posterior hemispheres more affected than anterior
  - *DCX* (a.k.a. *XLIS*) gene at Xq23: X-linked
    - Up to 12% of lissencephaly cases
    - Anterior hemispheres more affected than posterior

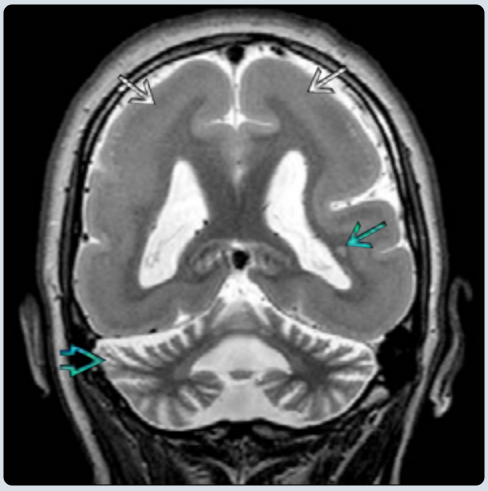
CLINICAL ISSUES

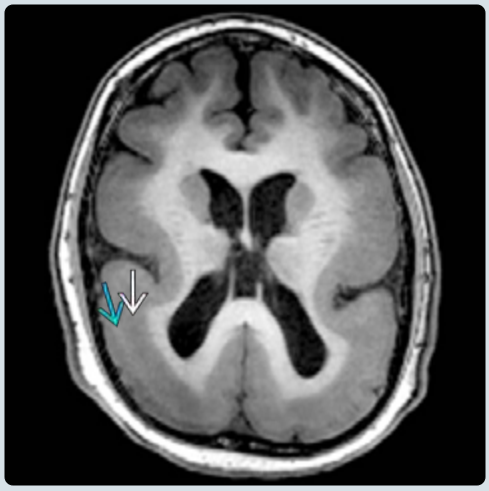
- Clinical profile depends on severity of malformation & associated anomalies





(Left) *Axial graphic depicts band heterotopia in the right cerebral hemisphere. The left hemispheric lissencephaly has a thick deep cellular layer of gray matter with a thin, smooth outer cortex.* **(Right)** *Axial T2WI MR in a 15-year-old girl with lissencephaly demonstrates absence of normal posterior gyri & sulci (posterior agyria) with less pronounced involvement of the frontal lobes (frontal pachygyria). The sylvian fissures are vertically oriented, giving an hourglass or figure 8 configuration to the brain.*





(Left) *Coronal T2WI MR in the same patient shows the extensive, thick cellular band of deep GM in addition to a tiny nodular focus of GM heterotopia. There is significant cerebellar atrophy, which has progressed from the prior studies & is likely related to antiepileptic medications.* **(Right)** *Axial T1WI MR in the same patient best delineates the abnormal cortical thickening with faint visualization of the cell sparse zone overlying the thicker inner cellular GM band.*

TERMINOLOGY

Definitions

- Extensive cortical malformation caused by slow or arrested neuronal migration; results in abnormally thick 4-layer cortex with smooth cerebral surface
- Many clinical & imaging phenotypes due to numerous underlying genetic mutations
  - Classical/type I lissencephaly
    - Spectrum from complete agyria to pachygyria
      - Regions of both types often present; may have regions of normal cerebrum
    - Overlap with gray matter (GM) band heterotopia (BH)
      - Isolated BH: Mildest form of classical lissencephaly
  - Lissencephaly syndromes with various other CNS &/or non-CNS anomalies

IMAGING

General Features

- Best diagnostic clue
  - Thick cortex with absence or ↓ number of sulci diffusely
  - Hourglass or figure 8 shape of cerebral hemispheres on axial images
    - Due to shallow, vertically oriented sylvian fissures
- Location
  - Gradients of anterior to posterior cerebral hemispheric involvement based on specific genetic mutations

MR Findings

- T1WI, T2WI, FLAIR
  - Smooth cortical surface
    - Spectrum of ↓ number & depth of sulci with large, broad gyri
      - Primitive rather than completely aberrant sulci
  - Thick abnormal cortex; 3 or 4 layers often seen on MR
    - Outer thin cortical GM layer
    - Thin white matter (WM) cell sparse zone
    - Inner thick cortical layer (appearance overlaps BH)
      - Thickness of band predicts smoothness of cortical surface
  - Deep WM abnormalities
    - ↓ volume with ↓ arborization
    - ± periventricular Ca^{2+} if due to congenital CMV
  - Enlarged lateral ventricles
  - Common associated findings
    - Callosal dysgenesis/agenesis
    - Pontocerebellar hypoplasia
    - Hippocampal anomalies

DIFFERENTIAL DIAGNOSIS

Microcephaly With Simplified Gyral Pattern

- Head circumference > 3 standard deviations below normal
- Broad gyri, ↓ number of sulci without cortical thickening
- Due to ↓ cell production or ↑ apoptosis

Congenital Muscular Dystrophies

- Cobblestone brain (formerly type 2 lissencephaly)
- Z-shaped hypoplastic brainstem on sagittal images
- Fukuyama, Walker-Warburg, muscle-eye-brain diseases

Immature Brain

- Premature neonate with sulcation pattern appropriate for gestational age

PATHOLOGY

General Features

- Genetics
  - Many identified mutations → spectrum of phenotypes
    - *LIS1* gene at 17p13.3: Autosomal dominant
      - Accounts for up to 60% of lissencephaly cases
      - Posterior dominant: More severe involvement of posterior hemispheres (agyria) vs. anterior hemispheres (pachygyria)
    - *DCX* (a.k.a. *XLIS*) gene at Xq23: X-linked
      - Accounts for up to 12% of lissencephaly cases
      - Classical lissencephaly in males (son), BH in females (mother)
      - Anterior dominant
    - *TUBA1A* gene at 12q12-q14.3: Autosomal dominant
      - Range of agyria to BH
      - Pontocerebellar hypoplasia, thin/absent CC with vertical splenium, hippocampal dysgenesis
    - *ARX* gene at Xp21.1: X-linked
      - Posterior dominant
      - Callosal agenesis, ambiguous genitalia
    - *RELN* gene at 7q22: Autosomal recessive
      - Anterior dominant

Microscopic Features

- 4-layer cortex in majority of cases (*LIS1* & *DCX*)
  - Superficial molecular layer
  - Thin outer cortical layer of neurons
  - Cell-sparse WM zone
  - Thick inner cellular layer of neurons

CLINICAL ISSUES

Presentation

- Clinical profile depends on severity of malformation & associated anomalies
  - Complete agyric lissencephaly: Neonatal hypotonia, gradual spasticity, severe epilepsy
  - Isolated BH: Mental retardation, seizures

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- If lissencephaly suspected in neonate, verify gestational age

Reporting Tips

- Describe regions & types of involvement + associated abnormalities to help in classification, clinical management

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Heterotopic Gray Matter

KEY FACTS

TERMINOLOGY

- Heterotopia (HTP): Abnormally located gray matter (GM) due to foreshortened or prolonged neuronal migration
  - Anywhere from periventricular germinal zone to pia

IMAGING

- Ectopic nodular or ribbon-like foci following GM signal intensity on every MR sequence
- Locations: Periventricular, subcortical, pial
  - Periventricular nodular HTP (most common)
  - Band HTP ~ laminar HTP, double cortex
  - Subcortical nodular HTP (focal or multinodular)
  - Pial: "Cobblestone" cortex
- Best MR sequences (with multiplanar reformats)
  - Myelinated brain: 3D T1 spoiled GRE (e.g., SPGR)
  - Unmyelinated brain: 3D T2 FSE (e.g., Cube/VISTA/SPACE)

TOP DIFFERENTIAL DIAGNOSES

- Tuberous sclerosis
- "Closed-lip" schizencephaly
- Ependymal spread of tumor
- Congenital cytomegalovirus

PATHOLOGY

- Genetic & acquired etiologies cause disturbed neuronal migration resulting in ectopic rests of GM
  - At germinal zone → periventricular HTP
  - Before reaching cortex → subcortical & band HTP
  - At pia → "cobblestone" brain

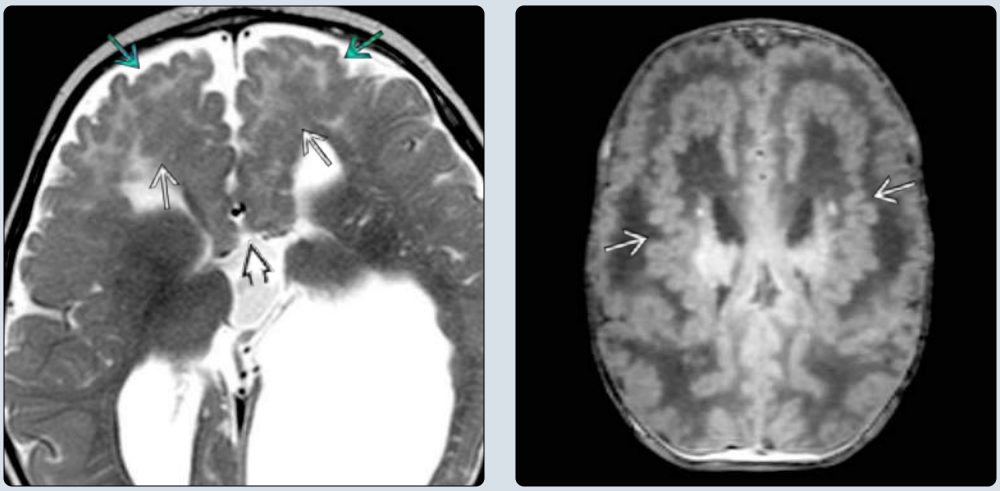
CLINICAL ISSUES

- Symptoms: Developmental delay, motor dysfunction, seizures (40% of intractable epilepsy cases)
- Palliative surgery reserved for intractable seizures

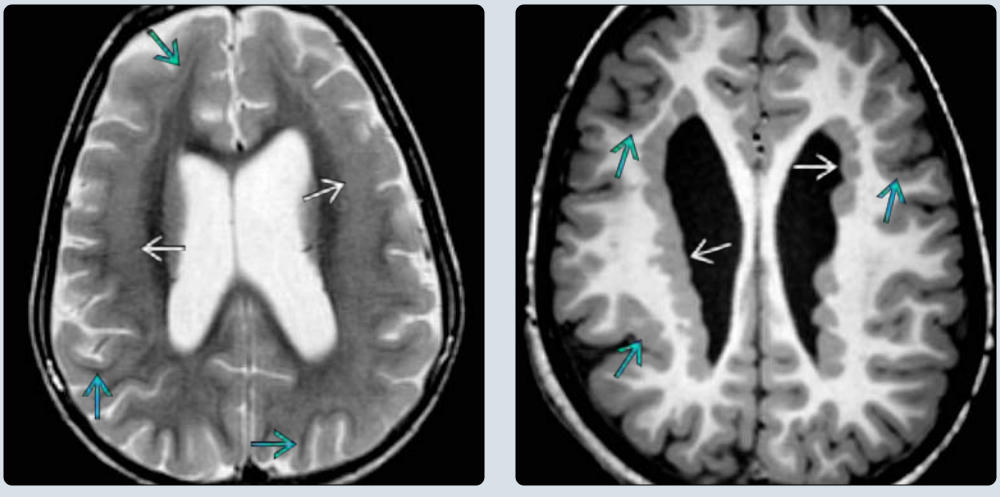
DIAGNOSTIC CHECKLIST

- Look closely for associated anomalies of adjacent cortex

(Left) *Axial T2 MR in an 18-month-old girl with Aicardi syndrome & seizures shows extensive bifrontal periventricular & subcortical gray matter heterotopia. Note the thin, irregular cortex overlying the heterotopia. The corpus callosum is absent.* **(Right)** *Axial T1 MR in a 2 month old with seizures shows extensive, symmetric heterotopic gray matter with a brain-in-brain appearance. The more superficial cortex is thinner than usual with poor sulcation.*



(Left) *Axial T2 MR in a 5-year-old boy with seizures demonstrates a band of gray matter of variable thickness within the deep & subcortical white matter, consistent with band heterotopia. Note the thin layer of intervening white matter between the band of heterotopic gray matter & cortex.* **(Right)** *Axial T1 MR in a 9-year-old boy with epilepsy shows the diffuse form of periventricular nodular heterotopia. Note the abnormal overlying cortex in multiple areas.*



TERMINOLOGY

Synonyms

- Gray matter (GM) heterotopia (HTP)
- Double cortex ~ band HTP

Definitions

- Abnormally located GM due to foreshortened or prolonged migration of neuronal groups
  - Occurs anywhere from periventricular germinal zone (GZ) to pia

IMAGING

General Features

- Best diagnostic clue
  - Nodules or ribbons of ectopic tissue that follow GM on all MR sequences
- Location
  - Periventricular, subcortical/transcerebral, pial

CT Findings

- **NECT:** Isodense with GM; rare dysplastic Ca^{2+}

MR Findings

- T1WI: Isointense to GM; well marginated
- T2WI: Isointense to GM
- FLAIR: Isointense to minimally hyperintense to GM
- DWI: Isointense to GM
- T1 C+: No enhancement
- Fetal MR: Nodular, irregular ventricular lining ± abnormal sulcation

Ultrasonographic Findings

- Nodular periventricular HTP may be evident as irregular/lobulated ventricular margin

Imaging Recommendations

- Protocol advice
  - Multiplanar reconstructions of
    - 3D T1 spoiled GRE MR (e.g., SPGR) in myelinated brain
    - 3D T2 FSE MR (e.g., Cube/VISTA/SPACE) in unmyelinated brain

DIFFERENTIAL DIAGNOSIS

Tuberous Sclerosis

- Subependymal nodules in regions of fetal germinal matrix
- Often calcify; may enhance
- Associated with cortical/subcortical tubers

"Closed-Lip" Schizencephaly

- Polymicrogyric GM lines apposed walls of pial-ependymal cleft with funnel-shaped outpouching of ventricular wall

Ependymal Spread of Tumor

- Nodular or crescentic foci along ventricular lining
- Usually enhance, may restrict diffusion

Congenital Cytomegalovirus

- Periventricular Ca^{2+} without protrusion into ventricle
- ± white matter injury, polymicrogyria, schizencephaly, cerebellar hypoplasia, microcephaly

PATHOLOGY

General Features

- Etiology
  - Genetic: Mutations alter molecular interactions at multiple migration points → migration arrest → HTP
  - Acquired: Fetal insult (ischemia, infection, etc.) → disturbed neuronal migration/cortical positioning
- Genetics
  - Diffuse periventricular nodular HTP often genetic
    - *FLNA* (Xq28), *ARFGEF2* (20q13.13), 5p15
  - Band HTP: Mild form of classic lissencephaly (agyria/pachygyria/double cortex)
    - *LIS1* (17p13.3), *DCX* (Xq22.3-q23), tubulin genes
- Associated abnormalities
  - Focal periventricular nodular HTP in 30% of Chiari II
- Embryology
  - Abnormal neuronal migration
    - At GZ → periventricular HTP
    - Before reaching cortex → subcortical & band HTP
    - At pia → "cobblestone" brain

Microscopic Features

- Multiple types of immature/dysplastic cells
  - Excitatory exceed inhibitory

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Developmental delay, seizures, motor dysfunction
  - Age of onset & severity of symptoms depend on location & extent of abnormalities

Demographics

- Epidemiology
  - Found in up to 40% of patients with intractable epilepsy

Natural History & Prognosis

- Prognosis depends on location & extent of HTP, associated malformations, & seizure severity
- Focal HTP can be incidental on imaging/autopsy

Treatment

- Palliative surgery reserved for intractable seizures

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- HTP commonly associated with other anomalies
  - Look closely for overlying cortical abnormalities
- GM HTP does not enhance

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3. Barkovich AJ et al: A developmental and genetic classification for malformations of cortical development: update 2012. Brain. 135(Pt 5):1348-69, 2012

Schizencephaly

KEY FACTS

IMAGING

- Transmantle cleft of brain parenchyma extending from cortical surface to ventricle
  - Cleft lined by dysplastic gray matter (GM)
    - Polymicrogyria, heterotopia
  - Closed-lip defect: Cleft walls closely apposed/fused
    - Dimple of lateral ventricle laterally at cleft
  - Open-lip defect: Cleft walls separated by CSF; widths & shapes of clefts variable
- Bilateral in up to 1/2 of patients
  - Unilateral often shows contralateral polymicrogyria
- ± hemosiderin staining of cleft, ventricles
- ± Ca^{2+} if due to in utero infection (e.g., CMV)
- Associated with absence of cavum septum pellucidum ± optic nerve hypoplasia (septooptic dysplasia)
  - Anomalies of hypothalamic-pituitary axis
  - Callosal hypogenesis, other midline anomalies
- MR protocol advice
  - Multiplanar sequences ± volumetric acquisition, particularly important for closed-lip type
  - Prior to myelination, GM-lined cleft best seen on T2WI

TOP DIFFERENTIAL DIAGNOSES

- Encephaloclastic porencephaly
- Hydranencephaly
- Alobar holoprosencephaly

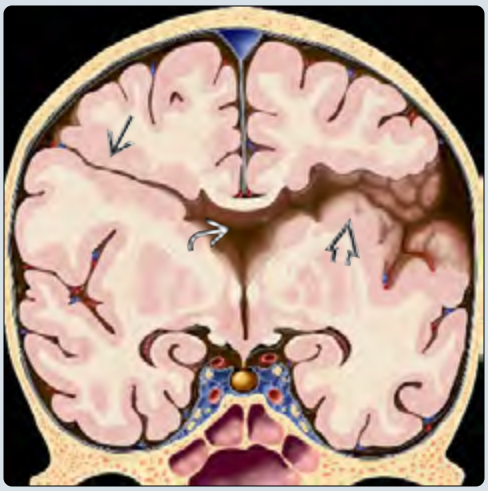
PATHOLOGY

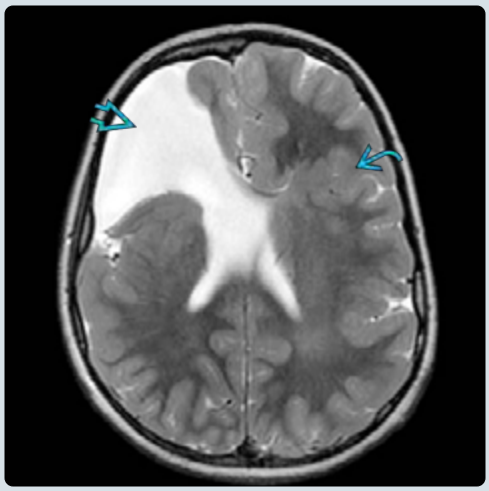
- In utero insult affecting neuronal migration
- Etiologies include: Vascular insult, infection (CMV, HSV), maternal trauma, toxins; possible genetic etiologies

CLINICAL ISSUES

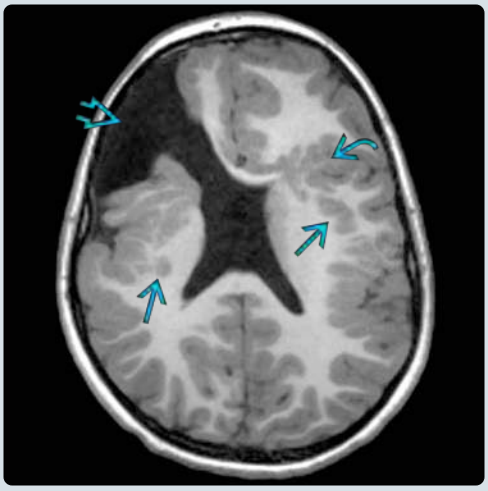
- Seizures, motor abnormalities, developmental delay
- Type & severity of clinical impairment determined by
  - Number, size, & location of clefts
  - Presence of associated malformations
- 1 in 3 children have non-CNS abnormalities

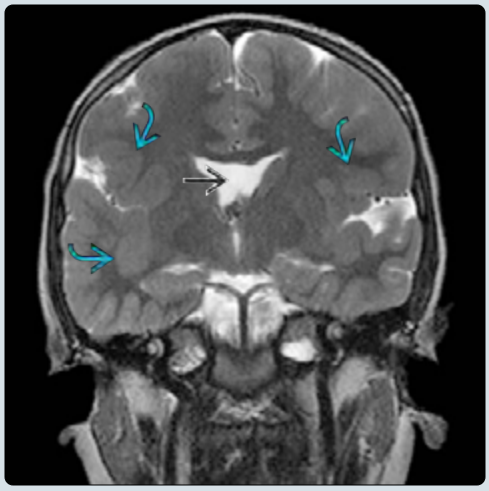
(Left) *Coronal graphic shows bilateral schizencephalic clefts, closed-lip on the right & open-lip on the left. Both clefts are lined by dysplastic gray matter. Note the absence of the septum pellucidum.* **(Right)** *Axial T2WI MR of a 7-year-old patient with schizencephaly shows bifrontal clefts, open-lip on the right & closed-lip on the left. Both clefts are lined by abnormal gray matter.*





(Left) *Axial T1WI MR in the same patient again shows the bifrontal schizencephaly with an open defect on the right & a closed defect on the left. Note the multiple areas of associated subcortical gray matter heterotopia.* **(Right)** *Coronal T2WI MR in the same patient demonstrates associated bilateral cortical thickening in a perisylvian distribution, right > left, compatible with polymicrogyria. Also note the absence of the cavum septum pellucidum.*





IMAGING

General Features

- Best diagnostic clue
  - Full thickness transmantle gray matter (GM)-lined cleft extending from pial to ependymal surfaces
    - Closed-lip (type I): Walls of cleft in direct contact
    - Open-lip (type II): CSF throughout cleft
      - Variable cleft widths
  - Up to 50% bilateral

CT Findings

- NECT
  - Cerebral cleft of CSF density (in open-lip type)
  - GM lining clefts may appear hyperdense
  - Dimple on lateral wall of lateral ventricle indicates ependymal margin of closed-lip cleft
  - Periventricular Ca^{2+} if due to infection (CMV)
  - Thinned, expanded calvarium with large open-lip clefts

MR Findings

- T1WI, T2WI, FLAIR
  - Closed type may appear as irregular tract of GM from cortical surface to ventricle (fused cleft)
    - Look for dimple in wall of lateral ventricle
  - Open type can be wide & wedge-shaped or have nearly parallel walls
    - CSF-filled cleft connects lateral ventricle with subarachnoid space
  - Abnormal GM lining cleft: Polymicrogyria or heterotopia
  - ± thin overlying membrane of pia or ependyma
    - More common on prenatal MR
- T2* GRE
  - ± hemosiderin staining of ventricles &/or cleft from prenatal insult
  - May show Ca^{2+} when associated with CMV

Imaging Recommendations

- Protocol advice
  - Multiplanar sequences (± volumetric acquisition) particularly important for closed-lip type
  - Prior to myelination, GM-lined cleft best seen on T2WI

DIFFERENTIAL DIAGNOSIS

Encephaloclastic Porencephaly

- Parenchymal cavity due to insult after migration complete
- Lined by gliotic white matter, not dysplastic GM

Hydranencephaly

- Destruction of middle & anterior cerebral artery territories
  - Residual tissue supplied by posterior circulation: Posterior fossa, occipital poles, medial temporal lobes

Alobar Holoprosencephaly

- Anterior monoventricle with large dorsal cyst

PATHOLOGY

General Features

- Etiology
  - In utero insult to germinal zone prior to neuronal migration
  - Etiologies may be hypoxic-ischemic, vascular, teratogenic (alcohol, Warfarin, & cocaine), infectious (CMV, HSV)
  - Many potential genetic etiologies
- Associated abnormalities
  - Septooptic dysplasia (SOD)
    - Heterogeneous disorder with 2 or more features of classic triad
      - Agenesis of septum pellucidum &/or corpus callosum
      - Optic nerve hypoplasia
      - Pituitary abnormalities
    - Schizencephaly in 30-50% of SOD patients
    - Septum pellucidum absent in large percentage of schizencephaly cases, especially bilateral clefts
  - GM heterotopia often seen adjacent to cleft
  - Contralateral polymicrogyria common
  - Hippocampal & callosal anomalies

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Seizures, hemiparesis, developmental delay
  - Bilateral clefts: Tetraparesis, blindness, more severe cognitive impairment

Demographics

- Epidemiology
  - Incidence: 1.5/100,000 births
  - 1 in 3 children have non-CNS abnormalities
    - > 50% likely due to vascular disruption
      - Gastroschisis, bowel atresias, & amniotic band disruption sequence
  - Associated with ↓ maternal age, lack of prenatal care

Natural History & Prognosis

- Type & severity of clinical impairment determined by
  - Number, size, & location of clefts
    - Bilateral & open defects have worse prognosis than unilateral & closed types
      - May change from open-lip to closed-lip forms between pre- & postnatal imaging, which could alter prognosis & prenatal counseling
      - Malformation stable postnatally
  - Presence of associated malformations

Treatment

- Surgery for medically intractable epilepsy

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Polymicrogyria

KEY FACTS

TERMINOLOGY

- Traditionally considered malformation of late neuronal migration & cortical organization resulting in abnormal distribution of neurons; disputed by some literature

IMAGING

- Excessively small & disorganized gyral convolutions with shallow sulci, creating appearance of nodular cortex
- Commonly perisylvian; may be unilateral or bilateral
- Imaging findings depend on patient age
  - Fetal: Premature appearance of sulci
  - 1-2 years: Abnormal sulcation pattern
  - > 2 years: Abnormally thick, irregular cortex
- NECT may detect Ca^{2+} in CMV
- Polymicrogyria (PMG) lines clefts of schizencephaly
- Optimal imaging sequence depends on brain maturation
  - Incomplete myelination (< 1 year): Thin-section T2 MR
  - Complete myelination (> 1-2 years): 3D SPGR T1 MR

TOP DIFFERENTIAL DIAGNOSES

- Microcephaly with simplified gyral pattern
- Hemimegalencephaly
- Pachygyria
- "Cobblestone" malformations

PATHOLOGY

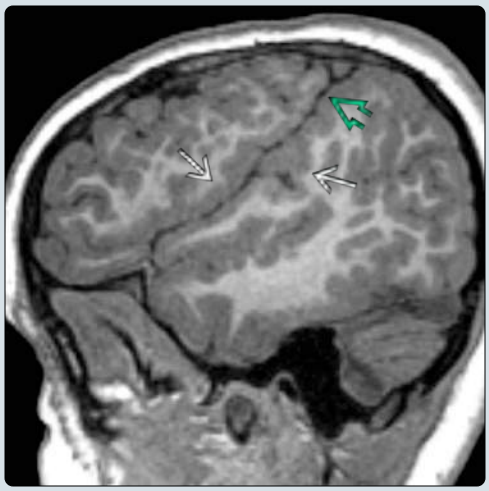
- Causes: Fetal infection, ischemia, or genetic/syndromic
- Associated syndromes: Aicardi, Zellweger, DiGeorge, Warburg micro syndromes

CLINICAL ISSUES

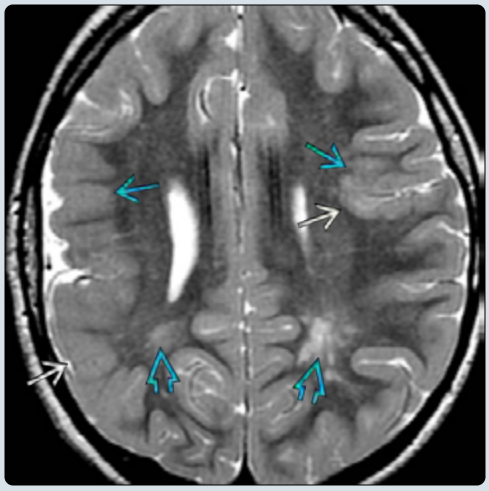
- Most common symptoms: Seizures, developmental delay, spasticity; varies with extent/location of PMG
- Age: Neonatal (in severe) vs. adolescence (in mild, focal)

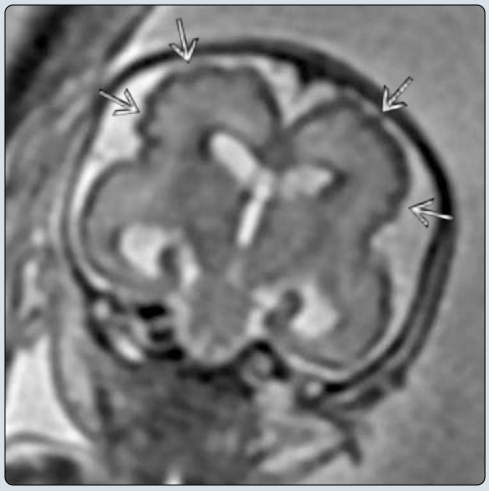
DIAGNOSTIC CHECKLIST

- Look for secondary findings to suggest underlying CMV or metabolic or syndromic diagnosis



(Left) *Coronal oblique graphic shows thickened "pebbly" or nodular gyri of polymicrogyria (PMG) involving the frontal & temporal opercula. Note the abnormal sulcation & irregular gray matter-white matter (WM) junctions in the affected regions.* **(Right)** *Sagittal T1 MR in an 11-year-old left-handed boy with new-onset seizures shows PMG predominantly in the perisylvian area. Note the continuation of the sylvian fissure into the superior parietal lobule, a characteristic appearance for PMG.*





(Left) *Axial T2 MR in a 4-year-old boy with hearing loss shows abnormal sulcation & nodular cortical thickening bilaterally (R > L), consistent with PMG. Also note the periatrial WM injury. The combination of hearing loss, PMG, & WM injury suggests congenital CMV infection.* **(Right)** *Coronal SSFSE MR in a 25-week gestation fetus shows bilateral foci of irregular cortex, consistent with extensive PMG. The cortex in these regions should appear smooth at this time in gestation.*

TERMINOLOGY

Definitions

- Polymicrogyria (PMG): Traditionally considered malformation of late neuronal migration & cortical organization resulting in abnormal distribution of neurons; disputed by some literature
  - Regardless, macroscopic result is nodular cortex with disorganized small undulating gyri & shallow sulci

IMAGING

General Features

- Best diagnostic clue
  - Excessively small & numerous gyri
- Location
  - Can be unilateral, bilateral, multifocal
  - Most common in perisylvian regions (60-70%)
- Size
  - Ranges from single gyrus to entire cerebrum
- Morphology
  - Small irregular gyri
  - Thick nodular cortex with irregular gray-white matter junctions
  - May appear as deep infolding of thick cortex
  - Schizencephalic clefts lined by PMG
  - Sylvian fissure may extend into parietal lobe

CT Findings

- Look for altered sulcation pattern (suggests PMG)
- Periventricular Ca^{2+} in CMV

MR Findings

- **T1WI**: Irregular cortical surface; thick-appearing cortex; deep infolding of irregular cortex
- **T2WI**: 2 imaging patterns depending on patient age
  - < 12 mo: Undulating cortex of normal thickness
  - > 18 mo: Thick, bumpy cortex (6-8 mm)

Nuclear Medicine Findings

- **PET**: Ictal hypermetabolism & interictal hypometabolism

Imaging Recommendations

- Protocol advice
  - Volumetric 3D SPGR (T1WI) MR in mature brain
  - Thin-section T2WI MR if unmyelinated

DIFFERENTIAL DIAGNOSIS

PMG Secondary to Inborn Errors of Metabolism

- Mitochondrial & pyruvate metabolism disorders
- Zellweger syndrome: Severe hypomyelination, PMG

Microcephaly With Simplified Gyral Pattern

- Normal cortex with primary & secondary sulci, but absent tertiary sulci

Hemimegalencephaly

- ↑ size of involved hemisphere (vs. ↓ in PMG)

Pachygyria

- Thick cortex (8-10 mm), smooth gray-white matter junction

"Cobblestone" Malformations

- Hypomyelination, cerebellar dysgenesis, pontine hypoplasia
- Often associated with congenital muscular dystrophy

PATHOLOGY

General Features

- Etiology
  - Intrauterine infection, ischemia, or gene mutations
- Genetics
  - Most common: 1p36.3 & 22q11.2 microdeletions
- Associated abnormalities
  - Aicardi, Zellweger, Warburg micro syndromes
  - Corpus callosum dysgenesis, periventricular nodular heterotopia, & subcortical heterotopia
  - Microcephaly (~ 50%); more common in generalized forms of PMG

Gross Pathologic & Surgical Features

- Multiple small gyri lie in haphazard orientation

Microscopic Features

- Derangements of normal 6-layered cortex
  - Reduced numbers of neurons
  - Areas of 2-, 4-, & 6-layered cortex described, even in different regions of same brain
    - Normal vs. abnormal laminar arrangement disputed

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Seizures, developmental delay, spasticity

Demographics

- Age
  - Symptom onset varies with extent/location of malformation
    - Neonatal manifestations in severely affected vs. 2nd decade of life for focal unilateral PMG
- Epidemiology
  - Malformations of cortical development found in ~ 40% of children with intractable epilepsy

Treatment

- Surgical options with medically refractory seizures
  - Focal PMG may be resected
  - Corpus callosotomy if bilateral or unresectable lesions

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Look for PMG in congenital hemiplegia with epilepsy
  - Schizencephaly always lined by PMG
  - Open sylvian fissures with thick cortex suggest PMG
- Look for secondary findings of CMV or syndrome

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Focal Cortical Dysplasia

KEY FACTS

TERMINOLOGY

- Cortical disorganization & abnormal laminar architecture

IMAGING

- Focal cortical dysplasia (FCD) type 1
  - MR imaging findings present in 22-64%, may be lower in children
    - White matter (WM) volume loss
    - Ill-defined WM increased signal
    - Blurring of gray-white matter junction
    - Cortical signal change
    - Abnormal sulcal & gyral pattern
- FCD type 2
  - MR imaging findings more commonly present (60-100%)
    - Focal signal abnormality in subcortical WM
    - Blurring of gray-white matter junction
    - Cortical thickening & signal change
    - Abnormal sulcal & gyral pattern

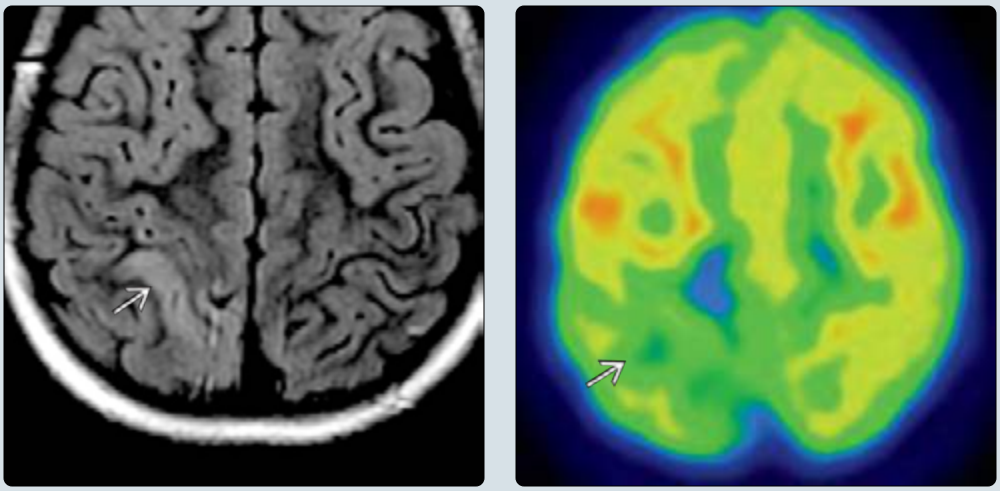
PATHOLOGY

- FCD type 1
  - Abnormal lamination of cortex **without** dysmorphic neurons
- FCD type 2
  - Abnormal lamination of cortex **with** dysmorphic neurons, **with or without** balloon cells
- FCD type 3
  - FCD associated with other structural lesions

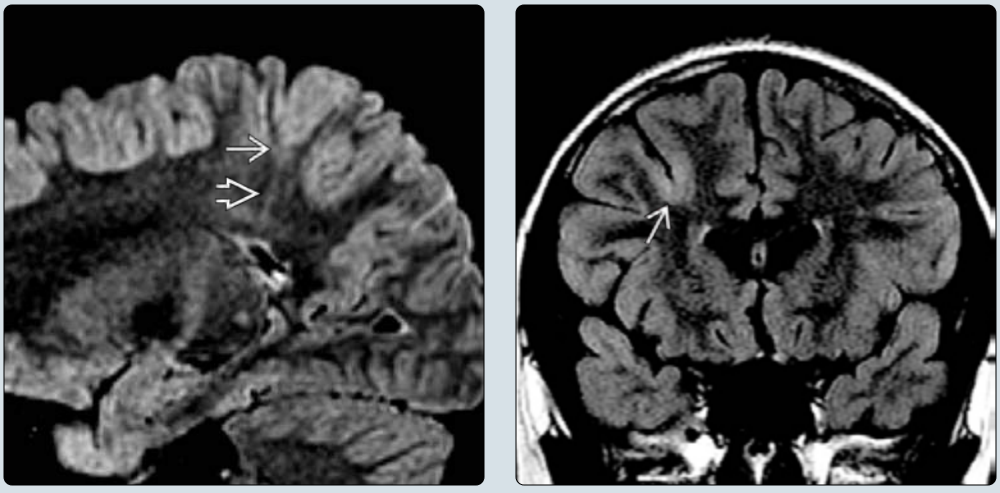
CLINICAL ISSUES

- FCD is most common pathology seen in pediatric patients with surgically treated epilepsy
  - Type 1 dysplasias more commonly seen in adults
- Epilepsy surgery effectiveness dependent on type of underlying pathology
  - Type 1 FCD: 13-21% seizure free
  - Type 2 FCD: 52-91% seizure free

(Left) *Axial FLAIR MR in an 8-year-old girl shows hyperintense signal ⇨ in the right parietal cortex. Pathology demonstrated type 2B focal cortical dysplasia (FCD).* **(Right)** *Axial color-coded FDG PET in the same patient shows focally decreased metabolism ⇨ in the area of MR signal abnormality.*



(Left) *Right sagittal 3D FLAIR MR in an 8-year-old girl with type 2B FCD shows blurring of the gray-white matter junction ⇨ & transmantle signal abnormality ⇨.* **(Right)** *Coronal oblique FLAIR MR in a 16-year-old girl with medically refractory epilepsy shows hyperintense, thickened cortex & gray-white matter blurring ⇨ at the bottom of an abnormally deep sulcus. This is a typical appearance for the "bottom-of-the-sulcus" morphology of FCD. Pathology showed type 2A FCD.*



TERMINOLOGY

Definitions

- Focal cortical dysplasia (FCD)
  - Cortical disorganization & abnormal laminar architecture

IMAGING

General Features

- FCD type 1
  - Only 22-64% have imaging findings, may be lower in children (12%)
  - When present, MR findings are often subtle
    - White matter (WM) volume loss
    - Ill-defined WM increased signal
    - Blurring of gray-white matter junction
    - Cortical signal change
    - Abnormal gyral & sulcal pattern
- FCD type 2
  - 60-100% have identifiable abnormalities on MR
    - Focal signal abnormality in subcortical WM
    - Blurring of gray-white matter junction
    - Increased cortical thickness
    - Cortical signal change
    - Abnormal gyral & sulcal pattern
  - MR findings highly suggestive of type 2
    - Transmantle sign: WM signal tapering from subcortical region to ventricular margin
    - Depth of sulcus morphology: Increased signal at depth of abnormal sulcus ± transmantle sign

Nuclear Medicine Findings

- PET
  - Decreased interictal vs. increased ictal metabolic activity
- Perfusion SPECT
  - Comparative ictal & interictal scans with Tc-99m HMPAO or Tc-99m ECD
    - Increased ictal vs. decreased interictal perfusion

Imaging Recommendations

- Protocol advice
  - Higher field strength (3T) preferred
  - 3D isotropic MR techniques (T1 & FLAIR) allow multiplanar reconstruction, PET/SPECT/MEG fusion, & intraoperative guidance

DIFFERENTIAL DIAGNOSIS

Tuberous Sclerosis

- Radial migration lines & cortical tubers share many imaging features with FCD, especially FCD type 2B
- Tubers & FCD type 2B share similar histopathology

Cortical Neoplasm

- Especially DNET, ganglioglioma, or other low-grade glioma
- Abnormal enhancement excludes FCD

Incomplete or Delayed Myelination

- Focal incomplete myelination may mimic FCD
- Follow-up imaging after myelin maturation helpful

PATHOLOGY

General Features

- Etiology
  - FCD type 1: Disturbance of cortical organization
  - FCD type 2: Disturbance of cellular proliferation

Staging, Grading, & Classification

- FCD type 1
  - Isolated alterations in cortical organization & lamination
    - 1A: Abnormal radial cortical lamination
    - 1B: Abnormal tangential cortical lamination
    - 1C: Abnormal radial & tangential cortical lamination
- FCD type 2
  - Isolated alterations in cortical organization & lamination **with dysmorphic neurons**
    - 2A: Presence of dysmorphic neurons
    - 2B: Dysmorphic neurons **& balloon cells**
- FCD type 3
  - FCD associated with another principal (non-FCD) lesion
    - 3A: FCD associated with **hippocampal sclerosis**
    - 3B: FCD associated with **glial or glioneuronal tumor**
    - 3C: FCD associated with **vascular malformation**
    - 3D: FCD associated with early-acquired **encephaloclastic lesion**

CLINICAL ISSUES

Presentation

- 10% of all epilepsy patients
- 50% of children undergoing surgery for intractable epilepsy

Natural History & Prognosis

- Seizure-free outcome following surgery much better in FCD type 2 (52-91%) compared to FCD type 1 (21%)

Treatment

- Focal resection ± preceding subdural EEG grid localization

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Chiari 1

KEY FACTS

TERMINOLOGY

- Chiari 1 malformation (CM1); synonyms: Chiari type I, Chiari 1 deformity, cerebellar tonsillar ectopia

IMAGING

- Pointed cerebellar tonsils extending ≥ 5 mm below foramen magnum (basion-opisthion/McRae line) with effacement of CSF spaces
- ± retroflexed odontoid, horizontal clivus, basilar invagination, C1 assimilation
- ± caudal descent of brainstem, brainstem compression, medullary kink
- ± syringohydromyelia, scoliosis

TOP DIFFERENTIAL DIAGNOSES

- Normal low-lying cerebellar tonsils
- Chiari 2 malformation
- Tonsillar herniation from increased intracranial pressure
- Intracranial hypotension

PATHOLOGY

- Most common cause believed to be small/underdeveloped posterior fossa; no association with myelomeningocele
- Can be result of premature closure of sutures
  - Causes include shunted infantile hydrocephalus, bone dysplasias, genetic syndromes

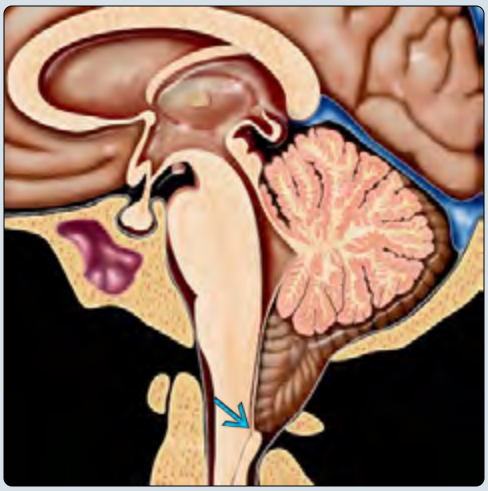
CLINICAL ISSUES

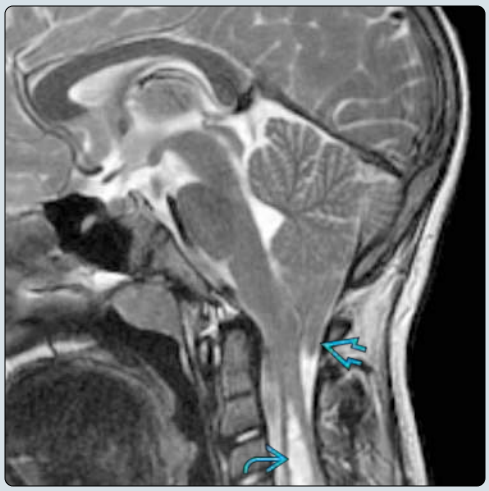
- Most common presenting symptom: Occipital headache
  - Up to 30% of patients asymptomatic
- Goal of surgery in symptomatic patients: Restore normal CSF flow at foramen magnum
  - Posterior fossa (suboccipital) decompression, resection of C1 posterior arch ± duraplasty, cerebellar tonsil cautery

DIAGNOSTIC CHECKLIST

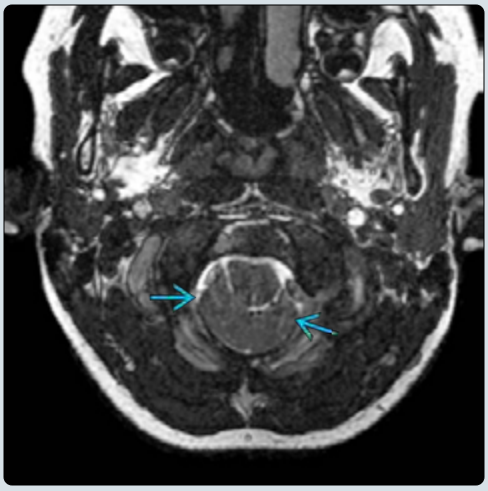
- Degree of tonsillar descent does not always correlate with symptoms: Chiari 1 frequently picked up incidentally

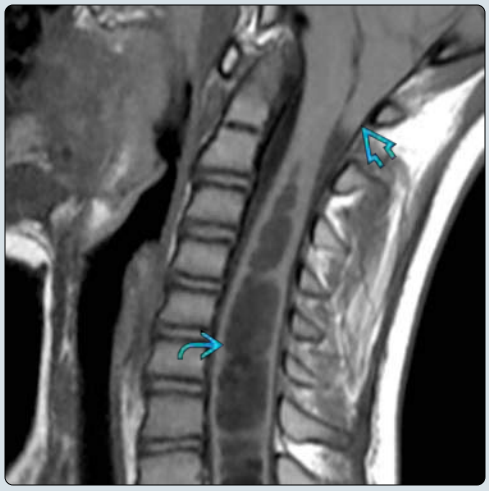
(Left) *Sagittal graphic demonstrates pointed tonsils extending below the foramen magnum to the inferior aspect of the C1 posterior arch. The obex is inferiorly displaced as well.* **(Right)** *Sagittal T2 MR from a 10-year-old patient demonstrates pointed cerebellar tonsils extending below the foramen magnum to the lower C1 level, typical of a Chiari 1 malformation. The CSF is largely effaced at the craniocervical junction, & a syrinx is seen in the cervical spinal cord.*





(Left) *Axial SSFP MR through the foramen magnum in the same patient shows crowding & effacement of the CSF spaces by the low cerebellar tonsils.* **(Right)** *Sagittal T1 MR of the cervical spine in the same patient further demonstrates the cerebellar tonsillar ectopia & large cervicothoracic spinal cord syrinx.*





TERMINOLOGY

Definitions

- Compressed & pointed cerebellar tonsils extending below foramen magnum with effacement of CSF spaces

IMAGING

General Features

- Best diagnostic clue
  - Pointed cerebellar tonsils extending ≥ 5 mm below foramen magnum (basion-opisthion line)
    - No consensus on exact definition

CT Findings

- Crowding of foramen magnum on axial NECT images can mimic cord expansion by tumor
  - Sagittal reconstructions helpful
- Most inferior head CT image may show top of syrinx
- Bone findings may include: Small posterior fossa with short horizontal clivus, retroverted dens, basilar invagination, C1 assimilation

MR Findings

- T1WI, T2WI, FLAIR
  - Pointed (not rounded) cerebellar tonsils extending ≥ 5 mm below foramen magnum
  - Crowded foramen magnum with small/effaced cisterns ± brainstem compression
  - ± small posterior fossa, elongated 4th ventricle
  - ± syringohydromyelia/syrinx, scoliosis
  - Other variants sometimes grouped separately
    - Chiari 1.5: Brainstem herniation
      - Obex located below foramen magnum
    - Complex Chiari: Medullary kink, retroflexed dens, abnormal clival-cervical angle, occipitalization of atlas, basilar invagination, platybasia
- MR cine
  - Restricted CSF flow through foramen magnum ± ↑ brainstem/cerebellar tonsil motion (pistoning)
    - Clinical utility of this sequence debatable

DIFFERENTIAL DIAGNOSIS

Normal Variation of Cerebellar Tonsil Position

- Tonsils may normally lie below foramen magnum
  - May be accentuated by certain head positions
- Tonsils retain normal rounded configuration

Chiari 2 Malformation

- Numerous intracranial findings centered around very small posterior fossa with hindbrain herniation in setting of open spinal dysraphism

Tonsillar Herniation From Increased Intracranial Pressure

- Neoplasm, hemorrhage, hydrocephalus, ischemia

Intracranial Hypotension

- Sagging midbrain, sunken hindbrain with diffuse dural thickening/enhancement, distended veins/dural sinuses, ± subdural hygromas

PATHOLOGY

General Features

- Etiology
  - Primary congenital malformation vs. secondarily acquired morphologic changes
  - Primary: Posterior fossa underdevelopment theory most common
    - Underdevelopment of endochondral occipital bone → small posterior fossa vault + downward hindbrain herniation
    - Not all Chiari 1 patients have small posterior fossa
  - Secondary: Premature closure of cranial sutures &/or generalized abnormal bone formation
    - Shunted infantile hydrocephalus
    - Calvarial thickening of bone dysplasias or thalassemia
    - Genetic syndromes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Occipital headache
    - Induced by cough, Valsalva, or physical exertion
  - Less common: Bulbar, cranial nerve, cord motor/sensory
  - 15-30% of adults with CM1 asymptomatic

Demographics

- Epidemiology: 0.5-3.5% of general population
- Age: Evenly distributed in adult & pediatric patients
- Gender: F > M (3:2)

Treatment

- Posterior fossa decompression: Suboccipital craniectomy with C1 laminectomy ± duraplasty, arachnoid opening/dissection, cerebellar tonsil cautery/resection
- Complex Chiari may also require odontoid resection or craniocervical junction fusion

DIAGNOSTIC CHECKLIST

Consider

- Degree of tonsillar descent does not always correlate with symptoms: Chiari 1 frequently picked up incidentally
  - High likelihood of symptoms if tonsils > 12 mm below foramen magnum

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Chiari 2

KEY FACTS

TERMINOLOGY

- Constellation of intracranial findings, mainly hindbrain herniation, due to open spinal dysraphism of myelomeningocele (MMC) or myelocele/myeloschisis

IMAGING

- Small posterior fossa: Cerebellum herniates downward through foramen magnum & upward through incisura
- Caudal brainstem herniation with cervicomedullary kink, tectal beaking
- ± ventriculomegaly, which may occur in utero or only after MMC repair
  - Colpocephalic configuration of lateral ventricles
  - Stenogyria (compact, narrow gyri) after shunting
- Falx insufficiency, thickened massa intermedia, callosal dysgenesis, ↓ white matter volume
- ± subependymal gray matter heterotopias, polymicrogyria
- Lacunar skull (lückenschädel) → undulations of inner calvarium in first 6 months of life

TOP DIFFERENTIAL DIAGNOSES

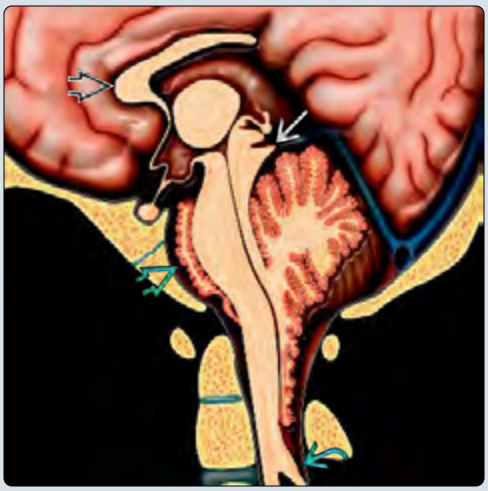
- Chiari 1 malformation
- Chiari 3 malformation
- Intracranial hypotension
- Severe, chronic shunted hydrocephalus (congenital)

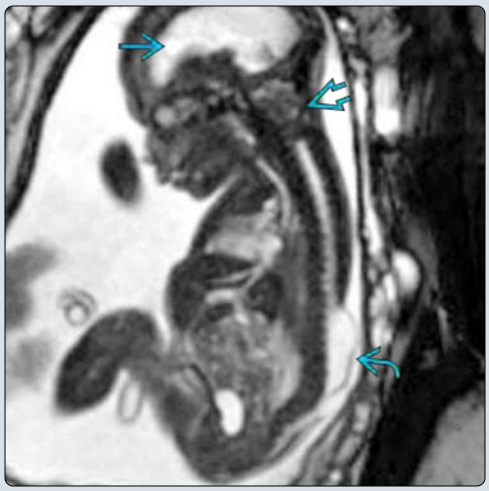
PATHOLOGY

- Secondary to sequelae of chronic in utero CSF leakage through open spinal dysraphism (4th fetal week)
- Folic acid supplementation during periconceptional period significantly ↓ risk

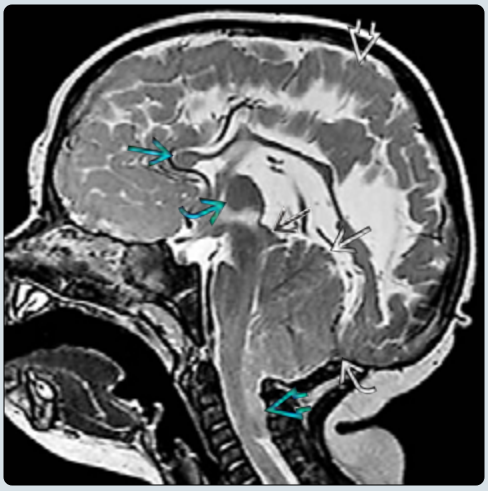
CLINICAL ISSUES

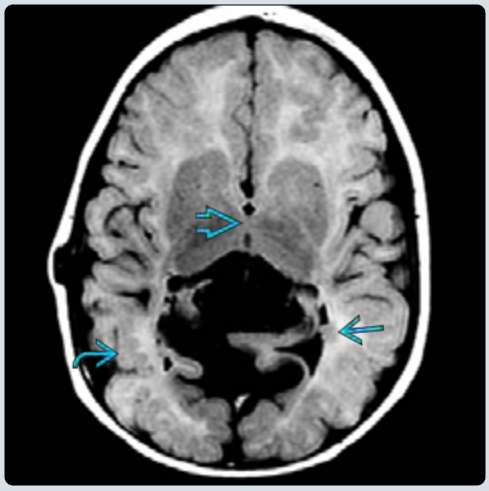
- Lower extremity paresis/spasticity, bowel/bladder dysfunction, symptoms of brainstem compression (swallowing difficulties, stridor, apnea), epilepsy
- In utero repair of MMC ↓ need for postnatal CSF diversion (shunting) & may improve neurologic outcomes in some patients





(Left) *Sagittal graphic shows characteristic findings of the Chiari 2 malformation (CM2), including caudal displacement of the cerebellum & brainstem, elongation of the 4th ventricle, anterior wrapping of the brainstem by the cerebellum, tectal beaking, cervicomedullary kinking, & callosal dysgenesis.* **(Right)** *Sagittal SSFP MR of a 22-week gestation fetus shows a lumbar myelomeningocele (MMC) with a CM2, including a small posterior fossa with cerebellar herniation & ventriculomegaly.*





(Left) *Sagittal T2 MR in the same patient at 8 months of age after postnatal MMC repair & CSF shunting demonstrates multiple findings of CM2 including cerebellar herniation, towering cerebellum, tectal beaking, low torcular herophili, enlarged massa intermedia, callosal dysgenesis, & stenogyria (narrow, compacted gyri).* **(Right)** *Axial T1 MR in the same patient demonstrates a thickened massa intermedia, polymicrogyria, & subependymal gray matter heterotopia.*

TERMINOLOGY

Definitions

- Chiari 2 malformation (CM2): Constellation of characteristic intracranial anomalies, mainly hindbrain herniation, due to open spinal dysraphism of myelomeningocele (MMC) or myelocele/myeloschisis
  - Vast majority of patients with MMC have CM2
  - Few case reports of CM2 without MMC have other congenital CSF leaks (e.g., patent craniopharyngeal duct)

IMAGING

General Features

- Best diagnostic clue
  - Small posterior fossa with inferior cerebellar & brainstem herniation in presence of MMC

Radiographic Findings

- Radiography
  - Lacunar skull (lückenschädel) → numerous undulations of inner calvarium
    - Usually resolves by 6 months of age
  - ± microcephaly

MR Findings

- Characteristic cerebellar morphology
  - Caudal descent of peg-like tonsils/vermis into foramen magnum
  - Towering appearance superiorly with herniation through widened incisura
  - Hemispheres wrap around brainstem
  - Elongated, effaced, inferiorly displaced 4th ventricle with flattened fastigium
  - Cerebellar atrophy over time; most severe form: Vanishing cerebellum
- Brainstem caudal displacement ± cervicomedullary kink
- Low torcular herophili
- Tectal beaking
- ± ventriculomegaly, often with colpocephalic appearance
  - May occur early in utero or only after MMC repair
- Midline anomalies: Large massa intermedia, callosal hypogenesis/dysgenesis, fused forniceal columns, absent septum pellucidum
- ± neuronal migrational anomalies: Heterotopia, polymicrogyria, rhombencephalosynapsis
- Falx insufficiency with interdigitation of hemispheric gyri
- Stenogyria (elongated, compact gyri) after shunting (differs from polymicrogyria)

Ultrasonographic Findings

- Grayscale ultrasound
  - Prenatal US key for early diagnosis
    - Characteristic skull/brain findings (lemon & banana signs) seen as early as 12 weeks
  - Postnatal head US
    - Findings follow MR, though posterior fossa more difficult to visualize due to distance from transducer
      - Mastoid fontanelle view may be helpful for posterior fossa visualization
    - Useful for following change in ventricular size to know if CSF diversion (shunting) required

DIFFERENTIAL DIAGNOSIS

Chiari 1

- Low pointed cerebellar tonsils effacing CSF at foramen magnum
- No association with MMC

Chiari 3

- Brainstem & cerebellum herniating through low occipital/high cervical encephalocele

Intracranial Hypotension

- Sunken hindbrain: Downward displacement of brain
- Dural enhancement, venous sinus engorgement, ± subdural collections
- Typically in setting of CSF leak with postural headaches

Severe Chronically Shunted Hydrocephalus

- May cause collapsed & dysmorphic brain with upward cerebellar herniation

PATHOLOGY

General Features

- Etiology
  - Unified theory of McLone & Knepper
    - Abnormal neurulation → CSF escapes through open neural tube defect (NTD) → failure to maintain 4th ventricular distention → hypoplastic posterior fossa chondrocranium → displaced/distorted PF contents

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 80-90% develop hydrocephalus
  - Varying degrees of lower extremity paresis/spasticity, clubfoot, bowel/bladder dysfunction
  - ± epilepsy, symptoms from brainstem compression (swallowing difficulties, stridor, apnea)

Demographics

- Epidemiology
  - Incidence: 0.44 per 1,000 births; ↓ risk with folate replacement therapy

Treatment

- Surgical management
  - MMC classically repaired in first 48 hours after delivery
  - CSF diversion/shunting ultimately required in 80-90%
  - Fetal MMC repair in select patients

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Neurofibromatosis Type 1

KEY FACTS

IMAGING

- Nonenhancing T2/FLAIR MR hyperintense lesions
  - 60-85% of children with neurofibromatosis type 1 (NF1)
  - Characteristic sites: Globus pallidus, cerebellar WM
  - Little/no mass effect, no enhancement
  - Higher lesion burden may ↓ cognitive functioning
  - ↓ with puberty, gone by adulthood
- Optic pathway gliomas (OPG)
  - 15-20% of children with NF1
  - Anywhere from optic nerve (ON) through optic radiations
  - Fusiform to lobular in shape, ± ON tortuosity
  - ± contrast enhancement
- Plexiform neurofibromas (PNF)
  - Lobular infiltrating soft tissue masses
  - Target appearance in cross section on T2 MR
  - Orbital PNFs associated with sphenoid wing defects
- Vascular dysplasia (moyamoya arteriopathy)
  - 3-7% of children with NF1
  - Vessel narrowing (especially distal ICA) with collaterals

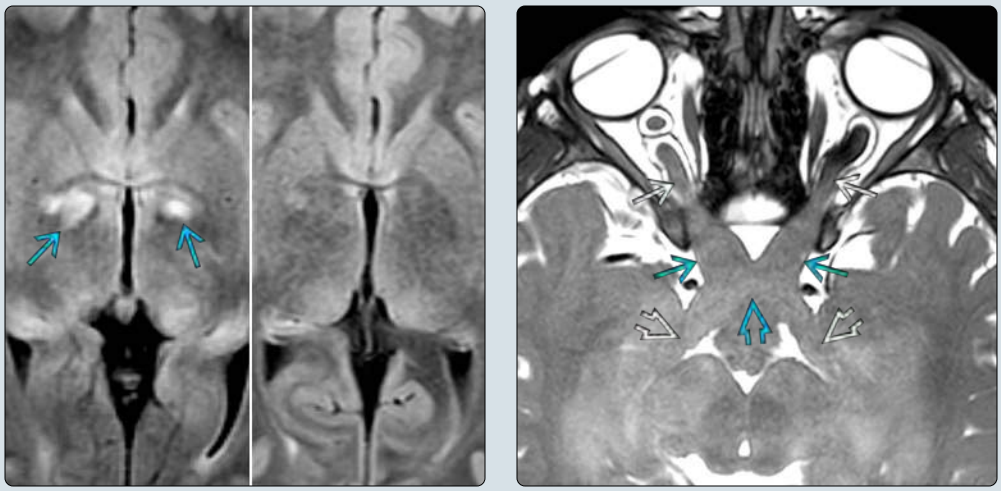
PATHOLOGY

- *NF1* gene locus → long arm of chromosome 17
  - Gene product → neurofibromin (inactivated in NF1)
  - Neurofibromin: RAS GTPase-activating protein responsible for tumor suppression
- Autosomal dominant; 50% new mutations
- Variable expression, virtually 100% penetrance

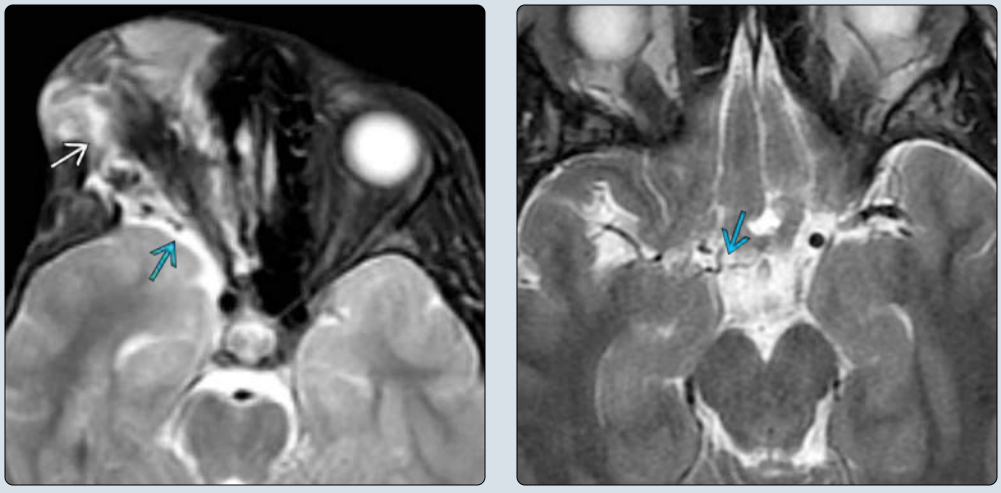
CLINICAL ISSUES

- 1:3,000-5,000 people have NF1
- NF1-related learning disability in 30-60%
- OPG & brainstem gliomas have more indolent behavior in NF1 compared to non-NF1 patients
  - OPG typically not progressive; treatment in ~ 15%
- Phenotype quite variable: Can be dominated by peripheral, paraspinal, or intracranial lesions

(Left) *Axial FLAIR MR images in a patient with neurofibromatosis type 1 (NF1) at 10 years (left) & 15 years (right) of age show near complete resolution of the characteristic nonenhancing signal abnormalities within the globus pallidus nuclei. It is typical for such lesions to resolve in the late teenage years.* **(Right)** *Axial MIP of a CISS MR in a 3-year-old girl with NF1 shows enlargement & tortuosity of the orbital & prechiasmatic optic nerves, optic chiasm, & optic tracts, consistent with an optic pathway glioma.*



(Left) *Axial T2 FS MR in a 12-year-old girl with NF1 shows a large hyperintense lesion in the right orbit, consistent with a plexiform neurofibroma (PFN). Note the absence of the greater sphenoid wing, consistent with sphenoid wing dysplasia. Such osseous changes in the sphenoid occur almost exclusively in the setting of a PFN.* **(Right)** *Axial T2 in a 13-year-old girl with NF1 shows absence of the right carotid terminus with a few collateral vessels noted, consistent with a moyamoya arteriopathy.*



TERMINOLOGY

Abbreviations

- Neurofibromatosis type 1 (NF1)

Synonyms

- von Recklinghausen disease

Definitions

- Neurocutaneous disorder (phakomatosis) characterized by
  - Nonenhancing T2/FLAIR MR signal abnormalities at characteristic brain locations
    - "Unidentified bright objects (UBOs)" or "focal abnormal signal intensities"
  - Plexiform neurofibromas (PNF) of soft tissues
    - Differ from diffuse & localized neurofibromas (NF)
  - Astrocytomas, primarily optic pathway gliomas (OPG)
  - Vascular dysplasias (moyamoya arteriopathy)
  - Skeletal dysplastic lesions
  - Café au lait spots of skin

IMAGING

General Features

- **Nonenhancing hyperintense T2/FLAIR MR lesions**
  - 60-85% of children with NF1
  - Characteristically found in deep gray matter (especially globus pallidus & thalamus), hippocampus, brainstem, cerebellar white matter (WM)
  - Hyperintense on T2/FLAIR, little/no mass effect
  - Often inapparent on T1WI & CT
  - No enhancement
  - Characteristic time course
    - Not present in first 1-2 years of life
    - ↑ in number/size in younger children
    - Diminish in teenage years; usually resolved by adulthood
- **Increased WM volume**
- **OPG**
  - Occur in ~ 15-20% of children with NF1
  - Anywhere from optic nerve (ON) through optic radiations
  - Fusiform enlargement ± enhancement
    - Tortuosity in orbit: Dotted i sign on axial images
- **PNF**
  - Numerous infiltrating soft tissue lobules along courses of peripheral nerves
  - Hyperintense on T2/STIR MR
    - Target sign in cross section (central hypointensity)
  - Variable enhancement, classically central
- **Sphenoid wing dysplasia**
  - Almost always associated with orbital PNF
  - Distortion/absence of lateral orbital wall with anterior expansion of middle cranial fossa
- **Lambdoid defect**
  - Absence of bone along lambdoid suture
- **Vascular dysplasia (moyamoya arteriopathy)**
  - Occurs in 3-7% of children with NF1
  - Vessel narrowing (especially distal internal carotid artery) with collateral formation

Imaging Recommendations

- Best imaging tool
  - Brain/orbits MR C+

PATHOLOGY

General Features

- Genetics
  - *NF* gene locus → long arm of chromosome 17
    - Gene product → neurofibromin, inactivated in NF1
    - Neurofibromin: RAS GTPase-activating protein responsible for tumor suppression
  - Autosomal dominant; 50% new mutations
- Associated abnormalities
  - Pheochromocytomas, malignant peripheral nerve sheath tumors (MPNST)

Staging, Grading, & Classification

- NF1 if ≥ 2 of following
  - ≥ 6 skin café au lait spots, > 5 mm
  - ≥ 2 NF or 1 PNF
  - Axillary/inguinal freckling
  - ON glioma
  - Distinctive bone lesion
  - 1° relative with NF1

CLINICAL ISSUES

Presentation

- Phenotype quite variable: Can be dominated by peripheral, paraspinal, or intracranial lesions

Demographics

- Epidemiology
  - Most common autosomal dominant disorder
  - 1:3,000-5,000 people have NF1

Natural History & Prognosis

- NF1-related learning disability in 30-60%
  - May be associated with ↑ number of characteristic T2/FLAIR MR parenchymal lesions
- 1-3% risk of other CNS glial tumors
  - Typically low grade in children, higher grade in adults
- 8-13% lifetime risk of MPNST
- Vascular dysplasia: If progressive, may cause ischemic injury
- OPG typically have indolent course with observation sufficient in most cases

Treatment

- PNF: Debulking surgery if symptomatic
- OPG: ~ 15% require chemotherapy for progression
- Moyamoya arteriopathy: Surgical revascularization

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Tuberous Sclerosis

KEY FACTS

TERMINOLOGY

- Hamartomas of multiple organs → central nervous system, skin, kidney, bone

IMAGING

- Cerebral tubers
  - Cortical/subcortical lesion expanding overlying gyrus
  - T2/FLAIR hyperintense, T1 hypointense after myelination
  - T1 hyperintense prior to myelination
- Cerebellar tubers
  - Wedge-shaped foci of volume loss
  - Often enhance & calcify
- Subependymal nodules
  - Elongated nodules in locations of fetal germinal matrix
  - Increasing Ca++ over time
  - 30-80% enhance
- Subependymal giant cell astrocytoma
  - Growing nodule at caudothalamic groove
  - WHO grade I neoplasm

TOP DIFFERENTIAL DIAGNOSES

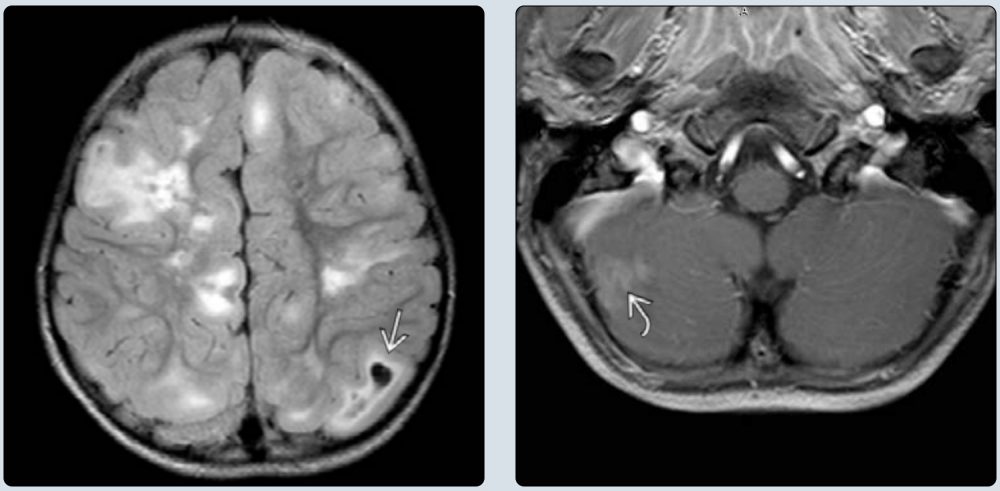
- Focal cortical dysplasia
- Dysembryoplastic neuroepithelial tumor
- Ganglioglioma
- TORCH infections that cause periventricular Ca++
- X-linked subependymal heterotopia

PATHOLOGY

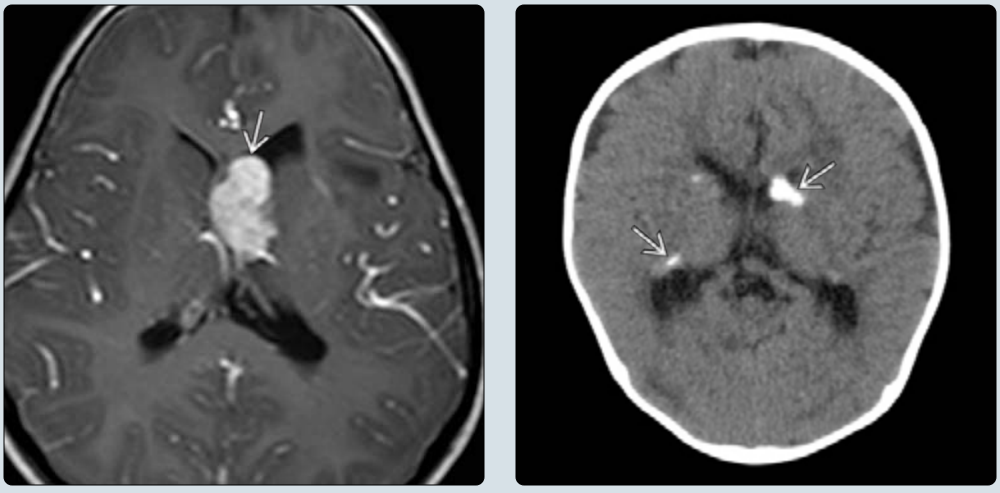
- 2 distinct gene loci
  - *TSC1* (9q34) encodes **hamartin**
  - *TSC2* (16p13) encodes **tuberin** → more severe phenotype

CLINICAL ISSUES

- Medical antiseizure therapy, resection of seizure focus
- mTOR inhibitors now 1st-line therapy for SEGA



(Left) *Axial FLAIR MR of a 3-year-old boy with tuberous sclerosis complex (TSC) shows a moderate to severe burden of cerebral tubers. Cystic change is seen in a left parietal lobe tuber.* **(Right)** *Axial T1 C+ MR in the same patient shows a wedge-shaped, enhancing right cerebellar tuber.*



(Left) *Axial T1 C+ MR in a 4-year-old girl with TSC shows a lobular, homogeneously enhancing mass in the left caudothalamic groove, consistent with a subependymal giant cell astrocytoma.* **(Right)** *Axial NECT in a 23-month-old girl with TSC shows multiple calcified subependymal nodules. Note that the location of the nodules adheres to the distribution of fetal germinal matrix with a preponderance in the caudothalamic grooves.*

TERMINOLOGY

Definitions

- Neurocutaneous syndrome: Hamartomatosis
  - Hamartomas of multiple organs → central nervous system (CNS), skin, kidney, bone

IMAGING

General Features

- Best diagnostic clue
  - Cerebral & cerebellar "tubers"
  - Subependymal nodules (SENs)
  - Subependymal giant cell astrocytoma (SEGA)

Tubers

- Cerebral
  - Cortical/subcortical tubers expand overlying gyri
  - Hyperintense on T2/FLAIR, hypointense on T1 MR
    - T1 hyperintense prior to myelination
  - Typically hypoattenuating on CT
  - Subcortical cystic degeneration in tubers → "empty gyrus"
  - Often associated with radial migration lines extending towards lateral ventricles
  - 3-4% enhance; 50% calcify (by age 10)
- Cerebellar
  - Typically wedge-shaped with volume loss & folia distortion
  - Many show enhancement (33-92%)
  - Many calcify (29%)

SEN

- Elongated subependymal nodules
- Located in areas of fetal germinal matrix with preponderance in caudothalamic grooves
- Caudothalamic groove > body/atrium > temporal horn
- Increasing Ca++ over time; 30-80% enhance

SEGA

- Growing nodule at caudothalamic groove
  - NAA ↓, choline ↑ compared to SEN
  - WHO grade I neoplasm
- Irregular & diffuse enhancement
- Can cause obstructive hydrocephalus

Other Lesions

- Cerebral aneurysms (0.78%) & dolichoectasia
- Retinal hamartoma
- Renal angiomyolipoma (RAM)
- Lymphangioleiomyomatosis (LAM)
- Cardiac rhabdomyoma

DIFFERENTIAL DIAGNOSIS

Infection

- TORCH infections that cause periventricular Ca++

Neoplasms

- Superficial tumors that can resemble tubers
- Intraventricular tumors

Cortical Dysplasia

- Focal cortical dysplasia, especially type II

X-linked Subependymal Heterotopia

- Gray matter heterotopia along lateral ventricle margins

PATHOLOGY

General Features

- Etiology
  - Tuberin & hamartin combine to form complex in vivo
    - Act together to regulate mTOR pathway
    - Regulate cell growth & proliferation
  - Mutations prevent them from downregulating mTOR
  - Affects germinal matrix → disordered neuronal migration & growth
- Genetics
  - 2 distinct gene loci
    - *TSC1* (9q34) encodes **hamartin**
    - *TSC2* (16p13) encodes **tuberin**
    - *TSC2* most common with severe phenotype
      - More likely to have complex partial seizures, infantile spasms, SEGAs, & intellectual disability

Staging, Grading, & Classification

- Diagnostic criteria: 2 major (definite) or 1 major + 1 minor (probable)
  - Major: Tuber, SEN, SEGA, cardiac rhabdomyoma, RAM, LAM, adenoma sebaceum, sub-/periungual fibroma, hypomelanotic macules, shagreen patch, retinal hamartoma
  - Minor: White matter lesions, dental pits, gingival fibromas, rectal polyps, bone cysts, nonrenal hamartoma, retinal achromic patch, confetti skin lesions, multiple renal cysts

CLINICAL ISSUES

Treatment

- Medical antiseizure therapy, resection of seizure focus
- mTOR inhibitors now 1st-line therapy for SEGA

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Sturge-Weber Syndrome

KEY FACTS

TERMINOLOGY

- Syndrome of abnormal cortical venous development
- Imaging features result from progressive venous occlusion, recruitment of alternate drainage pathways, & chronic venous ischemia

IMAGING

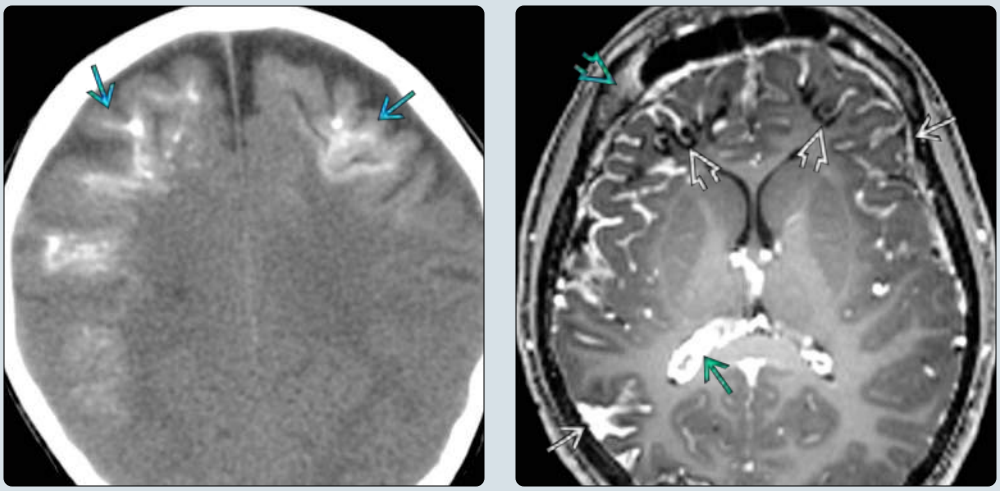
- **CT**: Gyral/subcortical Ca^{2+} (may be tram-track appearance)
  - ± calvarial thickening & sinus hyperpneumatization
- **MR**: Regions of atrophy ± abnormal myelination
  - **T2WI**: Flow voids in enlarged deep/transcerebral veins
  - **FLAIR**: Atrophied lobe(s) ± bright sulcal signal of leptomeningeal angiomatosis
  - **SWI/T2* GRE**: ↓ cortical/subcortical signal from Ca^{2+}
  - **T1WI C+**: Enhancing leptomeningeal angiomatosis
    - Abundant medullary & deep draining veins
    - Choroidal globe enhancement of angioma
  - **MRV**: Absent normal cortical veins in affected region
  - **PWI**: ↑ perfusion early, ↓ perfusion late
- **FDG-PET**: Progressive ↓ metabolism in affected brain

TOP DIFFERENTIAL DIAGNOSES

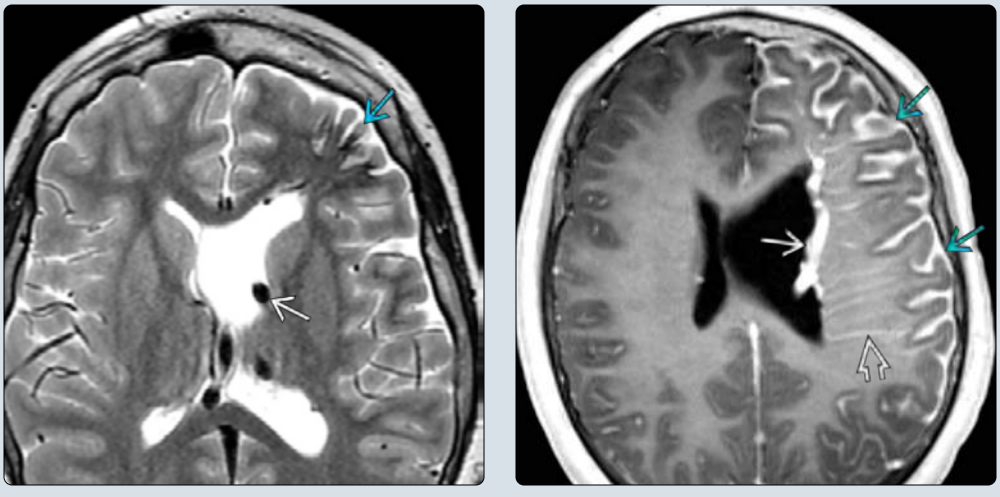
- Acquired meningeal processes
- PHACES syndrome
- Blue rubber bleb nevus syndrome
- Meningioangiomatosis

CLINICAL ISSUES

- Seizures (75-90%), hemiparesis (30-66%)
  - Holohemispheric/bilateral angiomatosis (10-20%) worse than focal involvement
- Forehead cutaneous capillary malformation ("port wine stain") in ~ 95%
- Choroidal angioma in 70% → glaucoma
- Treatment: Aggressive seizure control, resection/hemispherectomy for intractable epilepsy
- MR in early infancy may be normal: Recommend follow-up if patient at risk for Sturge-Weber syndrome clinically



(Left) *Axial NECT in an 11-year-old girl with intractable epilepsy shows bilateral (right > left) subcortical Ca^{2+}, characteristic of Sturge-Weber syndrome.* **(Right)** *Axial T1 C+ FS MR in the same patient shows bilateral leptomeningeal enhancement & enlargement of the right choroid plexus. Note the low signal within the subcortical white matter at the depth of sulci, consistent with Ca^{2+}. There is mildly increased right calvarial thickness secondary to underlying brain parenchymal volume loss.*



(Left) *Axial T2 MR in a 10-year-old boy with right-sided hemiplegia & seizures shows marked parenchymal volume loss & low signal corresponding to an area of cortical/subcortical Ca^{2+} seen on CT. Also note the ipsilateral enlarged deep draining vein.* **(Right)** *Axial T1 C+ MR in the same patient shows extensive left cerebral leptomeningeal enhancement with multiple prominent medullary veins & a large subependymal draining vein. Enlargement of the left lateral ventricle is secondary to left cerebral volume loss.*

TERMINOLOGY

Abbreviations

- Sturge-Weber syndrome (SWS)

Definitions

- Congenital (but not inherited) syndrome in which cortical veins fail to develop normally, leading to numerous long-term intracranial manifestations of chronic ischemia & expanded alternate routes of venous drainage

IMAGING

General Features

- Best diagnostic clue
  - Cerebral lobar atrophy with cortical/subcortical Ca^{2+}, leptomeningeal enhancement, & enlarged ipsilateral choroid plexus
- Location
  - Leptomeningeal angiomatosis unilateral in 80-90%, bilateral in 10-20%

CT Findings

- NECT
  - Gyral/subcortical Ca^{2+} (may be tram-track appearance)
    - Progressive process, usually not present in very young
  - Calvarial thickening & hyperpneumatization of sinuses

MR Findings

- **T1WI**: Altered ("accelerated") white matter (WM) myelination in young patients
- **T2WI**: Hypointense subcortical signal of Ca^{2+} at depth of sulci; ↓ WM signal of accelerated myelination &/or low oxygen tension; enlarged deep vein flow voids
- **FLAIR**: Gliosis often seen late in affected regions
- **SWI/T2* GRE**: ↓ cortical/subcortical signal (Ca^{2+})
  - SWI: Most sensitive to transcerebral medullary collaterals
- **T1WI C+**: Leptomeningeal enhancement (angiomatosis)
  - Late stage: ↓ leptomeningeal enhancement, atrophy
  - Choroidal globe enhancement (choroidal angioma)
- **MRA**: Normal early, but ↓ artery size in affected areas later
- **MRV**: Absent cortical veins in affected region
  - ↑ size of medullary & deep draining veins
- **Perfusion imaging (ASL & contrast perfusion)**
  - Early: ↑ perfusion in affected regions
  - Late: ↓ perfusion in affected regions

Nuclear Medicine Findings

- **FDG-PET**: Progressive hypometabolism in affected regions
- **SPECT**: Hyperperfusion (early), hypoperfusion (late) on interictal scans
- Note that ictal scans (which may be intentional on SPECT or unintentional on FDG-PET) will show ↑ activity

Imaging Recommendations

- Protocol advice
  - MR C+: May be normal at early age; consider follow-up

DIFFERENTIAL DIAGNOSIS

Acquired Meningeal Processes

- Meningitis, tumor spread, hemorrhage, high inspired oxygen content of CSF (usually with anesthesia)

PHACES Syndrome

- **P**osterior fossa malformations, infantile **H**emangiomas, **A**rterial anomalies, **C**oarctation of aorta, **C**ardiac, **E**ye, & **S**ternal anomalies
- Segmental hemangioma of face not fully present at birth

Blue Rubber Bleb Nevus Syndrome

- Multiple small soft tissue venous malformations + intracranial developmental venous anomalies

Klippel-Trenaunay Syndrome

- Extremity overgrowth with extensive capillary-veno-lymphatic malformations & abnormal deep venous system

Meningioangiomatosis

- Rare meningovascular hamartomatous plaque-like mass ± Ca^{2+} & cyst formation

PATHOLOGY

General Features

- Sporadic: Mosaic somatic mutation in gene *GNAQ* (9q21)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Seizures (75-90%), hemiparesis (30-66%)
  - Stroke-like episodes, neurological deficit, headaches, & intellectual disability
  - Facial capillary malformation ("port wine stain") in ~ 95%
    - V1 distribution classic
  - Choroidal angioma (70%), especially with eyelid stain
    - ↑ intraocular pressure/congenital glaucoma → buphthalmos

Demographics

- Rare: 1:20,000-50,000
- Seizures typically develop in 1st year of life

Natural History & Prognosis

- Seizures exacerbate vascular compromise to affected brain → progressive brain injury
- Holohemispheric &/or bilateral involvement worse compared with focal involvement

Treatment

- Presymptomatic: Some recommend low-dose aspirin ± anticonvulsant
- Symptomatic: Aggressive medical management of seizures
- Symptomatic & medically refractory: Consider surgical resection/hemispherectomy

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Infantile Hemangioma and PHACES Association

KEY FACTS

TERMINOLOGY

- **PHACES**: Association of segmental craniofacial infantile hemangioma (IH) & ≥ 1 additional feature
  - **P**osterior fossa malformations
  - **H**emangiomas (infantile)
  - **A**rterial lesions
  - **C**ardiac abnormalities/aortic coarctation
  - **E**ye abnormalities
  - **S**ternal defects or supraumbilical raphe

IMAGING

- Proliferating regional or midline cervicofacial IH: Lobulated or plaque-like, avidly enhancing mass with prominent vascularity
- Unilateral cerebellar hypoplasia & prominent retrocerebellar CSF space
- Widened IAC ± hemangioma ± persistent stapedial artery
- Hypoplasia, aplasia, aberrancy, ectasia, tortuosity, &/or stenoocclusive changes of major cervical/cerebral arteries

TOP DIFFERENTIAL DIAGNOSES

- Sturge-Weber syndrome
- Vestibular schwannoma

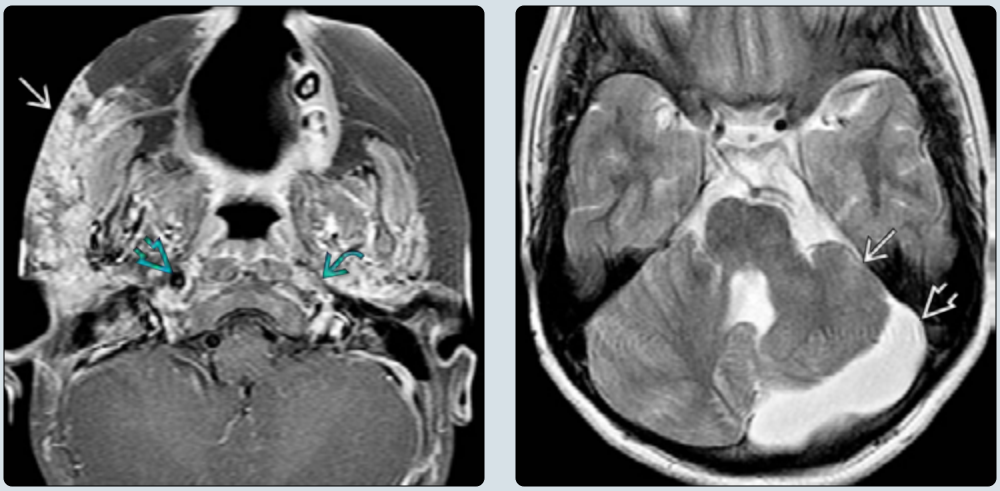
CLINICAL ISSUES

- Most common presentation due to cutaneous findings: Large regional or midline craniofacial IH appearing soon after birth
  - 20% have PHACES; 80-90% female
  - Propranolol (1st line) or steroids accelerate involution
- Prognosis depends on type & severity of anomalies
  - Progressive vascular phenomena leading to neurological deficits from stroke in some cases

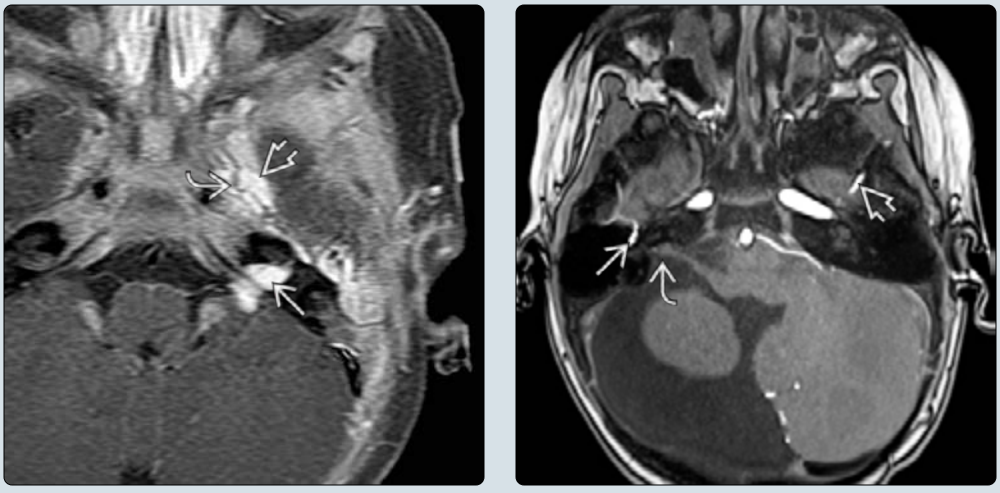
DIAGNOSTIC CHECKLIST

- Look for ipsilateral cerebellar anomaly of PHACES in patient clinically mistaken for Sturge-Weber syndrome (which has port-wine capillary stain, not IH)

(Left) *Axial T1 C+ FS MR shows an enhancing right facial infantile hemangioma (IH) with cutaneous & subcutaneous involvement. There is a normal right internal carotid artery (ICA) flow void. There is no left ICA flow void as a result of ICA atresia.* **(Right)** *Axial T2 MR in the same patient reveals cerebellar hypoplasia & cerebellar cortical malformation with a prominent retrocerebellar CSF space. The ventral aspect of the ipsilateral pons is flattened & small.*



(Left) *Three-week-old boy with a left facial palsy shows avidly enhancing masses within the left internal auditory canal (IAC) & cavernous sinus with a prominent flow void within the left cavernous sinus mass. The IAC mass mimics a schwannoma. These hemangiomas involuted after the 1st year of life.* **(Right)** *Axial MRA of an infant girl with PHACES & right cerebellar hypoplasia shows a right persistent stapedial artery. Note the normal left middle meningeal artery. The right IAC is widened.*



KEY FACTS

TERMINOLOGY

- Congenital phakomatosis characterized by giant or multiple cutaneous melanocytic nevi (CMN) + benign & malignant melanotic lesions of CNS
  - Parenchymal melanosis: Nonmalignant intraparenchymal foci
  - Leptomeningeal melanosis (LMs): Excess of benign melanotic cells in leptomeninges
  - Leptomeningeal melanoma (LMm): Malignant melanoma of leptomeninges
  - Primary CNS malignant melanoma (MM): Focal malignant melanoma of CNS

IMAGING

- Parenchymal melanosis: CMN + foci of intraaxial T1 MR hyperintensity (hyperdense on CT, echogenic on US)
  - Medial temporal lobe, cerebellum, pons, thalami, base of frontal lobes
- LMs & LMm: CMN + diffuse leptomeningeal enhancement
  - Typically develop communicating hydrocephalus
- MM: Focal intraparenchymal enhancing mass

PATHOLOGY

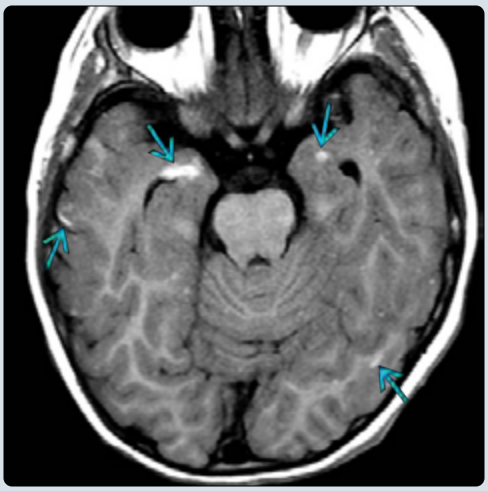
- Focal or diffuse proliferation of melanin-producing cells in both skin & leptomeninges
- Associated cerebellar hypoplasia ~ 10%

CLINICAL ISSUES

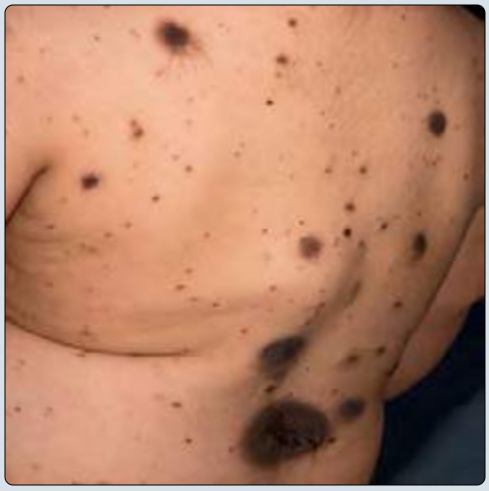
- Only ~ 10-15% of CMN patients have NCM
  - ~ 80-90% of have parenchymal melanosis
  - ~ 10% have LMs or LMm
  - ~ 1-5% have MM
- Screening recommended for CMN patients < 6 months
- Poor prognosis with LMs, LMm, or MM

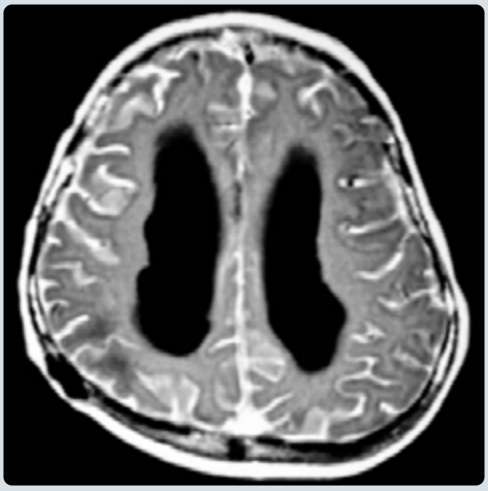
DIAGNOSTIC CHECKLIST

- Normal MR does not exclude diagnosis of NCM
- LMs cannot be distinguished from LMm by imaging



(Left) *Graphic shows localized dark (melanotic) pigmentation of the leptomeninges. Inset demonstrates extension of melanosis into the brain substance along the Virchow-Robin spaces.* **(Right)** *Axial T1 MR in a 4-year-old boy with multiple large congenital cutaneous nevi shows numerous foci of intraparenchymal hyperintense signal. The largest & most conspicuous lesions are located in the medial temporal lobes, the most common location for parenchymal melanosis.*





(Left) *Photograph of the back of a young child with neurocutaneous melanosis shows numerous cutaneous melanocytic nevi, many of which are several centimeters in size, qualifying for the distinction of "giant" nevi.* **(Right)** *Axial T1 C+ MR in a 6 year old shows diffuse leptomeningeal neurocutaneous melanosis. The pial melanosis involves virtually the entire surface of the brain, enhancing strongly & uniformly. There is moderate ventriculomegaly from communicating hydrocephalus.*

Colloid Cyst

KEY FACTS

TERMINOLOGY

- Benign epithelial-lined cyst of anterior 3rd ventricle

IMAGING

- Best clue: Round, homogeneous midline mass at foramen of Monro with lateral ventricular enlargement
  - Mean size: Asymptomatic 0.8 cm, symptomatic 1.4 cm
- CT: Hyperdense (70-80%); iso-/hypodense (20-30%)
- MR: Variable signal characteristics
  - T1 hyperintense (40-60%)
  - T2 hypointense (50-60%)
  - No FLAIR suppression
  - No restricted diffusion
  - No enhancement; rarely, thin rim enhancement
- Pillars of fornix straddle anterior aspect of cyst

TOP DIFFERENTIAL DIAGNOSES

- CSF flow artifact (MR pseudocyst)
- Subependymal giant cell astrocytoma (SEGA)
- Arachnoid cyst
- Neurocysticercosis
- Metastases

PATHOLOGY

- Derived from embryonic endoderm
  - Outer wall: Thin fibrous capsule
  - Inner lining: Columnar epithelium
  - Cyst contents: Proteinaceous material, exfoliated cells

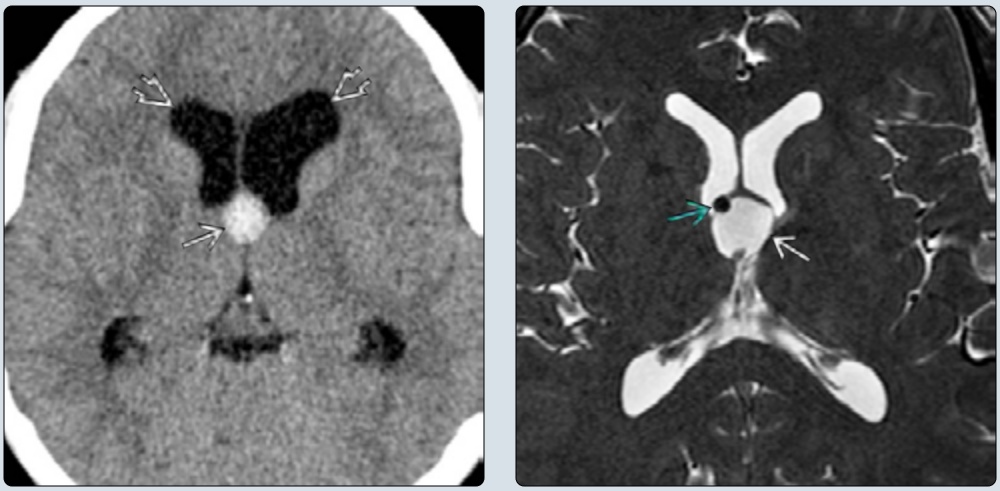
CLINICAL ISSUES

- 40-50% asymptomatic & discovered incidentally
- 90% stable over time
- Most common treatment: Complete surgical resection
  - Recurrence rare if resection complete
- Asymptomatic patients may be treated conservatively with clinical & imaging follow-up

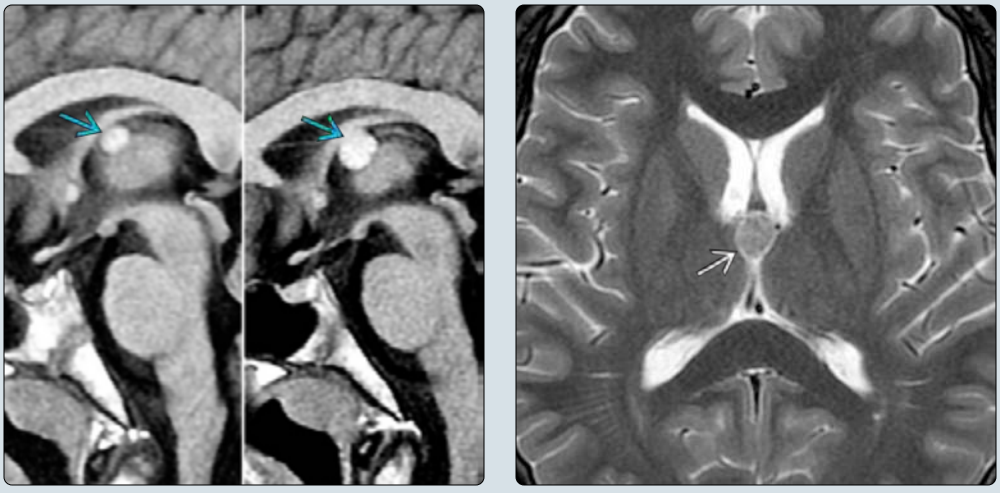
DIAGNOSTIC CHECKLIST

- NECT & T1 MR often more sensitive than T2, FLAIR, or DWI

(Left) *Axial NECT in a 14-year-old boy with headaches shows a hyperattenuating mass at the foramen of Monro, which is causing mild enlargement of the lateral ventricles. The mass was endoscopically resected; pathology confirmed a colloid cyst.* **(Right)** *Follow-up axial SSFP MR in the same patient shows recurrence of the colloid cyst, an uncommon occurrence. Note the small focus of hemosiderin from the prior endoscopic resection.*



(Left) *Sagittal T1 MR images in an asymptomatic boy at 10 (L) & 15 (R) years of age show an interval increase in the size of the colloid cyst in the anterior superior 3rd ventricle. Most colloid cysts are stable over years, but some will increase.* **(Right)** *Axial T2 MR in the same patient shows a foramen of Monro mass with contents that are isointense to gray matter. The majority of colloid cysts are hypo- or isointense to gray matter on T2 MR as decreased T2 signal correlates with increased cholesterol content & viscosity.*



TERMINOLOGY

Definitions

- Benign epithelial-lined, mucin-containing cyst; almost exclusively located in anterior superior 3rd ventricle

IMAGING

General Features

- Best diagnostic clue
  - Hyperdense midline foramen of Monro mass on NECT
- Location
  - 99% in anterior superior 3rd ventricle
    - Anterior to interthalamic adhesion
    - Posterior to fornices
    - Posteromedial but bulging into foramen of Monro
- Size
  - Few mm to 3 cm
    - Symptomatic mean size: 1.4 cm
    - Asymptomatic mean size: 0.8 cm
- Morphology
  - Usually spherical or ovoid

CT Findings

- NECT
  - Usually hyperdense (70-80%)
    - ↑ density correlates with ↑ T1 & ↓ T2 signal on MR
  - Minority iso-/hypodense (20-30%)
  - ± hydrocephalus

MR Findings

- **T1WI**: Hyperintense (40-60%); iso- (~ 20%); hypo- (~ 20%)
- **T2WI**: Hypointense (50-60%); iso-/hyperintense (~ 40%)
- **FLAIR**: Does not suppress like cerebrospinal fluid (CSF); usually iso-/hyperintense
- **DWI**: No diffusion restriction
- **T1WI C+**: Nonenhancing or rare peripheral (rim) enhancement

DIFFERENTIAL DIAGNOSIS

CSF Flow Artifact (MR Pseudocyst)

- Most pronounced on FLAIR MR
- Multiple different sequences & planes confirm artifact

Subependymal Giant Cell Astrocytoma

- Off-midline solid, lobulated, enhancing mass at foramen of Monro in child with tuberous sclerosis

Arachnoid Cyst

- Rare in ventricle; usually arises from suprasellar cistern
- Follows CSF signal intensity on all sequences

Craniopharyngioma

- Suprasellar location much more common than 3rd ventricle
- Often multilobulated with Ca^{2+} & rim/nodular enhancement

Metastases

- Dissemination of tumor in CSF, most commonly of primary CNS origin in children

Neurocysticercosis

- Single or multiple lesions within parenchyma & cisterns
  - Look for scolex along margin of cyst
- Ca^{2+} frequent

PATHOLOGY

General Features

- Outer wall: Thin fibrous capsule
- Inner lining
  - Pseudostratified epithelium with ciliated & goblet cells
  - Individual cells positive for cytokeratin & EMA
- Cyst contents
  - Amorphous proteinaceous ("colloid") material
  - Scattered exfoliated cells

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Headache (~ 75%), nausea & vomiting, visual symptoms
  - 40-50% asymptomatic & discovered incidentally
    - 3-, 5-, 10-year incidence of developing cyst-related symptoms: 0%, 0%, 8%, respectively
- Other signs/symptoms
  - Acute foramen of Monro obstruction may rarely lead to rapid onset hydrocephalus, herniation, death

Demographics

- Age
  - Mean: 30-40 years; 8% < 15 years at diagnosis

Natural History & Prognosis

- Factors associated with symptomatic cysts
  - Younger age
  - Ventricular dilation
  - Larger cyst size
  - ↑ signal intensity on T2 MR
- Rare cases of sudden death reported in literature
  - However, no reported cases of patients with previously identified colloid cysts resulting in sudden death

Treatment

- Symptomatic patients
  - Complete resection: Endoscopic vs. microsurgical
  - Recurrence rare, especially with complete resection
- Asymptomatic (incidental) patients
  - Conservative management: Clinical & imaging follow-up
  - However, with relatively low morbidity, surgery also considered for asymptomatic patients

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- NECT & T1 MR often more obvious than T2, FLAIR, or DWI
  - Pathology-weighted sequences can be insensitive, especially for smaller lesions

SELECTED REFERENCES

1. Sheikh AB et al: Endoscopic versus microsurgical resection of colloid cysts: a systematic review and meta-analysis of 1,278 patients. World Neurosurg. 82(6):1187-97, 2014

Arachnoid Cyst

KEY FACTS

TERMINOLOGY

- Focal extraaxial CSF collection lined by arachnoid

IMAGING

- Location
  - Middle cranial fossa: 50-70%
  - Retrocerebellar: 15-20%
  - Less common: Basilar cisterns, interhemispheric fissure, cerebral convexities
  - Rare: Intraventricular
- Displaces adjacent vessels & nerves
- Typically shows less mass effect than expected
  - Adjacent brain accommodates cyst
- Calvarial remodeling: Thinning/scalloping ± bulging
- **US**: Anechoic; no internal vascularity
- **NECT**: Isodense to CSF, unless hemorrhage occurs (rare)
- **MR**: Follows CSF signal intensity on all sequences
  - Fluid signal suppression on FLAIR
  - DWI/ADC signal equal to CSF

TOP DIFFERENTIAL DIAGNOSES

- Epidermoid cyst
- Chronic subdural hematoma
- Subdural hygroma
- Mega cisterna magna

PATHOLOGY

- Wall consists of flattened but normal arachnoid cells

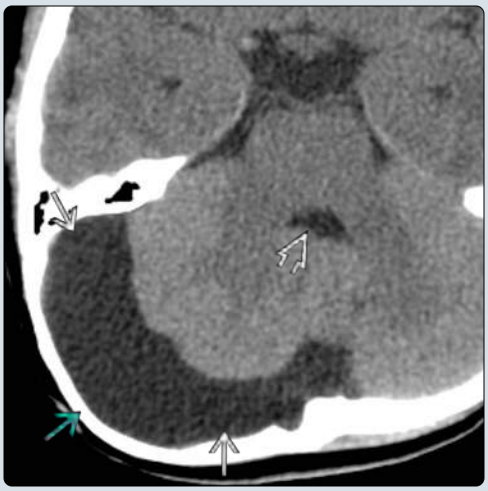
CLINICAL ISSUES

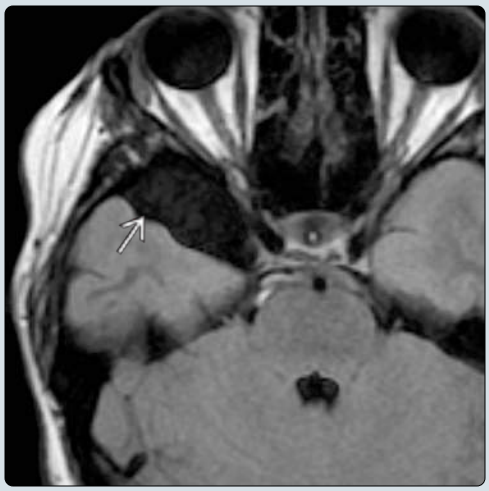
- Asymptomatic in vast majority (typically found incidentally)
- More likely to grow in younger patients (< 4 years of age)
- In young children: 80% stable, 10% enlarge, 10% decrease
- Surgery only indicated if symptoms directly attributable to arachnoid cyst

DIAGNOSTIC CHECKLIST

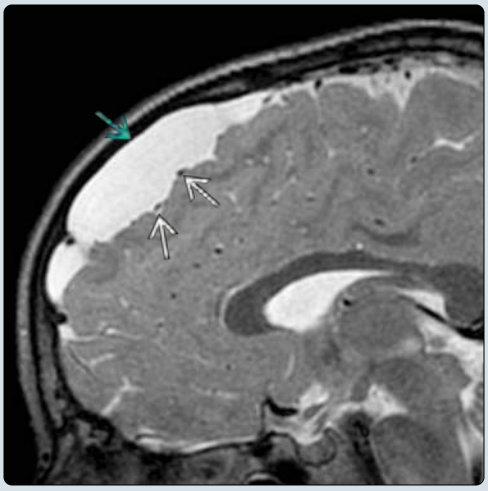
- FLAIR, DWI best MR sequences for distinguishing cystic-appearing intracranial masses

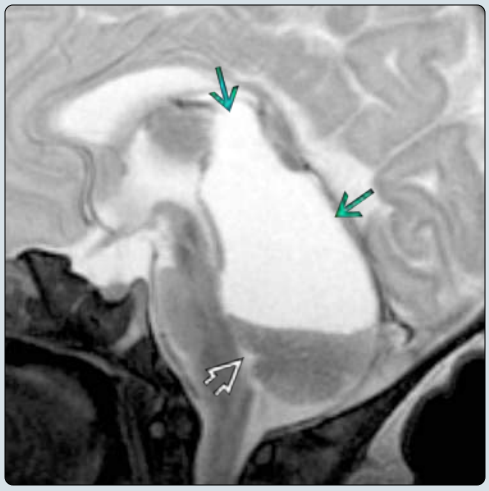
(Left) *Axial NECT in a 6-year-old boy shows a right retrocerebellar cystic collection. Note the characteristic imaging features of an arachnoid cyst (AC): Isoattenuating to CSF, bulging & thinning of the overlying calvaria, & relatively mild mass effect on the 4th ventricle.* **(Right)** *Axial FLAIR MR in a 14-year-old girl with headaches shows fluid signal suppression within the right middle cranial fossa arachnoid cyst. This is the most common location of ACs.*





(Left) *Sagittal T2 MR in a 9-year-old boy with a right frontal arachnoid cyst shows displacement of vessels & cortex with scalloping of the calvaria. The relative lack of mass effect despite the size of the lesion is characteristic of an AC.* **(Right)** *Sagittal T2 MR in a newborn male shows a large cyst in the quadrigeminal cistern with moderate mass effect on the brainstem, 4th ventricle, & cerebellum. While most ACs are incidental, larger cysts in the basilar cisterns can cause significant mass effect & symptoms.*





TERMINOLOGY

Definitions

- Arachnoid cyst (AC): Focal extraaxial CSF collection lined by arachnoid; generally lacks free communication with ventricles or subarachnoid space

IMAGING

General Features

- Location
  - Middle cranial fossa (MCF): 50-70%
  - Retrocerebellar: 15-20%
  - Less common locations: Cerebral convexities or interhemispheric fissure (5-10%), cerebellopontine angle (5-10%), quadrigeminal plate (1-5%), suprasellar (1-5%)
- Size
  - Mean diameter: 3 cm; ~ 20% > 5 cm
- Morphology
  - Sharply marginated extraaxial fluid collection
    - Lentiform, crescentic, ovoid, or lobular
    - Thin, nearly imperceptible wall
  - Less than expected mass effect
    - Adjacent brain accommodates cyst; parenchyma may be mildly hypoplastic
  - Thinning/scalloping ± bulging of overlying calvaria

CT Findings

- **NECT**: Isodense to CSF
- **CECT**: Nonenhancing cystic mass displaces vessels
- **CT cisternogram**: AC shows absent or delayed opacification relative to subarachnoid spaces

MR Findings

- **T1WI**: Isointense to CSF
- **T2WI**: Isointense to CSF
  - Displaced vascular flow voids; mild deformation of adjacent cortex
- **PD/intermediate**: Isointense to CSF
- **DWI**: Follows CSF signal
- **FLAIR**: Suppresses completely, similar to CSF
  - No edema in adjacent brain parenchyma
- **T1WI C+**: No cyst enhancement
- **Phase contrast cine**: Reduced/absent CSF flow compared to subarachnoid spaces (e.g., mega cisterna magna)

Ultrasonographic Findings

- Only useful in young infants with open fontanelles
- **Grayscale**: Anechoic with ↑ posterior acoustic shadowing
- **Color Doppler**: No internal vascularity

DIFFERENTIAL DIAGNOSIS

Epidermoid Cyst

- Does not follow CSF on all modalities/sequences
  - Markedly restricted diffusion (light-bulb bright) on MR

Chronic Subdural Hematoma

- Crescentic collection differing from CSF appearance
  - Hyperdense on CT/hyperintense on FLAIR, PD, & T1 MR relative to CSF (though difference may only be mild)

Subdural Hygroma

- CSF leak from subarachnoid space into subdural space
  - 2-7 days after trauma

Porencephalic Cyst

- Result of remote trauma or ischemic injury
- Surrounded by injured, chronically abnormal brain (vs. otherwise normal compressed/distorted brain)

Neuroepithelial Cyst

- Periventricular, intraventricular, or choroidal fissure cyst

Epidural Abscess

- Collection with enhancing wall & restricted diffusion
- Typically with relevant causative findings (e.g., sinusitis, penetrating injury)

Mega Cisterna Magna

- Communicates freely with subarachnoid space

PATHOLOGY

Microscopic Features

- Wall consists of flattened but normal arachnoid cells
- No inflammation or neoplastic changes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Vast majority asymptomatic & found incidentally
  - Associated symptoms vary with size & location of cyst

Demographics

- Age
  - Most symptomatic ACs diagnosed in children
  - Cysts in older children & adults usually incidental

Natural History & Prognosis

- More likely to enlarge in younger patients (< 4 years old)
  - In young children: 80% stable, 10% ↑, 10% ↓

Treatment

- Typically none
- Treatment only indicated if symptoms directly attributable to AC & potential benefits of surgery outweigh risks
- Surgical options
  - Long-term outcomes similar among different options
    - Endoscopic cyst fenestration
    - Open microsurgical cyst resection/fenestration
    - Cystoperitoneal shunt

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- FLAIR, DWI best MR sequences for distinguishing cystic-appearing intracranial masses

SELECTED REFERENCES

1. Huang JH et al: Analysis on clinical characteristics of intracranial Arachnoid Cysts in 488 pediatric cases. Int J Clin Exp Med. 8(10):18343-50, 2015
2. Ali ZS et al: Pediatric intracranial arachnoid cysts: comparative effectiveness of surgical treatment options. Childs Nerv Syst. 30(3):461-9, 2014
3. Al-Holou WN et al: Prevalence and natural history of arachnoid cysts in children. J Neurosurg Pediatr. 5(6):578-85, 2010

Dermoid and Epidermoid Cysts

KEY FACTS

TERMINOLOGY

- Intracranial inclusion cysts containing ectodermal tissue
  - Dermoids contain squamous epithelium & associated dermal appendages
  - Epidermoids consist of squamous epithelium

IMAGING

- Dermoids: Fatty mass with layered septations
  - NECT: Fat density may mimic gas; Ca^{2+} in 20%
  - MR: Striated/layered appearance, nonenhancing
    - T1WI: Iso-/hyperintense to brain
      - Incomplete fat suppression
    - T2WI: Heterogeneous signal
- Epidermoids: Nonenhancing, T2 hyperintense (~ CSF) mass
  - NECT: Iso- to slightly hyperdense to CSF
  - MR: Well-defined mass, lobulations on SSFP images
    - T1WI: Iso-/hyperintense to CSF on T1
    - T2WI: Isointense to CSF but no FLAIR suppression
    - DWI: Marked diffusion restriction

TOP DIFFERENTIAL DIAGNOSES

- Craniopharyngioma
- Arachnoid cyst
- Lipoma
- Teratoma

PATHOLOGY

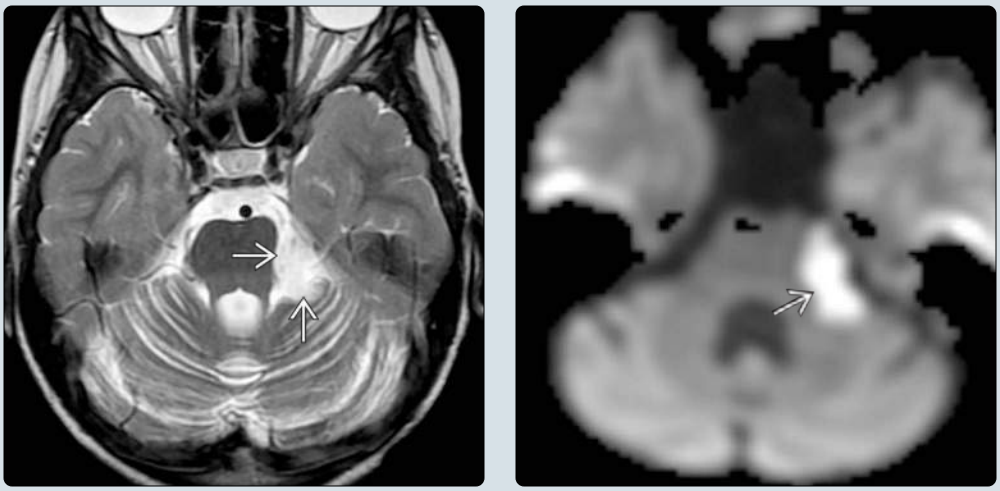
- Dermoids: Mixture of greasy lipid, cholesterol debris
- Epidermoids: Lobular mass with shiny (pearl-like) surface

CLINICAL ISSUES

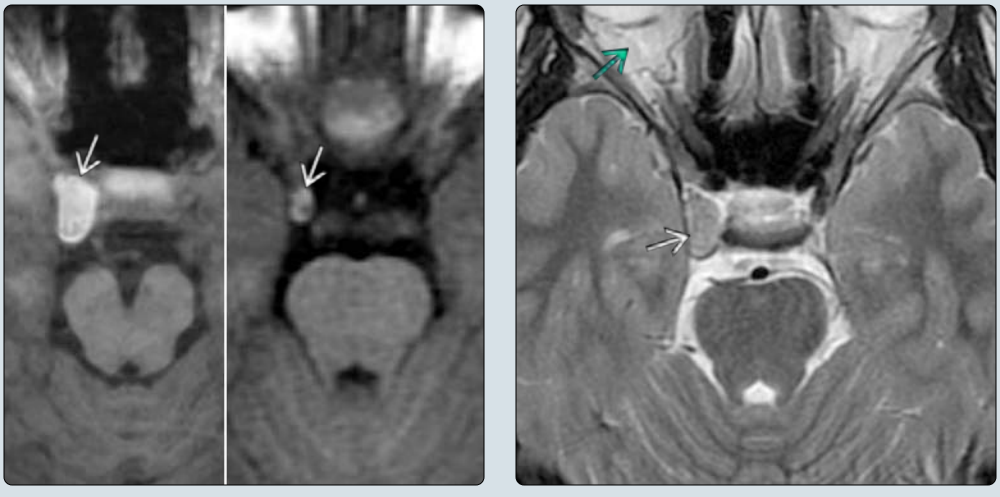
- Dermoid rupture → acute severe headache, collapse
- Dermoids rare: < 0.5% of primary intracranial tumors
- Epidermoids 5-10x more common

DIAGNOSTIC CHECKLIST

- Fat suppression of dermoid much less "clean" than lipoma
- IV contrast helps distinguish dermoid from craniopharyngioma



(Left) *Axial T2 MR in a 7-year-old child shows an epidermoid cyst in the left cerebellopontine angle. The T2 signal is essentially isointense to CSF. Without the associated mass effect on the brainstem & cerebellum, this lesion would not be visible on this sequence.* **(Right)** *Axial DWI MR in the same patient shows "light bulb" hyperintense signal within the mass, consistent with diffusion restriction. This is a characteristic feature of an epidermoid cyst, which was confirmed at surgery.*



(Left) *Axial T1 MR images in a patient at 1 year (right) & 15 years (left) show a hyperintense parasellar mass, which has increased in size over time. It can be difficult to distinguish a lipoma from a dermoid cyst, but substantial growth over time favors the latter.* **(Right)** *Axial T2 MR in the same patient at 15 years shows the mass to be isointense to gray matter & significantly darker than orbital fat, favoring a dermoid cyst over a lipoma. The lesion did not restrict diffusion (not shown), typical of a dermoid cyst.*

TERMINOLOGY

Definitions

- Intracranial inclusions of ectodermal tissue
- Dermoids contain squamous epithelium & associated dermal appendages
  - Sebaceous glands, dental enamel, hair follicles
- Epidermoids consist of squamous epithelium

IMAGING

General Features

- Best diagnostic clue
  - Dermoids: Fatty, nonenhancing mass with layered septations
  - Epidermoids: T2 hyperintense (~ CSF intensity), nonenhancing, diffusion-restricting mass
- Location
  - Dermoids: Usually midline, common in parasellar & frontonasal regions
  - Epidermoids: Most commonly in cerebellopontine angle & near 4th ventricle
    - Peripheral epidermoids typically located adjacent to cranial sutures

CT Findings

- NECT
  - Dermoids: Often have fat density; Ca^{2+} in 20%
    - Rupture → fat droplets within cisterns & sulci, fat/fluid levels in ventricles
  - Epidermoids: Usually iso- to hyperdense to CSF

MR Findings

- Dermoids: Striated or layered internal appearance
  - **T1WI:** Iso- to hyperintense to brain
    - Not as uniformly bright as lipomas
    - Suppress with fat saturation but not as uniformly as lipoma
  - **T2WI:** Heterogeneous signal
  - **DWI:** Variable but diffusion restriction typically < epidermoids
  - **T1WI C+:** No internal contrast enhancement
    - Rupture → chemical ventriculitis & ependymal enhancement
- Epidermoids: Small septations or lobules may be visible, especially on 3D SSFP imaging
  - **T1WI:** Iso- to slightly hyperintense to CSF
  - **T2WI:** Isointense to CSF (may mimic arachnoid cyst)
  - **FLAIR:** Does not suppress like CSF
  - **DWI:** Marked diffusion restriction ("light bulb bright")
  - **T1WI C+:** No internal contrast enhancement
  - "Atypical" epidermoids (< 5%)
    - T1 hyperintense & T2 hypointense
    - Atypical signal corresponds to hemorrhage

Imaging Recommendations

- Best imaging tool
  - MR with contrast
- Protocol advice
  - Use fat suppression, DWI, & FLAIR
  - Use MRA to assess vascular narrowing/encasement
  - Look for chemical shift artifact, especially with rupture

DIFFERENTIAL DIAGNOSIS

Craniopharyngioma

- T1 hyperintense foci do not change with fat suppression
- Enhancement in > 90%

Arachnoid Cyst

- Similar to epidermoids on NECT & T2WI MR
- Suppresses on FLAIR MR; do not restrict diffusion

Lipoma

- Fatty signal/attenuation more homogeneous

Teratoma

- Germ cell tumor that contains ≥ 2 embryologic layers
- Usually has enhancing components

PATHOLOGY

General Features

- Etiology
  - Congenital: Inclusion of cutaneous ectoderm during neural tube closure
  - Acquired: Displacement of epithelium into CNS during lumbar puncture, trauma, surgery

Gross Pathologic & Surgical Features

- Dermoids: Mixture of greasy lipid, cholesterol debris
  - Often contain hair, may contain enamel
- Epidermoids: Lobular mass with shiny (pearl-like) surface
  - Cyst contents: Soft, waxy, flaky material

CLINICAL ISSUES

Demographics

- Epidemiology
  - Dermoids rare: < 0.5% of primary intracranial tumors
  - Epidermoids 5-10x more common

Natural History & Prognosis

- Slowly growing, often asymptomatic
- Dermoid rupture can cause significant morbidity/mortality

Treatment

- Complete microsurgical excision
  - Residual capsule may lead to recurrence
  - Subarachnoid dissemination of contents may occur

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Use contrast to distinguish suprasellar dermoid from craniopharyngioma
- Fat suppression of dermoid much less "clean" than lipoma
- Epidermoids easily distinguished from arachnoid cysts with appropriate sequences
  - Use FLAIR, DWI, & 3D SSFP (FIESTA, CISS, etc.) MR

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1. Lynch JC et al: Surgical strategy for intracranial dermoid and epidermoid tumors: An experience with 33 Patients. Surg Neurol Int. 5:163, 2014
2. Ren X et al: Clinical, radiological, and pathological features of 24 atypical intracranial epidermoid cysts. J Neurosurg. 116(3):611-21, 2012

Pineal Cyst

KEY FACTS

TERMINOLOGY

- Nonneoplastic intrapineal glial-lined cyst

IMAGING

- Most common appearance: Single round/ovoid cyst < 1 cm
- Lobulated shape & internal septations common
- CT
  - Low-attenuation focus in pineal region
    - May be difficult to identify thin walls on CT
  - 25% with Ca^{2+} in cyst wall; more common with ↑ age
- MR
  - Cyst contents typically similar to CSF on T1, T2
  - Cyst contents do not suppress on FLAIR
  - Cyst contents do not restrict diffusion
  - ~ 90% show wall enhancement
    - Most common pattern: Thin rim (≤ 2 mm)

TOP DIFFERENTIAL DIAGNOSES

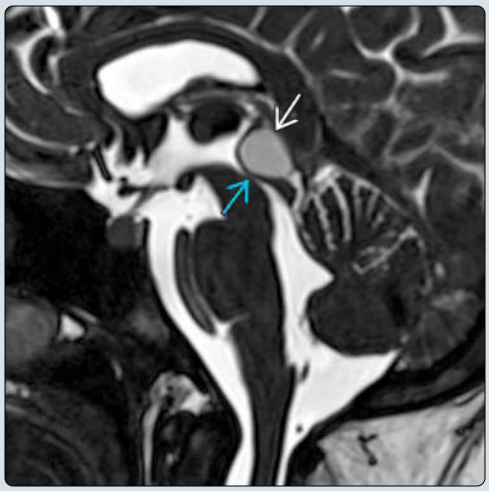
- Arachnoid cyst
- Epidermoid cyst
- Velum interpositum cyst
- Pineocytoma

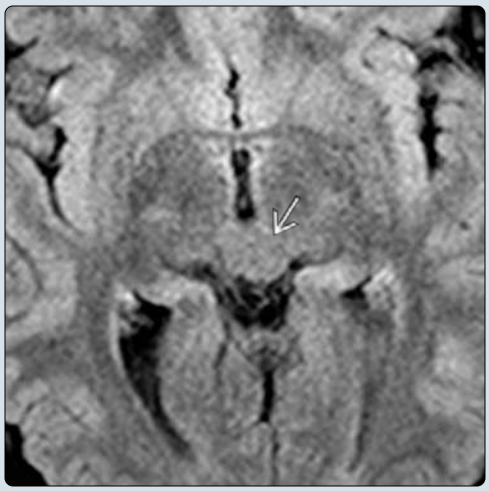
CLINICAL ISSUES

- Very common: > 50% prevalence with high resolution MR
- Occur at all ages but prevalence ↓ in older adults
- Most commonly incidental
- Large cysts (> 1 cm) may become symptomatic
  - Headache (aqueduct compression → hydrocephalus)
  - Parinaud syndrome (due to tectal plate compression)
  - In absence of hydrocephalus & visual changes, symptoms unlikely to be related to cyst

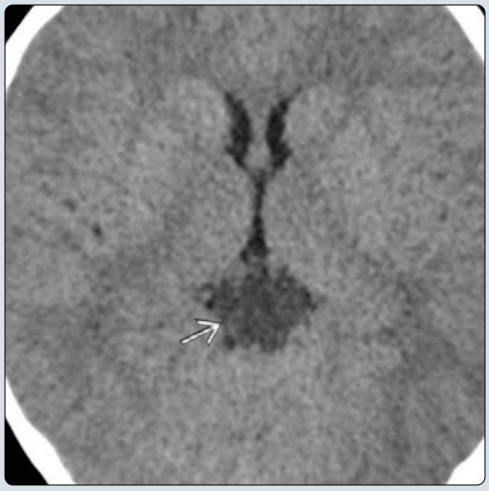
DIAGNOSTIC CHECKLIST

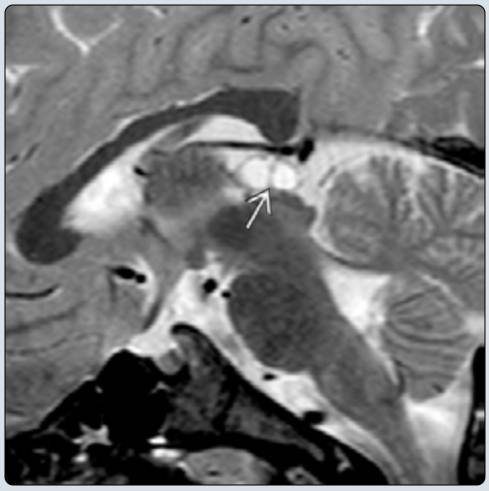
- Development of pineal gland cyst considered normal
- Imaging follow-up typically not necessary in children
  - Unless size or degree of enhancement unusual
- Pineocytoma appearance overlaps but rare in children





(Left) *Midline sagittal 3D SSFP MR in a 14 year old with vision changes shows a simple pineal cyst. The lesion is well defined, slightly hypointense to CSF, & contains no septations. There is mild mass effect on the tectum & cerebral aqueduct. The visual symptoms could reflect Parinaud syndrome, which can occur with larger pineal cysts.* **(Right)** *Axial FLAIR MR in the same 14 year old shows the cyst to be isointense to adjacent brain parenchyma. The signal intensity within pineal cysts does not typically suppress like CSF.*





(Left) *Axial NECT in a 9 year old with headaches shows a hypodense (but slightly hyperdense to CSF) lesion in the pineal region. It is difficult to clearly delineate the borders of the cyst on CT. Note the lack of ventriculomegaly.* **(Right)** *Sagittal T2 MR in a 6 year old with seizures shows a pineal cyst, either with a septation or representing 2 adjacent cysts. Septations are a common finding, seen in 25% or more of pineal cysts, & are especially evident when using high spatial resolution MR imaging.*

KEY FACTS

TERMINOLOGY

- Fluid-filled channels in subpial space that accompany penetrating arteries into brain

IMAGING

- Found in virtually all locations; conspicuity ↑ at 3T MR
  - Most commonly within inferior basal ganglia
  - Cerebral hemisphere white matter
- ↑ size & number with ↑ age
- Round/oval in basal ganglia; tubular in white matter
- "Giant" or "tumefactive" clusters of PVSs most often located in midbrain; may have associated mass effect
- **CT**: Well-defined low-attenuation lesion (~ CSF)
- **MR**: PVS contents follow CSF signal on all sequences
  - May see penetrating vessels within PVS
  - No significant adjacent parenchymal signal abnormality

TOP DIFFERENTIAL DIAGNOSES

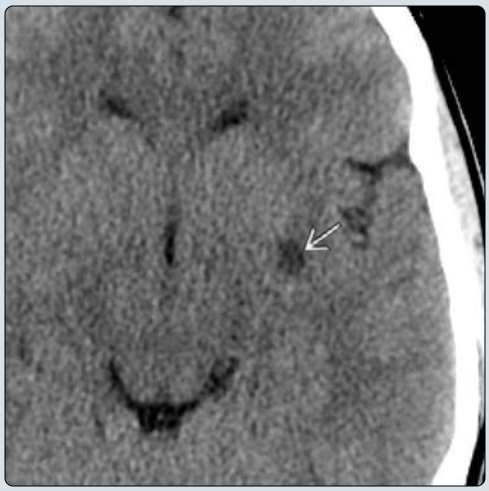
- Cystic neoplasms (e.g., DNET)
- Mucopolysaccharidoses
- Neurocysticercosis & other cyst-forming parasites
- Lacunar infarcts (typically in older patients)
- Cystic encephalomalacia

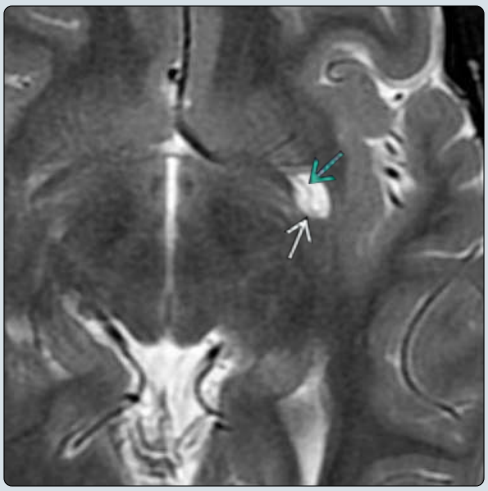
PATHOLOGY

- Actually contain interstitial fluid, not CSF
- Single or double layer of invaginated pia

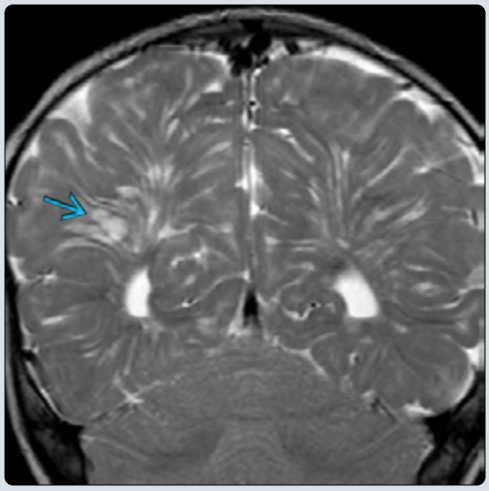
CLINICAL ISSUES

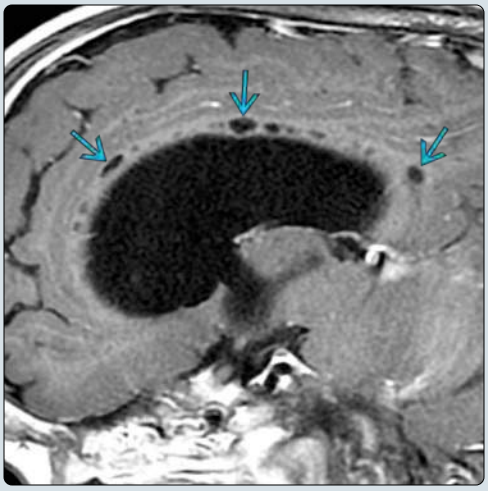
- Most common history: Incidentally identified area of low attenuation or "cystic neoplasm" on CT
- "Leave me alone" lesion should be distinguished from more serious considerations; MR ± contrast often diagnostic
  - PVSs usually stable in otherwise normal patients
- PVSs provide entry site to CNS in infectious, inflammatory, & neoplastic disease
- Recent studies in adults have shown PVSs to be imaging markers of cerebral small vessel disease





(Left) *Axial NECT in a 15-year-old girl with headache shows a focus of low attenuation in the left basal ganglia. While statistically this most likely represents an enlarged perivascular space (PVS), the finding is nonspecific on CT & warrants MR.* **(Right)** *Axial T2 MR in the same 15-year-old girl further characterizes the lesion seen on CT as an ovoid focus isointense to CSF & centered around a vessel, consistent with a PVS. The inferior basal ganglia are the most common location for PVSs.*





(Left) *Coronal T2 MR in a 1-year-old boy with Hurler syndrome shows multiple enlarged PVSs within the cerebral white matter. Most of the PVSs are tubular (as is typical in this location), but some have become more bulbous & cystic.* **(Right)** *Sagittal T1 C+ FS MR in a 2-year-old boy with Hurler syndrome shows multiple nonenhancing cystic spaces within the corpus callosum, consistent with enlarged PVSs. The PVSs tend to get larger over time in patients with mucopolysaccharidoses.*

Pilocytic Astrocytoma

KEY FACTS

TERMINOLOGY

- (Juvenile) pilocytic astrocytoma (JPA or PA)
  - WHO grade I astrocytic tumor
  - Most common primary brain tumor of children

IMAGING

- Location
  - Without neurofibromatosis type 1 (NF1): Cerebellum (midline or off-midline) > hypothalamus, brainstem, cerebral hemispheres > optic pathway (OP)
  - With NF1: OP most common
- Size: Cerebellar & cerebral lesions often > 5 cm
- > 95% enhance (patterns vary)
  - Nonenhancing cyst with enhancing mural nodule: 50%
  - Heterogeneous enhancement with smaller cysts: 40%
  - Solid, (typically) avid enhancement: 10%
- Ca^{2+} in 20%; hemorrhage uncommon
- May show little surrounding vasogenic edema
- Often cause obstructive hydrocephalus
- ↑ diffusivity typical (unlike highly cellular tumors)
- Paradoxical aggressive-appearing MR spectroscopy
  - High choline, low NAA, ± lactate

TOP DIFFERENTIAL DIAGNOSES

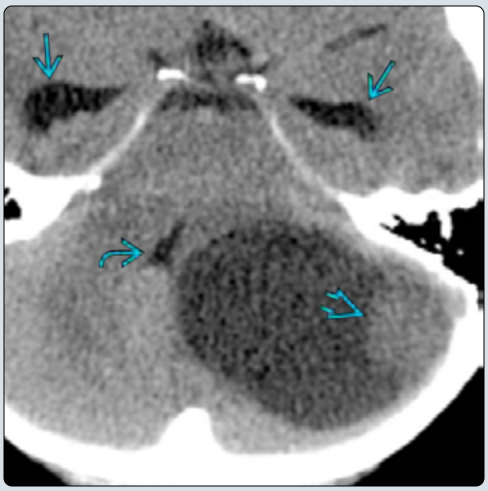
- Posterior fossa: Medulloblastoma, ependymoma
- Hypothalamus/OP: Pilomyxoid astrocytoma, optic neuritis

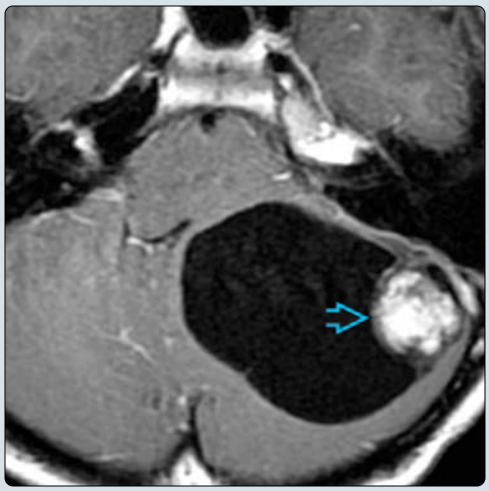
PATHOLOGY

- Sporadic > syndromic
  - 15% of NF1 patients develop PAs: OP > brainstem
    - 50-60% of patients with OP PAs have NF1

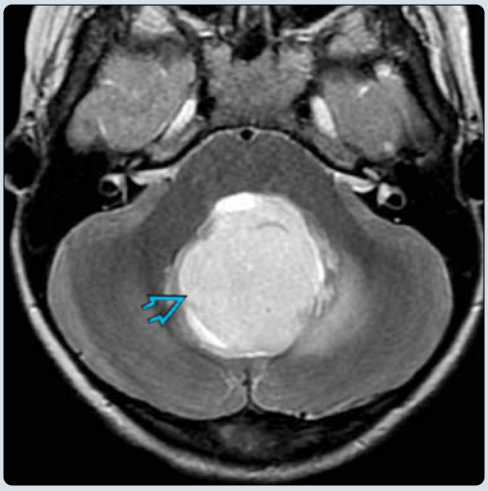
CLINICAL ISSUES

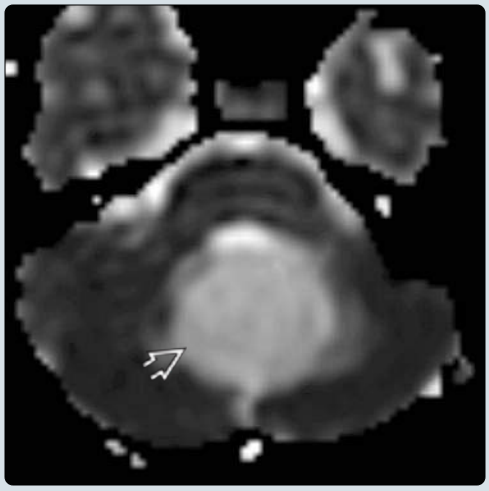
- Location determines presentation, treatment, prognosis
  - Headaches, nausea/vomiting, cerebellar signs, visual loss
  - Cerebellar & cerebral lesions cured with total resection; OP lesions (especially NF1) may be observed for growth or visual loss prior to chemotherapy or radiation
- 10-year survival > 90%





(Left) *Axial NECT in a 10-year-old boy with a pilocytic astrocytoma (PA) demonstrates a left cerebellar cystic mass with a solid mural nodule, which is isoattenuating to the adjacent white matter. Note the mass effect on the 4th ventricle with secondary dilation of the lateral ventricle temporal horns, consistent with obstructive hydrocephalus.* **(Right)** *Axial T1 C+ MR in the same patient shows a mildly heterogeneous, but overall moderate, enhancement of the solid nodule. No cyst wall enhancement is seen.*





(Left) *Axial T2WI MR in a 9-year-old girl with headaches shows a solid-appearing midline mass with increased signal intensity that is slightly lower than cerebrospinal fluid, typical for PA.* **(Right)** *Axial ADC map in the same patient shows signal intensity in the mass that is much greater than the adjacent brain parenchyma. This increased diffusivity is typical of a PA. Diffusion characteristics can be helpful in distinguishing PA from medulloblastoma, especially with midline tumors extending into the 4th ventricle.*

TERMINOLOGY

Abbreviations

- (Juvenile) pilocytic astrocytoma (JPA or PA)

IMAGING

General Features

- Location
  - In patients without neurofibromatosis type 1 (NF1): Cerebellum > hypothalamus, brainstem, cerebral hemispheres > optic pathway (OP)
  - With NF1: OP most common

CT Findings

- Mixed cystic/solid mass
  - Solid component: Iso- or hypoattenuating to white matter
  - Cystic component: Similar attenuation to cerebrospinal fluid (CSF)
- Ca^{2+} in 20%; hemorrhage rare
- Frequently cause obstructive hydrocephalus

MR Findings

- T1WI
  - Cyst contents iso- to slightly hyperintense to CSF
- T2WI
  - Solid portions hyperintense to gray matter (GM)
- FLAIR
  - Solid portions hyperintense to GM
  - Cyst contents iso- to hyperintense to nulled CSF
    - Can help determine relationship to 4th ventricle
  - May cause only mild surrounding vasogenic edema
- DWI
  - Variable, but classically bright on ADC (not DWI)
    - Increased diffusivity typical in PA
- T1WI C+
  - > 95% enhance (with exception of OP glioma)
    - 50% with large cyst & enhancing mural nodule
      - ± cyst wall enhancement
    - 40% with heterogeneous enhancement
    - 10% with solid homogeneous enhancement
  - Leptomeningeal spread of tumor very uncommon
- MRS
  - Paradoxical aggressive-appearing pattern
    - High Cho, low NAA ± lactate

DIFFERENTIAL DIAGNOSIS

Medulloblastoma

- Midline posterior fossa mass with hyperattenuation (CT) & diffusion restriction (MR)
- Younger patient age (median: 6 years)

Ependymoma

- "Plastic" tumor: Extends out 4th ventricle foramina
- Ca^{2+}, cysts, hemorrhage common

Pilomyxoid Astrocytoma

- More aggressive, less common tumor; usually suprasellar
- Hemorrhage & ↑ arterial spin labeling perfusion suggestive

PATHOLOGY

General Features

- WHO grade I, localized astrocytic neoplasm
  - Progression to higher grades uncommon
- Genetics
  - Sporadic (majority): *BRAF* gene (at chromosome 7q34) mutations most common, leading to activation of RAS/ERK/MAPK pathways
  - Syndromic: Loss of neurofibromin protein (*NF1* gene at 17q11.2) activates RAS pathway
    - 15-21% of NF1 patients develop PAs, usually of OP
      - 50-60% of OP PAs occur in NF1

Microscopic Features

- Typically biphasic histologic pattern
  - Dense fibrillar piloid with Rosenthal fibers
  - Hypofibrillar spongy tissue

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - ↑ intracranial pressure: Headaches, nausea, vomiting
  - Cerebellar lesions: Ataxia, dysdiadokinesia
  - OP lesions: Visual loss
- Clinical profile
  - Most common primary CNS tumor in children
  - Most common posterior fossa tumor in ages 5-19 years

Natural History & Prognosis

- > 90% survival at 10 years
- Poorer prognosis: Solid lesions, brainstem extension, hypothalamic location, leptomeningeal spread, anaplasia
  - Pathology still demonstrates WHO grade I tumor

Treatment

- Cerebellar or cerebral locations: Surgical
  - Prognosis related to success of total resection
    - Complete resection curative
  - Adjuvant chemotherapy or radiation only with residual progressive unresectable tumor
- OP: Often none
  - Decision to treat driven by vision loss & tumor growth
  - Radiation or chemotherapy for progressive disease

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Medulloblastoma

KEY FACTS

TERMINOLOGY

- Malignant (WHO grade IV), invasive, highly cellular embryonal tumor

IMAGING

- Round midline 4th ventricular mass
- Obstructive hydrocephalus in 95%
- CT: 90% hyperattenuating (due to ↑ cellularity)
- CT: Ca^{2+} in up to 20%; hemorrhage rare
- MR: Restricted diffusion reflects ↑ cellularity
- MRS: Taurine peak may be detected at 3.4 ppm
- Contrast and total spine MR to detect CSF spread
- 33% have subarachnoid metastatic disease at diagnosis
- 5% develop bone metastases, usually sclerotic

TOP DIFFERENTIAL DIAGNOSES

- Atypical teratoid/rhabdoid tumor (AT/RT)
- Ependymoma
- Pilocytic astrocytoma

PATHOLOGY

- 4 major molecular subgroups
  - Wnt-wingless (favorable prognosis)
  - SHH-Sonic Hedgehog (intermediate prognosis)
  - Group 3 (poor prognosis)
  - Group 4 (intermediate prognosis)
- Histology still important in treatment decisions
  - Classic, desmoplastic/nodular, and large cell/anaplastic

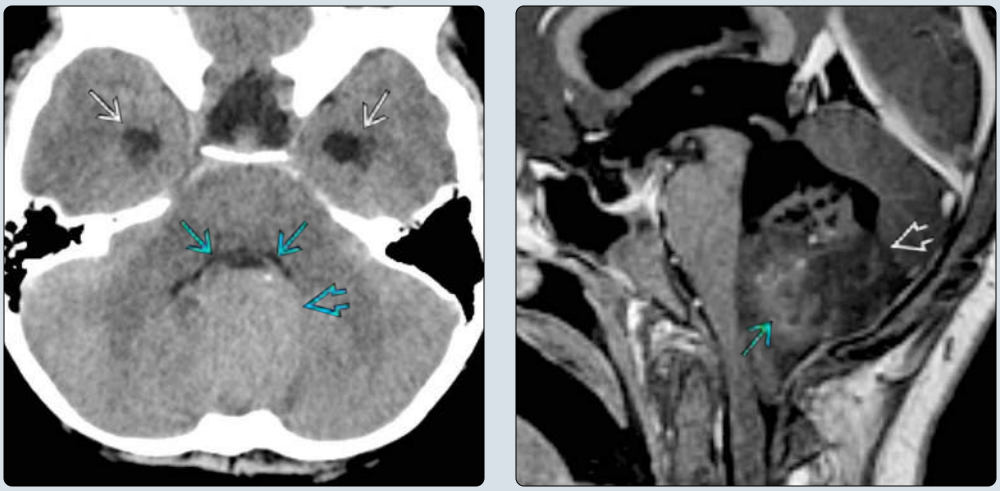
CLINICAL ISSUES

- Most common posterior fossa tumor in ages 0-4 years
  - 15-20% of all pediatric brain tumors
- Prognosis dependent on residual tumor status post resection and presence of metastatic disease

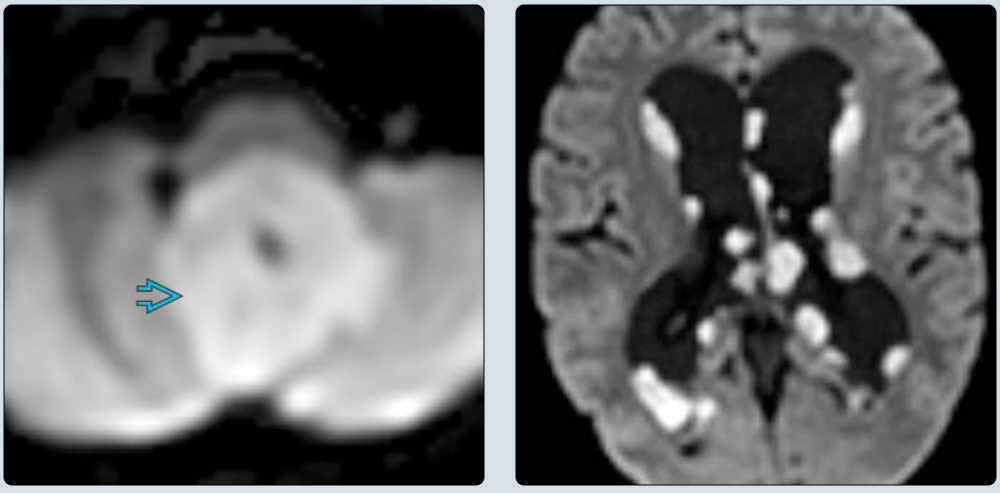
DIAGNOSTIC CHECKLIST

- 4th ventricular tumor arising from roof: Medulloblastoma
- 4th ventricular tumor arising from floor: Ependymoma
- Remember AT/RT in DDx in patients < 3 years

(Left) *Axial NECT in a 14 year old with Rubenstein-Taybi syndrome demonstrates a midline posterior fossa mass ⇨ effacing the 4th ventricle ⇨ from posterior to anterior. The mass is similar in attenuation to gray matter. The enlarged temporal horns ➡ are due to obstructive hydrocephalus.* **(Right)** *Sagittal T1 C+ in the same patient shows mild patchy enhancement of the mass ⇨. Note the indistinct interface with the posterior 4th ventricle (roof) ➡, typical of medulloblastoma.*



(Left) *Axial DWI in the same patient shows increased signal intensity of the mass ⇨. The ADC map (not shown) demonstrated decreased signal intensity, consistent with restricted diffusion and reflecting high tumor cellularity. DWI is a key discriminator of posterior fossa tumors.* **(Right)** *Axial DWI in a 13 year old with medulloblastoma status post gross total resection 3 years ago shows cerebrospinal fluid (CSF) dissemination of recurrent tumor along the ependymal lining of the lateral ventricles.*



TERMINOLOGY

Abbreviations

- Medulloblastoma (MB)

Definitions

- Malignant (WHO grade IV), invasive, highly cellular embryonal tumor

IMAGING

General Features

- Best diagnostic clue
  - Midline 4th ventricular mass in 1st decade of life
  - Hyperattenuating (CT), diffusion restricting (MR)
- Location
  - 75-90% occur in midline
    - 4th ventricular mass arising from roof (dorsal 4th ventricle)
    - Cerebellar hemisphere more frequent location in older children and adults
  - ~ 33% have subarachnoid metastatic disease at diagnosis
  - Bony metastases may occur in ~ 5%; usually sclerotic

CT Findings

- Hyperattenuation (90%) reflects high cellularity
- Ca^{2+} in up to 20%; hemorrhage rare
- Obstructive hydrocephalus in up to 95%

MR Findings

- **T1**: Hypointense to gray matter (GM)
- **T2**: Iso- to hyperintense to GM
- **FLAIR**: Hyperintense to brain
- **DWI**: Hyperintense on DWI; hypointense on ADC
  - Reflects high cellularity
- **MRS**: Taurine peak may be detected on MRS at 3.4 ppm
- **T1 C+**: Variable enhancement of primary tumor
  - Contrast improves detection of cerebrospinal fluid dissemination

Imaging Recommendations

- Best imaging tool
  - MR with DWI and postcontrast sequences
- Protocol advice
  - Sagittal images pre- and postcontrast often show site of origin (roof vs. floor of 4th ventricle)
  - Appropriate staging requires total spine imaging

DIFFERENTIAL DIAGNOSIS

Atypical Teratoid/Rhabdoid Tumor

- No differentiating imaging features; often more heterogeneous than MB
- Younger children (usually < 3 years of age)

Ependymoma

- Punctate Ca^{2} and hemorrhage more common than MB
- Extension through 4th ventricle foramina: "Plastic tumor"

Pilocytic Astrocytoma

- Cerebellar hemispheric lesion; often cystic
- Solid portion: ↑ ADC signal (↑ diffusivity, not ↓)

Choroid Plexus Papilloma

- 4th ventricle location less common
- Vigorous enhancement typical
- No diffusion restriction unless higher grade (carcinoma)

PATHOLOGY

General Features

- Most common posterior fossa tumor in ages 0-4 years
- Associated with many familial cancer syndromes

Staging, Grading, & Classification

- Molecular subgroups increasingly important
  - Wnt-wingless (least common): Favorable prognosis
    - Often located in cerebellopontine angle
  - SHH-Sonic hedgehog: Intermediate prognosis
    - 50% located in the cerebellar hemisphere
  - Group 3: Poor prognosis
    - *MYC* amplification common
    - Located in midline/4th ventricle
  - Group 4 (most common): Intermediate prognosis
    - i17q mutation common
    - Located in midline/4th ventricle

Microscopic Features

- Densely packed hyperchromatic cells with scant cytoplasm
- Histologic subtypes (still important in treatment decisions)
  - Classic (~ 70%)
  - Nodular/desmoplastic (~ 20%)
  - Anaplastic/large cell (~ 10%)

CLINICAL ISSUES

Presentation

- Ataxia, signs of ↑ intracranial pressure

Demographics

- M > F = 2-4:1
- 75% < 10 years of age

Treatment

- Surgical excision, adjuvant chemotherapy
- 5-year survival rate
  - No metastases or gross residual tumor status post resection: 60-100%
  - Presence of gross residual tumor after surgery or metastatic disease: 20%

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Atypical Teratoid/Rhabdoid Tumor

KEY FACTS

TERMINOLOGY

- Highly malignant (WHO grade IV) embryonal neoplasm

IMAGING

- Heterogeneous intracranial mass in infant/young child
  - Commonly contains cysts or hemorrhage
  - Solid components show ↑ attenuation on CT & restricted diffusion on MR due to high cellularity
- 47% infratentorial, 41% supratentorial, 12% both
- Often causes obstructive hydrocephalus
- Leptomeningeal spread common (15-20%)

TOP DIFFERENTIAL DIAGNOSES

- Medulloblastoma (MB)
- Supratentorial primitive neuroectodermal tumor (PNET)
- Ependymoma
- Desmoplastic infantile tumor
- Teratoma
- Choroid plexus tumor

PATHOLOGY

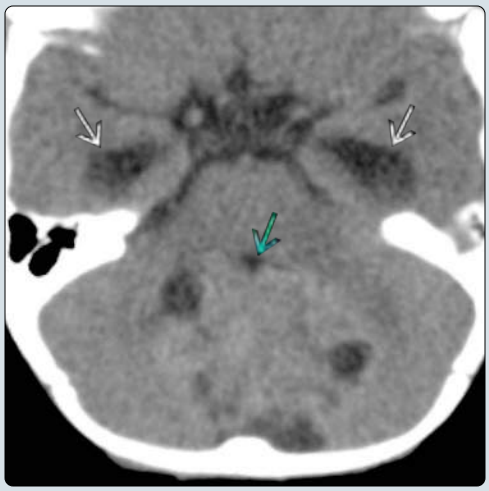
- Morphologic & immunophenotypic heterogeneity
- Divergent differentiation accounts for teratoid label
  - Sheets of primitive cells separated by fibrovascular septa
  - Large rhabdoid cells with eosinophilic cytoplasm
- Most rhabdoid tumors show lack of INI1 immunostain due to *SMARCB1* (hSNF5/INI1) mutation (22q11.2)
- Histology similar to renal & soft tissue rhabdoid tumors
  - May be concurrent with CNS AT/RT

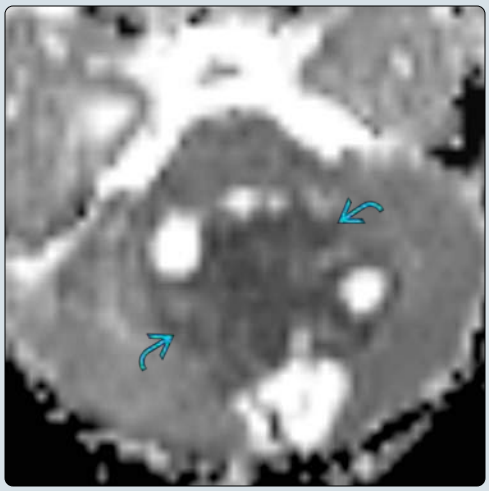
CLINICAL ISSUES

- Child < 3 yr old with increasing head size, vomiting
- Median survival: 12-18 months; overall survival rate: 23-37%

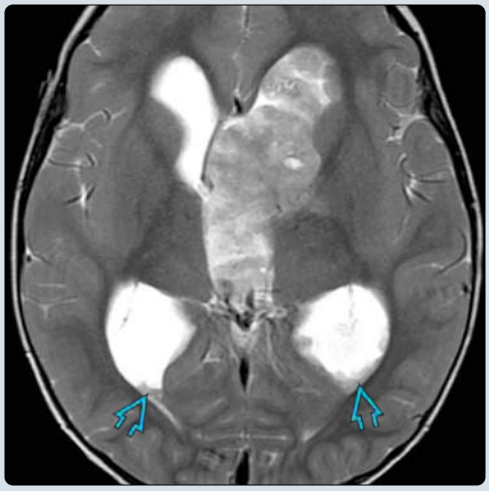
DIAGNOSTIC CHECKLIST

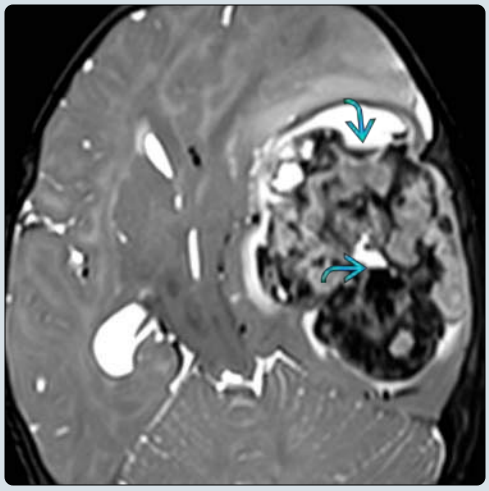
- Always consider AT/RT in young child (< 3 yr old) if
  - Large heterogeneous CNS tumor
  - MB or PNET being considered
  - Infant with new cranial nerve palsy + cisternal mass





(Left) *Axial NECT in a 6-month-old girl with atypical teratoid/rhabdoid tumor (AT/RT) shows a mixed cystic & solid mass centered in the vermis with effacement of the 4th ventricle & obstructive hydrocephalus. Note the mildly hyperattenuating solid component, reflecting the high cellularity of the tumor.* **(Right)** *Axial ADC map MR in the same patient demonstrates decreased signal intensity in the solid components of the tumor, consistent with restricted diffusion & reflecting the high cellularity of the tumor.*





(Left) *Axial T2 MR in a 2-year-old boy demonstrates a large intraventricular AT/RT with nodular masses along the ependyma of both lateral ventricles, consistent with CSF spread of disease.* **(Right)** *Axial T2 MR in an 11-month-old boy demonstrates a large heterogeneous mass centered in the left frontotemporal region. The fluid levels suggest prior intratumoral hemorrhage. AT/RT should be considered for any large hemorrhagic CNS mass in an infant or young child.*

TERMINOLOGY

Definitions

- Atypical teratoid/rhabdoid tumor (AT/RT): Malignant embryonal tumor of CNS in early childhood
  - Closely related to renal & soft tissue rhabdoid tumors

IMAGING

General Features

- Best diagnostic clue
  - Large solid & cystic tumor (± hemorrhage) with diffusion restriction in infant/young child
- Location
  - Infratentorial (47%)
  - Supratentorial (41%)
  - Both infra- & supratentorial (12%)
  - 15-20% have disseminated tumor at initial diagnosis

CT Findings

- NECT
  - Hyperattenuating mass
  - Commonly contains cysts &/or hemorrhage
  - May contain Ca^{2+}
  - Obstructive hydrocephalus common

MR Findings

- **T1WI**: Heterogeneous, ± hyperintense hemorrhage
- **T2WI**: Heterogeneous with hyperintense cystic foci
- **FLAIR**: Hyperintense cysts, ± peritumoral edema
- **T2* GRE/SWI**: Hemorrhagic foci causing exaggerated signal loss (blooming)
- **DWI**: Positive diffusion restriction (↓ ADC signal) due to high cellularity
- **T1WI C+**: Heterogeneous but avid enhancement

Imaging Recommendations

- Best imaging tool: MR of brain & spine with contrast
  - Leptomeningeal spread common (15-20%)

DIFFERENTIAL DIAGNOSIS

Medulloblastoma

- AT/RT more likely to have cysts than medulloblastoma (MB)

Primitive Neuroectodermal Tumor

- Cellular supratentorial tumor with diffusion restriction

Ependymoma

- Infratentorial tumor often extending out 4th ventricular foramina
- Supratentorial tumor often large & cystic

Desmoplastic Infantile Tumor

- Very young children (peak age: 3-6 months)
- Very large cystic components

Teratoma

- More often pineal or parasellar in location
- Heterogeneous due to Ca^{2+}, hemorrhage

Choroid Plexus Tumors

- Intraventricular mass
- Vigorous, homogeneous enhancement

PATHOLOGY

General Features

- Genetics
  - Loss of tumor suppressor gene *SMARCB1* (hSNF5/INI1) in most rhabdoid tumors, including CNS, renal, extrarenal
  - Germline mutation tumors present earlier than somatic
    - Rhabdoid tumor predisposition syndrome
      - Synchronous renal & CNS or bilateral renal tumors

Staging, Grading, & Classification

- WHO grade IV

Microscopic Features

- Hypercellular with morphologic & immunophenotypic heterogeneity
  - Divergent differentiation accounts for teratoid label: Neuroectodermal, mesenchymal, epithelial, & rhabdoid
    - Rhabdoid cells large with eosinophilic cytoplasm
- Lack of immunostaining for INI1 protein correlates with *SMARCB1* (hSNF5/INI1) mutation
  - Most discriminating finding for diagnosing AT/RT

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Signs of ↑ intracranial pressure
    - Lethargy, vomiting, ↑ head circumference
  - Other signs/symptoms: Torticollis, seizure, developmental regression, new cranial nerve palsy
- Clinical profile
  - Child < 3 yr (70-80%); median age: 1 yr

Natural History & Prognosis

- Median survival: 12-18 months
- Overall survival rate: 23-37%

Treatment

- Gross total resection alone: Early recurrence
- Intensive multimodal therapy has ↑ survival but prognosis remains poor
  - Maximal resection with high-dose systemic & intrathecal chemotherapy + radiation

DIAGNOSTIC CHECKLIST

Consider

- AT/RT in young children (usually < 3 yr of age) with tumor otherwise suggestive of primitive neuroectodermal tumor (PNET) or MB by imaging

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Ependymoma

KEY FACTS

TERMINOLOGY

- Slow-growing tumor resembling ependymal cells

IMAGING

- 1/3 supratentorial: Periventricular white matter
- 2/3 infratentorial: 4th ventricle/cisterns
- Soft or plastic tumor: Conforms to ventricle & extends through foramina into cisterns
- Variable heterogeneous enhancement
- Requires combination of imaging & clinical findings to distinguish from medulloblastoma (MB)

TOP DIFFERENTIAL DIAGNOSES

- Infratentorial
  - MB
  - Pilocytic astrocytoma
  - Choroid plexus papilloma
- Supratentorial
  - Numerous glial, neuronal, & mixed neoplasms

PATHOLOGY

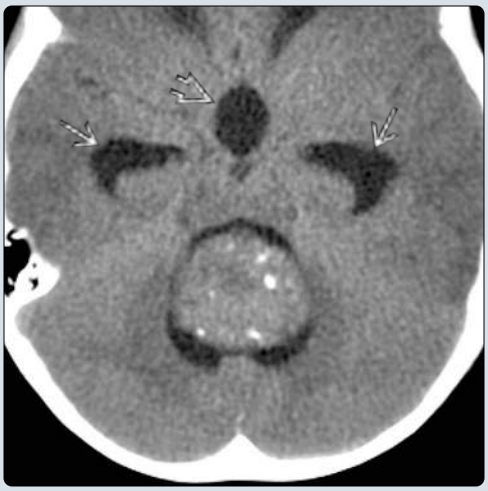
- Classic ependymoma: WHO grade II
- Supratentorial, posterior fossa, & spinal ependymomas histologically similar but genetically distinct
- 2 molecular subgroups of posterior fossa ependymoma
  - Group A: Younger children with poorer prognosis
    - More likely to have lateral location in posterior fossa
  - Group B: Often older children with better prognosis
    - More commonly confined to midline 4th ventricle

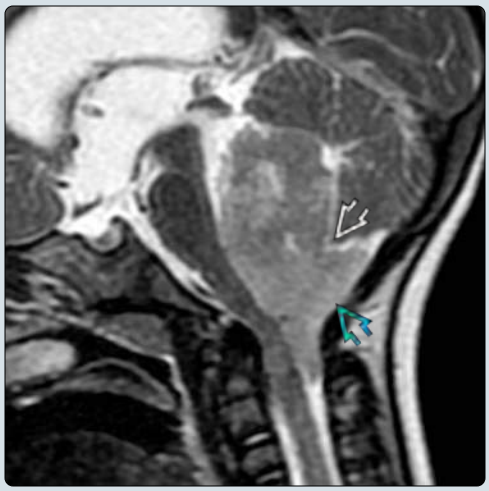
CLINICAL ISSUES

- 15% of posterior fossa tumors in children
- Peaks at 1-5 yr of age, but many cases in adolescents
- Overall 5-yr survival for brain lesions: 50-70%

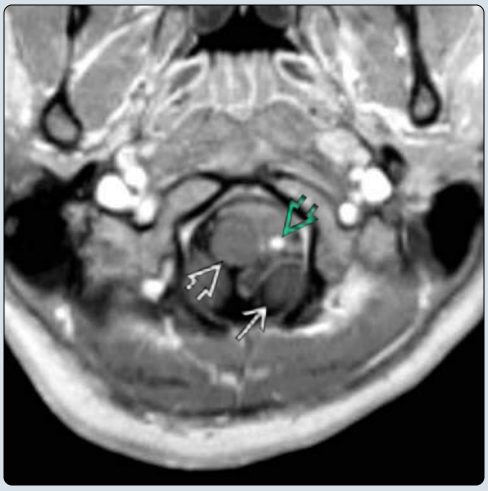
DIAGNOSTIC CHECKLIST

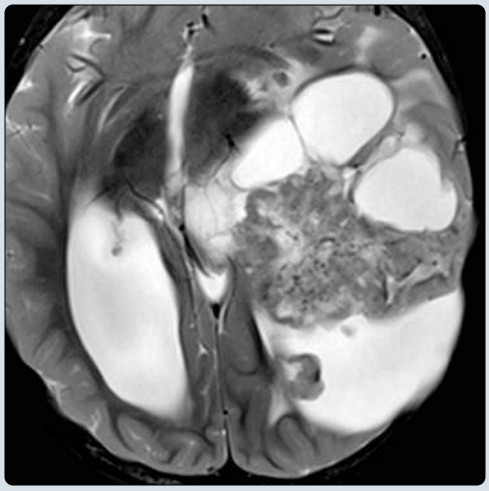
- Indistinct interface with 4th ventricular floor: Ependymoma
- Indistinct interface with 4th ventricular roof: MB





(Left) *Axial NECT in a 2-year-old girl shows a mass in the 4th ventricle containing scattered small Ca^{2+}. Note the enlargement of the temporal horns & 3rd ventricle, consistent with obstructive hydrocephalus.* **(Right)** *Midline sagittal T2 MR in the same patient demonstrates the mass expanding the 4th ventricle (displacing the cerebellum & brainstem) & extending through the foramen of Magendie into the cisterna magna.*





(Left) *Axial T1 GRE C+ MR in a 17-year-old girl demonstrates a lesion in the left cerebellomedullary cistern. The mass encases the enhancing left vertebral artery without narrowing it, further demonstrating its plastic nature as it extends between the left cerebellar tonsil & brainstem.* **(Right)** *Axial T2 MR in a 2-year-old patient demonstrates a large mixed cystic & solid mass. The large size, heterogeneous solid component, & cysts are typical of a supratentorial ependymoma.*

TERMINOLOGY

Definitions

- Slow-growing tumor resembling ependymal cells
- Other ependymal tumors genetically distinct; share some histologic features
  - Subependymoma, myxopapillary ependymoma, anaplastic ependymoma

IMAGING

General Features

- Best diagnostic clue
  - Plastic midline 4th ventricular tumor
    - Conforms to ventricle & extends through 4th ventricular outlets into cisterns
  - Indistinct interface with floor of 4th ventricle (brainstem)
  - Ca^{2+} in 50%; ± cysts, hemorrhage
- Location
  - ~ 2/3 infratentorial: 4th ventricle
  - ~ 1/3 supratentorial: Most at periventricular white matter

CT Findings

- Infratentorial: Hydrocephalus common
- Supratentorial: Typically large heterogeneous mass

MR Findings

- T1WI: Heterogeneous; hyperintense Ca^{2+} & hemorrhage
- T2WI: Hyperintense cystic foci; hypointense Ca^{2+} & hemorrhage
- FLAIR: Tumor cysts hyperintense to CSF
- DWI: Iso- to hypointense ADC compared to gray matter
  - Diffusion restriction depends on tumor grade
- GRE/SWI: Hypointense blooming of Ca^{2+} & blood products
- T1C+: Heterogeneous but variable enhancement
- MRS: ↓ NAA; ↑ Cho, lipid/lactate peaks

Imaging Recommendations

- Best imaging tool
  - MR of brain & spine with contrast
    - Spine imaging necessary for complete staging
- Protocol advice
  - High-resolution SSFP MR can visualize cranial nerve involvement by tumor extending into cisterns
  - MR angiography can delineate involvement of posterior fossa vessels by tumor extending into cisterns

DIFFERENTIAL DIAGNOSIS

Infratentorial

- Medulloblastoma
- Pilocytic astrocytoma
- Brainstem glioma
- Choroid plexus tumors

Supratentorial

- Pilocytic astrocytoma
- Oligodendroglioma
- Glioblastoma multiforme
- Astroblastoma

PATHOLOGY

General Features

- Etiology
  - Recent studies suggest radial glial cells as cells of origin
- Genetics
  - Supratentorial, infratentorial, & spinal ependymomas genetically distinct
  - Infratentorial tumors divided into 2 molecular subgroups
    - Group A: Common in younger children, poorer prognosis
      - Lateral posterior fossa location typical
    - Group B: Common in older children, better prognosis
      - Midline 4th ventricle or spinal cord typical

Staging, Grading, & Classification

- Standard ependymoma: WHO grade II
- Other ependymal tumors
  - Anaplastic ependymoma: WHO grade III
  - Subependymoma: WHO grade I
  - Myxopapillary: WHO grade I

Microscopic Features

- Moderately cellular with low mitotic activity, occasional nuclear atypia
- Perivascular pseudorosettes
- Immunohistochemistry: Positive for S100, glial fibrillary acidic protein, vimentin

CLINICAL ISSUES

Demographics

- Epidemiology
  - 15% of posterior fossa tumors in children
  - 3rd most common posterior fossa tumor in children

Natural History & Prognosis

- 3-17% have CSF dissemination
- Overall 5-yr survival for brain ependymomas: 50-70%
- Recurrence may be local or leptomeningeal
  - 5-yr survival after recurrence: 15%

Treatment

- Surgical resection ± chemo, radiation therapy
  - Extent of resection strongly correlates with outcome

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Indistinct interface with 4th ventricular floor = ependymoma
- Indistinct interface with 4th ventricular roof = medulloblastoma

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KEY FACTS

TERMINOLOGY

- Brainstem tumors (BST) distinguished by location, imaging appearance, & histology

IMAGING

- Diffuse intrinsic pontine glioma (DIPG): Expansile tumor centered in pons, effacing CSF cisterns & 4th ventricle; often encases basilar artery
  - Fibrillary tumors: Little or no enhancement
  - GBM: Focal enhancement with central necrosis
  - Most do not restrict diffusion
- PA: Exophytic, enhancing tumor anywhere in brainstem
- Tectal plate glioma: Nonenhancing mass in tectum
- Midbrain tumors: Heterogeneous group of tumors
- Best imaging study: MR with contrast
  - Advanced techniques ± helpful in DIPG prognosis
    - ↓ ADC, ↑ rCBV, ↑ Cho:NAA suggest ↓ survival

TOP DIFFERENTIAL DIAGNOSES

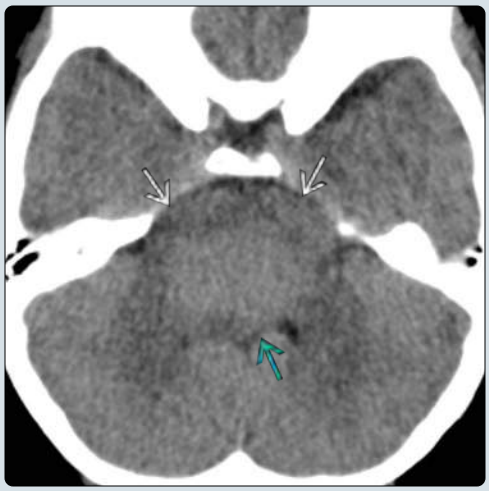
- Brainstem abscess, acute disseminated encephalomyelitis, neurofibromatosis type 1, osmotic demyelination syndrome, cavernous malformation

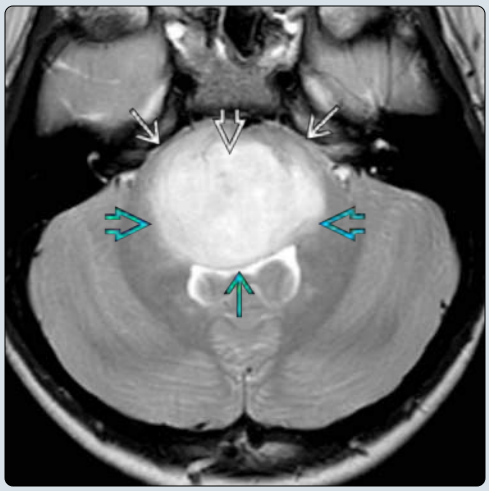
PATHOLOGY

- BST ~ 15% of pediatric brain tumors, 20-30% of pediatric posterior fossa tumors
  - Astrocytomas (WHO I-IV), PNETs, rare gangliogliomas
- DIPG diagnosis often made solely on imaging

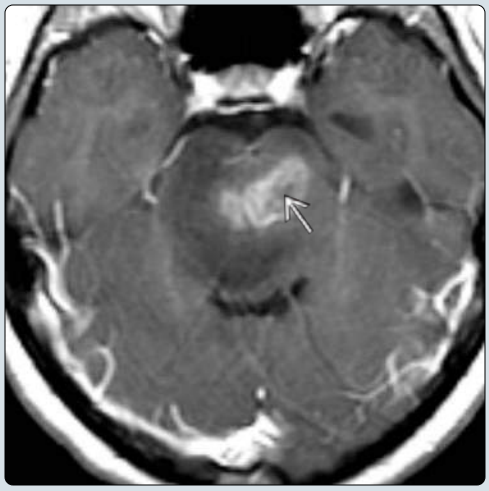
CLINICAL ISSUES

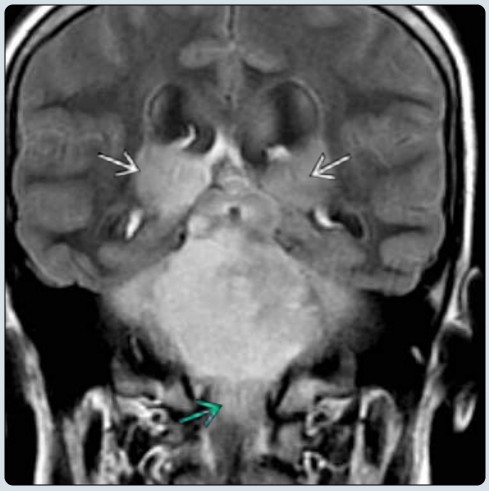
- Presentations include cranial nerve palsies, hemiparesis, gait disturbance, ataxia, headache, nausea, vomiting
- Prognosis depends on location & histology
  - Pons: Poor prognosis (unresectable)
    - DIPG: Median survival ~ 1 year
  - Medulla or midbrain: Variable prognosis
  - Tectum: Good prognosis (only requires CSF diversion)





(Left) *Axial NECT in a 7-year-old girl shows enlargement of the pons with obliteration of the prepontine cisterns & near complete effacement of the 4th ventricle. Mass effect on the cisterns & 4th ventricle may be the only clue to a diffuse intrinsic pontine glioma (DIPG) on CT.* **(Right)** *Axial T2 MR in the same patient shows a homogeneously hyperintense mass expanding the pons. The mass shows indistinct margins with mass effect on the cisterns & 4th ventricle, typical of DIPG.*





(Left) *Axial T1 C+ MR in a 9-year-old girl with DIPG demonstrates focal areas of enhancement with central nonenhancement, suggesting necrosis. Enhancement has been shown to be associated with shorter survival time in DIPG.* **(Right)** *Coronal FLAIR MR in a 9-year-old girl with DIPG shows extension of abnormal signal into the bilateral thalami & medulla. Such distant extension of signal abnormality typically suggests an infiltrative, higher grade tumor.*

TERMINOLOGY

Abbreviations

- Brainstem tumors (BST), diffuse intrinsic pontine glioma (DIPG)

Synonyms

- Pontine glioma, midbrain glioma, medullary glioma, dorsally exophytic medullary glioma

Definitions

- BST distinguished by location & imaging/histologic characteristics of tumor
  - DIPG: Glial tumor centered in pons (WHO grades II-IV)
    - Expansile, often poorly marginated, often lack enhancement
    - Glioblastoma multiforme (GBM) shows focal enhancement & central necrosis
  - Tectal gliomas
    - Present with hydrocephalus in 6-10 year olds
    - Rarely progressive
    - CSF diversion often only treatment required
  - Midbrain or mesencephalic tumors
    - All histologies occur (including PNET, atypical teratoid rhabdoid tumor)
  - Medullary tumors
    - PA most likely histology
    - Usually dorsally exophytic & solidly enhancing
  - BST associated with neurofibromatosis type 1 (NF1) → another distinct group
    - Rarely enlarge, often asymptomatic

IMAGING

General Features

- Best diagnostic clue
  - DIPG: Expansile lesion in pons
  - PA: Exophytic enhancing lesion anywhere in brainstem
  - Tectal plate glioma: Nonenhancing lesion in tectum
- Location
  - Cervicomedullary junction to cerebral peduncles
    - Medullary, pontine, midbrain, tectal
  - DIPG: By definition, must be centered in pons
- Size
  - Varies greatly, partly related to location
    - DIPG often very large at presentation
    - Tectal plate gliomas often small
- Morphology
  - Depends on histology; focal vs. large/infiltrative
    - Sometimes exophytic
    - DIPG: Signal extension into cerebellar peduncles, midbrain, or medulla → higher grade

CT Findings

- NECT
  - Brainstem enlargement with ↓ attenuation & effacement of adjacent CSF cisterns
    - Streak artifact from skull base can mimic tumor
  - Pontine tumors → flattening of anterior 4th ventricle

MR Findings

- T1WI
  - Mild to moderate hypointensity
  - Central areas of preserved signal may represent relatively preserved white matter (WM) tracts
- T2WI
  - Hyperintense mass (homogeneous to heterogeneous) & margins (edema vs. infiltrating tumor)
    - Treatment often results in ↑ heterogeneity
  - Basilar artery flow void engulfed by DIPG
- FLAIR
  - Hyperintense mass, ± better defined than on T2
- T2* GRE
  - Areas of signal loss typically represent intratumoral hemorrhage
    - Mineralization uncommon in BST
- DWI
  - DIPG typically does not show diffusion restriction; ADC signal usually greater than brain
    - ↓ ADC values associated with shorter survival time & higher grade tumor
  - Diffusion tractography (DTI) can show displacement of WM tracts by tumor
    - WM tracts typically infiltrated or displaced by DIPG (but not interrupted)
- PWI
  - ↑ relative cerebral blood volume (rCBV) associated with ↓ DIPG survival time
- T1WI C+
  - Fibrillary tumors: Variable enhancement, usually none or minimal
  - GBM: Often has focal areas of enhancement with central necrosis
  - PA: Solid portion enhances
    - ± solid, cystic with nodule, or rim enhancing; most often located dorsally with exophytic component
  - Enhancement change during DIPG treatment of unclear etiology
    - Altered enhancement during therapy may reflect steroid effect on blood-brain barrier, not necessarily change in tumor
  - Metastatic disease more common than initially reported
    - Especially late in disease or post bevacizumab therapy
- MRA
  - Basilar artery engulfed by DIPG but not typically narrowed
- MRS
  - ↑ Cho:NAA ratio associated with shorter survival
  - Presence of lactate implies necrosis & poor prognosis in some studies

Nuclear Medicine Findings

- PET-CT: FDG uptake in ≥ 50% of tumor area → shorter survival
  - Hypermetabolic tumors more likely to be GBM

Imaging Recommendations

- Best imaging tool
  - MR with contrast
- Protocol advice
  - DTI, perfusion imaging, & MRS have prognostic significance in DIPG & may help distinguish high vs. low grade tumors

DIFFERENTIAL DIAGNOSIS

Brainstem Abscess

- Listeria monocytogenes often implicated
  - Viral agents → West Nile virus, adenovirus, EBV, HSV
- More acute clinical course; often febrile

Acute Disseminated Encephalomyelitis/Other Autoimmune Inflammation

- Supratentorial & spinal sites often affected as well
- Delayed onset after viral prodrome or vaccination
- Behçet disease causes marked midbrain edema/swelling

Neurofibromatosis Type 1

- Asymptomatic, poorly defined T2 hyperintense foci in brainstem, cerebellum, globus pallidus
  - Develop in early childhood, diminish with age
- Cerebellar WM involvement more common than pons
  - Look for additional findings of NF1

Osmotic Demyelination Syndrome

- Central pontine T2 hyperintensity with sparing of periphery
- Classic clinical setting: Rapid correction of hyponatremia

Cavernous Malformation

- Low flow vascular malformation with locules containing fluid-fluid levels
- SWI/T2* GRE hypointensity classic
- Often associated with DVA

Histiocytosis

- Langerhans cell histiocytosis, hemophagocytic lymphohistiocytosis
- May cause signal abnormalities in pons & cerebellum
- Often associated with other sites of disease

PATHOLOGY

General Features

- Genetics
  - Mutations of *TP53* tumor suppressor gene (42-71%)
  - Histone-H3 mutations carry worse prognosis, regardless of histologic grade
- Associated abnormalities
  - Better prognosis of BST when associated with NF1
    - Medulla most common site in NF1

Staging, Grading, & Classification

- DIPG: Astrocytomas of varying grade (WHO II-IV)
  - Diagnosis often made solely on imaging without biopsy
    - ↑ molecular therapy targets will likely ↑ biopsy rates
- Pilocytic astrocytomas, PNETs, rare gangliogliomas
  - If suspected based on imaging, recommend biopsy

Gross Pathologic & Surgical Features

- Pontine swelling in DIPG
  - Diffuse tumor infiltration; craniocaudal extension along fiber tracts

Microscopic Features

- Variable cellularity & mitoses with pleomorphism & nuclear atypia, necrosis, & endothelial proliferation

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Cranial nerve palsies with long tract signs
  - Hemiparesis, ataxia, gait disturbance
  - Headache, nausea, vomiting

Demographics

- Age
  - Peak incidence ~ 3-10 years old
- Gender
  - M = F
- Epidemiology
  - ~ 15% of pediatric brain tumors
  - 20-30% of pediatric posterior fossa tumors

Natural History & Prognosis

- DIPG: Very poor prognosis
  - CSF dissemination in 50% prior to death
  - Median survival ~ 1 year
  - 20% survival at 2 years
- PA (dorsally exophytic): Fair to good prognosis, especially in setting of NF1
- Tectal plate gliomas: Good prognosis

Treatment

- DIPG: Radiation therapy if > 3 years old
  - Chemotherapy, though not particularly effective
- PA: Consider surgical resection based upon location
- Tectal plate glioma: Often only requires 3rd ventriculostomy followed by observation

DIAGNOSTIC CHECKLIST

Consider

- Rapid onset of symptoms: Abscess
- Blood products: Cavernous malformation (look for associated DVA)
- Lesions in other locations: Infection or demyelination
- Atypical appearance → consider biopsy

Image Interpretation Pearls

- Sagittal T2/FLAIR helpful to show extent of lesion
- Hydrocephalus usually late finding

Reporting Tips

- Pontine tumor signal extending to cerebellum, midbrain, or medulla suggests higher grade
- Solid exophytic enhancement most common in PA; may suggest better prognosis

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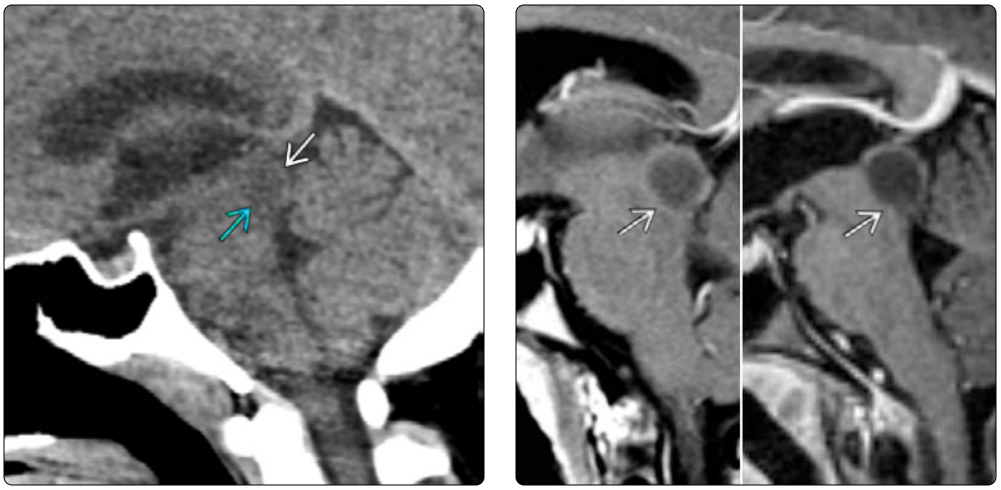
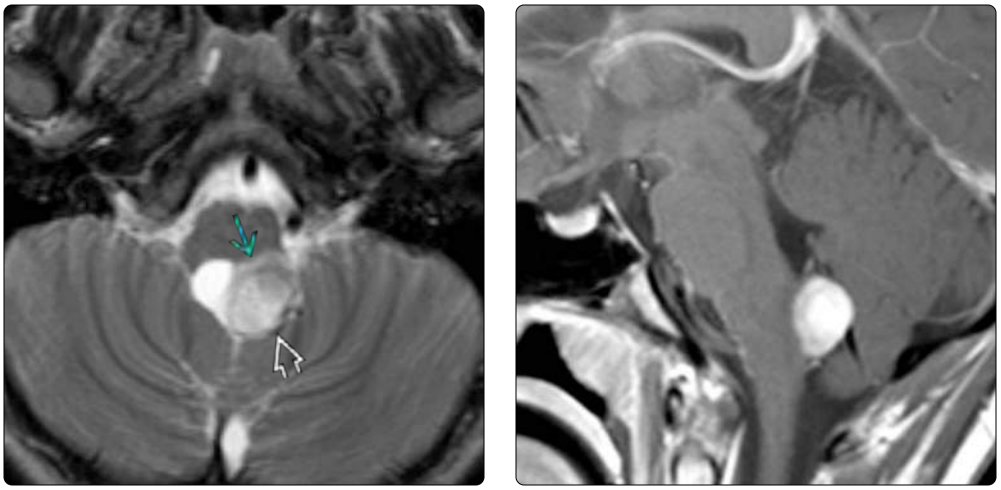
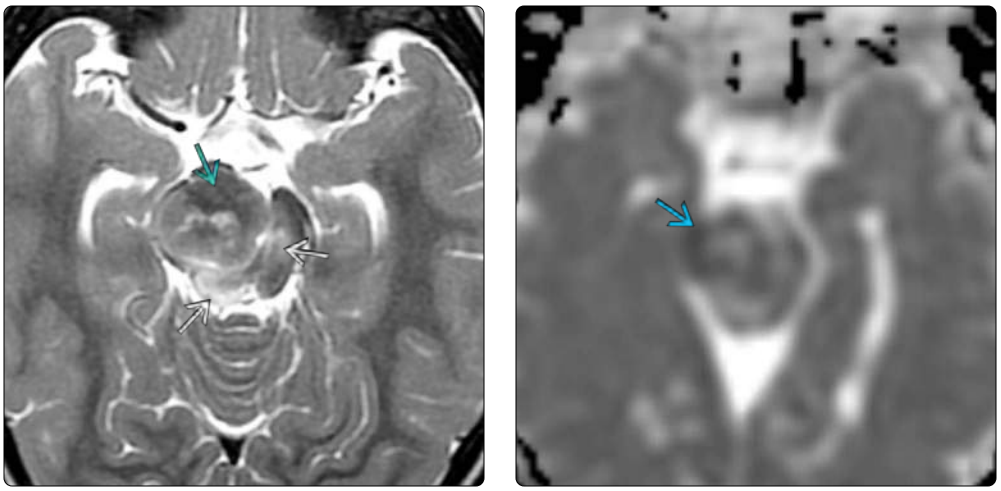
4. Jansen MH et al: Survival prediction model of children with diffuse intrinsic pontine glioma based on clinical and radiological criteria. Neuro Oncol. 17(1):160-6, 2015



(Left) *Midline sagittal NECT in a 16-year-old girl involved in a motor vehicle collision shows a low-attenuation expansile mass centered in the tectum with obliteration of the cerebral aqueduct.* **(Right)** *Midline sagittal T1 C+ MRs of the same patient at diagnosis (left) & 16 months later (right) show a nonenhancing expansile mass centered in the tectum. The lack of enhancement & lack of interval growth (or very slow growth over time) is typical for a tectal plate glioma.*



(Left) *Axial T2 MR in an 11 year old with a heterogeneously hyperintense mass at the inferior 4th ventricle shows an indistinct border of the mass with the medulla but a distinct border with the cerebellum, suggesting an exophytic origin from the dorsal medulla.* **(Right)** *Sagittal T1 C+ MR in the same patient shows homogeneous enhancement of the mass arising from the dorsal medulla, a typical pattern of enhancement for a pilocytic astrocytoma (confirmed histologically).*



(Left) *Axial T2 MR in a 2 year old with abnormal gait shows an exophytic, well-defined mass arising from the midbrain with relatively low signal intensity (~ white matter). The low T2 signal suggests high cellularity. Note the adjacent vasogenic edema in the left midbrain & right tectum.* **(Right)** *Axial ADC map MR in the same patient shows predominantly low signal (< brain parenchyma) within the midbrain mass, consistent with high cellularity. Pathology showed an atypical teratoid rhabdoid tumor.*

DNET

KEY FACTS

TERMINOLOGY

- Dysembryoplastic neuroepithelial tumor (DNET)
  - Benign mixed glial-neuronal neoplasm
  - Adjacent cortical dysplasia frequently present

IMAGING

- May occur in any region of supratentorial cortex
  - Medial temporal lobe most common
- Typically sharply demarcated, wedge-shaped
  - Often cystic ("bubbly")
  - Minimal or no mass effect
  - No peritumoral edema
- Some DNETs have less distinct margins
  - Associated cortical dysplasia more likely in these cases
- Faint focal punctate or ring enhancement in 20-30%
- Ca^{2+} in 6-36%
- Slow or no growth over years
- Remodel overlying bone in 20-44%

TOP DIFFERENTIAL DIAGNOSES

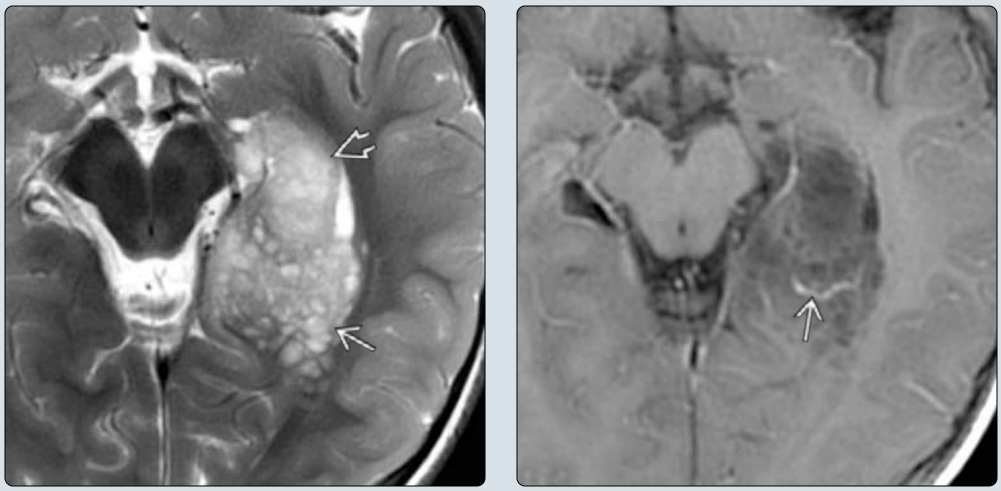
- Ganglioglioma
- Focal cortical dysplasia
- Neuroepithelial cyst
- Pleomorphic xanthoastrocytoma
- Angiocentric glioma

PATHOLOGY

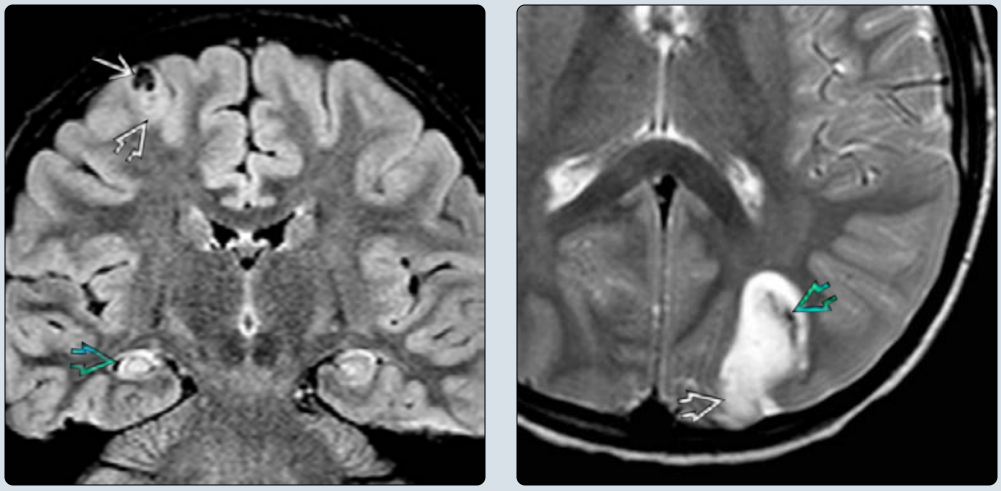
- WHO grade I
- Pathologic hallmark: Specific glioneuronal element
- Histologic subtypes: Simple, complex, and nonspecific
  - Complex: Associated cortical dysplasia present

CLINICAL ISSUES

- Typical history: Child or young adult with seizures
- Gross total resection (including adjacent dysplasia) usually curative and most important factor for seizure freedom
- Histology usually remains benign even with enhancing tumor recurrence



(Left) *Axial T2 MR in a 5-year-old boy with new onset of seizures shows a T2-hyperintense mass with multiple cystic spaces. The mass completely replaces and mildly enlarges the left hippocampus. The history, location, and cystic morphology are all typical for DNET.* **(Right)** *Axial T1 C+ MR in the same patient shows no tumor enhancement. The linear enhancement in the posterior tumor is due to an encased blood vessel. A minority (20-30%) of DNETs will demonstrate nodular or patchy contrast enhancement.*



(Left) *Coronal oblique FLAIR MR in a 12-year-old girl with intractable epilepsy for 2 years shows a small DNET with complete signal suppression of the cystic component. The noncystic component shows subtle hyperintensity. Note the associated right hippocampal sclerosis.* **(Right)** *Axial T2 MR in a 10-year-old boy with an incidental finding on a trauma CT shows a cortically based DNET that is of predominantly high T2 signal. The mass contains a low T2 signal focus (due to Ca^{2+} on the CT).*

KEY FACTS

TERMINOLOGY

- Well-differentiated, slowly growing neuroepithelial tumor composed of neoplastic ganglion & glial cells

IMAGING

- Best imaging clue: Partially cystic, partially enhancing, cortically based mass in child/young adult with temporal lobe epilepsy
- Supratentorial: 85-90%; infratentorial: 10-15%
- Most commonly involves cerebral cortex, especially temporal lobe (~ 50%)
- Ca^{2+} common (up to 50%)
- Enhancement in ~ 50%
- Superficial lesions may remodel overlying calvaria
- Ill-defined, adjacent, cortical signal abnormality suggests associated focal cortical dysplasia (FCD)

TOP DIFFERENTIAL DIAGNOSES

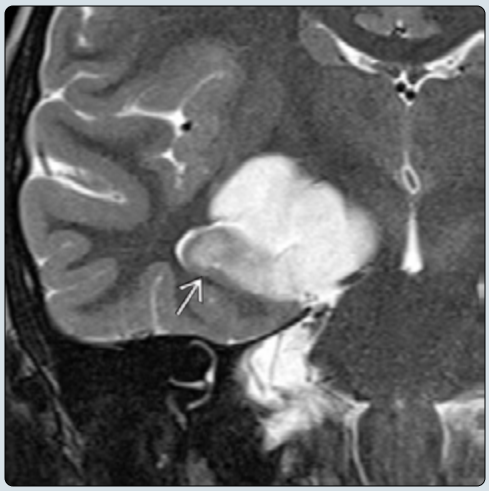
- Dysembryoplastic neuroepithelial tumor
- FCD
- Pleomorphic xanthoastrocytoma
- Pilocytic astrocytoma
- Diffuse astrocytoma (grade II)
- Oligodendroglioma

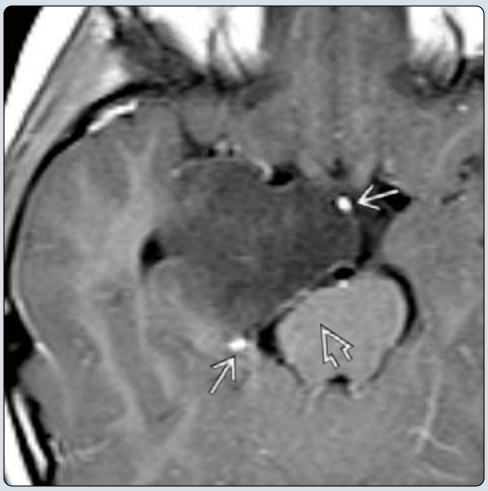
PATHOLOGY

- WHO grade I (80%) or II
- Uncommon: Anaplastic ganglioglioma (WHO III)
- Rare: Malignant with glioblastoma multiforme-like glial component (WHO IV)

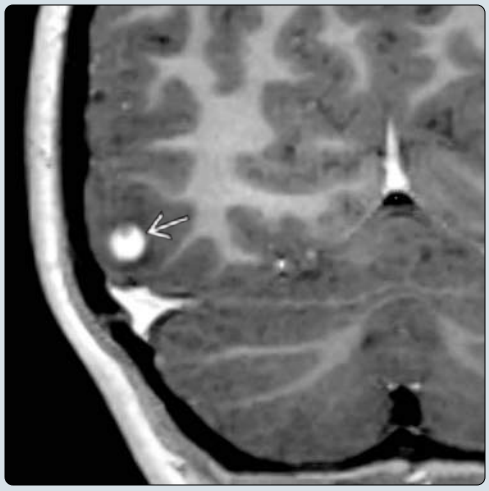
CLINICAL ISSUES

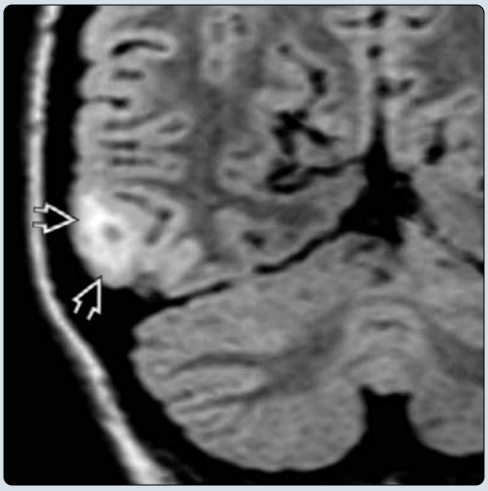
- Occurs at all ages; peak: 10-20 years
- 1-4% of all pediatric CNS neoplasms
- Most common glioneuronal tumor
- Most common neoplastic cause of temporal lobe epilepsy
- Excellent prognosis with complete resection
- Seizure freedom improved with early surgical intervention





(Left) *Coronal T2 MR in a 5-year-old boy with episodes of eyelid fluttering & left arm dystonia shows a well-defined, hyperintense solid mass in the right mesial temporal lobe causing enlargement of the hippocampus. This is a common location & appearance for ganglioglioma.* **(Right)** *Axial T1 C+ MR in the same patient shows a lack of enhancement within the lesion. There is mass effect upon the adjacent vessels & brainstem. Up to 50% of gangliogliomas will demonstrate no enhancement.*





(Left) *Coronal T1 C+ MR in a 12-year-old girl with new onset of nocturnal partial seizures shows a well-defined, round area of enhancement in the right posterior temporal lobe.* **(Right)** *Coronal reconstruction of a 3D FLAIR MR in the same patient shows abnormal signal within the adjacent cortex, a finding that suggests associated focal cortical dysplasia (FCD). Pathology confirmed dysplastic neurons of FCD in this adjacent cortex.*

Desmoplastic Infantile Tumors

KEY FACTS

TERMINOLOGY

- Large cystic tumors of infants involving superficial cerebral cortex & leptomeninges
- Desmoplastic infantile ganglioglioma (DIG)
  - Prominent desmoplastic stroma + neoplastic astrocytes & variable neuronal component
- Desmoplastic infantile astrocytoma (DIA)
  - Desmoplastic stroma + neoplastic astrocytes

IMAGING

- Peripheral supratentorial tumor with large cyst(s) & cortically-based solid nodule/plaque in child < 2 years of age
- Variable T2 signal of solid component, often markedly ↓
  - Peritumoral edema may be vasogenic or interstitial from obstructive hydrocephalus
- May have diffusion restriction despite low histologic grade
- Peripheral solid component often avidly enhances
- Broad dural base + enhancement of adjacent meninges
- Hemorrhage & Ca^{2+} uncommon

TOP DIFFERENTIAL DIAGNOSES

- Primitive neuroectodermal tumor
- Infantile glioblastoma multiforme
- Atypical teratoid/rhabdoid tumor
- Supratentorial ependymoma
- Pleomorphic xanthoastrocytoma
- Pilocytic astrocytoma

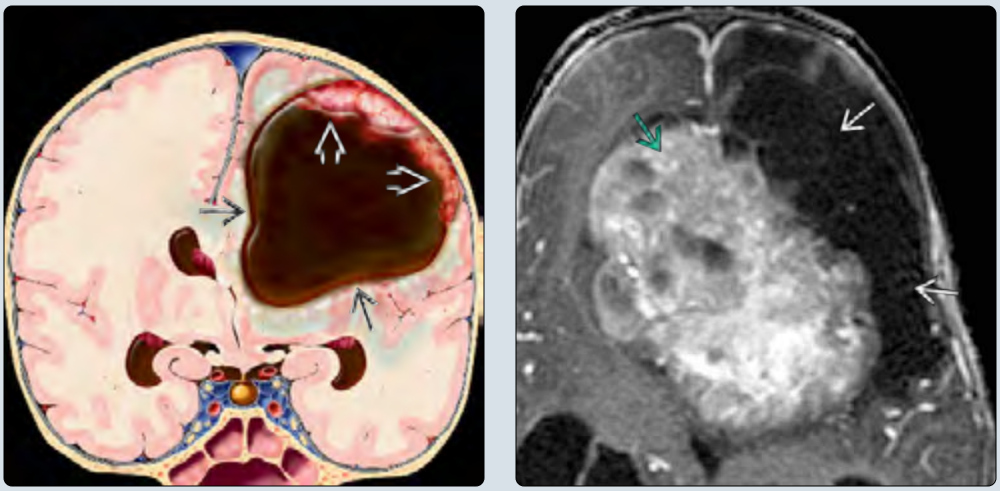
PATHOLOGY

- WHO grade I
- Areas of cellular proliferation, mitoses, & necrosis may cause misdiagnosis as higher grade tumor

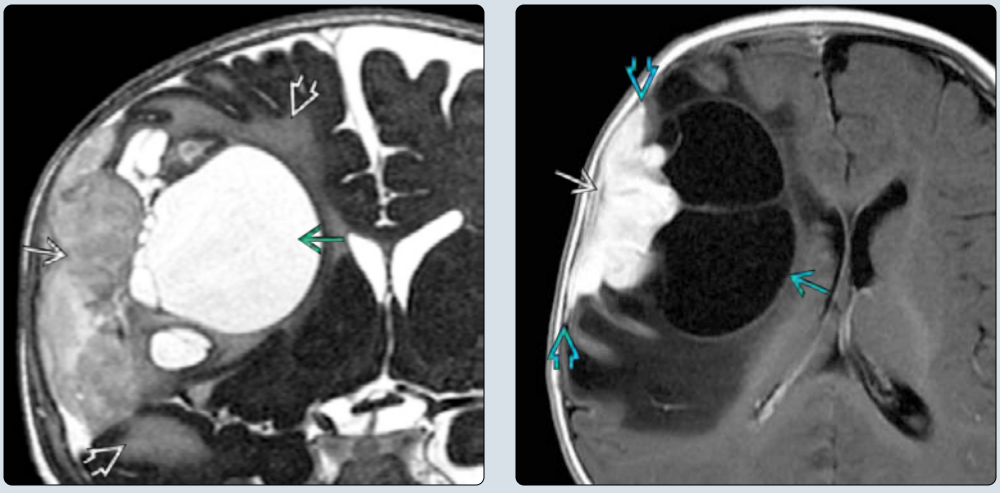
CLINICAL ISSUES

- Most found at 1-24 months of age (peak: 3-6 months)
- 16% of intracranial tumors in 1st year of life
- Median survival rate > 75% at 15 years
- Complete surgical resection typically curative
  - 40% require additional therapy beyond surgery

(Left) *Coronal graphic shows an infant with DIG/DIA. Note the dominant cystic component ⇨ with a dural-based plaque of desmoplastic stroma ➡. Mild surrounding edema & hydrocephalus is present.* **(Right)** *Axial T1 C+ MR in a 15-month-old girl with loss of developmental milestones shows a large central supratentorial mass with avidly enhancing solid ⇨ & complex cystic ➡ components. When a DIG arises from paracentral cortex, it may appear in a central rather than peripheral location.*



(Left) *Coronal 3D FIESTA MR in a 5-month-old girl with increasing head circumference shows a large right cerebral mass with a peripheral hyperintense solid component ➡ & a more central cystic component ⇨. Note also the peritumoral vasogenic edema ➡.* **(Right)** *Axial T1 C+ MR in the same 5-month-old shows homogeneous enhancement of the peripheral solid component ➡ & no significant enhancement along the margins of the central cystic component ⇨. Note the broad-based dural attachment ⇨ of this DIG.*



KEY FACTS

TERMINOLOGY

- Central nervous system primitive neuroectodermal tumor (CNS-PNET): Antiquated term for primitive cerebral embryonal tumor; no longer found in WHO 2016
- Now molecularly & genetically distinct entities considered under embryonal tumors (all WHO grade IV) include
  - Embryonal tumor with multilayered rosettes, C19MC altered
  - Embryonal tumor with multilayered rosettes, not otherwise specified (NOS)
  - Medulloepithelioma
  - CNS embryonal tumor, NOS
    - Most CNS-PNET lesions likely here

IMAGING

- Large (mean: 5 cm), complex-appearing hemispheric mass in infant/young child
- ~ 50% well defined with minimal peritumoral edema
- Ca^{2+} in 50-70%; hemorrhage common
- Heterogeneous enhancement
- Restricted diffusion of solid highly cellular components
- Subarachnoid spread common at presentation
  - Perform MR of entire neuraxis before surgery

TOP DIFFERENTIAL DIAGNOSES

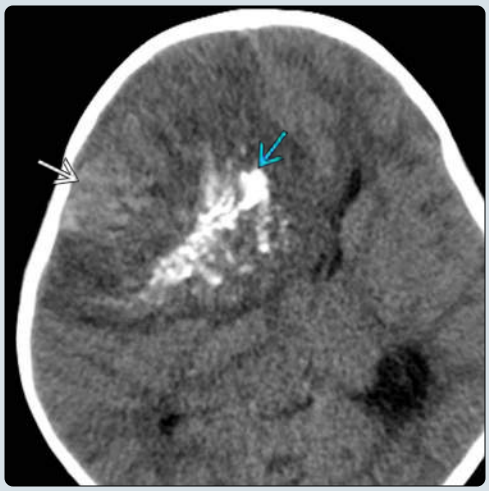
- Astrocytoma
- Ependymoma
- Atypical teratoid/rhabdoid tumor
- Choroid plexus carcinoma
- Giant cavernoma

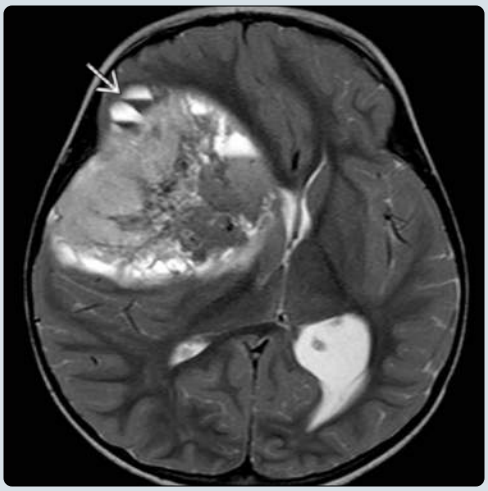
PATHOLOGY

- Histology similar to medulloblastoma but molecularly & genetically distinct

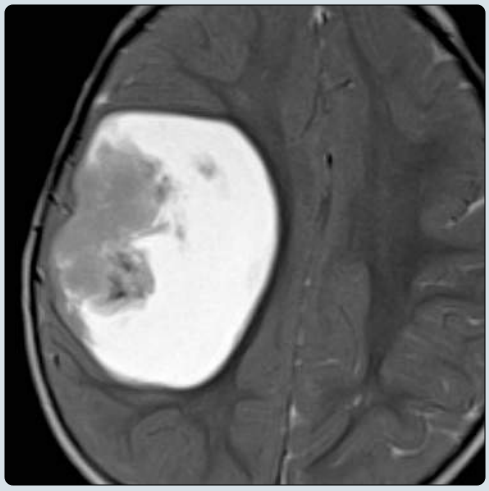
CLINICAL ISSUES

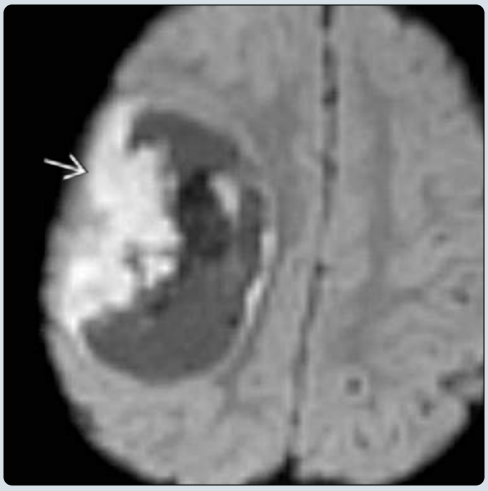
- Most common in younger children
- Treatment: Aggressive surgical resection, chemotherapy, craniospinal radiation (if of tolerable age)





(Left) *Axial NECT in a 2-year-old child with altered mental status shows a large heterogeneous intraaxial mass. Areas of attenuation equal to gray matter ➡ correspond to cellular areas of the tumor. Ca^{2+} (as seen in this mass ⇨) are present in > 50% of CNS-PNETs.* **(Right)** *Axial T2 MR in the same patient shows marked internal heterogeneity of the mass. Note cystic areas containing fluid-fluid levels ➡ in the anterior aspect of the lesion, consistent with foci of intracystic hemorrhage. Note the lack of vasogenic edema.*





(Left) *Axial T2 MR in a 3-year-old child with left-sided weakness & difficulty walking shows a large, well-demarcated mixed solid & cystic mass in the right cerebral hemisphere. Note the lack of surrounding vasogenic edema.* **(Right)** *Axial DWI MR in the same patient shows mildly high signal ➡ within the solid portions of the mass, consistent with diffusion restriction from the high cellularity of CNS-PNET. Restricted diffusion is a key diagnostic clue to making this diagnosis.*

Craniopharyngioma

KEY FACTS

IMAGING

- Best clue: Complex suprasellar cystic mass with Ca^{2+} & wall enhancement
- Location: Suprasellar 75%, suprasellar + intrasellar 21%
  - Larger tumors can extend into multiple cranial fossae
- "90% rule"
  - 90% cystic: MR signal intensity highly variable
    - Cystic components frequently T1 hyperintense
  - 90% calcified: SWI/T2* GRE MR helpful to identify Ca^{2+}
  - 90% enhance (wall & solid portions)
- Often develop obstructive hydrocephalus
- Optic chiasm, hypothalamus, & vessels often involved
  - Thin sagittal T2 or SSFP MR sequences help assess

TOP DIFFERENTIAL DIAGNOSES

- Rathke cleft cyst
- Pituitary adenoma
- Germinoma
- Hypothalamic-chiasmatic glioma

PATHOLOGY

- 2 clinically & pathologically distinct subtypes
  - Adamantinomatous: Cystic & solid, mostly in children
  - Squamous-papillary: Mostly solid tumor found in adults

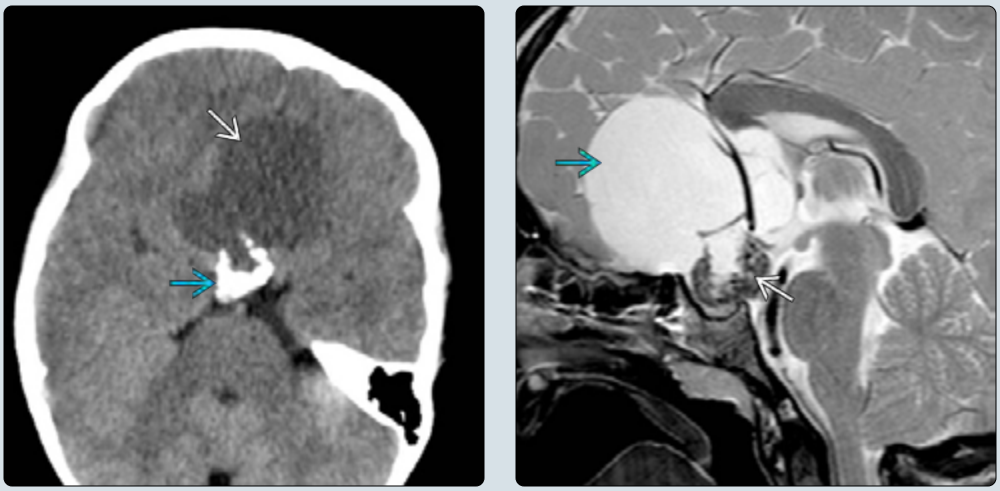
CLINICAL ISSUES

- Most common nonglial pediatric intracranial tumor
  - 6-9% of all pediatric intracranial tumors
- Symptoms: Visual changes, endocrine related, academic decline, headache/vomiting (obstructive hydrocephalus)
  - ~ 1/3 of patients with endocrine symptoms (↓ growth hormone, ↓ thyroid function, diabetes insipidus)
- Benign tumor with high rate of recurrence
- Poor prognostic factors: Hypothalamic involvement, ↑ tumor size

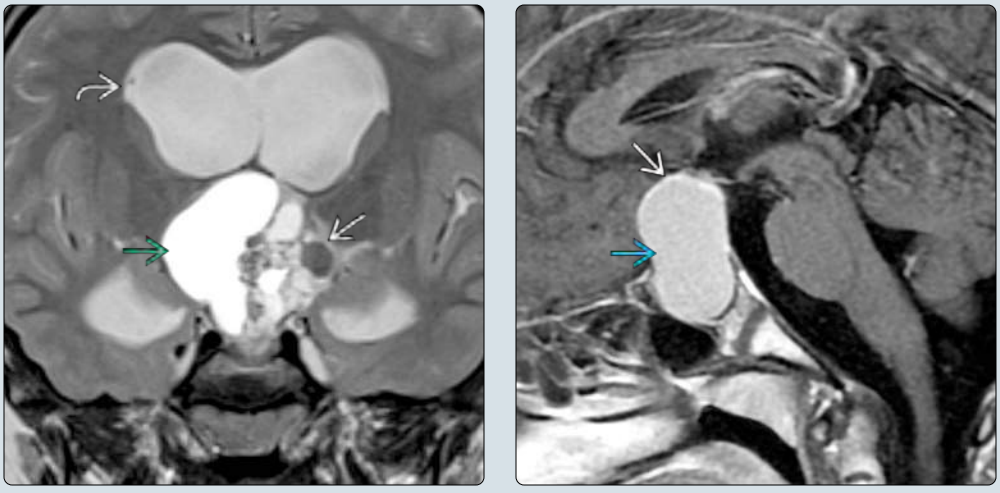
DIAGNOSTIC CHECKLIST

- Must identify relationship of tumor to optic chiasm, hypothalamus, & vessels

(Left) *Axial NECT in a 3-year-old boy with 2 months of headaches & vomiting shows a partially calcified ⇨, partially cystic ➡ lesion in the suprasellar region & interhemispheric fissure, characteristic of an adamantinomatous craniopharyngioma.* **(Right)** *Sagittal T2 MR in the same 3 year old shows hypointense signal in the posterior components ➡ of the mass, corresponding to Ca^{2+} on the CT. Frontal extension of the large cystic components ⇨ is typical of a prechiasmatic location.*



(Left) *Coronal T2 MR in an 8-year-old boy with visual changes shows a mixed solid ➡ & cystic ⇨ mass with obstructive hydrocephalus ➡. The visual changes are related to mass effect upon the optic chiasm.* **(Right)** *Sagittal T1 C+ MR in a 10-year-old boy with increasing headaches shows a predominantly cystic mass with a very thin rim of enhancement ➡. The majority of craniopharyngiomas have some solid component but not all. Cystic contents often show intrinsic T1 shortening ⇨ before contrast, as in this case.*



TERMINOLOGY

Definitions

- Craniopharyngioma (CP): Histologically benign epithelial tumor arising from squamous rests along involuted hypophyseal-Rathke duct

IMAGING

General Features

- Best diagnostic clue
  - Cystic suprasellar mass with Ca^{2+} & enhancing wall or mural nodule
- Location
  - Suprasellar: 75%, mixed suprasellar + intrasellar: 21%, intrasellar: 4%
  - Retrochiasmatic vs. prechiasmatic configuration key determinant for surgical approach
  - Larger tumors can extend into multiple cranial fossae
- Often large at presentation (> 5 cm)
  - Cyst typically largest component

CT Findings

- "90% rule": 90% cystic, 90% calcified, 90% enhance (wall & solid portions)
- May present with obstructive hydrocephalus from compression on 3rd ventricle & foramen of Monro

MR Findings

- T1WI: Cysts often very hyperintense, reflecting protein, cholesterol, &/or blood products in fluid
- T2WI: Heterogeneous solid tumor: Hypointense foci of Ca^{2+}, variable signal intensity of cysts
- FLAIR: Cysts typically do not suppress like CSF
- SWI/T2* GRE: Areas of signal loss correspond to Ca^{2+}
- DWI: Variable cystic signal, typically closer to CSF than brain
- T1WI C+: Solid portions enhance heterogeneously; cyst wall may show smooth or irregular enhancement

Imaging Recommendations

- Protocol advice
  - Volumetric T1 ± contrast; SWI/GRE for Ca^{2+}
  - Thin sagittal T2 or SSFP sequences help define relationship of tumor to adjacent structures
  - MRA helpful in preoperative identification of arterial involvement

DIFFERENTIAL DIAGNOSIS

Rathke Cleft Cyst

- Ca^{2+} uncommon; no nodular/solid component
- Cyst more homogeneous, although small T2-hypointense intracystic nodules may be present

Germinoma

- Predominantly solid & lobular
- Ca^{2+} very rare in suprasellar germinoma
- Strong association with diabetes insipidus

Pituitary Adenoma

- Solid tumor arising from adenohypophysis
- Diffuse & homogeneous enhancement

Hypothalamic-Chiasmatic Glioma

- More solid & homogeneous
- Extension into prechiasmatic optic nerves/tracts

PATHOLOGY

General Features

- 2 proposed sites of origin
  - In pars tuberalis at distal aspect of infundibulum
  - Along tract of involuted craniopharyngeal duct
- Over activation of WNT/β-catenin signaling pathway
- 2 clinically & pathologically distinct subtypes
  - Adamantinomatous (~ 90%): Classic calcified cyst with mural nodule seen in children
  - Papillary (~ 10%): Mostly solid tumor almost exclusively found in adults
  - Adamantinomatous & papillary: WHO grade I

CLINICAL ISSUES

Presentation

- Headache, vomiting, hydrocephalus, papilledema
- Visual disturbance, decline in school performance
- Endocrine symptoms in at least 1/3 of cases
  - Growth hormone deficiency, hypothyroidism, diabetes insipidus
  - Due to mass effect on pituitary/hypothalamus

Demographics

- Epidemiology
  - Most common nonglial pediatric intracranial tumor
    - > 50% of all pediatric suprasellar region tumors

Natural History & Prognosis

- 88% overall survival at 20 years
- Hypothalamic involvement: ↓ overall survival, quality of life
- Factors affecting recurrence
  - Gross total vs. partial resection
  - Radiation therapy (for residual tumor) ↓ recurrence
  - Tumor size (↑ recurrence in larger tumors)

Treatment

- Surgical: Complete resection ideal but must be weighed against high morbidity associated with extensive resection
- Radiation therapy for incomplete resection

DIAGNOSTIC CHECKLIST

Consider

- Rathke cleft cyst can be identical to small CP
  - Rathke cleft cyst unlikely to have Ca^{2+}, more homogeneous
  - Use NECT & SWI/GRE to identify Ca^{2+} in CP
- Relationship of tumor to optic chiasm & other adjacent structures important for surgical planning

SELECTED REFERENCES

1. Sterkenburg AS et al: Survival, hypothalamic obesity, and neuropsychological/psychosocial status after childhood-onset craniopharyngioma: newly reported long-term outcomes. Neuro Oncol. 17(7):1029-38, 2015
2. Plaza MJ et al: Conventional and advanced MRI features of pediatric intracranial tumors: posterior fossa and suprasellar tumors. AJR Am J Roentgenol. 200(5):1115-24, 2013

Germinoma, Brain

KEY FACTS

TERMINOLOGY

- Tumor of primordial germ cells, histologically identical to gonadal & nongonadal seminoma or dysgerminoma

IMAGING

- CT: Hyperattenuating solid components
  - Pineal region: Localized "engulfed" Ca^{2+}
  - Other sites: Ca^{2+} rare, but hemorrhage may occur
- MR: Mass in suprasellar, pineal, or basal ganglia regions
  - T2: Multiple small cysts not unusual (~ 25%)
  - DWI: Diffusion restriction in solid components
  - T1 C+: Avid enhancement, often "speckled"
  - Pineal: Bithalamic extension, peritumoral edema
  - Suprasellar: Absent T1 posterior pituitary bright spot
  - Metastatic CSF dissemination common (25-40%)

TOP DIFFERENTIAL DIAGNOSES

- Pineal region
  - Pineoblastoma
  - Nongerminomatous germ cell tumors (teratoma, etc.)
  - Tectal plate glioma
- Suprasellar
  - Craniopharyngioma
  - Hypothalamic/chiasmatic astrocytoma
  - Langerhans cell histiocytosis

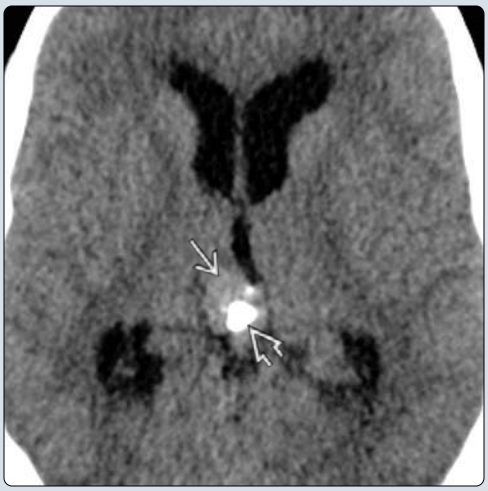
CLINICAL ISSUES

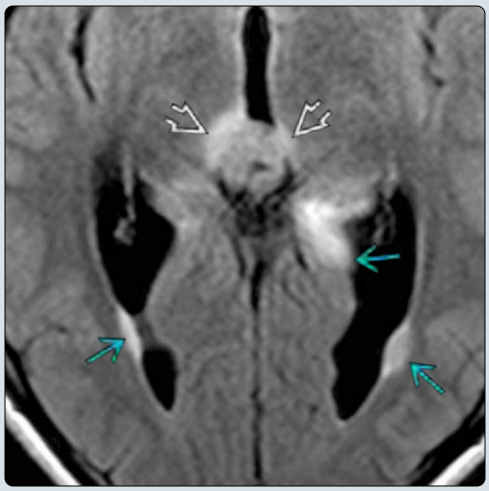
- M > F, especially in pineal region
- Mean age: 10-15 years
- Pineal: Headache, paralysis of upward gaze
- Suprasellar: Diabetes insipidus (DI), vision changes
- ± ↑ serum & CSF human chorionic gonadotropin
- Favorable prognosis (5-year survival > 90%)

DIAGNOSTIC CHECKLIST

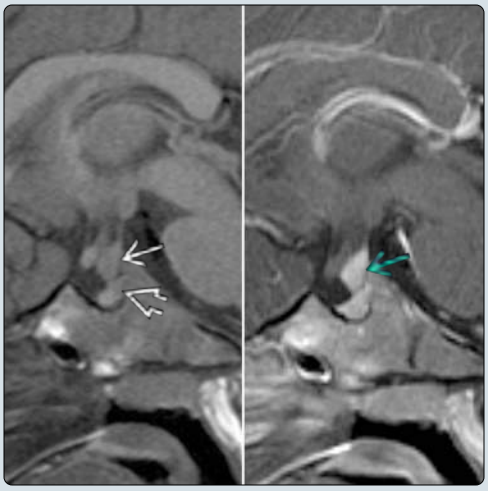
- Image entire neuraxis to detect CSF dissemination
- Occult germinoma possible in child with DI & normal MR
  - Repeat MR in 3-6 months to assess for growing mass

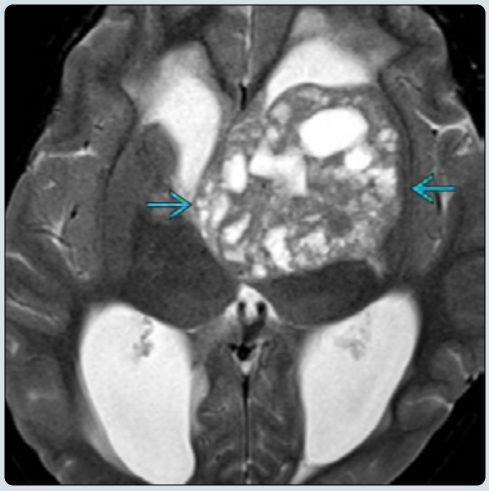
(Left) *Axial NECT in a 14-year-old boy with worsening headaches shows a hyperattenuating mass with localized Ca^{2+}. Calcifications are common in pineal germinoma & are thought to represent pineal Ca^{2+} "engulfed" by tumor.* **(Right)** *Axial FLAIR MR in the same patient shows parenchymal edema within the bilateral thalami (right > left). Peritumoral edema is a common finding (~ 40%) in germinoma, as are multiple ependymal metastases.*





(Left) *Sagittal T1 C+ MR (left) in a 4-year-old boy with central diabetes insipidus shows an infundibular mass & absence of the posterior pituitary bright spot. After contrast (right), the infundibular mass enhances homogeneously, typical of germinoma.* **(Right)** *Axial T2 MR in a 10-year-old girl with fatigue, headaches, & vomiting shows a large solid & cystic mass centered in the left basal ganglia. The basal ganglia are a less common (~ 15%) location for germinoma (though these tumors are often large).*





TERMINOLOGY

Definitions

- Tumor of primordial germ cells, essentially identical to gonadal or extragonadal seminoma & dysgerminoma

IMAGING

General Features

- Best diagnostic clue
  - Pineal mass with localized Ca^{2+} & bithalamic extension
  - Suprasellar mass with diabetes insipidus (DI)
- Location
  - Central nervous system (CNS) germinomas typically occur in midline near 3rd ventricle
    - Pineal region (40-50%)
    - Suprasellar (30-40%)
    - Basal ganglia (BG) tumors less common (5-15%)
  - CSF spread of metastases common (25-40%)
- Size
  - Typically small (1-3 cm)
  - Tiny or inapparent suprasellar germinoma may cause DI

CT Findings

- Sharply circumscribed heterogeneously hyperdense mass
- Localized ("engulfed") Ca^{2+} common in pineal tumors
- Ca^{2+} rare in suprasellar tumors
- Primary tumors & metastases usually enhance avidly

MR Findings

- T1WI: Isointense to hypointense
- T2WI: Hyperintense cystic components
- FLAIR: Sensitive for peritumoral edema
- DWI: Solid components typically show diffusion restriction
- GRE/SWI: Hypointense signal may represent Ca^{2+} (pineal) or hemorrhage (suprasellar & BG)
- T1C+: Most tumors show marked enhancement
  - Speckled pattern in tumors with small cysts
  - Most sensitive sequence for CSF metastases

DIFFERENTIAL DIAGNOSIS

Pineal Region Masses

- Pineoblastoma
  - Highly cellular pineal mass in young child
  - Scattered ("exploded") Ca^{2+} rather than localized ("engulfed") Ca^{2+} in germinoma
  - Less common than germinoma & typically younger age
- Tectal plate glioma
  - Little or no enhancement
  - High ADC signal (no diffusion restriction)

Sellar/Suprasellar Masses

- Craniopharyngioma
  - Dominant cystic components ± T1 shortening, Ca^{2+}
  - DI not usually present until after surgery
- Hypothalamic/chiasmatic astrocytoma
  - No diffusion restriction
  - Rarely associated with DI
- Langerhans cell histiocytosis (LCH)
  - Thickened infundibulum; look for osseus lesions

PATHOLOGY

General Features

- Associated abnormalities
  - ± ↑ serum & CSF human chorionic gonadotropin

Microscopic Features

- Sheets of large polygonal primitive germ cells
  - Large vesicular nuclei & prominent nucleoli
  - Clear, glycogen-rich cytoplasm (PAS-positive)
- Lymphocytic inflammatory component common

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Pineal region germinoma
    - Headache secondary to aqueduct obstruction & hydrocephalus
    - Parinaud syndrome (paralysis of upward gaze)
    - Precocious puberty
  - Suprasellar germinoma
    - DI
    - Hypothalamic-pituitary dysfunction (↓ growth, precocious puberty)
    - Visual symptoms

Demographics

- Age
  - Mean age in most studies: 10-15 years
- Gender
  - M > F, especially in pineal region

Natural History & Prognosis

- Pure germinoma has favorable prognosis
- Treatment with radiotherapy ± adjuvant chemotherapy
  - 5-year survival > 90%

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Child or adolescent with DI
  - Lobular enhancing suprasellar mass = germinoma
  - Thick, enhancing infundibulum = LCH or germinoma
  - No enhancing lesion = could be occult germinoma
- Adolescent with pineal mass = germinoma or nongerminomatous germ cell tumor (NGGCT)
- Child with pineal mass
  - Complex, cystic = also consider NGGCT
  - Homogeneous with restricted diffusion = germinoma vs. pineoblastoma

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Choroid Plexus Tumors

KEY FACTS

TERMINOLOGY

- Choroid plexus tumor (CPT)
  - Choroid plexus papilloma (CPP): WHO grade I
  - Atypical choroid plexus papilloma (aCPP): WHO grade II
  - Choroid plexus carcinoma (CPCa): WHO grade III

IMAGING

- Strongly enhancing, lobulated intraventricular mass
  - 60-70%: Lateral ventricles (most commonly atrium)
  - 20-30%: 4th ventricle (most common site in adults)
  - < 10%: 3rd ventricle, cerebellomedullary cistern
- Ventriculomegaly from obstruction or ↑ CSF production
- **CT**: Ca^{2+} in 25%
- **MR**: Iso- to hyperintense on T2; internal flow voids
- CPCa more likely than CPP to show heterogeneous enhancement, necrosis, brain invasion, & CSF spread

TOP DIFFERENTIAL DIAGNOSES

- Intraventricular hemorrhage
- Atypical teratoid rhabdoid tumor
- Sturge-Weber syndrome
- Medulloblastoma
- Ependymoma

PATHOLOGY

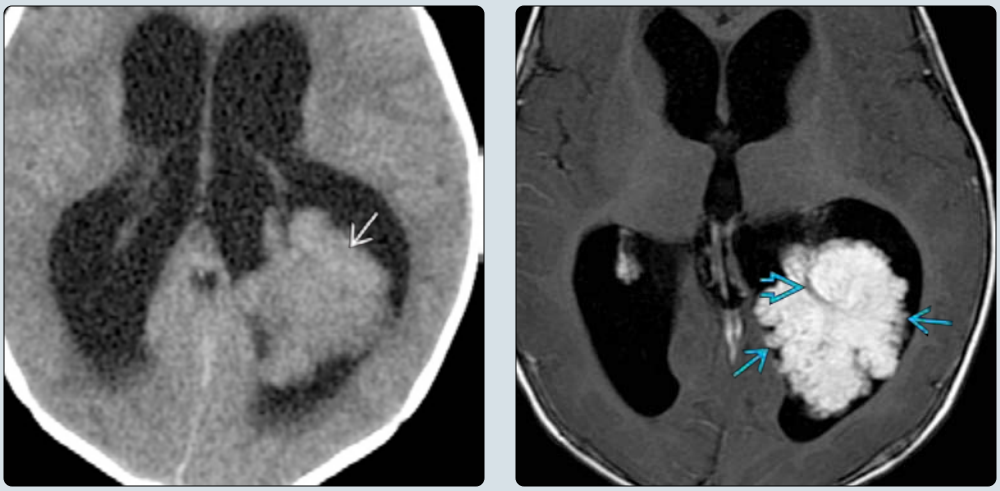
- ↑ p53 mutations in tumor correlate with poor prognosis
- Molecular subtype may be more prognostic than histology
  - Cluster 1: Pediatric, supratentorial, good prognosis
  - Cluster 2: Adult, infratentorial, good prognosis
  - Cluster 3: Pediatric, supratentorial, poor prognosis
- Associated with Li-Fraumeni & Aicardi syndromes

CLINICAL ISSUES

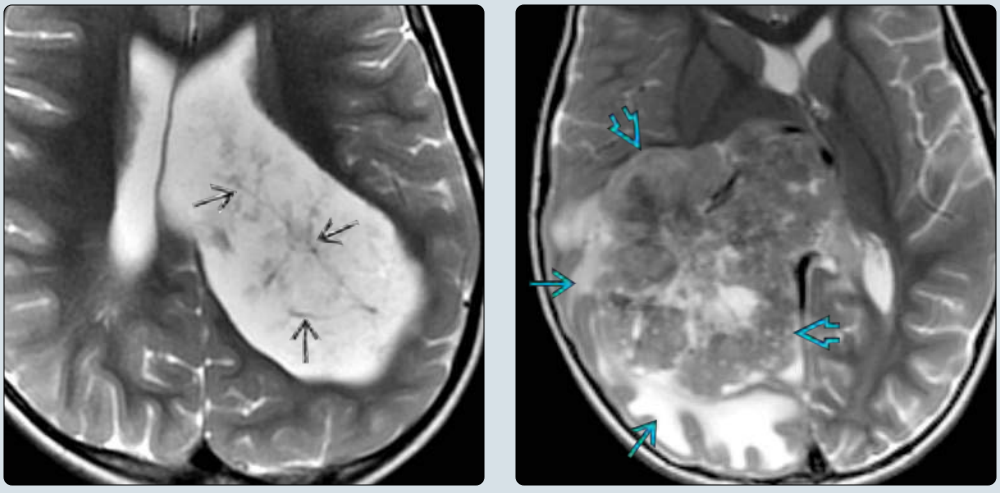
- ~ 25-33% present < 1 year of age; ~ 49-75% present < 5 years of age

DIAGNOSTIC CHECKLIST

- C+ MR of entire neuraxis before surgery
- Imaging not reliable for distinguishing CPP from CPCa



(Left) *Axial NECT in an 8-month-old girl with vomiting shows an isodense, lobulated mass ➡ centered in the atrium of the left lateral ventricle. Note the ventriculomegaly in the absence of ventricular obstruction (secondary to increased CSF production by the CPP).* **(Right)** *Axial T1 C+ MR in the same patient shows avid, homogeneous enhancement of the ventricular mass. Note the lobulated, frond-like margin ⇨ & large central flow void ⇨, typical of CPP.*



(Left) *Axial T2 MR in a 3-year-old girl shows predominantly hyperintense signal (equal to CSF) throughout a large mass that expands the ventricle. A few branching, hypointense foci ⇨ within the lesion represent high-flow vessels. CPP was proven upon resection.* **(Right)** *Axial T2 MR in a 3 year old shows heterogeneous signal throughout a large posterior intraventricular mass ⇨. There is extensive vasogenic edema ⇨ in the adjacent brain parenchyma. Pathology revealed a CPCa.*

TERMINOLOGY

Abbreviations

- Choroid plexus tumor (CPT)
  - Choroid plexus papilloma (CPP): WHO grade I
  - Atypical choroid plexus papilloma (aCPP): WHO grade II
  - Choroid plexus carcinoma (CPCa): WHO grade III

Definitions

- Intraventricular, papillary neoplasm derived from CP epithelium

IMAGING

General Features

- Best diagnostic clue
  - Strongly enhancing, lobulated intraventricular mass
  - Associated hydrocephalus may be obstructive or related to overproduction of CSF by tumor
- Location
  - 60-70%: Lateral ventricle (most commonly atrium)
  - 20-30%: 4th ventricle
  - < 10%: 3rd ventricle, cerebellomedullary cistern
- Size
  - Small to very large (CPCa typically larger)

CT Findings

- Iso- or hyperattenuating in 75%
- Ca^{2+} in 25%

MR Findings

- **T1WI**: Well-delineated, iso- to hypointense mass
- **T2WI**: Iso- to hyperintense mass
  - Internal dark signal: Ca^{2+} (nodular or irregular) vs. vascular flow voids (linear & branching)
- **FLAIR**: ↑ generalized periventricular signal → transependymal edema due to ventricular obstruction
  - Brain invasion causing focally abnormal signal more common in CPCa
- **SWI/T2* GRE**: ± foci of low signal → Ca^{2+} or hemorrhage
- **DWI**: Iso- to hyperintense on ADC
- **T1 C+**: Robust enhancement; frond-like margins
  - Heterogeneity with necrosis suggests CPCa
- **MRA**: May see arteries within vascular mass
- **Perfusion**: Typically elevated CBF & CBV

Imaging Recommendations

- Best imaging tool
  - MR with contrast of entire neuraxis prior to surgery

DIFFERENTIAL DIAGNOSIS

Intraventricular Hemorrhage

- Originates from germinal matrix in premature neonate
- Clot adherent to choroid can be echogenic & mass-like but lacks internal vascularity/enhancement

Atypical Teratoid Rhabdoid Tumor

- May be intraventricular & lobulated
- Shows less enhancement, more diffusion restriction

Sturge-Weber Syndrome

- Abnormal cortical venous drainage → ipsilateral cerebral atrophy, pial angiomatosis, choroid enlargement

Medulloblastoma

- Most common 4th ventricular neoplasm in children
- Shows less enhancement, more diffusion restriction

Ependymoma

- Ependymoma typically intraaxial when supratentorial
- 4th ventricular ependymoma similar to CPT

PATHOLOGY

General Features

- Genetics
  - 3 molecular subtypes by methylation pattern (which may be more predictive of prognosis than histology)
    - Cluster 1: Pediatric, supratentorial, good prognosis
      - CPP ~ aCPP histologically
    - Cluster 2: Adult, infratentorial, good prognosis
      - CPP > aCPP histologically
    - Cluster 3: Pediatric, supratentorial, poor prognosis
      - CPCa > aCPP > CPP histologically
  - Associated with Li-Fraumeni & Aicardi syndromes

Gross Pathologic & Surgical Features

- Brain parenchymal invasion more likely in CPCa
- Subarachnoid spread of tumor more likely in CPCa

CLINICAL ISSUES

Presentation

- Young child with signs & symptoms of ↑ intracranial pressure: Macrocrania, bulging fontanelle, vomiting, headache, ataxia, seizure

Demographics

- Age
  - CPP: 49% < 5 years; 26% < 1 year
  - CPCa: 75% < 5 years; 33% < 1 year
- Epidemiology
  - 2% of all pediatric brain tumors; CPP:CPCa ~ 2-3:1

Natural History & Prognosis

- CPP benign, slowly growing; 5-year survival ~ 100%
- CPCa usually large & invasive; 5-year survival ~ 60%

Treatment

- Gross total resection (GTR)

DIAGNOSTIC CHECKLIST

Consider

- Imaging alone not reliable for distinguishing CPP from CPCa
- 4th ventricular CPT less common than other pediatric posterior fossa tumors

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Child Abuse, Brain

KEY FACTS

TERMINOLOGY

- Nonaccidental trauma (NAT), abusive head trauma (AHT)
- Traumatic injury inflicted on infants & children by adults

IMAGING

- Direct impact injury: Direct blow to cranium or impact of skull on object
  - Calvarial (often complex) & skull base fractures
  - Focal brain injury deep to impact
- Shaking injury: Result of violent "to & fro" motion of head
  - Subdural hematomas (SDH) in 90-98%
  - Generalized parenchymal injuries (cytotoxic edema, lacerations, axonal injury)
  - Bridging vein injury & thrombosis common
- CT primary imaging tool in initial evaluation of AHT
  - Multiplanar reconstructions improve detection of
    - Small intracranial hemorrhages (ICH)
    - Fractures (with bone algorithm & 3D reformats)
- MR best for determining full extent of injury
  - DWI paramount for parenchymal injury
  - PD & SWI/T2* GRE for hemorrhage
  - T1 C+ for chronic SDH membranes

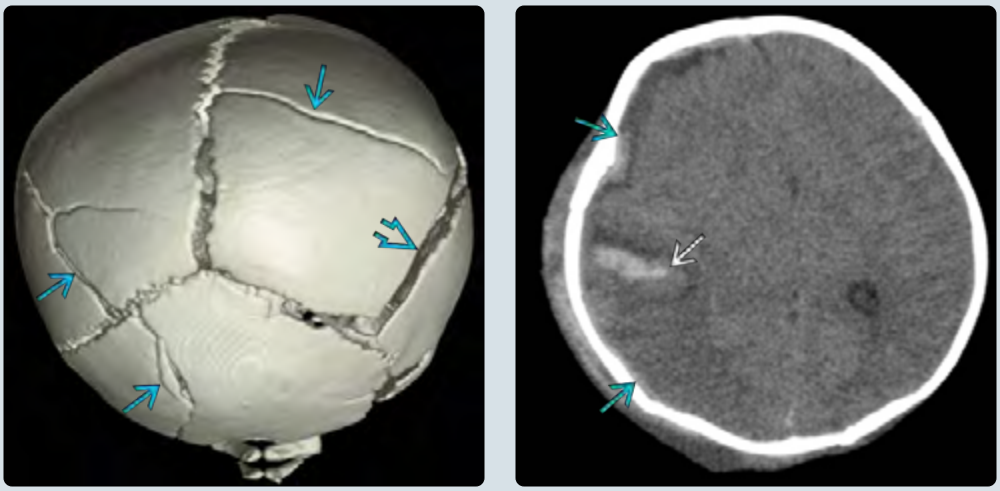
TOP DIFFERENTIAL DIAGNOSES

- Accidental trauma
- Benign macrocrania of infancy
- Mitochondrial encephalopathies
- Bleeding disorders

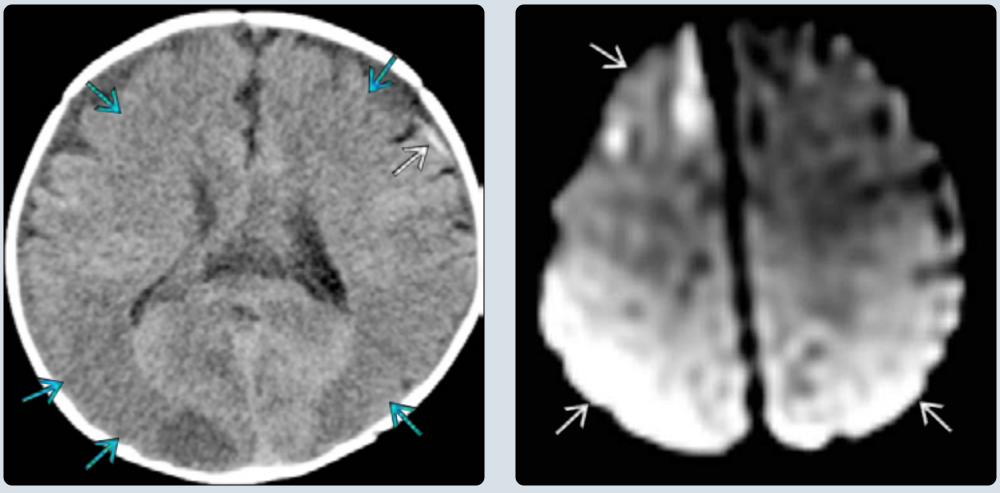
CLINICAL ISSUES

- Discordance between stated history & degree of injury
  - "Killer couch": Injuries blamed on infant rolling off couch
- Retinal hemorrhages in ~ 75%
- #1 cause of brain injury death in children < 2 years of age
  - 17-25:100,000 annual incidence
  - Cause of death in 80% of fatalities is brain swelling

(Left) *Posterior oblique view of a 3D NECT in a 9 week old who "fell off the couch" shows multiple complex skull fractures, including a displaced right parietal fracture. 3D renderings are helpful in improving the detection & characterization of skull fractures.* **(Right)** *Axial NECT in the same 9 week old shows a right subdural hematoma (SDH) & right opercular parenchymal laceration with significant midline shift & sulcal effacement. Parenchymal lacerations are seen in 10-15% of abusive head trauma (AHT).*



(Left) *Axial NECT in a 4-month-old boy with seizure activity shows multiple bilateral foci of low attenuation with loss of cortical differentiation as well as a left frontal SDH. There was no fracture, making these findings highly concerning for the shaking type of AHT.* **(Right)** *Axial DWI MR in a 2-month-old boy with AHT shows areas of diffusion restriction in the right frontal lobe & bilateral parietal lobes, consistent with parenchymal injury. MR is the most sensitive examination for parenchymal injury.*



TERMINOLOGY

Abbreviations

- Nonaccidental trauma (NAT), abusive head trauma (AHT), shaken-baby syndrome (SBS)

Definitions

- Traumatic injury inflicted on infants & children by adults

IMAGING

General Features

- 2 major groupings of injuries (but can occur together)
  - Direct impact injury: Result of direct blow to cranium or impact of skull on object
  - Shaking injury: Result of violent "to & fro" head motion
- Direct impact injury typified by skull fractures & injury to immediately underlying brain
  - Scalp laceration, hematoma, swelling strongly associated
  - High association with injuries to other organs
- Shaking injury typified by subdural hemorrhage (SDH) & generalized brain parenchymal injury
  - Cytotoxic brain injury not conforming to arterial territories
  - May see bridging vein injury ± thrombosis
  - Imaging findings may suggest injuries of differing ages

Radiographic Findings

- Sensitive in detection of linear skull fractures
  - CT (with appropriate techniques) better characterizes fractures; often being obtained to evaluate for intracranial hemorrhage
- Some fractures considered more suspicious for NAT
  - Evidence does not support this
  - Discordance with provided history best indicator

CT Findings

- NECT primary imaging tool for initial evaluation of AHT
- Intracranial hemorrhage (ICH)
  - SDH most common (90-98%)
    - Dominant feature of shaking injury
  - Subarachnoid hemorrhage common (> 50%)
  - Epidural hemorrhage uncommon but may occur
  - Use great caution if attempting to estimate "age" of ICH
- Subdural hygromas (SDHy) may develop after injury
- Bridging vein injury ± thrombosis common (40-50%)
  - Areas of ↑ density in paramedian high convexities
- Parenchymal ischemic injury often seen in shaking injuries
  - Areas of ↓ density (with loss of gray-white differentiation) & sulcal effacement not confined to arterial territories; may be diffuse
- Parenchymal laceration in 10-15%
- Shear injury (axonal injury) in ~ 15%
- Retinal hemorrhages rarely visualized

MR Findings

- DWI: Key for parenchymal injury
- T1WI: Bright foci of hemorrhage or evolving cortical injury
- T2WI: Loss of cortical ribbon & deep nuclei in neonates
- PD/intermediate echo sequences: Very sensitive for detection of small subdural collections
- SWI/T2* GRE: Detects small ICH ± retinal hemorrhages
- T1WI C+: Enhancing membranes best sign of chronic SDH

Imaging Recommendations

- Best imaging tool
  - NECT for acute evaluation
    - Sensitive in detection & characterization of fractures
    - Very sensitive in detection & characterization of ICH
  - MR after 24-48 hours to define extent of brain injury
- Protocol advice
  - NECT: Multiplanar reconstructions improve detection of
    - Small intracranial hemorrhages
    - Fractures (especially with bone algorithm & 3D)

DIFFERENTIAL DIAGNOSIS

Accidental Trauma

- Appropriate history for degree of injury

Benign Macrocrania of Infancy

- Self-limited communicating hydrocephalus

Mitochondrial Encephalopathies

- May cause atrophy with subdural collections
  - Glutaric acidurias (types I & II), Menkes syndrome

Bleeding Disorders

- von Willebrand, thrombocytopenia

PATHOLOGY

General Features

- ↑ vulnerability in infants due to
  - Large head:body ratio + weak neck muscles
- Retinal hemorrhage in ~ 75% (50-100% in literature)
  - Much less common in accidental head trauma (~ 6%)
- Retroclival collections can be seen in ~ 30% of AHT victims

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Discordance between stated history & degree of injury
    - "Killer couch": Severe injuries blamed on infant rolling off couch onto floor by perpetrator
  - Unprovoked seizures & apnea raise suspicion for AHT

Demographics

- Most common from 1-6 months of age

Natural History & Prognosis

- Mortality rate: 20-25%
- High rates of impairment for survivors

Treatment

- Notification of local Child Protection Agency
- Multidisciplinary child abuse & neglect team intervention

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Germinal Matrix Hemorrhage

KEY FACTS

TERMINOLOGY

- Hemorrhage that occurs in richly vascular, but fragile, germinal matrix in premature infants

IMAGING

- Can occur anywhere along germinal matrix
  - Most commonly at caudothalamic groove
- Look for intraventricular extension, ventriculomegaly, & secondary intraparenchymal hemorrhage
- US: Globular echogenic focus in caudothalamic groove
  - Acute blood echogenic; later clot retracts & becomes iso- to hypoechoic
  - May appear as abnormally thick choroid plexus but of slightly different echogenicity & lacking vascularity
  - Fluid-debris levels may be visible in dependent ventricles
  - Coronal & sagittal cine clips sweeping though ventricles help differentiate hemorrhage from normal choroid
- MR: Sensitive for detection of germinal matrix hemorrhage (GMH) & intraventricular hemorrhage (IVH)
  - ↑ T1 signal; ↓ SWI/T2* GRE
  - Best imaging modality for detection of associated parenchymal abnormalities

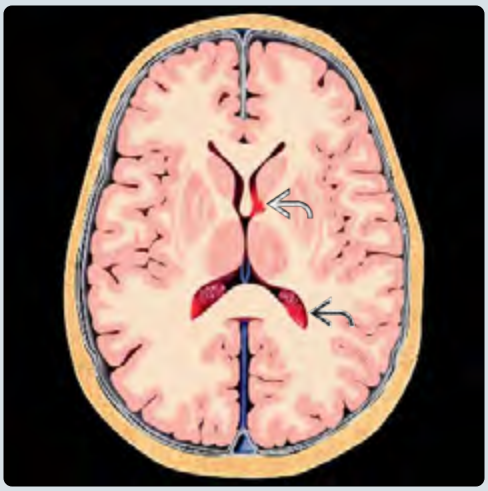
PATHOLOGY

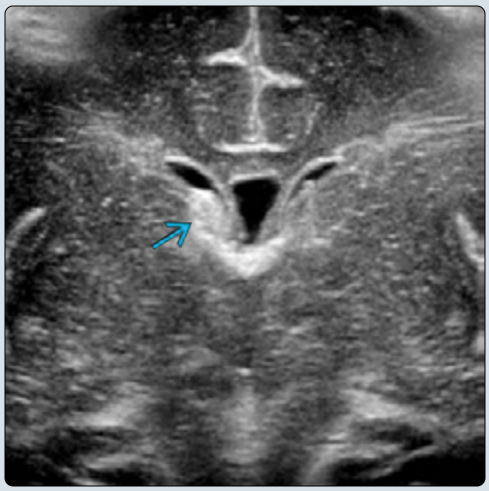
- Germinal matrix transiently present as region of fragile thin-walled vessels & migrating neuronal components
  - Involutes by 34 weeks of gestation
- In premature infants, perinatal stresses + poor cerebral autoregulation + germinal matrix → hemorrhage
- GMH-IVH grading system
  - Grade 1: GMH only
  - Grade 2: GMH + IVH, normal ventricle size
  - Grade 3: GMH + IVH + ventricular expansion
  - Grade 4: GMH-IVH + intraparenchymal hemorrhage
    - Venous compression leads to venous infarction

CLINICAL ISSUES

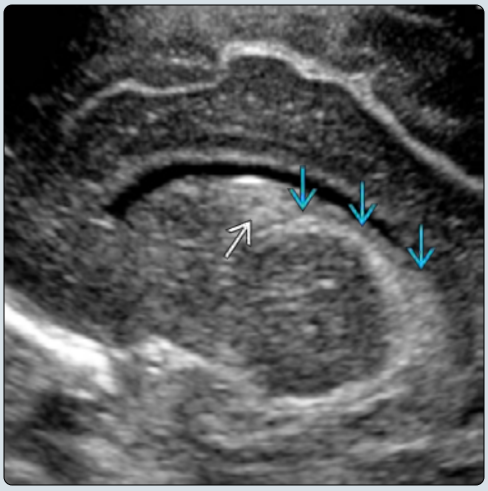
- Most common < 32 weeks of gestation & < 1,500 grams

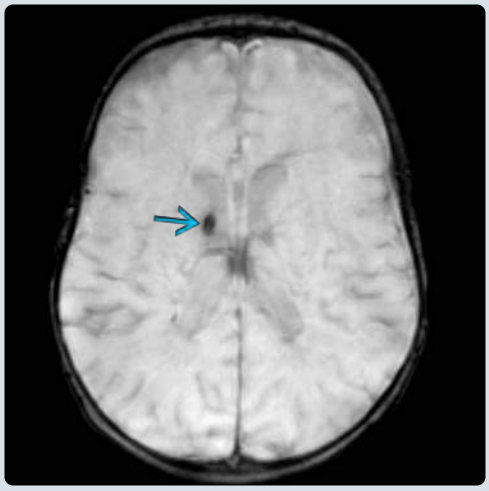
(Left) *Axial graphic shows a germinal matrix hemorrhage (GMH) (red) within the left caudothalamic groove. There is associated intraventricular hemorrhage (IVH) without ventriculomegaly, consistent with a grade 2 IVH.* **(Right)** *Coronal head US in a former 29-week male fetus 1 day after delivery shows an asymmetric globular echogenic focus in the right caudothalamic groove, consistent with a GMH. The lack of IVH makes this a grade 1 hemorrhage.*





(Left) *Sagittal head US in the same newborn shows increased echogenicity in the right caudothalamic groove, consistent with a grade 1 hemorrhage. Note the anterior tapering of the normal echogenic choroid plexus up to the caudothalamic groove.* **(Right)** *Axial SWI MR in the same patient at 3 months of age shows a focus of signal loss in the right caudothalamic groove, consistent with a remote GMH. There is no signal loss in the ventricles otherwise to suggest a component of IVH.*





TERMINOLOGY

Abbreviations

- Germinal matrix hemorrhage (GMH)

Synonyms

- Germinal matrix bleed, preterm caudothalamic hemorrhage, GMH-intraventricular hemorrhage (IVH)

Definitions

- Hemorrhage that occurs in very specific, richly vascular, stress-sensitive areas of premature brain

IMAGING

General Features

- Best diagnostic clue
  - Globular echogenic focus in caudothalamic groove, clearly asymmetric from contralateral normal choroid plexus
- Location
  - Caudothalamic groove
  - May extend posteriorly into ventricular system
    - Adherent to choroid plexus or layering in occipital horns
- Size
  - Variable
- Morphology
  - Globular & rounded when limited to caudothalamic groove (grade 1)
  - Lobulated or amorphous with posterior intraventricular extension (grade 2)

Ultrasonographic Findings

- Grayscale ultrasound
  - Hyperechoic acute hemorrhage; evolving clot retracts & becomes iso- to hypoechoic
  - IVH often adherent to choroid plexus
    - Fluid-debris levels may be visible in dependent ventricular occipital horns
    - Chemical ventriculitis in 2-3 days after IVH
      - Ependymal lining becomes thick & echogenic
  - Larger IVH will expand ventricle(s) (grade 3)
    - Further ventriculomegaly gradually develops due to
      - "Posthemorrhagic" hydrocephalus from obstruction of ventricular outlets & arachnoid granulations
      - White matter volume loss
  - Fan-shaped echogenic parenchymal focus next to site of grade 3 IVH due to venous hemorrhagic infarction (grade 4)
  - Examine remainder of brain for
    - Associated white matter injury
    - Lack of congenital anomalies
- Color Doppler
  - Useful to differentiate avascular hemorrhage from vascular choroid plexus

MR Findings

- Very helpful in identifying
  - Associated brain parenchymal injury that often accompanies GMH-IVH
    - White matter injury of prematurity
    - Cerebellar microhemorrhage
  - Underlying congenital anomalies
- Variable signal intensity based on age of blood
  - **T1WI**: ↑ signal in subacute blood, ↓ signal in chronic hemosiderin
  - **T2WI**: ↓ hypointense signal in most phases of hemorrhage evolution
  - **SWI**: Most sensitive sequence for identifying small or remote GMH-IVH

CT Findings

- Not typically used in premature neonate due to availability of US & superior soft tissue resolution of MR

Imaging Recommendations

- Best imaging tool
  - US primary imaging modality
    - Portability allows US to go to patient in NICU
    - Available sonographic window of anterior fontanelle
    - Lack of ionizing radiation
    - High sensitivity & specificity for GMH-IVH
  - MR useful adjunctive imaging modality
    - More sensitive, specific, & reproducible than US but not always practical in critically ill premature infants
    - MR rarely performed acutely; usually performed to evaluate extent of parenchymal injury
- Protocol advice
  - US: Use small-footprint, high-frequency linear transducer with multiple focal zones
    - Coronal & sagittal cine clips through ventricles particularly helpful in separating hemorrhage from normal choroid plexus
  - MR: Include SWI or T2* GRE to identify hemorrhage

DIFFERENTIAL DIAGNOSIS

Hypoxic-Ischemic Encephalopathy

- Deep or peripheral patterns of parenchymal injury due to perinatal insult to preterm or term infant
- Abnormal echogenicity of affected deep gray structures &/or white matter
- Often bilateral, may be asymmetric

Hemorrhage Infarction From Venous Thrombosis

- Superficial or deep venous thrombosis may lead to echogenic parenchyma ± discrete hematoma in neonate
  - Can liquefy & mimic abscess
- May be isolated or secondary to infection or other systemic processes

Choroid Plexus Cysts or Hematoma

- Small cysts &/or hemorrhages may occur in choroid plexus, sparing germinal matrix
- Often without consequence

Ventriculitis

- May be seen from infectious, toxic, & metabolic disorders without bleeding
- May lead to communicating hydrocephalus

Periventricular Calcifications

- Not typically seen in caudothalamic groove

- Related to TORCH infections most commonly

Periventricular Gray Matter Heterotopia

- Nodular heterotopia less echogenic than blood, isoechoic to brain
- Follows gray matter on all sequences

Tuberous Sclerosis

- Echogenic nodular periventricular foci
- Echogenic cortical/subcortical tubers distorting overlying gyri

PATHOLOGY

General Features

- Etiology
  - Germinal matrix only transiently present as region of thin-walled vessels, migrating neuronal components, & vessel precursors
    - Involutes by 34 weeks of gestation, such that hemorrhage becomes unlikely after this age
    - Most GMHs occur in 1st week of life
  - Germinal matrix prone to hemorrhage in premature infants due to
    - High density of fragile vascularized tissue
    - Poor cerebral autoregulation in premature infants
  - Hemorrhage occurs secondary to perinatal stresses: Labile blood pressure, hypoxia, hypercarbia, etc.

Staging, Grading, & Classification

- Germinal matrix grading system created by Burstein et al. in 1979
  - Performed on initial head US within days of delivery
  - Grading not changed on follow-up exams (even with progression)
- Grade 1: GMH only
- Grade 2: GMH, IVH, normal ventricle size
- Grade 3: GMH, IVH, & ventricular expansion from IVH
  - Secondary hydrocephalus occurring several days after grade 2 IVH should not be mislabeled as grade 3 IVH
- Grade 4: IVH + intraparenchymal hemorrhage
  - Intraparenchymal hemorrhage represents hemorrhagic venous infarction, not extension of GMH or IVH into parenchyma
    - Deep draining vein compression → venous congestion → venous infarction → hemorrhagic infarction
    - Debated whether or not ventricular dilation necessary
      - Is periventricular hemorrhagic infarction of premature infant with grade 1 or 2 GMH actually due to GMH vs. discrete focus of hypoxic-ischemic injury or venous thrombosis
  - Grade 4 may be called "posthemorrhagic venous infarction"

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Variable: Hypotonia, seizures, hyperreflexia, falling hematocrit, irritability, failure to thrive, paresis, acidosis, feeding difficulties
- Other signs/symptoms
  - GMH may occur in utero & follow same pathway of evolution & complications

Demographics

- Age
  - Premature infants in 1st week of life
  - 1/3-1/2 of all GMHs occur on 1st day of life
- Ethnicity
  - No predisposition
- Epidemiology
  - Most common in infants < 32 weeks of gestation & < 1,500 grams
  - Higher risk of GMH in premature infants with congenital heart disease, surgical procedures, severe respiratory distress
  - Incidence of GMH-IVH: 50% from 1975 to 1980 & then ↓ to ~ 15% after 2005
    - Improved modern outcomes attributed to prenatal steroids & surfactant, among other therapies

Natural History & Prognosis

- GMHs may progress or rebleed
- In general, clot retracts, lyses, & becomes hypoechoic, leaving behind cyst or area of porencephaly
- Prognosis
  - Grade 1 & 2 bleeds generally have good prognosis
  - Grade 3 & 4 bleeds have variable long-term deficits
    - Spastic diplegia, seizures, developmental delay

Treatment

- Prenatal
  - Prevent preterm delivery
  - Prenatal steroids
- Postnatal
  - Optimize neonatal resuscitation
  - Reduce fluctuations in cerebral blood flow
    - Minimize handling, gentle/synchronized ventilation, prompt treatment of patent ductus arteriosus, maintain normal oxygenation
- Posthemorrhagic hydrocephalus often requires CSF shunting

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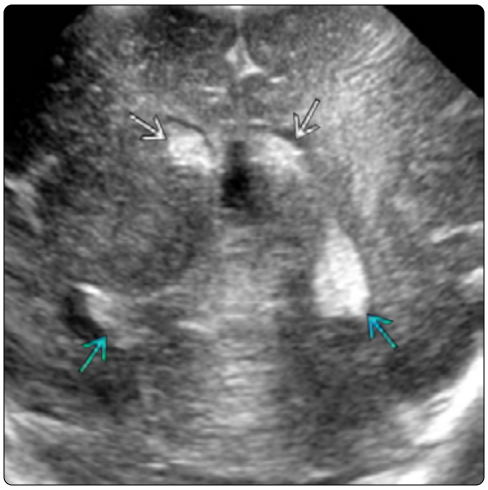
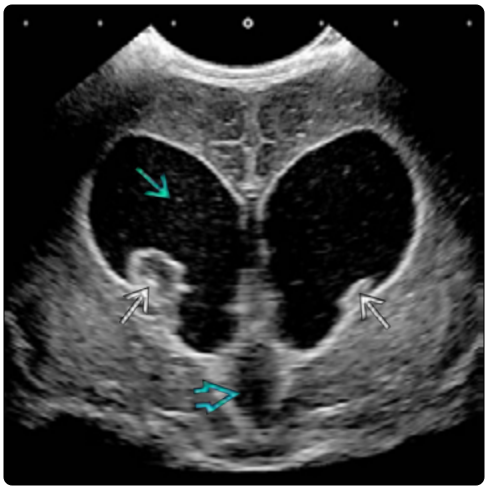
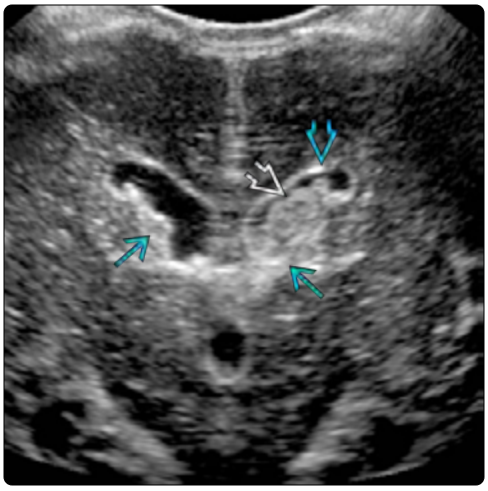
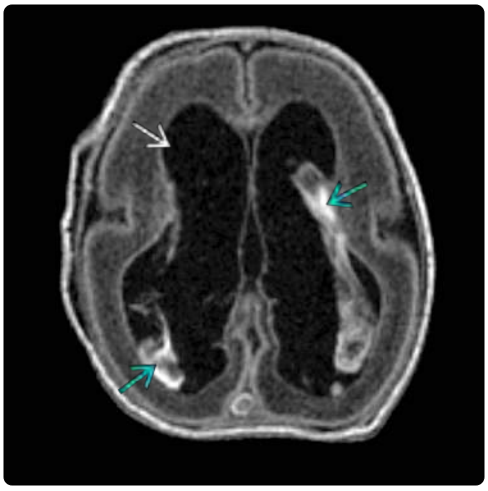


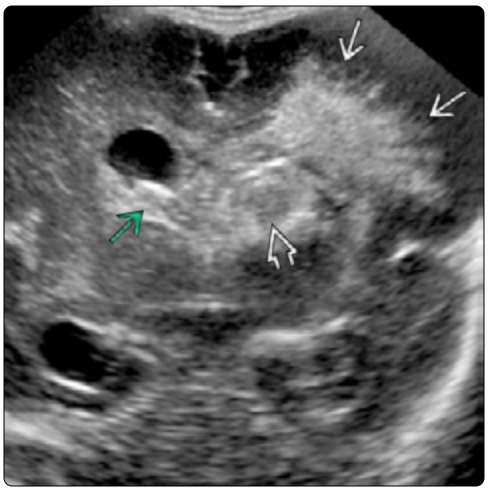


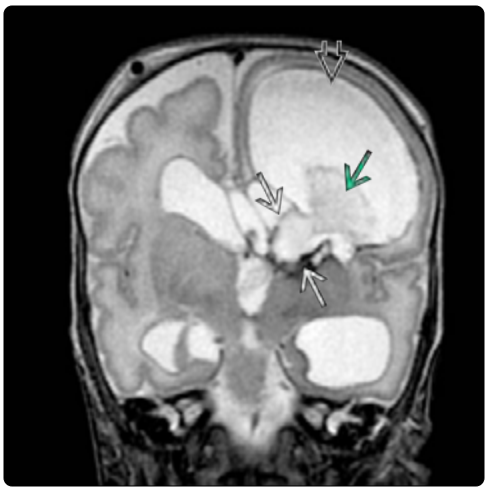
(Left) *Coronal head US in a former 30-week premature infant < 24 hours after delivery shows bilateral GMH ➡ & IVH ⇨ without ventriculomegaly, consistent with grade 2 IVH. Cine clips are the easiest way to distinguish GMH/IVH from normal choroid plexus (which is not easily done on this static image).* **(Right)** *Coronal head US in the same patient 3 weeks later shows evolution of the IVH ➡ with interval enlargement of the lateral ⇨ & 3rd ⇨ ventricles, consistent with posthemorrhagic hydrocephalus.*





(Left) *Coronal head US in a former 24-week premature infant (now 6 days old) shows bilateral GMHs ⇨. There is left intraventricular extension of the hemorrhage ➡ with expansion of the left frontal horn ⇨, consistent with a grade 3 IVH. A grade 2 IVH was seen on the right.* **(Right)** *Axial T1 MR 3 weeks later in the same patient shows foci of T1 shortening from the prior IVHs ⇨. Further ventriculomegaly ➡ has developed, consistent with posthemorrhagic hydrocephalus.*





(Left) *Coronal head US in a former 28-week premature infant (now 3 days old) shows a right GMH ⇨ & a left IVH, which fills & expands the lateral ventricle ➡. There is a fan-shaped hemorrhagic infarction ➡ in the left centrum semiovale, consistent with a grade 4 IVH.* **(Right)** *Coronal T2 MR in the same infant 6 weeks later shows residua of the left-sided IVH ➡ & intraparenchymal hemorrhage ⇨ with the development of cystic encephalomalacia ⇨, an expected evolution of grade IVH.*

White Matter Injury of Prematurity

KEY FACTS

TERMINOLOGY

- White matter (WM) injury of prematurity
- Brain injury occurring before 34 weeks gestation resulting in loss of periventricular WM

IMAGING

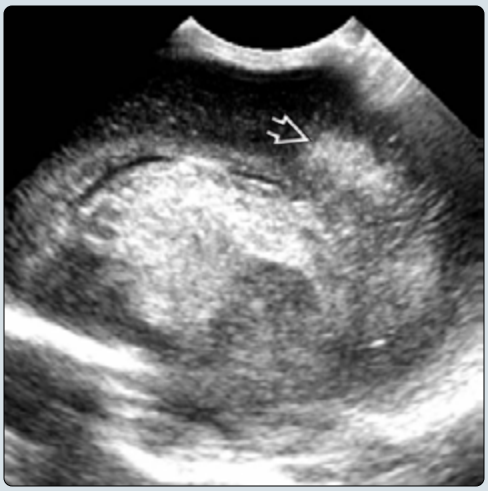
- Ultrasound: Reliable for more severe or late disease, less reliable for mild/moderate or early disease
  - Acute findings
    - Patchy, globular foci of ↑ echogenicity in periventricular/deep WM
  - Subacute/chronic findings
    - Clusters of periventricular cysts
- MR: Reliable for entire spectrum of disease
  - Acute findings
    - Hyperintense T1, hypointense T2 foci
    - Hypointense SWI/GRE foci with hemorrhage
    - ↓ ADC (may miss if imaging < 24 hours or > 5 days)
    - MRS: Lactate peak or ↑ excitatory neurotransmitters
  - Subacute findings
    - Periventricular cysts
  - Chronic finding
    - Periventricular/deep WM volume loss
    - Typically minimal associated gliosis

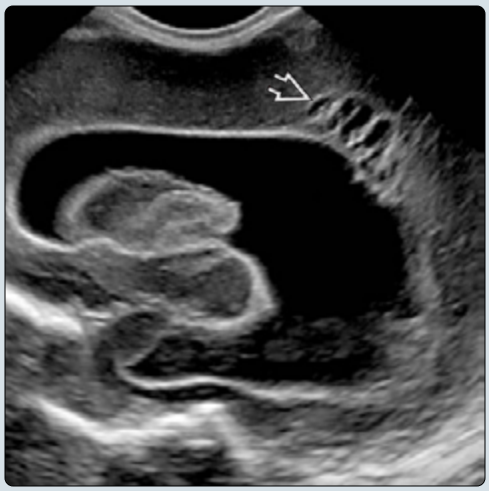
TOP DIFFERENTIAL DIAGNOSES

- Normal periventricular halo on ultrasound
- Infection
- Shunted hydrocephalus

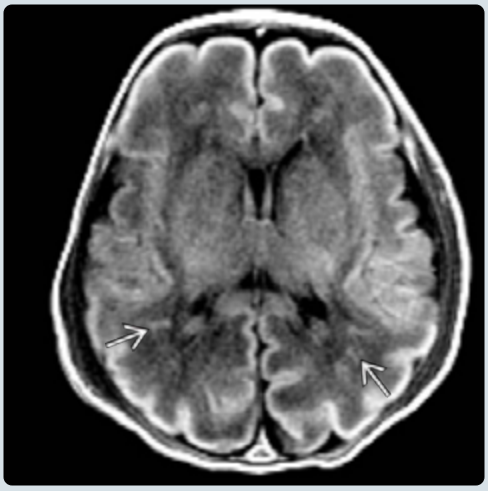
PATHOLOGY

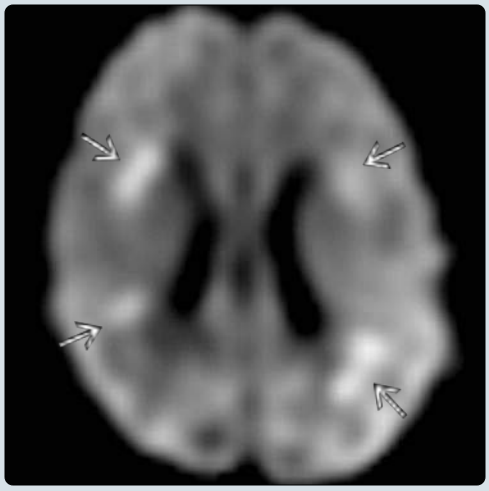
- Preceding inflammation (e.g., chorioamnionitis) + ischemia superimposed on vulnerable premature brain
- Characteristic pattern of injury reflects distribution of immature oligodendrocytes during vulnerable period
  - Selective injury to immature oligodendrocytes dramatically reduces WM volume in affected areas





(Left) *Sagittal head ultrasound of a 5-day-old former 27-week gestational age (GA) infant. There is globular increased echogenicity ➡ in the periventricular white matter, consistent with ischemic injury. Intraventricular hemorrhage and deep gray nuclei insult were also noted.* **(Right)** *Sagittal head ultrasound of the same infant 1 month later. The previously seen focus of increased echogenicity has now evolved into cystic encephalomalacia ➡. The new ventriculomegaly is likely due to volume loss and obstruction by blood products.*





(Left) *Axial T1 MR in an 18-day-old former 33-week GA neonate with tricuspid atresia demonstrates patchy foci of hyperintense signal ➡ in the bilateral parietooccipital periventricular white matter.* **(Right)** *Axial DWI in the same 18-day-old former 33-week GA neonate with tricuspid atresia shows corresponding foci of restricted diffusion ➡ (confirmed to have ↓ signal on the ADC map, not shown) in the bilateral periventricular white matter, consistent with ischemic injury.*

TERMINOLOGY

Abbreviations

- Periventricular leukomalacia (PVL)

Synonyms

- White matter (WM) injury of prematurity, perinatal white matter damage, punctate WM lesions

Definitions

- Perinatal WM injury occurring before 34 weeks gestation

IMAGING

Ultrasonographic Findings

- Reliable for more severe or late disease, less reliable for mild/moderate or early injury
- Acute findings
  - Patchy, globular foci of ↑ echogenicity in periventricular/deep WM
  - Loss of normal WM echotexture
- Subacute findings
  - Periventricular cysts

CT Findings

- Reliable for more severe or late disease, less reliable for mild/moderate or early injury
- Acute findings
  - ↓ attenuation in periventricular/deep WM
- Chronic findings
  - WM volume loss → angular ventricular margins

MR Findings

- T1WI
  - Acute: Areas of T1 hyperintensity
  - Chronic: WM volume loss, callosal thinning
- T2WI
  - Acute: Areas of T2 hypointensity
  - Subacute: Cystic changes in periventricular WM
  - Chronic: WM volume loss, ventriculomegaly
    - Cortical ribbon extending down to ventricular margin
    - Angular ventricles with "squared-off" trigones
- FLAIR
  - Chronic: Hyperintense signal in periventricular/deep WM
- T2* GRE
  - Hypointense SWI/GRE foci in areas of hemorrhage
- DWI
  - Reduced diffusion can precede ultrasound and MR abnormalities; ADC values may "normalize" after 5 days
- MRS
  - Lactate peak, ↓ NAA, ↑ excitatory neurotransmitters; alterations may antedate MR image abnormalities

Imaging Recommendations

- Best imaging tool
  - Ultrasound in perinatal period
    - Convenient and well tolerated by neonates
    - Poor sensitivity/specificity for WM injury, especially for mild, early, or noncavitary lesions
  - MR (including DWI and MRS)
    - Best performed 1-5 days after suspected injury

DIFFERENTIAL DIAGNOSIS

Normal Periventricular Halo

- Specular reflections of normal WM tract condensations
- Visualized in posterior periventricular WM but less echogenic than choroid plexus, not globular

Infection

- CMV: Microcephaly, periventricular Ca^{2+}, ± periventricular WM abnormalities, ± polymicrogyria

Shunted Hydrocephalus

- Ventricular distortion ± abnormal WM after shunting

PATHOLOGY

General Features

- Etiology
  - Predisposing factors of premature brain: Unique cellular vulnerability, impaired cerebrovascular autoregulation, periventricular arterial end zones
  - Superimposed insult
    - Chorioamnionitis → vasculitis of chorionic plate → ↑ inflammatory cytokines
    - With pre-/perinatal hypoxia → WM damage via inflammatory response, oxidative stress linked to reoxygenation during perinatal period

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - May be clinically silent initially ± EEG findings
  - Spastic diplegia, visual and cognitive impairment
- Clinical profile
  - Risk factors for WM injury of prematurity
    - Pregnancy: Low gestational age/weight, previous preterm birth, spontaneous preterm labor
    - Intrapartum: Preeclampsia, premature rupture of membranes, chorioamnionitis, group B streptococcus infection
    - Perinatal: Respiratory distress, PDA, ↓ PaCO2, sepsis, anemia, apnea, bradycardia, cardiac arrest

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Hypoxic-Ischemic Encephalopathy

KEY FACTS

TERMINOLOGY

- Brain injury in neonate caused by hypoxic ischemic insult

IMAGING

- US may be used for screening, particularly in acute setting or with concern for hemorrhage
- MR best imaging test for parenchymal injury
- Deep or central pattern of injury
  - Basal ganglia, thalamus, ± brainstem
- Peripheral pattern of injury
  - Injury to watershed zones of hemispheres
- Patterns may overlap with combination of patterns
- **T1WI & T2WI**: Usually normal in 1st few days
  - T1WI: ↑ signal in affected regions
  - T2WI: ↑ signal in affected regions
- **DWI**: Best sequence to define extent of injury
  - ↑ DWI, ↓ ADC in affected regions from 2-7 days
- **MRS**: ↓ NAA & ↑ lactate in affected areas
- **PWI**: ↑ ASL in affected deep gray nuclei during 1st week

PATHOLOGY

- Central pattern due to severe hypoxia of relatively brief (10-25 minutes) duration
  - Injury to regions of greatest metabolic demand
- Peripheral pattern due to less severe hypoxia over longer period of time
  - Regions of greatest metabolic demand protected
- Preterm infants more susceptible to equivalent ischemic injury compared to term infants
- Preterm neonates more likely to have associated white matter injury & hemorrhage

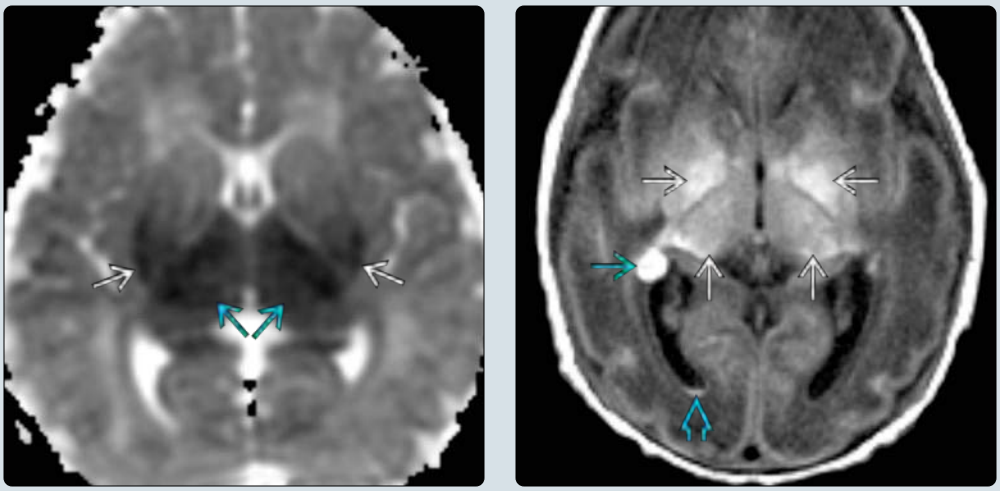
CLINICAL ISSUES

- Deep pattern associated with dyskinetic cerebral palsy
- Peripheral pattern associated with spastic cerebral palsy

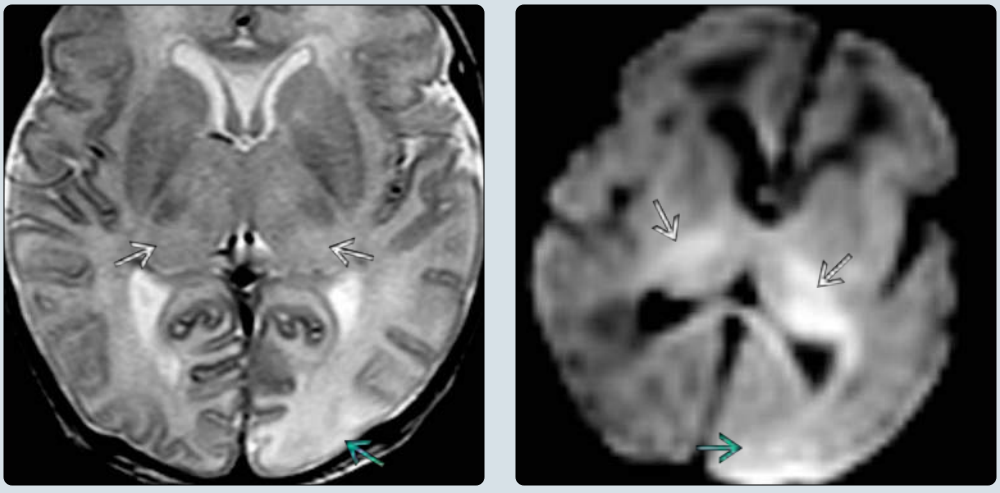
DIAGNOSTIC CHECKLIST

- Findings may be inapparent/subtle in first 24 hours
- Image at 2-7 days for best MR evaluation of injury extent

(Left) *Axial ADC MR in a 3-day-old term male delivered via emergent C-section (with Apgar scores of 1, 1, & 1) shows diffusion restriction in the thalami & posterolateral putamina, consistent with the deep injury pattern of profound HIE.* **(Right)** *Axial T1 MR in a 31-week premature infant 11 days following placental abruption shows irregular hyperintense signal in the bilateral thalami & nuclei of the globus pallidus. Also note the GMH & small layering IVH, findings often seen in premature infants with HIE.*



(Left) *Axial T2 MR in a 4-day-old term male delivered by emergent C-section for fetal bradycardia shows increased signal with loss of cortical differentiation in the left occipital lobe + ↑ signal in the thalami.* **(Right)** *Axial DWI MR in the same 4-day-old boy best demonstrates the full extent of injury with definite involvement of the bilateral thalami & the left occipital lobe suggesting a more severe injury with components of both the central & peripheral patterns.*



TERMINOLOGY

Definitions

- Hypoxic ischemic encephalopathy (HIE): Neonatal brain injury caused by perinatal hypoxic ischemic insult

IMAGING

General Features

- 2 patterns of injury have been described
  - Deep or central pattern
    - Injury to basal ganglia, thalami, ± brainstem
    - Profound hypoxic-ischemic injury (e.g., asystole)
    - More severe motor/cognitive outcomes
  - Peripheral pattern
    - Injury to watershed zones of cerebral hemispheres
    - Prolonged partial ischemia (e.g., fetal bradycardia)
    - Less severe motor/cognitive outcomes
  - Can overlap with combination of patterns
  - Late findings: Atrophy, gliosis/encephalomalacia

CT Findings

- Loss of gray-white differentiation
- Indistinct, hypodense deep gray nuclei
- Parenchymal Ca^{2+} & volume loss months after injury

MR Findings

- **T1WI**: Usually normal during 1st few days
  - ↑ signal in affected areas after ~ 2-3 days
  - Loss of normal bright T1 signal in posterior limb of internal capsule
- **T2WI**: Usually normal during 1st few days
  - Indistinct deep gray nuclei & loss of cortical ribbon
  - ↑ signal in affected areas after ~ 2-3 days
- **PD**: ↑ signal in affected areas
  - May be more conspicuous than T1 & T2
- **SWI**: Hemorrhage uncommon in term neonates but common in preterm neonates with HIE
- **DWI**: Most important sequence
  - Restricted diffusion within 1-7 days
  - Pseudonormalization begins at 8-10 days
- **MRS**: ↓ NAA & ↑ lactate in affected areas
  - ↑ Lac:NAA ratio correlates with more severe injury
- **PWI**: ↑ ASL in affected deep gray nuclei during 1st week

Ultrasonographic Findings

- Findings often less conspicuous compared to MR
- Slit-like lateral ventricles & sulcal effacement
  - Not specific for injury in newborn; ↓ Doppler resistive index in anterior cerebral artery supports injury
- Accentuated gray-white matter (WM) differentiation (with patchy, coarse ↑ echogenicity of WM)
- Hyperechoic foci in deep gray nuclei

Imaging Recommendations

- Best imaging tool
  - MR with DWI & MRS
- Protocol advice
  - Findings may be inapparent/subtle in first 24 hours
  - Image at 2-7 days for best MR evaluation of injury extent
    - ADC changes peak at ~ 5 days

DIFFERENTIAL DIAGNOSIS

Urea Cycle Disorders

- Elevated ammonia, present at 24-28 hours
- Diffuse cerebral edema

Maple Syrup Urine Disease

- Edema in myelinated areas
- Brainstem, perirolandic & cerebellar WM

Hypoglycemia

- Characteristic occipital cortex injury

Arterial Ischemic Stroke

- Focal arterial territory injury without hypoxic event

Venous Injury

- Edema, hemorrhage, or ischemia in venous distribution

TORCH Infections

- Microcephaly, migration anomalies, Ca^{2+}

PATHOLOGY

General Features

- Etiology
  - Central pattern associated with severe hypoxia of relatively brief (10-25 minutes) duration
    - Causative (sentinel) event usually identifiable
    - Injury to regions of greatest metabolic demand
  - Peripheral pattern associated with less severe hypoxia over longer period
    - Causative event often cryptic
    - Regions of greatest metabolic demand protected
    - Watershed regions susceptible
  - Preterm infants more susceptible to equivalent ischemic injury as compared to term infants
- Associated abnormalities
  - Maternal: Infection, preeclampsia, diabetes, cocaine

Staging, Grading, & Classification

- Sarnat stage (based on clinical & EEG findings)
  - I (mild): Hyperalert/irritable, mydriasis, ↑ heart rate, EEG normal
  - II (moderate): Lethargy, hypotonia, miosis, ↓ heart rate
  - III (severe): Stupor, flaccid, reflexes absent, seizures

CLINICAL ISSUES

Natural History & Prognosis

- Deep pattern associated with dyskinetic cerebral palsy
- Peripheral pattern associated with spastic cerebral palsy

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Childhood Stroke

KEY FACTS

TERMINOLOGY

- Acute alteration of neurologic function due to loss of vascular integrity

IMAGING

- NECT: ↓ attenuation of affected gray matter
- NECT: Insular ribbon sign → loss of distinct insular cortex
- NECT: Hyperdense middle cerebral artery (MCA) sign → thrombosed MCA
- MR: ↓ diffusion within ~ 30 minutes of arterial occlusion
- MR: Cytotoxic edema evident in affected territory on FLAIR/T2 by 4-6 hours after arterial occlusion
- MR: Enhancement of infarct typically occurs after 5-7 days
- CTA/MRA: Critical for early evaluation & identification of possible etiology (e.g., dissection, arteriopathy)
- MR perfusion imaging can provide valuable information regarding region at risk in setting of acute stroke
  - MR: Arterial spin labeling can provide useful perfusion information without contrast administration

TOP DIFFERENTIAL DIAGNOSES

- Seizure-related injury
- Acute encephalitis
- Mitochondrial encephalopathies
- Posterior reversible encephalopathy syndrome

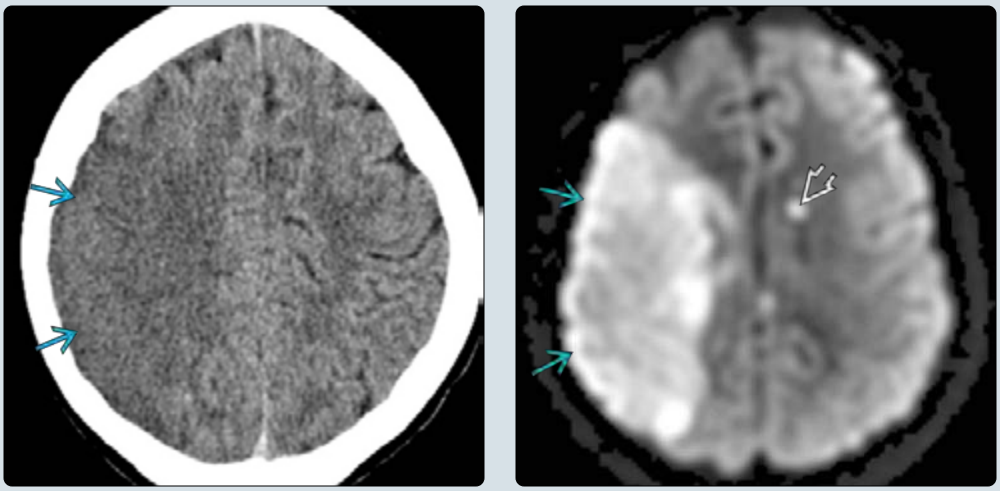
PATHOLOGY

- Major causes: Cardiac disease (~ 25%), moyamoya-type arteriopathy, dissection, vasculitis, hematologic/metabolic
- No underlying cause discovered in ~ 25% of cases

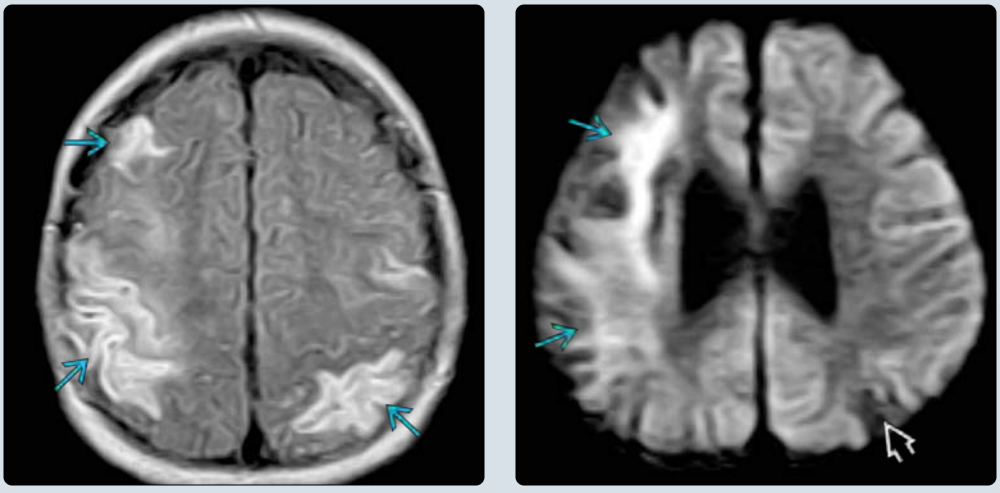
CLINICAL ISSUES

- Incidence: 2-3/100,000 per year in USA
  - Mortality: 0.6/100,000
- Children typically present later than adults (> 24 hours)
- Focal deficit may be masked by lethargy, coma, irritability
- Treatment in pediatric acute stroke usually conservative
- Capacity for recovery in children much > adults

(Left) *Axial NECT in a 15-year-old girl with dilated cardiomyopathy shows a large area of low attenuation in the right middle cerebral artery (MCA) territory. Note the sulcal effacement & loss of the gray-white matter differentiation.* **(Right)** *Axial DWI MR in the same patient confirms restricted diffusion in the right MCA territory. Also note the focus of restricted diffusion in the left periventricular region. Multiple infarcts in multiple vascular territories should raise suspicion of a proximal embolic source.*



(Left) *Axial FLAIR MR in a 2-year-old girl shows multiple areas of cytotoxic edema in both cerebral hemispheres in this patient with moyamoya-type vasculopathy.* **(Right)** *Axial DWI MR in the same 2-year-old girl with moyamoya-type vasculopathy shows diffusion restriction in the right frontoparietal foci of signal abnormality, suggesting an acute/subacute infarct. However, there is no diffusion restriction in the left parietal region, suggesting this infarct is of an older age.*



TERMINOLOGY

Definitions

- Acute alteration of neurologic function due to loss of vascular integrity
  - This chapter specifically addresses arterial ischemia beyond perinatal period

IMAGING

CT Findings

- NECT
  - ↓ attenuation of affected gray matter with loss of normal gray-white matter differentiation
    - ↓ in white matter attenuation less pronounced
    - Often wedge-shaped & localized to 1 arterial territory
  - Insular ribbon sign → loss of distinct of insular cortex
  - Hyperdense middle cerebral artery (MCA) sign → ↑ density of acutely thrombosed MCA
  - Hemorrhagic transformation (HT)
    - Symptomatic HT in 3%; asymptomatic HT in 30%
- CTA
  - Invaluable for demonstrating focal vascular abnormalities in acute setting

MR Findings

- **T1WI**: Acute: ↓ signal with gyral swelling
  - Chronic: ± ↑ signal in cortical laminar necrosis
- **T2WI**: Loss of flow void in thrombosed vessel
- **FLAIR**: ↑ signal with gyral swelling (within 4-6 hours)
  - Abnormal sulcal ↑ signal (climbing ivy sign) of chronic slow flow collaterals in setting of longstanding proximal vascular occlusion
- **DWI**: Most sensitive for early detection of ischemia
  - Acute: Restricted diffusion (↑ DWI, ↓ ADC signal) ≤ 30 minutes after ischemic insult
  - Subacute (7-14 days): Pseudonormalization of signal
  - Chronic: Facilitated diffusion in gliotic brain
- **T1WI C+**: Cortical & leptomeningeal enhancement seen after 5-7 days following acute infarct
  - Enhancing climbing ivy sign
- **MRA**: Can detect arterial occlusion & stenosis in large- & medium-sized cerebral vessels
- **PWI**: Provides valuable information about affected brain
  - Ischemic penumbra: ↓ perfusion, no DWI change (PWI-DWI mismatch)
- **MRS**: ↑ lactate hallmark of ischemia/infarct

Imaging Recommendations

- Best imaging tool
  - CT initial imaging test for signs/symptoms of stroke; excellent for excluding hemorrhagic stroke (more common in children vs. adults)
  - MR with DWI, MRA, PWI

DIFFERENTIAL DIAGNOSIS

Seizure-Related Injury

- Swelling & restricted diffusion secondary to persistent seizure activity
- Differentiation by clinical presentation & EEG

Acute Encephalitis

- Acute parenchymal inflammation secondary to infectious agents, typically viral
- Slower onset with encephalopathy

Mitochondrial Encephalopathies

- Symmetric basal ganglia involvement common
- Usually have manifestations beyond CNS

Posterior Reversible Encephalopathy Syndrome

- Patchy cortical/subcortical edema most common in parietal & occipital lobes, typically in setting of hypertension
- Diffusion restriction uncommon

PATHOLOGY

General Features

- 6 major causes of arterial stroke in children
  - Cardiac disease (~ 25%)
  - Moyamoya-type arteriopathy
  - Arterial dissection (e.g., trauma)
  - CNS vasculitis
  - Hematologic/metabolic (e.g., coagulopathy)
  - Idiopathic (~ 25%)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depends on patient age, etiology, & involved artery
    - < 1 year: Seizures, encephalopathy > focal neurologic
    - > 1 year: Usually focal neurologic (e.g., hemiplegia)
    - Speech difficulties, gait abnormality, seizure
    - Embolic cause: Sudden onset of symptoms
    - Stenoocclusive cause: Gradual/intermittent (e.g., TIA)
  - Focal deficit may be masked by lethargy, coma, irritability
- Children typically present later than adults (> 24 hours)

Demographics

- Epidemiology
  - Incidence: 2-3/100,000 per year in USA
    - Mortality: 0.6/100,000
  - Underrecognized as significant source of morbidity in pediatric population

Natural History & Prognosis

- Capacity for recovery better than in adults, due to
  - Better compensatory mechanisms, collateral recruitment, neuronal plasticity
  - Fewer concomitant risk factors

Treatment

- Clinical window of opportunity/benefit not as well understood in children as compared to adults
- Mainstay of chronic therapy for fixed vascular lesions & vasculopathies: Aspirin
- Transfusion therapy for at risk children with sickle cell

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Moyamoya

KEY FACTS

TERMINOLOGY

- Progressive narrowing of distal internal carotid artery (ICA) & proximal circle of Willis (COW) vessels → characteristic adjacent clusters of collateral flow appearing as "puff of smoke" on real-time angiography
- Moyamoya disease = primary (idiopathic) moyamoya
- Moyamoya arteriopathy (a.k.a. moyamoya syndrome or secondary moyamoya) due to other disorders

IMAGING

- Absent or narrowed distal ICA & abnormal COW
- Excessive tiny collaterals in basal ganglia & cisterns
  - "Puff of smoke" (moyamoya in Japanese) of lenticulostriate & thalamoperforator collaterals
- Prominent collaterals in sulci
  - Ivy sign on FLAIR & T1 C+ MR
- Acute & chronic infarcts
- CT/CTA: Acute use for ischemia or hemorrhage
- MR C+/MRA: Vascular protocol with DWI & perfusion
  - DWI: Helpful to identify "acute on chronic" injury

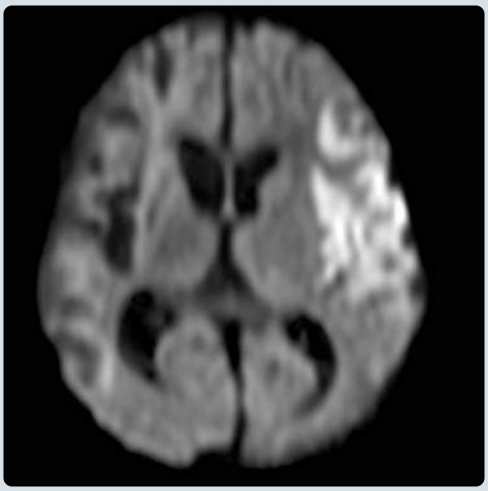
PATHOLOGY

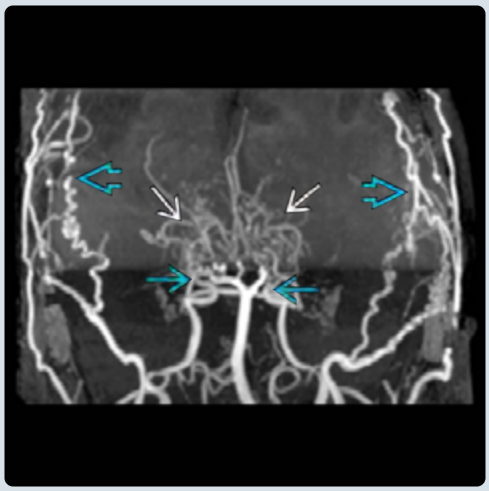
- Moyamoya disease: Inherited idiopathic disorder
- Moyamoya arteriopathy: Secondary process
  - Sickle cell disease, neurofibromatosis type 1, radiation therapy, trisomy 21, Alagille syndrome, morning glory syndrome, TB meningitis, among others

CLINICAL ISSUES

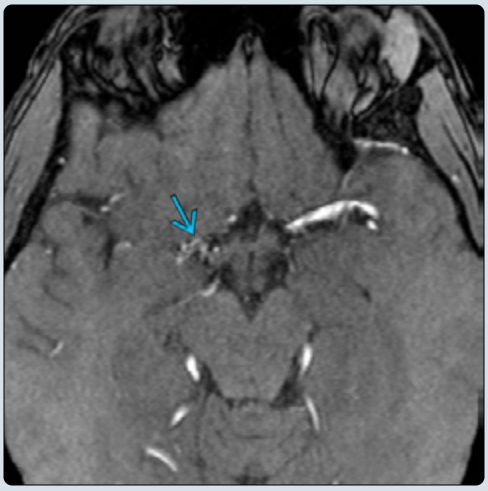
- Bimodal age peaks: 6 & 35 years
- Most frequent cause of stroke in Asian children
- Presentation (children): Transient ischemic attacks (TIAs), alternating hemiplegia (exacerbated by crying), headache
- Presentation (adults): TIAs, hemorrhage (~ 30%), & cerebral infarct
- Prognosis depends on etiology, ability to form collaterals, age/stage at diagnosis
- Treatment: Indirect (more common in children) or direct (more common in adults) vascular bypass

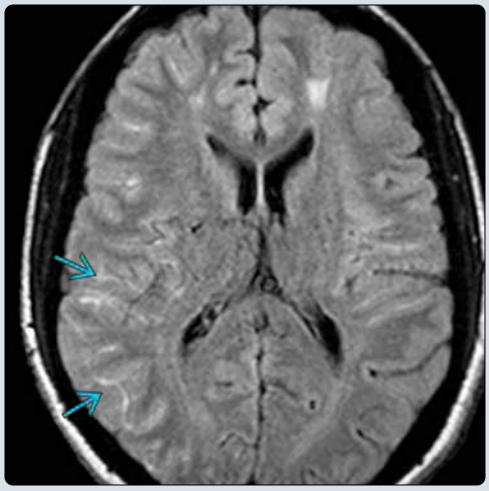
(Left) *Axial DWI MR in a 13 month old with increasing seizures shows left MCA distribution ischemia as well as a remote infarct in the right MCA territory. This is a typical acute on chronic ischemic pattern of moyamoya.* **(Right)** *Anterior 3D TOF MRA in the same patient at 8 years of age shows occlusions of the terminal ICAs, absence of the MCAs, & numerous lenticulostriate collaterals forming a "puff of smoke." The PCAs are also occluded. Note the enlarged ECA collaterals status post synangiosis & dural inversion.*





(Left) *Axial TOF MRA in a 14 year old with NF1 shows absence of the right internal carotid terminus & MCA. In the expected location of the carotid terminus & MCA, there are multiple small leptomeningeal collaterals.* **(Right)** *Axial FLAIR MR in a 21 year old with sickle cell disease shows high signal within the right MCA distribution sulci, the so-called ivy sign. This abnormal signal corresponds to engorged pial collateral vessels & is often seen in moyamoya.*





TERMINOLOGY

Definitions

- Progressive narrowing of distal internal carotid artery (ICA) & proximal circle of Willis (COW) vessels → characteristic adjacent clusters of collateral flow appearing as "puff of smoke" on real-time angiography
- Moyamoya disease: Primary (idiopathic) moyamoya
- Moyamoya arteriopathy (a.k.a. moyamoya syndrome or secondary moyamoya) occurs in association with other disorders or after radiation treatment

IMAGING

General Features

- Best diagnostic clue: Multiple enhancing punctate dots (CECT) & flow voids (MR) in basal ganglia & cisterns
- Arterial occlusions: Distal ICA, COW, branches
  - Anterior > posterior circulation
- Leads to prominent clusters of nearby collaterals
  - "Cloud-like" lenticulostriate & thalamoperforator collaterals on angiography: "Puff of smoke" ("moyamoya" in Japanese)
- Also leads to prominent sulcal collaterals distally

CT Findings

- NECT
  - Children: Acute ischemia ± old infarcts
  - Older children/adults: Usually ischemia but may present with intracranial hemorrhage
- CTA: Abnormal COW + basilar net-like collaterals

MR Findings

- **T2WI**: ↑ signal in gliotic areas from prior infarcts
  - Collateral vessels: Net-like cisternal flow voids
- **FLAIR**: Bright sulci = leptomeningeal ivy sign
  - Slow flowing engorged pial collateral vessels, thickened arachnoid membranes
  - Correlates with ↓ cerebral vascular reserve
- **SWI/T2* GRE**: Hemosiderin if prior hemorrhage
- **DWI**: Very useful for "acute on chronic" infarcts
- **T1WI C+**: Lenticulostriate collaterals → enhancing "dots" in BG & net-like thin vessels in cisterns
  - Leptomeningeal enhancement (ivy sign)
  - Vessel wall imaging shows concentric enhancement
- **MRA**: Narrowed/occluded distal ICA & COW vessels
- **PWI**: ↓ perfusion in affected territories
  - May be used to measure response to revascularization

Angiographic Findings

- Predominantly (not exclusively) anterior circulation
  - Narrow proximal COW & ICA (early phase)
  - Lenticulostriate & thalamoperforator collaterals (intermediate phase)
  - Transdural/transosseous external carotid artery (ECA)-ICA collaterals (late phase)
- Dilation & branch extension of anterior choroidal artery predicts adult hemorrhagic events

Imaging Recommendations

- Best imaging tool: MR C+/MRA
- Catheter angiography defines anatomy prior to bypass

DIFFERENTIAL DIAGNOSIS

Ivy Sign

- Leptomeningeal metastases, subarachnoid hemorrhage, meningitis, ↑ inspired oxygen, collateral veins of Sturge-Weber or other chronic venous occlusion

Large Vessel Inflammatory Vasculitis

- Postvaricella vasculitis, lupus, & other CNS vasculitides
- May be reversible with treatment

Severely Attenuated Circle of Willis

- Subarachnoid hemorrhage (spasm), meningitis, tumor encasement

PATHOLOGY

General Features

- Etiology
  - Moyamoya disease
    - Inherited polygenic or autosomal dominant
  - Moyamoya arteriopathy (a.k.a. moyamoya syndrome or secondary moyamoya)
    - Sickle cell disease, neurofibromatosis type 1, radiation therapy, trisomy 21, Alagille syndrome, morning glory syndrome, tuberculous meningitis, many others

Staging, Grading, & Classification

- Staging criteria (Suzuki)
  - Stage 1: Narrowing of ICA bifurcation
  - Stage 2: ACA, MCA, PCA dilated
  - Stage 3: Maximal basal collaterals; small ACA/MCA
  - Stage 4: Fewer collaterals (vessels); small PCA
  - Stage 5: Further ↓ in collaterals; absent ACA/MCA/PCA
  - Stage 6: Extensive ECA-pial collaterals

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Children: Transient ischemic attacks (TIAs), alternating hemiplegia (exacerbated by crying), headache
  - Adults: TIAs, hemorrhage (~ 30%), cerebral infarct

Natural History & Prognosis

- Pediatric cases usually advance to stage 5 in < 10 years
- Hemorrhagic moyamoya more common in older patients

Treatment

- Aspirin therapy
- Direct bypass: Superficial temporal artery (STA)-MCA more common in adults
- Indirect bypass
  - Pial synangiosis & encephaloduroarteriosynangiosis with STA more common in children
  - Dural inversion with middle meningeal artery

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3. Kim SK et al: Pediatric moyamoya disease: an analysis of 410 consecutive cases. Ann Neurol. 68(1):92-101, 2010

Vein of Galen Aneurysmal Malformation

KEY FACTS

TERMINOLOGY

- High-flow arteriovenous fistula (AV) between deep choroidal arteries & median prosencephalic vein (MPV) of Markowski
- Not actually aneurysm of true vein of Galen, which fails to form because of fistula

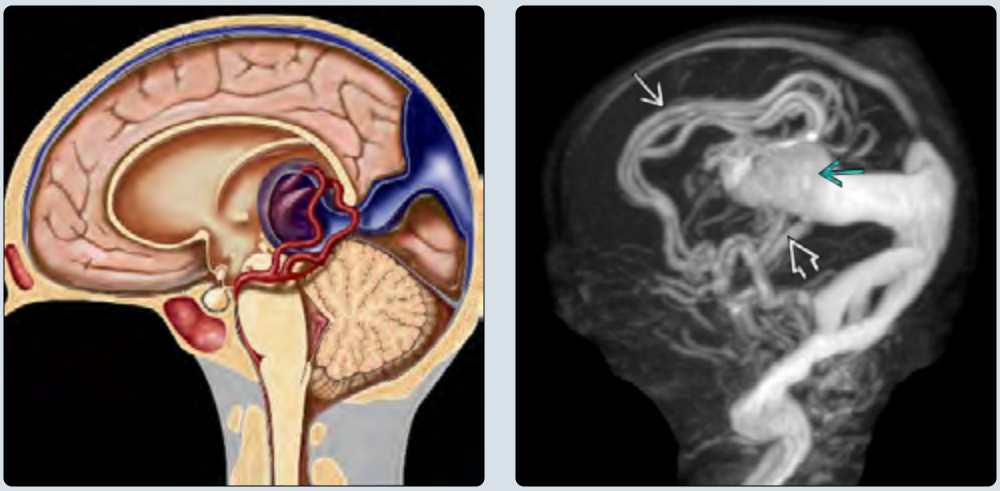
IMAGING

- 2 major goals of imaging
  - Assess degree of brain injury
  - Define VGAM architecture to help plan embolization
- Some degree of ventriculomegaly typical
  - ↓ CSF resorption due to venous hypertension
  - Chronic volume loss
    - Due to venous hypertension & arterial steal
  - Obstruction from compression of tectum
- Venous outflow stenosis affects end-organ damage
  - Sigmoid/jugular stenoses protect heart, harm brain
- Classification system for VGAMs (Lasjunias)
  - Choroidal type: Multiple primitive choroidal arteries pass through nidus before entering MPV
  - Mural type: 1 or more (usually multiple) direct AV fistulas within wall of MPV

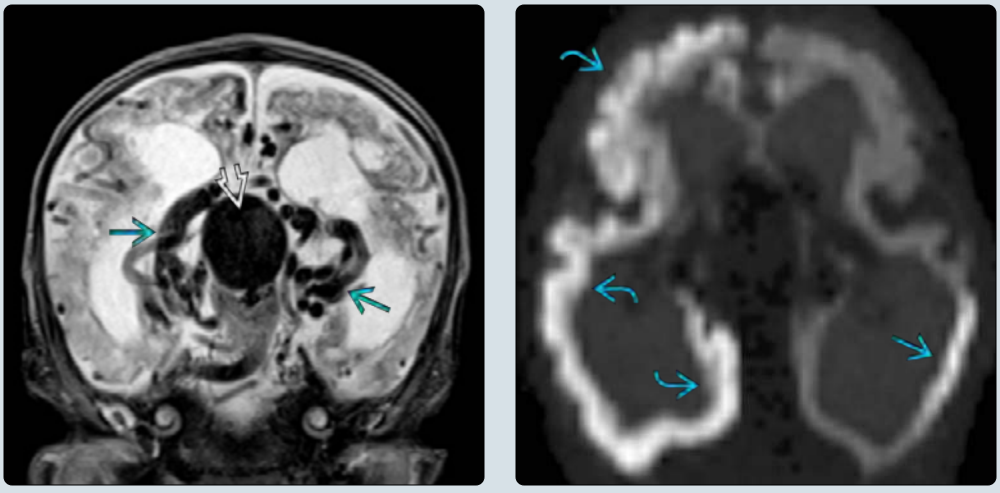
CLINICAL ISSUES

- Most common extracardiac cause of high-output heart failure in newborn
- Preferred treatment: Transcatheter embolization at 4-6 months with occlusion of fistula, ideally from arterial side
  - May require staged embolizations
- Bicêtre neonatal evaluation score guides therapy
  - Assesses end-organ damage
- Prognosis related to volume of shunt & timing/success of treatment
  - High-volume shunts requiring treatment in newborn period have worse prognosis
  - Ability to delay treatment until 4-6 months associated with better outcome

(Left) *Sagittal graphic shows medial & lateral posterior choroidal arteries with a fistulous connection to an enlarged midline vein [median prosencephalic vein (MPV)], which drains through a persistent falcine sinus to the superior sagittal sinus. This would be a mural-type vein of Galen malformation (VGAM).* **(Right)** *Lateral 3D MRA with contrast in a newborn with high-output cardiac failure shows enlarged choroidal & pericallosal arteries feeding a large MPV & straight sinus. No venous outflow stenosis is identified.*



(Left) *Coronal T2 MR in the same newborn with a large VGAM shows a large MPV & multiple enlarged tortuous feeding arteries. The brain parenchyma is thinned with loss of normal gray-white matter differentiation, a poor prognostic sign.* **(Right)** *Axial DWI MR in the same neonate shows extensive right & focal left cerebral diffusion restriction, representing severe brain injury ("melting brain") related to venous hypertension & arterial steal from the VGAM. No treatment was recommended for this patient.*



TERMINOLOGY

Synonyms

- Vein of Galen malformation (VGAM), vein of Galen aneurysm, galenic varix

Definitions

- High-flow arteriovenous (AV) fistula between deep choroidal arteries & median prosencephalic vein (MPV) of Markowski
  - Not actually aneurysm of true vein of Galen, which fails to form because of fistula

IMAGING

General Features

- 2 major goals of imaging
  - Assess brain injury to determine if treatment warranted
  - Delineate malformation architecture for embolization

CT Findings

- NECT
  - White matter Ca^{2+} from chronic venous ischemia
- CTA
  - Excellent preangiography delineation of vessels

MR Findings

- Large flow void of varix (unless slow/turbulent flow)
- Phase-encoding pulsation artifact from varix
- Prominent flow voids of feeding arteries around varix
- Some degree of ventriculomegaly typical; ± ↑ T2 signal of white matter
  - ↓ CSF resorption due to venous hypertension
  - Obstruction from compression of tectum
  - Chronic volume loss
- Restricted diffusion in areas of acute ischemia/infarction
  - Due to venous hypertension & arterial steal phenomenon
  - ± focal or extensive loss of cortical ribbon on T2WI
- MRV: High flow through veins → venous stenoses
  - Affects end-organ damage
    - Venous stenosis protects heart at expense of brain
    - Jugular stenosis/atresia associated with poor outcome
- MRA: Key to define architecture of lesion
  - Major arterial feeders: Targets for embolization
  - Unaffected by embolics (ideal for posttreatment evaluation)
- Perfusion: Arterial spin labeling can assess AV shunt reduction following embolization
- Fetal MR can identify malformation in 2nd & 3rd trimester & provide information about end-organ injury

Angiographic Findings

- Choroidal or mural classification (Lasjunias) based on angioarchitecture of VGAM
  - Choroidal type: Primitive vessel morphology with multiple choroidal feeding arteries passing through nidus network before draining into MPV
  - Mural type: 1 or more (usually multiple) direct AV fistulas within wall of MPV
- Frequent venous abnormalities
  - Embryonic falcine sinus drains MPV in ~ 50%
  - Stenoses at sigmoid-jugular junction

DIFFERENTIAL DIAGNOSIS

Secondary Vein of Galen Aneurysmal Dilation

- AV malformation drains into true vein of Galen

Childhood Dural Arteriovenous Fistula

- High-flow fistula with venous varices

Giant Aneurysm

- Not associated with venous abnormalities

PATHOLOGY

General Features

- Embryology
  - Abnormal connection of choroidal arteries to MPV occurs at 6-11 weeks gestation
  - Flow through fistula prevents normal regression of MPV

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Neonate: High-output CHF, cranial bruit
  - Infant: Macrocrania (hydrocephalus), seizures, or neurocognitive delay
  - Older children/adults: Usually compensated, may present with hemorrhage

Natural History & Prognosis

- Prognosis related to volume of shunt & timing/success of treatment
  - High-volume shunts requiring treatment in newborn period have worse prognosis
- Severe cases → ischemia & cerebral atrophy: "Melting brain"
  - Due to venous insufficiency & arterial steal

Treatment

- Treatment in neonatal period based upon Bicêtre neonatal evaluation score
  - 21 point system based upon cardiac (5), cerebral (5), respiratory (5), hepatic (3), & renal (3) function
  - High score → good function; low score → poor function
    - < 8: No treatment
    - 8-12: Emergent embolization
    - > 12: Medical management initially followed by delayed embolization at 4-6 months
- Transcatheter embolization
  - Permanent occlusion of fistula, ideally from arterial side
    - Liquid agents or coils
  - Outcome following embolization: 75% normal; 15% moderate retardation; 10% severe retardation

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2. Chow ML et al: Radiological and clinical features of vein of Galen malformations. J Neurointerv Surg. 7(6):443-8, 2015

KEY FACTS

TERMINOLOGY

- Vascular malformation with arteriovenous shunting through complex nidus of arterioles & venules (without intervening capillary bed)

IMAGING

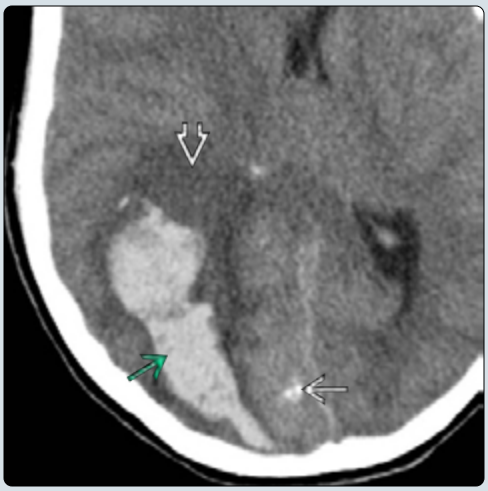
- **NECT**: Parenchymal hematoma most common
  - Small unruptured AVMs often not visible
  - May see Ca^{2+}
- **CTA**: Enhancing feeding arteries, nidus, & draining veins
  - May be negative if AVM compressed by hematoma
- **MR**
  - T1WI: Bright hemorrhage
  - T2WI: Prominent dark flow voids
  - T1 C+ (volumetric gradient): Enhancing abnormal vessels; important for micro-AVMs
- **MRA**: Gross depiction of AVM components
  - Flow-related signal in draining veins reflects AV shunt
- **Digital subtraction catheter angiography**
  - Gold standard with high temporal & spatial resolution
  - Best identifies all 3 components of AVM
    - Multiple arterial feeding vessels (internal-external carotid, vertebrobasilar)
    - Deep vs. superficial venous drainage
    - Associated arterial/nidus aneurysms, venous stenoses

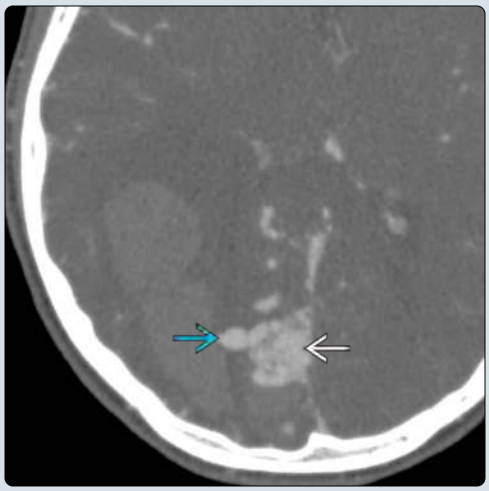
PATHOLOGY

- Most AVMs sporadic; small % related to hemorrhagic telangiectasia
- Spetzler-Martin surgical risk grading system (1-5 points)
  - Size: < 3 cm (1 point), 3-6 cm (2), > 6 cm (3)
  - Location: Eloquent (1) vs. noneloquent (0)
  - Venous drainage: Deep (1) vs. superficial (0)

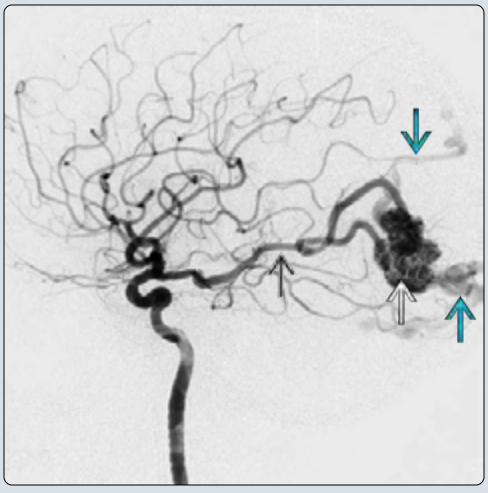
CLINICAL ISSUES

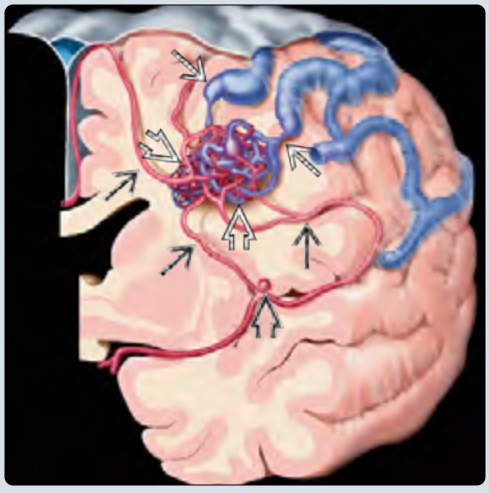
- Spontaneous parenchymal hemorrhage in child should be considered AVM until proven otherwise
- Annual bleeding risk = 2-4%





(Left) *Axial NECT in a 15-year-old girl who was found unconscious with dilated pupils shows extensive right posterior temporal & occipital intraparenchymal hemorrhage. Note the surrounding parenchymal edema. A few small calcific foci in the medial occipital lobe suggest an associated vascular malformation.* **(Right)** *Axial CT angiogram in the same patient shows a tightly packed central nidus with dilated peripheral veins, consistent with an arteriovenous malformation (AVM).*





(Left) *Lateral DSA image (right ICA injection) in the same 15-year-old girl shows the enlarged arterial feeder arising from the right ICA via the PCOM artery. The tightly packed nidus is well defined. Note the 2 separate superficial draining veins that opacify early during the arterial phase, typical of an AVM.* **(Right)** *Graphic shows a cerebral AVM with a centrum semiovale nidus fed from cortical & central arteries (with a proximal flow-related aneurysm) & drained by enlarged cortical veins.*

TERMINOLOGY

Definitions

- Arteriovenous malformation (AVM): High-flow vascular malformation with AV shunting through complex nidus of arterioles & venules (without intervening capillary bed)

IMAGING

CT Findings

- **NECT**: Unruptured AVM often occult, unless large; ± Ca^{2+}
  - Ruptured AVM: Spontaneous parenchymal hemorrhage
- **CTA**: Enlarged feeding arteries, nidus, draining veins
  - Nidus may be compressed by hematoma

MR Findings

- **T1WI**: Hematoma in ruptured AVM typically of ↑ signal
- **T2WI**: Arterial feeder, nidal, & draining vein flow voids
- **FLAIR**: May see adjacent gliosis, especially if prior bleed
- **SWI/T2* GRE**: Blooming with hemorrhagic blood products
  - Hyperintense (arterial) SWI signal in draining veins
- **DWI**: Usually normal on initial imaging studies
  - ± ischemic complications following intervention
- **T1WI C+**: Avid enhancement of nidus & draining veins with GRE T1 (not spin echo)
  - Important for small peripheral AVMs
- **MRA/MRV**: Gross depiction of AVM components
  - Flow-related signal in draining veins reflects AV shunt
- **ASL**: High signal in AVM nidus & draining veins
  - Sensitive sequence for detection of micro-AVMs

Angiographic Findings

- DSA depicts 3 components of AVMs with high spatial & temporal resolution
  - Arterial feeders
    - Enlarged; often from multiple arteries (internal-external carotid, vertebrobasilar)
    - Feeding artery aneurysms in 10-15%
  - AVM nidus
    - Central tangle of vessels where AV shunting occurs
    - Small aneurysms within nidus (~ 50%)
  - Venous drainage
    - Enlarged, tortuous veins
    - Must identify drainage as deep &/or superficial
    - ± venous varix, which indicates distal stenosis

Imaging Recommendations

- Best imaging tool
  - CTA rapidly obtained with acute presentation; typically provides diagnosis & adequate detail to direct therapy
- Protocol advice
  - If initial study negative, repeat when hematoma resolves
    - Small AVM may be compressed by initial hematoma

DIFFERENTIAL DIAGNOSIS

Cavernous Malformation

- "Angiographically occult" lesion
- Popcorn appearance on MR with blood-fluid levels, hemosiderin staining, ± Ca^{2+}

Hemorrhagic Tumor

- Often enhances on MR; tumor blush on DSA
- No enlarged draining veins

Arteriovenous Fistula

- Direct AV shunting without nidus

Arterial Aneurysm

- No nidus or enlarged draining veins
- Subarachnoid blood common

PATHOLOGY

General Features

- Etiology
  - Most AVMs sporadic; can develop in previously normal regions of brain
- Associated abnormalities
  - Syndromic AVMs (2% of cases)
    - HHT: 40-50% are micro-AVMs of brain
      - Additional lung & hepatic AVMs; cutaneous & mucosal telangiectasias

Staging, Grading, & Classification

- Spetzler-Martin scale predicts surgical morbidity: ↑ score = ↑ risk of open surgery
  - Size: < 3 cm (1 point), 3-6 cm (2), > 6 cm (3)
  - Location: Noneloquent (0) or eloquent (1)
  - Drainage Superficial (0) or deep (1)

CLINICAL ISSUES

Presentation

- ~ 50% acute hemorrhage (headache, ↓ consciousness)
  - Spontaneous parenchymal hemorrhage in child should be considered AVM until proven otherwise
- ~ 25% seizure; ~ 15% focal neurologic findings; ~ 10% incidental

Natural History & Prognosis

- Annual bleeding risk = 2-4%
- Factors that ↑ annual bleeding risk
  - Prior AVM rupture; exclusive deep venous drainage; deep brain location

Treatment

- Acute surgical decompression often required for mass effect from hematoma
- Optimal treatment for underlying AVM based on variety of factors; options include
  - Microsurgical resection
  - Endovascular embolization
  - Stereotactic radiosurgery
- Recent trial (ARUBA-2014) suggested medical management may be superior to interventional therapy
  - Controversial study due to short time course (3.3 years) & methodologies

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Cavernous Malformation

KEY FACTS

TERMINOLOGY

- Benign vascular lesion with dilated sinusoids lined by thin immature walls & containing blood products of various ages

IMAGING

- Occur throughout CNS
- Vary in size from microscopic to giant (> 6 cm)
- **CT**: Hyperdense lesion ± Ca^{2+}; 50% not visible
- **MR**: Heterogeneous core with T2 hypointense rim
  - Popcorn ball appearance with internal fluid-fluid levels
  - Adjacent ↑ FLAIR signal suggests recent hemorrhage
  - SWI/T2* GRE most sensitive for small lesions

TOP DIFFERENTIAL DIAGNOSES

- Diffuse axonal injury
- Intracerebral hematoma
- Neoplasm
- Capillary telangiectasia

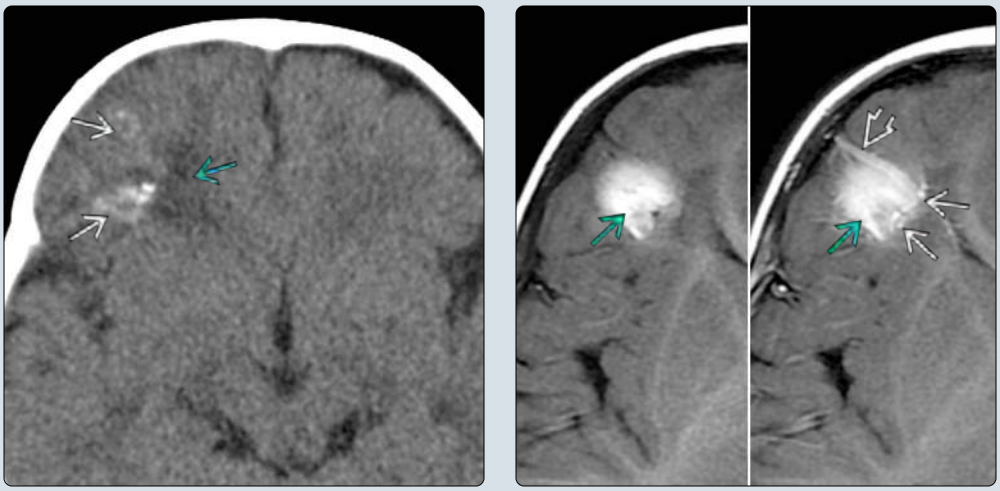
PATHOLOGY

- Etiology: Initial parenchymal microhemorrhage followed by neoangiogenesis & recurrent hemorrhage
  - ↑ size from recurring intralesional hemorrhages
- Multiple (familial) CM syndrome: Autosomal dominant
  - *CCM1, CCM2, CCM3* genes; variable penetrance
- 25% of CMs associated with DVAs

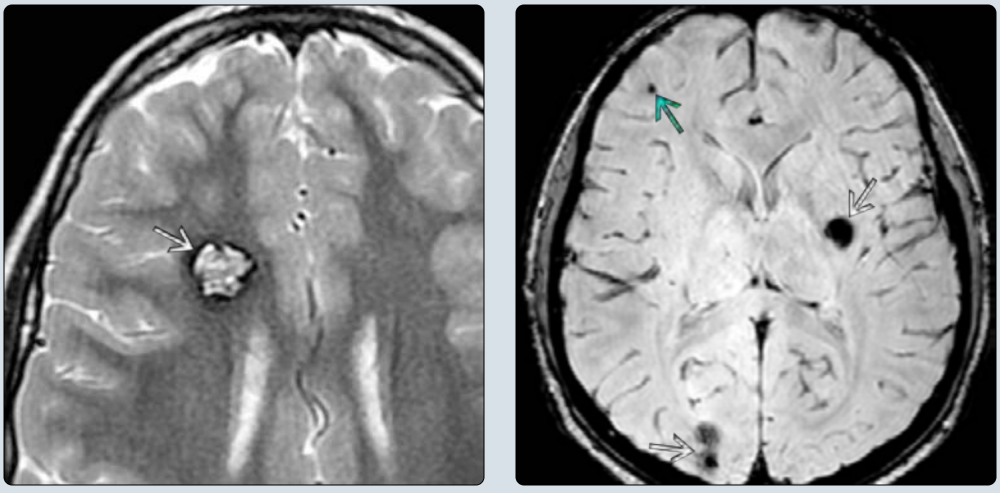
CLINICAL ISSUES

- 0.6% incidence in children undergoing brain MR
- Symptoms: 1/2 incidental; 1/4 seizures; 1/4 neurologic deficits
- 70% solitary; 30% multiple (in familial & prior XRT)
- Factors that ↑ annual risk of hemorrhage are
  - Zabramski MR classification (I & II > III, IV)
  - Prior hemorrhage
  - Brainstem location
  - Associated developmental venous anomaly (DVA)
- Treatment: Total surgical excision, sparing DVA

(Left) *Axial NECT in a 1 year old with focal facial seizures shows heterogeneous high attenuation in the right frontal lobe with low attenuation (edema) deep to the lesion.* **(Right)** *Axial T1 (left) & T1 C+ (right) MR images in same 1 year old demonstrate intrinsically high signal (Zabramski type I) in the lesion with multiple small vessels along the medial margin coalescing into a draining vein, consistent with a developmental venous anomaly (DVA). Twenty-five percent of CMs are associated with DVAs.*



(Left) *Axial T2 MR in a 15 year old with a headache shows the typical popcorn appearance of a Zabramski type II CM with a reticulated mixed signal core & hypointense rim.* **(Right)** *Axial SWI MR in a 15 year old with multiple familial CM syndrome shows 2 larger lesions in the left basal ganglia & right occipital lobe plus a smaller lesion (Zabramski IV) in the right frontal lobe. The 2 larger lesions were visible on T2, but the smaller lesion was only visible on the more sensitive SWI sequence.*



TERMINOLOGY

Definitions

- Cavernous malformation (CM): Benign vascular lesion arising in CNS
- Consists of dilated sinusoids containing blood products of various ages & lined by thin immature walls

IMAGING

General Features

- Location: Can be found throughout CNS
  - Supratentorial ~ 80%
  - Infratentorial ~ 20%
  - Rarely in spinal cord, except in multiple CMs
- Vary in size from microscopic to giant (> 6 cm)

CT Findings

- **NECT**: Round or irregular hyperdense lesion
  - ± hemorrhage & associated perilesional edema
  - Often shows small areas of Ca^{2+}
  - CT negative in up to 50%, especially for small lesions
- **CECT**: No enhancement of actual lesion
  - ± associated developmental venous anomaly (DVA)

MR Findings

- **T1WI**: Heterogeneous with blood of varied ages & Ca^{2+}
- **T2WI**: Mixed signal core with complete hypointense rim corresponding to hemosiderin
  - Numerous septations or locules
  - ± fluid-fluid levels of layering blood products
  - Small lesions may appear as focal hypointensities
- **FLAIR**: Surrounding edema suggests recent hemorrhage
- **SWI/T2* GRE**: Exaggerated signal loss (blooming)
  - Most sensitive for detection of small CMs
    - Numerous punctate hypointense foci (black dots)
- **DWI**: Usually normal; may show susceptibility effect
- **T1WI C+**: Minimal or no enhancement
  - Associated enhancing DVA in ~ 25%
- **MRA**: Normal
  - CMs "angiographically occult"

Imaging Recommendations

- Protocol advice
  - Use SWI or T2* GRE for detection of small lesions
  - Contrast administration will show associated DVA

DIFFERENTIAL DIAGNOSIS

Diffuse Axonal Injury

- Multiple small foci of hemosiderin deposition
- History of prior trauma with severe neurologic impairment

Venous Infarction

- More often hemorrhagic compared to arterial infarction
- Less complex-appearing; no hemosiderin ring if acute

Intracerebral Hematoma

- Usually more simple-appearing
- No hemosiderin at acute presentation

Neoplasm

- Hemorrhage or Ca^{2+} in primary brain tumor
- Often has enhancing soft tissue component

Neurocysticercosis

- May mimic familial CM with many small CNS Ca^{2+}

Capillary Telangiectasia

- Subtle enhancement following contrast administration

PATHOLOGY

General Features

- Etiology
  - Initial microhemorrhage from variety of conditions followed by neoangiogenesis & recurrent hemorrhage
    - ↑ vascular permeability (familial CMs); ↑ venous flow/pressure (CMs associated with DVAs); small vessel injury (CMs associated with XRT)
- Genetics
  - Multiple (familial) CM syndrome: Autosomal dominant
    - 3 separate loci implicated: *CCM1*, *CCM2*, *CCM3* genes
- Associated abnormalities
  - Developmental venous anomaly (DVA): 25% of CMs have associated DVA; < 5% of DVAs have associated CM

Staging, Grading, & Classification

- Zabramski classification of CMs
  - Type I: Subacute hemorrhage (↑ T1; ↑ or ↓ T2)
  - Type II: Reticulated mixed T1 & T2 with ↓ T2 rim
  - Type III: Chronic hemorrhage (iso/↓ T1; ↓ T2)
  - Type IV: Tiny microhemorrhages only seen on SWI/T2*

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Identified incidentally (40-50%)
  - Seizures (20-25%)
  - Neurologic deficit from hemorrhage (20-25%)

Demographics

- ~ 0.6% prevalence in all children undergoing brain MR
- ~ 70% solitary, 30% multiple

Natural History & Prognosis

- Factors that ↑ risk of symptomatic hemorrhage in children
  - Prior hemorrhage: 11.3% annually
  - Brainstem location: 16.7% annually
  - Associated DVA: 9.7% annually
- Annual risk of hemorrhage also predicted by MR imaging
  - Zabramski I & II: 23.4% (5-60% in literature)
  - Zabramski III: 3.4% (0-6% in literature)
  - Zabramski IV: 1.3%

Treatment

- Total removal via microsurgical resection
- If DVA present, venous drainage should be preserved
  - DVA removal → venous injury & venous infarct

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Childhood Aneurysms, Brain

KEY FACTS

IMAGING

- Majority of pediatric cerebral aneurysms present with SAH
- Location: Most located away from circle of Willis
  - ~ 80% anterior, ~ 20% posterior
- Morphology: Majority saccular, but fusiform aneurysms more common in children than adults
- Size: 15-20% "giant" (aneurysm > 2.5 cm)
  - May be partially thrombosed with onion-skin thrombus
- **NECT**: ± Ca^{2+}, especially giant aneurysms
- **CTA**: > 95% sensitivity for aneurysms > 2 mm
- **MR**: May be heterogeneous due to turbulent flow & layers of thrombus
  - Pulsation artifact (in phase-encoding direction) helps distinguish giant aneurysm from mass
- **3D TOF MRA**: > 90% sensitivity for aneurysms > 3 mm

TOP DIFFERENTIAL DIAGNOSES

- Arterial infundibulum
- Congenital cerebral aneurysmal arteriopathies

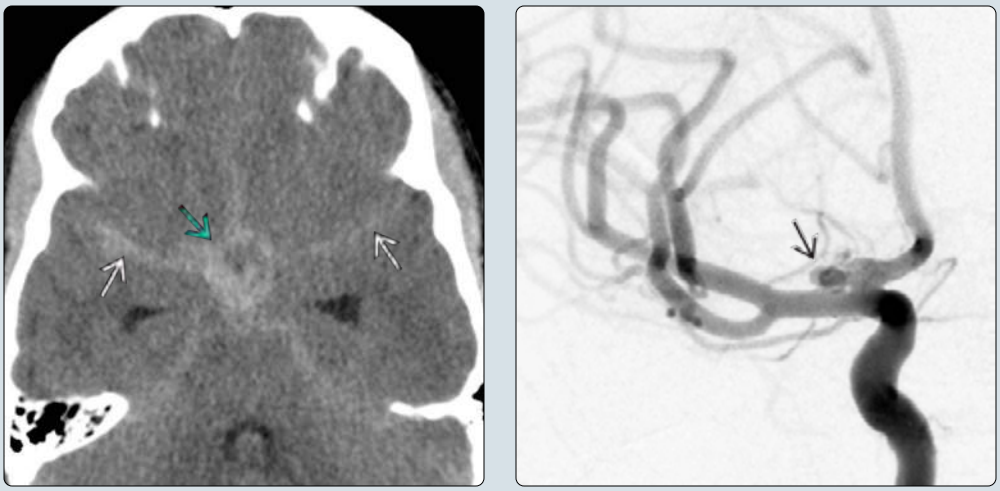
PATHOLOGY

- Idiopathic (~ 45%)
- Traumatic (~ 20%)
- Vasculopathic (~ 10%)
- Infectious (mycotic) (~ 5%)
- Familial, excessive hemodynamic stress, noninfectious inflammatory, & oncotic aneurysms less common

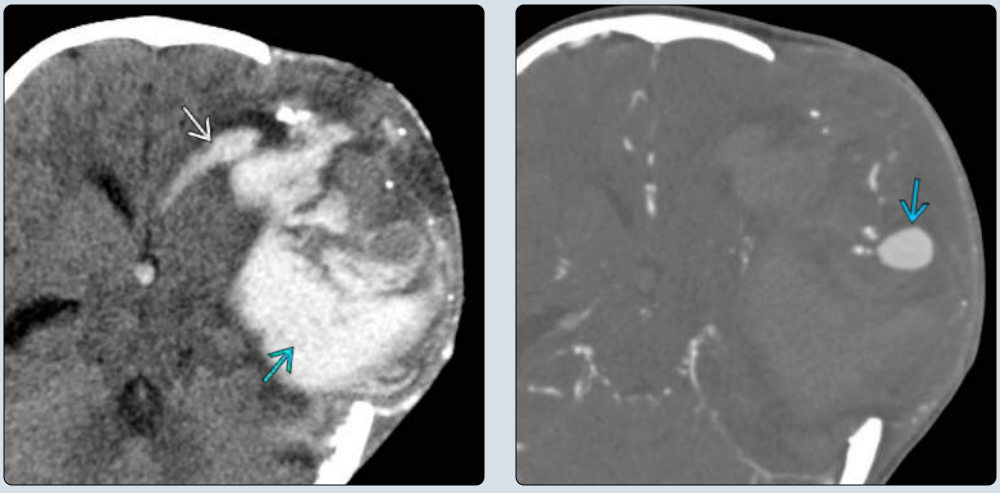
CLINICAL ISSUES

- < 5% of intracranial aneurysms occur < 18 years of age
- ~ 15% of hemorrhagic strokes occur in young patients
- Presentation: Sudden severe headache most common
  - Seizure, loss of consciousness, limb weakness, & cranial nerve deficits less common
- Prognosis: Severity of clinical presentation at time of rupture correlates with eventual outcome
- Treatment: Endovascular coiling or surgical clipping
  - Observation may be appropriate for certain types & locations of unruptured aneurysms

(Left) *Axial NECT image in a 13-year-old boy with aortic coarctation & an abrupt onset of severe headache shows hyperattenuation within the suprasellar cistern, sylvian fissures, & adjacent subarachnoid spaces, greater on the right. This is a typical location for subarachnoid hemorrhage (SAH) related to aneurysm rupture.* **(Right)** *Frontal oblique DSA image shows the focal vessel outpouching from the proximal A1 segment, consistent with a saccular aneurysm.*



(Left) *Axial NECT image in a 4-year-old girl status post gunshot wound & craniectomy shows a large intraparenchymal & intraventricular hemorrhage. Patients with such a history are at high risk for traumatic pseudoaneurysm formation, & a search for a traumatic aneurysm should be performed with a CT angiogram.* **(Right)** *Axial CT angiogram in the same patient shows a large ovoid contrast-opacified lesion within the parenchymal hematoma, consistent with a traumatic aneurysm.*



TERMINOLOGY

Definitions

- Focal arterial outpouching or dilation presenting < 18 years

IMAGING

General Features

- Best diagnostic clue
  - Enlargement or focal deformity of artery on vascular imaging (CTA, MRA, catheter angiography)
- Location
  - Anterior circulation (~ 80%)
  - Posterior circulation (~ 20%)
- Size
  - Giant aneurysms (> 2.5 cm) more common in children
    - ~ 15-20% in children vs. < 5% in adults
- Morphology
  - 27-78% saccular
  - 10-58% fusiform
- Multiple aneurysms less common in children (~ 15%)
  - Usually vasculopathic or infectious when multiple

CT Findings

- NECT
  - ~ 50-70% present with hemorrhage [most commonly subarachnoid hemorrhage (SAH)]
    - Intraventricular & parenchymal much less common
  - ± Ca^{2+} (especially giant aneurysms)
- CTA
  - Excellent means of detecting & delineating aneurysms
    - CTA sensitivity for aneurysms > 2 mm: > 95%
  - Luminal imaging only; thrombosed aneurysms may be missed or mischaracterized

MR Findings

- Appearance may be heterogeneous due to turbulent flow, flow-related enhancement, or layered thrombus
- Partially thrombosed aneurysms may have lamellated appearance on MR
  - Central T1/T2 flow void (on spin echo-based techniques) with peripheral layers of mixed signal
  - Central lumen & wall may enhance with contrast
- Pulsation artifact key feature to distinguish giant aneurysms from other masses
- 3D TOF MRA sensitivity for aneurysms > 3 mm: > 90%

Angiographic Findings

- Most diagnosed & characterized prior to angiography
- Catheter angiography usually performed in concert with endovascular therapy
- Both carotid & vertebral arteries must be injected to search for additional aneurysms

Imaging Recommendations

- Best imaging tool
  - NECT & CTA for emergent evaluation of SAH, intraventricular, or parenchymal hemorrhage
  - 3D TOF MRA may be used to follow known aneurysms
- Protocol advice
  - Thick MIP & 3D CTA reconstructions ↑ sensitivity for small peripheral aneurysms

DIFFERENTIAL DIAGNOSIS

Arterial Infundibulum

- Focal dilation at origin of arterial branch from circle of Willis
- Broad base, < 3-mm size, with vessel arising from apex

Congenital Cerebral Aneurysmal Arteriopathies

- Loeys-Dietz syndrome, Alagille syndrome, PHACES, autosomal dominant polycystic kidney disease, Ehlers-Danlos type IV, & others

PATHOLOGY

Staging, Grading, & Classification

- Classified by etiology
  - Idiopathic (~ 45%): No underlying predisposing factor
    - Usually distal to circle of Willis, saccular, & large
  - Traumatic (~ 20%): Blunt or penetrating head injury
    - Usually anterior circulation with saccular morphology
  - Vasculopathic (~ 10%): Disorders with vascular dysplasia
    - Often multiple, large, & fusiform
  - Infectious "mycotic" (~ 5%): Septicemia & endocarditis
    - Usually peripheral, saccular, & often multiple
  - Familial (~ 5%): Family history of nonsyndromic intracranial aneurysms
    - Small, anterior circulation, saccular aneurysms
  - Due to excessive hemodynamic stress (< 5%)
    - e.g., arteriovenous malformation, sickle cell disease
    - Usually small, solitary, & saccular in flow-loaded vessel
  - Noninfectious inflammatory (uncommon)
    - e.g., autoimmune vasculitis (lupus)
    - Usually multifocal, fusiform, & proximal

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - 10-15% of hemorrhagic strokes in children/young adults
  - Majority (32-89% in literature) present with SAH
    - Headache most common symptom
    - Seizure, loss of consciousness, limb weakness, & cranial nerve deficits

Demographics

- < 5% of intracranial aneurysms occur < 18 years of age
- More common with increasing patient age

Natural History & Prognosis

- Hunt & Hess grading system: Grades 1-5 based on clinical severity at time of SAH
  - ↑ grade associated with ↑ mortality & morbidity

Treatment

- Endovascular coiling
- Surgical clipping

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3. Aeron G et al: Clinical and imaging features of intracranial arterial aneurysms in the pediatric population. Radiographics. 32(3):667-81, 2012

KEY FACTS

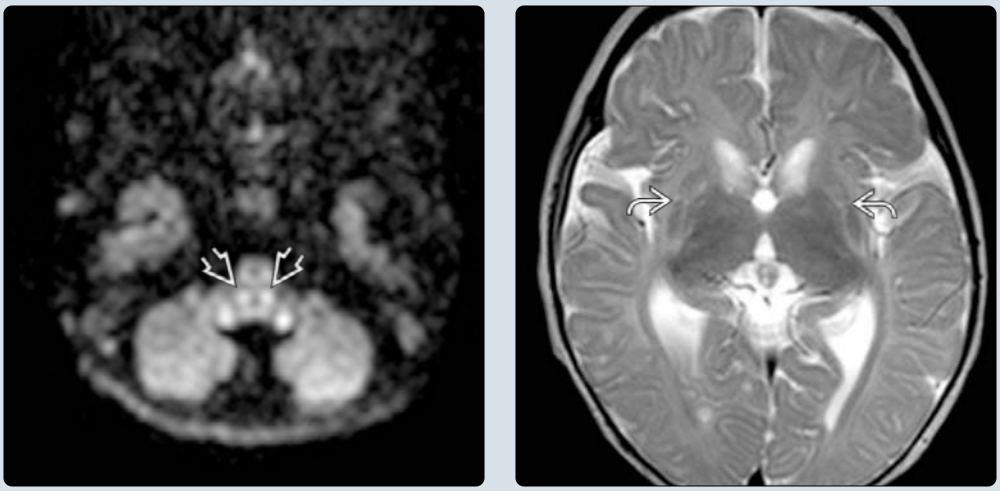
TERMINOLOGY

- Inborn errors of metabolism affecting brain
  - Localized &/or systemic accumulation of metabolites
  - Exclusive of mitochondrial encephalopathies & leukodystrophies
- Mucopolysaccharidoses (MPS)
  - Hunter, Hurler, Sanfilippo, Morquio, Maroteaux-Lamy, Sly, Natowicz syndromes
- Organic & aminoacidopathies
  - Maple syrup urine disease (MSUD)
  - Phenylketonuria
  - Nonketotic hyperglycinemia (NKH)
- Urea cycle disorders
  - Deficiency of enzymes in urea cycle
- Fatty acid oxidation disorders
  - Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
  - Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
  - Long-chain acyl-CoA dehydrogenase deficiency (LCAD, VLCAD)
  - Carnitine palmitoyl transferase deficiencies (CPT1, CPT2)

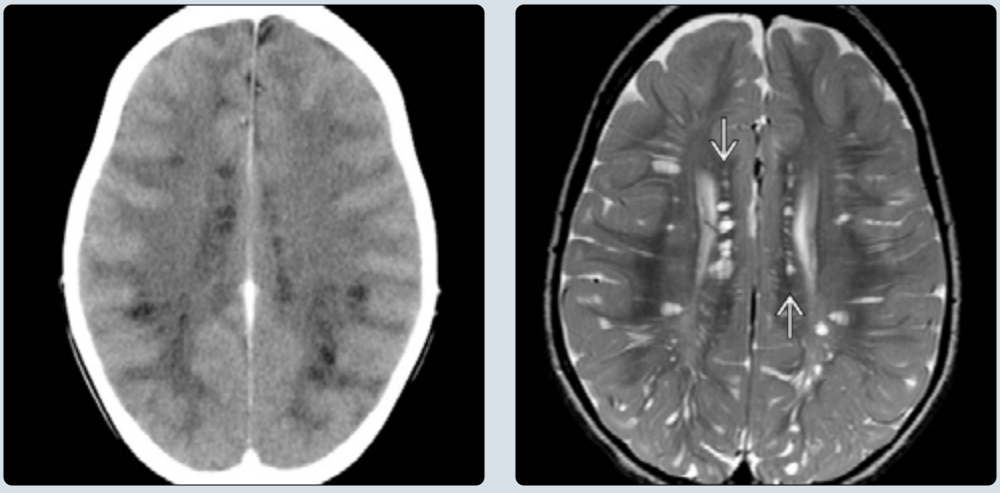
IMAGING

- MPS
  - Enlarged perivascular spaces
  - Often with patchy ↑ WM signal on T2WI
- MSUD
  - Edema prominently involving brainstem & cerebellar white matter
- NKH
  - Restricted diffusion & abnormal signal in posterior limb of internal capsule, dorsal midbrain, & cerebellar white matter in neonate
- Urea cycle disorders
  - Diffuse brain edema, not sparing basal ganglia or thalami
- MCAD deficiency
  - Cerebral cortical edema

(Left) *DWI MR through the pontomedullary junction in a neonate with nonketotic hyperglycinemia shows characteristic restricted diffusion in the dorsal tegmental tracts* ➪. **(Right)** *Axial T2 MR in a 4 month old with ornithine transcarbamylase deficiency shows volume loss & abnormal signal in the basal ganglia* ➪ *as a result of a prior episode of hyperammonemia.*



(Left) *CECT in a 14 year old with Hurler syndrome (mucopolysaccharidosis MPS-I) shows multiple foci of decreased attenuation in the cerebral white matter reflecting enlarged perivascular spaces.* **(Right)** *Axial T2 MR in a 2 year old with Hunter syndrome (MPS-II) demonstrates markedly enlarged perivascular spaces throughout the cerebrum, to include the body of the corpus callosum* ➪.



TERMINOLOGY

Definitions

- Inborn errors of metabolism affecting brain
  - Exclusive of mitochondrial encephalopathies & leukodystrophies
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- Organic & aminoacidopathies
  - Maple syrup urine disease (MSUD)
  - Phenylketonuria (PKU)
  - Nonketotic hyperglycinemia (NKH)
- Urea cycle disorders
  - Deficiency of enzymes in urea cycle
- Fatty acid oxidation disorders
  - Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
  - Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
  - Long-chain acyl-CoA dehydrogenase deficiency (LCAD, VLCAD)
  - Carnitine palmitoyl transferase deficiencies (CPT1, CPT2)

IMAGING

Key Imaging Findings

- MPS
  - CNS imaging abnormalities due to accumulated glycosaminoglycan (GAG)
  - Hunter syndrome prototypical
    - Enlarged perivascular spaces (supratentorial & infratentorial)
    - Often with patchy ↑ white matter (WM) signal on T2WI
  - Not all MPS will show abnormalities
- MSUD
  - Edema prominently involving brainstem & cerebellar WM
  - Hypointense on T1WI, hyperintense on T2WI, hypodense on CT
- PKU
  - ↑ signal on T2WI in periventricular WM
- NKH
  - Restricted diffusion & abnormal signal in posterior limb of internal capsule, dorsal midbrain, cerebellar WM in neonate
- Urea cycle disorders
  - Diffuse brain edema, not sparing basal ganglia or thalami
- MCAD deficiency
  - Cerebral cortical edema

PATHOLOGY

MPS

- Deficiencies of various enzymes required to break down GAG → accumulation of GAG in multiple organs, including brain & connective tissues
  - MPS I (Hurler): α-L-iduronidase
  - MPS II (Hunter): Iduronate 2-sulfatase
  - MPS III (Sanfilippo): Heparan sulfamidase
  - MPS IV (Morquio): Galactose 6-sulfatase
  - MPS VI (Maroteaux-Lamy): Arylsulfatase B
  - MPS VII (Sly): β-glucuronidase
  - MPS IX (Natowicz): Hyaluronidase

Organic & Aminoacidopathies

- MSUD: ↓ activity of branched chain α-keto acid dehydrogenase complex → accumulation of branched chain amino acids, particularly L-leucine
- PKU: ↓ phenylalanine dehydroxylase → accumulation of phenylalanine in brain (toxic to developing brain)
- NKH: Defect in 1 of 4 protein components (usually P-protein) of glycine cleavage system → accumulation of glycine (toxic to developing brain)

Urea Cycle Disorders

- Deficiencies of specific urea cycle enzymes
  - Ornithine transcarbamylase
  - Carbamoyl phosphate synthetase I
  - Argininosuccinate synthetase (citrullinemia)
  - Arginase (argininemia)
  - N-acetylglutamate synthetase
- Enzyme deficiencies prevent conversion of ammonia into urea → hyperammonemia, neurotoxicity

Fatty Acid Oxidation Disorders

- MCAD deficiency: Enzyme deficiency prevents oxidization of medium-chain fatty acids → hypoketotic hypoglycemia when fatty acid oxidation required to supplement cellular energy stores

CLINICAL ISSUES

MPS

- Progressive coarsening of features, developmental delay, skeletal dysplasia

Organic & Aminoacidopathies

- Catastrophic illness in neonatal period

Urea Cycle Disorders

- Hyperammonemia & coma in neonatal period

Fatty Acid Oxidation Disorders

- Severe hypoglycemia in response to illness or fasting

DIAGNOSTIC CHECKLIST

Consider

- Remember to consider metabolic diseases when encountering acutely ill neonate

Image Interpretation Pearls

- Characteristic laboratory abnormalities may provide clue
  - Hyperammonemia → urea cycle disorder
  - Severe hypoglycemia → MCAD deficiency

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Mitochondrial Encephalopathies

KEY FACTS

TERMINOLOGY

- Genetically based disorders of mitochondrial function resulting in progressive or intermittent brain injury
  - Subacute necrotizing encephalomyelopathy or Leigh syndrome (LS)
  - Pantothenate kinase-associated neurodegeneration (PKAN) (previously Hallervorden-Spatz syndrome)
  - Glutaric acidurias, type 1 (GA-1) & type 2 (GA-2)
  - MELAS (myopathy, encephalopathy, lactic acidosis, & stroke-like episodes)
  - Menkes disease (trichopoliodystrophy)

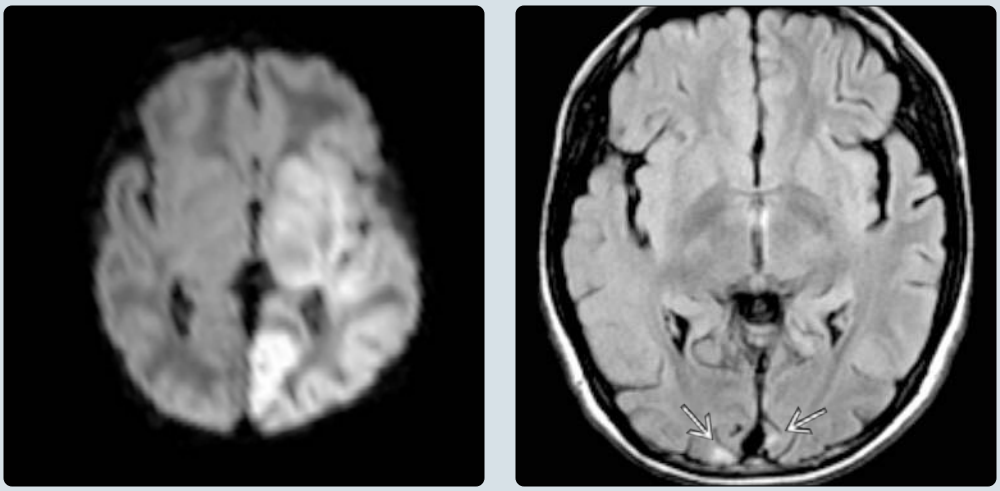
IMAGING

- Broad range of imaging appearances, characterized by regions of edema, brain destruction, volume loss, &/or mineralization
- Most disorders of mitochondrial function will cause lesions in basal ganglia (BG)
  - Typically bilateral & symmetric
- LS → linear striations in diffusely hyperintense BG
- PKAN → eye of tiger sign of gliosis & iron
- MELAS causes peripheral stroke-like lesions
- GA-1, Menkes → volume loss + subdural collections
- Menkes also notable for cerebral arterial tortuosity

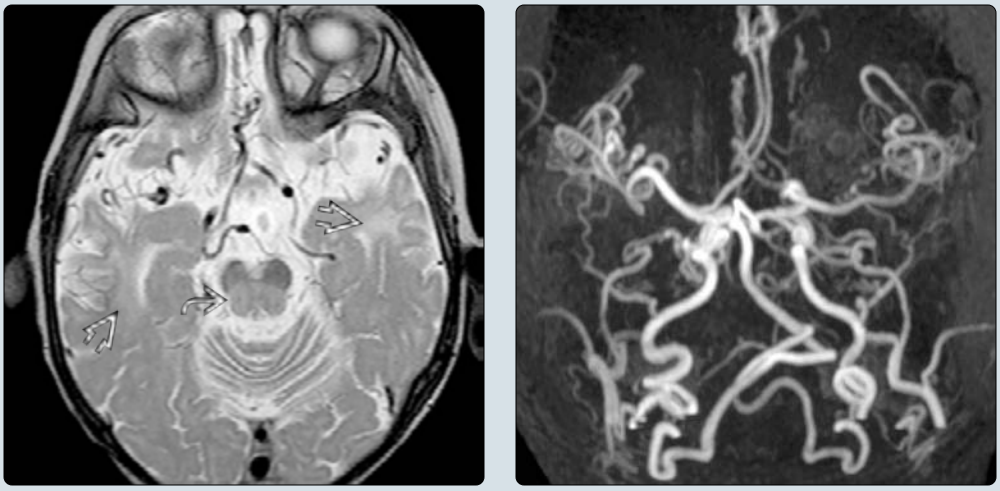
PATHOLOGY

- LS → group of disorders caused by defective terminal oxidative metabolism
- PKAN ("classic" form) → defects on chromosome 20p13 (*PANK2* gene)
- MELAS → mitochondrial DNA defects (*trnA* gene)
- GA-1 → deficiency of glutaryl-coenzyme A dehydrogenase; gene on chromosome 19p13.2
- Menkes → X-linked, Xq12-q13.3
- Friedreich ataxia → *FXN* gene on chromosome 9q13
- Alpers → mutations of mitochondrial DNA polymerase-γ subunits

(Left) *Axial DWI MR in a neonate with Leigh syndrome shows extensive ischemic injury throughout much of the left cerebrum to include the thalamus & basal ganglia. This is an atypical pattern for neonatal ischemic injury & should raise suspicion for metabolic brain disease.* **(Right)** *Axial FLAIR MR shows small foci of ischemic injury in each medial occipital lobe cortex in this 12 year old with MELAS (myopathy, encephalopathy, lactic acidosis, & stroke-like episodes).*



(Left) *Axial T2 MR in a 22 month old with Menkes syndrome shows prominence of the subarachnoid spaces & marked tortuosity of the cerebral arteries. The extent of bright signal in the dorsal brainstem & temporal lobes is greater than can be attributed to incomplete myelination.* **(Right)** *Axial oblique MIP from TOF MR angiography in the same patient more completely demonstrates the extent of abnormal arterial tortuosity, a typical finding in Menkes disease.*



TERMINOLOGY

Definitions

- Genetically based disorders of mitochondrial function resulting in progressive or intermittent brain injury
  - Subacute necrotizing encephalomyelopathy or Leigh syndrome (LS)
  - Pantothenate kinase-associated neurodegeneration (PKAN)
  - Glutaric acidurias, type 1 (GA-1) & type 2 (GA-2)
  - MELAS (myopathy, encephalopathy, lactic acidosis, & stroke-like episodes)
  - Kearns-Sayre syndrome (KSS)
  - Menkes disease (trichopoliodystrophy)
  - Alpers disease
  - Friedreich ataxia
- Characteristically due to deficiencies/defects of enzymes affecting respiratory (electron-transport) chain, Krebs cycle, &/or other components of energy production by mitochondria

IMAGING

General Features

- Best diagnostic clue
  - Broad range of imaging appearances, characterized by regions of edema, brain destruction, volume loss, &/or mineralization
  - Typically affect both gray & white matter (GM & WM)
  - Most disorders of mitochondrial function cause lesions in basal ganglia (BG)
    - Typically bilateral & symmetric
  - MELAS causes peripheral stroke-like lesions
- Edema/swelling characteristic of acute lesions; volume loss characteristic of late disease
- ↑ density can be seen on CT in BG in PKAN, KSS
- GA-1, Menkes → volume loss with subdural collections
  - Subdural collections in GA-1 mimic subdural hematomas from child abuse
- Mitochondrial encephalopathies (MEs) typically cause hyperintense lesions on T2WI & FLAIR
  - LS typically causes speckled pattern in deep nuclei
    - Sparing (islands of preserved signal) around vessels
- PKAN causes characteristic T2 hypointensity in globus pallidus (GP)
  - Due to excess & premature iron deposition
  - Central area of hyperintensity reflects gliosis
  - Combination of hypo- & hyperintense lesions → eye of tiger sign
- MEs may or may not cause foci of restricted diffusion
  - DWI not reliable for detection or exclusion of MEs
- Detection of lactate characteristic of MEs
  - Absence of lactate does not exclude diagnosis, however
- Menkes notable for cerebral arterial tortuosity
- Friedreich ataxia → cerebellar, spinal atrophy

Imaging Recommendations

- Best imaging tool
  - MR is modality of choice in investigation of suspected metabolic disease of any sort
- Protocol advice
  - MRS can be helpful although often nonspecific
  - ↑ lactate & restricted diffusion can be clue to true etiology
    - Especially if found in normal-appearing regions

DIFFERENTIAL DIAGNOSIS

Hypoxic Ischemic Encephalopathy

- Central pattern of injury affects ventrolateral thalamus & BG

Neurofibromatosis Type 1

- Signal abnormalities in GP most common brain manifestation in children

Encephalitis

- Acute disseminated encephalomyelitis can affect BG, mimic MELAS/myoclonic epilepsy with ragged red fibers (MERRF)

PATHOLOGY

General Features

- Genetics
  - LS → group of disorders caused by defective terminal oxidative metabolism
  - PKAN ("classic" form) → defects on chromosome 20p13 (*PANK2* gene)
  - MELAS → mitochondrial DNA defects (*trnA* gene)
  - GA-1 → deficiency of glutaryl-coenzyme A dehydrogenase; gene on chromosome 19p13.2
  - Friedreich ataxia → *FXN* gene on chromosome 9q13
    - ↓ levels of mitochondrial protein frataxin → causes deficiency of respiratory chain complexes I-III
  - Menkes → X-linked, Xq12-q13.3
  - Alpers → mutations of mitochondrial DNA polymerase-γ subunits
- Broad phenotypic presentations due to varied distribution of mitochondria throughout various cell types

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Think of MEs when encountering atypical presentation of stroke, severe encephalitis, or seizure
- Remember to consider MEs when infant presents with subdurals
  - Can also have retinal hemorrhages
- Tortuous arteries → think Menkes or Loeys-Dietz syndrome

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KEY FACTS

TERMINOLOGY

- Inherited disorders resulting in abnormal production or maintenance of myelin

IMAGING

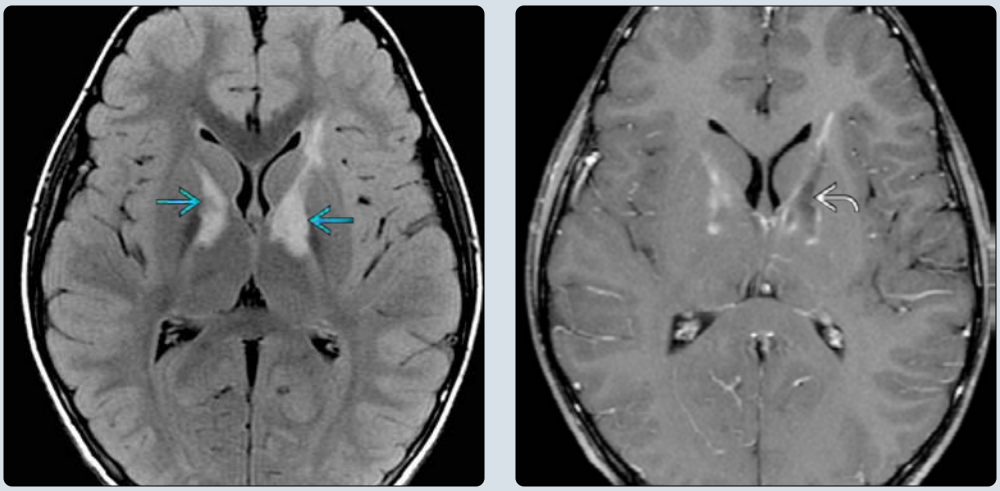
- Key tools in diagnosis: C+ MR, MRS
- Adrenoleukodystrophy (ALD): Characteristic early involvement of parietal periventricular white matter (WM)
- Metachromatic leukodystrophy (MLD): Early sparing of subcortical U-fibers & perivascular WM
- Globoid cell leukodystrophy (GLD): ↑ density on NECT in basal ganglia
- Alexander disease: Enhancement of ventricular lining, periventricular rim, frontal WM
- Canavan disease: ↑ NAA on MRS (1 of few conditions where this occurs)
- Megalencephalic leukoencephalopathy (MLC): Megalencephaly with swelling & ↑ signal in WM + anterior temporal subcortical cysts
- Vanishing WM (VWM): Diffuse ↑ WM on T2WI, ↓ on T1WI

PATHOLOGY

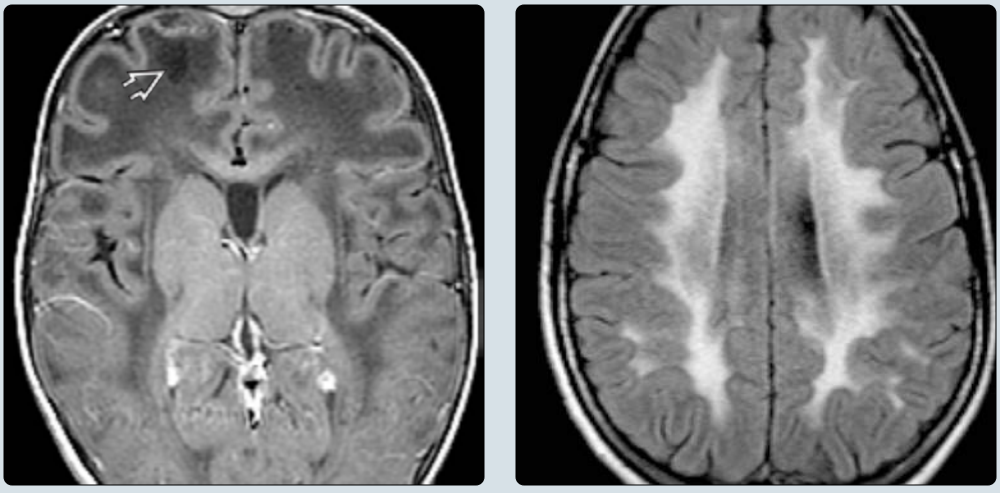
- ALD: Absent/deficient peroxisomal enzyme acyl-CoA synthetase → accumulation of long-chain fatty acids
- GLD: Absent/deficient lysosomal enzyme galactosylceramidase I → accumulation of psychosine & cerebroside
- MLD: Absent/deficient lysosomal enzyme arylsulfatase A → accumulation of sulfatide
- MLC: Mutations in *MLC1* or *HEPACAM* genes
- Alexander: Mutations in gene for glial fibrillary acidic protein → excess Rosenthal fibers in WM
- Canavan: Absence/deficiency of aspartoacyclase → accumulation of NAA
- VWM: Defects in *EIF2B* → episodic WM cavitation after febrile infections or minor head trauma

CLINICAL ISSUES

- Bone marrow/stem cell transplantation may stop progress



(Left) *Axial FLAIR MR in a 9-year-old boy with new onset of psychosis shows abnormal bright signal extending from the globus pallidus into the anterior limb of the internal capsule & periventricular white matter on each side.* **(Right)** *Axial T1 C+ FS MR in the same child shows enhancement along the periphery of the lesions with hypointensity in the central "burned out" zone on the left. Laboratory studies showed very long chain fatty acids, & genetic analysis confirmed the ABCD1 mutation of X-linked adrenoleukodystrophy.*



(Left) *Axial T1 C+ FS MR in a 5-year-old child with megalencephalic leukoencephalopathy with subcortical cysts shows swelling & hypointense signal (without enhancement) in the cerebral white matter, concentrated in the frontal lobes. Note the subcortical cyst on the right.* **(Right)** *Axial FLAIR MR in a 12-year-old child with metachromatic leukodystrophy shows the typical grainy pattern of dysmyelination seen in this condition.*

TERMINOLOGY

Definitions

- Inherited disorders resulting in abnormal production or maintenance of myelin
  - X-linked adrenoleukodystrophy (ALD)
  - Metachromatic leukodystrophy (MLD)
  - Krabbe disease/globoid cell leukodystrophy (GLD)
  - Alexander disease
  - Canavan disease
  - Megalencephalic leukoencephalopathy with subcortical cysts (MLC)
  - Vanishing white matter (VWM) disease
- Effects on white matter (WM) can be put into 3 categories
  - Dysmyelination: Formation of abnormal myelin
    - Typically results in demyelination
  - Demyelination: Destruction of myelin
  - Hypomyelination: Failure to form myelin

IMAGING

General Features

- Best diagnostic clue
  - Abnormal signal in WM, usually diffuse & symmetric
  - Failure to achieve myelination milestones
- Location
  - Cerebral WM > cerebellar WM
  - Some leukodystrophies have characteristic distributions, especially early in disease course
- ALD
  - Characteristic early involvement of parietal periventricular WM
  - Enhancement of zone of active inflammation
- MLD
  - ↑ signal in hemispheric WM on T2WI
  - Early sparing of subcortical U-fibers & perivascular WM
- GLD
  - Increased density on CT in basal ganglia
    - Globoid cell accumulation with Ca^{2+}
- Alexander disease
  - Megalencephaly with ↑ signal in frontal WM, thalami, & brainstem structures on T2WI
  - Enhancement of ventricular lining, periventricular rim, frontal WM, & more
- Canavan disease
  - Megalencephaly with ↑ signal in occipital WM early, progressing to entire brain
  - ↑ NAA on MRS (1 of few conditions where this occurs)
- VWM
  - Diffuse ↑ WM signal on T2WI, ↓ on T1WI
  - DTI may show progressive loss of anisotropy
- MLC
  - Megalencephaly with swelling & ↑ signal in cerebral > cerebellar WM
  - Subcortical cysts in anterior temporal &/or frontal WM

Imaging Recommendations

- Best imaging tool
  - Contrast-enhanced MR with MRS
- Protocol advice
  - MRS: Sample abnormal & normal-appearing WM

DIFFERENTIAL DIAGNOSIS

Pelizaeus-Merzbacher Disease

- Lack of myelination without myelin destruction

Radiation-Induced Leukomalacia

- WM injury associated with radiation therapy

PATHOLOGY

General Features

- Etiology
  - ALD: Absent/deficient peroxisomal enzyme acyl-CoA synthetase → accumulation of long-chain fatty acids
  - GLD: Absent/deficient lysosomal enzyme galactosylceramidase I → accumulation of psychosine & cerebroside
  - MLD: Absent/deficient lysosomal enzyme arylsulfatase A → accumulation of sulfatide
  - MLC: Mutations in *MLC1* or *HEPACAM* genes (function of encoded proteins presently unknown)
  - Alexander: Mutations in gene for glial fibrillary acidic protein → excess Rosenthal fibers in WM
  - Canavan: Absence/deficiency of aspartoacyclase → accumulation of NAA
  - VWM: Defects in eukaryotic initiation factor 2B → episodic WM cavitation after febrile infections or minor head trauma

CLINICAL ISSUES

Demographics

- Epidemiology
  - Rare diseases that can be ↑ in incidence in closed communities
  - Cumulative incidence as group > 1:10,000

Treatment

- Bone marrow & stem cell transplantation may arrest progress

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Key tools: Contrast-enhanced MR & MRS

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TORCH Infections

KEY FACTS

TERMINOLOGY

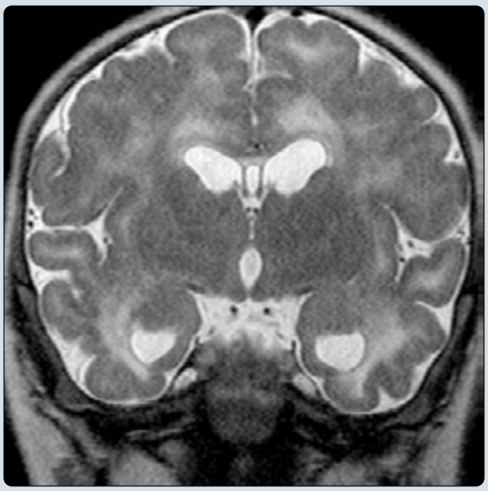
- TORCH/TORCHES: Acronym for congenital infections caused by transplacental transmission of pathogens
  - **T**oxoplasmosis (toxo) → *Toxoplasma gondii*
  - **O**ther (Zika → Zika virus)
  - **R**ubella → rubella virus
  - **C**ytomegalovirus (CMV) → most common TORCH infection
  - **He**rpes → herpes simplex virus 2 (HSV-2)
  - Human immunodeficiency virus (HIV)
  - **S**yphilis → *Treponema pallidum*

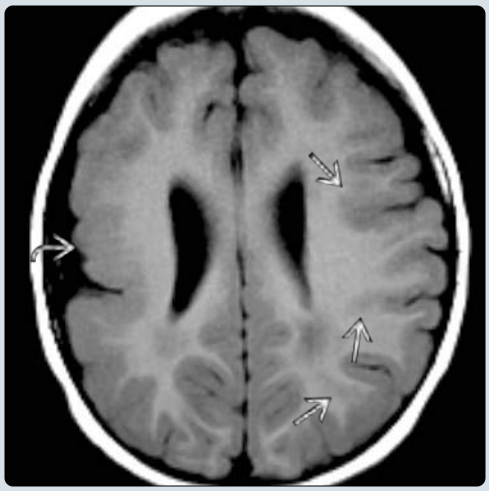
IMAGING

- Toxo, CMV, HIV, Zika, & rubella all cause parenchymal Ca^{2+}
- CMV causes migrational defects, white matter gliosis/hypomyelination, & cystic foci in temporal poles
- Rubella, Zika, & HSV cause lobar destruction/encephalomalacia
- ± microcephaly (not unique to Zika virus)

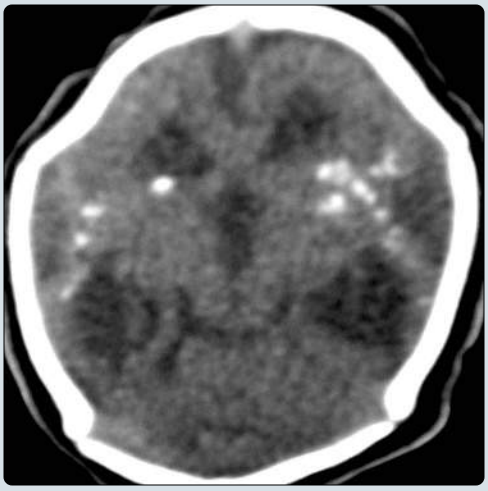
PATHOLOGY

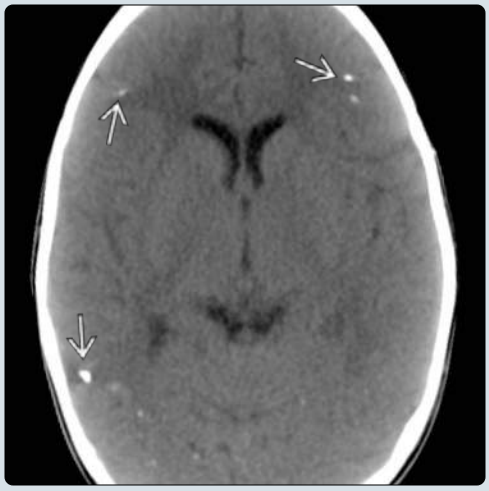
- CMV
  - Ubiquitous DNA virus of herpesvirus family
- Zika
  - Arbovirus of flavivirus family
  - Most commonly spread by *Aedes aegypti* mosquito
  - Primary infection typically mild but associated with postinfectious Guillain-Barré syndrome
- Herpes encephalitis
  - 75-90% of congenital herpes encephalitis
  - Most commonly from transmission during delivery
- Toxoplasmosis
  - Active infection in pregnancy → 20-50% congenital infection
- HIV
  - 30% of pregnancies in HIV-positive women will result in transmission unless preventative measures taken





(Left) *Coronal T2 MR in a 1 year old with congenital CMV infection & sensorineural hearing loss shows diffusely abnormal white matter (WM) signal & irregularity of the gray-W junction, reflecting migrational abnormalities.* **(Right)** *Axial T1 MR in a 4 year old with congenital CMV infection shows a large region of abnormal cortical thickening & diminished sulcation in the right hemisphere reflecting abnormal neuronal migration. More subtle migration abnormalities can be seen in the left hemisphere.*





(Left) *Axial NECT from an infant with in utero Zika virus infection shows periventricular & subcortical Ca^{2+} with marked volume loss resulting in microcephaly (despite the presence of ventriculomegaly). (Courtesy T. Fazecas, MD.)* **(Right)** *Axial CT in a 13 month old with congenital toxoplasmosis infection shows multiple peripheral punctate Ca^{2+} without superimposed WM destruction or migration abnormalities.*

TERMINOLOGY

Definitions

- Acronym for congenital infections caused by transplacental transmission of pathogens
  - **T**oxoplasmosis (toxo) → *Toxoplasma gondii*
  - **O**ther (Zika → Zika virus)
  - **R**ubella → rubella virus
  - **C**ytomegalovirus (CMV) → most common TORCH infection
  - **He**rpes → herpes simplex virus 2 (HSV-2)
  - Human immunodeficiency virus (HIV)
  - **S**yphilis → *Treponema pallidum*

IMAGING

General Features

- Toxo, CMV, HIV, Zika, & rubella all cause parenchymal Ca^{2+}
  - Toxo Ca^{2+} typically less extensive than CMV
- CMV causes migrational defects, white matter (WM) gliosis/hypomyelination, & cystic foci in temporal poles
- Rubella, Zika, & HSV cause lobar destruction/encephalomalacia
- Unless complicated by hydrocephalus, volume loss caused by TORCH infections typically results in microcephaly
  - Zika not unique in this regard
- Zika also associated with macular atrophy

Imaging Recommendations

- Best imaging tool
  - MR brain to completely characterize abnormalities
  - CT may be of benefit in confirming Ca^{2+}

DIFFERENTIAL DIAGNOSIS

Tuberous Sclerosis

- Subependymal Ca^{2+} characteristic

Neurocysticercosis (Cysticercosis)

- Most common cause of cerebral Ca^{2+} under 30

Pseudo-TORCH Syndromes

- Baraister-Reardon, Aicardi-Goutieres (CSF pleocytosis, ↑ CSF α-interferon)
  - Basal ganglia Ca^{2+}, ± periventricular Ca^{2+}

PATHOLOGY

General Features

- Etiology
  - CMV
    - Ubiquitous DNA virus of herpesvirus family
  - Zika
    - Arbovirus of flavivirus family
    - Endemic in Africa & Asia, first reached western hemisphere in 2014
    - Most commonly spread by *Aedes aegypti* mosquito
    - Primary infection typically mild but associated with postinfectious Guillain-Barré syndrome
  - Herpes encephalitis
    - Active HSV-2 genital infection during delivery can pass directly to child
      - 75-90% of congenital herpes encephalitis
    - Transplacental infection can be from HSV-1 or HSV-2
  - Rubella
    - Togaviridae family of viruses
  - Toxoplasmosis
    - Active infection in pregnancy → 20-50% congenital infection
  - HIV
    - 30% of pregnancies in HIV-positive women will result in transmission unless preventative measures taken
  - Syphilis
    - Transplacental transmission from mother with primary or secondary syphilis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - CMV can present at birth (10%) with microcephaly, hepatosplenomegaly, petechial rash
    - 55% with systemic disease have central nervous system involvement
  - Congenital toxoplasmosis usually inapparent at birth, presenting at 2-3 months
  - HSV acquired during delivery typically presents at 3-15 days with seizures, lethargy
    - HSV acquired in utero (5%) typically presents at birth
  - Zika typically apparent at birth due to microcephaly

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- CMV most frequently encountered TORCH infection in USA
- Congenital CMV encephalitis should be considered when MR shows
  - Microcephaly & cerebellar hypoplasia
  - Cortical gyral abnormalities or schizencephaly with Ca^{2+}
  - WM gliosis & subcortical temporal cysts
- Zika virus causes brain destruction & dysmorphism in addition to microcephaly with Ca^{2+}

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Brain Abscess

KEY FACTS

TERMINOLOGY

- Focal pyogenic infection of brain parenchyma

IMAGING

- Varies with stage of abscess development
- Early cerebritis: Ill-defined hypodense (CT)/hyperintense (T2 MR) subcortical lesion with mass effect & patchy enhancement
- Late cerebritis: T2 hypointense rim (MR) with irregular peripheral rim enhancement (CT/MR)
- Early capsule: T2 hypointense rim (MR) with well-defined, thin enhancing wall (CT/MR)
- Late capsule: Cavity shrinks, edema & mass effect diminish, capsule thickens (CT/MR)
- DWI MR: Restricted diffusion in cerebritis & abscess

TOP DIFFERENTIAL DIAGNOSES

- Resolving hematoma
- Pilocytic astrocytoma
- Demyelinating disease

PATHOLOGY

- Early cerebritis (3-5 days)
  - Unencapsulated mass of leukocytes & edema
- Late cerebritis (4-5 days up to 2 weeks)
  - Necrotic foci coalesce
- Early capsule (begins at around 2 weeks)
  - Well-delineated collagenous capsule
- Late capsule (weeks to months)
  - Central cavity shrinks

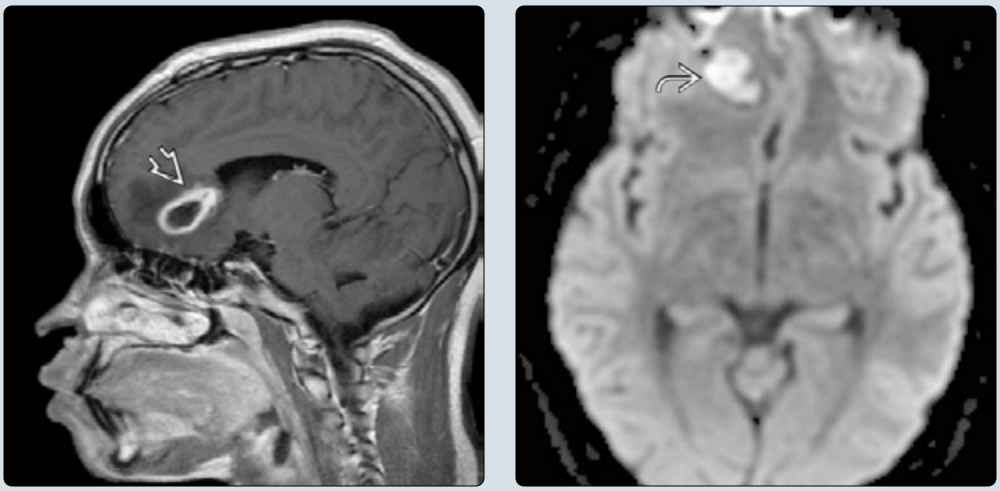
CLINICAL ISSUES

- 25% occur in patients < 15 years
- Headache most common symptom (up to 90%)
  - Fever in only 50%

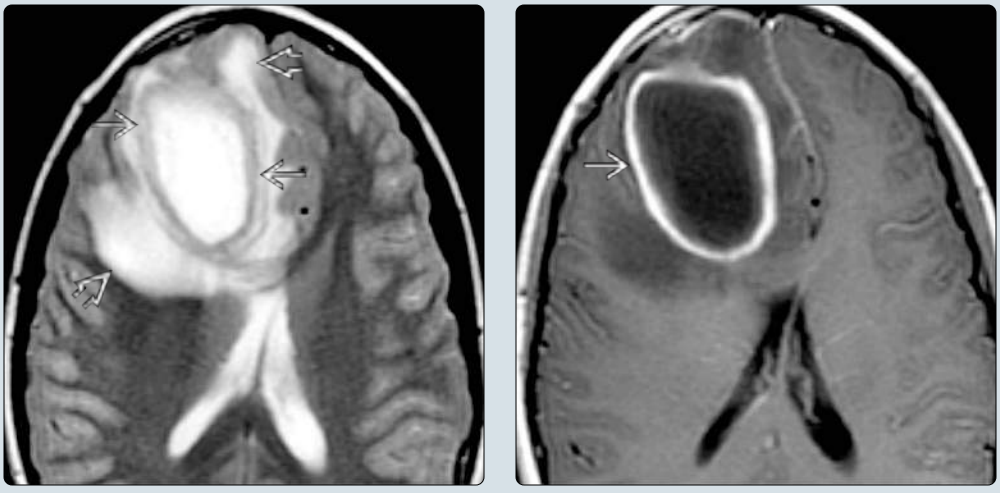
DIAGNOSTIC CHECKLIST

- Search for local cause such as sinusitis, mastoiditis

(Left) *Sagittal T1 C+ MR of a 12 year old who suffered penetrating trauma to the right frontal lobe from falling on a stick shows a rim-enhancing abscess with a thick wall that is characteristic of the late capsule stage.* **(Right)** *Axial DWI MR in the same patient shows markedly restricted diffusion within the central lesion, characteristic of abscess.*



(Left) *Axial T2 MR in a 13 year old with headaches & dizziness shows an ovoid hyperintense lesion with a hypointense wall & moderate surrounding vasogenic edema.* **(Right)** *Axial T1 C+ MR in the same patient shows a smooth, continuous outer rim of enhancement to the lesion. The constellation of findings is typical for a cerebral abscess. A necrotic tumor would typically show more heterogeneous & irregular enhancement without a T2 hypointense rim.*



TERMINOLOGY

Definitions

- Focal pyogenic infection of brain parenchyma
  - Typically bacterial; fungal or parasitic less common

IMAGING

General Features

- Best diagnostic clue
  - Ring-enhancing lesion
  - T2 hypointense rim with surrounding edema
  - High signal (restricted diffusion) on DWI with low ADC
- Location
  - Supratentorial > > infratentorial
    - Frontal & parietal lobes most common

CT Findings

- Early cerebritis: Ill-defined hypodense subcortical lesion with mass effect
- Late cerebritis: Irregular peripheral rim enhancement
- Early capsule: Thin, distinct enhancing capsule
  - Capsule thickest near cortex
- Late capsule: Cavity shrinks, capsule thickens

MR Findings

- Early cerebritis: Ill-defined T2 hyperintense mass with patchy enhancement
- Late cerebritis: Hypointense rim on T2; intense but irregular rim enhancement
- Early capsule: Hypointense rim on T2; well-defined, enhancing thin wall
- Late capsule: Edema & mass effect ↓; ↑ of thick wall
- Restricted diffusion in cerebritis & abscess
  - Markedly ↓ signal centrally on ADC map
- Central necrotic area may show presence of acetate, lactate, alanine, succinate, pyruvate on MRS

Imaging Recommendations

- Best imaging tool
  - Contrast-enhanced MR + DWI

DIFFERENTIAL DIAGNOSIS

Resolving Hematoma

- Rim enhancement starts ~ 7 days after event

Pilocytic Astrocytoma

- Cyst with enhancing mural nodule
- Low signal of cyst contents on DWI

Demyelinating Disease

- Multiple sclerosis, acute disseminated encephalomyelopathy
- May have incomplete rim enhancement
- Characteristic lesions elsewhere in brain

PATHOLOGY

General Features

- Etiology
  - Hematogenous from extracranial location
  - Extension from sinus infection via valveless emissary veins
  - Direct extension from calvarial or meningeal infection
  - Right-to-left shunts (congenital cardiac malformations, pulmonary arteriovenous fistulas)
  - 20-30% have no identifiable source → cryptogenic
  - Often polymicrobial → streptococci, staphylococci, anaerobes

Staging, Grading, & Classification

- Early cerebritis (3-5 days)
  - Unencapsulated mass of leukocytes & edema
- Late cerebritis (4-5 days up to 2 weeks)
  - Necrotic foci coalesce
- Early capsule (begins at ~ 2 weeks)
  - Well-delineated collagenous capsule
  - Liquefied necrotic core
- Late capsule (weeks to months)
  - Central cavity shrinks

CLINICAL ISSUES

Presentation

- Headache most common symptom (up to 90%)
  - Fever in only 50%

Demographics

- 25% occur in patients < 15 years

Natural History & Prognosis

- Complications of inadequately or untreated abscesses
  - Intraventricular rupture, ventriculitis (may be fatal)
  - Meningitis, daughter lesions
  - Mass effect, herniation
- Mortality variable, 0-30%

Treatment

- Antibiotics only, if small (< 2.5 cm) or early cerebritis
- Steroids to treat edema & mass effect
- Surgical drainage or excision may be required
- Drainage of adjacent source (e.g., infected paranasal sinuses, mastoids)

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Search for local cause such as sinusitis, mastoiditis
- T2 hypointense abscess rim resolves before enhancement in successfully treated patients

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Acute Encephalitis

KEY FACTS

TERMINOLOGY

- Acute brain inflammation caused by infectious agents, most commonly viruses

IMAGING

- Most viral encephalitides → wide ranging, nonspecific imaging findings
  - Poorly defined white matter signal abnormalities
  - Variable gray matter involvement
  - Bilateral symmetric basal ganglia or thalamic signal abnormalities
  - Little or no parenchymal enhancement
  - Minimal leptomeningeal enhancement
- Some pathogens have characteristic imaging features
  - Herpetic encephalitis (HSV1) reactivation pattern
    - Temporal & subfrontal hemorrhage
    - Most children with HSV1 do not follow this pattern but have multilobar distribution
- Some features associated with variety of pathogens
  - Focal splenium signal abnormality
  - Acute necrotizing encephalitis of childhood

TOP DIFFERENTIAL DIAGNOSES

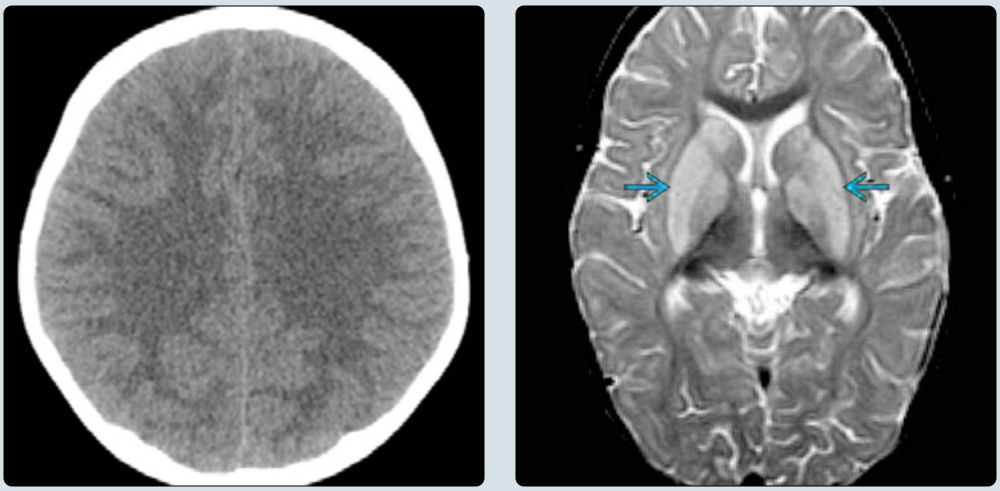
- Acute ischemia
- Gliomatosis cerebri
- Status epilepticus
- Mitochondrial encephalopathy

PATHOLOGY

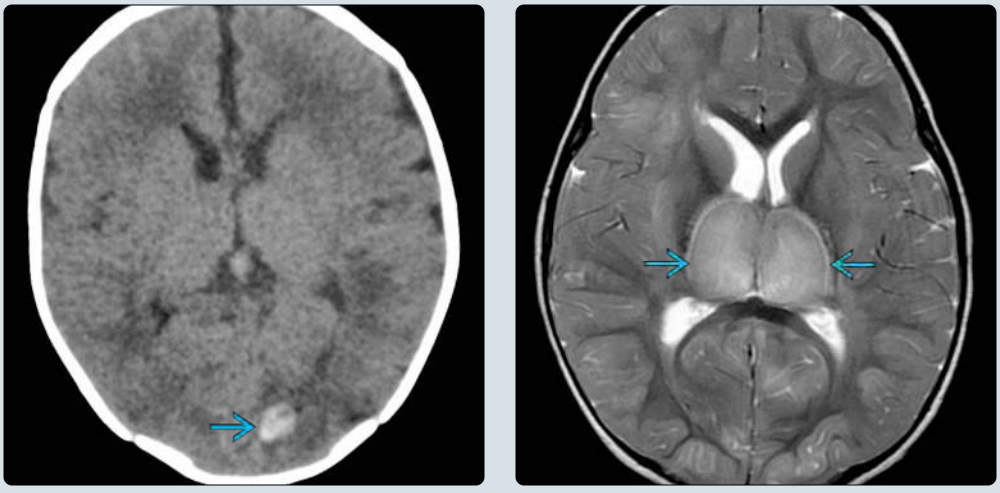
- Herpes: Most common sporadic viral encephalitis
- Japanese encephalitis: Most common endemic encephalitis in Asia

CLINICAL ISSUES

- Presentation varies widely: Slight meningeal to severe encephalitic symptoms, ± fever, prodrome
- Rapid diagnosis & early treatment with antiviral or antibacterial agents can ↓ mortality, may improve outcome



(Left) *Axial NECT in a 6 year old with West Nile virus encephalitis shows diffuse effacement of sulci with preservation of gray-white matter differentiation. Peripheral sulci should be visible over the convexities at any age; their absence indicates some degree of diffuse cerebral swelling.* **(Right)** *Axial T2 MR shows bilateral symmetric lesions in the basal ganglia of this 1 year old with EBV encephalitis. EBV is 1 of many viruses that can cause symmetric basal ganglia or thalamic signal abnormalities.*



(Left) *Axial NECT in a 13 day old with disseminated HSV shows a hemorrhagic lesion in the occipital pole. Perinatal HSV infection is typically from HSV-2 acquired during passage through an infected birth canal. Older infants & children are more likely to have HSV-1 infection.* **(Right)** *Axial T2 MR in a 15 month old with acute necrotizing encephalitis complicating influenza infection shows bilateral severe swelling of thalami, which is characteristic of this fulminant & frequently fatal complication of viral encephalitis.*

TERMINOLOGY

Definitions

- Acute brain inflammation caused by infectious agents, most commonly viruses

IMAGING

General Features

- Most viral encephalitides → wide ranging, nonspecific imaging findings
  - St. Louis, LaCrosse, California, eastern equine
    - Poorly defined signal abnormalities in white matter
    - Variable gray matter involvement
    - Bilateral symmetric basal ganglia or thalamic signal abnormalities
      - Diffusion restriction
    - Little or no parenchymal enhancement
    - Minimal leptomeningeal enhancement
- Some pathogens have characteristic imaging features
  - Herpetic encephalitis (HSV1) reactivation pattern
    - Temporal & subfrontal involvement, typically spares basal ganglia
    - Early hemorrhagic conversion
    - Characteristic in adults
      - Majority of children with HSV1 encephalitis do not follow this pattern but have multilobar distribution instead
  - Some features associated with variety of pathogens
    - Focal splenium signal abnormality
      - Ovoid focus of diffusion restriction in splenium of corpus callosum
      - Associated with influenza & other viral encephalitides
    - Acute necrotizing encephalitis of childhood
      - Fulminant encephalitis associated with influenza & Japanese encephalitis
      - Symmetric thalamic & basal ganglia necrosis

Imaging Recommendations

- Best imaging tool
  - MR with FLAIR, DWI, & contrast most sensitive
    - SWI or GRE to detect hemorrhage

DIFFERENTIAL DIAGNOSIS

Acute Ischemia

- Cytotoxic edema in typical vascular distribution; DWI positive

Gliomatosis Cerebri

- Lobar or hemispheric infiltration & expansion with subacute onset

Status Epilepticus

- Hyperintense on T2/FLAIR + diffusion restriction

Mitochondrial Encephalopathy

- Symmetric basal ganglia or thalamic involvement common

PATHOLOGY

General Features

- Etiology
  - Herpesviruses include HSV1, HSV2, CMV, EBV, varicella-zoster virus (VZV), B virus, HSV6, HSV7
  - Arboviruses transmitted by mosquitoes & ticks
    - Include eastern & western equine encephalitis, St. Louis encephalitis, Lacrosse encephalitis, California encephalitis
  - Enteroviruses include Coxsackie viruses A & B, poliovirus, echoviruses, enteroviruses 68 to 71
  - Rabies: Reaches CNS by retrograde axoplasmic flow

Microscopic Features

- Infiltration by polymorphonuclear cells, lymphocytes, plasma cells, & mononuclear cells
- May see inclusion bodies (i.e., Negri bodies in rabies)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Varies widely: Slight meningeal to severe encephalitic symptoms; ± fever, prodrome
  - Varicella & herpes zoster: Different clinical manifestations of infection by same virus (VZV)
    - Varicella encephalitis: Fever, headache, vomiting, seizures, altered mental status days to weeks after onset of (chicken pox) rash
    - Zoster: Immunocompetent patient with CN & peripheral nerve palsies in dermatomes involved by skin lesions
  - Enterovirus encephalitis (EV 71)
    - Hand, foot, & mouth disease: Fever, vesicles on hands, feet, elbows, knees, lips

Demographics

- Epidemiology
  - Herpes: Most common cause of sporadic (nonepidemic) viral encephalitis
  - Japanese encephalitis: Most common endemic encephalitis in Asia
  - CNS involvement in EBV uncommon (< 10% of cases)

Natural History & Prognosis

- Rapid diagnosis & early treatment with antiviral or antibacterial agents can ↓ mortality, may improve outcome

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KEY FACTS

TERMINOLOGY

- Acquired demyelinating processes characterized by inflammation
  - Multiple sclerosis (MS)
  - Acute disseminated encephalomyelitis (ADEM)
  - Lyme disease
  - Neuromyelitis optica (NMO) (a.k.a. Devic disease)

IMAGING

- MS
  - Multiple lesions, typically discrete
  - Can be mass-like (tumefactive MS)
- ADEM
  - Frequent brainstem & thalamic involvement
- Lyme disease
  - May be accompanied by optic neuritis or other cranial nerve inflammation
- NMO
  - Optic neuritis with spinal cord lesions

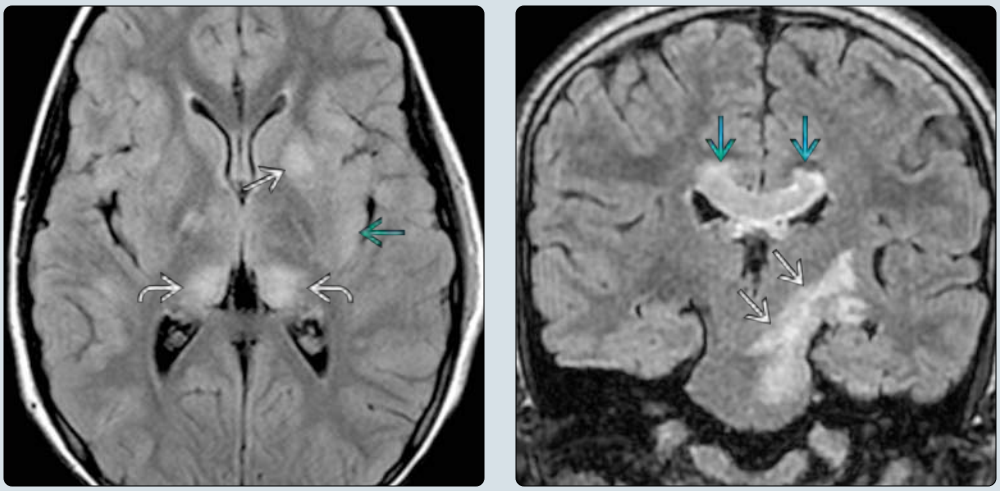
PATHOLOGY

- MS
  - Possibly viral-incited autoimmune reaction in genetically susceptible individuals
  - Activated T cells attack myelinated axons
- ADEM
  - Autoimmune mediated demyelination
  - Subsequent to infection (viral respiratory) or vaccination
- NMO
  - Associated with serum aquaporin-4 immunoglobulin G antibodies
  - 20-30% of cases are seronegative
- Lyme disease
  - Caused by spirochete *Borrelia burgdorferi*

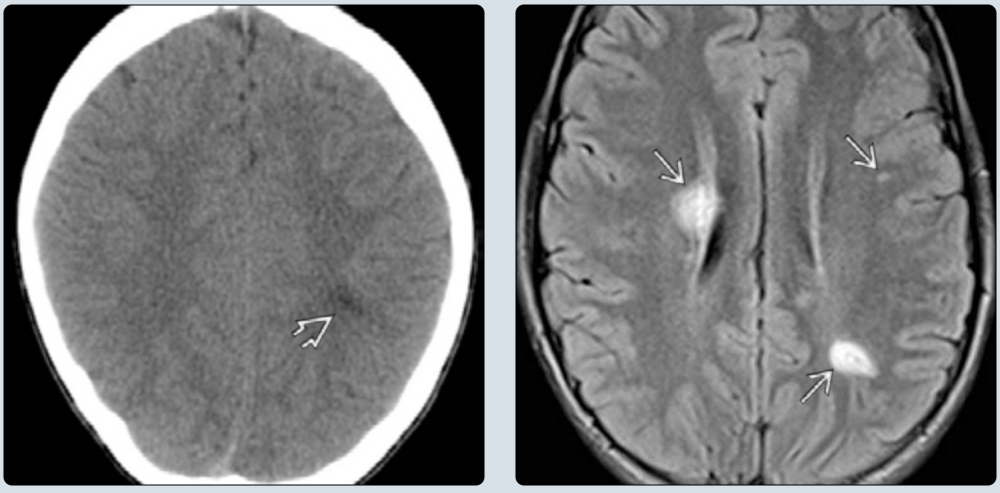
CLINICAL ISSUES

- Estimated 2,500,000 people in world have MS
- ADEM more frequently diagnosed in children
- 20% of childhood MS initially diagnosed as ADEM

(Left) *Axial FLAIR MR in a 9-year-old patient with altered mental status & hyperreflexia shows ill-defined hyperintense lesions in the thalami, basal ganglia, & insula. Involvement of the deep nuclei is a relatively common feature of acute disseminated encephalomyelitis.* **(Right)** *Coronal FLAIR MR in a 12-year-old patient with neuromyelitis optica & bladder dysfunction shows large lesions extending across the corpus callosum & left cerebral peduncle.*



(Left) *Axial NECT in a 14-year-old patient with vomiting shows a nonspecific low-attenuation lesion in the left posterior frontal subcortical white matter.* **(Right)** *Axial FLAIR MR of the same patient acquired the next day shows several ovoid multiple sclerosis plaques. Active lesions will also show contrast enhancement & restricted diffusion.*



TERMINOLOGY

Definitions

- Acquired demyelinating processes characterized by inflammation
  - Multiple sclerosis (MS)
  - Acute disseminated encephalomyelitis (ADEM)
  - Lyme disease
  - Neuromyelitis optica (NMO)
    - Neuromyelitis optica spectrum disorders (NMOSD)

IMAGING

General Features

- MS
  - Multiple lesions, typically discrete
    - Callosal involvement, hemispheric white matter
    - Perpendicular to ventricle margin; perivenular distribution
    - Can be mass-like: Tumefactive MS
  - Hyperintense on T2WI & FLAIR
    - "Black holes" on T1WI much more likely to be seen in MS than ADEM
  - Variable enhancement
    - Presumed to reflect active demyelination
    - Nodular, diffuse, or ring-like
  - Diffusion restriction in acute lesions
- ADEM
  - Hyperintense on T2WI & FLAIR, hypointense on T1WI
  - Frequent brainstem & thalamic involvement
- Lyme disease
  - Similar lesion appearance to MS & ADEM
  - May be accompanied by optic neuritis or other cranial nerve inflammation
    - Bell palsy characteristic
  - Associated meningitis
- NMOSD
  - Optic neuritis with longitudinally extensive transverse myelitis (> 3 segments); may have extensive brain lesions

DIFFERENTIAL DIAGNOSIS

Posterior Reversible Encephalopathy Syndrome

- Subcortical vasogenic edema in association with hypertension

Autoimmune-Mediated Vasculitis

- Enhancing lesions spare callososeptal interface

Susac Syndrome

- Classic triad: Encephalopathy, branch retinal artery occlusions, hearing loss

Leukodystrophies

- Alexander disease, Canavan, X-linked adrenoleukodystrophy

Tuberous Sclerosis Complex

- Gyral deformation by cortical/subcortical tubers

PATHOLOGY

General Features

- Etiology
  - MS
    - Idiopathic; possibly viral-incited autoimmune reaction in genetically susceptible individuals
  - ADEM
    - Autoimmune mediated demyelination; subsequent to infection (viral respiratory) or vaccination
  - NMOSD
    - Associated with serum aquaporin-4 immunoglobulin G antibodies; 20-30% of cases seronegative
      - May have antibodies to myelin oligodendrocyte glycoprotein
  - Lyme disease
    - Caused by spirochete *Borrelia burgdorferi*; tickborne infection

Staging, Grading, & Classification

- Major clinical subtypes of MS
  - Relapsing-remitting (85% initial presentation)
  - Primary-progressive, a.k.a. chronic progressive (5-10%)
  - Secondary-progressive, a.k.a. relapsing progressive
  - Progressive-relapsing
  - Clinically isolated syndrome: Single episode > 24 hours; vast majority progress to MS after number of years

CLINICAL ISSUES

Demographics

- Most common disabling CNS disease of young adults: 1 in 1,000 in Western world
- 3-5% of MS diagnosed before age 15
- ADEM more frequently diagnosed in children (20% of childhood MS initially diagnosed as ADEM)

Natural History & Prognosis

- MS: 45% of MS patients not severely affected, nearly normal
  - > 80% with "probable" MS & positive MR progress to clinically definite MS
- ADEM: Characteristically monophasic, although multiphasic subtypes now included
- Lyme disease: 11% develop neurologic manifestations; no evidence that antibiotic therapy alters natural history
- NMOSD: Typically relapsing-remitting

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- 95% with definite MS clinically have positive MR

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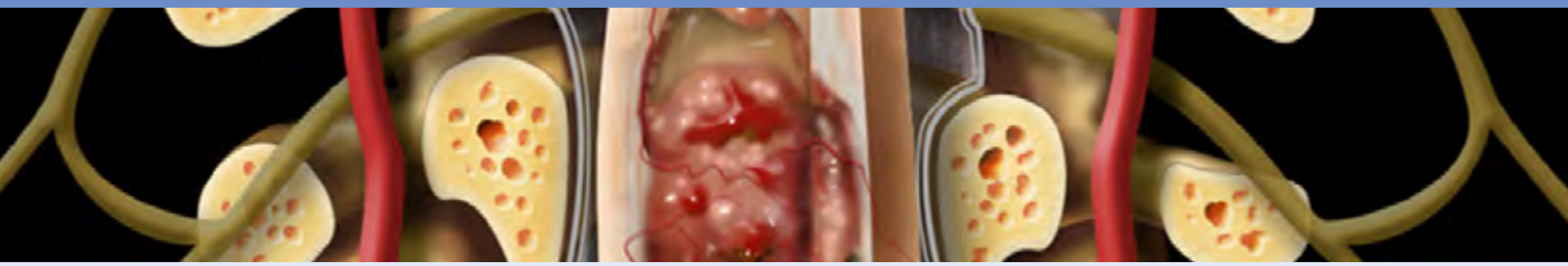
SECTION 8

Spine



Pathology-based Imaging Issues

The key to performing an optimal imaging evaluation in any circumstance is a clear understanding of the clinical questions to be addressed & the capabilities of the technology at your disposal. This is no less true in imaging of the pediatric spine, where the range & distribution of pathology are vastly different from what is encountered in the adult. Although degenerative disc & joint pathology certainly occurs in children, it is a small percentage of the overall pathologic spectrum, whereas it is the most commonly investigated category of disease in the adult. Accordingly, it is an error to attempt to adapt adult imaging protocols to the evaluation of the pediatric spine. The approach should rather be to devise imaging protocols that will maximize the ability to identify & assess the diseases that occur in children.

For young children with suspected congenital spine malformations, US can provide valuable information up to 6 months of age but will always need to be supplemented by MR when pathology is discovered. There is little benefit to attempting US on older infants, as the increasing bone formation limits acoustic windows. US can provide vital real-time information in the operating suite to guide resection of intramedullary tumors & the access of syrinx cavities.

Speed, accessibility, & high bony detail make CT ideal for the initial evaluation of spinal trauma, but MR is essential for the assessment of neural & ligamentous injury. For chronic back pain in the older child, spondylolysis is a commonly encountered pathology that is often more confidently evaluated by CT, especially in combination with nuclear medicine SPECT. Primary osseous lesions can sometimes be more accurately characterized by investigation of their appearance on CT.

However, MR is the mainstay of spinal imaging in children, as it provides the most complete & accurate assessment of congenital, inflammatory, & neoplastic disease. For the latter 2 categories, the administration of IV contrast is essential, but it rarely provides additional insight in congenital malformations. It is important to recognize that orthopedic stabilization devices may cause marked susceptibility artifact on MR, severely hampering the ability to evaluate the spine after surgery. This should not prevent the use of MR in evaluation of the postoperative instrumented spine; however, in many cases, the key features needed to make an accurate diagnosis can still be discerned despite the presence of marked image distortion by metal instrumentation.

Imaging Protocols

There is a temptation to always increase spatial resolution when performing medical imaging; after all, why wouldn't you want the greatest detail possible in all situations? This ignores the fact that increases in spatial resolution come at the cost of decreases in signal:noise ratios. Although some of the increasing image noise can be mitigated, it is often more rewarding to back away from higher spatial resolution in the interest of improved image quality. It is important to remember that the interpretation of a diagnostic image requires both identification of the abnormality & the ability to confidently characterize it, & the latter relies on optimal signal:noise ratios.

To that end, it can be counterproductive to image the pediatric spine with smaller fields of view (FOVs). In young infants, you may achieve the best imaging quality with a single sagittal FOV, whereas, in the majority of older children, you are best served by using 2 overlapping FOVs. T2WI should be the mainstay of most spine imaging protocols, providing the best assessment of both the spinal cord & of the normal pediatric intervertebral disc. Fluid-sensitive sequences (such as STIR or T2WI FS) diminish the signal of normal fat that may interfere with the assessment of pathologic fluid in fat-containing tissues; these sequences are essential in cases of trauma &/or pain in which marrow & soft tissue edema can point to associated fractures & ligamentous disruptions. Precontrast spin-echo-based T1WI provides an excellent assessment of normal bone marrow as well as any fat-containing intraspinal pathology. Postcontrast T1WI should be performed with fat-saturation techniques when possible, although this can be time intensive. CSF pulsation tends to be more vigorous in children than in adults, & the associated artifacts can be minimized by the use of gradient-echo sequences. High spatial resolution sequences, such as CISS, T2 DRIVE, or FIESTA, can be included in protocols to increase confidence in the detection of smaller lesions. Diffusion-weighted imaging of the spine is still in evolution, as technique adjustments like reduced FOV are necessary to improve image quality.

There are limited indications for myelography in pediatric spinal imaging (e.g., dural leaks, arachnoid cysts, absolute contraindications to MR). When it is performed, it should always include CT imaging with multiplanar reformatting.

Embryology

A unique challenge to performance of pediatric spine imaging is the prevalence of congenital spinal abnormalities. In order to confidently & accurately diagnose these malformations, it is necessary to have a working understanding of spinal embryology. Congenital spinal malformations can be classified into neural tube defects, split-cord or notochord malformations, & abnormalities of the caudal cell mass. Neural tube defects result from abnormalities in closure of the neural tube (neurulation), which occurs between 17 & 27 days of gestation. Failure of the neural ectoderm to separate from the cutaneous ectoderm will result in an open neural tube defect, which can be large in the case of a myelocele or myelomeningocele or small in the case of a dorsal dermal sinus. If disjunction occurs but is complicated by inclusion of mesoderm within the neural tube (perhaps a result of premature separation), a lipomatous malformation (lipomyelocele or lipomyelocystocele) will result. If the notochord splits during migration from Hensen node (perhaps due to an adhesion of the ectoderm & endoderm), a split cord malformation, such as a diastematomyelia or neurenteric cyst, is the consequence. Finally, if there is a disruption of the normal canalization & retrogressive differentiation of the caudal cell mass, distal lesions such as a terminal myelocystocele or syndrome of caudal regression will develop. The nomenclature for these congenital lesions is confusing & often based on observations & assumptions that are no longer valid. Recognizing this, the neuroradiologist should take great care to identify & describe all the salient characteristics of the malformation, rather than assuming a diagnosis & adjusting the interpretation to fit.

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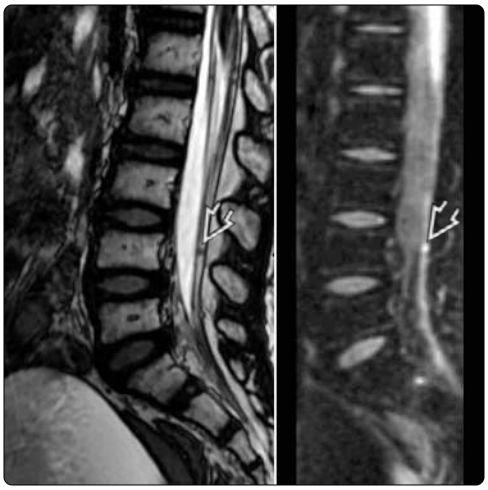
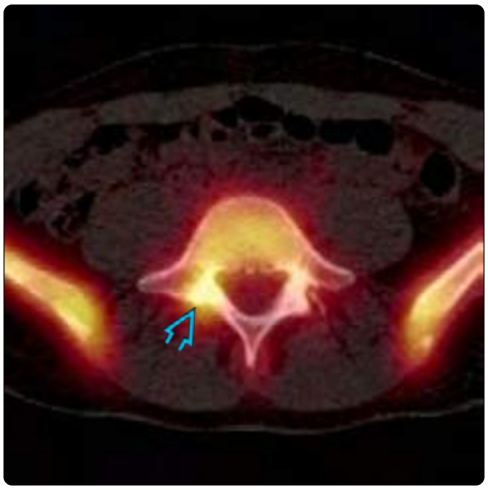
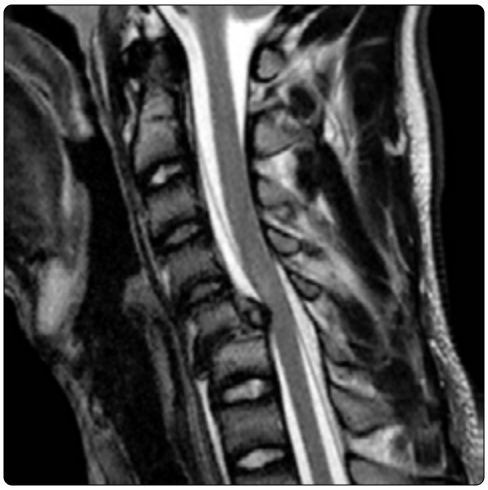
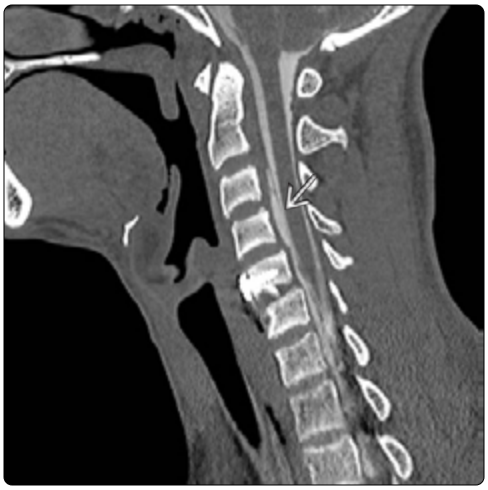


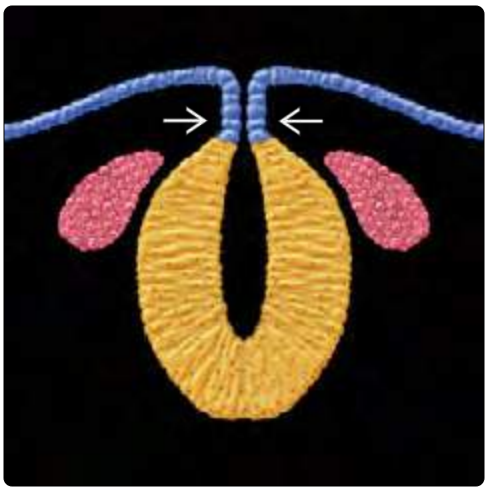


(Left) *Sagittal FIESTA (L) & DWI (R) MR through the lumbar canal show a 2-mm metastatic nodule adherent to the cauda equina in this child with medulloblastoma.* **(Right)** *Axial fused Tc-99m SPECT-CT through the lumbosacral junction shows increased radiotracer uptake on the right reflecting active bone response to instability caused by spondylolysis. Appropriate use of complimentary modalities can dramatically increase the clinical utility of spine imaging.*





(Left) *Sagittal T2 MR in a 10 year old with neck pain & paraparesis after a motor vehicle collision shows a fracture-dislocation with ligamentous disruption at C5-C6.* **(Right)** *Sagittal CT myelography in the same patient after stabilization & internal fixation shows extravasation of contrast from the thecal sac through a large ventral posttraumatic defect in the dura. The provocative nature of myelography allows it to demonstrate lesions that may be subtle or inapparent on MR.*



(Left) *The normal embryologic development of the spinal canal requires disjunction of the neural tube from the overlying surface ectoderm, as seen in the lower image.* **(Right)** *Failure of this disjunction allows continued communication of the central canal of the neural tube with the amniotic fluid resulting in an open neural tube defect. In nearly all instances, this prevents normal formation of the posterior fossa causing a Chiari II malformation.*

Myelomeningocele

KEY FACTS

IMAGING

- Open neural tube defect leading to Chiari II malformation
- Fetal US: Lumbosacral dysraphism with thin-walled, fluid-filled myelomeningocele sac + intracranial findings of Chiari II (lemon sign, banana sign, ± ventriculomegaly)
- Postnatal spine imaging typically only performed after repair: Expect soft tissue covering of defect
  - Posterior osseous dysraphic defects persist
  - Elongation of spinal cord (low conus) persists
  - Neural placode (distal nonneurulated segment of cord) inserts onto dorsal aspect of covered distal thecal sac

TOP DIFFERENTIAL DIAGNOSES

- Myelocele (a.k.a. myeloschisis)
- Closed (occult) spinal dysraphism
- Postoperative pseudomeningocele

PATHOLOGY

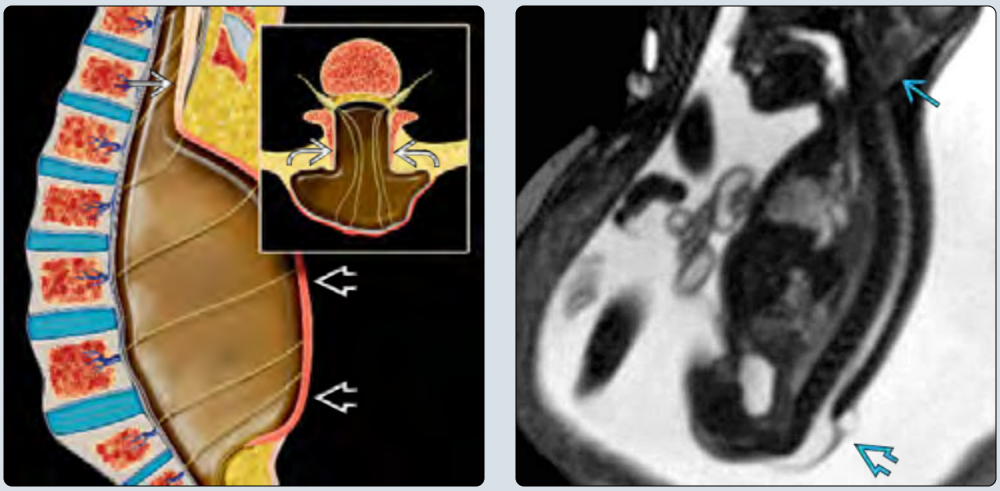
- Failure of neural tube closure during 3rd gestational week
- Association with maternal folate deficiency or abnormal folate metabolism

CLINICAL ISSUES

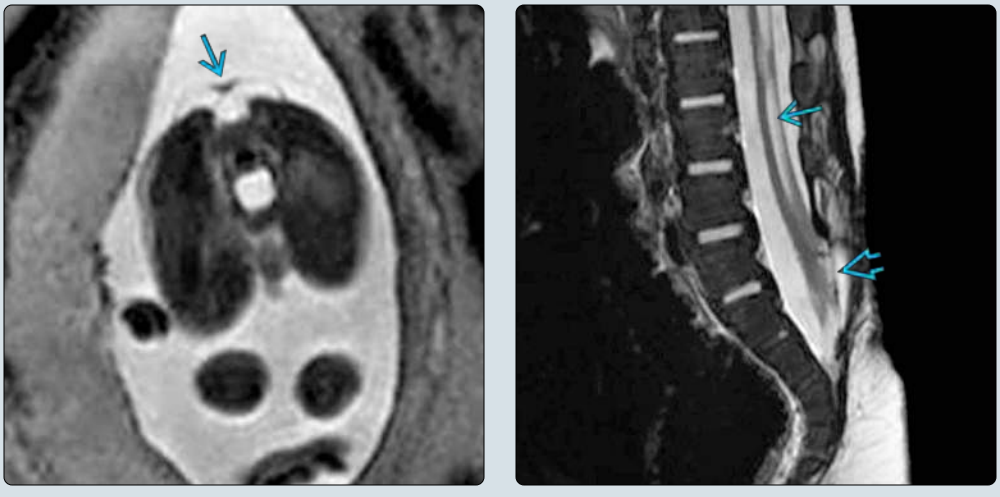
- Most common neurologic deficits: Lower extremities, bowel & bladder dysfunction
- Stable deficits expected following repair
- Subsequent neurological deterioration prompts imaging evaluation for other findings such as dural ring constriction, cord ischemia, syringohydromyelia, dermoid/epidermoid, arachnoid cyst, brainstem compression, shunt malfunction
- ~ 80-85% have hydrocephalus requiring CSF diversion, though lower incidence reported after fetal repair
- Mortality: 10-30% die before adulthood
  - Brainstem/cerebellar dysfunction or shunt malfunction

DIAGNOSTIC CHECKLIST

- Most common spinal cause of delayed neurologic deterioration: Cord retethering (clinical diagnosis)
  - Spine MR (± brain imaging) to exclude for other causes



(Left) *Sagittal graphic shows ballooning of the meninges & neural placode through a dysraphic spinal defect. The low-lying spinal cord terminates in the red neural placode. Axial insert shows the origin of spinal roots from the ventral placode as well as the protrusion of the meninges & placode through dysraphic posterior elements.* **(Right)** *Sagittal SSFP MR of a 22-week gestational age fetus with a lumbosacral myelomeningocele also shows posterior fossa findings of a Chiari II malformation.*



(Left) *Axial T2 SSFSE MR from the same 22-week gestational age fetus with a lumbosacral myelomeningocele shows the neural placode (or nonneurulated distal cord) exposed along the dorsal surface of the myelomeningocele sac.* **(Right)** *Sagittal T2 MR of the spine in the same patient at 6 months of age shows postoperative changes from the myelomeningocele repair with persistent elongation of the spinal cord (low conus). This patient developed a spinal cord syrinx, which is a common complication.*

TERMINOLOGY

Definitions

- Myelomeningocele (MMC): Open (lacking skin covering) spinal dysraphism in which neural placode (exposed flattened end of elongated spinal cord) protrudes beyond skin surface dorsal to spinal canal

IMAGING

General Features

- Best diagnostic clue
  - Splayed posterior osseous elements, distorted low-lying spinal cord/roots, postoperative skin closure changes
    - Rarely imaged before repair due to infection risk
  - > 90% have intracranial findings of Chiari II malformation

MR Findings

- Fetal MR
  - Abnormal elongation of spinal cord with exposed neural placode at dorsal surface of bulging MMC sac
  - Intracranial findings of Chiari II malformation
- Postnatal MR: T1WI, T2WI
  - Postoperative soft tissue changes of MMC closure
  - Elongation of spinal cord extending to flat neural placode adherent to posterior thecal sac
    - Loss of normal posterior epidural fat at level of defect
  - Postoperative complications
    - Constricting dural ring
    - Spinal cord compression by dermoid/epidermoid or arachnoid cyst
    - Cord ischemia from vascular compromise
    - Syringohydromyelia
  - Associated anomalies: Split cord malformation (31-46%) (may cause hemimyelomeningocele), dorsal dermal sinus, segmentation anomalies

Ultrasonographic Findings

- Obstetrical ultrasound → antenatal diagnosis
  - Open neural arch, flared laminae, protruding MMC sac
  - ± club foot, ± lower extremity movement

DIFFERENTIAL DIAGNOSIS

Myelocele (a.k.a. Myeloschisis)

- Open spinal dysraphism in which neural placode is flush with skin surface
- No measurable bulging MMC sac

Closed (Occult) Spinal Dysraphism

- Skin-covered osseous defect
- Many types: Lipomyelomeningocele, lipomyelocele, myelocystocele, meningocele

Postoperative Pseudomeningocele

- History & clinical exam allow for differentiation

PATHOLOGY

General Features

- Etiology
  - Failure of neural tube closure in 3rd gestational week
    - Likely multifactorial: Folate deficiency, toxins, genetics
  - Neurologic deficits worsened by chronic mechanical injury & amniotic fluid exposure (chemical trauma)
- Associated abnormalities
  - Kyphoscoliosis: Neuromuscular imbalance ± vertebral segmentation anomalies
  - Split cord malformation, dorsal dermal sinus
  - Syringohydromyelia (~ 50%)

CLINICAL ISSUES

Presentation

- Most common neurologic deficits: Lower extremities, bowel & bladder dysfunction
  - Higher level spine defect correlates with greater motor & somatosensory deficits
- ~ 80-85% have hydrocephalus requiring CSF diversion, though lower incidence reported with fetal repair
- Latex hypersensitivity common (~ 1/3)

Natural History & Prognosis

- Stable postoperative motor deficit seen in majority (~ 73%)
  - Neurological deterioration suggests complication
    - Tethering by scar (clinical diagnosis of exclusion) most common; symptoms of retethering in ~ 1/3
    - Imaging to look for other causes
      - Syringohydromyelia → disassociated sensory loss
      - Cervical myelopathy → spasticity & weakness of upper extremities
      - Brainstem compression → respiratory difficulties, cranial nerve dysfunction, aspiration/choking
      - Shunt malfunction
- ~ 2/3 have normal intelligence, ~ 23% have ≥ 1 seizure
- Bladder, bowel, & sexual dysfunction
  - ~ 80% bladder continent (most with catheterization)
  - ~ 38% require bowel regimen
- Lower extremity dysfunction
  - ~ 46% able to maintain ambulation into adult years
- Mortality: 10-30% die before adulthood

Treatment

- Folate supplementation to pregnant/conceiving women
- MMC closure < 48 hours after delivery to stabilize neurologic deficits & prevent infection
- In utero surgical repair: ↓ need for shunting, ↓ Chiari II findings; may improve neurologic function
- Subsequent management of postoperative complications
  - Cord untethering, CSF diversion

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Lipomyelomeningocele

KEY FACTS

TERMINOLOGY

- Subtype of closed (skin-covered) spinal dysraphism
  - Neural placode of elongated spinal cord attaches to lipomatous mass
  - Neural, fatty, & meningeal components extend through dysraphic posterior elements

IMAGING

- Thinned, elongated conus tethered dorsally to lipoma
  - Does not move dependently/ventrally in prone position
  - Subarachnoid space expanded ventral to neural placode
- Dorsal herniation of neural placode, lipoma, & meninges through splayed osteocartilaginous posterior elements of lumbosacral spine
- Lipoma blends with subcutaneous (SQ) fat
  - Variable size/extent of intradural/epidural components
- ± syrinx, split cord malformation, lumbosacral anomalies
- Anorectal & genitourinary tract malformations in 5-10%

TOP DIFFERENTIAL DIAGNOSES

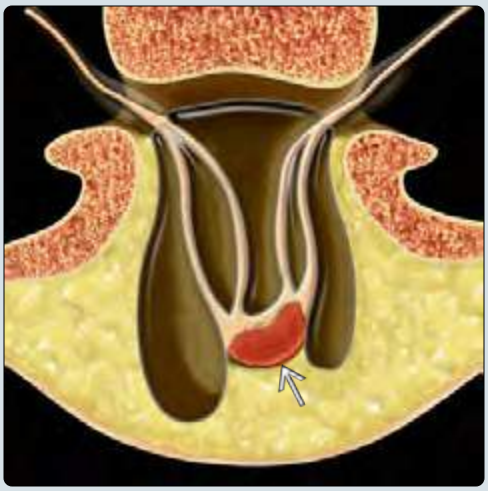
- Other closed spinal dysraphisms
  - With SQ mass: Lipomyelocele, meningocele, terminal myelocystocele
  - Without SQ mass: Intradural lipoma, dorsal dermal sinus
- Open spinal dysraphisms
  - Myelomeningocele, myeloschisis
- Sacrococcygeal teratoma

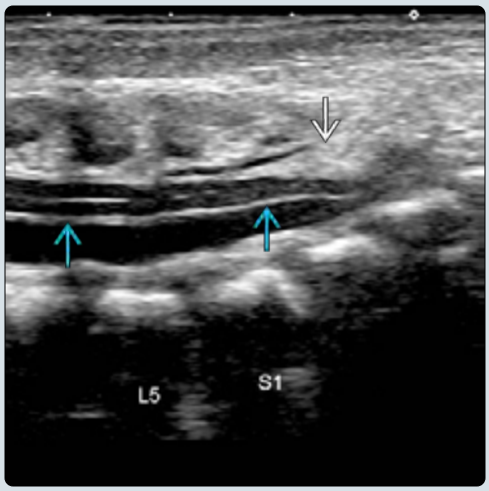
PATHOLOGY

- Premature disjunction of neural ectoderm from cutaneous ectoderm → retained mesenchyme induced to form fat (lipoma)

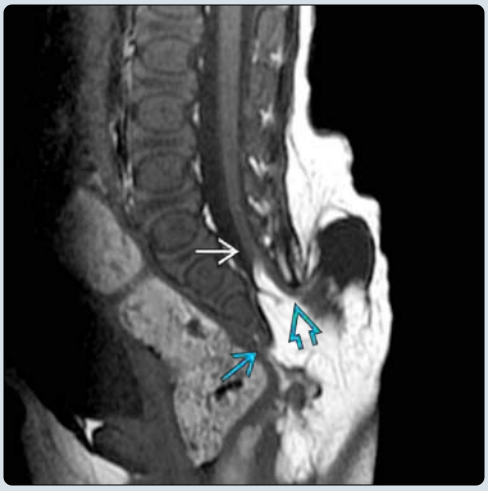
CLINICAL ISSUES

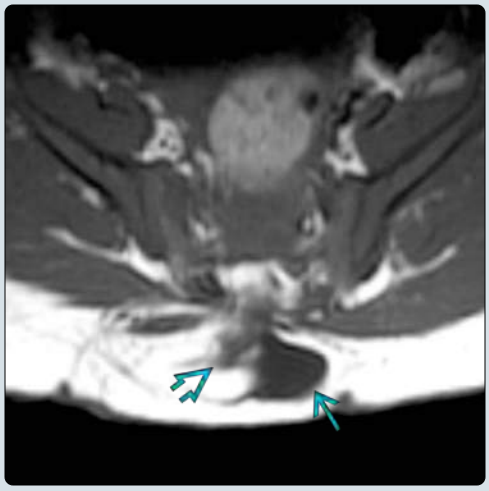
- Presents with soft skin-covered mass above buttocks
- 40-60% neurologically normal at birth
- Variable development of back/leg pain, lower extremity weakness, sensory loss, bladder/bowel dysfunction





(Left) *Axial graphic of a lipomyelomeningocele (LMMC) shows the neural placode ➡ tethered into a fatty mass that is continuous (through a dorsal dysraphic defect) with the subcutaneous (SQ) fat. The lesion is skin covered.* **(Right)** *Longitudinal ultrasound in a newborn with a lipomatous sacral appendage shows an elongated nondependent spinal cord ⇨ tethered to an echogenic fatty mass ➡. The mass extends dorsally through dysraphic posterior elements into the SQ tissues, consistent with an LMMC.*





(Left) *Sagittal T1 MR of an LMMC in a newborn demonstrates a low-lying conus medullaris ➡ with the placode-lipoma interface ⇨ extending dorsally through the abnormal posterior elements. Associated sacral hypogenesis & coccygeal aplasia are noted ⇨.* **(Right)** *Axial T1 MR in the same patient with an LMMC shows the placode-lipoma interface ⇨ outside of the spinal canal with expansion of the adjacent meninges ⇨.*

KEY FACTS

TERMINOLOGY

- Midline/paramidline subcutaneous sinus tract extending from skin surface toward spinal canal
- Tract lined by stratified squamous epithelium
- Located anywhere along neuraxis from cranium to intergluteal cleft; most common location (~ 70%) in lumbosacral region above intergluteal cleft

IMAGING

- Linear midline/paramidline sagittal T1 hypointense tract in subcutaneous tissues
- Terminates anywhere from subcutaneous tissues (blind-ending) to spine (epidural, intradural/intrathecal, or even intramedullary)
- ± epidermoid/dermoid cysts along tract (~ 50%)
  - DWI, FLAIR, or thin-section fluid-sensitive sequences helpful for intrathecal mass visualization
- ± tethered cord with low-lying conus medullaris, intradural lipoma, split cord malformation

TOP DIFFERENTIAL DIAGNOSES

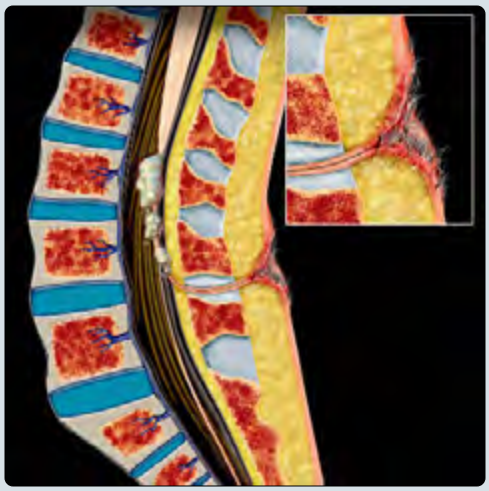
- Coccygeal pit/simple sacral dimple
- Pilonidal sinus/cyst
- Epidermoid/dermoid without sinus tract
- Lipomyelomeningocele (or other closed dysraphisms)

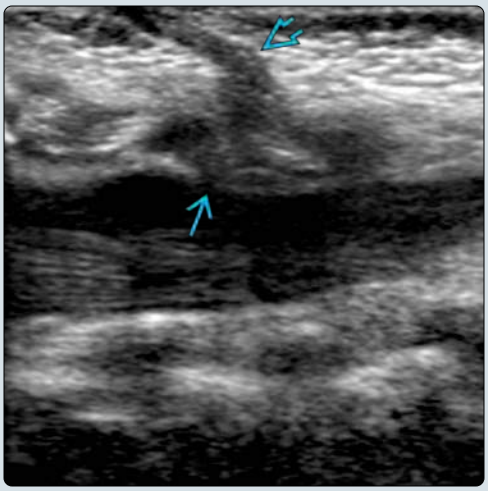
PATHOLOGY

- Closed spinal dysraphism due to incomplete dysjunction/separation of cutaneous ectoderm from neural ectoderm during neurulation at 3-8 weeks gestational age

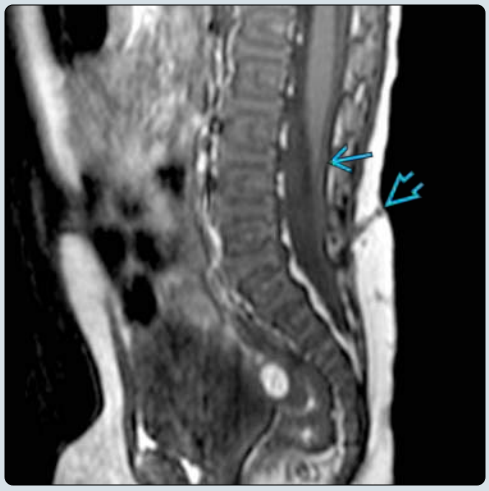
CLINICAL ISSUES

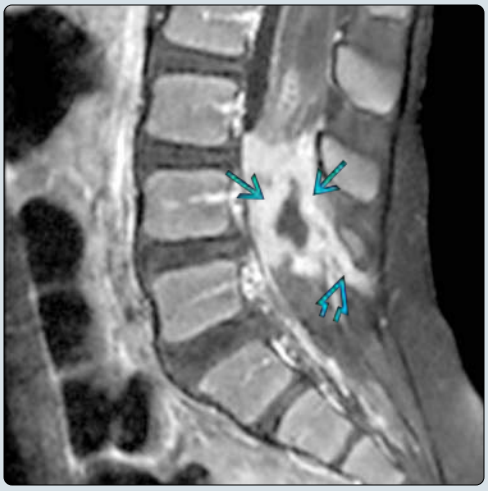
- Presentations include
  - Asymptomatic (incidentally noted skin dimple)
  - Infection (meningitis, abscess)
  - Neurologic deficits from cord tethering or compression
- Dorsal dermal sinus typically requires excision of entire tract
  - Intraspinal extension may be occult on imaging (necessitating operative exploration of dura)





(Left) *Sagittal graphic shows a dermal sinus extending from the skin surface into the spinal canal to terminate with epidermoid cysts at the conus medullaris. In this case, the sinus opening is marked by a skin dimple with a hairy tuft & capillary stain.* **(Right)** *Sagittal ultrasound image of the spine in an infant with a cutaneous lumbar dimple demonstrates a hypoechoic dorsal dermal sinus tract ⇨ in the subcutaneous tissues. A dural defect ⇨ confirms communication of the tract with the thecal sac & subarachnoid space.*





(Left) *Sagittal T1 MR in the same patient shows a T1 hypointense dorsal dermal sinus tract ⇨ in the subcutaneous tissues. The conus medullaris ⇨ is low lying at the L3-4 disc space level. No associated intraspinal mass or cyst was identified.* **(Right)** *Sagittal T1 C+ FS MR in the same patient at 3 years of age demonstrates the interval development of a rim-enhancing intraspinal abscess ⇨. Note that there is also enhancement along the dorsal dermal sinus tract ⇨ entering the spinal canal.*

Caudal Regression

KEY FACTS

TERMINOLOGY

- Synonyms: Caudal regression syndrome, caudal agenesis, lumbosacral hypogenesis

IMAGING

- Agenesis/dysgenesis of portion of caudal spine, frequently in conjunction with spinal cord ± GI/GU anomalies
- Spectrum ranges in severity from isolated coccygeal aplasia to complete lumbosacral (& even lower thoracic) spine agenesis
- 2 main types
  - Group 1: High (L1 or higher) blunted distal spinal cord
    - "Double-bundle" nerve roots
    - More severe distal osseous anomalies
  - Group 2: Tapered, low-lying distal cord
    - Other signs of cord tethering: ↓ nerve root movement, intradural lipoma, thick filum
    - Less severe osseous anomalies

TOP DIFFERENTIAL DIAGNOSES

- Tethered spinal cord
- Other closed spinal dysraphisms
- Occult intrasacral meningocele
- Currarino triad

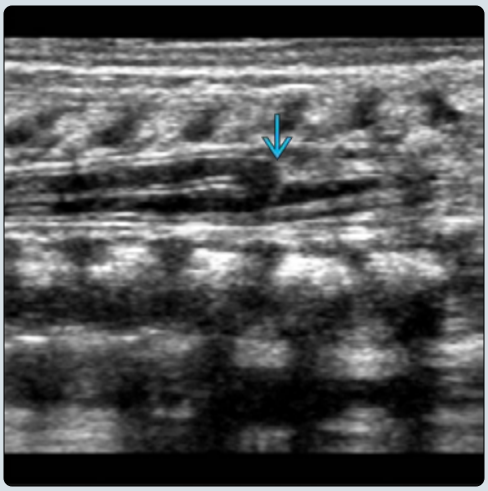
CLINICAL ISSUES

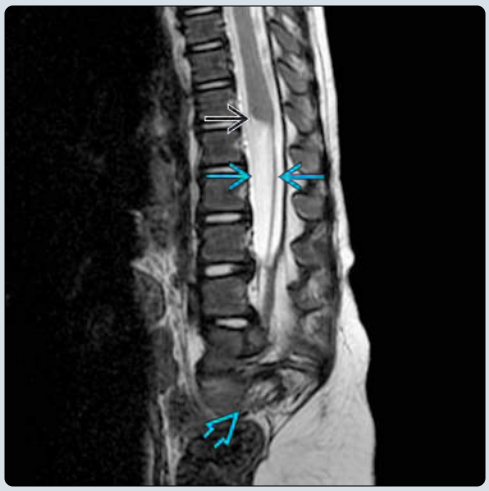
- 200-400x more common in infants of diabetic mothers
- Clinical spectrum ranges from neurologically normal → severely impaired
- Group 2 patients more likely to require neurosurgical intervention (cord detethering) than group 1
- Usually sporadic though can be seen in various syndromes
  - OEIS, VACTERL, Currarino triad

DIAGNOSTIC CHECKLIST

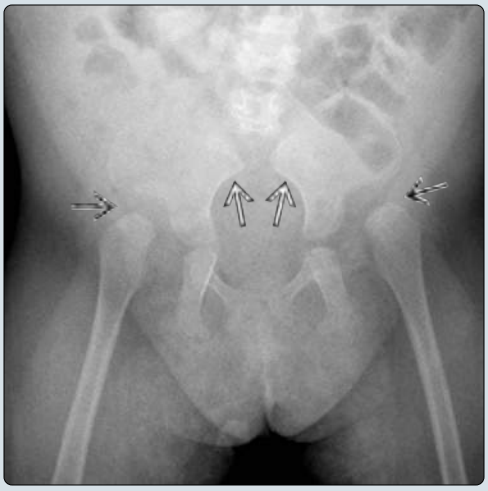
- Look for caudal spine anomalies in patients with genitourinary &/or anorectal malformations

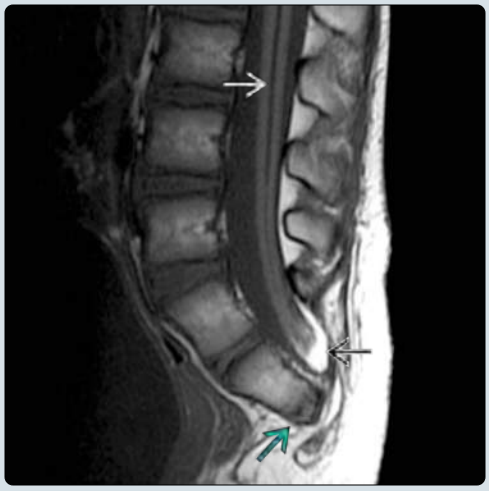
(Left) *Longitudinal thoracolumbar spinal ultrasound of an infant shows a blunt wedge-shaped termination of the spinal cord ⇨ at the T11 level, typical of group 1 caudal regression syndrome (CRS).* **(Right)** *Sagittal T2 MR of the lumbosacral spine in an infant with group 1 CRS shows an absence of the sacrum ⇨, a blunted wedge-shaped conus ⇨, & a "double-bundle" appearance of the spinal nerve roots ⇨ clumped anteriorly & posteriorly.*





(Left) *Frontal radiograph of the same infant with group 1 CRS demonstrates a complete absence of the sacrum with a narrow pelvis & medial positioning of the iliac bones ⇨. There is hypoplasia of the lower lumbar spine as well as bilateral hip dysplasia with superolateral dislocations ⇨.* **(Right)** *Sagittal T1 MR in a 5 year old with group 2 CRS shows sacral hypoplasia ⇨ with a low-lying conus attached to an intradural lipoma ⇨. Note the mild dilation of the central canal diffusely ⇨.*





KEY FACTS

TERMINOLOGY

- Traditional terms diastematomyelia & diplomyelia out of favor as these entities remain difficult to differentiate

IMAGING

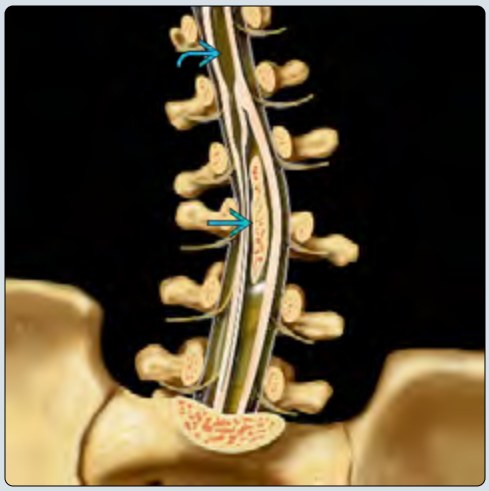
- Split cord malformation (SCM): Sagittal division of spinal cord into 2 hemicords
  - Hemicords have variable symmetry & may or may not unite below cleft
- Divided into 2 types
  - Type I SCM: 2 separate dural tubes, usually with bony/cartilaginous cleft
  - Type II SCM: Single dural sac, usually with thin separating fibrous band
- Location most commonly thoracolumbar (> 85%)
- Radiographs detect septum in < 50% of cases, may show widening of interpediculate distances
- ~ 85% of cases associated with other spinal anomalies
  - Open or closed spinal dysraphisms
  - Hydromyelia, lipoma, dermal sinus, (epi)dermoid, tethering adhesions (meningocele manqué)
  - Segmentation anomalies, kyphoscoliosis

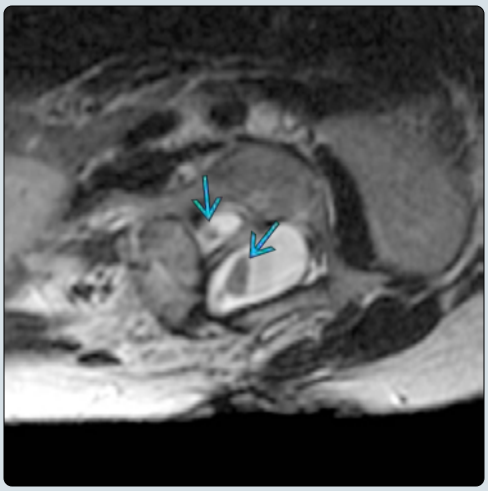
PATHOLOGY

- Believed to be result of adhesion between ectoderm & endoderm during gastrulation

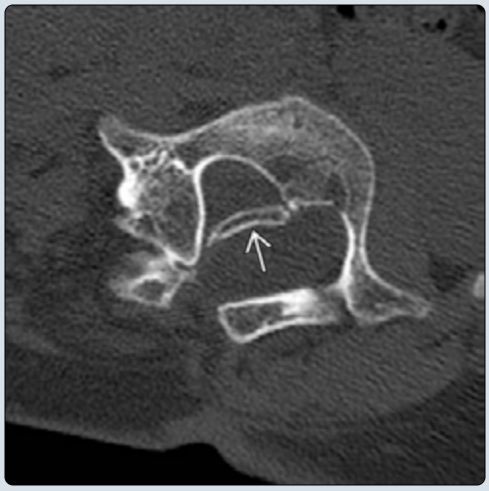
CLINICAL ISSUES

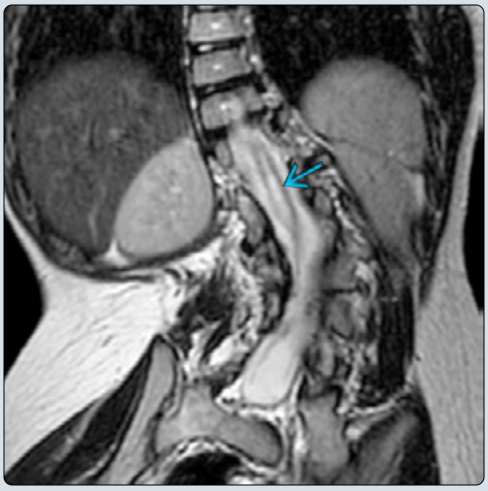
- Usually presents in early childhood
  - Cutaneous stigmata on back in > 50%
    - Hypertrichosis (hairy patch) most common
  - Progressive kyphoscoliosis in older children & adults
  - Orthopedic foot problems (~ 50%)
  - Bladder & bowel dysfunction
- Prophylactic surgery to prevent symptom progression, especially type I SCM
  - Resection of spur, lysis of adhesions, detethering





(Left) *Coronal graphic of the lumbar spine demonstrates a type I split cord malformation (SCM) with an osseous spur splitting the low-lying syringomyelic spinal cord. Both hemicords continue below the S1 level.* **(Right)** *Axial T2 MR in a 6 year old with a type I SCM at the level of the mid lumbar spine demonstrates 2 hemicords within dural sacs that are separated by a bony spur. The hemicords are asymmetric in size.*





(Left) *Axial bone CT in the same patient with a type I SCM at the level of the mid lumbar spine more clearly delineates the bony septum*. **(Right)** *Coronal T2 MR in the same patient demonstrates a spinal cord syrinx above the level of the SCM. There is also marked scoliosis.*

Terminal Myelocystocele

KEY FACTS

IMAGING

- Closed (skin-covered) spinal dysraphism with bulging subcutaneous cystic mass
- Hydromyelic, low-lying spinal cord extends through osseous defect of lumbosacral spine into meningocele sac & attaches to subcutaneous fat
  - "Trumpet flaring" of distal cord
  - Cyst-within-cyst appearance of distal cord dilated central canal surrounded by meningocele
    - Cord cyst does not communicate with subarachnoid space
- ± T1-hyperintense lipoma (lipomyelocystocele), holocord syrinx; rarely, Chiari II malformation

TOP DIFFERENTIAL DIAGNOSES

- Simple dorsal meningocele
- Lipomyelomeningocele
- Other closed spinal dysraphisms
- Sacrococcygeal teratoma

PATHOLOGY

- Exact etiology unclear; may be due to anomaly of caudal cell mass during secondary neurulation
- Associated malformations: OEIS (omphalocele, cloacal exstrophy, imperforate anus, spinal anomalies), caudal agenesis, Chiari II malformation (not as common as open spinal dysraphisms)

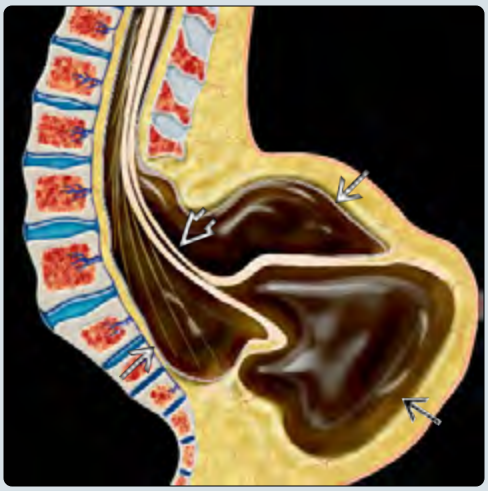
CLINICAL ISSUES

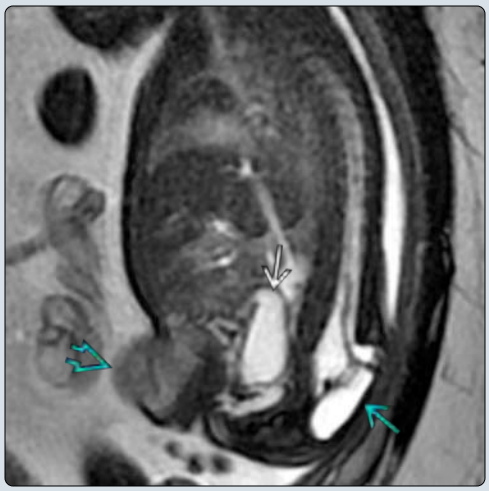
- Presents at birth with lumbosacral skin-covered mass
- Can be neurologically intact at birth with progressive lower extremity sensorimotor deficits
- Early neurosurgical intervention recommended

DIAGNOSTIC CHECKLIST

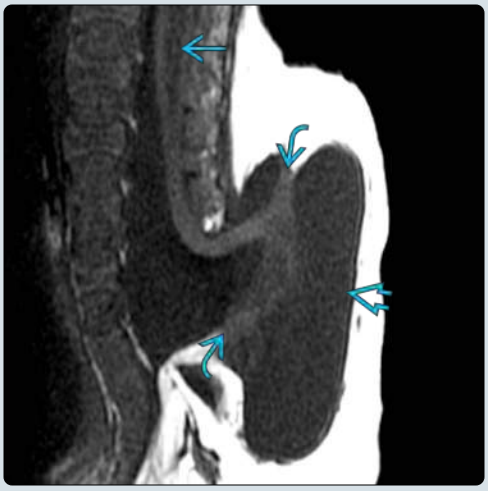
- Early diagnosis & surgery → best chance for normal neurological outcome
- Nonneurological prognosis largely linked to severity of associated anomalies

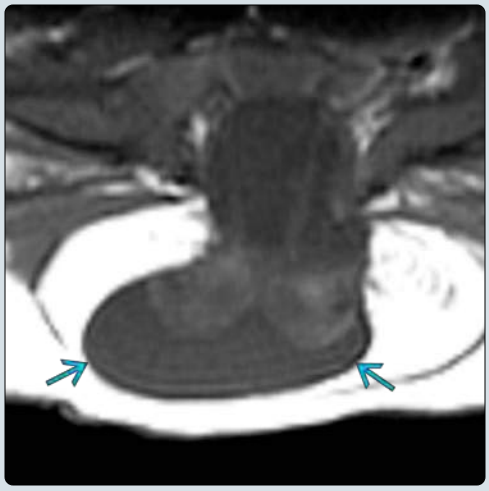
(Left) *Sagittal graphic displays a closed (skin-covered) dysraphism with a low-lying, hydromyelic spinal cord piercing an expanded subarachnoid space (meningocele) & terminating in a myelocystocele.* **(Right)** *Sagittal SSFP MR of a fetus at 28 weeks gestational age demonstrates a terminal myelocystocele. There are additional findings of the OEIS complex, including a low omphalocele & an obstructed distal colon.*





(Left) *Postnatal T1 MR in the same patient 11 weeks after delivery confirms the presence of a skin-covered terminal myelocystocele with splaying of the neural elements around a fluid collection that does not communicate with the surrounding subarachnoid space. There is also syringohydromyelia.* **(Right)** *Axial T1 MR in the same patient shows the myelocystocele, which is covered by skin & fat.*





Neurenteric Cyst

KEY FACTS

TERMINOLOGY

- Cyst along neuraxis lined with mucin-secreting cuboidal or columnar epithelium resembling alimentary tract
  - Derived from displaced endodermal tissue
- Synonyms: Enteric, enterogenous, or endodermal cyst

IMAGING

- Cyst along neuraxis (usually intraspinal) ± vertebral anomalies
- Most are intradural, extramedullary simple unilocular cysts ventral to spinal cord
- Can be associated with other closed spinal dysraphisms
- ± associated fistulae & mediastinal/abdominal cysts

TOP DIFFERENTIAL DIAGNOSES

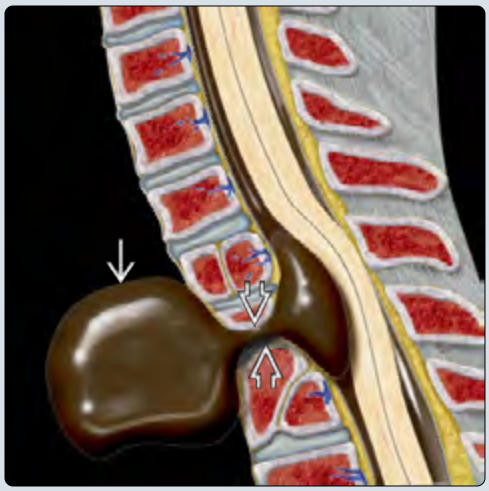
- Arachnoid cyst
- (Epi)dermoid cyst
- Anterior thoracic meningocele

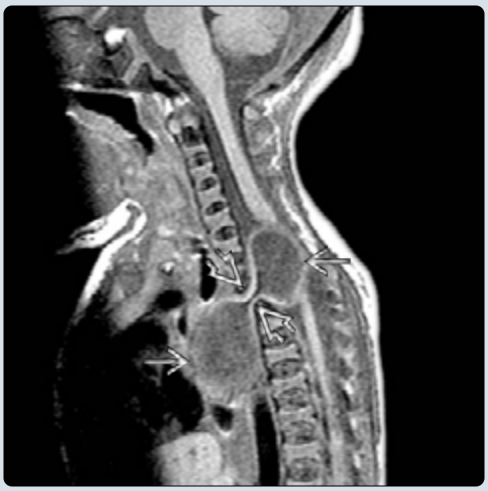
PATHOLOGY

- Histologic diagnosis, though diagnosis can be suggested in presence of vertebral anomalies
- Enteric & spinal structures connected through persistent neurenteric canal (a.k.a. canal of Kovalevsky)
- Subgroup of split notochord syndrome spectrum
  - Sporadic or syndromic (Klippel-Feil, VACTERL, OEIS)
  - Associated with vertebral anomalies, split cord malformation, lipoma, dermal sinus tract, & tethered spinal cord

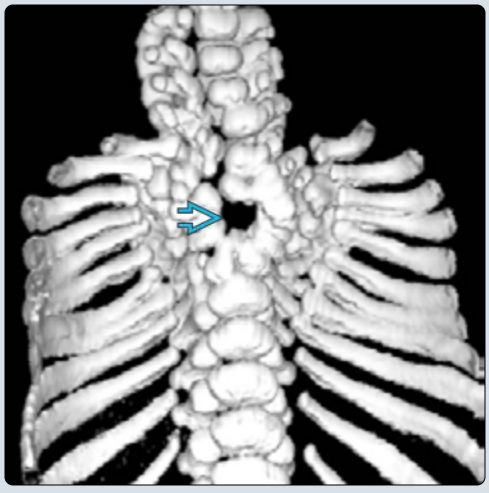
CLINICAL ISSUES

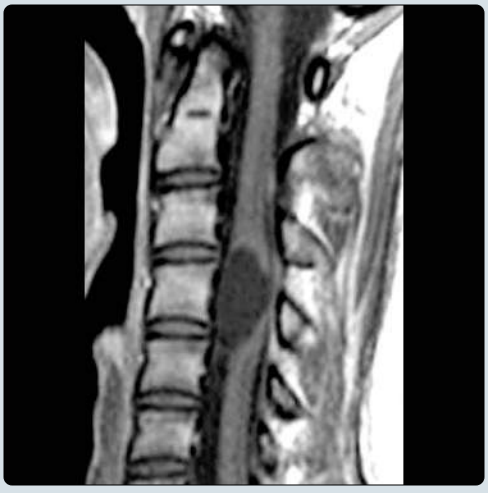
- Some asymptomatic but most show progressive neurological deterioration
  - Location, size of cyst, degree of cord compression, & severity of associated anomalies determine prognosis
- Usually present in adolescents; not typically symptomatic in infants unless large
- Best treatment (not always possible): Complete excision





(Left) *Sagittal graphic shows a large mediastinal enteric cyst ⇨ extending into the ventral spinal canal through a persistent neurenteric canal (a.k.a. canal of Kovalevsky) ⇨. There is dorsal displacement & compression of the thoracic spinal cord.* **(Right)** *Sagittal T1 MR demonstrates a large, dumbbell-shaped neurenteric cyst ⇨ extending from the mediastinum into the spinal canal through a persistent neurenteric canal ⇨. There is marked spinal cord compression. (Courtesy S. Blaser, MD.)*





(Left) *3D coronal bone CT in the same patient with a complex thoracic enteric cyst & spinal canal extension shows extensive vertebral segmentation anomalies, multiple abnormal fused ribs, & a large persistent neurenteric canal ⇨. (Courtesy S. Blaser, MD.)* **(Right)** *Sagittal T1 C+ MR in a different patient demonstrates a well-defined cystic lesion at the C4 level. The cyst invaginates into the ventral spinal cord but shows no abnormal enhancement or adjacent vertebral body remodeling.*

Tethered Spinal Cord

KEY FACTS

IMAGING

- Best clue: Low-lying conus medullaris
  - May have thickened filum terminale ± fatty infiltration or lipomatous mass
  - Accompanied by clinical signs & symptoms of tethering
    - Lower extremity weakness, spasticity, & ↓ sensation; abnormal gait; bladder dysfunction
- General imaging features
  - Conus below L2-3 disc level
  - May appear taut or directly apposed to dorsal thecal sac
    - Lack of conus motion with CSF pulsations
    - Lack of dependent ventral shift of conus when prone
  - Filum > 2 mm thick (at L5-S1 on axial/transverse images)
  - ± echogenic (US) or T1 (MR) bright fatty mass at conus
  - ± bony/soft tissue dysraphism
- Ultrasound to screen infants (< 6 months old) at ↑ risk of spinal anomalies (as suggested by certain cutaneous stigmata or associated systemic anomalies)
- MR to define underlying anomalies for surgical planning in symptomatic patients

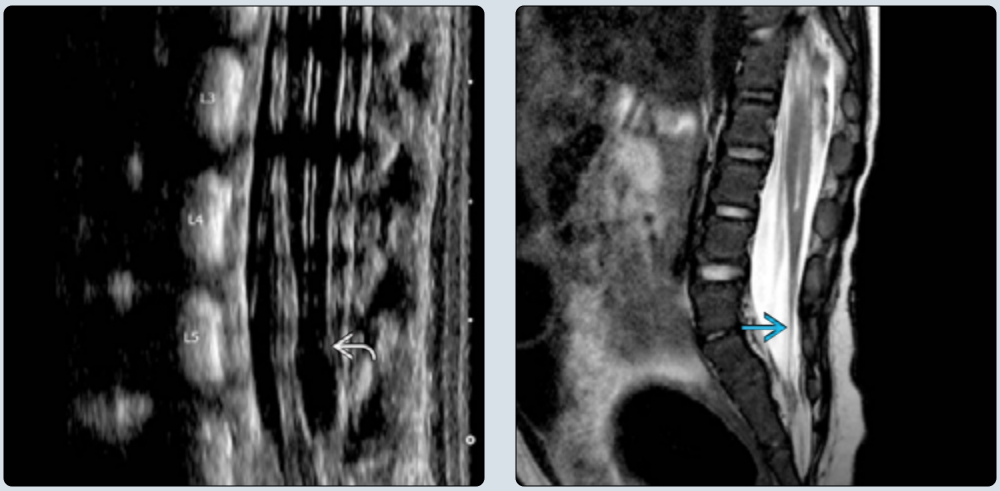
PATHOLOGY

- Cutaneous stigmata in up to 50%
  - Hairy patch, hemangioma, skin tag, atypical dimple
- Tethering also found in clinically apparent open & closed spinal dysraphism
- Tethered filum histologically abnormal
  - ↑ connective tissue with dense collagen fibers, hyalinization, & dilated capillaries

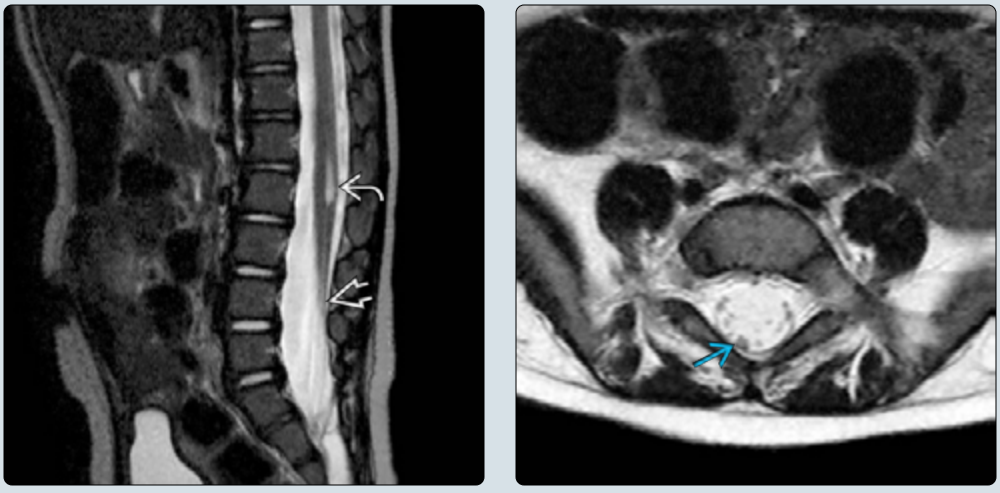
CLINICAL ISSUES

- Tethered cord syndrome: Clinical diagnosis; imaging in specific screening circumstances or to detect associated anomalies for surgical planning
- Symptomatic presentation most common during rapid growth (4-8 years of age & adolescent growth spurt)
- Majority show improvement or stabilization of neurological deficits following surgical untethering

(Left) *Sagittal ultrasound of the lumbar spine in an 8-day-old infant with the VACTERL association shows that the conus medullaris extends caudally to the level of L5-S1. The conus remains dorsally positioned despite the patient being prone.* **(Right)** *Sagittal T2 MR in the same child again shows the inferior position of the conus with a thickened filum terminale, findings that suggest tethering. No mass is identified. Up to 40% of children with the VACTERL association will be diagnosed with a tethered cord.*



(Left) *Sagittal T2 MR in a 5 month old with a cutaneous birthmark over the lower spine shows an abnormally low & dorsal position of the conus medullaris. The mild prominence of the distal spinal cord central canal is a ventriculus terminalis, a normal finding in infants.* **(Right)** *Axial T2 MR at the level of L5-S1 in the same infant further demonstrates the abnormal thickening of the filum terminale, which is directly apposed to the dorsal aspect of the thecal sac. These imaging findings are typical of tethering.*



KEY FACTS

TERMINOLOGY

- Primary curvature: Curvature with greatest angulation
- Secondary or compensatory curvature: Smaller curve, which balances primary curvature
- Structural curve: Curvature that will not correct with ipsilateral bending; included in fusions
- Terminal vertebra: Most tilted vertebra in single curve

IMAGING

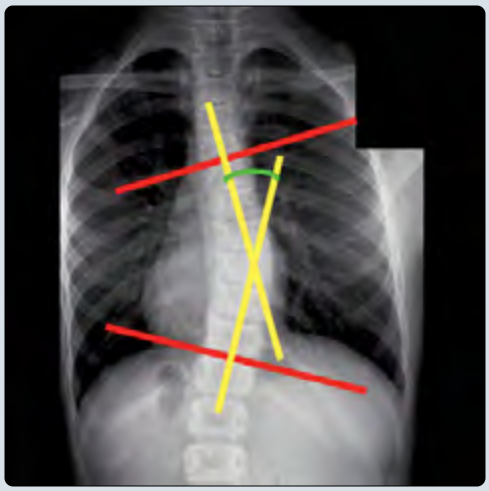
- Method of Cobb standard for measuring scoliosis
  - Draw lines parallel to endplates of terminal vertebrae
  - Draw intersecting lines perpendicular to 1st lines
  - Cobb angle lies at superior or inferior intersection
    - Represents angle between terminal vertebrae
- CT best tool for anatomic bony detail
  - 3D reconstructions helpful for surgical planning
- MR best tool for spinal cord & intraspinal contents
  - Assess for syrinx, tumor, tethering lesions in atypical curves

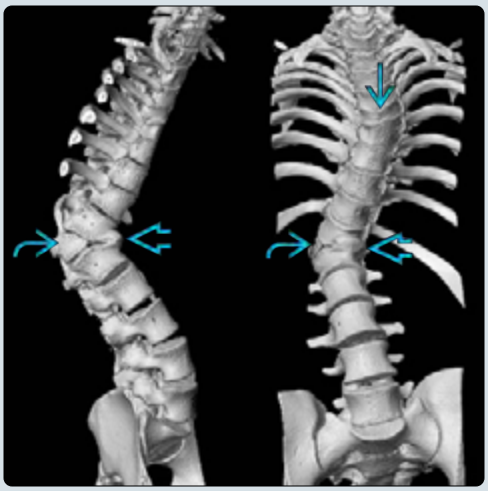
TOP DIFFERENTIAL DIAGNOSES

- Idiopathic scoliosis
  - Infantile, juvenile, adolescent
  - 80% of scoliosis with adolescent most common by far
    - Infantile tends to be stable
    - 5% of adolescent onset progress
    - Up to 95% of juvenile onset will progress
- Congenital scoliosis
  - Osteogenic or neuropathic
  - Progresses in 75%
- Neuromuscular scoliosis
  - Neurologic disorders, muscular dystrophies
- Syndromic scoliosis
  - Neurofibromatosis, Marfan syndrome, Down syndrome

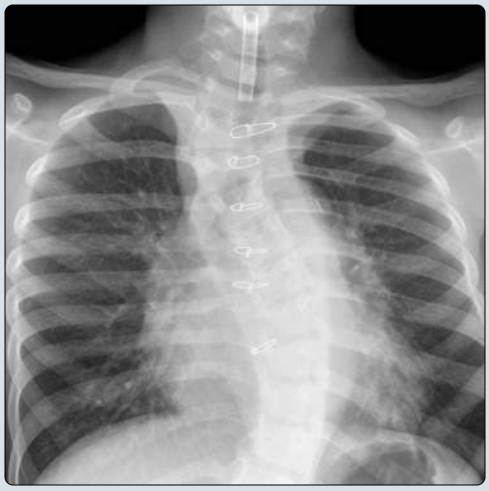
CLINICAL ISSUES

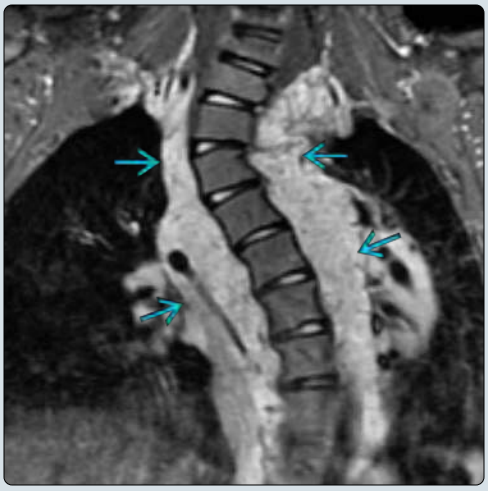
- Bracing for curves > 25°
- Fusion for rapidly progressive curves or curves > 45°





(Left) *AP radiograph (viewed from the orthopedist's perspective) shows the method of Cobb for measuring scoliosis angles: Lines are drawn parallel to the superior & inferior endplates of terminal vertebrae (red) with intersecting lines (yellow) then drawn perpendicular to the 1st lines. This provides the Cobb angle (green).* **(Right)** *Lateral & frontal 3D CT images of a teenager with scoliosis show a T11 hemivertebra with fusion of the adjacent vertebral bodies. There is also a T4 butterfly vertebra.*





(Left) *PA chest radiograph in a teenager with neurofibromatosis type 1 (NF1) shows a short segment upper thoracic curve. Short curves (< 6 segments) often indicate underlying pathology.* **(Right)** *Coronal STIR MR in the same patient shows extensive paraspinal plexiform neurofibromas associated with the scoliosis. Up to 1/3 of children with NF1 have spinal deformities (nearly always associated with paraspinal neurofibromas).*

Syringomyelia

KEY FACTS

IMAGING

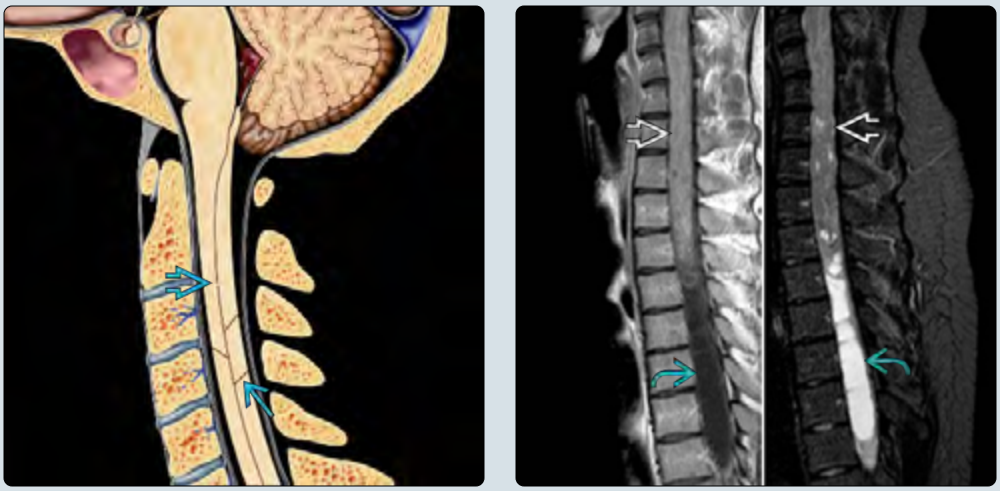
- Overlap between incidental (nonprogressive) & pathological central canal dilation
  - Diameter of normal central canal can be up to 4 mm
  - Do not label central canal of 1-2 mm as syrinx
- Cystic cavity typically centrally located on axial images
  - Off-midline location ↑ concern for neoplastic, traumatic, or inflammatory etiology
- Extent & morphology best demonstrated on T2 MR
  - Hyperintense intramedullary cavity ± adjacent gliosis, myelomalacia
  - Axial images more reliable for measurement
  - Volumetric/3D sequences may demonstrate adhesions
    - SSFP (e.g., CISS, FIESTA) also reduces flow artifact
  - Carefully examine for features suggesting neoplastic or inflammatory etiology
    - Nodularity/mass, signal abnormality in adjacent cord
    - Give contrast if atypical features present
- US useful for screening < 6 months of age

PATHOLOGY

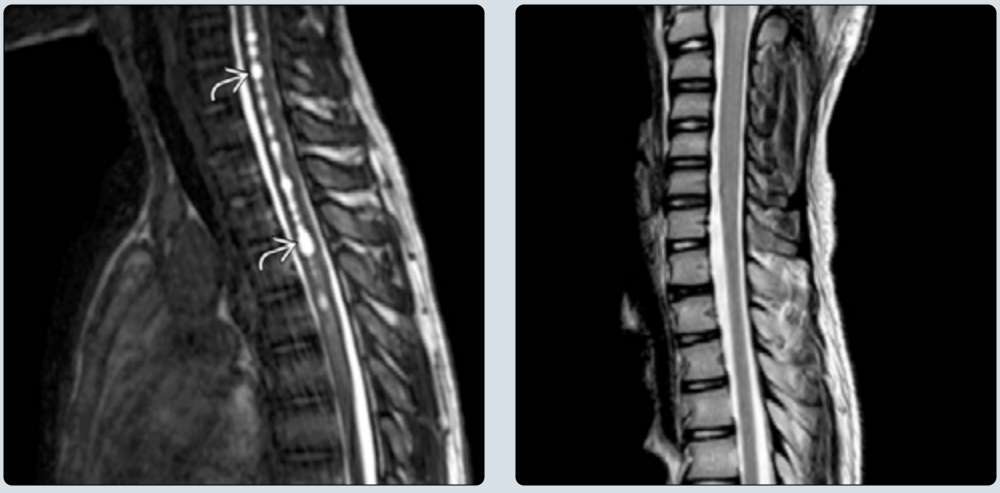
- Lesions obstructing both central canal & subarachnoid space (SAS) can cause syrinx
  - Tumor, inflammation, Chiari 1 or 2
    - Up to 45% of primary spinal cord tumors have syrinx at presentation
- Posttraumatic or postinflammatory myelomalacia can lead to cavitation
- Majority of pediatric causes congenital
  - 10-15% of children with Chiari 1 present with syrinx
  - Combination of scoliosis + Chiari 1 associated with larger cavities

CLINICAL ISSUES

- Suboccipital decompression can reverse/resolve syrinx associated with Chiari malformations



(Left) *The normal central canal has multiple small obstructions, but it also has small channels that lead from the central canal to the subarachnoid space, allowing for the transmission of CSF between the 2 compartments. Because of this, flow must be impeded in both compartments for a syrinx to develop.* **(Right)** *Sagittal T1 C+ FS (left) & STIR (right) MR images in a teenager with an ependymoma show a syrinx caudal to the tumor. The tumor obstructs both the central canal & the subarachnoid space.*



(Left) *Sagittal SSFP/FIESTA MR in a 12 year old with a Chiari 1 malformation (not shown) demonstrates a multiloculated syrinx involving the cervical & thoracic cord.* **(Right)** *Sagittal T2 MR in the same patient 5 months later (after a suboccipital decompression surgery to relieve the obstruction at the foramen magnum caused by the Chiari 1 malformation) shows complete resolution of the syrinx.*

IMAGING

General Features

- Location
  - Intramedullary spinal cord
  - Typically centrally located on axial images
    - Off-midline location of ↑ concern for neoplastic, traumatic, or inflammatory etiology
  - Thoracic > cervicothoracic > cervical
- Size
  - Cavity diameter: Small → markedly dilated
    - Overlap between incidental (nonprogressive) & pathological central canal dilation
      - Normal central canal may be up to 4 mm
      - True syrinx associated with cord expansion
  - Cavity length: Short segment (1 or 2 levels) to holocord
- Morphology
  - CSF-filled cavity in spinal cord
    - Follows CSF in signal & attenuation
      - May have flow artifact (seen in larger lesions)
    - May be septated & lobulated or smooth & simple

MR Findings

- T2WI
  - Hyperintense cystic intramedullary cavity ± adjacent gliosis, myelomalacia
  - ± flow artifact with poorly defined, heterogeneously hypointense signal throughout larger cavities
  - ± adhesions on 3D/volumetric sequences
- T1WI C+
  - Simple syrinx does not enhance; enhancement suggests inflammatory or neoplastic lesion

Imaging Recommendations

- Best imaging tool
  - MR: Although syrinx may be seen on CT, extent & associated abnormalities best assessed by MR
- Protocol advice
  - Extent & morphology best demonstrated on T2WI
    - Axial plane most reliable for diameter measurement
  - Volumetric/3D sequences may demonstrate adhesions
    - SSFP (e.g., CISS, FIESTA) also reduces flow artifact
  - Carefully examine for features suggesting neoplastic or inflammatory etiology
  - Give contrast to look for underlying lesion if atypical features present

PATHOLOGY

General Features

- Etiology
  - Central canal of spinal cord transmits CSF in rostral direction toward obex
    - Normal canal has multiple sites of obstruction/adhesion/collapse (↑ with age)
      - May function like valves in venous system
    - Multiple microscopic channels lead from canal to subarachnoid space (SAS)
      - Similar to perivascular spaces in brain
      - Allow CSF to be driven into canal from SAS
    - Lesions obstructing both central canal & SAS → syrinx
      - Tumor, inflammation
      - Chiari malformation
    - Posttraumatic or postinflammatory myelomalacia can also cause cavitation
      - Analogous to porencephaly in brain
- Associated abnormalities
  - Congenital anomalies (frequent in children)
    - Chiari 1 malformation (not always congenital)
      - 10-15% of children with Chiari 1 present with syrinx
      - May develop syrinx over time
      - Combination of scoliosis + Chiari 1 associated with larger cavities
    - Chiari 2 malformation
  - Scoliosis
  - Tumors
    - Up to 45% of primary spinal cord tumors have syrinx at presentation
      - More common in adults than children
      - Most common in hemangioblastoma, ependymoma

CLINICAL ISSUES

Presentation

- Distal upper extremity weakness, gait instability
- Cloak-like pain & temperature sensory loss
- May be asymptomatic

Natural History & Prognosis

- Variable; dependent on underlying etiology
  - Those associated with underlying malformations (Chiari 1 or 2, tethering lesions) more likely to progress
  - Spontaneous resolution rare

Treatment

- Suboccipital decompression can reverse/resolve syrinx associated with Chiari malformations
- Consider syrinx drainage with indwelling catheter only if efforts to restore normal cord CSF dynamics unsuccessful

DIAGNOSTIC CHECKLIST

Consider

- Image entire CNS for associated abnormalities
- Syrinx etiology influences treatment approach

Image Interpretation Pearls

- Do not label central canal of 1- to 2-mm diameter as syrinx
- Simple syringomyelia does not require contrast
  - Give contrast if associated cord signal abnormalities, complex morphology, focal mass, or off-midline location

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KEY FACTS

TERMINOLOGY

- Spinal cord neoplasm arising from ependymal cells

IMAGING

- Heterogeneous expansile mass, typically intramedullary
- Intratumoral or peripheral cysts or syrinx in 50-90%
- Hemorrhagic foci common in or at margins of tumor
  - Typically hyperintense on T1WI, hypointense on T2WI
  - Gradient-echo sequences ↑ sensitivity
  - ± subarachnoid hemorrhage (especially myxopapillary type) with superficial siderosis
- Heterogeneous enhancement
- Locations: Cervical > thoracic > conus
  - Myxopapillary classically at conus medullaris & filum terminale; may also occur in soft tissues of sacrococcygeal region

TOP DIFFERENTIAL DIAGNOSES

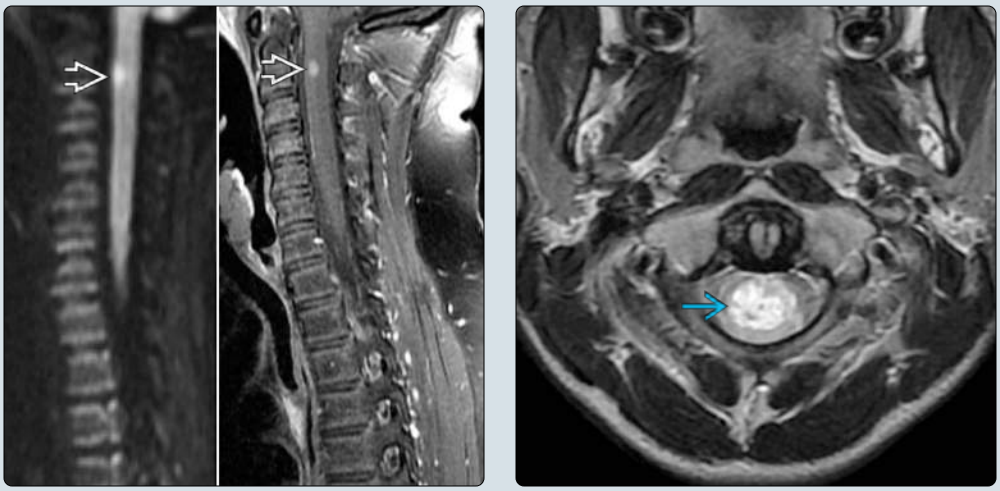
- Astrocytoma
- Cavernous malformation
- Demyelinating disease
- Cord contusion

PATHOLOGY

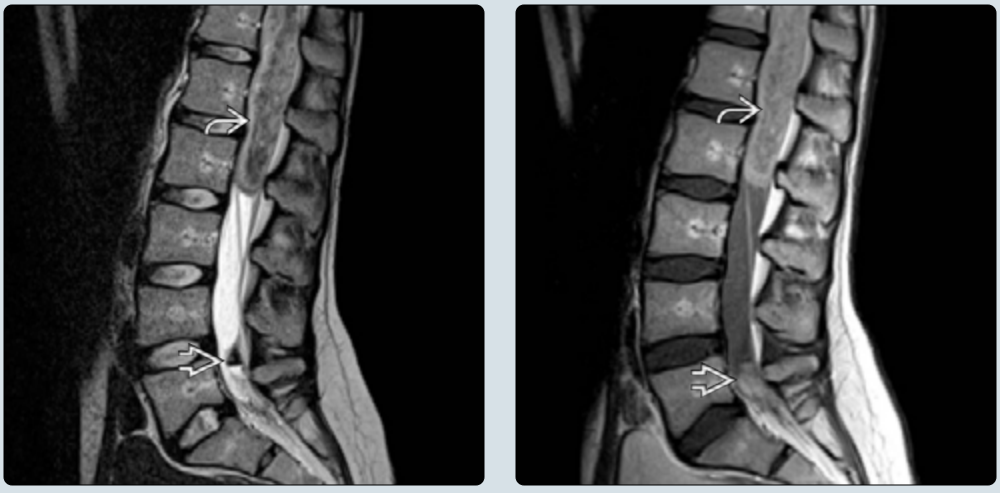
- Genetically distinct from intracranial ependymoma
- 4 subtypes: Cellular, papillary, clear-cell, tanycytic
- Mostly WHO grade II
  - Myxopapillary: WHO grade I
  - Anaplastic (rare): WHO grade III
- May occur in setting of neurofibromatosis type 2

CLINICAL ISSUES

- Neck or back pain, progressive paraparesis, paresthesias
- Gross total resection in > 85% of cases
  - Grade III, subtotal resection, or recurrent tumors require adjuvant radiation treatment
  - 5- & 10-year survival rates of 50-100% following combination of surgery & radiation



(Left) *Sagittal DWI (left) & T1 C+ (right) MR images in an 11 year old with NF2 show a small, enhancing, intramedullary lesion with diffusion restriction in the cervical cord. Ependymoma is the most common intramedullary cord lesion in NF2.* **(Right)** *Axial T2 MR through the cervical cord shows the characteristic heterogenous signal of a spinal ependymoma. These tumors are much more likely to bleed or have cystic components than spinal astrocytoma.*



(Left) *Sagittal T2 MR in a 15 year old with progressive back pain & lower extremity weakness shows a large, heterogeneous tumor enveloping the conus medullaris. There is also a hemorrhagic focus in the nerve roots at L5.* **(Right)** *Sagittal T1 C+ MR in the same patient shows mild heterogeneous enhancement of the conus & subarachnoid lesions. Myxopapillary ependymoma typically arises at the conus & often presents with hemorrhage.*

TERMINOLOGY

Definitions

- Spinal cord neoplasm arising from ependymal cells

IMAGING

General Features

- Best diagnostic clue
  - Heterogeneous spinal cord mass
- Location
  - Cervical > thoracic > conus
  - Most intramedullary
    - Myxopapillary type may present in extramedullary sites about conus medullaris & filum terminale as well as in soft tissues of sacrococcygeal region
- Size
  - Multisegmental: Typically 3-4 segments
- Morphology
  - Well-circumscribed
  - Symmetric cord expansion
  - May have exophytic component
- Typical MR features
  - Isointense to hypointense on T1WI
  - Heterogeneous but mostly hyperintense on T2WI
    - May have cystic foci
      - May be within tumor, at rostral & caudal margins, or due to reactive syrinx
  - Hemorrhagic foci in or at margins of tumor
    - Typically hyperintense on T1WI, hypointense on T2WI
    - Gradient-echo sequences ↑ sensitivity
    - Subarachnoid hemorrhage (myxopapillary type)
      - May have superficial siderosis
  - Heterogeneous enhancement

Imaging Recommendations

- Best imaging tool
  - Sagittal & axial T2WI & T1WI C+

DIFFERENTIAL DIAGNOSIS

Astrocytoma

- Most common primary cord neoplasm in children
- Less heterogeneous than ependymoma
  - Hemorrhage & cysts less common
- Often larger, can be holocord

Cavernous Malformation

- Focal hemorrhagic lesion
- Little or no enhancement
- Rare below cervical cord

Hemangioblastoma

- Older patients
- 1/3 with von Hippel-Lindau disease
- Cyst with enhancing highly vascular nodule
  - Flow voids may be present

Multiple Sclerosis

- Often multifocal; 90% have brain lesions
- Lesions more often peripheral, posterolateral
- Typically < 2 vertebral segments in length

Idiopathic Transverse Myelitis

- Cord expansion less pronounced
- Diagnosis of exclusion

Cord Infarction

- Sudden onset of symptoms
- Central gray matter typically involved

Cord Contusion

- History of trauma

PATHOLOGY

General Features

- Genetics
  - Different from intracranial ependymoma
- Associated abnormalities
  - Neurofibromatosis type 2

Staging, Grading, & Classification

- Most are WHO grade II
- Myxopapillary are WHO grade I
- Rarely WHO grade III: Anaplastic ependymoma

Microscopic Features

- Arises from ependymal cells of central canal
- 4 subtypes: Cellular, papillary, clear-cell, tanycytic
  - Cellular most common intramedullary tumor subtype

CLINICAL ISSUES

Presentation

- Neck or back pain, progressive paraparesis, paresthesias
- Delay in diagnosis due to slow growth

Demographics

- Epidemiology
  - 2nd most common primary spinal cord tumor in children
  - 30% of all ependymomas are spinal

Treatment

- Prognosis of grade I & II tumors largely dependent upon extent of resection
  - Gross total resection in > 85% of cases
- Radiation for subtotal resection, grade III, or recurrent disease
- Chemotherapy for failed surgery & radiotherapy
- Most series report 5- & 10-year survival rates of 50-100% following combination of both surgery & postoperative radiation treatment

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KEY FACTS

TERMINOLOGY

- Primary intramedullary neoplasm of spinal cord originating from astrocytes

IMAGING

- Cervical > thoracic > lumbar locations
- Usually 1-3 cm, < 4 segments in length
- Oblong, fusiform expansion of cord
- 40% have tumor cysts or syringohydromyelia
- Solid portion hypo-/isointense on T1WI, hyperintense (but not fluid bright) on T2WI MR
- Mild to moderate enhancement
- Rarely hemorrhagic

TOP DIFFERENTIAL DIAGNOSES

- Ependymoma
- Ganglioglioma
- Multiple sclerosis
- Transverse myelitis

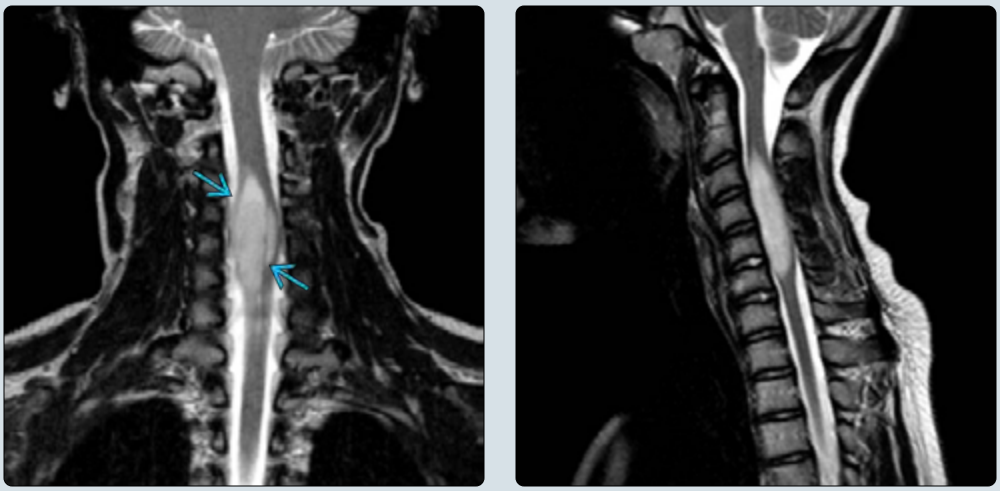
PATHOLOGY

- 80-90% low grade
  - Pilocytic astrocytoma = WHO I
    - Rosenthal fibers
    - Low prevalence of nuclear atypia/mitoses
  - Fibrillary astrocytoma = WHO II
    - ↑ cellularity, variable atypia/mitoses
    - Parenchymal infiltration
- 10-15% high grade
  - Anaplastic astrocytomas = WHO III

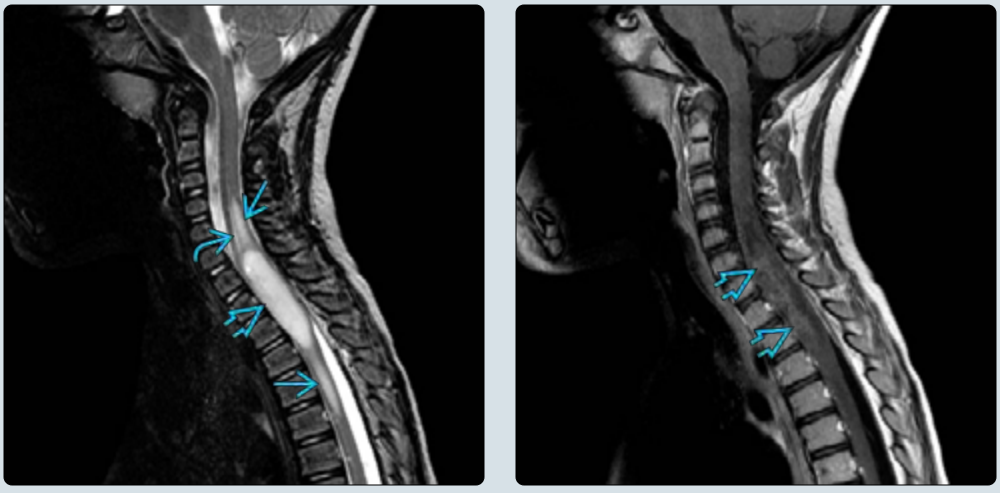
CLINICAL ISSUES

- Most common spinal cord tumor in children
- Typically in preadolescents, 5-10 years old
  - Scoliosis, pain, myelopathy, muscle wasting
- Gross total resection: Definitive treatment for pilocytic tumors
- Survival varies with tumor histology/grade & degree of tumor resection; 80% 5-year survival for low grade

(Left) *Coronal T2 MR in an 11 year old with progressive right upper extremity weakness shows an expansile tumor in the midcervical cord. The homogeneous imaging appearance is typical for a spinal cord astrocytoma.* **(Right)** *Sagittal T2 MR in the same child shows the tumor extending from C2-C3 down to C6-C7 with somewhat ill-defined superior & inferior margins. The poor delineation of the tumor from the normal cord makes complete surgical excision difficult.*



(Left) *Sagittal T2 MR in a 3 year old with intermittent torticollis shows a typical spinal astrocytoma expanding the cord at the cervicothoracic junction. Note the early dilation of the central canal & the signal extending away from the tumor into the cord. The latter may represent perilesional edema or infiltration of tumor.* **(Right)** *Sagittal T1 C+ MR in the same child shows irregular faint enhancement of the cervical spinal cord mass, which is typical of astrocytoma.*



TERMINOLOGY

Definitions

- Primary intramedullary neoplasm of spinal cord originating from astrocytes

IMAGING

General Features

- Location
  - Cervical > thoracic > lumbar
- Size
  - Usually 1-3 cm, < 4 segments
- Morphology
  - Oblong, fusiform expansion of cord
- Typical MR features
  - 40% have tumor cysts or syringohydromyelia
    - Cyst fluid slightly hyperintense to CSF
  - Solid portion hypo-/isointense on T1WI, hyperintense (but not fluid bright) on T2WI
  - Rarely hemorrhagic
  - Mild to moderate enhancement

DIFFERENTIAL DIAGNOSIS

Ependymoma

- Less common in children
- Hemorrhage common
  - May present with superficial siderosis

Ganglioglioma

- Similar imaging features
- Comprise up to 30% of cord tumors in children < 3 years old

Schwannoma

- Rarely presents as intramedullary mass

Syringohydromyelia

- Cystic intramedullary cavity without enhancement or solid component (in absence of underlying tumor)

Multiple Sclerosis

- Short-segment lesions involving < 1/2 of cord cross section, predominantly in dorsal aspect of cord

Transverse Myelitis

- Absent or mild enhancement
- Preceding vaccine or viral prodrome

PATHOLOGY

General Features

- Associated abnormalities
  - Some ↑ risk in patients with neurofibromatosis (NF1, NF2)
  - Ependymoma & extramedullary tumors more common in NF2

Staging, Grading, & Classification

- 80-90% low grade
  - Pilocytic astrocytoma = WHO I
  - Fibrillary astrocytoma = WHO II
- 10-15% high grade
  - Anaplastic astrocytomas = WHO III

Microscopic Features

- 2 main histologic subtypes: Pilocytic & fibrillary
- Pilocytic astrocytoma
  - Rosenthal fibers
  - Glomeruloid/hyalinized vessels
  - Low prevalence of nuclear atypia/mitoses
- Fibrillary astrocytoma
  - ↑ cellularity, variable atypia/mitoses
  - Parenchymal infiltration

CLINICAL ISSUES

Presentation

- Scoliosis, pain, myelopathy, muscle wasting
- Cervicomedullary lesions can cause hydrocephalus

Demographics

- Typically in preadolescents, 5-10 years old
- Most common spinal cord tumor in children
  - 30-35% of intraspinal neoplasms in children
  - 60% of primary spinal cord tumors in children

Natural History & Prognosis

- Most are slow growing
- Survival varies with tumor histology/grade & degree of tumor resection
  - 80% 5-year survival for low grade

Treatment

- Gross total resection is goal in pilocytic & low-grade tumors
  - Definitive treatment for pilocytic tumors
  - Intraoperative evoked potentials helpful for maximal resection without damaging normal tissue
  - Intraoperative US can show extent of tumor vs. syrinx

DIAGNOSTIC CHECKLIST

Consider

- Remember inflammatory lesions in differential

Image Interpretation Pearls

- Image entire spinal cord
- Subtotal surgical resection may be unimpressive on imaging
  - Residual lesion "fills in" resection cavity

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Sacrococcygeal Teratoma

KEY FACTS

TERMINOLOGY

- Teratomas made up of various parenchymal cell types from > 1 germ layer, usually all 3
- Sacrococcygeal teratoma (SCT) arises from coccyx, potentially grows both internally & externally (with latter more common)
- Both benign & malignant (17%) varieties

IMAGING

- Always with presacral components; exophytic extension more common than internal growth
- Heterogeneous mixed solid & cystic masses
  - Ca^{2+}, fat, hemorrhage, cysts, various soft tissues in mass
  - Solid components may show moderate to high vascularity
- MR best for identifying intraspinal extension

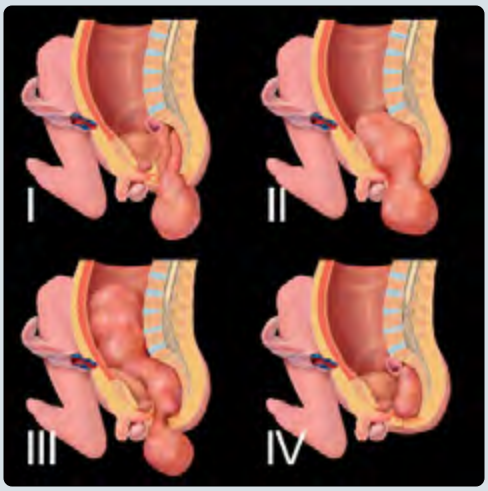
PATHOLOGY

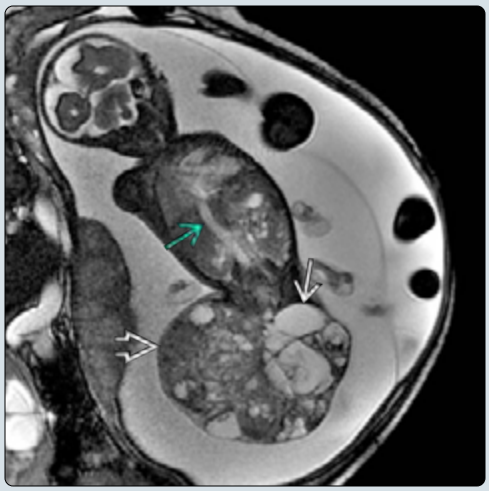
- Malignant characteristics ↑ with age, type IV
- Currarino triad: Presacral mass (usually anterior meningocele or SCT), anorectal malformation, sacral anomalies
- **American Association of Pediatric Surgery Section classification**
  - Type I (47%): Primarily external in location
  - Type II (34%): Dumbbell shape, equal pelvic & external components
  - Type III (9%): Primarily located within abdomen/pelvis
  - Type IV (10%): Entirely internal, no external component

CLINICAL ISSUES

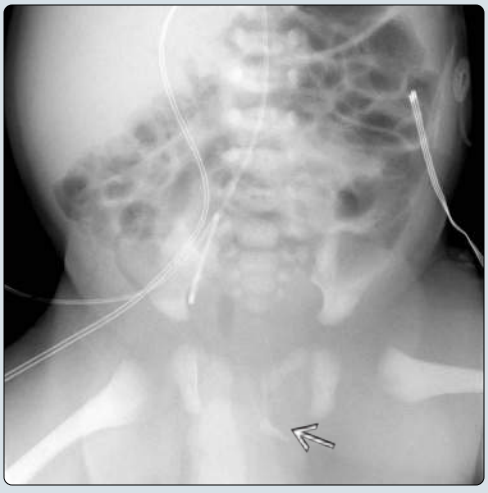
- Prognosis excellent in patients with benign tumors after birth
  - Fetal SCT can be quite vascular & grow rapidly → hydrops, intratumoral hemorrhage, rupture → demise
    - Hydrops before 30 weeks gestation > 90% mortality
- Complete surgical resection to include coccyx
- 5-15% risk of recurrence

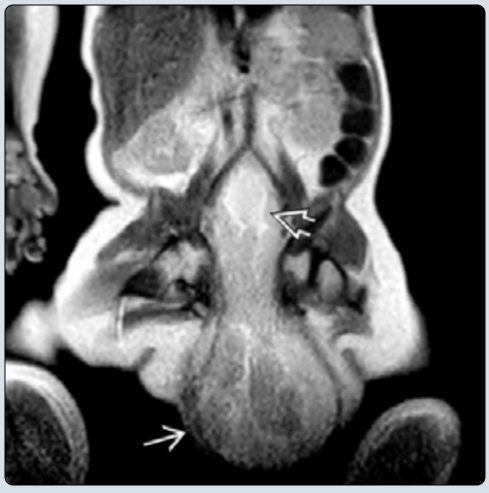
(Left) *Graphic shows tumor classification scheme: Type I is primarily exophytic, type II has equivalent size internal & external masses, type III has a larger intraabdominal component, & type IV is entirely internal.* **(Right)** *Coronal SSFP MR of a twin gestation shows a fetus with a large, predominantly exophytic mass extending from the perineum. Note the solid ⇨ & cystic ⇨ areas within the mass. The inferior vena cava ⇨ is prominent due to the amount of blood flow coursing through this tumor. However, no hydrops is seen.*





(Left) *AP radiograph in a newborn boy with a protruding buttock mass shows soft tissue fullness in the pelvis & perineum with irregular Ca^{2+} ⇨ below the pubic bones.* **(Right)** *Coronal T2 MR in the same infant shows a heterogeneous solid mass, which protrudes externally ⇨ with dumbbell extension internally ⇨, splaying the aortic bifurcation in this case of a type II sacrococcygeal teratoma (SCT).*





TERMINOLOGY

Abbreviations

- Sacrococcygeal teratoma (SCT)

Synonyms

- Teratoma, germ cell tumor of coccyx

Definitions

- Teratomas made up of various parenchymal cell types from > 1 germ layer, usually all 3
  - Tumors may contain hair, teeth, cartilage, & fat, amongst other tissues
- SCT arises from coccyx, potentially grows both internally & externally (with latter more common)
- Both benign & malignant varieties

IMAGING

General Features

- Best diagnostic clue
  - Large solid, cystic, or mixed mass with large exophytic perineal/buttock component & relatively small presacral component
  - May see Ca^{2+}, bone, fat, fluid levels, various soft tissues in mass
- Location
  - Origin from coccyx, but growth can occur in any direction
    - Presacral components always present
  - Look for intraspinal extension
- Size
  - Ranges from only few mL to massive in volume, potentially exceeding fetal weight
- Morphology
  - Classically heterogeneous: Multiple tissue types, Ca^{2+}

Radiographic Findings

- Typically show large mass extending outside infant
- Ca^{2+} may be present

Fluoroscopic Findings

- Type IV SCT may be found incidentally during fluoroscopy studies for constipation or voiding problems
- Patients rarely have long-term sequela of bladder outlet obstruction

CT Findings

- NECT
  - Demonstrates fatty components, Ca^{2+}, & fluid levels well
  - Heterogeneous mass wrapped around coccyx but typically without bony destruction
- CECT
  - Variable enhancement pattern in solid & cystic components

MR Findings

- Heterogeneous, similar to CT
- Fat best confirmed on T1WI without & with FS
- Variable enhancement with gadolinium that does not predict malignancy
- Best study for identifying intraspinal extension

Ultrasonographic Findings

- Grayscale ultrasound
  - Ultrasound may be limited due to large size & internal Ca^{2+}
  - Heterogeneous echotexture mass
    - Ca^{2+} & fat cause highly echogenic foci while cystic areas are hypo- to anechoic
- Color Doppler
  - Solid components may show moderate to high vascularity
  - Arterial steal may be seen in nearby vessels to supply tumor

Nuclear Medicine Findings

- PET
  - Used more often in evaluation of malignant recurrence than in initial diagnosis

Imaging Recommendations

- Best imaging tool
  - Prenatal sonography most common initial diagnostic modality
  - Prenatal or postnatal MR to determine full extent of mass & aid surgical planning

DIFFERENTIAL DIAGNOSIS

Pelvic Rhabdomyosarcoma

- Solid mas without Ca^{2+}, fat, or substantial cysts (usually)

Neuroblastoma

- May arise in pelvis at organ of Zuckerkandl
- Solid mass with Ca^{2+}

Myelomeningocele or Myelocystocele

- Dorsal dysraphism; neural placode continuous with abnormal spinal cord

Other Intrapelvic Masses

- Consider Burkitt lymphoma, ovarian tumors, lymphatic malformation, hematometrocolpos

PATHOLOGY

General Features

- Etiology
  - Probably results from rests of pluripotential cells at caudal end of notochord/spine
- Genetics
  - Not inherited
- Associated abnormalities
  - 10% of SCTs associated with other congenital anomalies, primarily defects of hindgut & cloacal region, which exceeds baseline rate of 2.5% expected in general population
  - SCT 2nd most common lesion after anterior meningocele in familial disorder of Currarino (autosomal dominant triad of presacral mass, partial sacral agenesis, & anorectal defects)
  - Familial tendency reported, prompting some to recommend screening asymptomatic siblings
- Malignant characteristics increase with
  - Age at diagnosis

- Internal subtype (type IV worst)
- Male gender
- Presence of necrosis or hemorrhage

Staging, Grading, & Classification

- **American Association of Pediatric Surgery Section classification**
  - Type I (47%)
    - Primarily external in location
  - Type II (34%)
    - Dumbbell shape, equal internal/external components
  - Type III (9%)
    - Primarily located within abdomen/pelvis
  - Type IV (10%)
    - Entirely internal, no external component visible

Gross Pathologic & Surgical Features

- Typical of all teratomas: Multiple tissue types in varying stages of maturation & differentiation
- Solid & cystic components common

Microscopic Features

- Tumors can be mature or immature
- Only 17% of SCT have malignant features

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Frequently diagnosed in utero
    - Large fetal tumors require C-section delivery or in utero surgical intervention (open fetal surgery, radiofrequency or thermal ablation of tumor)
  - Most other tumors diagnosed within 1st few days of life
    - Exophytic masses easily diagnosed clinically
    - Entirely internal masses may have delayed diagnosis, presenting with urinary symptoms or constipation
- Other signs/symptoms
  - In utero complications: Fetal SCT can be quite vascular & grow rapidly → hydrops, intratumoral hemorrhage or rupture → fetal demise
    - Hydrops before 30 weeks gestation > 90% mortality rate
      - Solid masses more likely to develop hydrops
      - Doppler parameters may help predict hydrops
    - Maternal mirror syndrome (preeclampsia) may develop

Demographics

- Age
  - Typically diagnosed in fetus or newborn
  - Delayed diagnoses possible in 1st year of life, rarely even later
- Epidemiology
  - Prevalence: 1 in 14,000 (Sweden) to 40,000 births

Natural History & Prognosis

- Prognosis excellent in patients with benign tumors after birth
  - Gait abnormalities & early arthrosis reported with extensive pelvic muscle resection
  - Rarely with long-term bladder & bowel issues
- 5-15% risk of recurrence
  - Intraoperative tumor spillage & incomplete resection ↑ risk
- Masses diagnosed after 1st birthday & located only internally have higher malignant potential
  - Prognosis variable in malignant tumors
  - Serum α-fetoprotein is useful tumor marker postoperatively
- Malignant components may be found at presentation or at time of recurrence
  - Yolk sac tumor most common
  - Embryonal carcinoma 2nd most common
  - With intensive chemotherapy, 5-year relapse-free survival rate reported between 76-90%
- Study from Netherlands showed normal fertility in adult female survivors

Treatment

- Complete surgical resection to include coccyx
- Benign tumors do not require additional therapy
- Malignant tumors treated with chemotherapy (platinum-based agents) & radiation
- Consideration of in utero intervention with development of hydrops; must weigh benefits with risks of prematurity

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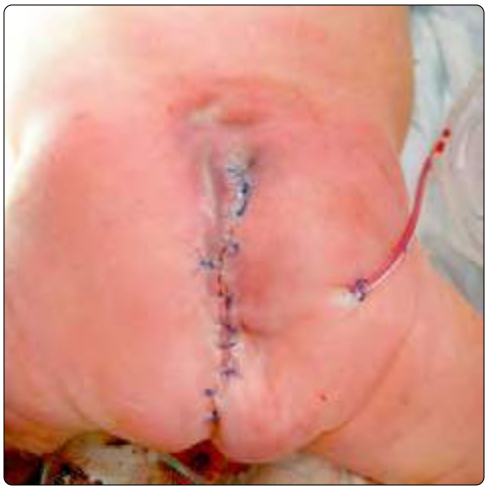
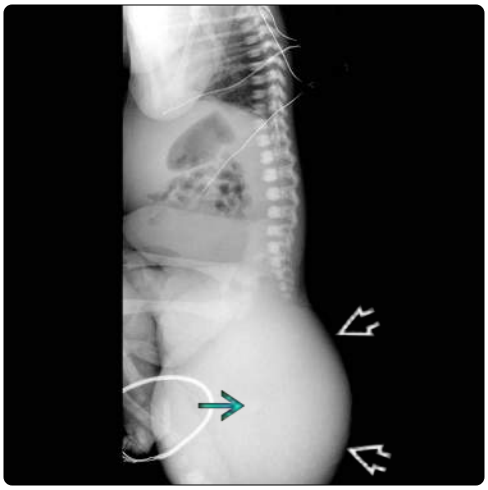
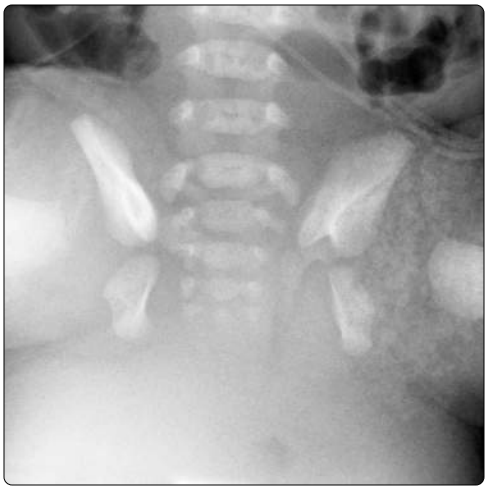
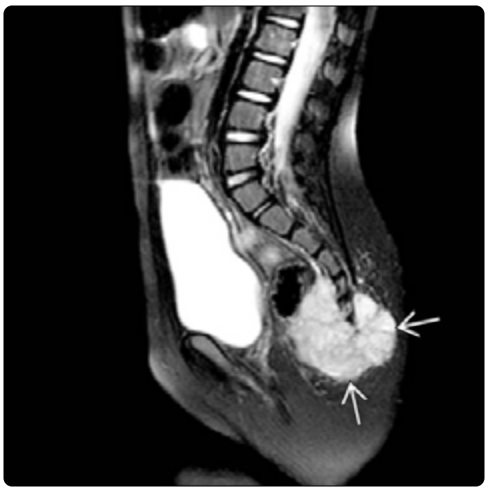


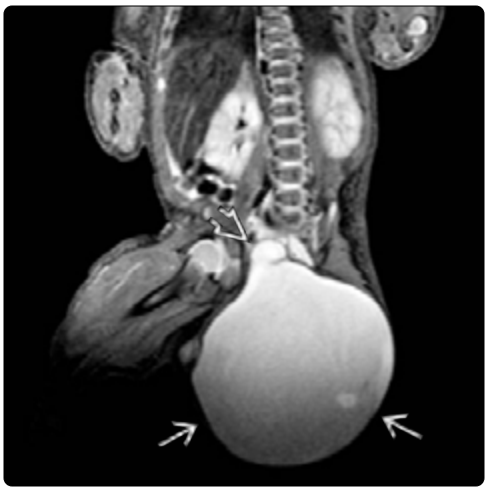
(Left) *Clinical photograph shows an infant prior to resection of a type I SCT. Cross-sectional imaging is helpful to delineate the intrapelvic/intraabdominal extent of tumor & develop an appropriate surgical plan.* **(Right)** *Clinical photograph shows the immediate postoperative appearance of the buttocks following SCT resection. After several months, the gluteal crease & buttocks assumed a nearly normal appearance.*





(Left) *Lateral radiograph shows a newborn infant with a large exophytic mass ⇨ extending from the sacrum & coccygeal region. A few calcifications ⇨ are visible within the mass, typical of a SCT.* **(Right)** *Frontal radiograph of the same infant shows widely spaced pubic & ischial bones, which are splayed by the large tumor.*





(Left) *Sagittal T2 FS MR shows a 3-year-old boy who was imaged for the changing appearance of his sacral dimple with firmness to palpation of the area. A high signal intensity solid mass ⇨ surrounds the coccyx but does not reach the skin surface.* **(Right)** *Coronal T2 FS MR in an infant shows a largely cystic external component ⇨ & a relatively small intrapelvic component ⇨ to a type I SCT.*

Discitis/Osteomyelitis

KEY FACTS

TERMINOLOGY

- Bacterial suppurative infection of intervertebral disc & adjacent vertebrae

IMAGING

- Disc-centered process: Disc space narrowing & adjacent endplate irregularity in young children
  - Lumbosacral (75%) > thoracic > cervical spine
- Early: MR imaging most sensitive, specific modality
  - Abnormal disc signal & enhancement
  - Ill-defined abnormal marrow signal & enhancement
  - Paraspinal & epidural phlegmon or abscess
- Subacute
  - Endplate destruction/erosion
- Chronic
  - Increased bone density ± vertebral fusion with healing

TOP DIFFERENTIAL DIAGNOSES

- Langerhans cell histiocytosis
- Spinal metastases
- Chronic recurrent multifocal osteomyelitis (CRMO)
- Degenerative endplate changes

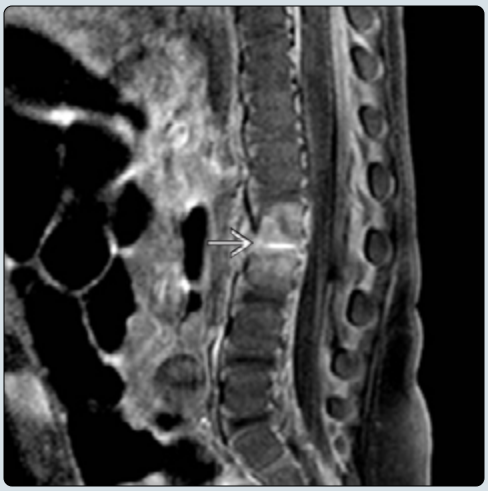
PATHOLOGY

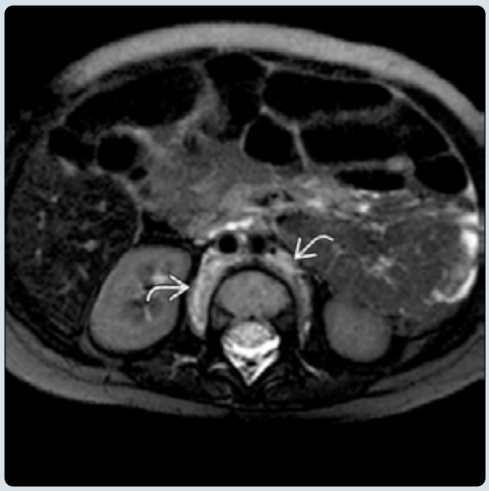
- Hematogenous spread to vascularized disc or subchondral vertebral growth plate in children
- *Staphylococcus aureus* is most common pathogen

CLINICAL ISSUES

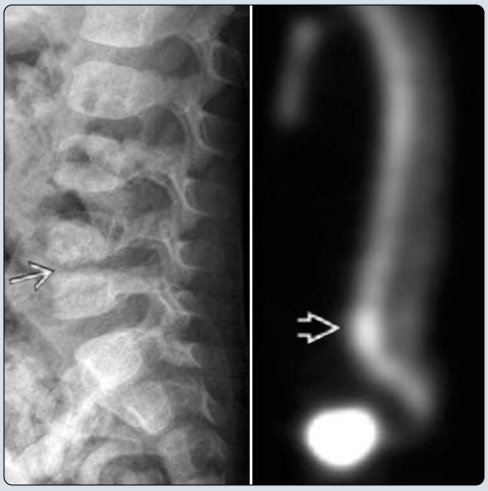
- Peak age: 6 months to 4 years
- Variable & nonspecific symptoms
  - May delay diagnosis for weeks in children
- Elevated erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), white blood cell count (WBC)
- Treat early with empiric IV antibiotics with broad-spectrum coverage until causative pathogen isolated, then organism-specific parenteral antibiotics for 6-8 weeks

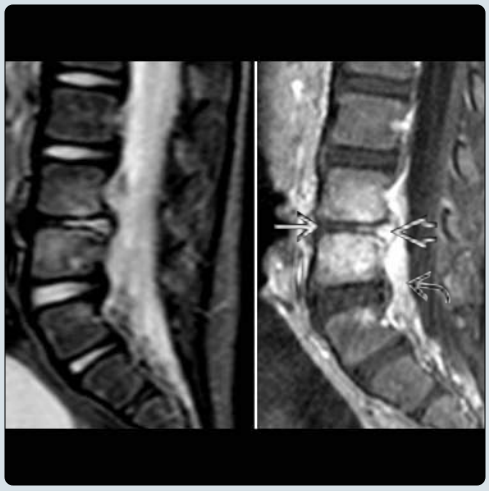
(Left) *Midline sagittal T1 C+ FS MR in a 7-month-old shows poor definition & abnormal enhancement of the L2-3 disc space. Patchy abnormal enhancement is also noted in the marrow of the adjoining vertebral bodies. No drainable collection is identified within the adjacent paraspinal soft tissues or epidural space.* **(Right)** *Axial T2 MR in the same patient shows abnormal thickening & poorly defined fluid signal in the anterior & lateral paraspinal soft tissues in this patient with spondylodiscitis.*





(Left) *Lateral radiograph (left) shows L4-5 disc space narrowing & endplate irregularity in this 1 year old with back pain. Sagittal SPECT bone scan (right) confirms abnormal uptake in the vertebral bodies, typical of spondylodiscitis.* **(Right)** *Sagittal STIR (left) & T1 C+ FS (right) MR images in a 2 year old show disc space narrowing & enhancement with abnormal signal/enhancement of the L4-5 vertebral marrow. Enhancing soft tissue in the epidural space is due to contiguous inflammation.*





TERMINOLOGY

Definitions

- Bacterial suppurative infection of intervertebral disc & adjacent vertebrae

IMAGING

Radiographic Findings

- Radiography
  - Negative up to 2-8 weeks after onset of symptoms
  - Initial endplate & vertebral osteolysis
  - Loss of disc space height
  - Increased bone density ± intervertebral fusion with successful treatment
  - Total vertebral destruction/collapse with treatment delay/failure

MR Findings

- Abnormal disc space
  - Loss of normal disc height, signal, & morphology
  - T1 hypointense, variable T2 signal intensity
  - Diffuse or rim enhancement with contrast
- Vertebral marrow signal abnormality abutting disc
  - T1 hypointense
  - Poorly defined increased fluid signal, most conspicuous on fat-saturated (FS) T2 or STIR images
  - Typically enhances with contrast unless necrotic
- Paraspinal & epidural phlegmon or abscess
  - Thickened T2 hyperintense tissues
  - Poorly defined fluid signal & stranding of fat
  - Diffuse enhancement vs. rim enhancing collections
- Follow-up: No single MR finding predicts clinical status
  - Abnormal imaging persists for months, does not necessarily equate residual/recurrent infection

Nuclear Medicine Findings

- Bone scan
  - Increased uptake can localize nonspecific signs & symptoms

Imaging Recommendations

- Best imaging tool
  - MR provides excellent diagnostic detail
  - NM bone scan can localize nonspecific signs & symptoms
- Protocol advice
  - STIR or FS T2 MR for marrow & soft tissue involvement
  - FS T1 C+ MR to evaluate for drainable paraspinal & epidural fluid collections

DIFFERENTIAL DIAGNOSIS

Langerhans Cell Histiocytosis

- Enhancing marrow lesion ± lytic destruction, enhancing soft tissue mass; disc sparing
- Single level vs. noncontiguous multifocal involvement
- Collapse of vertebra → vertebra plana

Chronic Recurrent Multifocal Osteomyelitis

- Autoimmune inflammatory disorder of bone
- Little disc involvement

Spinal Metastases

- Rare in children → neuroblastoma, leukemia
- Marrow abnormalities with disc space preservation

Degenerative Changes

- Rare in children
- Normal marrow ± vertebral endplate preservation

PATHOLOGY

General Features

- Etiology
  - Pathophysiology
    - Intervertebral disc & vertebral growth plates highly vascularized in young children
    - Hematogenous seeding of intervertebral disc/subchondral bone most common source
      - Lumbosacral (75%) > thoracic > cervical spine
  - Most common pathogen: *Staphylococcus aureus*

CLINICAL ISSUES

Presentation

- Clinical profile
  - Peak age range: 6 months to 4 years
  - Clinical symptoms variable & nonspecific in children
    - Difficulty walking, back/hip pain, fever (< 50%), irritability
  - Elevated ESR, CRP, WBC

Treatment

- Early empiric IV antibiotics, broad-spectrum coverage until causative pathogen isolated
- Organism-specific parenteral antibiotics for 6-8 weeks
- Spinal immobilization with bracing for 6-12 weeks
- CT-guided or open biopsy may be indicated if blood cultures negative & conservative treatment fails
- Surgery rarely indicated except in advanced infection

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Disc-centered process: Abnormal morphology, signal, & enhancement of disc & adjacent marrow is highly suggestive of discitis/osteomyelitis in children

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Guillain-Barré Syndrome

KEY FACTS

TERMINOLOGY

- Group of disorders characterized by acute onset of weakness & diminished reflexes secondary to inflammatory polyradiculopathy
  - Diagnosis primarily clinical (with supporting imaging findings)

IMAGING

- Smooth pial enhancement of cauda equina & conus medullaris
  - Ventral roots > dorsal roots
  - May be slightly thickened but not nodular
- May see cranial nerve enhancement

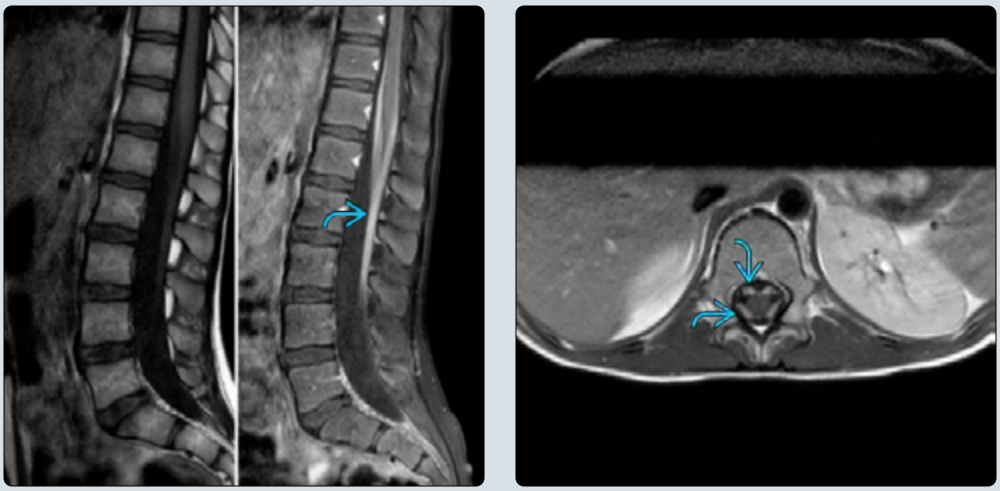
TOP DIFFERENTIAL DIAGNOSES

- Physiological nerve root enhancement
- Subarachnoid metastases
- Hereditary polyneuropathies
- Subacute & chronic demyelinating polyneuropathies

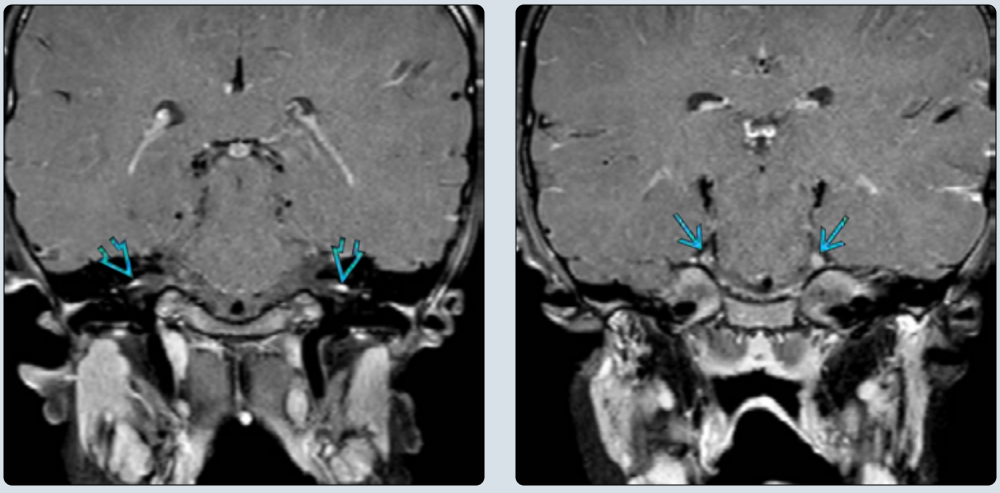
PATHOLOGY

- Inflammatory immune-mediated demyelination
  - Cross reactivity of antibodies to pathogen with specific gangliosides & glycolipids
- Antecedent event or "trigger" in 70% of Guillain-Barré syndrome (GBS) cases
  - *Campylobacter jejuni* infection
  - CMV infection
  - Temporal link has been suggested with vaccines but no correlation has been shown
- Subtypes include
  - Acute inflammatory demyelinating polyradiculoneuropathy: Most common form in USA
  - Acute motor axonal neuropathy: Pure motor form
  - Miller-Fisher variant: Ophthalmoplegia, ataxia, areflexia, normal extremity strength, & CSF protein
    - 5% of all cases of GBS

(Left) *Sagittal T1 (left) & T1 C+ FS (right) MR images show marked enhancement of the cauda equina nerve roots in this 10-year-old patient with Guillain-Barré syndrome (GBS).* **(Right)** *Axial T1 C+ MR in the same patient shows a striking degree of enhancement of the nerves relative to the conus medullaris. Some nerve root enhancement can be normal, but this degree is not physiologic.*



(Left) *Coronal T1 C+ FS MR through the internal auditory canals in a 3-year-old patient with the Miller-Fisher variant of GBS shows abnormal enhancement of the facial nerve on each side.* **(Right)** *Coronal T1 C+ FS MR more anteriorly in the same patient shows bilateral & symmetric abnormal enhancement of the trigeminal nerves.*



TERMINOLOGY

Abbreviations

- Guillain-Barré syndrome (GBS)

Synonyms

- Acute inflammatory demyelinating polyradiculoneuropathy (AIDP)

Definitions

- Group of disorders characterized by acute development of weakness & diminished reflexes secondary to inflammatory polyradiculopathy
- Primarily clinical diagnosis, with supporting imaging findings

IMAGING

MR Findings

- T1WI C+
  - Avid enhancement of cauda equina: Ventral roots > dorsal roots; may be slightly thickened (but not nodular)
  - Pial surface of distal cord & conus enhances variably; conus not enlarged
  - Cranial nerve enhancement in Miller-Fisher variant

DIFFERENTIAL DIAGNOSIS

Physiological Nerve Root Enhancement

- Much more subtle enhancement of normal roots

Subarachnoid Metastases

- Typically more nodular

Hereditary Polyneuropathies

- Charcot-Marie-Tooth, Dejerine-Sottas

Subacute or Chronic Demyelinating Polyneuropathies

- Clinical syndromes of inflammatory demyelinating polyradiculoneuropathy that have slower onset & longer duration than GBS

Acute Transverse Myelitis

- Cranial nerves always spared

Chemical or Postsurgical Arachnoiditis

- Hemorrhage-induced arachnoidal inflammation

Bacterial or Granulomatous Meningitis

- Acute onset

PATHOLOGY

General Features

- Etiology
  - Inflammatory immune-mediated demyelination; cross reactivity of antibodies to pathogen with specific gangliosides & glycolipids
  - Antecedent event or "trigger" in 70% of GBS cases
    - *Campylobacter jejuni* infection: 1/3 to 2/3 of cases
    - CMV infection: Up to 15% of cases
    - Multiple other infectious agents have been associated: EBV, HIV, *Mycoplasma*, varicella-zoster virus, Zika
  - Temporal link has been suggested with vaccines but no correlation has been shown

Staging, Grading, & Classification

- AIDP: Most common form in USA
  - Monophasic, nonfebrile, with ascending weakness & hyporeflexia
- Acute motor axonal neuropathy
  - Pure motor form; 1/3 may be hyperreflexic
- Acute motor sensory axonal neuropathy
  - Sensory root involvement; muscle wasting common
- Miller-Fisher variant
  - Cranial nerve involvement; Ophthalmoplegia, ataxia, areflexia, normal extremity strength, & CSF protein
  - 5% of all cases of GBS
- Acute panautonomic neuropathy
  - Affects sympathetic & parasympathetic nervous systems
- Pure sensory GBS
  - Sensory loss, ataxia, & areflexia
- Chronic polyneuropathies
  - Subacute inflammatory demyelinating polyradiculoneuropathy (SIDP)
    - Disease course between 4 weeks & 2 months
    - Bridge from AIDP to chronic inflammatory demyelinating polyradiculopathy (CIDP)
  - CIDP: Insidious onset over weeks or months; relapsing & remitting course

CLINICAL ISSUES

Presentation

- Acute flaccid paralysis or distal paraesthesias rapidly followed by ascending paralysis
  - Ascent up to brainstem may involve cranial nerves

Demographics

- Epidemiology
  - Most common cause of paralysis in Western countries
  - Incidence: 1.2-3 cases per 100,000 people per year
  - Affects all ages, races, socioeconomic statuses
    - Typically children, young adults

Natural History & Prognosis

- Most patients somewhat better by 2-3 months
  - 30-50% have persistent symptoms at 1 year
  - Permanent deficits in 5-10%
- Relapse in 2-10%

Treatment

- Medical management with plasma exchange or intravenous immunoglobulin (IVIg)
  - Severe GBS may benefit from plasma exchange after IVIg
- May require prolonged respiratory support in severe cases

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Transverse Myelitis

KEY FACTS

TERMINOLOGY

- Poorly understood inflammatory disorder of spinal cord resulting in bilateral motor, sensory, & autonomic dysfunction
- Requires exclusion of other definable causes of spinal cord inflammation: Neuromyelitis optica, MS, acute disseminated encephalomyelitis, Lyme disease

IMAGING

- Normal imaging in up to 40%
- Thoracic more common than cervical cord
- > 2 vertebral segments in length (often much longer)
- Smooth cord expansion with CSF effacement
- Bright cord signal on T2WI MR
  - Involves > 2/3 of cross-sectional area of cord
  - Centrally located with peripheral cord sparing
- No enhancement in up to 1/2 of cases
- Affected cord may atrophy over time

TOP DIFFERENTIAL DIAGNOSES

- Spinal cord astrocytoma
- Multiple sclerosis
- Neuromyelitis optica
- Spinal cord infarction

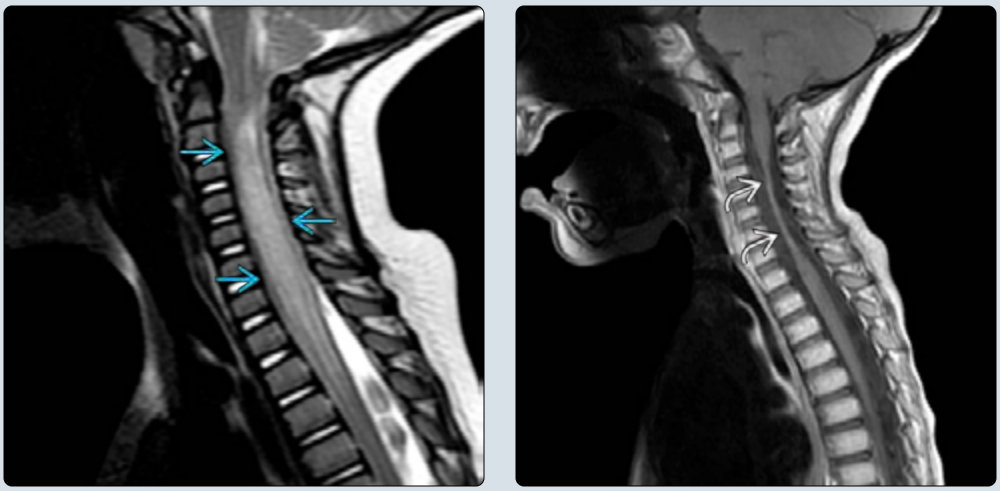
PATHOLOGY

- Clinical syndrome that can be idiopathic or associated with systemic disease or prodrome
- No etiology defined in up 1/3 of cases

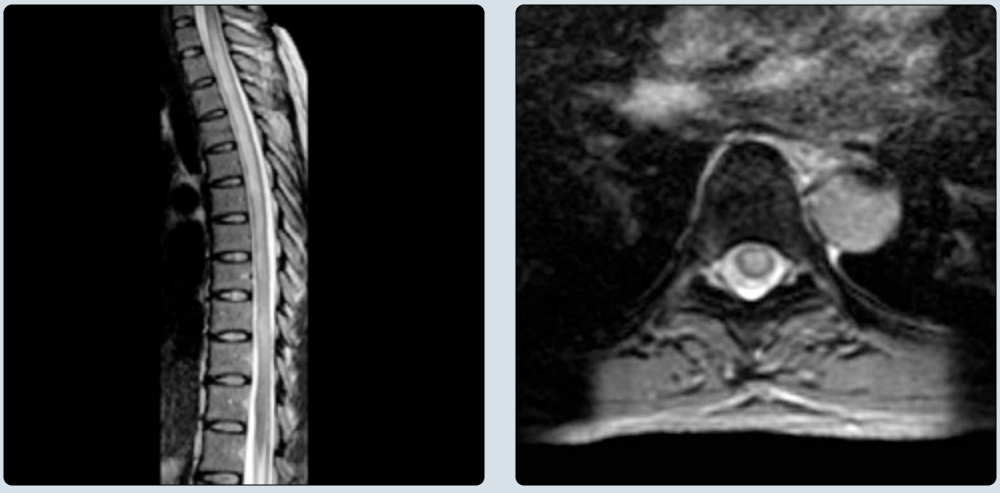
CLINICAL ISSUES

- 2 age peaks: 10-19 & 30-39 years old
- Typically monophasic
- Recovery variable: 1/3 good, 1/3 fair, 1/3 poor
- High-dose intravenous steroid pulse therapy
- Plasmapheresis for cases that fail to respond

(Left) *Sagittal T2 MR in a 2 year old who developed quadriparesis 6 days after receiving an influenza vaccine shows swelling & hyperintense signal of the cervical & upper thoracic cord with effacement of the subarachnoid CSF spaces at the cervical levels* ⇨. **(Right)** *Sagittal T1 C+ MR in the same patient 7 months after presentation shows marked atrophy of the cervical cord* ➡. *Up to 1/3 of patients with transverse myelitis will have persistent fixed neurological deficits, usually accompanied by imaging abnormalities.*



(Left) *Sagittal T2 MR shows a long segment of abnormal hyperintensity & expansion of the visualized spinal cord, characteristic of transverse myelitis.* **(Right)** *Axial T2* GRE MR in the same patient shows diffuse hyperintensity within the central cord. Note the peripheral sparing of the cord, a typical appearance of transverse myelitis.*



TERMINOLOGY

Definitions

- Transverse myelitis (TM): Inflammatory disorder involving both halves of spinal cord, resulting in bilateral motor, sensory, & autonomic dysfunction
- Must exclude other causes of spinal cord inflammation

IMAGING

General Features

- Location
  - Thoracic > cervical cord
  - Central cord location on axial imaging
- Size
  - > 2/3 of cross-sectional area of cord on axial imaging
  - > 2 vertebral segments in length
- Typical MR features
  - Normal imaging in up to 40%
  - Smooth cord expansion in up to 50%
  - Bright signal on T2WI & STIR MR
  - No enhancement in up to 1/2 of cases
  - ↓ fractional anisotropy (FA) centrally in lesions

DIFFERENTIAL DIAGNOSIS

Spinal Cord Astrocytoma

- Homogeneous cord expansion
- Often involves 3-4 vertebral segments, rarely holocord
- Slower clinical progression

Multiple Sclerosis

- Peripheral lesions ± central enhancement
- < 2 vertebral segments in length
- < 1/2 of cross-sectional area of cord
- 90% with associated intracranial lesions

Neuromyelitis Optica

- Fulminant optic neuritis + spinal cord lesions
  - Extensive (> 3 vertebral segments) T2 hyperintensity in cord + optic nerve enhancement
- CSF shows neuromyelitis optica (NMO)-IgG antibodies, specifically AQP4-specific antibodies

Spinal Cord Infarction

- Immediate onset

PATHOLOGY

General Features

- Etiology
  - Clinical syndrome that can be idiopathic or associated with systemic disease or prodrome
    - Previous infection or vaccination in some cases
    - Neoplastic, paraneoplastic, collagen vascular, & iatrogenic etiologies
  - Autoimmune phenomenon with formation of antigen-antibody complexes
  - Associated demyelinating process
  - No etiology defined in up to 1/3 of cases

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Sensory, motor, or autonomic dysfunction attributable to spinal cord
    - Bilateral signs & symptoms
    - Well-defined sensory level
    - Band-like dysesthesia
    - Loss of pain & temperature sensation
    - Paraplegia or quadriplegia
    - Bladder & bowel dysfunction
- Other signs/symptoms
  - Hypotonia & hyporeflexia initially
  - Spasticity & hyperreflexia over time

Demographics

- Age
  - 2 age peaks: 10-19 & 30-39 years old

Natural History & Prognosis

- 1/3 of patients experience good to complete recovery
  - Symptomatic improvement 2-12 weeks after onset
  - Children with slightly better prognosis than adults
- 1/3 fair recovery
  - Residual spasticity & urinary dysfunction
- 1/3 poor recovery
  - Persistent complete deficits
- Typically monophasic
  - Recurrence rate reported between 24-40%
  - If recurrent, must consider
    - Multiple sclerosis (MS): Progression to MS in 2-8% of cases of TM
    - SLE, antiphospholipid syndrome

Treatment

- High-dose intravenous steroid pulse therapy
- Plasmapheresis for refractory cases

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- TM is diagnosis of exclusion
  - Consider all differential possibilities before settling on diagnosis
  - Exclude intracranial lesions associated with MS, acute disseminated encephalomyelitis, or NMO

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KEY FACTS

TERMINOLOGY

- Traumatic injury to upper cervical region: Occiput to C2

IMAGING

- Normal measurements in adults may not translate to children due to dynamic skeletal changes in early life
- Radiography: Initial screening test of choice
  - > 20% of cervical fractures missed by radiography alone
  - 10% of craniocervical junction (CCJ) fractures shown by CT, not radiography
- CT: Asymmetry & ↑ joint space measurements
- CT: Look for retroclival hematoma & perimedullary hemorrhage
- MR: T2WI for ligament integrity & cord edema
- MR: STIR for bone marrow & soft tissue edema
- MR: SWI/T2* GRE for cord hemorrhage
- MRA: Vertebral artery injury may be associated with CCJ injuries; often clinically occult

TOP DIFFERENTIAL DIAGNOSES

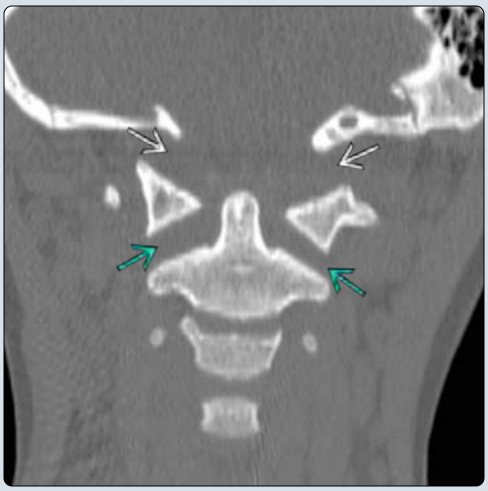
- Pseudosubluxation
- Os odontoideum
- Ligament instability
- Congenital fusion & segmentation anomalies

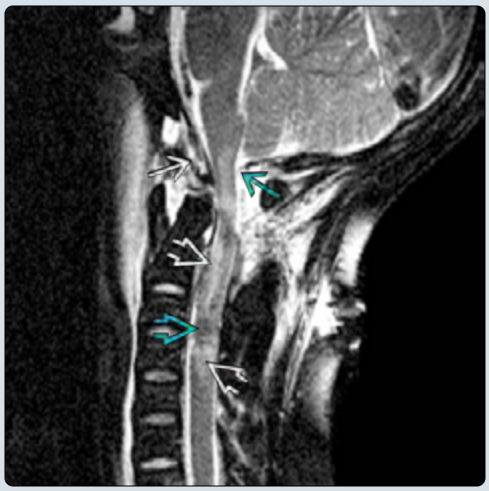
CLINICAL ISSUES

- Motor vehicle accidents, sports-related injuries, & falls
  - Higher mortality rate than adults
- Injury at CCJ most common in 1st decade
  - Disproportionate head size, ligamentous laxity, & immature muscles may contribute
- Neurological status at presentation predicts outcome

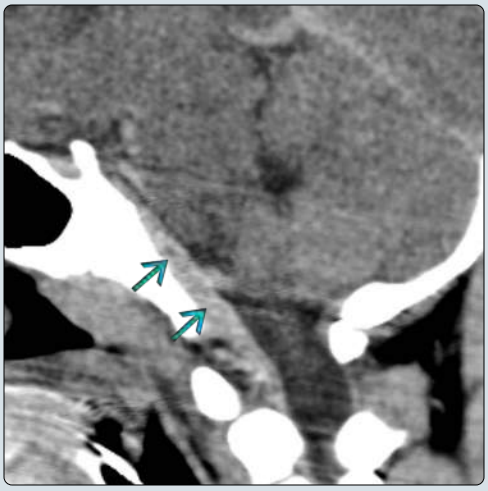
DIAGNOSTIC CHECKLIST

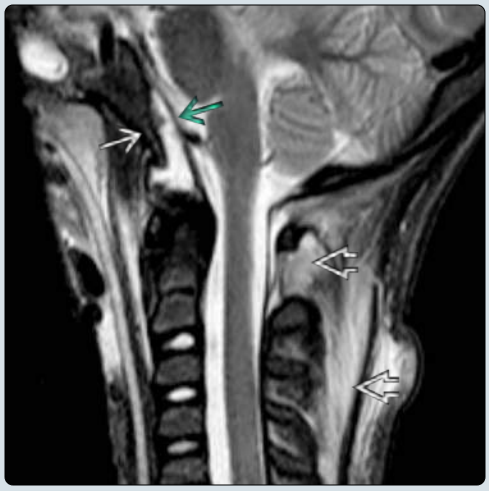
- Include C1-C2 on all head trauma CT studies
- MR in patients with normal radiographs/CT who continue to have neurologic symptoms
- STIR & MRA on all cervical spine trauma MR studies





(Left) *Coronal NECT in 9-year-old girl involved in high-speed MVA shows increase of the condyle-C1 interval (CCI) as well as the LMI, consistent with severe atlantooccipital dissocation (AOD) & atlantoaxial dissociation.* **(Right)** *Midline sagittal STIR MR in the same patient with AOD shows likely disruption of the tectorial membrane & complete transection of the posterior atlantooccipital ligament. The spinal cord shows areas of hyperintense signal (likely contusion) as well as a few foci of low signal (possibly hemorrhage).*





(Left) *Sagittal NECT in a 7-year-old girl involved in a MVA shows a hyperintense collection (hemorrhage) between the clivus & the tectorial membrane. A retroclival hematoma indicates AOD until proven otherwise.* **(Right)** *Sagittal STIR MR in the same patient shows that the linear hypointense tectorial membrane has been lifted off the clivus. Also note the extensive abnormal signal in the posterior neck soft tissues, suggestive of a severe flexion-related injury. The spinal cord appears normal.*

TERMINOLOGY

Abbreviations

- Craniocervical junction (CCJ) injuries

Definitions

- Traumatic injury to upper cervical region: Occiput to C2
  - Much more common in 1st decade than in older children & adults

Types of CCJ Injuries

- Atlantooccipital dissociation (AOD)
- Atlantoaxial dissociation
- Jefferson fracture (C1 ring)
- Odontoid (C2) fracture
- Hangman (C2) fracture
- Atlantoaxial rotatory fixation
- Spinal cord injury without radiographic abnormality

IMAGING

General Features

- Best diagnostic clue
  - Abnormal alignment, joint space enlargement, abnormal fluid collection, or obvious fracture involving occiput to C2
- Size
  - Numerous normal measurements have been proposed as signs of CCJ injury
  - **Key point**: Most measurements have been shown to be helpful in severe injuries but insensitive for mild to moderate injuries
  - **Key point**: Normal measurements in children change significantly with age & normal osseus development
  - **Occipital condyle-C1 interval (CCI) > 4 mm → AOD**
    - Found to be superior (most sensitive & specific) to all other measures for diagnosis of AOD but still insensitive to mild/moderate injury
    - Requires averaging multiple CCI measurements
    - Normal CCI varies with age; largest (> 2 mm) at 2-4 years, smallest (< 1 mm) after late teenage years
  - **Power ratio > 1.0 → AOD**
    - Power ratio calculated as BC/OA
    - BC: Basion to posterior C1 arch
    - OA: Opisthion to anterior C1 arch
    - Difficult to calculate in very young due to incomplete C1 ring ossification
  - **Basion-dens interval (BDI) > 1.2 cm or < 0 → AOD**
    - ↓ with age as dens ossifies
    - May cause false-positive in very young
  - **Interspinous ratio > 2.5 → AOD**
    - Interspinous ratio = C1-C2/C2-C3
  - **Lateral atlantodental space > 5-mm asymmetry → C1 ring fracture**
    - Some asymmetry is normal with head rotation/angulation
  - **Atlantodental interval > 6 mm → atlantoaxial dislocation**
    - Mean: ~ 2 mm in children
  - **Lateral mass interval (LMI) > 5 mm → atlantoaxial dislocation**
    - ↓ with age (mean: ~ 3 mm in children, ~ 1 mm in adults)

Radiographic Findings

- Prevertebral soft tissue thickening often seen in severe CCJ injuries but often absent in mild to moderate injuries
- AOD
  - Severe: ↑ CCI, power ratio > 1, abnormal BDI, ↑ interspinous ratio
  - Mild to moderate: Typically normal radiographs
- Atlantoaxial dislocation
  - Severe: ↑ joint space; ± joint space asymmetry on frontal
  - Mild to moderate: Typically normal radiographs
- Jefferson (C1 ring) fracture
  - Lateral displacement of C1 lateral mass in relation to C2 lateral mass on AP/open mouth views
    - Highly specific sign, especially when bilateral
  - Lateral atlantodental space > 5-mm asymmetry
    - Usually due to positioning/rotation rather than fracture
    - Often leads to false-positive radiograph interpretation
- Odontoid (C2) fracture
  - Type I, II, III based upon level of fracture
  - Typically occur at C2 synchondroses, especially in younger children
  - When severe will lead to abnormal alignment at CCJ
- Hangman (C2) fracture
  - Fracture of pars interarticularis on lateral radiographs
  - ± anterolisthesis of C2 on C3
- Rotation injuries & torticollis
  - Rotation of mandible over upper cervical spine on lateral view

CT Findings

- AOD
  - Abnormal CCJ measurements & asymmetry
  - Retroclival hematoma
  - Perimedullary hemorrhage
- Atlantoaxial dislocation
  - ↑ LMI ± asymmetric LMI
- C1 & C2 fractures clearly delineated
  - > 20% of cervical fractures missed by radiography alone
  - 10% of CCJ fractures are shown by CT & not radiography
  - Sagittal & coronal reformats essential
- Rotation injuries & torticollis
  - 3D reconstruction ideal for rotational injuries
  - Dynamic CT for fixed atlantoaxial rotatory subluxation
    - Axial images through C1-C2 in neutral position & with maximal voluntary rotation in each direction
    - Measure degree of rotation of C1 relative to C2
    - If no significant change in C1/C2 relationship with rotation → fixed rotatory subluxation

MR Findings

- T1WI
  - Helpful in identifying blood products
  - Fat saturation to differentiate blood from fat in soft tissues
- T2WI
  - Best sequence for cord edema
    - Axial & sagittal to confirm subtle cases

- Best conventional sequence for ligament integrity
- STIR
  - Best sequence for bone marrow & soft tissue edema
- CISS (constructive interference in steady state)
  - High spatial resolution with near complete CSF flow artifact suppression
  - Ideal for detection of intraspinal nerve root avulsion
  - Ideal for ligamentous injury at CCJ
- DWI
  - ADC values correlate with severity of spinal cord injury & can predict cavity formation
- SWI/T2* GRE
  - Detects blood products in cord
- MRA
  - Key adjunct in cervical spine injuries
  - Vertebral artery injury in up to 25% of adults with cervical spine trauma
    - May be clinically occult

Imaging Recommendations

- Best imaging tool
  - Imaging algorithm based upon ability to clinically clear C-spine
  - Radiographs: Almost universal initial screening test
  - NECT with reformats: Further evaluation of abnormal radiographs or persistent clinical symptoms in face of normal radiographs
  - MR + MRA: Optimal evaluation of ligamentous, spinal cord, & vascular injury
- Protocol advice
  - Include C1-C2 on all head trauma CT studies
  - Perform MR when clinical symptoms or subtle CT clues present (even when CCJ measurements normal on CT)

DIFFERENTIAL DIAGNOSIS

Pseudosubluxation

- Normal anterior translation of C2 relative to C3 in children < 5 years of age
- Only significant if posterior ring of C2 translates anterior to line from posterior ring of C1 to posterior ring of C3 (spinolaminar line)

Os Odontoideum

- Result of aberrant embryogenesis vs. fracture through odontoid synchondrosis prior to fusion (5-6 years)
  - Can be source of instability
  - More common in children with skeletal dysplasias
- Ossiculum terminale → nonfusion of distal ossification center
  - Smaller & without clinical significance

Ligament Instability

- Abnormal laxity of ligaments stabilizing C1-C2
- Grisel syndrome → atlantoaxial instability associated with inflammation in adjacent soft tissues
- Down syndrome → laxity of transverse ligament
  - 10-20% incidence

Congenital Fusion & Segmentation Anomalies

- C1-occiput, C1-C2
- Klippel-Feil spectrum

PATHOLOGY

General Features

- Etiology
  - Motor vehicle accidents (MVA) most common in young children
  - Sports-related injuries predominate in 2nd decade of life

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - AOD
    - Many die prior to reaching medical care
    - > 1/2 in cardiorespiratory arrest
    - 25-30% neurologically intact at presentation
  - Jefferson (C1) fracture
    - Axial loading injury (e.g., diving into shallow pool)
  - Odontoid & hangman fractures
    - Flexion or extension with distraction (usually MVA)
  - Rotational injuries
    - Present with painful torticollis

Demographics

- Age
  - 0-9 years: High incidence of upper cervical spine injury compared to adolescents & adults
    - Disproportionate head size, ligamentous laxity, & immature muscles have all been postulated as causes
- Gender
  - M > F in adolescents & teens
- Epidemiology
  - 5% of spinal injuries occur in children
  - 18-21:1 million population

Natural History & Prognosis

- Neurological status at presentation best predictor of outcome
  - Intact or incomplete injury → likely good outcome
  - Complete loss of function at level of injury → poor chance of recovery
- Spinal cord hemorrhage → worse potential for recovery

Treatment

- Support of impaired systemic function
- Stabilization of spine to prevent further injury
- Internal or external surgical stabilization
  - May impair ability to assess with MR
- Emergent decompression of spinal cord impingement
- Administration of steroids for acute spinal cord injury no longer recommended

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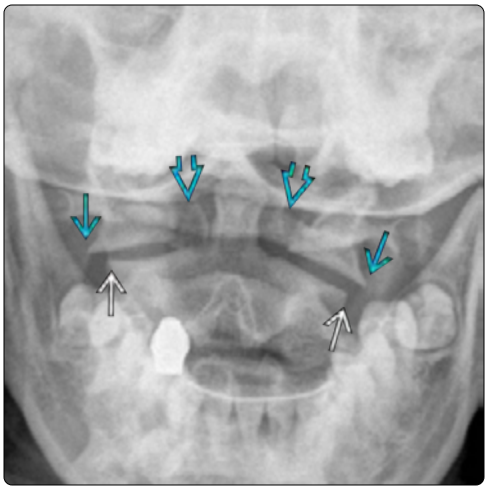
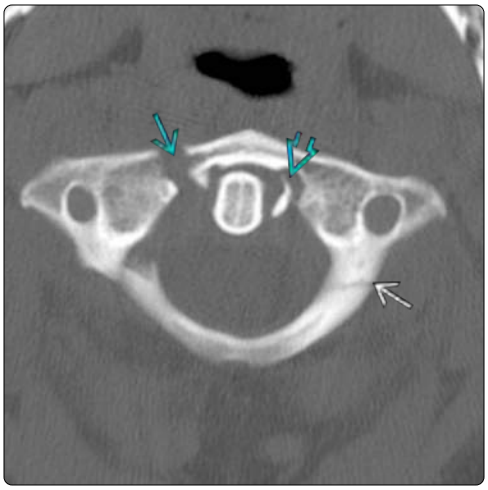
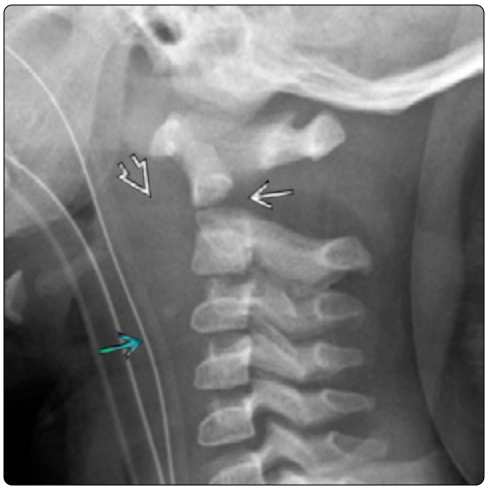
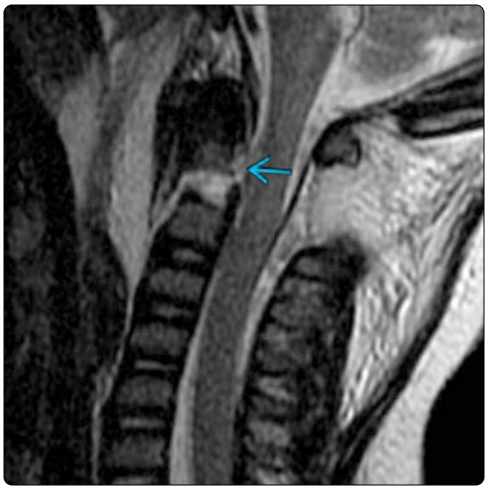


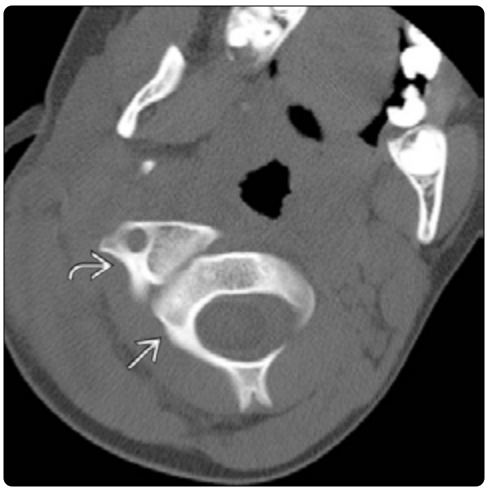


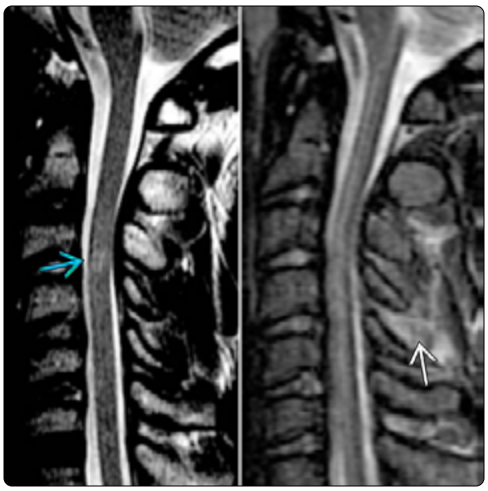
(Left) *Frontal open mouth radiograph of a 17-year-old girl who fell while performing a back flip shows bilateral step-off of the C1 lateral masses ⇨ in relation to the C2 lateral masses ➡ as well as widening of the lateral atlantodental spaces ⇨, consistent with a Jefferson fracture.* **(Right)** *Axial NECT in the same patient shows a displaced fracture of the right anterior arch of C1 ⇨, a nondisplaced fracture of the left posterior arch of C1 ➡, & a posterior cortical avulsion fracture of the left anterior arch of C1 ⇨.*





(Left) *Lateral radiograph in a 2-year-old girl in an MVA shows a displaced fracture ➡ of the odontoid at the synchondrosis, separating the odontoid & C2 body. Also note the extensive prevertebral soft tissue swelling ➡ & displacement of the nasoenteric tube ⇨.* **(Right)** *Midline sagittal T2 MR in the same patient shows disruption of the posterior longitudinal ligament ⇨, a commonly associated ligamentous injury in the setting of an odontoid fracture.*





(Left) *Axial NECT in a child with painful torticollis shows an anterior dislocation of the C1 right lateral mass ➡ relative to the C2 lateral mass ➡ (atlantoaxial rotational dislocation).* **(Right)** *Sagittal T2 (left) & STIR (right) MRs in a 15-year-old football player with motor & sensory deficits in both arms (but normal cervical spine radiographs) show increased signal ⇨ at the level of C3-C4, consistent with cord contusion. There is also interspinous soft tissue/ligamentous edema ➡. The presentation & imaging are typical for SCIWORA.*

Chance Fracture

KEY FACTS

TERMINOLOGY

- Synonyms: Flexion-distraction injury, seat belt fracture
- Definition: Compression injury of anterior column of spine with distraction of middle & posterior columns
  - Anterior column: Anterior longitudinal ligament, anterior 1/2 of vertebral body, anterior annulus fibrosis
  - Middle column: Posterior longitudinal ligament, posterior 1/2 of vertebral body, posterior annulus fibrosis
  - Posterior column: Posterior neural arch, facet joint capsular ligaments, ligamentum flavum, inter- & supraspinous ligaments
- May be completely osseous, ligamentous (rare "Chance equivalent"), or mixed (most frequent)
- Extension across 3 columns may invoke osseous & soft tissue structures at multiple adjacent levels

IMAGING

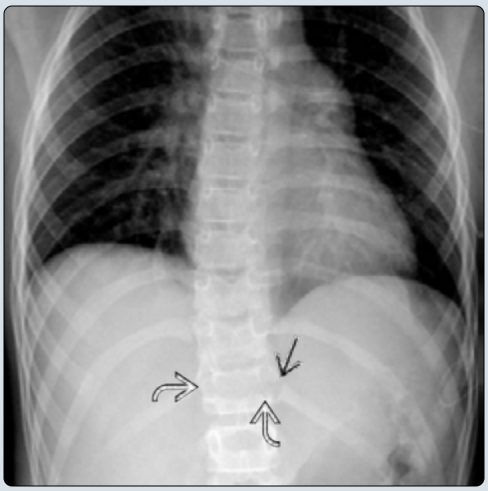
- 78% occur between T12-L2
- Wedging of anterior vertebral body
- Posterior bony & ligamentous distraction injuries result in
  - Focal kyphosis
  - Wide "splitting" of pedicles & laminae on AP view
  - Separation of facet joints with ↑ interspinous distance
  - Artifactual ↑ in radiolucency of vertebral body on AP radiograph secondary to splayed spinous processes
- Vertebral marrow edema & posterior ligamentous disruptions best seen on STIR or T2 FS MR

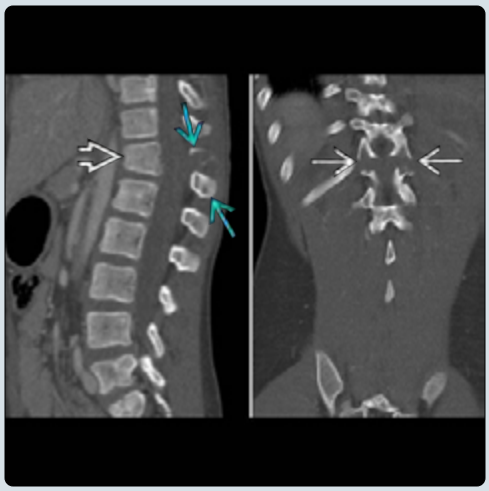
CLINICAL ISSUES

- Classic history: MVC with lap belt restraint & cutaneous seat belt sign over abdomen
- Up to 80% have significant abdominal injuries
  - Bowel/mesentery (most common), aorta

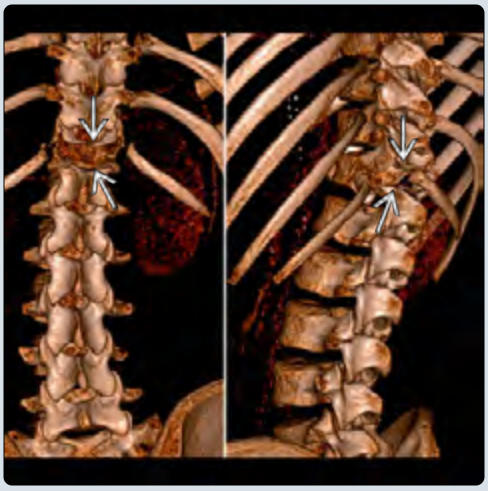
DIAGNOSTIC CHECKLIST

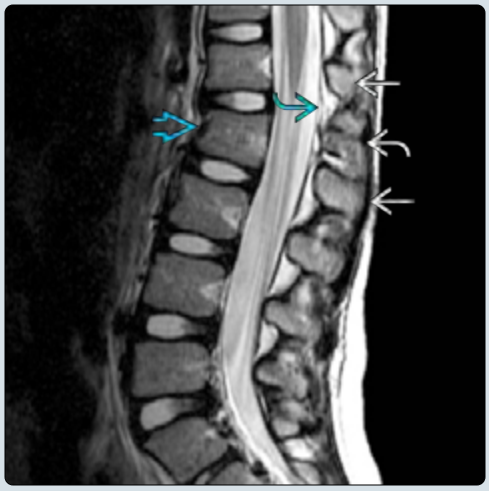
- Crucial to create sagittal & coronal bone reformatted images from abdominal CT exams in trauma patients





(Left) *AP chest radiograph of a teenager injured in a motor vehicle collision while wearing a seat belt shows a horizontal lucency splitting the T12 pedicles with widening of the costo-vertebral joints, indicative of a Chance fracture.* **(Right)** *Sagittal (left) & coronal (right) bone windowed CECT in the same patient show anterior wedging of the T12 vertebral body with a horizontal fracture plane splitting the posterior elements. Note the splaying/distraction of the T12 spinous process fragments.*





(Left) *3D posterior oblique CT in the same patient clearly show the horizontal splitting & distraction of the T12 posterior elements, characteristic of the posterior column injury in a Chance fracture.* **(Right)** *Sagittal T2 MR in a 7 year old with a primarily ligamentous injury shows mild anterior wedging of the L2 vertebra with disruption of the ligamentum flavum & interspinous ligaments resulting in splaying of the L1 & L2 posterior elements.*

TERMINOLOGY

Synonyms

- Flexion-distraction injury, seat belt fracture

Definitions

- Compression injury of anterior column of spine with distraction of middle & posterior columns
  - Anterior column: Anterior longitudinal ligament (ALL), anterior 1/2 of vertebral body, anterior annulus fibrosis
  - Middle column: Posterior longitudinal ligament, posterior 1/2 of vertebral body, posterior annulus fibrosis
  - Posterior column: Posterior neural arch, facet joint capsular ligaments, ligamentum flavum, inter- & supraspinous ligaments

IMAGING

General Features

- Location
  - Usually occurs at T11-L3; 78% occur between T12-L2
  - May have anterior injury at 1 level with posterior injury (fracture &/or soft tissue disruption) at adjacent level

Radiographic Findings

- Wedging of anterior vertebral body with focal kyphosis
  - ± sclerotic transverse fracture line due to trabecular impaction
  - Vertebral body height loss often > 40-50%
- Bony & ligamentous injuries to posterior column
  - "Splitting" of pedicles & laminae on AP view
  - Empty body sign on AP view: Artifactual ↑ in radiolucency of vertebral body secondary to splayed spinous processes
  - Separation of facet joints with ↑ interspinous distance
- No subluxation of vertebral body

CT Findings

- Vertebral body fracture often comminuted
  - May have mild retropulsion of posterior cortex
- Transversely oriented posterior element fracture &/or posterior ligamentous injury
  - ↑ interspinous distance
  - Separation of facet joints: Naked facet sign

MR Findings

- Varying degrees of marrow edema (poorly defined ↑ T2/STIR signal intensity) surround fracture plane
  - May be seen in multiple adjacent vertebral bodies with radiographically occult fractures/contusions
- Low signal ALL not discontinuous
  - May be partially stripped from vertebral attachment
- Posterior element fractures difficult to see if splaying absent
- Low signal interspinous & supraspinous ligaments may be disrupted by high fluid signal
- Poorly defined fluid in surrounding soft tissues

Imaging Recommendations

- Best imaging tool
  - CT scan
    - Data typically acquired as part of abdominal CECT
  - MR indicated to investigate neurologic deficit, ligamentous injuries
- Protocol advice
  - Coronal/sagittal reformations essential for CT
    - Define fracture planes, bony relationships
  - STIR &/or T2 FS crucial on MR
    - Detect marrow & soft tissue edema

DIFFERENTIAL DIAGNOSIS

Distraction Injury

- Vertebral body displacement & ALL disruption distinguish from Chance fracture

Traumatic Compression Fracture

- No posterior element fracture

Burst Fracture

- Also involves posterior vertebral body cortex
- Vertically oriented fractures of posterior elements

PATHOLOGY

General Features

- Etiology
  - Typical setting: Motor vehicle collision or fall
  - Classic pattern: Lap seat belt without shoulder strap
    - Seat belt, in high position, serves as fulcrum anterior to vertebral column → compression of anterior column + distraction of posterior column
    - Up to 40% have 3-point restraint (not just lap belt)
- Associated abnormalities
  - Up to 80% have significant abdominal injuries
    - Bowel/mesentery (most common)
      - Look for unexplained mesenteric & peritoneal fluid with focal bowel wall thickening/dilation (± free air)
    - Aorta > solid organs

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Radiographic recognition essential → prompts further evaluation for abdominal injury
- Search for spine injury on sagittal & coronal reformats from abdominal CT exams in trauma patients
  - Especially with clinical evidence of lap belt injury
- Chance fracture may have retropulsion of posterior vertebral body cortex mimicking burst fracture

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KEY FACTS

TERMINOLOGY

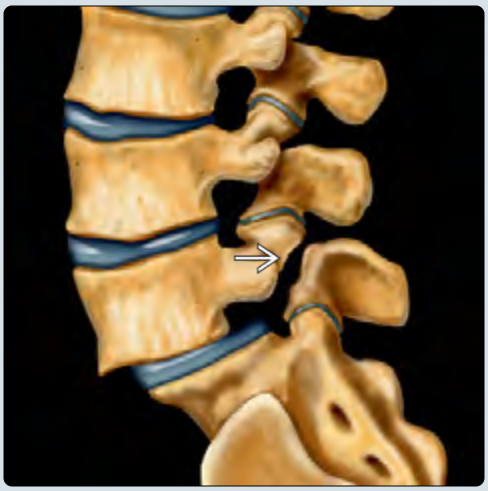
- Spondylolysis: Defect/break in pars interarticularis
  - Repetitive microtrauma likely etiology
- Spondylolisthesis: Spondylolysis + anterior slippage of vertebra in relation to vertebra below

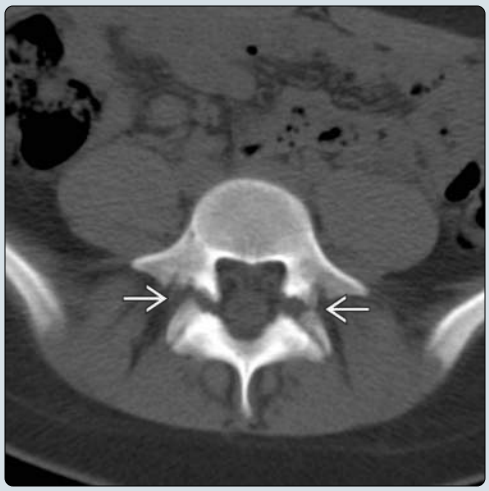
IMAGING

- Bone CT
  - Linear lucency or defect in pars interarticularis
    - Sagittal or oblique sagittal reformatted imaging vital in assessment
  - Incomplete ring sign on axial imaging ± distraction
    - May simulate "extra" or double facet joints
    - May be confused with facet joint
      - Lysis located anterior to facet joint
      - Lysis more transverse in orientation on axial images compared to facet joint, which is more oblique
      - Facet joint is smooth, while lysis typically is irregular/fragmented
  - Spondylolisthesis & foraminal narrowing on sagittal reformatted images
  - Secondary finding of sclerosis &/or hypertrophy of contralateral pedicle & lamina
  - May be insensitive to early stress injury (edema with microtrabecular fracture)
- SPECT bone scan imaging helpful for diagnosis
  - Intense focal uptake in posterior elements, unilateral or bilateral
  - Triangular pattern of uptake on sagittal images
  - Remote or healed may be occult (normal)
- SPECT/CT: Confirms diagnosis with anatomy & physiology
- MR for lysis: Adjust spine protocol towards musculoskeletal system by adding STIR/T2 FS (↑ conspicuity of marrow & soft tissue edema)

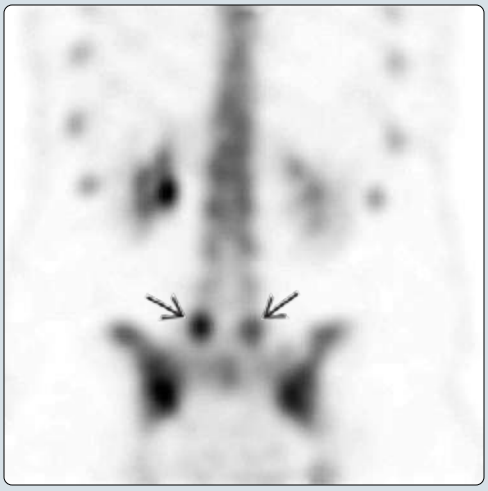
CLINICAL ISSUES

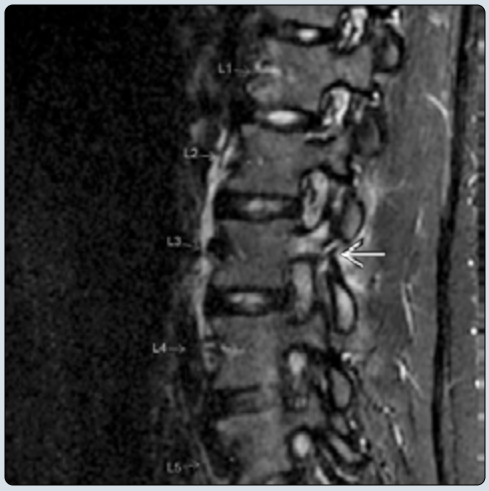
- 40% incidence in children with lower back pain (LBP)
  - L5: 85%; L4: 5-15%





(Left) *Lateral graphic shows a separated defect within the pars interarticularis (spondylolysis) at L5. Notice the resultant anterior slippage (spondylolisthesis) of L5 on S1.* **(Right)** *Axial NECT in a 14-year-old girl who presents with severe left flank pain for renal stone evaluation shows an incomplete ring with bilateral distracted pars interarticularis defects (spondylolysis) at L5.*





(Left) *Coronal SPECT bone scan in a 15 year old who presents with lower back pain shows asymmetric uptake in the posterior elements of L5, right > left. NECT images (not shown) demonstrated right L5 spondylolysis with reactive stress changes at the left L5 pars interarticularis.* **(Right)** *Sagittal STIR MR in 14 year old with back pain shows a pars defect with edema in the marrow of the posterior elements & adjacent soft tissues. STIR images can be helpful in detecting marrow edema associated with spondylolysis.*

TERMINOLOGY

Synonyms

- Lysis, isthmic spondylolysis

IMAGING

General Features

- Location
  - L5: 85%; L4: 5-15%
  - Cervical spine usually congenital
  - 10-15% unilateral

Radiographic Findings

- Classic: Break in neck of "Scotty dog" (pars interarticularis defect on oblique views of standing lumbar spine)
  - ± anterolisthesis
  - Positive predictive value of 57%

MR Findings

- T1WI: Discontinuity (sagittal images best), ↓ signal in pars interarticularis
- T2WI: ↓ signal in pars interarticularis (reactive sclerosis), ↑ signal in pars interarticularis (marrow edema)
- STIR/T2 FS: ↑ conspicuity of surrounding marrow & soft tissue edema, suggesting spondylolysis at classic location even if fracture not seen
- MR for lysis: Sensitivity: 57-86%; specificity: 81-82%

Nuclear Medicine Findings

- ↑ uptake best seen on SPECT bone scan

Imaging Recommendations

- Best imaging tool
  - If suspicious for this diagnosis
    - Targeted helical/volumetric bone CT with multiplanar reformats
    - Bone scan SPECT/CT better than SPECT alone
      - Anatomic & physiologic confirmation
  - If source of back pain not clear, MR may be more beneficial
    - Include STIR or T2 FS for marrow & soft tissue edema

DIFFERENTIAL DIAGNOSIS

Spectrum of Radiologic Findings in Back Pain

- Musculoskeletal
  - Normal (muscular)
  - Scoliosis, vertebral or sacral fracture, spinous process avulsion, Baastrup disease, facet degeneration, Bertolotti syndrome, degenerative disc disease, ring apophyseal injury, Scheuermann disease
- Infection
  - Discitis, osteomyelitis, sacroiliitis, paraspinal inflammation/abscess, pyelonephritis, pelvic inflammatory disease
- Tumor
  - Osteoid osteoma, osteoblastoma, Langerhans cell histiocytosis, leukemia, lymphoma, metastatic disease, neurofibroma
- Inflammatory
  - Ankylosing spondylitis, psoriatic arthritis, Reiter disease, inflammatory bowel disease

PATHOLOGY

General Features

- Etiology
  - Believed to be caused by repetitive microtrauma, resulting in stress fracture
  - Participation in gymnastics, weightlifting, wrestling, cricket, & football at young age
- Associated abnormalities
  - Spondylolisthesis (50%), scoliosis, Scheuermann disease, spina bifida occulta

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Asymptomatic (80%)
  - 40% incidence in children with lower back pain (LBP)
- Other signs/symptoms
  - Tight hamstring muscles
    - Waddling gait secondary to tight hamstring muscles
  - Back spasms or radiating pain
  - Back pain exacerbated by rigorous activities
  - Radiculopathy & cauda equina syndrome in lysis with high-grade spondylolisthesis

Demographics

- Gender
  - M:F = 2-4:1
- Epidemiology
  - Incidence: 3-6% of Caucasian population
  - Higher incidence in competitive athletes, especially males

Treatment

- Mainly conservative 1st; surgical if conservative treatment fails or subluxation progresses

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Sagittal or oblique sagittal bone CT reformats & sagittal STIR/T2 FS MR images are most important for diagnosis
- Identify complete ring at each lumbar level on axial bone CT or MR
- Typical bone scan findings with normal radiographs or bone CT suggests stress reaction or early spondylolysis

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SECTION 9

Head and Neck

Summary Thoughts: Congenital Lesions of Head & Neck

Neck masses are a common indication for imaging the pediatric head & neck (H&N). The majority of neck masses in children are either congenital or inflammatory in origin, with only 5% of childhood neoplasms occurring in the H&N. The most common extrathyroid, nonneoplastic solid neck masses in children are related to inflammatory disease & do not require imaging unless there is concern for deep neck infection or abscess. The most common cystic masses in the pediatric H&N are congenital lesions secondary to abnormal embryogenesis involving the thyroglossal duct (TGD), the branchial apparatus, or the vascular endothelium. **TGD cysts (TGDC)** account for 70-90% of all congenital neck abnormalities in children. The most common **branchial apparatus lesion** is the 2nd branchial cleft or apparatus cyst (BCC/BAC), & **lymphatic malformations** are the most common vascular malformations in the H&N.

Whenever a cystic neck mass in a child is encountered on an imaging study, a very reasonable differential diagnosis can be made based on location (midline, paramidline, or lateral, as well as location relative to carotid sheath), imaging appearance (simple cyst, complicated cyst, enhancement, ± solid component), & clinical presentation (present since birth or acute onset, ± clinical evidence of infection).

Terminology

TGDC or tract is an anomalous remnant of the TGD, which, during normal development, completely involutes. Cysts are located anywhere from the midline posterior tongue at the foramen cecum to the thyroid bed in the lower neck.

Branchial apparatus anomalies may be in the form of cysts, sinus tracts, or fistulae. **Cysts** are fluid filled with well-defined walls secondary to the failure of obliteration of a branchial cleft or pouch. **Sinus tracts** are congenital tracts with 1 opening, either externally to the skin surface (or external auditory canal) or internally to the pharynx (2nd branchial cleft), superolateral hypopharynx (3rd branchial cleft), or pyriform sinus (4th or 3rd branchial pouch). **Fistulae** are congenital tracts with 2 openings, 1 internally & 1 externally, secondary to failure of obliteration of the branchial cleft & pouch.

Imaging Techniques & Indications

US, CT, & MR are all reasonable options for initial imaging of neck masses in children. Modality choice depends on the clinical presentation, the referring clinical service, & the modalities available at the time of imaging. For instance, if there is concern that a child with cellulitis & cervical adenitis has a drainable abscess deep to a palpable neck mass, US may be very helpful in determining the presence of underlying suppurative adenitis or frank abscess. Likewise, if a child is thought to have a TGDC, US is the imaging modality of choice to evaluate the suspected TGDC & prove the presence of a normal-appearing thyroid gland in the lower neck. Furthermore, US is the imaging modality of choice in patients with suspected infantile hemangioma, as color Doppler findings can be quite characteristic. However, CT is the initial imaging modality of choice to evaluate the total extent of disease in children with suspected deep neck infection & to assess for a possible underlying pyriform sinus tract in children with left-sided neck abscesses that involve the left thyroid lobe.

In children who are not presenting with signs & symptoms of infection, CT or MR may be used as the initial imaging modality. The choice of which modality to use may depend on the need for sedation/anesthesia, which is usually required for MR in children under the age of 5 or 6 years. MR is the preferred imaging modality in children with suspected vascular malformations due to its excellent tissue contrast, variety of methods of interrogation, & lack of limitations regarding depth.

Embryology

The thyroid gland migrates from the foramen cecum at the midline posterior tongue to the paramidline location in the lower neck via the path of the **TGD**. The TGD normally involutes during the 5th or 6th week of gestation. However, remnant epithelium anywhere along the tract may persist & form a **TGDC**. As the TGD diverticulum descends caudally, it passes along the anterior surface of the developing hyoid bone; therefore, remnants may be found anterior to the preepiglottic space of the larynx.

Branchial apparatus structures develop between the 4th & 6th week of gestation & consist of 6 pairs of mesodermal arches separated by 5 paired endodermal pouches internally & 5 paired ectodermal clefts externally. During the 6th week of gestation, the 2nd branchial arch overgrows the 3rd & 4th branchial arches, resulting in a combined 2nd, 3rd, & 4th branchial cleft, termed the "cervical sinus of His." Anomalies of the branchial apparatus include **cysts, sinus tracts, & fistulae**. The most common lesions for which imaging is indicated are **BCC/BAC**, & most of these are related to anomalous development of a branchial cleft. However, a few are related to anomalous development of a branchial pouch.

The location of the lesion is the single most important determinant of the origin of a branchial apparatus anomaly. **First BCC** account for ~ 8% of all branchial anomalies & are located in or around the external auditory canal, ear lobe, or parotid gland & may extend inferiorly to the angle of the mandible. **Second BCC** account for up to 95% of all branchial apparatus anomalies & are subclassified by location using the Bailey system: Type 1 cysts are located deep to the platysma muscle/anterior to the sternocleidomastoid (SCM) muscle; type 2 cysts are posterior to the submandibular gland/anterior to the SCM (& are the most common); type 3 cysts protrude between the external & internal carotid arteries; & type 4 cysts are directly adjacent to the pharyngeal wall (thought to be a remnant of the 2nd pouch). Fistulas are rare; however, a 2nd branchial apparatus fistula is occasionally identified with an internal opening at the level of the pharynx & an external skin opening anterior to the lower aspect of the SCM.

Third BCC are rare, located in the posterior compartment of the upper neck or anterior compartment of the lower neck. Third pharyngeal pouch remnants are more common than cleft remnants & are related to descent of the thymic primordium from the lateral margins of the pharynx to the upper anterior mediastinum (via the thymopharyngeal duct) during the 6th to 9th weeks of gestation. If the duct does not undergo normal involution, a cervical thymic cyst may form that is in close association with the anterior margin of the carotid sheath. The cyst formation is from interactions between the endodermal primordia & neural crest cells during normal thymic development & migration. Histologically, Hassall corpuscles will be identifiable within the cyst wall.

Types of Congenital Head & Neck Lesions

Thyroglossal Duct Lesions
Thyroglossal duct cyst: Tongue base to thyroid bed; embedded in strap muscles when infrahyoid
Ectopic thyroid tissue: Lingual location most common
Branchial Apparatus Lesions
1st branchial cleft cyst (type 1): Located anterior, inferior, or posterior to external auditory canal
1st branchial cleft cyst (type 2): Located in or adjacent to parotid gland, may extend inferiorly to angle of mandible
2nd branchial cleft cyst: Most common location posterior to submandibular gland, lateral to carotid space, & anteromedial to SCM muscle
3rd branchial cleft cyst: Posterior cervical space upper neck, anterior to SCM lower neck
3rd branchial pouch remnant → thymopharyngeal duct cyst or ectopic thymus: Along course from pharynx to upper mediastinum
4th branchial pouch remnant → pyriform sinus tract: Patients present with left-sided abscess involving thyroid gland
Vascular Malformations
Venous malformation: Slow flow enhancing lesion, phleboliths common
Lymphatic malformation: Nonenhancing unilocular or multilocular ± fluid-fluid levels
Venolymphatic malformation: Mixed enhancing venous & nonenhancing lymphatic components

SCM = sternocleidomastoid.

Ectopic foci of solid thymic tissue may also be deposited along the remnant duct.

Pyriform sinus tracts, or rarely fistulae, represent a unique congenital anomaly of the 4th (or 3rd) pharyngeal pouch. This lesion should be suspected in any child presenting with neck infection involving the left thyroid lobe. The inflammation can frequently be traced superiorly to an asymmetric pyriform sinus apex. A postbarium swallow CT study is frequently helpful to define the barium-filled tract extending from the pyriform sinus to the anterior lower neck.

Vascular malformations are congenital malformations of endothelial development that can be divided into capillary, lymphatic, venous, venolymphatic, & arteriovenous malformations based on the predominant endothelial characteristics of the lesion. The most common vascular malformations identified in the H&N are lymphatic malformations, venous malformations, & combined venolymphatic lesions.

Imaging Anatomy

Recognizing the defined location of a congenital abnormality, particularly congenital cysts, is key to arriving at the correct diagnosis or differential diagnosis. **TGD** remnants may be in the form of cysts or solid ectopic thyroid tissue anywhere from the midline tongue base to infrahyoid paramidline thyroid bed. **First BCCs** are in or around the EAC or parotid gland. **Second BCCs** are most commonly posteromedial to the submandibular gland & anteromedial to the SCM. Cysts in the posterior triangle of the upper neck may be **3rd BCCs or lymphatic malformations**. Cysts along the lower anterior margin of the SCM may be from the 2nd or 3rd branchial cleft or may be lymphatic malformations. Cysts or solid masses in close association with the carotid sheath along the tract of the thymopharyngeal duct (from the angle of the mandible to the upper mediastinum) should raise the question of **cervical thymic cyst or ectopic thymus**.

Differential Diagnosis

TGD Lesions

- TGDC: Tongue base to thyroid bed
- Ectopic thyroid: Lingual thyroid most common

Branchial Apparatus Lesions

- 1st BCC: Type 1 cysts anterior, inferior, or posterior to external auditory canal
- 1st BCC: Type 2 cysts in superficial, parotid, or parapharyngeal space may extend as low as posterior submandibular space
- 2nd BCC: Most common posterolateral to submandibular gland, lateral to carotid space, & anteromedial to SCM
- 3rd BCC: Posterior cervical space of upper neck or along anterior border of SCM in mid & lower neck
- 3rd branchial pouch remnant: Thymopharyngeal duct cyst or ectopic thymus
- 4th branchial pouch remnant: Pyriform sinus tract → recurrent thyroiditis or thyroid abscess, usually left sided; recent literature suggests this may be 3rd branchial pouch remnant

Vascular Malformations

- Venous malformations: Slow flow lesions with moderate patchy & gradual postcontrast enhancement + phleboliths
- Lymphatic malformations: Unilocular or multilocular; single space or transspatial; nonenhancing multiseptated cystic mass + fluid-fluid levels
- Venolymphatic malformations: Nonenhancing lymphatic & enhancing venous elements ± phleboliths

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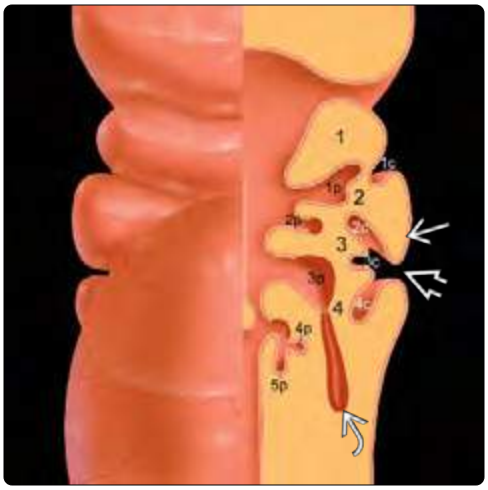
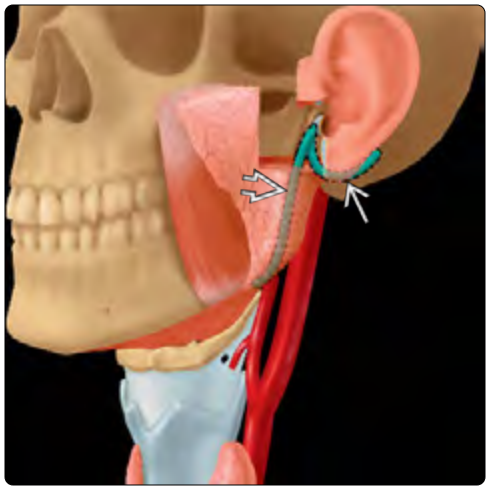
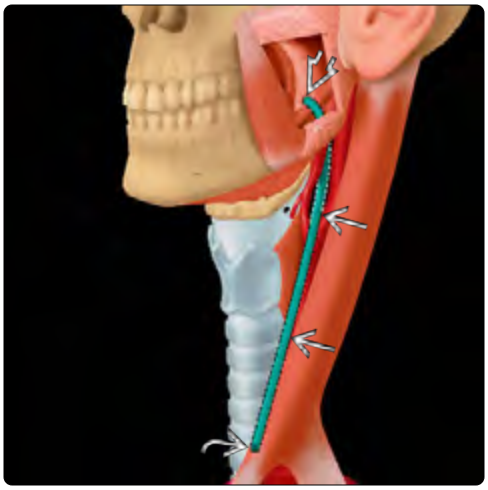
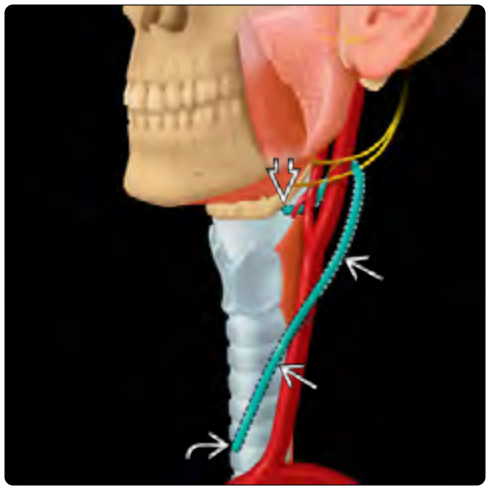
(Left) *Anterior graphic of a 6-week embryo shows the 2nd branchial arch ➡ growing inferiorly over the 3rd & 4th arches resulting in the cervical sinus of His ➡ that combines the 2nd, 3rd, & 4th branchial clefts (BC). Note that the thymopharyngeal duct is a remnant of the 3rd branchial pouch ➡.* **(Right)** *Oblique graphic shows the tract of a type I 1st BC anomaly ➡ from the medial bony external auditory canal (EAC) toward the retroauricular area. The tract of a type II 1st BC anomaly ➡ connects the EAC to the angle of the mandible.*



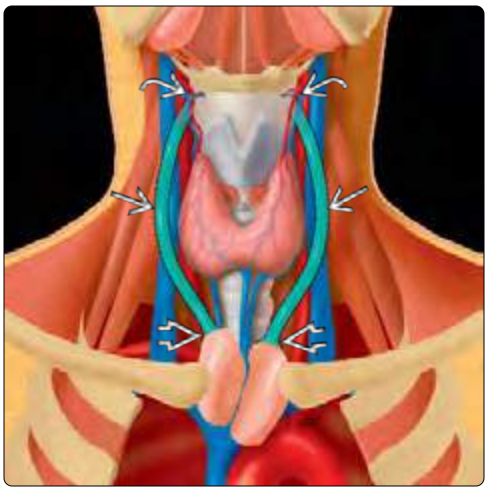


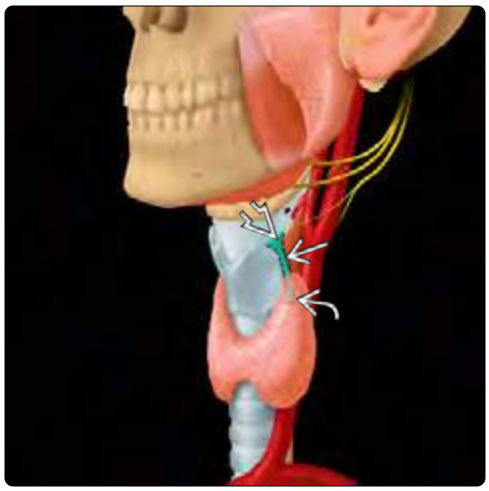
(Left) *Oblique graphic of the tract of a 2nd BC fistula ➡ shows a proximal opening ➡ in the faucial tonsil & a distal opening in the anterior supraclavicular neck ➡.* **(Right)** *Oblique graphic illustrates the tract of a 3rd BC anomaly ➡ extending from the cephalad aspect of the lateral hypopharynx ➡ to the supraclavicular anterior neck skin ➡.*

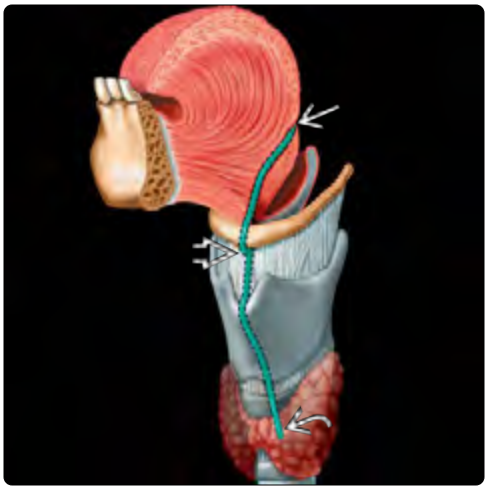


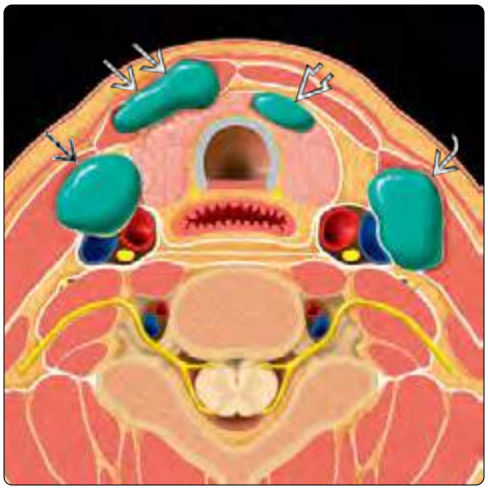


(Left) *Anterior graphic shows both thymopharyngeal duct tracts ➡ extending from the lateral hypopharyngeal area ➡ to the location of the normal lobes of the thymus ➡ in the superior mediastinum.* **(Right)** *Oblique graphic of the neck shows the tract of a 4th BC anomaly ➡ extending from the hypopharynx ➡ to the location of the left thyroid lobe ➡. This relationship explains why this lesion often presents with thyroiditis.*

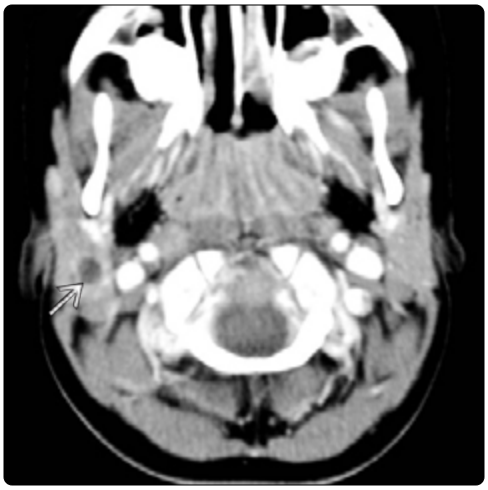


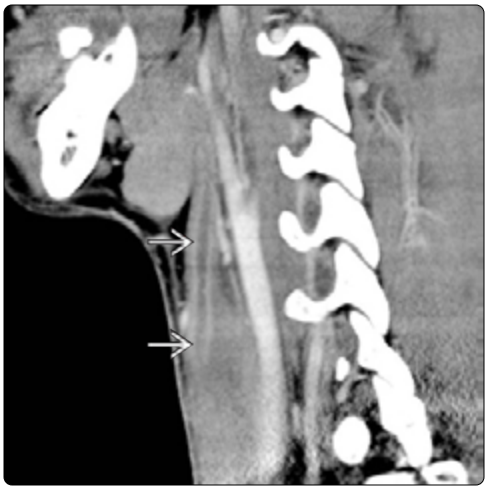




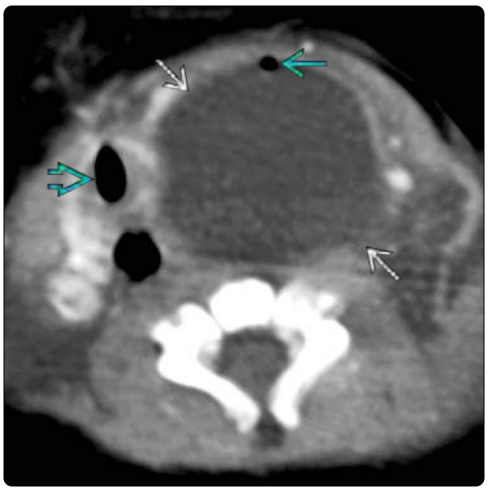


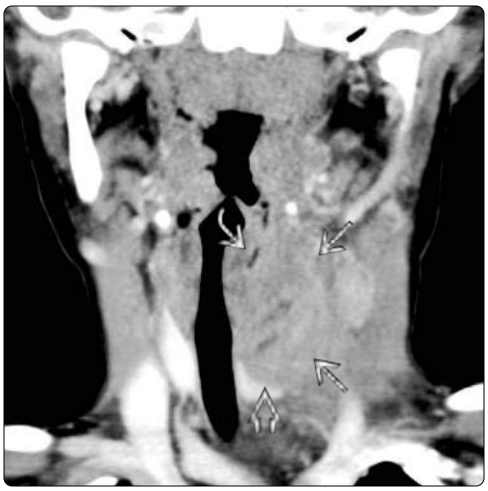
(Left) *Oblique graphic illustrates the tract of the thyroglossal duct descending from the foramen cecum (at the tongue base) to the midline hyoid bone before tracking off midline to the thyroid lobe.* **(Right)** *Axial graphic demonstrates the locations of 4 major congenital cystic lesions of the head & neck. Shown here are the infrahyoid 2nd & 3rd BC cysts, infrahyoid thyroglossal duct cyst, cervical thymic cysts, & 4th branchial apparatus sinus tracts.*





(Left) *Axial CECT in a 2-year-old boy shows an intraparotid 1st BC cyst with mild peripheral enhancement & associated mild enlargement of the ipsilateral parotid gland, consistent with parotitis.* **(Right)** *Sagittal reformatted CECT shows the typical location of a 2nd branchial apparatus fistula extending from the anterior lower neck skin opening (not included) toward the pharynx in a teenager with intermittent purulent drainage from the skin opening.*





(Left) *Axial CECT in an infant shows a large, well-defined cystic mass in the left neck that extends into the mediastinum, typical of a cervical thymic cyst. The internal bubble of gas is concerning for infection. Note the airway deviation.* **(Right)** *Coronal CECT in a 4-year-old boy shows a phlegmonous mass in the left neck involving the left thyroid lobe. There is a tract of inflammation coursing toward the left pyriform sinus, consistent with an inflamed 4th (or 3rd) branchial sinus tract.*

Nasal Dermal Sinus

KEY FACTS

TERMINOLOGY

- Defective embryogenesis of anterior neuropore resulting in any mixture of dermoid cyst, epidermoid cyst, &/or sinus tract in frontonasal region

IMAGING

- Midline location anywhere from nasal tip to anterior skull base at foramen cecum
- CT
  - Bifid crista galli with large foramen cecum
  - Fluid attenuation tract (sinus)/cyst or fat-containing mass (dermoid) from nasal dorsum to skull base within nasal septum
- MR
  - T2: Fluid signal tract in septum from nasal dorsum to skull base (sinus)
  - T1: Focal low-signal (epidermoid) or high-signal (dermoid) mass between tip of nose & apex of crista galli

TOP DIFFERENTIAL DIAGNOSES

- Fatty marrow in crista galli
- Nonossified foramen cecum
- Frontoethmoidal cephalocele
- Nasal glioma (nasal glial heterotopia)

PATHOLOGY

- Intracranial extension of nasal dermal sinus (NDS) in 20%
  - May rarely lead to meningitis
- Associated craniofacial anomalies in 15%

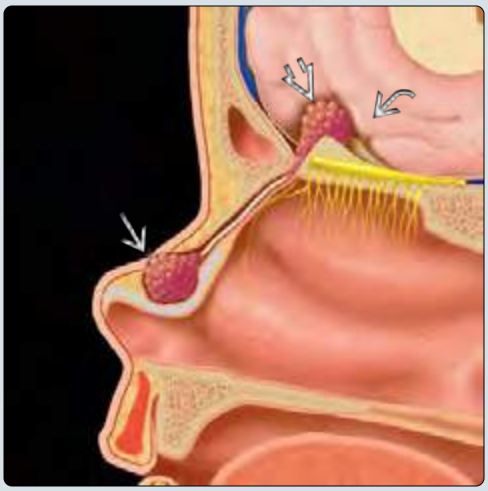
CLINICAL ISSUES

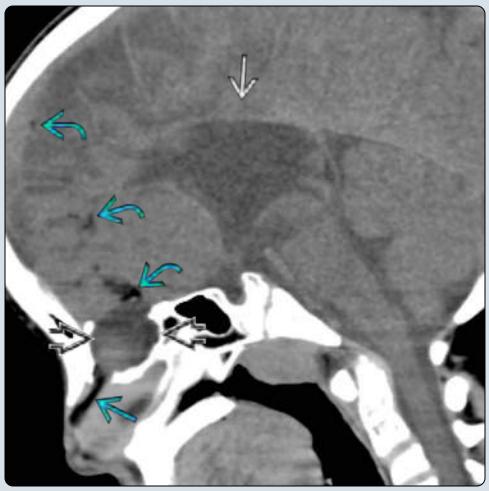
- Nasoglabellar mass (30%)
- Pit (± protruding hair) on skin of nasal bridge at osteocartilaginous nasal junction

DIAGNOSTIC CHECKLIST

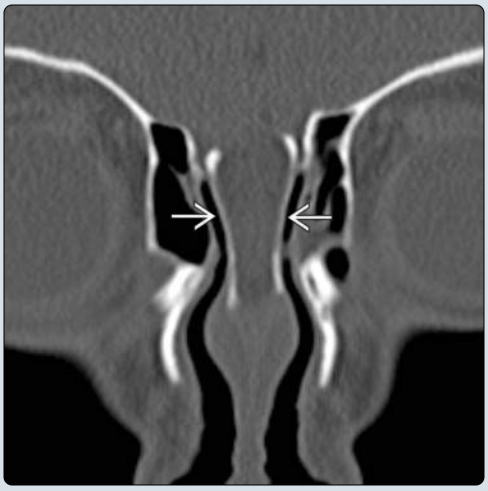
- Nasoglabellar mass or pit on nose sends clinician in search of NDS with intracranial extension

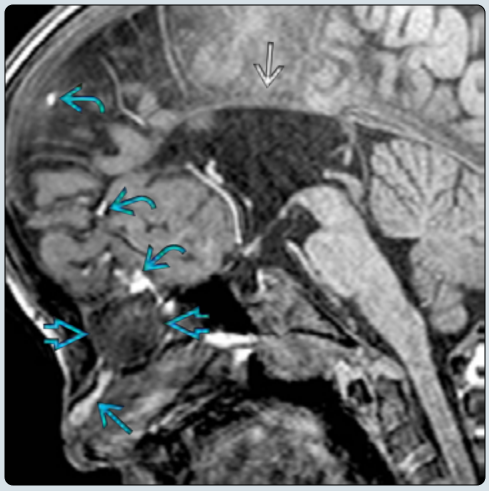
(Left) *Lateral graphic depicts a nasal dermal sinus with 2 dermoids. An extracranial dermoid is present just deep to a cutaneous nasal pit. An intracranial dermoid splits a bifid crista galli.* **(Right)** *Sagittal NECT in an 8 year old with known midline anomalies shows a fat-lined nasal dermal sinus deep to the nasal bones. The sinus leads to a large fat & fluid attenuation dermoid. There is a large defect in the cribriform plate with fatty foci extending along the falx. Agenesis of the corpus callosum is also noted.*





(Left) *Coronal bone CT in the same patient shows the bony nasal septum split by a large, fat & fluid attenuation dermoid.* **(Right)** *Sagittal T1 MR in the same patient shows the fat-lined nasal dermal sinus, large mixed signal intensity dermoid, & falcine foci of fat (consistent with intracranial extension & rupture). Agenesis of the corpus callosum is again noted. The dermoid was ultimately resected. Note that the size & extent of findings in this case is greater than typically seen.*





TERMINOLOGY

Abbreviations

- Nasal dermal sinus (NDS)

Synonyms

- Nasal dermoid, nasal dermal cyst, anterior neuropore anomaly

Definitions

- Defective embryogenesis of anterior neuropore resulting in any mixture of dermoid cyst, epidermoid cyst, &/or sinus tract in frontonasal region

IMAGING

General Features

- Best diagnostic clue
  - CT
    - Bifid crista galli with large foramen cecum
    - Fluid attenuation tract (sinus)/cyst or fat-containing mass (dermoid)
      - From nasal dorsum to skull base within nasal septum
  - MR
    - Fluid signal tract in septum from nasal dorsum to skull base (sinus)
    - Focal T1 low-signal (epidermoid) or high-signal (dermoid) mass found between tip of nose & apex of crista galli
- Location
  - Midline lesion anywhere from nasal tip to anterior skull base at foramen cecum
- Size
  - 5 mm to 2 cm for dermoid/epidermoid
- Morphology
  - Ovoid mass ± tubular sinus tract

CT Findings

- Bone CT
  - Focal tract (sinus) or mass (dermoid or epidermoid) anywhere from nasal bridge to crista galli
    - Fluid density tract = sinus
    - Fluid density mass = epidermoid
    - Fat-density mass = dermoid
  - Signs of intracranial extension
    - Large foramen cecum with bifid or deformed crista galli or cribriform plate

MR Findings

- T1WI
  - ↓ signal tract = sinus
  - ↑ signal mass = dermoid
  - ↓ signal mass = epidermoid
- T2WI
  - ↑ signal in sinus, epidermoid, or dermoid
  - Coronal plane shows septal lesions to best advantage
- DWI
  - ↑ signal = epidermoid more likely than dermoid
  - Susceptibility artifacts at skull base may obscure signal from epidermoid

Imaging Recommendations

- Best imaging tool
  - MR more sensitive for delineating sinus tract, characterizing epidermoid/dermoid lesions, & detecting intracranial extension
  - Bone CT optimal for identifying skull base defect & crista galli deformity
- Protocol advice
  - Imaging "sweet spot" is small & anterior
    - Focus imaging from tip of nose to back of crista galli
    - Inferior end of axial imaging is hard palate
    - Contrast helps with infectious complications or consideration of other lesions, not primary dermoid/epidermoid diagnosis
  - CT
    - Thin-section (1-2 mm) bone & soft tissue axial & coronal images
  - MR
    - Sagittal plane displays course of sinus tract from nasal dorsum to skull base
    - Fat-suppressed T1 images confirm fat presence in dermoids
    - DWI important additional sequence

DIFFERENTIAL DIAGNOSIS

Fatty Marrow in Crista Galli

- No nasoglabellar mass or pit on nose
- CT & MR otherwise normal

Nonossified Foramen Cecum

- Closes postnatally by 5 years of age
- Crista galli not deformed or bifid

Frontoethmoidal Cephalocele

- Bone dehiscence typically larger, involving broader area of midline cribriform plate or frontal bone
- Direct extension of meninges, subarachnoid space ± brain can be seen projecting into cephalocele on sagittal MR

Nasal Glioma

- Solid mass of dysplastic glial tissue separated from brain by subarachnoid space & meninges
- Preferred term = nasal glial heterotopia
- Most commonly projects extranasally onto paramedian bridge of nose
- Less commonly intranasal & along anterior nasal septum, off midline

PATHOLOGY

General Features

- Etiology
  - Anterior neuropore anomaly: General term for anomalous anterior neuropore regression; 3 main types
    - NDS
    - Nasal glioma (nasal glial heterotopia)
    - Anterior cephalocele
  - Embryology-anatomy: Development of anterior neuropore in 4th gestational week

- Dural stalk passes from area of future foramen cecum to area of osteocartilaginous nasal junction & then regresses completely
- Failure of involution may leave neuroectodermal remnants along tract of dural stalk
- Results in dermoid or epidermoid alone or in association with NDS tract

- Genetics
  - Familial clustering
- Associated abnormalities
  - Intracranial extension of NDS seen in 20%
  - Craniofacial anomalies (15%)

Gross Pathologic & Surgical Features

- Sinus = tube of tissue can be followed through bones
- Epidermoid = well-defined cyst; dermoid = lobular, well-defined mass

Microscopic Features

- Sinus = midline epithelial-lined tract
- Epidermoid cyst contains desquamated epithelium
- Dermoid cyst contains epithelium, keratin debris, skin adnexa

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Nasoglabellar mass (30%)
  - Pit on skin of nasal bridge at osteocartilaginous nasal junction
    - ± protruding hair
- Other signs/symptoms
  - Intermittent sebaceous material discharge from pit
  - < 50% have broadening nasal root & bridge
  - If nasal sinus tract present, recurrent meningitis may occur (rare)
- Clinical profile
  - Child (mean age: 32 months) with nasal pit ± nasoglabellar mass
  - Rarely presents in adult population
    - Episode of meningitis may be 1st problem leading to diagnosis

Demographics

- Age
  - Newborn to 5 years old
- Gender
  - Male patients with dermal sinus more likely to have intracranial extension
- Epidemiology
  - Congenital midline nasal lesions rare (1 in 20,000-40,000 births)
    - Nasal dermoids most common

Natural History & Prognosis

- Isolated problem when surgical correction successful
- Untreated patients have nasal bridge broadening ± recurrent meningitis

Treatment

- 80% require extracranial excision only
  - Local procedure to remove pit
  - Any associated dermoid or epidermoid also simultaneously removed from nasal bridge
    - Open rhinoplasty vs. transnasal endoscopic excision
- 20% undergo combined extracranial & intracranial resection
  - Biorbitofrontal nasal craniotomy approach
    - Dermoid or epidermoid along with involved dura & crista galli removed
    - Primary closure of surgical margins of dura

DIAGNOSTIC CHECKLIST

Consider

- Nasoglabellar mass or pit on nose sends clinician in search of NDS with intracranial extension
- Focused thin-section MR key to radiologic diagnosis
  - Axial coverage from cephalad margin of crista galli to hard palate
  - Coronal coverage from tip of nose to crista galli
- Add bone CT if NDS with intracranial extension found on MR

Image Interpretation Pearls

- If dermal sinus tract reaches dura of anterior cranial fossa → crista galli will be bifid with large foramen cecum
- If foramen cecum large but crista galli not bifid & tract not seen → foramen cecum normal & not yet closed
  - Foramen cecum may not close before age of 5 years
  - Do not overcall "large foramen cecum," or unnecessary craniotomy may result
  - Repeat imaging in 6-12 months to confirm foramen cecum closure acceptable approach in difficult cases

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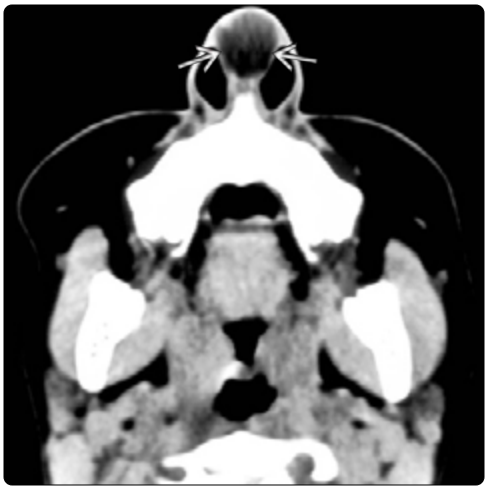
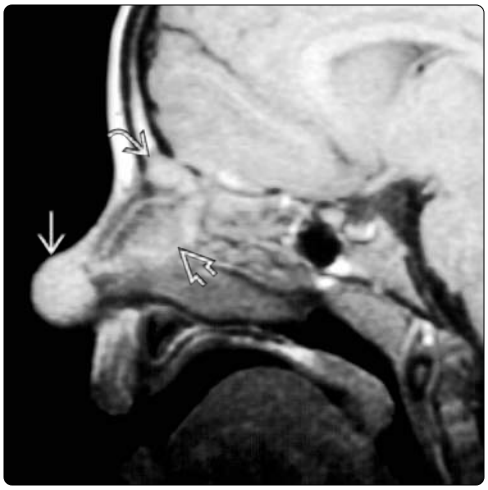
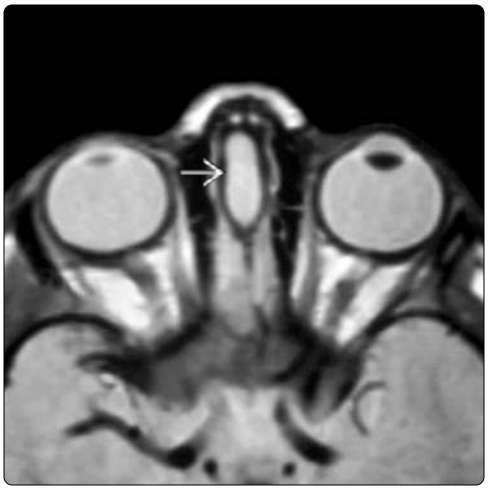
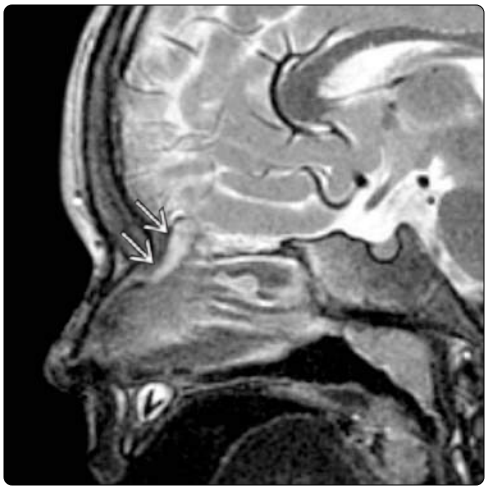


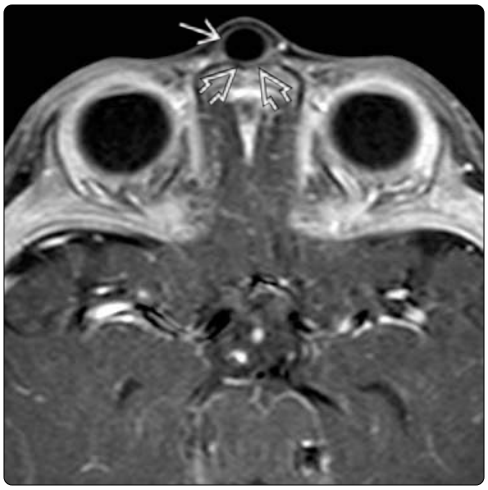


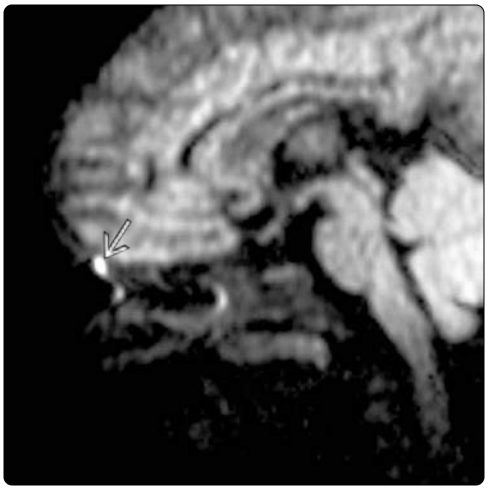
(Left) *Axial NECT shows a well-circumscribed mass consistent with a dermoid at the nasal tip. The mass is slightly higher in attenuation than adjacent fat. Dermoids were also seen in the septum & at the foramen cecum in this patient.* **(Right)** *Sagittal T1 MR in the same patient shows the dermoid at the nasal tip. Additional dermoids are noted in the nasal septum & at the skull base.*





(Left) *Axial PD MR shows the typical features of a nasal dermal sinus tract within the nasal septum. The sinus in this infant extended from the skull base to the nasal tip.* **(Right)** *Sagittal T2 MR in a 3-year-old boy with a bump on the tip of his nose shows a hyperintense sinus tract extending from the anterior skull base into the nasal septum. The features are characteristic of a dermal sinus.*





(Left) *Axial T1 C+ FS MR in a 4-month-old infant with a glabella mass demonstrates a nonenhancing, fluid signal intensity dermoid cyst with mild remodeling of the underlying nasal bones.* **(Right)** *Sagittal DWI MR reveals an epidermoid in the region of the foramen cecum as a high-signal focus of restricted diffusion. A 2nd (extracranial) lesion is not visible due to air-bone susceptibility artifact.*

Juvenile Angiofibroma

KEY FACTS

TERMINOLOGY

- Benign, vascular, nonencapsulated, locally invasive mass originating in nasal cavity

IMAGING

- Centered in posterior nasal cavity near sphenopalatine foramen; extends into nasopharynx, sphenoid sinus, pterygopalatine fossa, infratemporal fossa, orbit, skull base
- CT findings in juvenile angiofibroma (JAF)
  - Heterogeneous vs. diffuse, avid enhancement
  - Bone remodeling & destruction
    - Posterior wall of maxillary sinus bowed anteriorly
    - ± skull base invasion, intracranial extension
- MR findings in JAF
  - Tubular signal voids on spin echo-based sequences due to fast flow in enlarged vessels
  - Intense enhancement, diffuse or heterogeneous
- Angiography typically performed at time of preoperative embolization; shows tumor blush
  - Most common feeding artery: Internal maxillary branch of external carotid artery

TOP DIFFERENTIAL DIAGNOSES

- Rhabdomyosarcoma
- Nasopharyngeal carcinoma
- Antrochoanal polyp
- Esthesioneuroblastoma

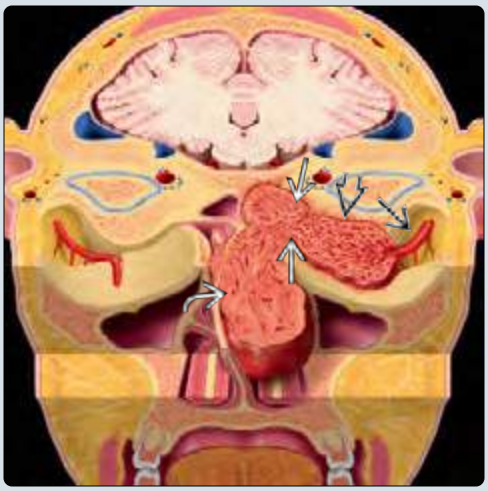
CLINICAL ISSUES

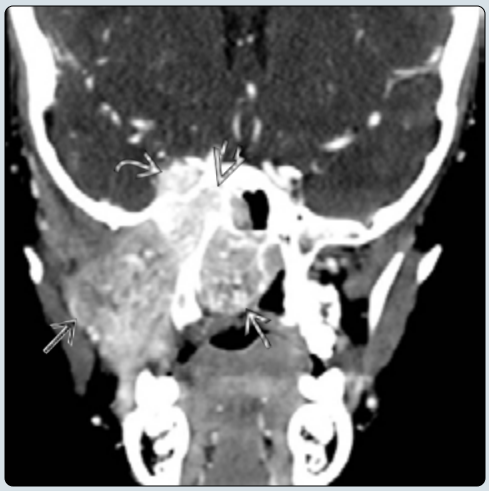
- Unilateral nasal obstruction (90%), epistaxis (60%)
  - Occurs almost exclusively in male patients
- Preferred treatment: Complete surgical resection using preoperative embolization to ↓ blood loss
  - ± adjuvant radiation therapy after surgery or as primary treatment in some cases

DIAGNOSTIC CHECKLIST

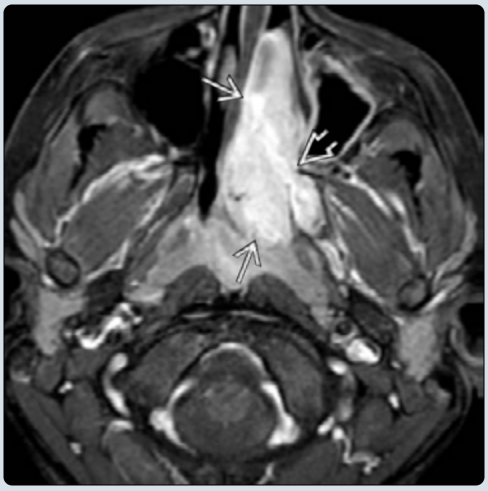
- Look for JAF extension into surrounding structures
- Consider other diagnoses in female patient

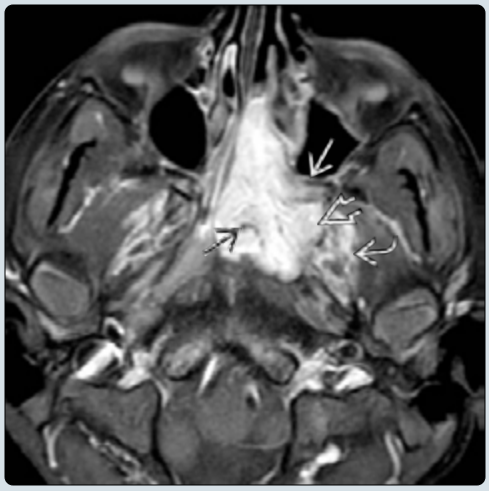
(Left) *Tiered graphic shows the classic features & location of a juvenile angiofibroma (JAF). The site of origin is the sphenopalatine foramen (SPF) ➡ with tumoral extension into the pterygopalatine fossa (PPF) ⇨ & nasal cavity ➡. The internal maxillary artery ⇨ is the dominant feeding vessel of this vascular mass.* **(Right)** *Coronal CECT in a 13-year-old patient shows the typical growth pattern of a large JAF, occluding the nasopharynx ➡ & extending into the right sphenoid ⇨, middle cranial fossa ➡, & masticator space ⇨.*





(Left) *Axial T1 C+ FS MR in a 15-year-old patient with nasal obstruction shows an enhancing JAF ➡ filling the left nasal cavity & extending into the nasopharynx. The presumed site of origin is the nasopalatine foramen ⇨.* **(Right)** *Axial T1 C+ FS MR in the same patient shows a few intralesional high-flow vessels ⇨ with extension of the JAF into the widened PPF ➡. There is destruction of the left pterygoid ⇨ with extension of the mass into the masticator space ➡.*





TERMINOLOGY

Abbreviations

- Juvenile angiofibroma (JAF)

Synonyms

- Juvenile nasopharyngeal angiofibroma (JNA); fibromatous or angiofibromatous hamartoma
  - JAF of nasal cavity is more correct terminology
  - JNA is commonly used term, but tumor begins in nasal cavity, not in nasopharynx

Definitions

- Benign, vascular, nonencapsulated, locally invasive nasal cavity mass

IMAGING

General Features

- Best diagnostic clue
  - Intensely enhancing soft tissue mass originating at sphenopalatine foramen (SPF) in adolescent male patient
- Location
  - Centered in posterior wall of nasal cavity off midline, at margin of SPF
    - Extends from posterior nasal cavity into nasal cavity, nasopharynx, & pterygopalatine fossa (PPF)
      - Penetrates PPF early (90%) with involvement of upper medial pterygoid lamina
    - Sphenoid sinus extension (60%)
    - May extend into maxillary (43%) & ethmoid sinuses (35%), masticator space (infratemporal fossa), inferior orbital fissure
    - 5-20% extend into middle cranial fossa via vidian canal or foramen rotundum
- Size
  - Usually 2-6 cm but may become massive
- Morphology
  - Lobular, usually well-circumscribed mass
    - Large lesions have infiltrating margins

Radiographic Findings

- Lateral facial plain film shows anterior displacement of posterior wall of maxillary antrum (bow sign)
- Associated with nasal cavity opacification
- ± nasal cavity/nasopharyngeal soft tissue mass

CT Findings

- CECT
  - Heterogeneous vs. diffuse avid enhancement of soft tissue mass originating near SPF with extension into adjacent nasopharynx & PPF
  - ± opacified sphenoid sinus (obstructed secretions vs. tumor infiltration)
  - ± intracranial extension
- Bone CT
  - Bone remodeling ± destruction
    - Posterior wall maxillary sinus bowed anteriorly
    - Ipsilateral nasal cavity & PPF enlarged
    - ± skull base invasion
- CTA
  - Enlarged ipsilateral external carotid artery (ECA) & internal maxillary (IMAX) artery

MR Findings

- T1WI
  - Heterogeneous, intermediate signal
  - Signal voids represent flow in enlarged vessels
- T2WI
  - Heterogeneous, intermediate- to high-signal mass
  - Punctate & serpentine flow voids within tumor
- T1WI C+
  - Intense enhancement ± flow voids
- T1WI C+ FS
  - Coronal plane shows cavernous sinus, sphenoid sinus, or skull base extension
- MRA
  - Enlarged ipsilateral ECA & IMAX artery
  - Lesional vessels may be too small to evaluate with MRA

Angiographic Findings

- Conventional angiography typically performed at time of preoperative embolization
- Intense capillary tumor blush is fed by enlarged feeding vessels from ECA
  - Most common feeding vessels: IMAX & ascending pharyngeal arteries from ECA
  - If skull base or cavernous sinus extension, internal carotid artery (ICA) supply common
  - ± supply from contralateral ECA branches

Imaging Recommendations

- Best imaging tool
  - Maxillofacial bone-only NECT in axial & coronal planes for evaluating bone remodeling vs. destruction
  - Postcontrast MR optimal for mapping lesion extent & determining vascularity
  - Catheter angiography of both ECA & ICA
    - Often in conjunction with embolization therapy
    - Helps plan surgery & ↓ intraoperative blood loss
- Protocol advice
  - Maxillofacial MR with T1 C+ FS in axial & coronal planes
    - Multiplanar imaging for evaluating extension into sphenoid sinus, orbit, skull base
  - CECT may be helpful for evaluating residual disease in postoperative period

DIFFERENTIAL DIAGNOSIS

Rhabdomyosarcoma

- Intermediate- to mildly high-signal mass with variable enhancement (often mild to moderate) ± bone destruction
- Can arise in many locations (not necessarily nasal cavity)
- Rarely penetrates SPF into PPF

Nasopharyngeal Carcinoma

- Invasive mass of lateral nasopharynx
- Mild to moderate enhancement
- 90% with nodal metastases

Antrochoanal Polyp

- Maxillary antrum opacified

- Homogeneous mass herniates into anterior nasal cavity, then nasopharynx; PPF not involved
- Peripheral enhancement only

Encephalocele

- Nasoethmoidal type presents as intranasal mass
- Connection to intracranial cavity seen on imaging
- No enhancement
- Usually more anterior in position

Esthesioneuroblastoma

- 1st incidence peaks in 2nd decade; F > M
- Presenting symptoms same as JAF
- Nasal cavity mass near cribriform plate
- Cystic intracranial components characteristic

PATHOLOGY

General Features

- Etiology
  - Source of fibrovascular tissue of JAF is not known
  - Best current hypothesis: Primitive mesenchyme of SPF is source of JAF

Staging, Grading, & Classification

- Staging systems based on tumor size (< or > 6 cm), invasion to PPF anterior &/or posterior to pterygoid plates, & skull base/intracranial invasion

Gross Pathologic & Surgical Features

- Reddish-purple, compressible, mucosa-covered mass
- Cut surface has spongy appearance

Microscopic Features

- Unencapsulated, highly vascular polypoid mass of angiomatous tissue in fibrous stroma
- Myofibroblast thought to be cell of origin
- Estrogen, testosterone, or progesterone receptors may be present

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Unilateral nasal obstruction (90%), epistaxis (60%)
- Other signs/symptoms
  - Nasal voice, nasal discharge, anosmia, pain or swelling in cheek, proptosis, serous otitis media
- Clinical profile
  - Adolescent male patient with nasal obstruction & epistaxis
  - Nasal endoscopy: Vascular-appearing nasal cavity mass
    - Biopsy in outpatient setting should be avoided due to risk of hemorrhage

Demographics

- Age
  - 10-25 years reported range
    - Average age at onset = 15 years
- Gender
  - Almost exclusively occurs in male patients
    - If found in female patient, genetic testing may reveal mosaicism
- Epidemiology
  - 0.5% of all head & neck neoplasms
  - 5-20% of JAF extends to skull base & may have skull base erosion

Natural History & Prognosis

- May rarely spontaneously regress
- Local recurrence rate with surgery: 6-24%
  - Local recurrence more common with large lesions (> 6 cm), intracranial spread, previous treatment

Treatment

- Preferred: Complete surgical resection using preoperative embolization to ↓ blood loss
- Multiple surgical approaches
  - Open resection (midface degloving) vs. endoscopic removal ± laser assistance
  - Endoscopic resection associated with ↓ bleeding & shorter hospital stay
- Radiation therapy (RT)
  - Adjuvant to surgery for unresectable intracranial disease & cavernous sinus involvement
    - 78% control rates reported
  - RT alone for cure used in some institutions
  - Used with caution in young patients due to potential to induce malignancies
- Hormonal therapy (estrogen) controversial
  - Not routine, as complete tumor regression does not occur
  - Feminization side effects undesirable in adolescent male patient

DIAGNOSTIC CHECKLIST

Consider

- JAF in adolescent male patient with epistaxis & enhancing posterior nasal cavity mass
- Consider other diagnoses in female patient

Image Interpretation Pearls

- Be sure to look for JAF extension into surrounding structures

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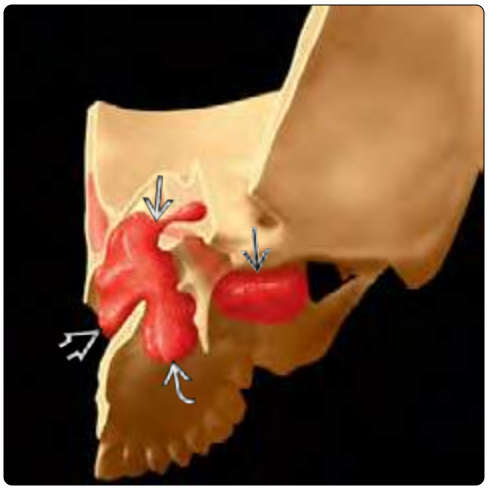
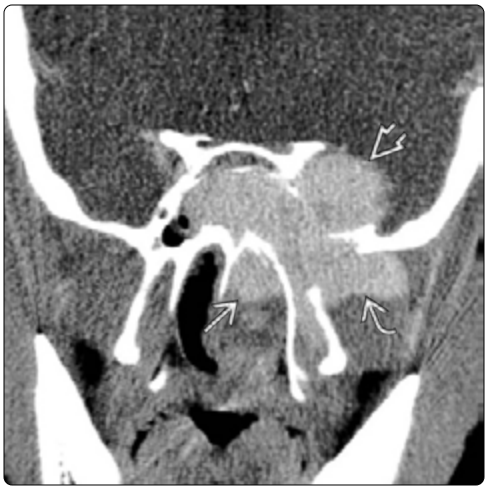
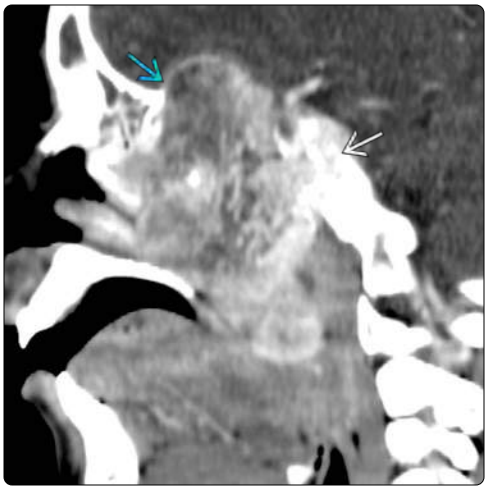
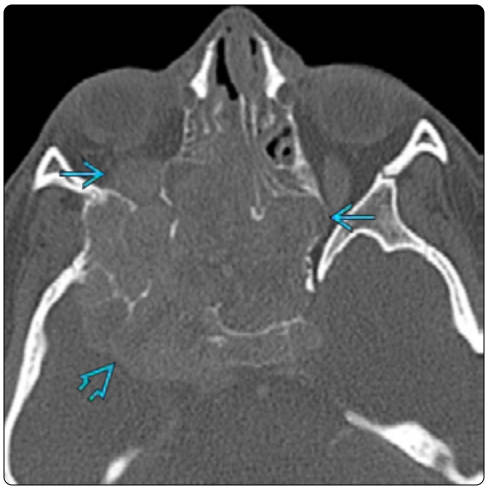


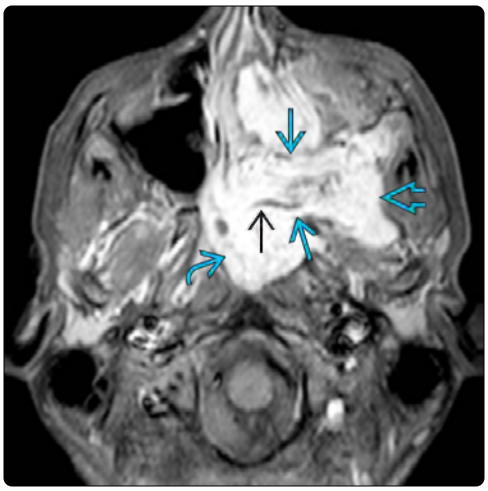


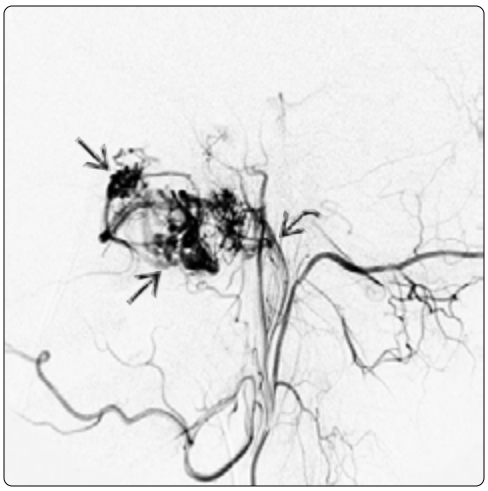
(Left) *Posterior oblique sagittal graphic shows the spread patterns of a JAF. The lesion originates at the SPF & extends into the nasal cavity, nasopharynx/oropharynx, & infratemporal fossa.* **(Right)** *Coronal CECT shows a large JAF extending into the nasopharynx, infratemporal fossa, & middle cranial fossa. The sphenoid sinus is replaced by the tumor. As seen in this case, the JAF classically shows avid enhancement.*





(Left) *Right paramidline CECT of the face in an 11-year-old boy with epistaxis shows a large, multilobulated, heterogeneously enhancing mass of the nasal cavity, consistent with a JAF. There is intracranial extension through the floor of the anterior cranial fossa, sella, & clivus.* **(Right)** *Axial CECT (in bone algorithm) in the same patient shows extensive bone destruction & remodeling by the JAF. There is invasion of the posterior ethmoid & sphenoid sinuses, right greater than left orbits, & right middle cranial fossa.*





(Left) *Axial T1 C+ FS MR shows avid enhancement throughout a JAF. The mass is centered at the SPF & extends laterally into the masticator space & medially into the nasopharynx. Several serpiginous flow voids are noted in the mass consistent with enlarged feeding vessels.* **(Right)** *Lateral DSA image from an external carotid artery injection shows areas of dense tumor blush within a JAF prior to embolization. As is typical, the main arterial feeding vessel for this JAF is the internal maxillary artery.*

Acute Rhinosinusitis

KEY FACTS

TERMINOLOGY

- Acute bacterial rhinosinusitis (ABRS), acute rhinosinusitis (ARS), viral rhinosinusitis (VRS)
- Acute inflammatory sinonasal process lasting ≤ 4 weeks

IMAGING

- ARS is clinical diagnosis & imaging rarely necessary
- Radiography: Inaccurate & should be supplanted by CT
- NECT: Confirms diagnosis, evaluates when medical therapy has failed, delineates anatomic variants (especially presurgical)
  - Axial < 1-mm acquisitions with coronal & sagittal reconstructions
    - Can review 2- to 3-mm slices
  - Best sign: Air-fluid level ± aerosolized secretions with mucosal thickening
    - Most common in ethmoid & maxillary sinuses
    - Often asymmetric sinus involvement
- CECT vs. MR: If complications suspected
- Consider dictating: "Imaging findings support clinical diagnosis of VRS or ABRS"

TOP DIFFERENTIAL DIAGNOSES

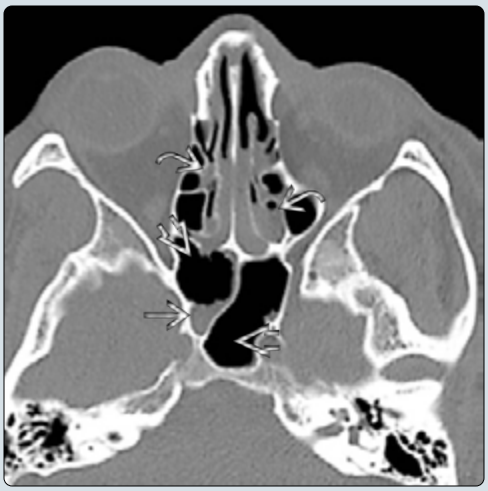
- Pseudofluid level from large maxillary polyp/cyst
- Posttraumatic blood level
- Postobstructive noninfected secretions

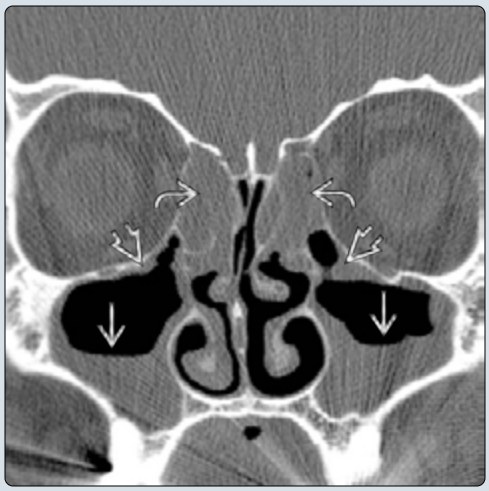
PATHOLOGY

- Most cases follow viral upper respiratory infection

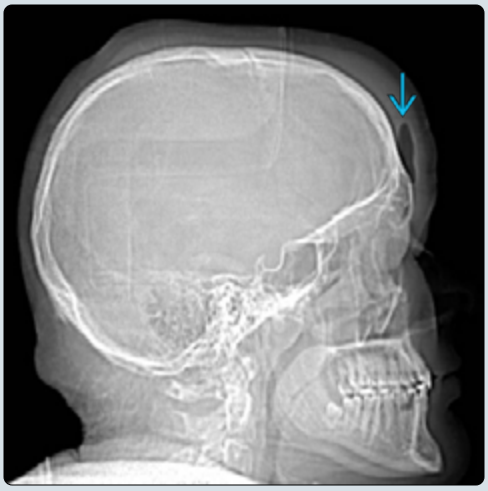
CLINICAL ISSUES

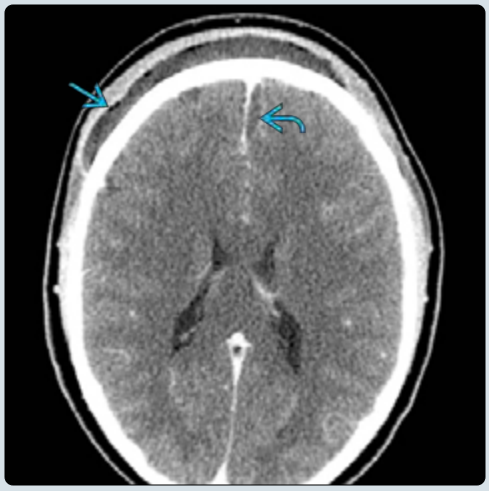
- Cardinal signs/symptoms of ARS in children: Purulent nasal drainage & obstruction, facial pain, fever, & cough
- VRS usually self-limited
- ARS complications rare
  - Orbital cellulitis, subperiosteal or subgaleal abscess, meningitis, subdural empyema, brain abscess, venous sinus thrombosis





(Left) *Axial NECT in a patient with purulent nasal discharge & headaches shows patchy bilateral ethmoid sinus disease, an air-fluid level in the right sphenoid sinus, & frothy secretions in the bilateral sphenoid sinuses corroborating the clinical history of acute bacterial rhinosinusitis (ABRS).* **(Right)** *Coronal bone CT shows acute bilateral maxillary & ethmoid sinusitis. Note the air-fluid levels & mucosal thickening in the maxillary sinuses with complete opacification of the ethmoid sinuses.*





(Left) *Lateral scout image from a head CT in a teenager with forehead swelling, fever, & confusion shows a gas collection in the edematous frontal scalp.* **(Right)** *Axial CECT in the same patient shows a large subgaleal abscess. There is diffuse effacement of cerebral sulci (due to edema) plus a left frontal parafalcine subdural collection. The frontal sinuses (not shown) contained air-fluid levels. The subdural empyema was emergently evacuated in this patient with the Pott puffy tumor of complicated sinusitis.*

TERMINOLOGY

Abbreviations

- Acute rhinosinusitis (ARS)
- Acute bacterial rhinosinusitis (ABRS)
- Viral rhinosinusitis (VRS)

Definitions

- Acute inflammatory process of sinonasal mucosa lasting ≤ 4 weeks
  - Classified into VRS vs. ABRS based on clinical presentation
- Generally clinical diagnosis; imaging only needed if complications suspected

IMAGING

General Features

- Best diagnostic clue
  - Air-fluid level ± bubbly/frothy secretions within sinus
    - Mucosal thickening often present as well but does not connote ABRS in isolation
    - Difficult to separate fluid from mucosal thickening on NECT if sinus completely opacified
- Location
  - Most common in ethmoid & maxillary sinuses, typically asymmetric
- Size
  - Normal sinus volume
    - No sinus expansion (mucocele) or reduced volume (chronic rhinosinusitis)

Radiographic Findings

- Mucosal thickening or sinus opacification ± air-fluid level
- Inaccurate & should be supplanted by CT

CT Findings

- NECT
  - Air-fluid level, bubbly or frothy-appearing secretions
  - Moderate mucosal thickening, generally > 1 cm in sinus cavity, ostium, or nasal cavity
  - May see polypoid inflammatory tissue obstructing drainage pathways
- CECT
  - Indicated when orbital or intracranial complications suspected clinically
  - Inflamed sinus mucosa enhances but thin, linear soft tissue deep to mucosa does not
  - Central secretions do not enhance
- Bone CT
  - Bone destruction not typical for acute infection
    - If present, suspect aggressive invasive sinus infection or neoplasm
  - Osteoneogenesis (sinus wall sclerosis & thickening) usually indicates chronic inflammation

MR Findings

- T1WI
  - Mucosal thickening isointense to muscle
  - Air/fluid level
  - Hyperintense secretions when chronic sinusitis present
- T2WI
  - Thickened edematous mucosa, especially in maxillary & ethmoids
  - Air/fluid level in maxillary, frontal, or sphenoid sinus
    - Ethmoid cells generally too small for discrete fluid level
  - Hypointense secretions when proteinaceous, chronic, or fungal
- T1WI C+
  - Enhancing mucosa lining sinus cavity
  - Central secretions do not enhance

Imaging Recommendations

- Best imaging tool
  - ARS is clinical diagnosis
  - Radiographs inaccurate with little role in clinical practice
  - CT: Consider only if evaluating for complications, failed medical therapy, alternative diagnoses, or surgical candidate
    - NECT delineates anatomic variants prior to functional endoscopic sinus surgery
  - CECT or MR indicated for orbital or intracranial complications; MR for invasive fungal disease or possible neoplasm
- Protocol advice
  - Bone CT: Axial ≤ 1-mm slice thickness with coronal & sagittal reconstructions to evaluate drainage pathways
    - Can review thicker (2-3 mm) reconstructions from data
  - CECT: Emergent presentations of complications
  - MR: Multiplanar T1 & T2 sequences necessary; T1 C+ FS best for intracranial/orbital complications

DIFFERENTIAL DIAGNOSIS

Pseudofluid Level

- Mucus retention cyst mimics air-fluid level
- Anterior margin typically convex, does not fill sinus wall-to-wall

Posttraumatic Blood Level

- Clinical history of recent facial injury
- ↑ attenuation of layering fluid (blood)
- Associated sinus wall fractures

Postobstructive Secretions

- Lesion obstructs sinus drainage pathway → noninfected trapped fluid
- MR differentiates tumor from obstructed secretions
  - Neoplasm generally hypointense on T2 compared to hyperintense secretions

PATHOLOGY

General Features

- Etiology
  - ABRS uncommonly follows VRS (0.5-2.0% of cases)
    - Upper respiratory infection (URI) → mucosal swelling → sinus outflow obstruction → static secretions in sinus → infection
    - Viral symptoms usually improve in 7-10 days
    - Symptoms > 10 days or worsening after 5-7 days suggest bacterial superinfection

- Common organisms: *Streptococcus pneumonia*, *Haemophilus influenzae*, *Moraxella catarrhalis*
- Odontogenic sinusitis: Apical periodontitis with dehiscence or transosseous infection extension into maxillary sinus
- Genetics
  - Cystic fibrosis (autosomal recessive disorder) predisposes to rhinosinusitis & polyps
- Associated abnormalities
  - Structural abnormalities may narrow drainage pathways
    - Anatomic variants of septum, uncinate process, middle turbinate, frontal recess, ethmoid sinuses
    - Polyps, either isolated or associated with allergic sinusitis with diffuse polyposis
    - Benign or malignant neoplasms
  - Predisposing systemic disorders: Allergies, immunoglobulin deficiency, immotile cilia syndrome, cystic fibrosis, vitamin D deficiency

Staging, Grading, & Classification

- Can be classified according to etiology: Viral, bacterial, allergic, vasomotor

Gross Pathologic & Surgical Features

- Edematous, erythematous mucosa with ostial obstruction, purulent secretions

Microscopic Features

- Tissue-invasive bacteria
- Luminal exudate of neutrophils, eosinophils

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Cardinal signs/symptoms of ARS in children: Purulent nasal drainage & obstruction, facial pain, fever, & cough
    - VRS: Signs/symptoms last < 10 days, does not progress
    - ABRS: Signs/symptoms fail to improve within 10 days or worsen within 10 days after initial improvement
  - Irritability common in children
- Other signs/symptoms
  - Fever, cough, malaise, hyposmia, anosmia, facial/dental pain, ear pressure/fullness
- Clinical profile
  - Pediatric or adult patient with nasal discharge & obstruction following viral URI lasting ≤ 4 weeks
  - Laboratory results
    - Nasal/nasopharynx cultures poorly correlate with sinus cultures, do not differentiate ABRS from VRS
      - Often contaminated with *Staphylococcus aureus*
    - Endoscopic middle meatus aspiration more specific but generally not necessary in uncomplicated ARS

Demographics

- Age
  - Generally adult disease but can be seen in children
    - Typically follows viral URI in children
- Epidemiology
  - Sinonasal inflammatory disease ubiquitous
  - Rhinosinusitis affects nearly 31 million patients (all ages) in USA annually
    - 12% (1 in 8 adults) of USA population annually
    - > 1 billion physician visits & > $3 billion in ARS healthcare expenditures/year
  - VRS & ARS often follow/coexist with common cold

Natural History & Prognosis

- VRS usually self-limited
- ABRS may resolve without antibiotics
- ABRS course may be shortened by medical therapy, surgical drainage, & possibly saline irrigation
- If ABRS untreated, rarely complications may ensue
  - Orbital cellulitis, subperiosteal or subgaleal abscess, meningitis, subdural empyema, brain abscess, venous sinus thrombosis

Treatment

- Medical therapy
  - Saline nasal sprays & irrigants, mucolytics to thin secretions
  - Decongestants, antihistamines, antibiotics
  - Nasal steroids
- Surgical therapy
  - More often performed for chronic rhinosinusitis
  - Drainage procedures may be performed in acute disease (frontal & sphenoid) to prevent development of complications

DIAGNOSTIC CHECKLIST

Consider

- Bone CT for alternative diagnoses, failed therapy, complications, or surgical candidate
  - Acquire ≤ 1-mm axial slices with coronal & sagittal reformations
  - Give contrast for suspected orbital or intracranial complications
- CT limitations
  - Cannot differentiate viral from bacterial disease
  - High incidence of sinus mucosal abnormalities in asymptomatic patients

Image Interpretation Pearls

- Air-fluid levels not always present but most specific indicator
- In correct clinical setting, even severe mucosal thickening can indicate ABRS
- Normal nasal mucosal cycle may be impossible to distinguish from ARS mucosal thickening
- Look for signs of invasive fungal sinusitis in immunocompromised patient
- Consider dictation stating, "in correct clinical setting, CT findings consistent with ARS or ABRS"

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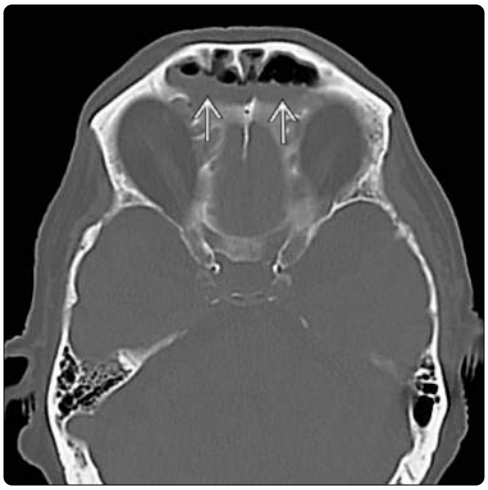
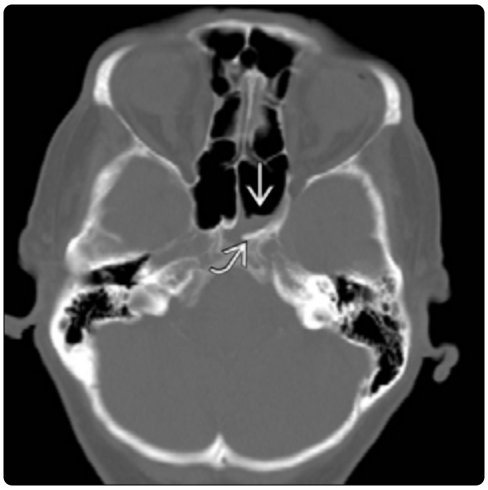
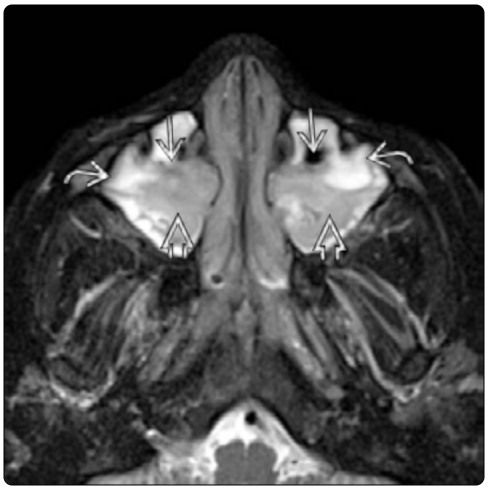
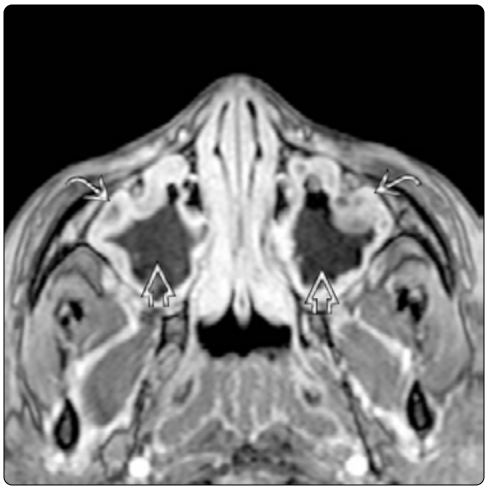


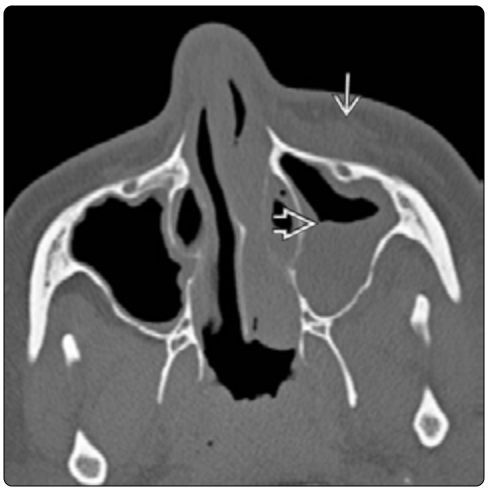


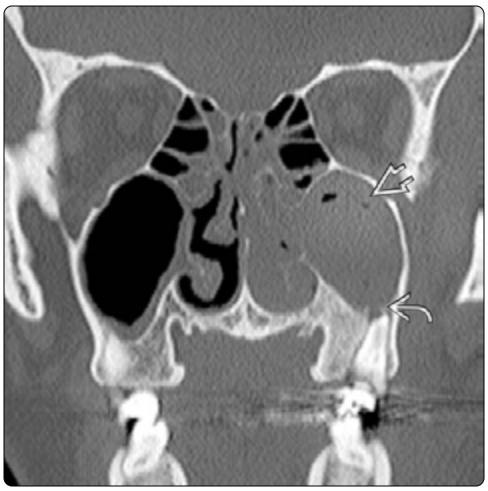
(Left) *Axial NECT performed for headache & fever reveals frothy air-fluid levels in both frontal sinuses. Although acute rhinosinusitis (ARS) is a clinical diagnosis, imaging may be performed to rule out suspected complications.* **(Right)** *Axial NECT in a patient with a clinical diagnosis of ABRS demonstrates an air-fluid level in the left sphenoid sinus. Note the osteoneogenesis, or sclerotic sinus wall, suggesting chronic sinus inflammatory disease as well as acute ABRS.*





(Left) *Axial T2 FS MR in a pediatric patient with ABRS shows near complete opacification of the maxillary sinuses with discrete air-fluid levels. Note the circumferential hyperintense edematous mucosa. The fluid is relatively hypointense secondary to increased protein content.* **(Right)** *Axial T1 C+ SPGR MR in same patient shows avid enhancement of the inflamed sinus mucosa relative to the nonenhancing central secretions. This patient had a clinical diagnosis of ABRS.*





(Left) *Axial NECT in a patient with ABRS shows left maxillary sinus opacification with an air-fluid level. The premaxillary fat soft tissue edema heralds complicated ABRS with soft tissue infection & facial cellulitis. Note the edematous mucosa in the left nasal cavity with near-complete obstruction.* **(Right)** *Coronal NECT shows opacification of the left maxillary sinus. Apical periodontitis with osseous dehiscence may contribute an odontogenic component to the ARS.*

Orbital Cellulitis

KEY FACTS

TERMINOLOGY

- Preseptal cellulitis: Infection anterior to orbital septum
- Postseptal cellulitis: Infection posterior to orbital septum
- Orbital septum: Periosteal reflection from bony orbit to tarsal plates

IMAGING

- Thickening & edema of orbital/periorbital soft tissues
- Preseptal cellulitis: Limited to anterior tissues
- Postseptal cellulitis
  - Low-attenuation, rim-enhancing collection
    - Drainable subperiosteal abscess (SPA) in majority
    - 20% without drainable abscess (phlegmon)
  - Associated myositis common with swollen extraocular muscles, ± abnormal enhancement
  - ± thrombosed superior ophthalmic vein
- ± extraorbital complications of sinusitis
  - Frontal osteomyelitis, meningitis, subdural/epidural effusion or empyema, cerebritis, parenchymal abscess

TOP DIFFERENTIAL DIAGNOSES

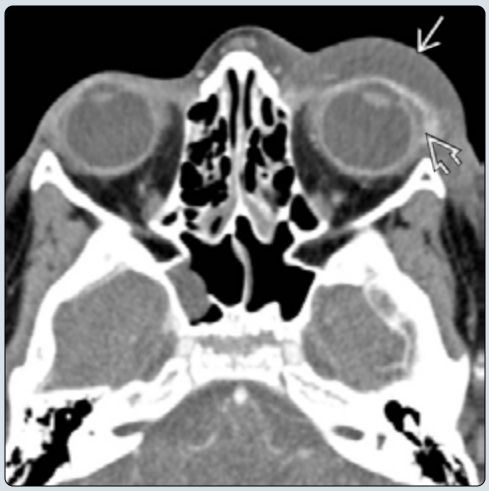
- Idiopathic orbital inflammatory pseudotumor
- Langerhans cell histiocytosis
- Orbital soft tissue or osseous neoplasm
  - Rhabdomyosarcoma
  - Metastatic neuroblastoma
- Orbital vascular anomaly
  - Infantile hemangioma
  - Venous malformation

PATHOLOGY

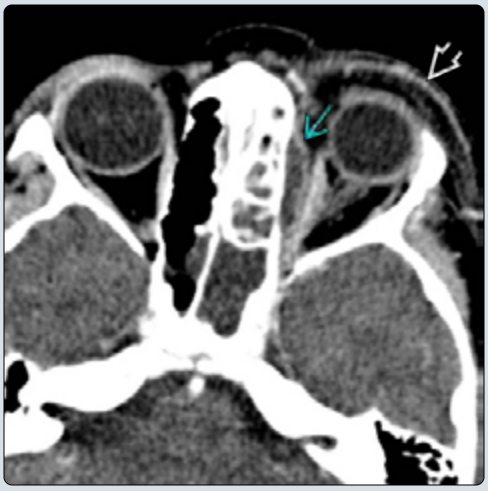
- Sinusitis most common cause of pediatric orbital cellulitis
- Other causes: Trauma, foreign body, skin infection, nasolacrimal duct mucocele; rarely retinoblastoma

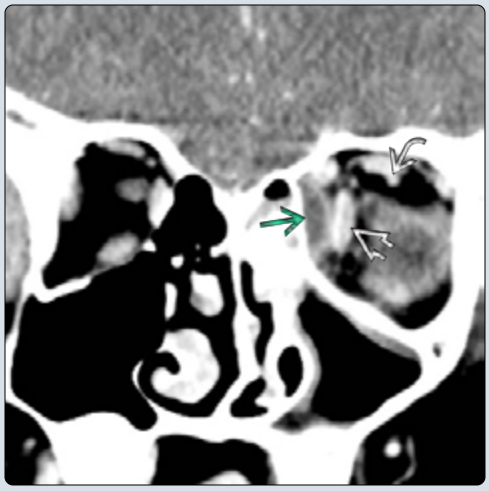
CLINICAL ISSUES

- Treatment: Intravenous antibiotics ± sinus drainage procedures, SPA drainage



(Left) *Axial graphic shows the spread of infection from the ethmoid sinuses through the lamina papyracea into the medial orbit, resulting in a subperiosteal abscess (SPA) & putting the optic nerve at risk.* **(Right)** *Axial CECT in a 7 year old with neutropenia & eyelid swelling demonstrates significant left preseptal periorbital soft tissue edema. There is a low-attenuation crescent with rim enhancement marginating the anterolateral globe, consistent with chemosis.*





(Left) *Axial CECT in a 7 year old demonstrates extensive, primarily left-sided ethmoid & sphenoid sinus disease with a medial rim-enhancing SPA. This collection causes lateral deviation of the left medial rectus muscle. There is mild associated preseptal cellulitis.* **(Right)** *Coronal CECT in the same patient shows the left medial SPA & better defines the thickening of the deviated medial rectus muscle. The left superior ophthalmic vein remains patent.*

TERMINOLOGY

Definitions

- Preseptal cellulitis: Inflammation anterior to orbital septum
- Postseptal cellulitis: Inflammation posterior to orbital septum
- Orbital septum: Periosteal reflection from bony orbit to tarsal plates of eyelids

IMAGING

General Features

- Best diagnostic clue
  - Thickening & edema of periorbital &/or orbital soft tissues = cellulitis
  - Low-attenuation, rim-enhancing subperiosteal collection in medial or superior orbit
    - Drainable subperiosteal abscess (SPA) in majority
    - Phlegmon in 20% without drainable abscess
- Location
  - Preseptal: Disease limited to periorbital soft tissues, anterior to orbital septum
  - Postseptal: Disease posterior to orbital septum
    - Intraconal: Within cone formed by extraocular muscles (EOMs)
    - Extraconal: Postseptal disease between bony orbit & EOMs
    - Subperiosteal: Between bony orbital wall & orbital periosteum
    - Associated myositis common
      - Swollen EOMs ± abnormal enhancement
  - ± extraorbital complications of sinusitis
    - Frontal osteomyelitis, meningitis, subdural/epidural effusion or empyema, cerebritis, parenchymal abscess

CT Findings

- CECT
  - Infiltration of periorbital &/or intraorbital fat with poorly-defined diffuse mild enhancement
  - ± focal elongated/lentiform rim-enhancing SPA or intraorbital abscess
    - Medial > > superior orbit
    - ± foci of gas in abscess
  - ± EOM enlargement due to myositis; EOM deviation by inflammatory collections
  - ± enlarged, centrally nonenhancing superior ophthalmic vein due to thrombosis
  - Careful review of anterior & middle cranial fossae for fluid collection or findings of parenchymal edema

MR Findings

- T1WI: Hypointense infiltration of normal fat
- T2WI FS: Heterogeneous hyperintensity of fat ± fluid in focal collection
- DWI: Restricted diffusion suggests abscess
- T1WI C+ FS: Heterogeneous enhancement of inflamed tissues ± rim-enhancing SPA

Imaging Recommendations

- Best imaging tool
  - CECT: Axial & coronal reformatted images
  - MR with contrast: Best for evaluation of intracranial complications of sinusitis

DIFFERENTIAL DIAGNOSIS

Idiopathic Orbital Inflammatory Pseudotumor

- Subacute onset
- Poorly marginated, mass-like enhancing soft tissue involving any region of orbit
  - Unilateral or bilateral

Langerhans Cell Histiocytosis

- Classic punched-out round or geographic lytic bone lesion
- Bone lesion often filled with homogeneously enhancing soft tissue mass ± intra- & extracranial extension

Orbital Neoplasm

- Variably enhancing mass originating in bone or soft tissues
- Bone-centered lesions (primary or metastatic) show aggressive permeative destruction & periosteal reaction, particularly in superolateral orbit
  - Metastatic neuroblastoma: Perpendicular radiating spicules of new bone; may show mild osseous expansion
- Soft tissue neoplasms often well-defined without surrounding edema

Orbital Vascular Anomaly

- Infantile hemangioma
  - Well-circumscribed, lobular diffusely enhancing mass
  - Superficial lesions show characteristic cutaneous features (raised red "strawberry mark")
    - Appear in first few months of life with rapid growth prior to gradual involution
- Venous malformation
  - Infiltrative pre- &/or postseptal fluid signal lesion with septated mass-like components &/or tubular channels
  - ± fluid-fluid levels, low signal thrombi, patchy enhancement
  - ± bluish skin discoloration that bulges with Valsalva
    - Present at birth

PATHOLOGY

General Features

- Etiology
  - Most common cause of periorbital/orbital cellulitis in children: Sinusitis
    - Up to 3% of sinusitis patients develop cellulitis (most common complication)
      - May precede signs & symptoms of sinusitis
      - Usually secondary to ethmoiditis
    - Spread of sinus infection to orbit
      - Direct extension via thin bone with acquired dehiscence &/or normal foramina in lamina papyracea
      - Valveless venous system (diploic veins of Breschet) connects orbital circulation with ethmoid, frontal, & maxillary sinus circulation
    - Occasional underlying cause of sinusitis
      - Antrochoanal polyp
      - Sinonasal foreign body
      - Odontogenic sinusitis

- Other causes: Trauma, foreign body, skin infection, nasolacrimal duct mucocele; retinoblastoma rarely presents as orbital cellulitis
- Associated abnormalities
  - Superior ophthalmic vein &/or cavernous sinus thrombosis
    - Expanded with central heterogeneous or nonenhancing thrombus
  - Frontal osteomyelitis (Pott puffy tumor)
    - Forehead cellulitis, phlegmon ± subgaleal abscess
    - Frontal bone lytic lesion may be difficult to detect
  - Meningitis
    - ± abnormal meningeal contrast enhancement
      - Absence of enhancement does not exclude meningitis with clinical suspicion
  - Effusions
    - Epidural (lenticular) or subdural (crescentic) fluid collections without rim enhancement or restricted diffusion
  - Empyema
    - Epidural (lenticular) or subdural (crescentic) collection of pus
    - Usually shows peripheral dural enhancement
    - Restricted diffusion on MR (↑ signal on DWI with ↓ signal on ADC map)
  - Cerebritis
    - Amorphous parenchymal edema without rim enhancing collection
  - Abscess
    - Round or ovoid collection of pus within brain
    - Ring-enhancing wall may be thicker superficially; ± low T2 signal rim on MR
    - Hyperintense DWI, hypointense ADC on MR

Staging, Grading, & Classification

- Chandler classification: Orbital complications of sinusitis
  - Preseptal cellulitis
    - Inflammation anterior to orbital septum
    - Eyelid edema
    - No tenderness, visual loss, or impaired EOM motility (ophthalmoplegia)
  - Orbital cellulitis without abscess
    - Diffuse postseptal edema of orbital fat
  - Orbital cellulitis with SPA
    - ± proptosis, impaired vision, or limited EOM motility
  - Orbital cellulitis with abscess in orbital fat
    - Usually severe proptosis, ↓ vision, & limited EOM motility
  - Cavernous sinus thrombosis secondary to orbital phlebitis: Unilateral or bilateral

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Depend on degree of inflammation
    - Fever, eyelid swelling, erythema, tenderness, chemosis, proptosis, ophthalmoplegia resulting in diplopia, ↓ visual acuity
- Other signs/symptoms
  - Cranial nerve palsies (III-VI) with cavernous sinus thrombosis
  - Seizures, mental status changes with intracranial complications

Demographics

- 50% of affected children < 4 years of age

Natural History & Prognosis

- Excellent with appropriate treatment
- Rare cause of blindness if untreated

Treatment

- Medical management: Intravenous antibiotics
- Surgical management
  - Chandler classification + imaging characteristics ↑ ability to predict surgical need
  - SPA: Not absolute surgical indication
    - Younger children may only require antibiotics with more aggressive surgical drainage in older children
    - Emergent surgery if visual disturbance from optic nerve or retinal compromise
  - Drainage of orbital fat abscess
  - Sinus drainage procedures
  - Intracranial empyema classically considered surgical emergency, particularly with neurologic signs/symptoms
    - Collection may resolve with antibiotics

DIAGNOSTIC CHECKLIST

Consider

- Imaging indications: CECT
  - Impairment in visual acuity or ophthalmoplegia
  - No improvement or worsening of symptoms on appropriate antibiotics
  - Severe eyelid edema prohibiting evaluation of vision & EOM motility
- Imaging indications: MR with contrast
  - Suspected intracranial complications
    - Fluid collection presumed infected if neurologic symptoms (seizure, altered mental status) present
    - Clinical & CT findings may dictate operative intervention before MR

Image Interpretation Pearls

- Distinguishing SPA from phlegmon may be difficult
- Cavernous sinus thrombosis may be subtle
- Look for underlying cause of sinusitis & extraorbital complications

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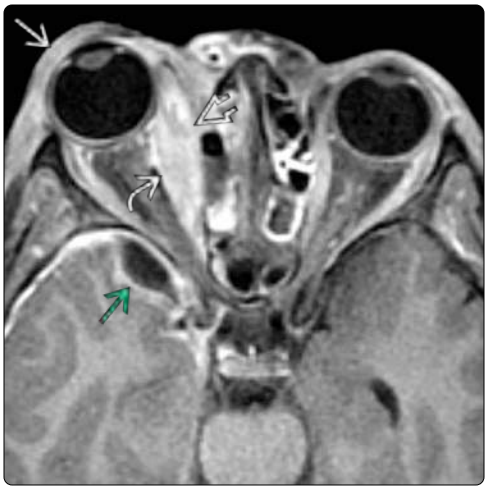
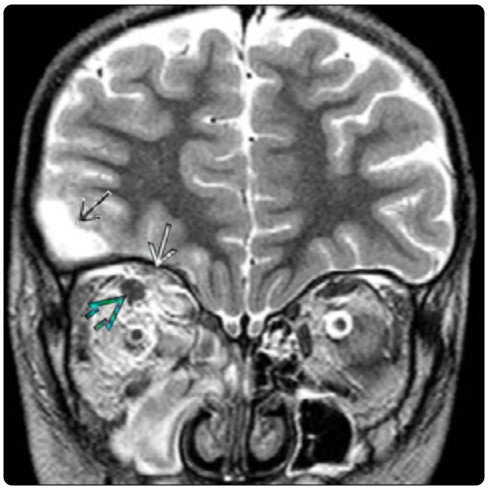
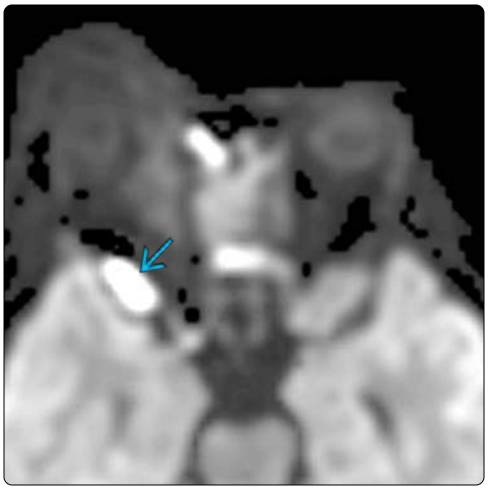
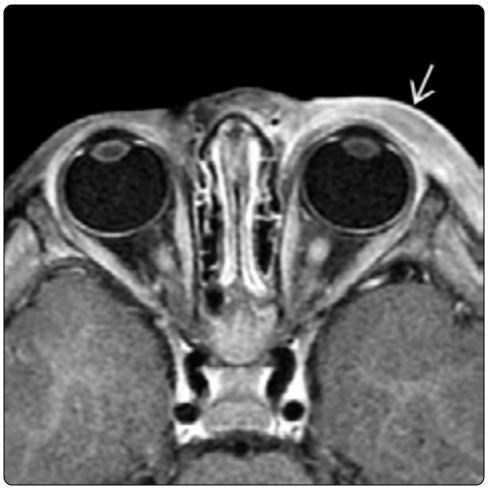


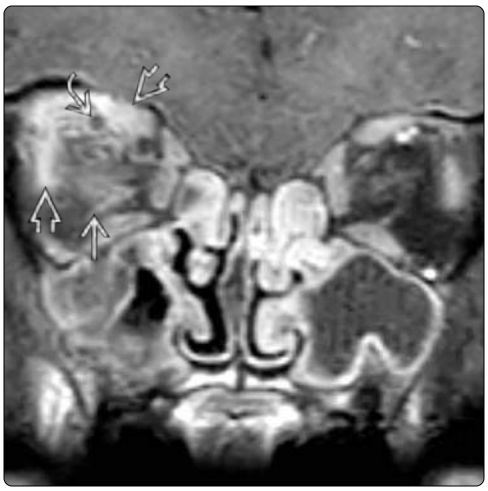


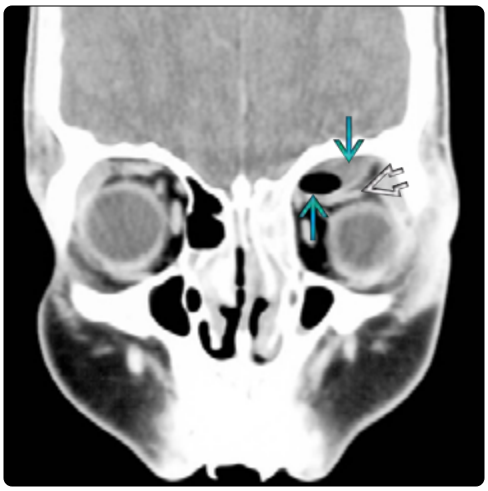
(Left) *Axial T1 C+ FS MR in a 5-year-old child with proptosis & seizures shows ethmoid sinus disease, right preseptal orbital cellulitis, medial extraconal postseptal phlegmon adjacent to an enlarged right medial rectus muscle, & a middle cranial fossa epidural abscess.* **(Right)** *Coronal T2 MR in the same child shows additional extraconal disease in the superior right orbit with inferior displacement of the right superior rectus muscle complex. There is an extraaxial fluid collection in the right anterior cranial fossa.*





(Left) *Axial DWI MR in the same child demonstrates restricted diffusion in the right middle cranial fossa fluid collection, consistent with epidural empyema.* **(Right)** *Axial T1 C+ FS MR in a 7 year old imaged for follow-up of an intracranial abnormality demonstrates new, unsuspected left preseptal cellulitis. In the absence of significant sinus disease, this finding should prompt further investigation for prior injury, lesion, or allergic reaction. In this case, the cellulitis was secondary to a bee sting 1 day prior.*





(Left) *Coronal T1 C+ FS MR in a child with extensive sinusitis shows hazy increased enhancement of the right intraconal fat, thickening of the superior & lateral rectus muscles consistent with myositis, & enlargement plus nonenhancement of the superior ophthalmic vein, consistent with thrombosis.* **(Right)** *Coronal CECT in a child with left ethmoid sinusitis shows an ipsilateral superior orbital gas-containing SPA. The left superior rectus muscle complex & globe are deviated inferiorly.*

KEY FACTS

TERMINOLOGY

- Retinoblastoma (RB): Malignant primary retinal neoplasm
- Trilateral/quadrilateral RB: Bilateral ocular RB + pineal tumor ± suprasellar tumor

IMAGING

- Unilateral in 60%, bilateral in 40%
- Trilateral or quadrilateral disease rare
- Extraocular extension in < 10%: Poor prognosis
- CT: Ca^{2+} in > 90%
- MR: Assess extent of intraocular tumor + presence of optic nerve, orbital, &/or intracranial involvement
  - T1: Mild hyperintensity
  - T2: Moderate to marked hypointensity
  - T1 C+: Moderate to marked heterogeneous enhancement

TOP DIFFERENTIAL DIAGNOSES

- Persistent hyperplastic primary vitreous
- Coats disease
- Retinopathy of prematurity
- Orbital toxocariasis

PATHOLOGY

- Primitive neuroectodermal tumor
- Sporadic (nongermline) *RB1* mutation: Most unilateral
- Inherited (germline) *RB1* mutation: Multilateral > unilateral
  - ↑ risk of 2nd remote malignancy

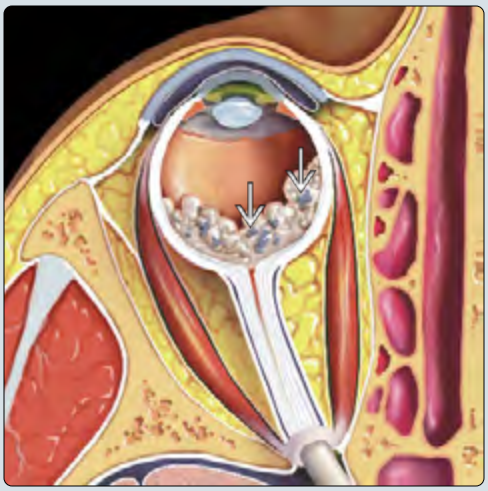
CLINICAL ISSUES

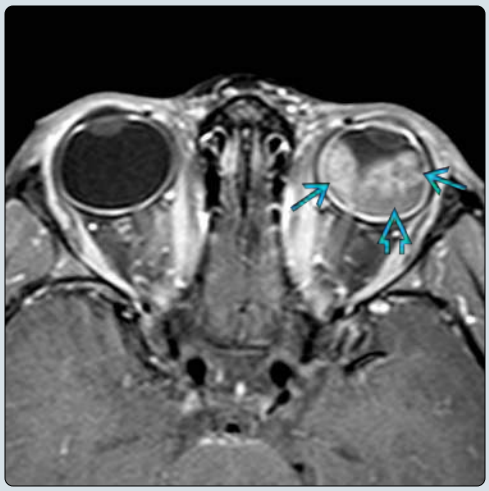
- Most common intraocular tumor of childhood
- Leukocoria in 50-60%
- 90-95% diagnosed by age 5 years

DIAGNOSTIC CHECKLIST

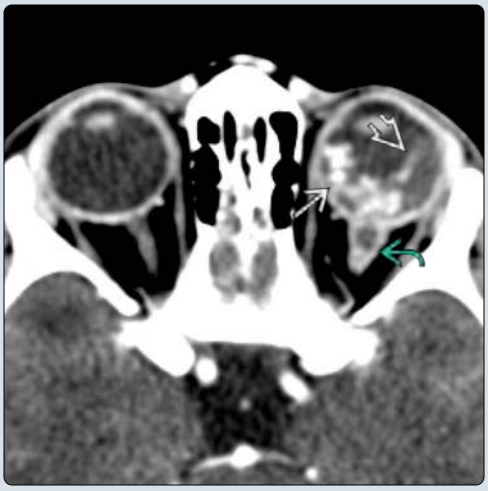
- Calcified intraocular mass in child: RB until proven otherwise

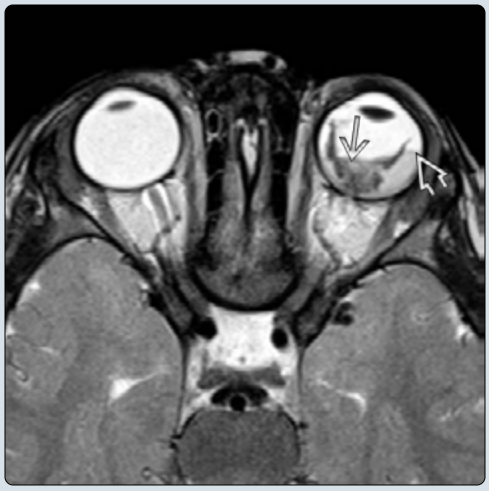
(Left) *Axial graphic depicts a retinoblastoma (RB) with lobulated tumor extending through the limiting membrane into the vitreous. Characteristic punctate Ca^{2+} ➡ are shown.* **(Right)** *Axial T1 C+ FS MR in a 3 year old with leukocoria demonstrates a large, moderately enhancing, bilobed left RB ⇨. Mild decreased signal intensity posteriorly ⇨ represents subretinal fluid secondary to retinal detachment.*





(Left) *Axial CECT in a 3 year old with leukocoria shows a partially calcified RB in the left globe ➡ with a focus of retinal detachment &/or noncalcified subretinal tumor at the temporal aspect of the globe ➡. There is also optic nerve sheath invasion ⇨, an uncommon finding on CT imaging.* **(Right)** *Axial T2 MR in the same child shows hypointensity ➡ in the calcified portions of the ocular mass with a lateral subretinal fluid level ➡ secondary to ocular detachment.*





TERMINOLOGY

Abbreviations

- Retinoblastoma (RB)

Definitions

- Malignant primary retinal neoplasm
- Trilateral RB: Bilateral ocular tumors + midline intracranial neuroblastic tumor, typically pineal
- Quadrilateral/tetralateral RB: Bilateral disease + pineal & suprasellar tumors

IMAGING

General Features

- Best diagnostic clue
  - Intraocular calcified mass in young child
  - Diagnosis typically with ophthalmoscopy & ultrasound
    - MR for tumor mapping & prognostication
- Location
  - Unilateral in 60%, bilateral in 40%
  - Trilateral or quadrilateral disease rare
  - Extraocular extension in < 10%
    - Spreads along scleral vessels into orbit & along optic nerve to subarachnoid space
  - Predictors for metastatic disease: Involvement of optic nerve, choroid, anterior chamber, or orbit
    - Anterior chamber enhancement reflects neoangiogenesis → associated with more aggressive tumor behavior
  - Role of MR imaging
    - Exclude pseudoneoplastic lesions
    - Assess intraocular (choroid, sclera, prelaminar optic nerve), extraocular (postlaminar optic nerve, orbital), & intracranial (pineal, parasellar, metastatic) involvement
- Morphology
  - Irregular, solid, & heterogeneous
- Growth patterns
  - Endophytic form (45%)
    - Inward protrusion into vitreous
    - Associated with vitreous seeding
  - Exophytic form (45%)
    - Outward growth into subretinal space, typically with hemispherical configuration
    - Associated retinal detachment & subretinal exudate
  - Mixed endophytic & exophytic (10%)

CT Findings

- NECT
  - Punctate or finely speckled Ca^{2+} (> 90-95%)
- CECT
  - Moderate to marked heterogeneous enhancement

MR Findings

- T1WI
  - Variable, mildly hyperintense (relative to vitreous)
- T2WI
  - Moderately to markedly hypointense (relative to vitreous)
    - Helps distinguish from hyperintense lesions (e.g., persistent hyperplastic primary vitreous, Coats disease)
  - Best for identifying subretinal fluid ± vitreous hemorrhage
- T1WI C+
  - Moderate to marked heterogeneous enhancement
  - Best to assess extent of intraocular disease & presence of optic nerve or extraocular invasion
    - Choroidal invasion: Localized thickening & heterogeneous contrast enhancement near tumor
    - Scleral invasion: Interruption in thin hypointense zone surrounding enhancing choroid
    - Optic nerve invasion: Thickening of optic disc (prelaminar), enhancement of nerve (postlaminar)
      - MR shown to have low sensitivity & specificity in assessing optic nerve invasion

Ultrasonographic Findings

- A-scan: Highly reflective spikes at Ca^{2+}
- B-scan: Echodense, irregular mass with focal shadows

Imaging Recommendations

- Best imaging tool
  - T1 C+ FS & T2 MR images best for tumor mapping
  - Intraocular Ca^{2+} on CT relatively specific in child
- Protocol advice
  - Include whole brain to assess for intracranial disease

DIFFERENTIAL DIAGNOSIS

Persistent Hyperplastic Primary Vitreous

- Small globe, hyperdense tissue, no Ca^{2+}
- Hyperintense on T2 MR with enhancing retrolental stalk

Coats Disease

- Normal size globe, hyperdense exudate, rare Ca^{2+}
- Hyperintense on T1 & T2 MR

Retinopathy of Prematurity

- Retrolental fibroplasia; associated with excess oxygen & premature retinal vessels
- Bilateral, small globes, hyperdense tissue, Ca^{2+} if advanced

Retinal Astrocytic Hamartoma

- Rare lesion often associated with tuberous sclerosis; ± Ca^{2+}

Toxocariasis

- Uveoscleral enhancement with no Ca^{2+} acutely

Other Causes of Leukocoria

- Retinal detachment
  - Subretinal hemorrhage, retinal folds
- Choroidal osteoma
- Choroidal angioma
- Retinal dysplasia

PATHOLOGY

General Features

- Etiology
  - Primitive neuroectodermal tumor
  - Sporadic (nongermline): 60% of RB
    - Majority (85%) of unilateral disease

- Inherited (germline): 40% of RB
  - Essentially all bilateral & multilateral disease
    - Minority (15%) of unilateral disease
  - Autosomal dominant with 90% penetrance
  - Positive family history in 5-10%
  - New germline mutations in 30-35%
- Genetics
  - *RB1* gene: Chromosome 13q14
  - Somatic mosaicism in 10-20% of RB patients
- Associated abnormalities
  - ↑ risk of 2nd malignancy in germline disease
    - Sarcoma, melanoma, CNS tumors, epithelial tumors (lung, bladder, breast)
    - 20-30% in nonirradiated patients
    - 50-60% in irradiated patients
    - Occur within 30 years; average 10-13 years
  - 13q deletion syndrome: RB + multiple organ anomalies

Staging, Grading, & Classification

- Reese-Ellsworth classification
  - Groups 1-5
  - Based on size, location, & multifocality
  - More useful in radiation therapy management
- International (Murphree) Classification of Retinoblastoma
  - Groups A-E
  - Based on size, retinal location, subretinal or vitreous seeding, & several specific prognostic features
  - More useful in chemotherapy management

Gross Pathologic & Surgical Features

- Yellowish-white irregular pedunculated retinal mass

Microscopic Features

- Small round cells, scant cytoplasm, & large nuclei
- Flexner-Wintersteiner rosettes & fleurettes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Leukocoria in 50-60%
- Other signs/symptoms
  - Severe vision loss
  - Strabismus with macular involvement or retinal detachment
  - Proptosis with significant orbital disease
  - Rubeosis iridis (redness of iris secondary to neovascularization) correlates with anterior chamber enhancement on MR
  - Inflammatory signs in 10%

Demographics

- Age
  - Congenital but usually not apparent at birth
  - Average age of RB diagnosis: 18 months
    - Unilateral: 24 months; bilateral: 13 months
    - Earlier with family history & routine screening
  - 90-95% diagnosed by age 5 years
- Epidemiology
  - Most common intraocular tumor of childhood
  - Incidence of 1:17,000 live births (↑ in past 60 years)
  - 3% of cancers in children < 15 years old
  - 1% of cancer deaths; 5% of childhood blindness

Natural History & Prognosis

- Degree of nerve involvement correlates with survival
  - Superficial or no invasion: 90%
  - Invasion to lamina cribrosa (prelaminar): 70%
  - Invasion beyond lamina cribrosa (postlaminar): 60%
  - Involvement at surgical margin: 20%
- Poor prognosis for extraocular disease
  - < 10% 5-year disease-free survival
- Poor prognosis for trilateral disease or CSF spread
  - < 24-month survival

Treatment

- > 95% of children with RB in United States cured with modern techniques
  - Maintaining eye & vision remains problematic
- Therapy based on tumor volume & localization, intraocular tumor extent, & extraocular stage of disease
- Enucleation: Used in advanced disease with no chance of preserving useful vision
- External beam radiation therapy: Indicated for bulky tumors with seeding
  - Unfavorable complications (e.g., arrested bone growth, radiation-induced tumors)
- Chemotherapy ("chemoreduction"): Currently favored 1st-line therapy for lower grade intraocular tumors
  - Intraarterial & intravitreous chemotherapy gaining traction with high globe salvage rates
    - Limits need for systemic chemotherapy, external radiation, & enucleation
- Plaque radiotherapy: Locally directed I-125 or other isotope
  - Selected solitary or small tumors
- Cryotherapy: Primary local treatment of small anterior tumors
- Photocoagulation: Primary local treatment of small posterior tumors

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Calcified intraocular mass in child: RB until proven otherwise
- Assess for intraocular & extraocular spread
- Check for intracranial trilateral or quadrilateral disease in pineal & suprasellar regions

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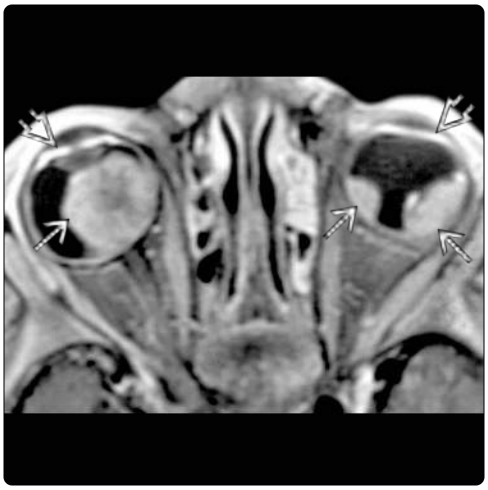
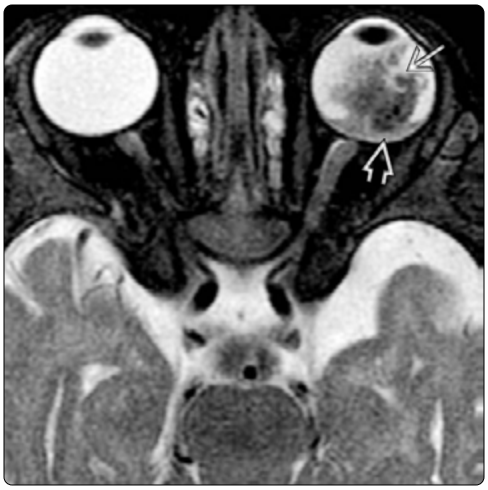
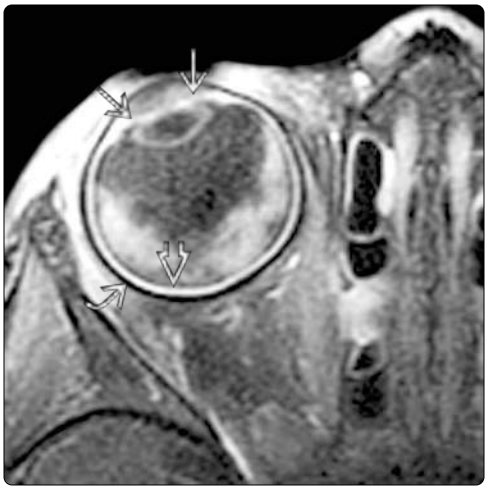
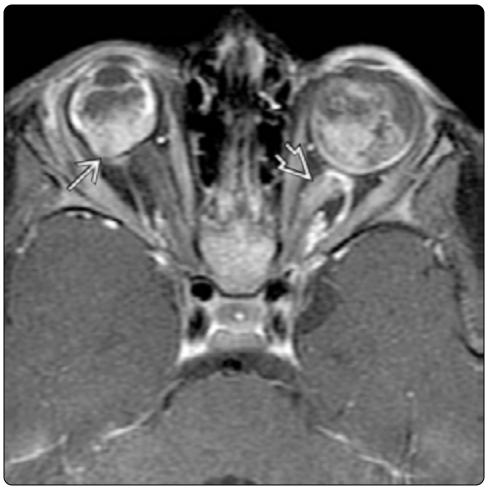


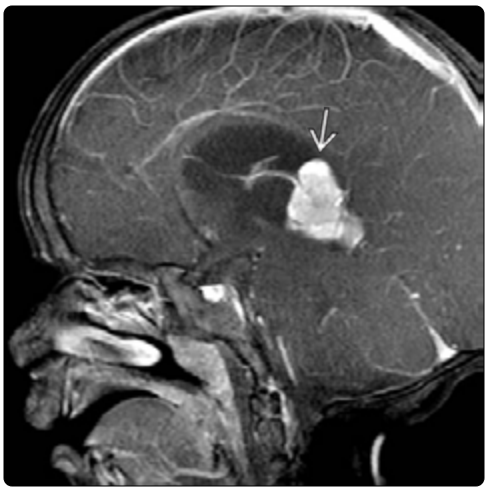


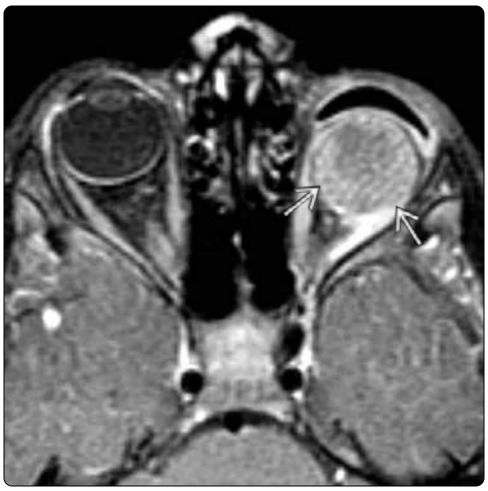
(Left) *Axial T1 C+ FS MR shows bulky enhancing intraocular masses compatible with bilateral RB. Note the prominent enhancement of each iris. Anterior segment enhancement is associated with more aggressive tumor behavior.* **(Right)** *Axial T2 FS MR reveals a lobular, hypointense mass filling much of the vitreous compartment of the left globe. Note the posterior fluid level indicating vitreous hemorrhage associated with an endophytic form of RB.*





(Left) *Axial T1 C+ FS MR shows a moderately enhancing RB associated with prominent anterior segment enhancement. Note the intact thin lines of enhancing choroid & hypointense sclera, indicating absence of invasion.* **(Right)** *Axial T1 C+ FS MR shows interruption of linear hypointensity posteriorly with focal contour abnormality, indicating scleral invasion by RB on the right. The left postlaminar optic nerve demonstrates asymmetric enlargement & enhancement, consistent with tumor extension.*





(Left) *Sagittal T1 C+ FS MR in a 3 year old with bilateral RB demonstrates a lobulated, enhancing mass in the region of the pineal gland. This mass was proven to represent pineoblastoma/PNET, typical of trilateral RB.* **(Right)** *Axial T1 C+ FS MR in a child after left globe enucleation for treatment of a large RB shows the hydroxyapatite intraorbital component with a well-defined, spherical prosthesis. There is typical moderate, heterogeneous postcontrast enhancement secondary to fibrovascular tissue infiltration.*

Congenital Auricle Malformations

KEY FACTS

TERMINOLOGY

- Congenital aural dysplasia (CAD): Congenital external ear malformation
- Outer ear: Auricle (pinna) & external auditory canal (EAC)
- Microtia: Congenital malformation; underdeveloped, misshapen auricle

IMAGING

- Small, malformed, ± low-set pinna
- EAC stenosis/atresia & middle ear (ME)/ossicular anomalies
- Degree of microtia correlates with degree of EAC stenosis/atresia & ME anomalies

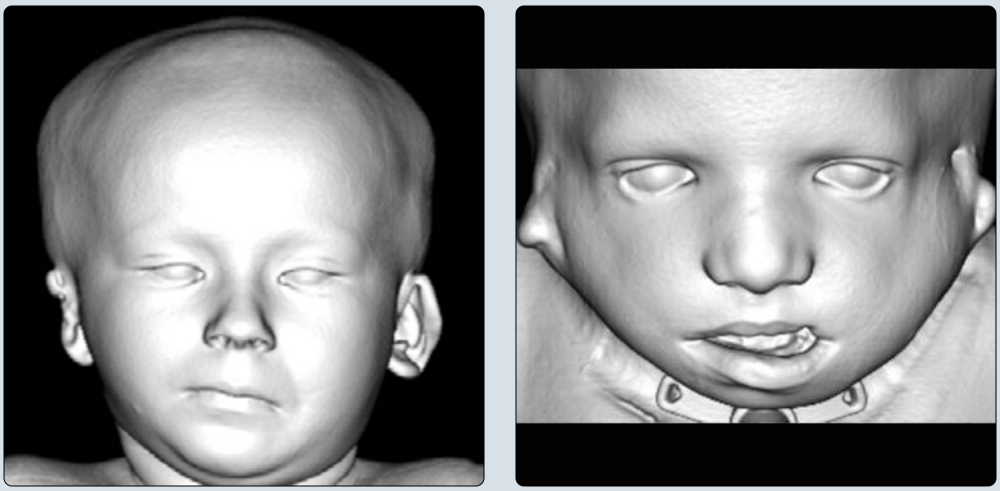
TOP DIFFERENTIAL DIAGNOSES

- Syndromes with auricle malformations
  - Treacher Collins syndrome
  - Hemifacial microsomia
  - Branchiootorenal syndrome
  - CHARGE syndrome

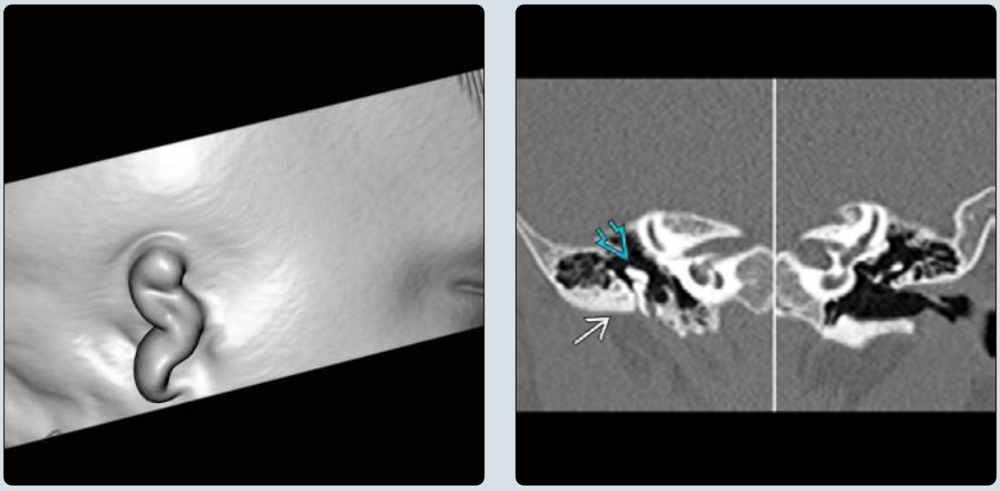
PATHOLOGY

- Weerda classification of auricular dysplasia
  - 1st-degree dysplasia: Macrotia, prominent ear, pocket ear, absence of upper helix, absence of tragus, clefts, lobular deformities, & cup ear types I & II
  - 2nd-degree dysplasia: Cup ear type III, mini-ear
  - 3rd-degree dysplasia: Auricular remnant with no recognizable structures of normal auricle
- Minor microtia: 1st & 2nd degree
  - Associated with EAC stenosis
- Major microtia: 3rd degree
  - Associated with EAC atresia (75%)
- Associated temporal bone abnormalities
  - EAC stenosis or atresia & malformed ossicles
  - ± ↓ ME & mastoid pneumatization
  - 1/3 with CAD have inner ear malformations
  - Anomalous CNVII canal ± oval window anomaly
  - Congenital or acquired ME cholesteatoma

(Left) *Frontal 3D soft tissue surface-rendered NECT reformation in a 3-year-old girl with hemifacial microsomia demonstrates a right-sided 2nd-degree microtia, a small right mandible, & a normal left pinna.* **(Right)** *Frontal 3D soft tissue surface-rendered NECT reformation in a child with Treacher Collins syndrome shows bilateral 3rd-degree microtia, downward-slanting palpebral fissures, & symmetric micrognathia.*



(Left) *Lateral 3D soft tissue surface-rendered NECT reformation shows a moderately deformed right pinna (2nd-degree malformation) in a 10 month old with right CEMEM.* **(Right)** *Coronal temporal bone CTs in the same patient clearly show the atretic right EAC. The deformed right malleus & incus are fused to the lateral epitympanic cavity, & the middle ear cavity is underdeveloped. All the findings are easier to identify when compared to the normal contralateral left temporal bone.*



TERMINOLOGY

Synonyms

- Congenital aural dysplasia: Congenital external ear malformation
- Outer ear: Auricle & external auditory canal (EAC)
- Auricle: Pinna

Definitions

- Microtia: Congenital malformation; underdeveloped & misshapen auricle
- Anotia: Absent auricle

IMAGING

General Features

- Best diagnostic clue
  - Small, malformed, ± low-set pinna
    - Associated EAC stenosis/atresia, middle ear (ME) & ossicular malformation
  - Degree of microtia correlates with degree of EAC stenosis/atresia & ME anomalies
- Location
  - Auricle, EAC, ME, ± inner ear (IE)
  - Right > left (60:40); 1/3 of cases bilateral

CT Findings

- Bone CT
  - Identifies congenital external & middle ear malformation (CEMEM)
  - 3D soft tissue reconstruction demonstrates microtia

Imaging Recommendations

- Best imaging tool
  - Temporal bone CT with multiplanar & 3D reconstruction

DIFFERENTIAL DIAGNOSIS

Syndromes With Auricle Malformations

- Treacher Collins syndrome
- Hemifacial microsomia
- Branchiootorenal syndrome
- Pierre Robin sequence
- CHARGE syndrome

PATHOLOGY

General Features

- Etiology
  - CEMEM secondary to anomalous 1st & 2nd branchial arch development 1st trimester
  - Genetic or teratogenic insult
- Associated abnormalities
  - < 30% of CEMEM have IE malformations
  - Up to 30% of CEMEM have syndromic etiology
  - ↓ ME & mastoid pneumatization in 25% of minor & 66% of major microtia
  - Abnormal course of CNVII canal ± oval window atresia
  - Congenital or acquired cholesteatoma behind atretic plate in up to 10% of patients with CEMEM
  - Micrognathia with low-set pinna in syndromic cases

Staging, Grading, & Classification

- Weerda classification of auricular dysplasia
  - Minor microtia
    - 1st-degree dysplasia: Macrotia, prominent ear, pocket ear, absence of upper helix, absence of tragus, clefts, lobular deformities, & cup ear types I & II
    - 2nd-degree dysplasia: Cup ear type III, mini-ear
      - ± low, anterior position of pinnae
      - 75% of minor microtia patients have EAC stenosis
  - Major microtia
    - 3rd-degree dysplasia: Auricular remnant with no recognizable structures of normal auricle
      - Includes anotia
      - Severely dysplastic ears frequently anterior & inferior in position secondary to incomplete embryologic ascent in neck
      - 75% of patients with major microtia have EAC atresia; remainder have EAC stenosis

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Microtia or anotia & stenotic/absent EAC
  - Conductive or mixed hearing loss
- Other signs/symptoms
  - Micrognathia: Unilateral (hemifacial microsomia), bilateral (Treacher Collins) with low-set pinna

Demographics

- Overall incidence of CEMEM: 1/3,300-10,000 live births

Treatment

- Complicated surgical correction of pinnae
  - Autogenous rib construction: 2 or more stages
    - More durable, less prone to infection
  - Alloplastic porous high-density polyethylene reconstruction: 2 stages
    - More aesthetic, less morbidity, & can be done at younger age
- ± drilling new EAC &/or ossicle reconstruction

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- Degree of microtia correlates with underdevelopment of EAC, ME, & mastoid air cells
- Look for erosive opacity due to associated cholesteatoma

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Large Vestibular Aqueduct (IP-II)

KEY FACTS

TERMINOLOGY

- Large vestibular aqueduct (LVA) houses enlarged endolymphatic sac (ES) & duct
- IP-II: Incomplete partition between middle & apical cochlear turns is typically seen with LVA
- IP-II + LVA: Mondini anomaly (historic terminology)

IMAGING

- Axial CT: LVA ≥ 1 mm at midpoint &/or ≥ 2 mm at operculum; short-axis CT reformat: LVA ≥ 1.2 mm at midpoint, ≥ 1.3 mm at operculum
- MR: Enlarged ES & duct
- Cochlea: Abnormal in ~ 75% of LVA cases
  - Absent septation between middle & apical turns
  - Deficient modiolus, asymmetric scalar chambers
- Vestibule & semicircular canal: Normal or mildly enlarged

TOP DIFFERENTIAL DIAGNOSES

- Cystic cochleovestibular malformation (IP-I)
- Cochlear hypoplasia
- CHARGE syndrome (funnel-shaped LVA)
- Branchiootorenal syndrome (funnel-shaped LVA)

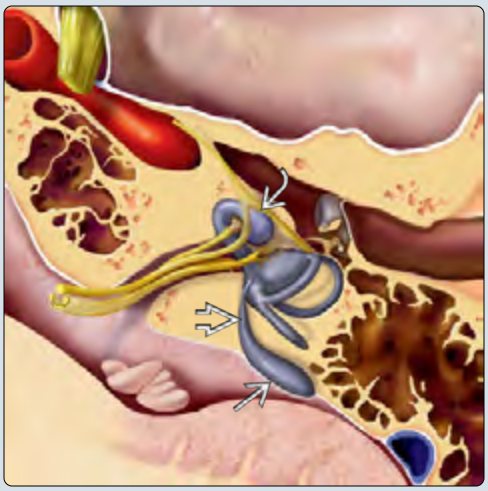
PATHOLOGY

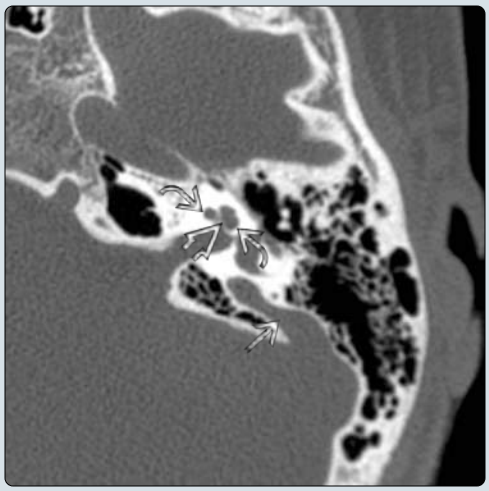
- *SLC26A4* mutations
  - Autosomal recessive, ~ 5-10% of cases of prelingual hearing loss (HL)
  - Syndromic deafness: Pendred syndrome
  - Nonsyndromic deafness: DFNB4
  - ~ 50% of LVA patients have *SLC26A4* mutations

CLINICAL ISSUES

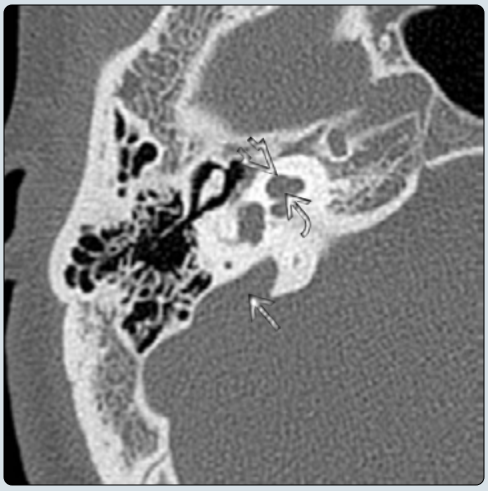
- Most common imaging abnormality in pediatric SNHL
- Bilateral anomaly (most)
- Congenital cause of acquired SNHL or mixed HL
- Progressive/fluctuating SNHL
- Avoid contact sports & try to prevent head trauma
- Cochlear implantation for profound bilateral SNHL

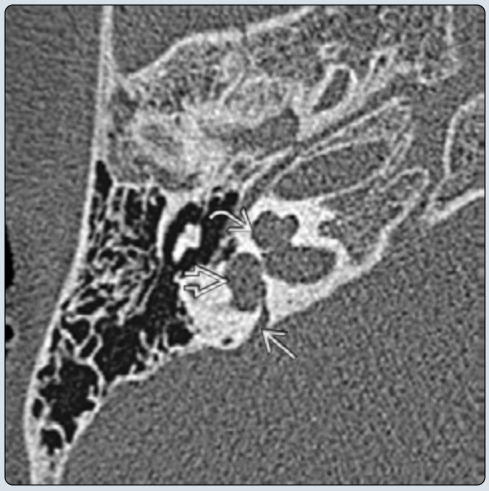
(Left) *Axial graphic of left inner ear shows the large endolymphatic sac epidural & intraosseous components. The cochlea is malformed with absence of septation between the middle & apical turns, which appear bulbous.* **(Right)** *Axial temporal bone CT in a 15-year-old boy with SNHL reveals a large vestibular aqueduct (LVA). The interscalar septum is present between the apical & middle cochlear turns, but the modiolus is narrow & the scalar chambers are asymmetric (posterior > anterior).*





(Left) *Axial temporal bone CT in a 17-year-old girl with Pendred syndrome shows an LVA & absent septation between the middle & apical cochlear turns with absence of the modiolus. This is typical of incomplete partition type II, which results in a baseball cap-shaped cochlea.* **(Right)** *Axial temporal bone CT in a 3 year old with SNHL & LVA shows a typical IP-II cochlear anomaly with plump cochlear middle & apical turns, absent modiolus, & no apical septation. The vestibule is mildly enlarged.*





TERMINOLOGY

Definitions

- Large vestibular aqueduct (LVA): Enlarged bony vestibular aqueduct (VA) houses large endolymphatic sac (ES) & duct associated with variable cochlear malformation
- IP-II: Incomplete partition (deficient interscalar septum) between middle & apical cochlear turns is usually seen with LVA

IMAGING

General Features

- Size
  - Axial temporal bone CT: LVA ≥ 1 mm at midpoint, ± ≥ 2 mm at opercular margin perpendicular to long axis of VA
    - Short-axis reformat, parallel to plane of superior semicircular canal (SCC): ≥ 1.2 mm at midpoint, ≥ 1.3 mm at operculum
- Morphology
  - Axial temporal bone CT: V-shaped, enlarged bony VA
  - Axial T2 MR: Large ES along posterior wall of petrous bone, lateral to dural reflection

CT Findings

- Bone CT
  - LVA: May scallop posterior margin of petrous bone
  - Cochlea: Abnormal on CT in ~ 75% of LVA cases
    - Normal basal turn
    - Normal or deficient septation between plump apical & middle turns (IP-II)
    - Asymmetric scalar chambers: Anterior < posterior
    - Deficient (typical), absent, or normal modiolus
  - Vestibule: Normal or mildly enlarged; vestibular volume usually increased
  - SCC: Normal or mildly dilated
  - Middle ear space & ossicles: Normal

DIFFERENTIAL DIAGNOSIS

Cystic Cochleovestibular Malformation (IP-I)

- Cystic cochlea without internal structure & cystic vestibule

Cochlear Hypoplasia

- Small cochlea, usually < 2 turns

CHARGE Syndrome

- Funnel-shaped LVA, small vestibule, & small/absent SCC
- Cochlear nerve canal stenosis/atresia & thickened modiolus; occasional cochlear hypoplasia

Branchiootorenal Syndrome

- Funnel-shaped LVA, tapered basal turn, & small, offset middle & apical turns

High Jugular Bulb

- Communicates with jugular foramen, not vestibule
- Normal variation of jugular foramen anatomy

PATHOLOGY

General Features

- Genetics
  - *SLC26A4* mutations (*PDS* gene, chromosome 7)
    - ~ 50% of LVA patients have *SLC26A4* mutations
    - Syndromic deafness: Pendred syndrome
      - Sensorineural hearing loss (SNHL) + thyroid organification defect ± goiter
      - Autosomal recessive; biallelic *SLC26A4* mutation
      - ~ 10% of hereditary deafness

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - SNHL [or mixed hearing loss (MHL)]
    - Bilateral or unilateral; severe or profound
    - Fluctuating or progressive course
    - SNHL precipitated by minor head trauma
- Other signs/symptoms
  - Tinnitus, vertigo, dizziness
  - Pendred syndrome: Hypothyroidism in ~ 50%, ± goiter in adolescence

Demographics

- Age
  - Prelingual or early postlingual SNHL (or MHL)
  - Congenital cause of acquired SNHL
- Epidemiology
  - LVA: Most common CT/MR abnormality in pediatric SNHL
  - Bilateral (most) or unilateral anomaly

Natural History & Prognosis

- If bilateral, inevitably → profound SNHL
- SNHL (or MHL)
  - Hearing loss (HL) may not be present until early adult life
  - Fluctuating or progressive course
- ± linear relationship observed between VA width & progressive SNHL
- Prognosis best for unilateral HL or late-onset HL

Treatment

- Avoid contact sports & try to prevent head trauma
- Profound bilateral SNHL: Cochlear implantation
  - No ↑ in cochlear implant complications

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Congenital Cholesteatoma

KEY FACTS

TERMINOLOGY

- Congenital cholesteatoma (CCh): Benign mass secondary to epithelial rests of embryonal origin
- CCh most commonly occurs in middle ear (ME) behind intact tympanic membrane (TM) in patient without history of surgery, chronic otitis media (COM), or otorrhea

IMAGING

- Majority in ME (CCh-ME)
  - Anterosuperior tympanic cavity near eustachian tube or stapes most common
  - Posterior epitympanum at tympanic isthmus
  - Other sites include petrous apex & mastoid
- Temporal bone CT findings
  - Small well-circumscribed ME lesion medial to ossicles
  - Large mass may erode ossicles, ME wall, lateral semicircular canal, or tegmen tympani
- MR findings: Peripherally enhancing ME mass with diffusion restriction in larger lesions

TOP DIFFERENTIAL DIAGNOSES

- Acquired cholesteatoma
- Rhabdomyosarcoma
- Langerhans cell histiocytosis
- Glomus tympanicum paraganglioma

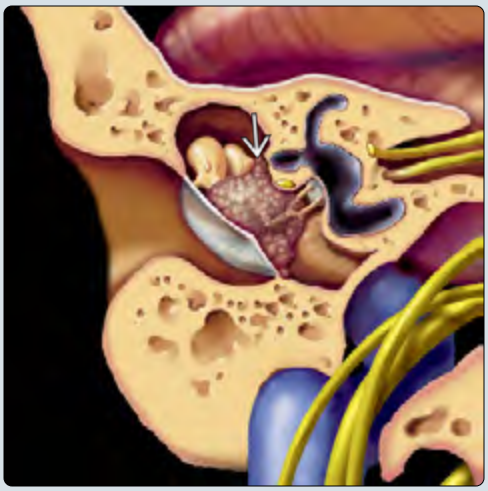
CLINICAL ISSUES

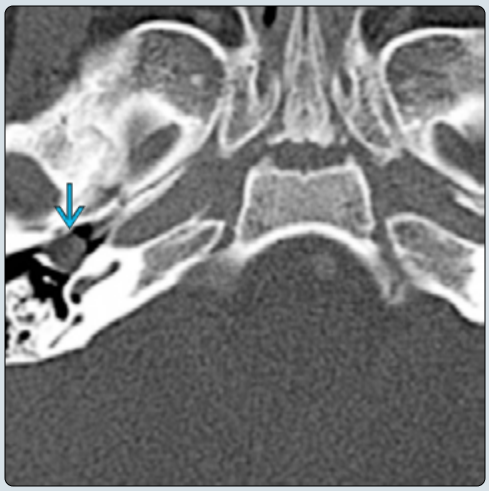
- Avascular pearly white ME mass behind intact TM without prior history of inflammation or trauma
- Unilateral conductive hearing loss in 30%
- Large ME lesions can obstruct eustachian tube → ME effusion & infection
- Complete surgical extirpation = treatment of choice

DIAGNOSTIC CHECKLIST

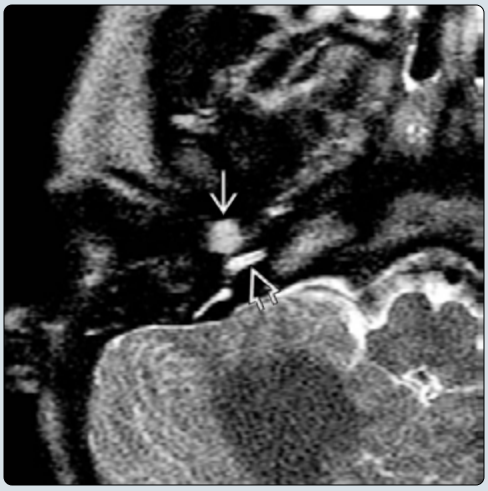
- Child with ME mass medial to ossicles, normal mastoid aeration, & no history of COM = CCh-ME

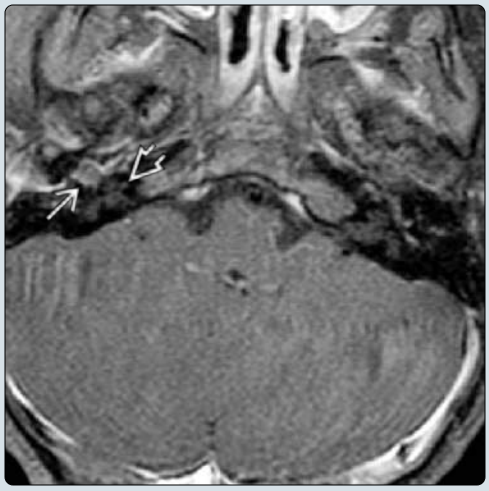
(Left) *Coronal graphic shows a congenital cholesteatoma (CCh) involving the middle ear (ME) cavity. Notice that the lesion has extended medial to the ossicles ➡ as it engulfs the entire ossicular chain. The tympanic membrane (TM) is intact, a typical finding in congenital lesions, as opposed to acquired cholesteatomas (which are associated with TM perforation).* **(Right)** *Axial bone CT in a 3-year-old child without a history of chronic otitis media shows a typical CCh ⇨ anteromedial to the malleus manubrium.*





(Left) *Axial T2 MR in the same patient shows the CCh as an intermediate signal intensity well-defined ME mass ➡ anterolateral to the basal turn ➡ of the right cochlea.* **(Right)** *Axial T1 C+ FS MR in the same patient shows the centrally nonenhancing CCh with a thin rim of peripheral enhancement ➡ adjacent to the cochlea ➡. This is a typical appearance & location for a CCh.*





TERMINOLOGY

Abbreviations

- Congenital cholesteatoma (CCh)

Synonyms

- Primary cholesteatoma, epidermoid

Definitions

- Benign mass secondary to epithelial rests of embryonal origin
- Most commonly occurs in middle ear (ME), behind intact tympanic membrane (TM) in patient without history of surgery, chronic otitis media, or otorrhea

IMAGING

General Features

- Best diagnostic clue
  - Smooth, well-circumscribed ME mass ± ossicular erosions
- Location
  - Majority in ME (CCh-ME)
    - Anterosuperior tympanic cavity near eustachian tube or stapes: Most common
    - Posterior epitympanum at tympanic isthmus (between ME cavity & attic)
  - Other locations: External auditory canal (EAC), mastoid, petrous apex, cerebellopontine angle, facial nerve canal
- Size
  - Usually small (presents on otoscopic exam)
  - Rarely fills entire ME cavity
- Morphology
  - Round, lobular, discrete ME mass

CT Findings

- Bone CT
  - Appearance depends on size of lesion & location
  - Small CCh-ME: Well-circumscribed lesion
  - Large CCh-ME: Mass may erode ossicles, ME wall, lateral semicircular canal, or tegmen tympani [similar to acquired cholesteatoma (ACh)]
    - Bone erosion < ACh; occurs late in disease
    - Ossicular erosion unusual with anterior mesotympanum involvement
    - Long process of incus & stapes superstructure are most commonly destroyed ossicles
    - Labyrinthine extension may occur late
    - If aditus ad antrum occluded, mastoid air cells opacify with retained secretions
  - Common locations of CCh-ME
    - Anterosuperior ME, adjacent to eustachian tube & anterior tympanic ring, medial to ossicles
    - Inferior but adjacent to tensor tympani muscle; mimics pars tensa-ACh-ME that also often ends up medial to ossicles
    - Near stapes
    - Posterior epitympanum, at tympanic isthmus
  - Petrous apex: Expansile mass with smooth bone remodeling/erosion
  - Mastoid: Expansile mass with smooth bone erosion
  - EAC: CCh rare, ↑ in patients with EAC malformations

MR Findings

- T1WI: Iso- to hypointense mass
- T2WI: Intermediate signal intensity mass
  - Larger ME lesions → aditus ad antrum obstruction → high signal retained secretions in mastoid air cells
- DWI: Larger lesions demonstrate diffusion restriction
- T1WI C+: Peripherally enhancing ME mass
  - CCh-ME nonenhancing material surrounded by thin, subtle rim enhancement
  - If lesion longstanding, associated scar may lead to area of enhancement adjacent to CCh-ME

Imaging Recommendations

- Exam of choice: Temporal bone CT
- T1 C+ MR complementary in certain circumstances
  - Recommended for recurrent or large cholesteatoma
  - Recommended if diagnosis uncertain: Glomus tumor enhances

DIFFERENTIAL DIAGNOSIS

Acquired Cholesteatoma

- Otoscopy reveals retraction pocket & pars flaccida or pars tensa TM perforation
- CT findings
  - Pars flaccida-ACh
    - Scutum erosion with lesion in Prussak space of lateral epitympanum
    - Ossicular chain & lateral semicircular canal more likely eroded
    - Chronic inflammatory changes
  - Pars tensa ACh
    - Lesion enlarges medial to ossicles
    - Ossicular erosion common

Rhabdomyosarcoma

- Destructive mass in ME & mastoid
  - ± lateral extension into EAC
  - ± medial extension into internal auditory canal
  - ± cephalad extension into middle cranial fossa
  - ± posterior extension into posterior cranial fossa
  - ± inferior extension into temporal mandibular joint &/or nasopharyngeal, masticator, or parotid spaces

Langerhans Cell Histiocytosis

- Conductive hearing loss ± otorrhea
- Enhancing soft tissue mass with bone destruction
  - Temporal bone, orbit, maxilla, mandible, cervical spine, & skull

Glomus Tympanicum Paraganglioma

- Pulsatile, vascular mass behind TM
  - Unusual in pediatric & adolescent patients
- Enhancing mass on cochlear promontory; no bone erosion

Middle Ear Cholesterol Granuloma

- Otoscopy reveals blue TM
- ME mass with high signal intensity on T1WI MR
  - Ossicular erosion common

PATHOLOGY

General Features

- Etiology
  - 2 principal theories of CCh-ME
    - Congenital ectodermal rest in ME cavity left behind at time of neural tube closure (3rd-5th weeks of gestation)
    - Lack of regression of epidermoid formation
      - Epidermoid formation: Point of epithelial transformation between tympanic cavity & eustachian tube
      - When fails to regress → mass-like ME accumulation of stratified epithelial squamous cells→ anterosuperior CCh-ME
- Associated abnormalities
  - EAC atresia may have associated CCh of ME or EAC
  - Rarely associated with 1st branchial cleft remnant

Staging, Grading, & Classification

- CCh-ME staging system
  - Stage 1: Single quadrant; no ossicular involvement or mastoid extension
  - Stage 2: Multiple quadrants; no ossicular involvement or mastoid extension
  - Stage 3: Ossicular involvement; no mastoid extension
  - Stage 4: Mastoid extension

Gross Pathologic & Surgical Features

- Circumscribed, pearly white mass with capsular sheen
- If detected early, no associated inflammatory changes

Microscopic Features

- Identical to epidermoid inclusion cyst
- Stratified squamous epithelium with progressive exfoliation of keratinous material
- Contents rich in cholesterol crystals

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Avascular pearly white ME mass behind intact TM without prior history of inflammation or trauma
- Other signs/symptoms
  - Unilateral conductive hearing loss in 30%
  - Large ME lesions can obstruct eustachian tube → ME effusion & infection
  - May be discovered surgically after chronic ME effusion unresponsive to tympanostomy tubes
  - Mastoid or petrous apex mass on imaging, with symptoms related to involvement of adjacent structures
  - Rarely EAC mass with bone destruction

Demographics

- Age
  - Average age of presentation or detection
    - Anterior or anterosuperior: 4 years
    - Posterosuperior & mesotympanum: 12 years
    - Attic & mastoid antrum: 20 years
- Gender
  - M:F = 3:1
- Epidemiology
  - 2-5% of temporal bone cholesteatomas congenital

Natural History & Prognosis

- CCh-ME: Smaller, anterior lesions have better outcome with complete surgical resection
- If untreated, keratin debris accumulates over time → enlarging mass
  - Enlarging, cyst-like CCh-ME may rupture, extending throughout ME cavity
  - Eustachian tube obstruction → ME effusions & otomastoiditis
  - Larger lesions with infection difficult to differentiate from ACh
- Large lesions or posterior epitympanic CCh have recurrence rates as high as 20%
  - Staged surgical resection often used for large lesions

Treatment

- Complete surgical extirpation = treatment of choice
  - Tympanoplasty for small, well-encapsulated CCh-ME
  - Tympanoplasty + canal wall down mastoidectomy for large CCh-ME
- ± ossicle chain reconstruction

DIAGNOSTIC CHECKLIST

Consider

- CCh-ME when mass seen behind intact TM
- CCh-ME with no history of recurrent ME infections
- CCh-ME when ME opacified with wall erosion in patient with congenital external ear dysplasia

Image Interpretation Pearls

- Young patient with ME mass medial to ossicles + normal mastoid pneumatization, without history of infection = CCh-ME

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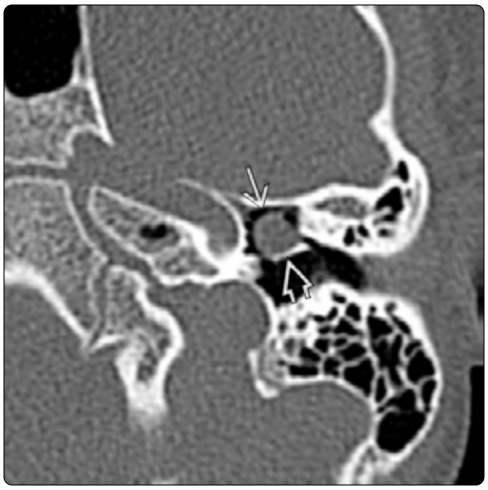
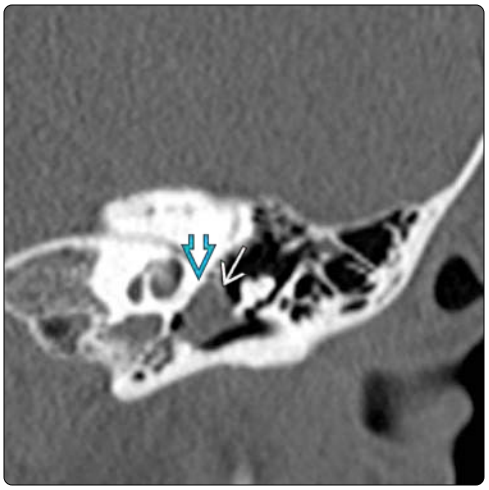
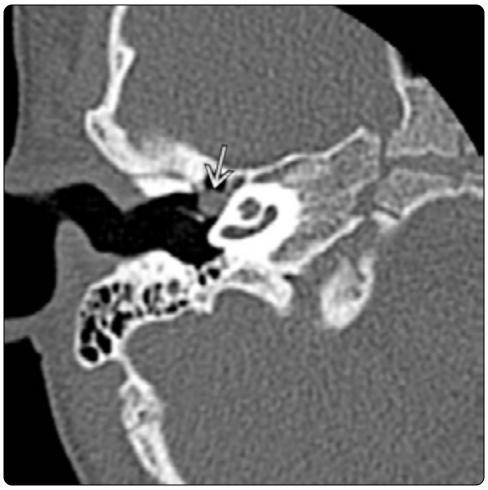
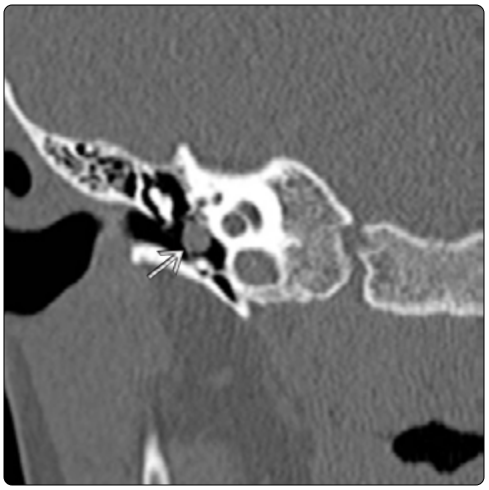


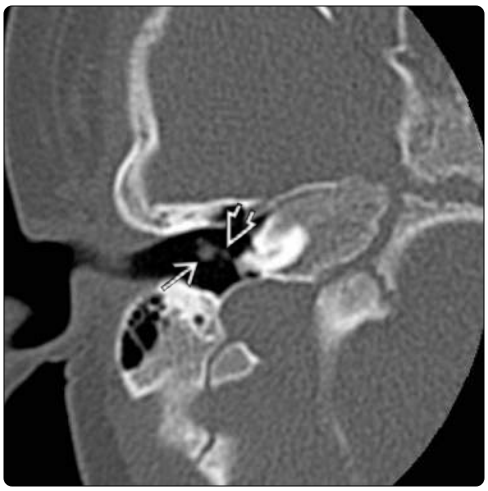


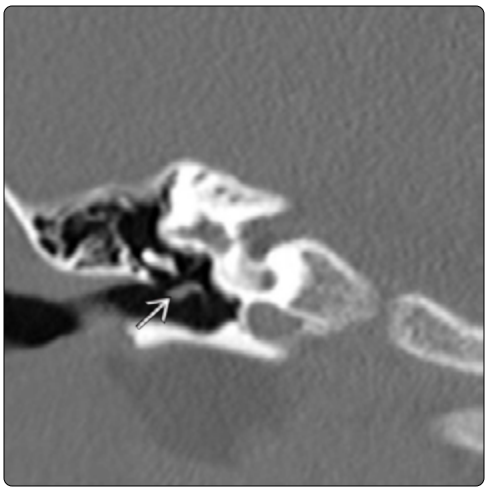
(Left) *Axial bone CT in a 4-year-old patient with left congenital hearing loss demonstrates a well-defined, round soft tissue mass in the left ME cavity abutting the anterior margin of the malleus manubrium.* **(Right)** *Coronal bone CT in the same patient shows the typical appearance of a ME cavity CCh medial to the ossicles & abutting the cochlear promontory. There is no osseous erosion.*





(Left) *Axial bone CT in a 5-year-old patient (in whom the pediatrician noticed a pearly white ME mass during otoscopic evaluation) shows a small CCh just anterior to the malleus without erosion.* **(Right)** *Coronal bone CT in the same patient shows the typical appearance of a soft tissue mass in the mesotympanum, lateral to the cochlear promontory.*





(Left) *Axial bone CT in an 18-month-old patient with a suspected CCh shows a very small soft tissue mass dorsal to the lowermost aspect of the malleus.* **(Right)** *Coronal bone CT in the same patent shows the ME cavity mass in the mesotympanum, inferior & lateral to the ossicles & abutting the tympanic membrane. This mass was pathologically proven to be a cholesteatoma. This is an unusual case involving the TM as most CChs are located more superiorly, medial to the ossicles.*

Acquired Cholesteatoma

KEY FACTS

TERMINOLOGY

- Secondary or acquired cholesteatoma (ACh)
  - Tympanic membrane (TM) retraction or perforation → accumulation of stratified squamous epithelial cells in middle ear (ME) → mass-like keratin ball
- Pars flaccida cholesteatoma (PFC) (80%)
- Pars tensa cholesteatoma (PTC) (15%)
- Sinus cholesteatoma = PTC in sinus tympani

IMAGING

- Best modalities
  - Noncontrast bone CT: Axial & coronal
    - Ossicular & adjacent bone evaluation
  - Coronal T1 C+ FS MR
    - Suspected intracranial extension/infection
- Nonenhancing ME soft tissue mass + ossicular erosion
  - PFC: In Prussak space with scutum erosion
    - ± tegmen tympani, lateral semicircular canal, facial nerve canal, or sigmoid sinus plate dehiscence
  - PTC: In posterior mesotympanum medial to ossicles
    - ± ossicular erosion, involvement of sinus tympani, facial recess, aditus ad antrum, or mastoid
- Restricts diffusion on DWI MR
- Associated granulation tissue or scar may enhance

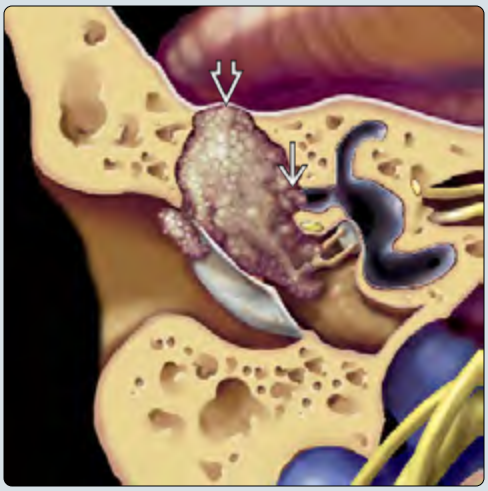
TOP DIFFERENTIAL DIAGNOSES

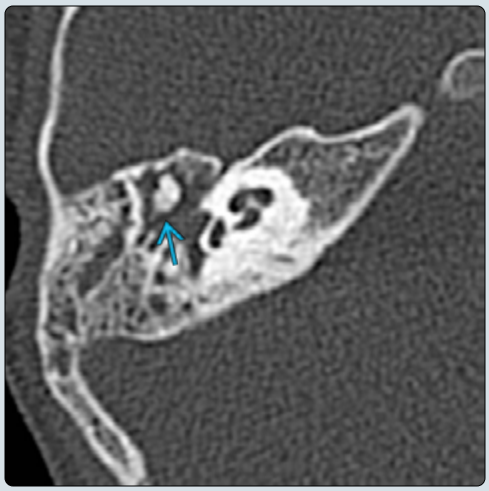
- Congenital cholesteatoma
- Chronic otitis media with ossicular erosion
- Acute coalescent otomastoiditis with abscess
- Langerhans cell histiocytosis
- Rhabdomyosarcoma

CLINICAL ISSUES

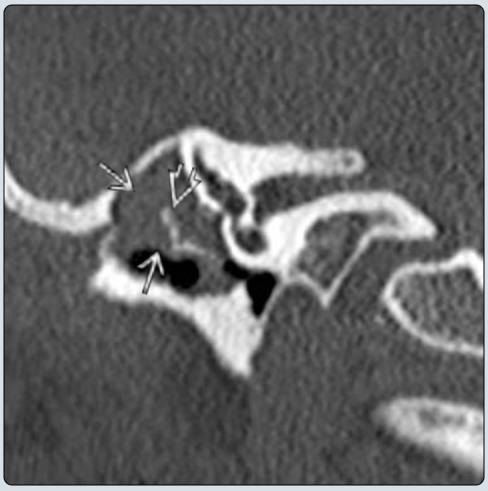
- Recurrent or chronic ME infections with TM perforation or retraction pocket
- Conductive hearing loss
- Affects children & adults
  - Unusual in children < 4 years of age

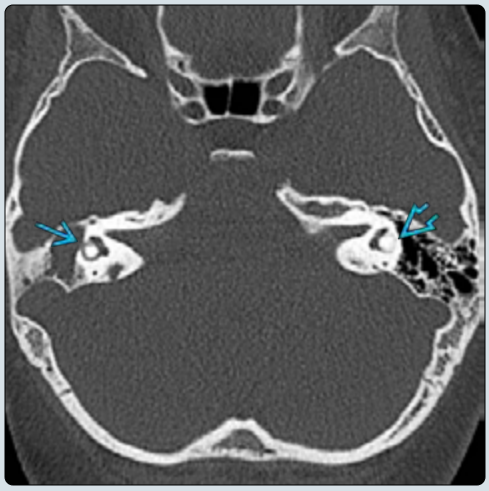
(Left) *Coronal graphic shows a large acquired cholesteatoma of the pars flaccida. Complications include erosion of the ossicles & lateral semicircular canal (SCC) ➡ & thinning of the tegmen tympani ➡.* **(Right)** *Axial bone CT in a 5 year old with chronic otitis media & conductive hearing loss shows complete opacification of the right middle ear cavity & mastoid air cells. There is truncation of the incus ⇨ without a definable long process.*





(Left) *Coronal bone CT in a 10 year old with chronic right otitis media & otorrhea shows a large PFC ➡ nearly filling the mesotympanum & epitympanum. The demineralized/partially eroded ossicles ➡ are deviated inferior & medial.* **(Right)** *Axial bone CT in the same child demonstrates focal dehiscence of the right lateral SCC ⇨ compared to the normal covering of the left lateral SCC ⇨. Also note the underdevelopment of the right mastoid air cells relative to the left, a common finding in chronic inflammatory disease.*





TERMINOLOGY

Abbreviations

- Secondary or acquired cholesteatoma (ACh)

Definitions

- Stratified, squamous epithelium-lined sac filled with exfoliated keratin debris in middle ear (ME)
  - "Attic" or Prussak space ACh = pars flaccida cholesteatoma (PFC)
  - Pars tensa ACh (PTC) in posterior mesotympanum, medial to ossicles

IMAGING

General Features

- Best diagnostic clue
  - PFC: Nonenhancing soft tissue mass in Prussak space with scutum erosion
    - ± tegmen tympani, lateral semicircular canal, facial nerve canal, or sigmoid sinus plate dehiscence
  - PTC: Nonenhancing erosive mass in posterior mesotympanum, medial to ossicles
    - ± ossicular erosion, involvement of sinus tympani, facial recess, aditus ad antrum, or mastoid
- Location
  - PFC: 80% of all cholesteatomas
    - 2° to tympanic membrane (TM) perforation or retraction pocket in anterior superior pars flaccida portion of TM (also called Shrapnell membrane)
  - PTC: 15% of all cholesteatomas
    - Secondary to TM perforation or retraction pocket in inferior pars tensa portion of TM

CT Findings

- Bone CT
  - PFC
    - Soft tissue mass in Prussak space, lateral to malleus head; scutum erosion characteristic
    - Ossicular erosion in 70%: Most commonly involves incus long process, less commonly incus body & malleus head
    - ± posterolateral extension to aditus ad antrum & mastoid antrum or inferiorly to posterior ME recesses
    - May also erode lateral semicircular canal, facial nerve canal, tegmen tympani, &/or sigmoid sinus plate
  - PTC
    - Erosive mass in posterior mesotympanum medial to ossicles
    - ± sinus tympani, facial recess, aditus ad antrum, &/or mastoid involvement
    - Ossicular erosion common (90%): Especially medial aspect of incus long process, stapes suprastructure, & malleus manubrium

MR Findings

- ↓ T1, mildly ↑ T2 (< than trapped secretions) ME mass
- PFC & PTC do not enhance
- Associated granulation tissue or scar may enhance
- If tegmen tympani dehiscent, coronal may show dural enhancement adjacent to bony defect
- Restricted diffusion on DWI

Imaging Recommendations

- Noncontrast bone CT: Axial & coronal
- Coronal T1 C+ MR useful adjunct when cephalocele, intracranial extension, or intracranial infection suspected

DIFFERENTIAL DIAGNOSIS

Congenital Cholesteatoma

- Well-defined ME mass behind intact TM

Chronic Otitis Media With Ossicular Erosion

- Noncholesteatomatous ossicular erosion

Acute Coalescent Otomastoiditis With Abscess

- Rim-enhancing fluid collection adjacent to opacified mastoid air cells = intracranial or extracranial abscess

Langerhans Cell Histiocytosis

- Enhancing soft tissue mass with punched-out bone destruction

Rhabdomyosarcoma

- Soft tissue mass with variable contrast enhancement & permeative bone destruction

PATHOLOGY

General Features

- Etiology
  - TM retraction or perforation → accumulation of stratified squamous epithelial cells in ME → mass-like keratin ball
  - Gradually increasing, ± erosion of adjacent bone
  - Inflammation may cause further bone erosion

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Recurrent or chronic ME infections with TM perforation or retraction pocket
  - Conductive hearing loss (CHL)
  - ME mass with TM perforation on otoscopy

Demographics

- Unusual in children < 4 years of age

Treatment

- Surgical excision, mastoidectomy, & ossicular chain reconstruction if needed

DIAGNOSTIC CHECKLIST

Image Interpretation Pearls

- When ME & mastoid opacified, difficult to differentiate effusion from ACh
- Presence of ossicular erosion favors ACh (but may also occur in noncholesteatomatous chronic otitis media)

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Acute Otomastoiditis With Abscess

KEY FACTS

TERMINOLOGY

- Acute coalescent (or confluent) otomastoiditis (ACOM)
- ACOM definition: Acute middle ear-mastoid infection with progressive bony resorption & demineralization due to intramastoid empyema

IMAGING

- Bone CT findings
  - Opacified mastoid cells
  - Erosion of mastoid cortex ± trabecula (confluent mastoiditis)
- Contrast-enhanced CT or MR findings
  - **Subperiosteal abscess**: Periauricular fluid collection
  - **Bezold abscess**: Walled-off pus in & around sternocleidomastoid muscle
  - **Middle cranial fossa abscess** (epidural or temporal lobe abscess)
  - **Posterior fossa abscess** (epidural or cerebellar abscess)
  - **Dural sinus thrombosis**: Adjacent venous filling defect

TOP DIFFERENTIAL DIAGNOSES

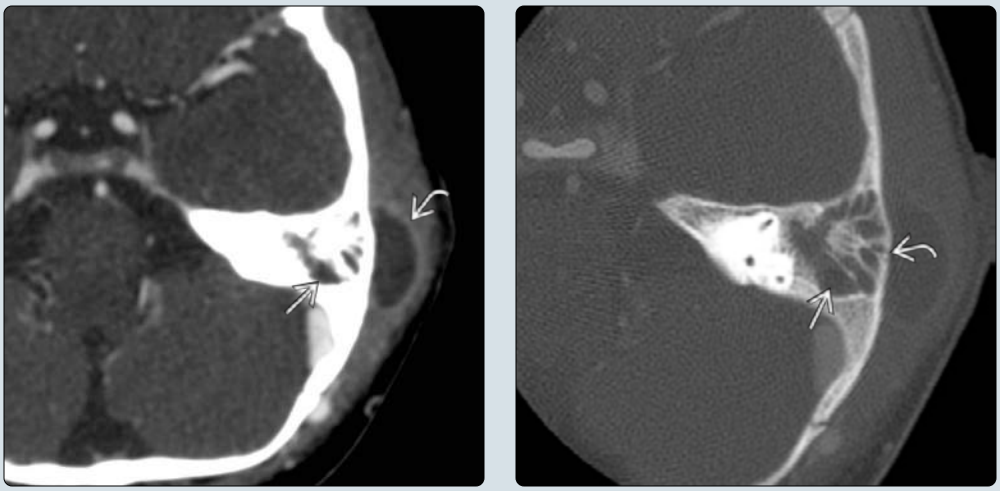
- Acquired cholesteatoma
- Apical petrositis
- Langerhans cell histiocytosis
- Rhabdomyosarcoma

PATHOLOGY

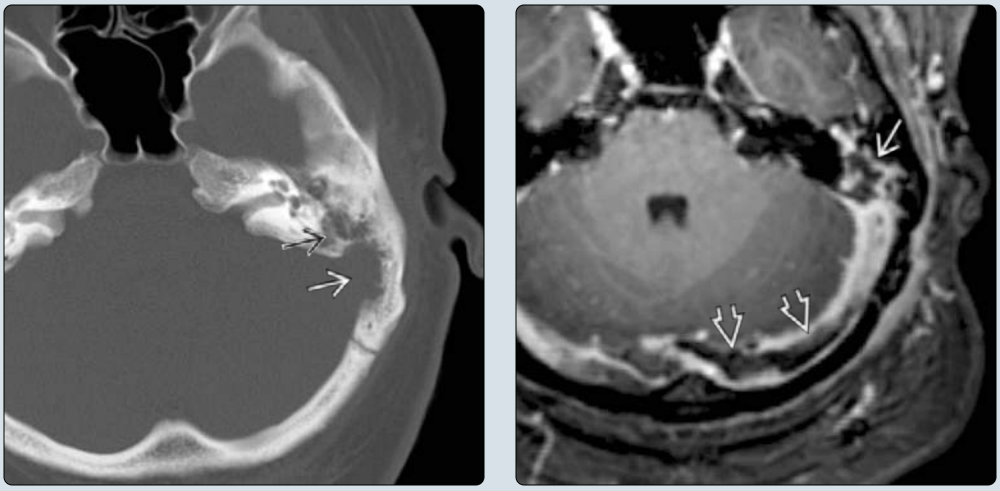
- Common pathophysiology
  - Granulation tissue or cholesteatoma blocks aditus ad antrum & prevents mastoid air cell drainage
- Less common pathophysiology
  - Mastoid cortex remains intact with septic thrombophlebitis of emissary veins seeding periosteum

CLINICAL ISSUES

- Young child with 1 day to 1 week history of otalgia, posterior auricular swelling, fever, & otorrhea



(Left) *Axial CECT in an 8-month-old girl shows a rim-enhancing fluid collection superficial to opacified left mastoid air cells, consistent with a subperiosteal abscess. There is edema of the surrounding soft tissues.* **(Right)** *Axial bone CT in the same patient shows opacification of the mastoid air cells without septal destruction. Subtle bony dehiscence is suggested deep to the abscess. However, bony destruction need not be present to establish a diagnosis of acute mastoiditis.*



(Left) *Axial NECT (bone window) in a 17-year-old boy shows complete opacification of the left mastoid air cells & middle ear cavity with extensive destruction of the mastoid septa & medial wall, consistent with acute coalescent otomastoiditis.* **(Right)** *Axial FS T1 C+ SPGR MR in the same patient shows extensive intramastoid enhancement & a large, hypointense filling defect in the left transverse sinus, consistent with secondary venous thrombosis.*

TERMINOLOGY

Abbreviations

- Acute coalescent (or confluent) otomastoiditis (ACOM)
- Acute mastoiditis (AM)
- Acute otitis media (AOM)

Synonyms

- Coalescent (or confluent) otomastoiditis with abscess

Definitions

- AM: Active infection in mastoid without destruction of mastoid septations or cortex
- ACOM: Acute middle ear-mastoid infection with progressive bony resorption & demineralization due to intramastoid empyema
- ACOM + abscess: Coalescent mastoiditis with resultant **intracranial** or **extracranial** abscess

IMAGING

General Features

- Best diagnostic clue
  - Rim-enhancing fluid collection adjacent to opacified mastoid cells with eroded cortex
- Location
  - Abscess location depends on direction of mastoid cortical dehiscence
    - **Lateral mastoid wall**
      - Postauricular abscess (cortical bone thin here)
      - Pre- or periauricular abscess
    - **Inferior mastoid wall**
      - Mastoid tip → Bezold abscess
      - Other inferior mastoid cortical dehiscence → trans-spatial abscess
    - **Tegmen mastoideum** → temporal lobe abscess
    - **Medial mastoid wall** → epidural or cerebellar abscess

CT Findings

- CECT
  - **Subperiosteal abscess**
    - Periauricular fluid collection
      - Thick, enhancing lateral wall represents inflamed periosteum
  - **Bezold abscess**
    - Walled-off pus in & around sternocleidomastoid muscle
  - **Middle cranial fossa abscess** (epidural or temporal lobe abscess)
    - Epidural or intratemporal lobe rim-enhancing fluid
  - **Posterior fossa abscess** (epidural or cerebellar abscess)
    - Epidural or intracerebellar rim-enhancing fluid
  - **Dural sinus thrombosis**
    - Filling defect within venous sinus
- Bone CT
  - Middle ear & mastoid opacification
  - Variable trabecular & cortical erosions (confluent disease)
  - Subtle to obvious cortical dehiscence deep to abscess
    - Lateral mastoid cortex → subperiosteal abscess
    - Mastoid tip cortex → Bezold abscess
    - Tegmen mastoideum cortex → epidural or temporal lobe abscess
    - Medial mastoid cortex → epidural or cerebellar abscess

MR Findings

- T2WI
  - High signal fills middle ear & mastoid
    - Complete opacification more common in children (90%)
  - High signal fluid in epidural or parenchymal abscess
- DWI
  - Restricted diffusion in abscess
- T1WI C+
  - Variable enhancement of middle ear & mastoid
    - More common in children (90%)
  - Perimastoid dural enhancement
    - More common in children (80%)
  - Rim-enhancing pus in subperiosteal, epidural, or brain parenchymal abscess
    - Subperiosteal abscess more common in children (50%)
- MRV
  - May show dural sinus thrombosis (DST)
    - Focus of diminished or absent vascular signal on flow-sensitive sequences

Imaging Recommendations

- Best imaging tool
  - Temporal bone CT defines bony changes (confluence, cortical dehiscence)
    - CECT will define most infectious complications
  - Enhanced temporal bone MR more sensitive for intracranial complications (DST, meningitis, subdural empyema, abscess)
- Protocol advice
  - Thin-section SE T1 MR with FS gives best evaluation of bone
  - 3D gradient-echo T1 C+ allows optimal delineation of epidural abscess & venous sinus thrombosis
  - Non-EPI DWI techniques may decrease artifact

DIFFERENTIAL DIAGNOSIS

Acquired Cholesteatoma

- Clinical: Retraction or rupture of tympanic membrane; may be superinfected
- Imaging: CT shows erosive mass with poor enhancement
- May lead to ACOM & associated complications

Apical Petrositis

- Clinical: CNV6 palsy, retroauricular pain, AOM
- Imaging: CT shows coalescent changes in petrous apex
  - T1-weighted C+ MR shows enhancing meninges & focal walled-off fluid in petrous apex
- Usually no associated intracranial abscess

Langerhans Cell Histiocytosis

- Clinical: Child with draining ear & periauricular mass
- Imaging: Extensive, often bilateral, mastoid destruction with homogeneously enhancing soft tissue

- Look for additional lesions (e.g., punched-out well-defined lytic calvarial lesions)

Rhabdomyosarcoma

- Clinical: Neurologic deficits common, including CNVII palsy
- Imaging: CT shows extensive bone lysis & permeation, intracranial extension
  - T1-weighted C+ MR shows variably enhancing soft tissue mass emanating from middle ear

PATHOLOGY

General Features

- Etiology
  - Inflammation, granulation tissue, or cholesteatoma block aditus ad antrum & prevent mastoid air cell drainage
  - Local hyperemia-acidosis creates enzymatic resorption of trabeculae (confluent mastoiditis)
  - Subtle or obvious cortical dehiscence conveys infection into adjacent tissues
  - Less common pathophysiology: Mastoid cortex remains intact with septic thrombophlebitis of emissary veins seeding periosteum
- Macewen triangle
  - Surgical access point to mastoid antrum at posterosuperior EAC
  - Weak bone, loose periosteum allow breakout of infection in postauricular location

Gross Pathologic & Surgical Features

- Pus in mastoid, adjacent abscess cavity
- Granulation tissue or cholesteatoma occasionally identified in middle ear-mastoid

Microscopic Features

- *Streptococcus* species most common
- Polymicrobial aerobes & anaerobes less common

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Otalgia (ear pain)
  - Other signs/symptoms
    - Fever
    - Otorrhea (ear drainage)
    - Postauricular pain & swelling
    - Lateralized auricle (ear pushed outward by abscess)
    - Conductive > sensorineural hearing loss
    - ↑ WBC, ↑ ESR
- Clinical profile
  - Young child with 1 day to 1 week history of otalgia, postauricular swelling, fever, & otorrhea
  - 35-70% of patients have received antibiotics for AOM
  - Postauricular edema (Griesinger sign) common in uncomplicated acute mastoiditis (85%)

Demographics

- Age
  - Infants & young children
  - Older age group affected when ACOM results from acquired cholesteatoma
- Epidemiology
  - 46% of children have > 2 episodes AOM by age 3
  - 0.24% of patients with AOM develop ACOM

Natural History & Prognosis

- Isolated extracranial subperiosteal abscess
  - Excellent prognosis with prompt therapy
  - Worse if prior incomplete antibiotic therapy, virulent organism, or immunocompromised host
- Intracranial abscess (temporal lobe most common)
  - Worse prognosis
  - If concomitant complications, even worse prognosis
    - Venous sinus thrombosis
    - Epidural abscess or subdural empyema

Treatment

- Intravenous antibiotics ± tympanocentesis with myringotomy tube placement
- Surgical treatment
  - I&D of extracranial subperiosteal abscess
  - Simple mastoidectomy typically reserved for patients that fail initial conservative management
  - Radical mastoidectomy if cholesteatoma present

DIAGNOSTIC CHECKLIST

Consider

- Seek other complications of ACOM
  - Temporal bone findings (T1 C+ MR)
    - Facial nerve paralysis: Enhancing CNVII
    - Labyrinthitis: Enhancement within membranous labyrinth
    - Apical petrositis: Enhancing apical air cells on MR
  - Intracranial findings (T1 C+ MR)
    - Epidural or brain abscess
    - Subdural empyema, meningitis ± DST

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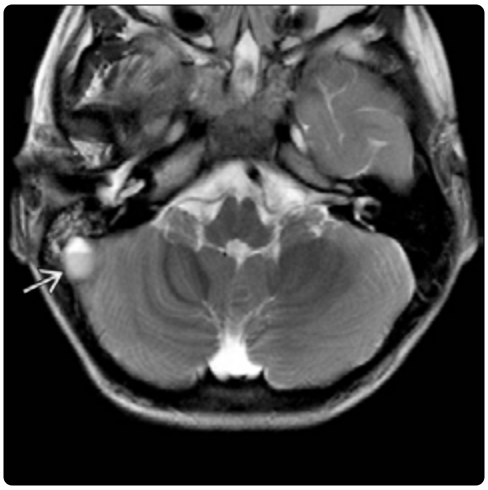
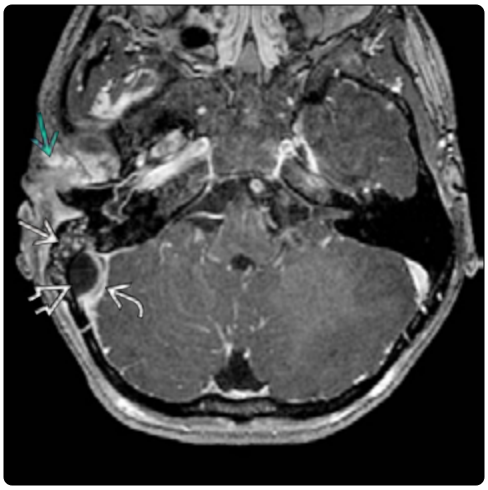
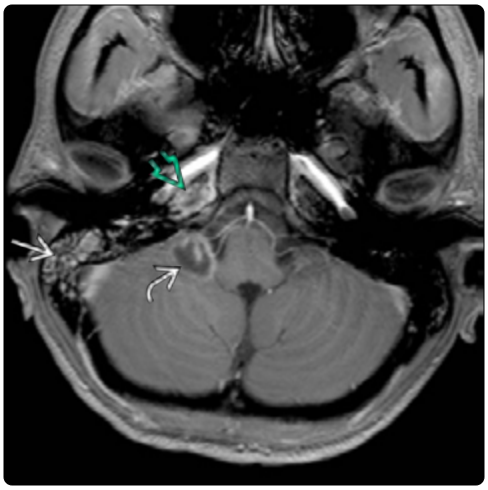
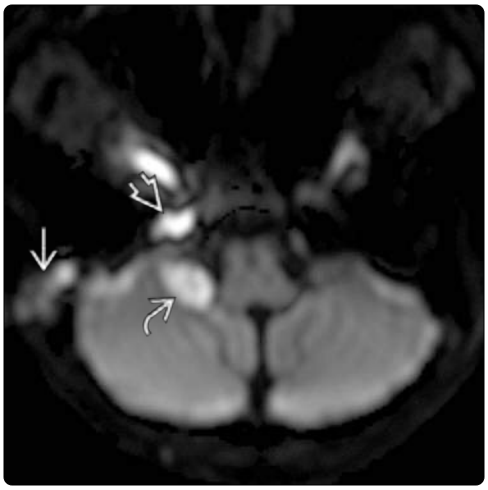


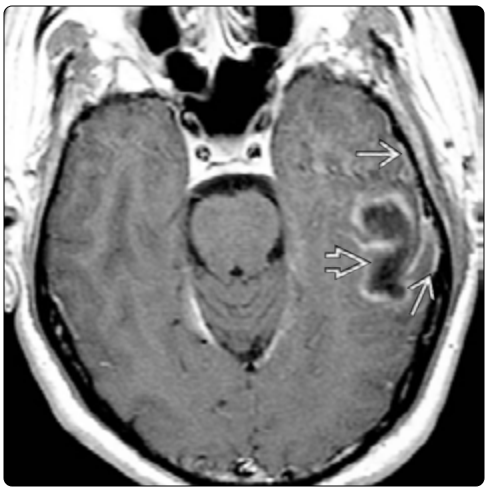


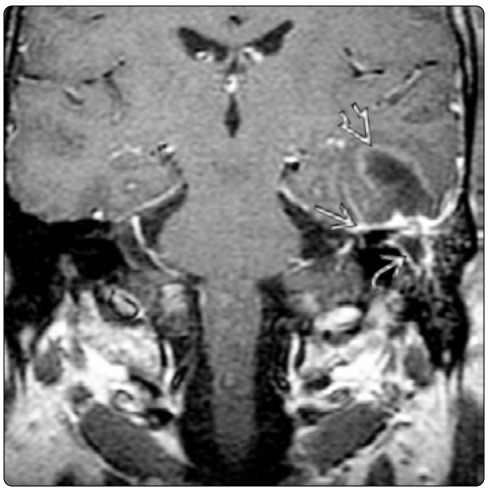
(Left) *Axial T2 MR in this 7-year-old boy with acute mastoiditis shows an epidural abscess in the right jugular fossa. Note the fluid-fluid level, which is most frequently due to blood products.* **(Right)** *Axial FS T1 C+ SPGR MR in the same patient shows enhancement of the right mastoid air cells with right periauricular soft tissue swelling. There is an epidural abscess in the right sigmoid fossa. The dura is medially displaced with near complete occlusion of the right sigmoid sinus.*





(Left) *Axial T1 C+ MR in a 14-year-old boy with right-sided acute mastoiditis & petrous apicitis shows extensive mastoid & petrous apex enhancement with an intraparenchymal abscess involving the right flocculus.* **(Right)** *Axial DWI MR in the same patient demonstrates restricted diffusion in the right mastoid & petrous apex as well as the right flocculus intraparenchymal abscess.*





(Left) *Axial T1 C+ MR in an adult with decreased consciousness & fever demonstrates a bilobed, rim-enhancing temporal lobe abscess along with meningeal enhancement secondary to meningitis.* **(Right)** *Coronal T1 C+ MR in the same patient reveals acute otomastoiditis with direct continuity between the mastoid & temporal lobe abscesses. Associated meningitis is again noted.*

CHARGE Syndrome

KEY FACTS

TERMINOLOGY

- **CHARGE**
  - **C**oloboma
  - **H**eart anomaly
  - **A**tresia of choanae
  - **R**etardation: Mental & somatic development
  - **G**enital hypoplasia
  - **E**ar abnormalities
- Major signs: Coloboma, choanal atresia, semicircular canal (SCC) hypoplasia/aplasia, cranial nerve (CN) involvement
- Minor signs: Hindbrain, external/middle ear, cardiac/esophageal malformations, hypothalamo-hypophyseal dysfunction, mental retardation

IMAGING

- Choanal atresia, coloboma (variable), cleft lip/palate
- Hypoplastic vestibule & hypoplastic/absent SCC
- Mildly flattened cochlear apical ± middle turns + thickened modiolus or single cochlear turn/hypoplasia
- Stenotic/atretic cochlear nerve canal
- Stenotic/atretic oval window & overlying anomalous tympanic segment of CNVII
- Large emissary veins, hypoplasia of basiocciput, basilar invagination, & vertebral anomalies
- Hypoplastic pons, uplifted vermis ± cerebellar malformation
- CN hypoplasia/aplasia (mainly CNI, VII, & VIII)

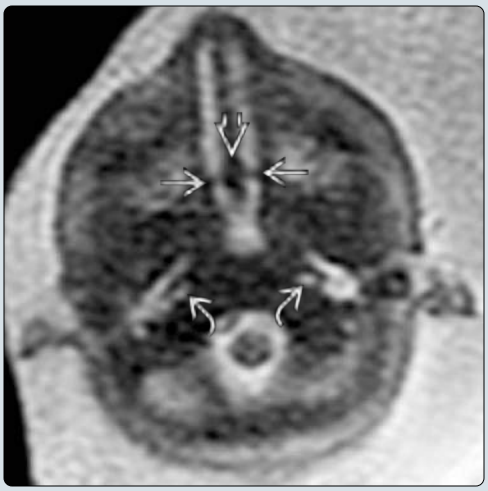
TOP DIFFERENTIAL DIAGNOSES

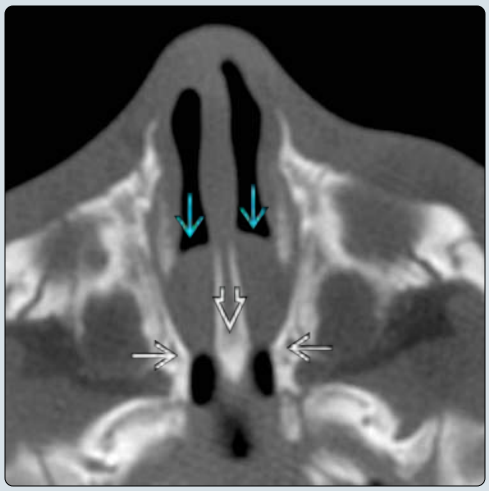
- Kallmann syndrome
- VACTERL association
- Branchiootorenal syndrome

PATHOLOGY

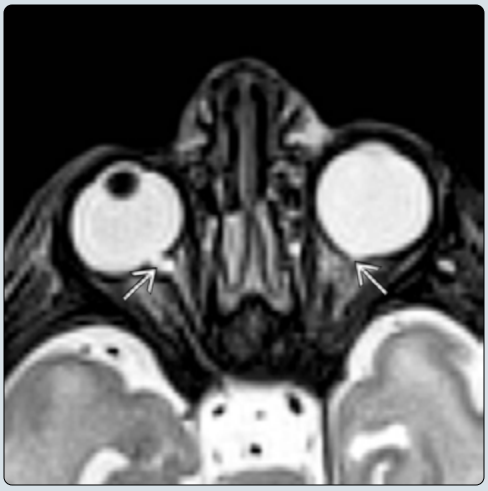
- *CHD7* mutation in 60%
- Highly predictive of CHARGE: Cup-shaped pinna, agenesis/hypoplasia of SCC, arrhinencephaly

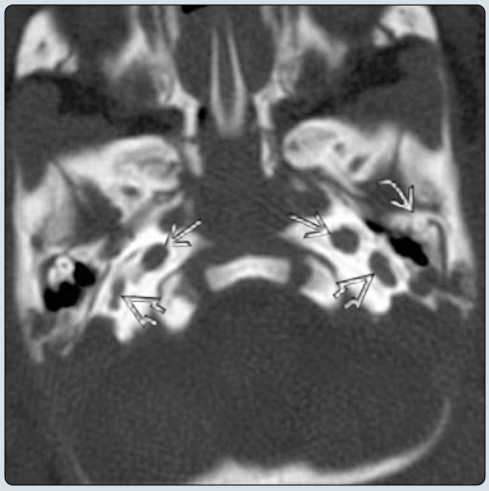
(Left) *Axial SSFSE T2 fetal MR at 32 weeks gestation shows bilateral choanal atresia with linear hypointensities extending from the thickened vomer to the lateral nasal walls. A single cochlear turn is seen bilaterally, & the cochleae appeared isolated from the internal auditory canals.* **(Right)** *Axial bone CT in the same neonate shows medial deviation of the lateral nasal walls & a thickened vomer with bilateral bony & membranous choanal atresia. Fluid & secretions layer anterior to the obstructing membranes.*





(Left) *Axial T2 MR in the same neonate reveals right microphthalmia & bilateral colobomas. A CHD7 gene mutation was found, confirming CHARGE syndrome.* **(Right)** *Axial bone CT in the same child confirms incomplete cochlear partitioning, hypoplastic vestibules, & absent semicircular canals. The ossicles are dysmorphic, & the left malleus is fused to the attic. It is unusual to see the ocular, ear, & nasal findings of CHARGE all in the same patient.*





KEY FACTS

TERMINOLOGY

- Branchiootorenal syndrome
  - Deafness & ear anomalies
  - Branchial anomalies/preauricular pits
  - Renal abnormalities

IMAGING

- Face & neck US/CT findings
  - Branchial cleft cyst/fistula
  - Variable, asymmetric micrognathia
- Temporal bone CT findings
  - Dilated eustachian tubes
  - External ear: Variable stenosis/atresia
  - Middle ear: Dysmorphic with fused, malformed ossicles
  - Cochlea: Tapered basal turn, hypoplastic & offset middle/apical turns (unwound appearance)
  - Semicircular canal anomalies
  - Dilated, bulbous vestibular aqueduct
  - Flared internal auditory canal; anomalous CNVII canal
- Abdominal US/CT/MR findings
  - Renal cysts, dysplasia, agenesis

TOP DIFFERENTIAL DIAGNOSES

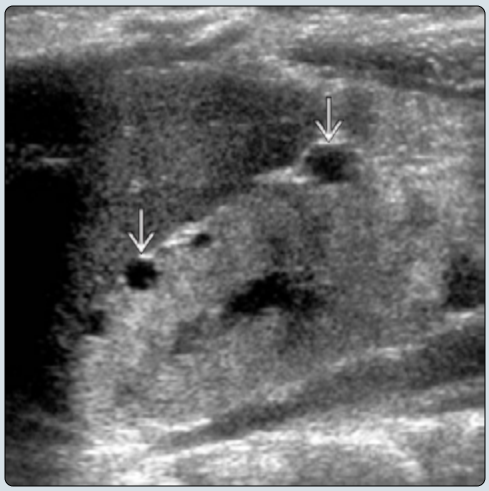
- Branchiootic syndrome
- Otofaciocervical syndrome
- Nonsyndromic congenital external ear & middle ear malformation

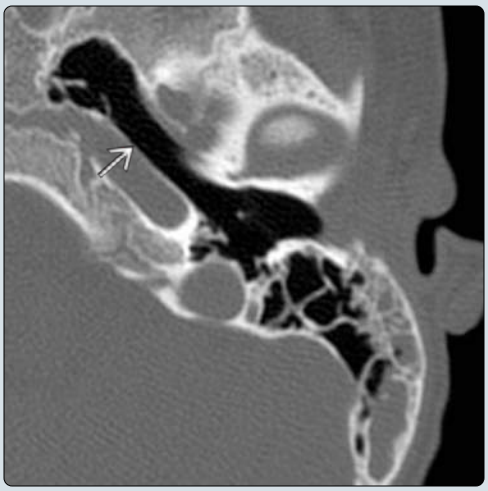
PATHOLOGY

- Autosomal dominant syndrome
  - BOR 1: 8q13.3 locus, *EYA1* gene mutation
  - BOR 2: 19q13.3 locus, *SIX* gene mutation

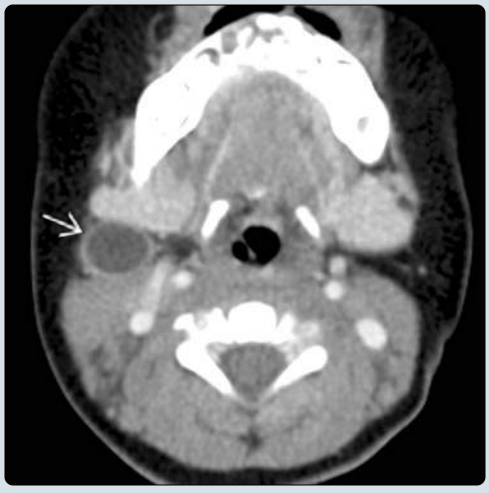
CLINICAL ISSUES

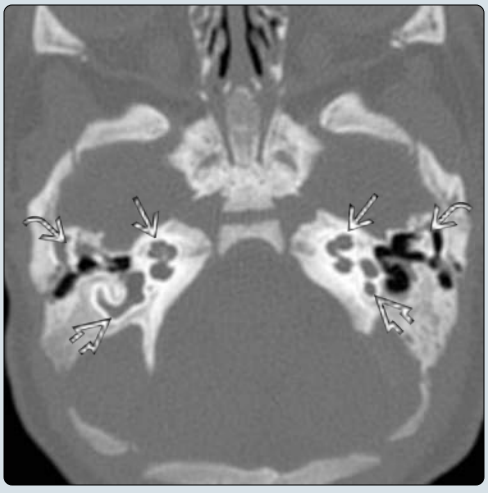
- Clinical presentation
  - Hearing loss (sensorineural, conductive, mixed) (~ 98%)
  - Preauricular tag or pit (~ 84%)
  - Branchial anomalies (~ 70%)
  - Renal anomalies (~ 40%)





(Left) *Longitudinal US of a 4-month-old boy presenting with Potter syndrome shows a solitary echogenic kidney with numerous peripheral cysts ➡. This patient also had a branchial cleft cyst & microtia, consistent with branchiootorenal (BOR) syndrome.* **(Right)** *Axial temporal bone CT in a 4-year-old boy with BOR syndrome shows dilation of an anomalous eustachian tube ➡ terminating in the sphenoid bone.*





(Left) *Axial CECT in a 5-month-old boy with preauricular pits & a family history of renal & ear anomalies shows a round hypodense lesion ➡ in a typical location for a 2nd branchial cleft cyst. The patient also had characteristic ear anomalies.* **(Right)** *Axial bone CT in the same patient shows unwound or offset hypoplastic middle & apical cochlear turns ➡ & deficiency of the right cochlear modiolus. The posterior semicircular canals (SCC) ➡ & ossicles ➡ are malformed. The CT findings are characteristic of BOR syndrome.*

Treacher Collins Syndrome

KEY FACTS

TERMINOLOGY

- Treacher Collins syndrome (TCS): Craniofacial malformation with down-slanting palpebral fissures, micrognathia, zygomatic & malar hypoplasia, microtia/anotia
- Nager syndrome: TCS like + limb anomalies

IMAGING

- Temporal bone
  - External auditory canal (EAC): Stenosis/atresia
  - ↓/absent mastoid pneumatization
  - Hypoplastic/atretic middle ear space
  - Malformed or absent ossicles ± fixation
  - Oval window stenosis/atresia
  - Facial nerve canal anomalies/dehiscence
  - Normal or malformed cochlea (flattened turns)
  - Normal or malformed lateral semicircular canal ± vestibule
- Face/orbits: Relatively symmetric micrognathia, zygomatic/malar hypoplasia, coloboma

TOP DIFFERENTIAL DIAGNOSES

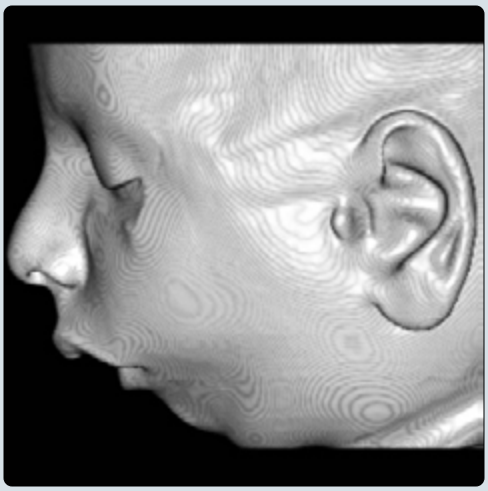
- Bilateral facial microsomia
- Nonsyndromic congenital external & middle ear malformation
- Branchiootorenal syndrome

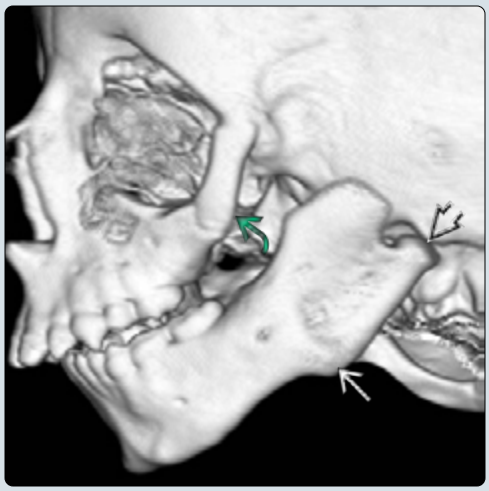
PATHOLOGY

- Autosomal dominant >> recessive, phenotypic variability & genetic heterogeneity, "ribosomopathy"

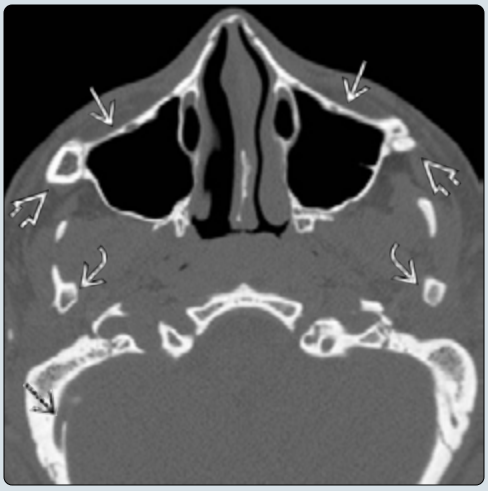
CLINICAL ISSUES

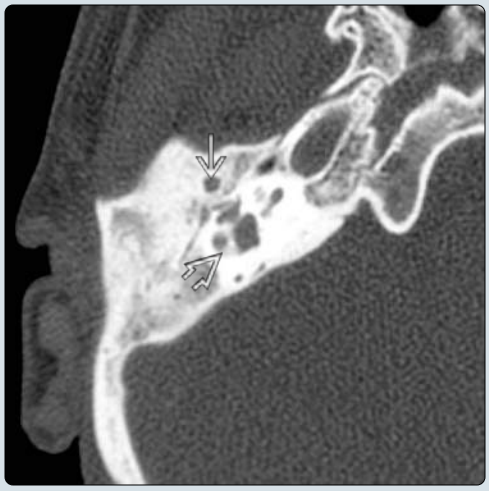
- Airway obstruction, deafness
- Down-slanting palpebral fissures (100%)
- Nager limb anomalies: Malformed thumbs, radial hypoplasia/aplasia, radioulnar synostosis
- Treatment: Airway support, reconstructive surgery, hearing aids, developmental support





(Left) *Lateral view of a CT with 3D soft tissue surface-rendered reformation in an infant with Nager syndrome shows micrognathia, malar flattening, mildly low-set ears, & external auditory meatus atresia.* **(Right)** *Lateral view 3D bone CT in the same infant with Nager syndrome shows micrognathia with an obtuse mandibular angle & marked hypoplasia of the neck & condyle of the mandible. There is hypoplasia of the midface & zygomatic complex with absence of the zygomatic arch. EAC atresia is also noted.*





(Left) *Axial bone CT in a 13-year-old girl with TCS shows zygomatic complex hypoplasia with posteriorly slanted maxillae, absent zygomatic arches, & hypoplastic mandibular condyles. There is EAC atresia, absent mastoid pneumatization, & an enlarged mastoid emissary vein.* **(Right)** *Axial temporal bone CT in a 16-year-old girl with TCS shows EAC atresia, absent middle ear space & ossicles, ventrally placed descending facial nerve canal, & a malformed lateral semicircular canal (SCC) with a small bone island.*

KEY FACTS

TERMINOLOGY

- Pierre Robin sequence (PRS)
- PRS triad: Micrognathia, glossoptosis, respiratory distress

IMAGING

- Bilateral, usually symmetric micrognathia
- Glossoptosis: Elevated, posteriorly displaced tongue
- Posterior U-shaped cleft palate
- Additional features depend on syndromic etiology
- Temporal bone
  - EAC: Normal, stenotic, or atretic [e.g., Treacher Collins syndrome (TCS)]
  - Middle ear & mastoid: Normal or hypoplastic ± opacification
  - Ossicles: Normal, mildly malformed [e.g., stapes in velocardiofacial syndrome (VCFS)], or severely malformed ± fixation (e.g., TCS)
  - Inner ear: Normal or malformed (e.g., small semicircular canal bone island/anlage anomaly in VCFS & TCS)

TOP DIFFERENTIAL DIAGNOSES

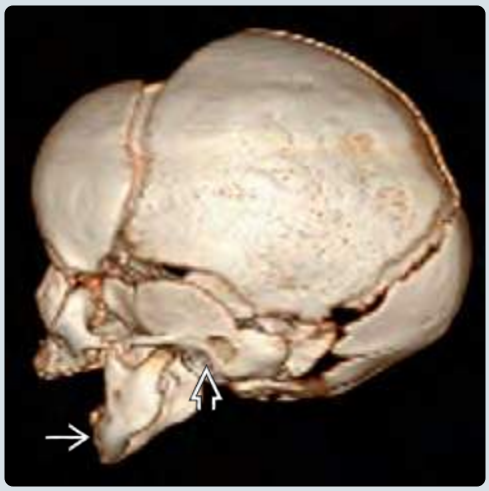
- Stickler & related syndromes (18% of PRS)
- VCFS (~ 7% of PRS)
- TCS (~ 5% of PRS)

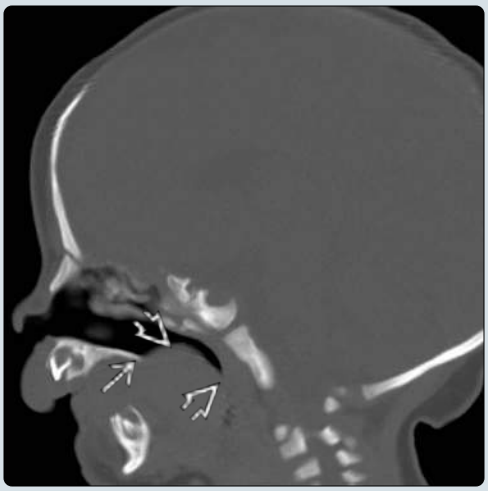
PATHOLOGY

- Primary micrognathia → glossoptosis → failure of palatal shelf elevation & fusion
- Collagen (COL) gene mutations: Stickler syndromes
- 22q11.2 deletion: VCFS

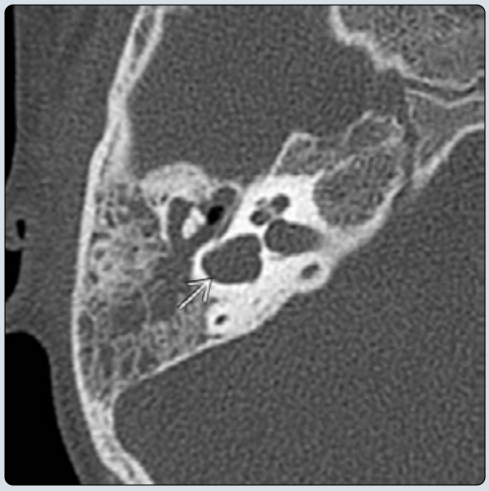
CLINICAL ISSUES

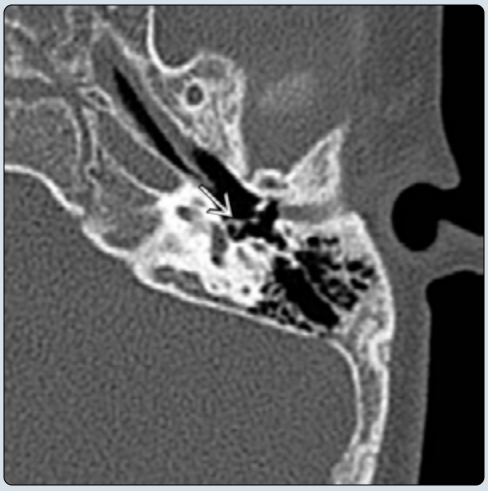
- Feeding & breathing difficulties, failure to thrive
- Stickler: Progressive myopia, joint degeneration
- VCFS: Cardiac anomalies, adenoidal hypoplasia, velopharyngeal insufficiency, medial deviation of cervical internal carotid arteries, learning difficulties
- TCS: Malar flattening, downslanting palpebral fissures, coloboma





(Left) *Lateral 3D CT reformation of a 3-week-old girl with Pierre Robin sequence (PRS) shows moderate, symmetric micrognathia. Note that the zygomatic arch & EAC are present.* **(Right)** *Sagittal CT reconstruction in the same patient reveals a shortened hard palate & glossoptosis (abnormal downward or backward displacement of the tongue). The tongue, which protrudes above & behind the palate, obstructs the naso- & oropharynx resulting in difficulty with breathing & feeding.*





(Left) *Axial bone CT in a 5-year-old boy with velocardiofacial syndrome (VCFS) shows an anlage anomaly where the lateral semicircular canal & vestibule form a single globular space without a bone island. There is diffuse opacification of the middle ear space & mastoid air cells.* **(Right)** *Axial bone CT in a 7-year-old girl with VCFS & hearing loss shows mild thickening of the anterior crus of the stapes & mildly reduced mastoid pneumatization.*

Thyroglossal Duct Cyst

KEY FACTS

TERMINOLOGY

- TGDC: Cystic remnant of embryologic thyroglossal duct (TGD)

IMAGING

- Best diagnostic clue: Round or ovoid midline suprahyoid or midline/paramidline infrahyoid cystic neck mass
- Suprahyoid neck: ~ 20-25%, typically midline
- At hyoid bone: ~ 50%
- Infrahyoid neck: ~ 25%, midline or paramidline
  - Embedded in strap muscles: Claw sign
- ± wall enhancement, soft tissue stranding if infected

TOP DIFFERENTIAL DIAGNOSES

- Dermoid or epidermoid
- Lingual thyroid
- Lymphatic malformation
- 4th branchial apparatus anomaly
- Thymic cyst

PATHOLOGY

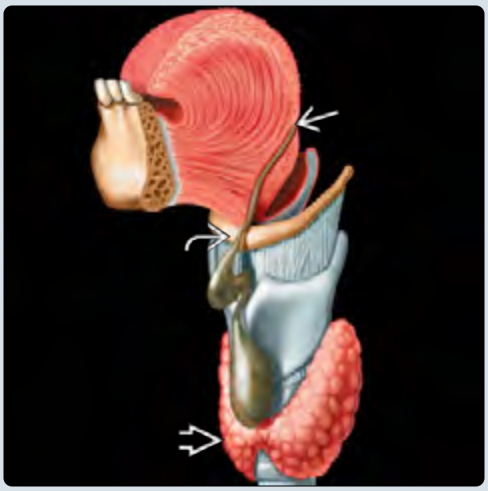
- Failure of involution of TGD + persistent secretion of epithelial cells lining duct → TGDC
- Lies anywhere along TGD route of thyroid anlage descent from foramen cecum at tongue base to thyroid bed in infrahyoid neck

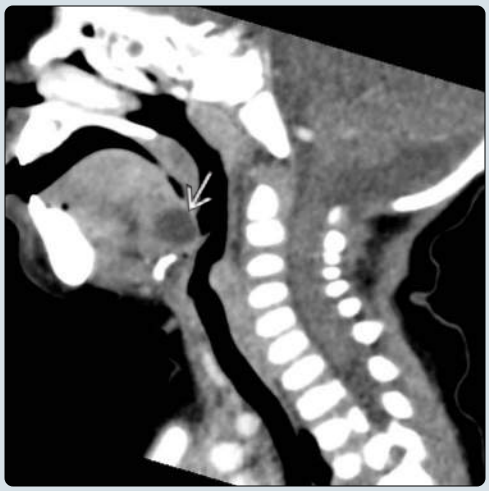
CLINICAL ISSUES

- Most common congenital neck lesion
- Treatment: Sistrunk procedure (excision of cyst, tract, & midline hyoid bone) → ↓ recurrences

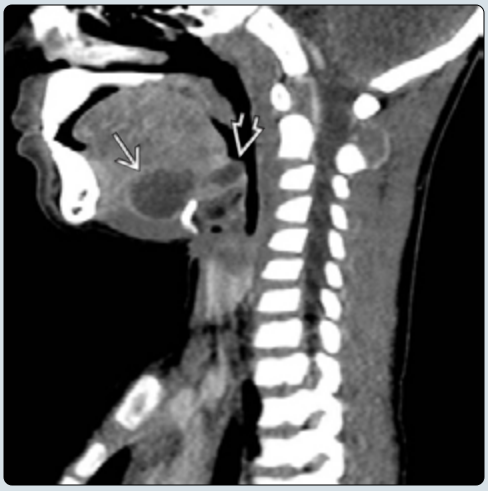
DIAGNOSTIC CHECKLIST

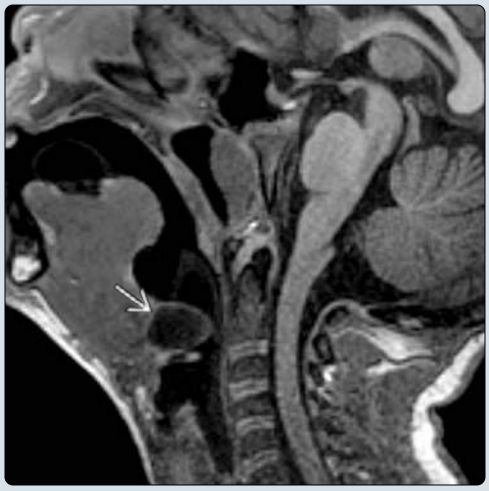
- Relationship to hyoid bone important to note: Suprahyoid, hyoid, or infrahyoid in location
- Any nodularity or Ca^{2+} suggests associated thyroid carcinoma
- Confirm normal thyroid by ultrasound prior to TGDC or lingual thyroid resection





(Left) *Sagittal oblique graphic shows the potential sites of a thyroglossal duct cyst (TGDC) from the foramen cecum ➡ to the thyroid bed ➡. Note the close relationship of the midportion of the hyoid bone ➡ to this pathway. A cyst can occur anywhere along this tract.* **(Right)** *Sagittal CECT shows a well-defined, cystic-appearing mass ➡ at the midline base of the tongue. This mass was incidentally found on a CT performed to evaluate the extent of a deep neck infection (not shown). The mass was subsequently proven to be a TGDC.*





(Left) *Sagittal CECT in a child with sore throat & difficulty swallowing shows a lobulated rim-enhancing, fluid-attenuation mass ➡ in the midline sublingual space with a small posterior extension at the tongue base ➡. Histologically, this proved to be an inflamed TGDC.* **(Right)** *Sagittal T1 MR in a child with a recurrent TGDC demonstrates a cystic mass ➡ at the midline tongue base. Note the distortion of the tongue & superior aspect of the mass by the laryngeal mask airway used during anesthesia.*

TERMINOLOGY

Synonyms

- Thyroglossal duct cyst (TGDC)
- Thyroglossal duct remnant

Definitions

- Remnant of embryologic thyroglossal duct (TGD) found between foramen cecum at tongue base & thyroid bed in infrahyoid neck

IMAGING

General Features

- Best diagnostic clue
  - Anterior midline suprahyoid or midline/paramidline infrahyoid cystic neck mass
- Location
  - Suprahyoid neck: ~ 20-25%, typically midline
    - Base of tongue or within posterior floor of mouth
  - At hyoid bone: ~ 50%
    - Usually abutting anterior hyoid bone
    - May project into preepiglottic space
  - Infrahyoid neck: ~ 25%, midline or paramidline embedded in strap muscles
    - Further inferior TGDC more likely to be off-midline
- Size
  - Variable, usually 2-4 cm
- Morphology
  - Round or ovoid cyst

CT Findings

- CECT
  - Low-attenuation cystic midline neck mass with thin rim of peripheral enhancement
    - ± wall enhancement, soft tissue stranding if infected
    - Occasional septations
  - Paramidline infrahyoid TGDC embedded in strap muscles may show claw sign
  - < 1% contain associated thyroid carcinoma (usually papillary carcinoma)
    - Solid eccentric mass, often with Ca^{2+}, within cyst
    - May only be microscopic & not identifiable prospectively with imaging
    - Majority occur in adults, but may occur in teenagers
      - Youngest reported: 10 years old

MR Findings

- T1WI
  - Usually hypointense; hyperintense if proteinaceous fluid
- T2WI
  - Homogeneously hyperintense
- T1WI C+
  - Nonenhancing cyst
    - Rim enhancement if infected

Ultrasonographic Findings

- Anechoic or hypoechoic midline neck mass
  - ± internal echoes
    - May or may not be due to hemorrhage or infection
- Must image lower neck to prove presence of normal-appearing, bilobed thyroid

Imaging Recommendations

- Children: TGDCs have classic clinical presentation
  - Sonography only to confirm normal thyroid gland
  - CT or MR if infected or diagnosis uncertain
- Adults: CT or MR if cyst suprahyoid or infected
- Nuclear scintigraphy helpful if ectopic thyroid suspected

DIFFERENTIAL DIAGNOSIS

Oral Cavity Dermoid or Epidermoid

- Dermoid: Fat, fluid, or mixed
- Epidermoid: Fluid
- Submandibular space, sublingual space, or root of tongue
- Neither directly involves hyoid bone

Lingual Thyroid

- Solid round hyperattenuating mass on NECT

Lymphatic Malformation

- Unilocular or multilocular
- Focal or transspatial
- Fluid-fluid levels common secondary to hemorrhage
- Nonenhancing unless infected or part of combined venolymphatic vascular malformation

4th Branchial Apparatus Anomaly

- Left thyroid lobe abscess from tract to pyriform sinus

Thymic Cyst

- Congenital cyst at lateral infrahyoid neck at level of thyroid
- Close association with carotid sheath

Mixed Laryngocele

- Off-midline fluid- or air & fluid-containing mass
- Traces back to laryngeal origin
- Not embedded within strap muscles

Delphian Chain Necrotic Node

- Can be difficult to differentiate from infected TGDC
  - Rare in children

PATHOLOGY

General Features

- Genetics
  - Thyroid developmental anomalies often occur in same family
- Associated abnormalities
  - Thyroid agenesis, ectopia, or pyramidal lobe
  - Occasionally associated with carcinoma
    - Most commonly papillary carcinoma within TGDC
- Embryology/anatomy
  - TGD originates near foramen cecum at posterior 3rd of tongue
  - Thyroid anlage arises at base of tongue → descends around or through hyoid bone → descends along strap muscles → final position in thyroid bed, anterior to thyroid cartilage or cricoid cartilage
  - At 5-6 gestational weeks, TGD usually involutes; foramen cecum & pyramidal thyroid lobe may be left as normal remnants
  - Failure of TGD involution with persistent secretory activity of epithelial cells lining duct → TGDC

- TGDC or ectopic thyroid tissue may occur anywhere along TGD

Gross Pathologic & Surgical Features

- Smooth, benign-appearing cyst with tract to hyoid bone ± foramen cecum

Microscopic Features

- Cyst lined by respiratory or squamous epithelium
- Small deposits of thyroid tissue with colloid commonly associated
- ± thyroid carcinoma (papillary carcinoma most common)

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Midline or paramidline doughy, compressible, painless neck mass in child or young adult
  - Cyst elevates when tongue protrudes if TGDC located around hyoid bone
- Other signs/symptoms
  - Recurrent midline neck mass with upper respiratory tract infections or trauma
  - ± multiple prior incision & drainage procedures for "neck abscess"
  - Rarely, lingual TGDC may lead to airway obstruction in infants
  - Small lesion may be recognized as incidental finding on brain MR, especially at base of tongue

Demographics

- Age
  - < 10 years at presentation (up to 90%)
- Gender
  - M < F in uncommon familial forms
- Epidemiology
  - Most common congenital neck lesion
    - 90% of nonodontogenic congenital cysts
    - 3x as common as branchial cleft cysts
  - At autopsy > 7% of population will have TGD remnant somewhere along course of tract

Natural History & Prognosis

- Recurrent, intermittent swelling of mass, usually following minor upper respiratory infection
- Rapidly enlarging mass suggests either infection or differentiated thyroid carcinoma (< 1%)
  - 85% papillary carcinoma

Treatment

- Complete surgical resection: Sistrunk procedure → ↓ recurrence rate from 50% to < 4%
  - Tract to foramen cecum dissected free
  - Entire cyst & midline portion of hyoid bone resected
    - Even if imaging shows no obvious connection to hyoid bone
    - Exception: Low infrahyoid neck TGDC
- Isolated lingual TGDC may be treated endoscopically
- Prognosis excellent with complete surgical resection
- Recurrences (from incomplete resection) often complicated & lateral
  - ↑ risk of recurrence in patients with postoperative infection

DIAGNOSTIC CHECKLIST

Consider

- Relationship to hyoid bone important to note: Suprahyoid, hyoid, or infrahyoid in location
- Any nodularity or Ca^{2+} suggests associated thyroid carcinoma
- Image thyroid bed with ultrasound to confirm presence of normal thyroid gland prior to TGDC or lingual thyroid excision

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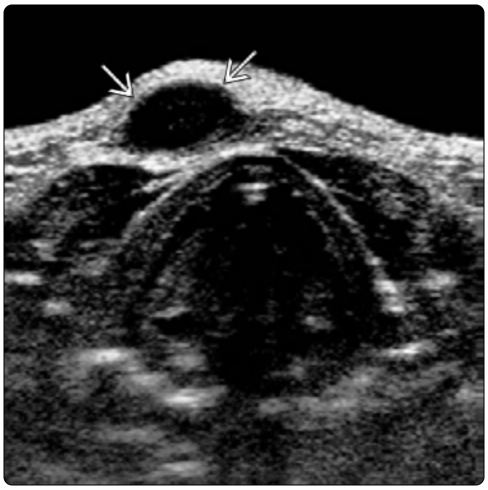
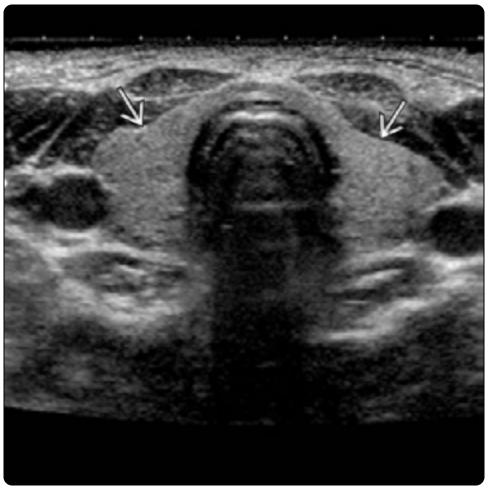
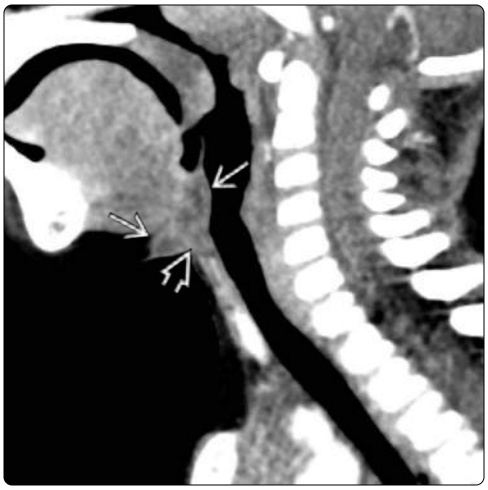
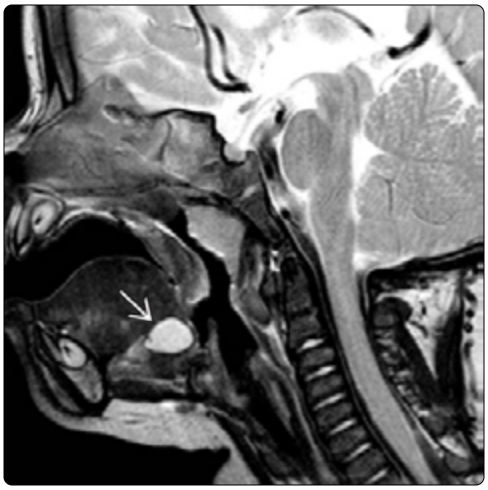


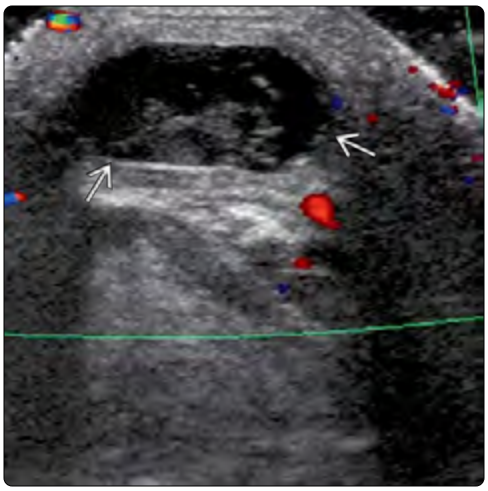


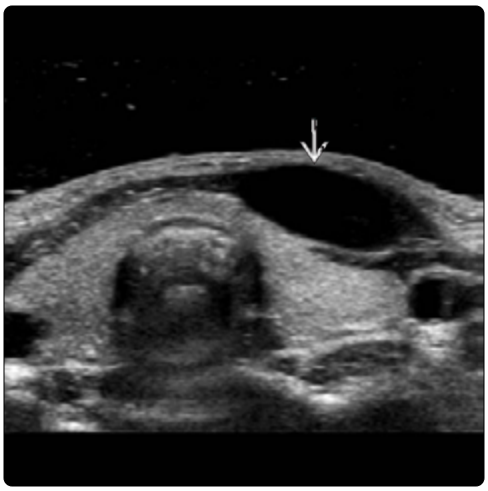
(Left) *Transverse ultrasound of the anterior neck shows a well-defined, subcutaneous, right paramidline, hypoechoic mass ➡ ventral to the strap muscles. This was surgically removed & confirmed to be a TGDC.* **(Right)** *Transverse ultrasound in a child with a suspected TGDC (not shown) demonstrates a normal-appearing, bilobed thyroid ➡ at the expected location in the lower anterior midline neck. It is important to document the presence of normal thyroid tissue in all patients imaged for the work-up of a TGDC.*





(Left) *Sagittal CECT in a child with a proven recurrence of a TGDC demonstrates a lobulated, heterogeneous mass ➡ in the midline suprahyoid neck. By imaging, this is indistinguishable from a postsurgical collection. Note the absence of a midline hyoid bone ➡, consistent with a prior Sistrunk procedure.* **(Right)** *Sagittal T2 MR in a child undergoing brain imaging for the work-up of seizures shows an incidental, well-defined, hyperintense mass ➡ at the midline tongue base, subsequently proven to be a TGDC.*





(Left) *Transverse color Doppler US in a child with the new onset of a paramidline anterior left neck mass shows a heterogeneously hypoechoic lesion ➡ with internal echoes concerning for dermoid or carcinoma within a TGDC. No internal vascularity is seen. Histologically, the lesion proved to be an uncomplicated TGDC.* **(Right)** *Transverse US in an 8 year old shows a typical infrahyoid TGDC as an anechoic, paramidline mass ➡ causing mild compression of an otherwise normal-appearing thyroid lobe.*

KEY FACTS

TERMINOLOGY

- Branchial cleft cyst, branchial apparatus cyst (BAC), branchial apparatus anomaly (BAA)
- Anomaly/remnant of embryologic endodermal pouch, mesodermal arch, or ectodermal cleft
  - May be cyst, fistula, or sinus tract

IMAGING

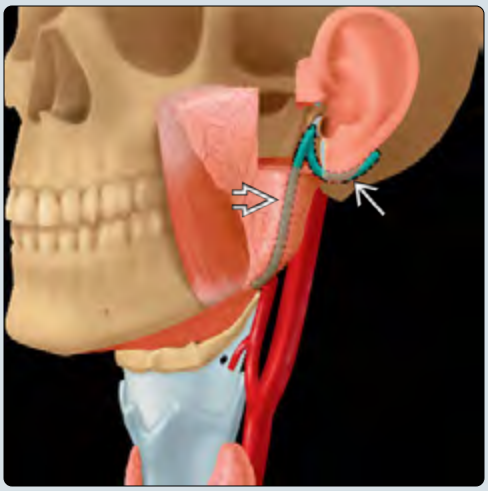
- Well-defined, smooth-walled unilocular cyst of face/neck
  - ± wall thickening, enhancement, & septation with surrounding edema if infected
- Location
  - 1st BAC: Periauricular, in or adjacent to parotid
  - 2nd BAC (most common): Anterior to sternocleidomastoid muscle, posterior to submandibular gland, lateral to carotid sheath
  - 3rd BAC: Posterior triangle in upper neck or anterior triangle in lower neck
  - Cervical thymic cyst: Anywhere along thymopharyngeal duct from lateral hypopharynx to location of normal lobes of thymus in superior mediastinum
  - 4th BAA: Typically sinus tract or fistula extending from apex of pyriform sinus to anterior lower neck, usually in or adjacent to left thyroid lobe

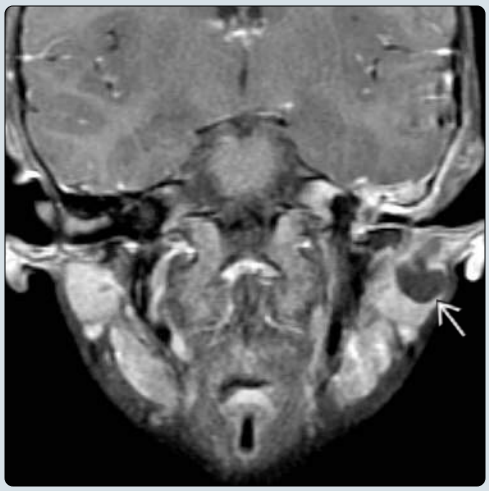
TOP DIFFERENTIAL DIAGNOSES

- Suppurative lymph node or abscess
- Thyroglossal duct cyst
- Lymphatic malformation
- Dermoid & epidermoid

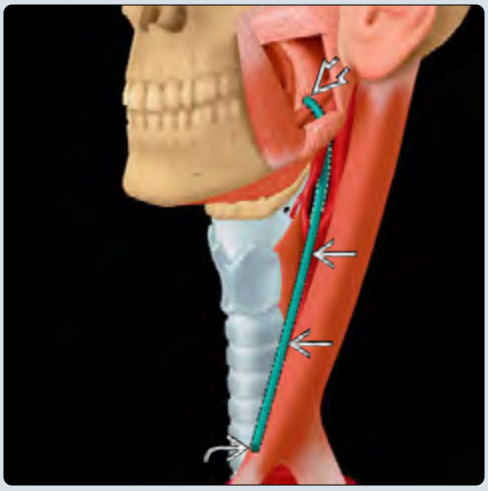
CLINICAL ISSUES

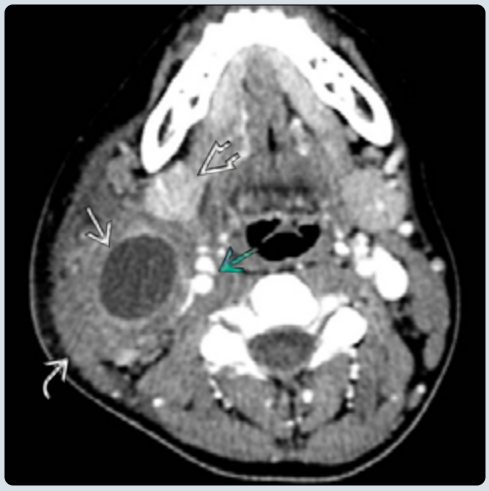
- Typical presentations include soft, painless mass vs. recurrent infections or drainage
  - 1st BAC: ± otorrhea or recurrent parotid abscess
  - 4th BAA: ± left thyroid lobe abscess
- Treatment requires complete surgical excision of cyst & associated fistula or sinus tract, if present





(Left) *Oblique graphic shows the 2 forms of a 1st branchial apparatus cyst (BAC). Type I ➡ courses from the medial bony external auditory canal (EAC) toward the retroauricular area. The tract of a type II BAC ➡ connects the EAC to the angle of the mandible.* **(Right)** *Coronal T1 C+ FS MR in a 3-year-old patient with a 1st BAC shows a lobulated, nonenhancing periparotid cyst ➡ extending towards the junction of the cartilaginous & bony EAC.*





(Left) *Oblique graphic of the tract of a 2nd BAC ➡ shows a proximal opening ➡ in the faucial tonsil & a distal opening in the anterior supraclavicular neck ➡.* **(Right)** *Axial CECT in a teenager with an enlarging mass shows a cystic-appearing lesion ➡ in the typical location of a 2nd BAC (dorsal to the submandibular gland ➡, anterior to the SCM ➡, & lateral to the carotid sheath vessels ➡). Note the thick, irregular rim & overlying soft tissues, consistent with superimposed cyst infection plus cellulitis/myositis.*

TERMINOLOGY

Synonyms

- Branchial cleft cyst, branchial apparatus cyst (BAC), branchial apparatus anomaly (BAA)

Definitions

- Anomaly/remnant of embryologic endodermal pouch, mesodermal arch, or ectodermal cleft
- May be cyst, fistula, or sinus tract

IMAGING

General Features

- Best diagnostic clue
  - Well-defined, smooth-walled cyst (types 1-3)
    - ± surrounding enhancement if infected
  - Left thyroid lobe abscess: Suspect pyriform sinus fistula or sinus tract (type 4)
    - CT after barium swallow best identifies sinus tract or fistula
- Location
  - 1st BAC (8%)
    - Periauricular (type I)
    - Periparotid (type II): May extend to angle of mandible
  - 2nd BAC (95%): 4 locations based on Bailey classification
    - Deep to platysma (type I): Rare
    - Anterior to sternocleidomastoid (SCM) muscle, posterior to submandibular gland, lateral to carotid sheath (type II): Most common
    - Protrude between internal carotid artery & external carotid artery (type III)
    - Adjacent to lateral pharyngeal wall (type IV, probably remnant of 2nd pharyngeal pouch)
  - 3rd BAC
    - Posterior triangle in upper neck, anterior triangle in lower neck
  - Cervical thymic cyst
    - 3rd pharyngeal pouch remnant: Thymopharyngeal duct remnant from lateral hypopharynx to location of normal lobes of thymus in superior mediastinum
    - ± solid ectopic thymus along same tract
    - Remnants may be mixed cystic & solid
  - 4th BAA
    - Sinus tract or fistula extending from apex of pyriform sinus to anterior lower neck, usually adjacent to left thyroid lobe
    - 4th pharyngeal pouch remnant (or recent literature suggests 3rd pouch remnant)
- Size
  - Variable from small to several cm
- Morphology
  - Most cysts well-defined, unilocular, & round or ovoid

CT Findings

- CECT
  - Low-density cyst with nonenhancing wall in typical location
    - If infected: Thickened, enhancing wall with surrounding cellulitis
  - Pyriform sinus fistula (extends to skin surface) or sinus tract (does not extend to skin surface)
    - Abscess in or adjacent to left thyroid lobe
    - May only show subtle inflammation around apex of pyriform sinus
    - Sinus tract or fistula best demonstrated on neck CT obtained immediately after swallowing oral contrast

MR Findings

- Without infection: Thin-walled, fluid-filled cyst
- If infected: Thick wall with variable signal intensity of fluid + surrounding cellulitis
- Ideal to demonstrate small sinus tract or fistula associated with 1st BAC

Ultrasonographic Findings

- Thin-walled hypoechoic cyst ± mobile internal echoes
- Remnant ectopic thymus shows characteristic "dot & dash" thymic tissue

Imaging Recommendations

- Best imaging tool
  - Ultrasound helpful to confirm cystic nature & location
    - May not show total extent of lesion
  - MR best to evaluate suspected 1st BAC
  - CECT in acute setting (typically infected cyst)
- Protocol advice
  - Suspected 1st BAC: Cover periauricular tissues through angle of mandible
    - MR best identifies associated fistula or sinus tract
    - Coronal images help evaluate relationship to external auditory canal (EAC)
  - Suspected 2nd or 3rd BACs: Image entire neck to upper mediastinum
  - Suspected pyriform sinus fistula or sinus tract: Helpful to image after swallowing oral contrast

DIFFERENTIAL DIAGNOSIS

Suppurative Lymph Nodes

- Thick, enhancing nodal wall with central hypoattenuation/hypoechogenicity = nodal necrosis/intranodal abscess
- Edema in surrounding fat (cellulitis)
- ± thickening of muscles (myositis)
- Associated nonsuppurative adenopathy
- Nontuberculous mycobacterial adenitis lacks surrounding inflammatory change

Abscess

- Conglomeration of suppurative nodes or rupture of node → extranodal abscess
- Superficial: Anterior or posterior cervical or submandibular spaces (SMSs)
- Deep: Retropharyngeal, parapharyngeal, or tonsillar; may grow rapidly → airway compromise ± mediastinal extension
- If anterior to left thyroid lobe, think of 4th BAA
- Exclude dental infection & salivary gland calculus as cause of head & neck infection

Thyroglossal Duct Cyst

- Remnant of thyroglossal duct, anywhere from foramen cecum/base of tongue to thyroid bed in infrahyoid neck
- Cyst with minimal rim enhancement

- Location: 50% at hyoid, up to 25% suprahyoid, up to 25% infrahyoid
  - Suprahyoid usually midline
  - Infrahyoid may be off midline, embedded in strap muscles
- ± nodularity or Ca^{2+} if associated thyroid carcinoma (usually adults)

Lymphatic Malformation

- Congenital slow flow vascular malformation composed of variably sized abnormal lymphatic channels
- Often multilocular transspatial mass
  - Thin rim/septal enhancement; ↑ enhancement if infected, microcystic, or combined with venous malformation
- Present at birth, grows with child
- Hemorrhage → sudden ↑ in size with internal debris or fluid-fluid levels

Ranula

- Simple: Postinflammatory retention cyst with epithelial lining; arises in sublingual gland (or minor salivary gland) in sublingual space (SLS)
  - Appears as well-defined, elongated cyst in SLS
- Diving: Ruptures out of SLS into SMS
  - Cyst in both SLS & SMS
  - May see "tail" if SLS component collapsed

Dermoid & Epidermoid

- Dermoid: Epithelial & dermal elements
- Epidermoid: Epithelial elements only
- Well-defined cyst filled with fluid = dermoid or epidermoid
- Well-defined cyst with fatty material, mixed fluid ± Ca^{2+} = dermoid
- Diffusion restriction on DWI = dermoid or epidermoid

PATHOLOGY

General Features

- Associated abnormalities
  - Bilateral branchial apparatus fistulas/cysts & preauricular tag or pit associated with branchiootorenal syndrome
    - Autosomal dominant inheritance
    - Profound mixed hearing loss: Cochlear & semicircular canal malformations, stapes fixation
    - Renal anomalies: Cysts, dysplasia, agenesis
    - Patulous eustachian tubes

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Soft, painless, compressible mass
  - Fever & cellulitis if associated infection or pyriform sinus fistula (or sinus tract)
  - Drainage at skin opening if fistula present
- Other signs/symptoms
  - Large thymic cysts may lead to airway compromise in neonates

Natural History & Prognosis

- If not recognized & treated, 1st BAC may present with recurrent otorrhea or parotid abscess
- If not recognized & completely excised, 4th BAA/BAC anomalies may present with recurrent thyroiditis

Treatment

- Complete surgical excision of cyst & associated fistula or sinus tract, if present
  - Proximity of facial nerve to cyst puts nerve at risk during surgical excision of 1st BACs
  - Large cervical thymic cysts may require otolaryngology & thoracic surgery

DIAGNOSTIC CHECKLIST

Consider

- 1st BAC if cyst in or around EAC or parotid gland
  - May present with otorrhea or recurrent parotid gland abscess
- 2nd BAC if cyst anterior to SCM, posterior to submandibular gland, & lateral to carotid sheath
- Thymic cyst if cystic mass extends from neck to superior mediastinum &/or associated with anterior carotid sheath
- Pyriform sinus fistula or sinus tract if abscess identified in or adjacent to left thyroid lobe

Image Interpretation Pearls

- If "abscess" seen in typical location, think of infected preexisting BAC/BAA
- If patient over 30 years of age, consider cystic nodal metastasis in differential diagnoses of 2nd or 3rd BAC

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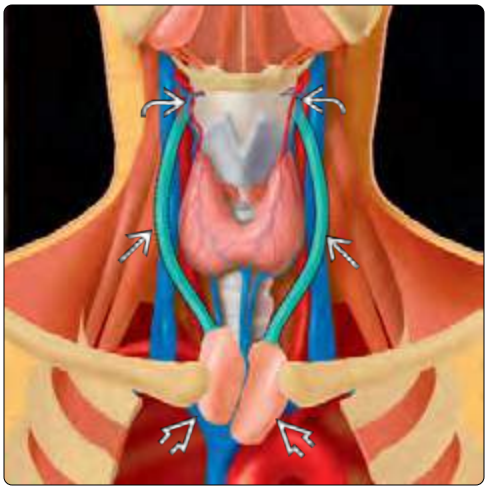
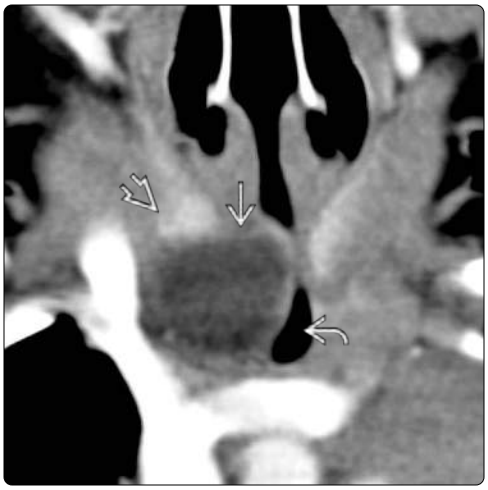
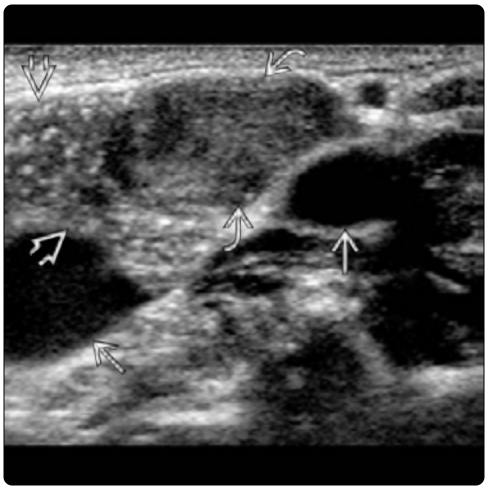
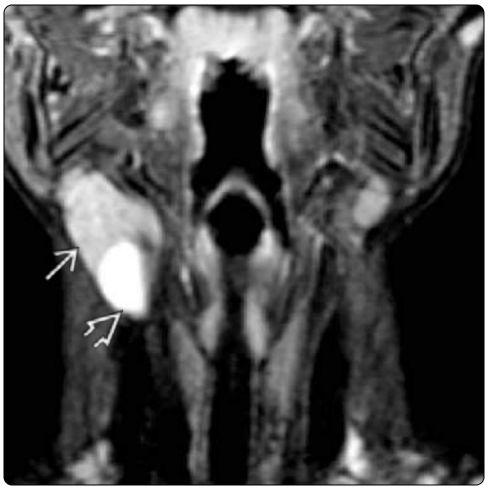


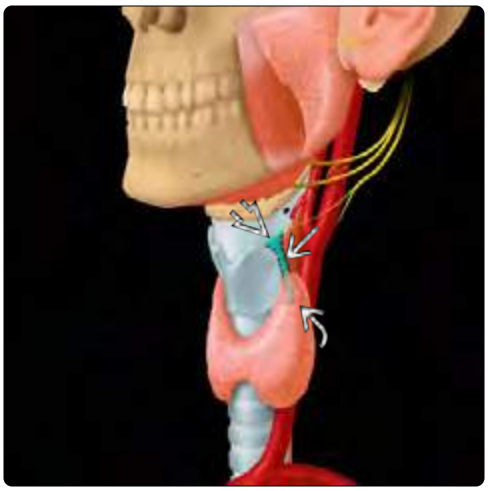


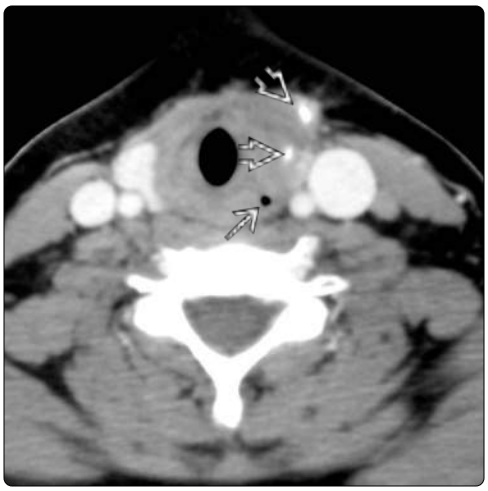
(Left) *Coronal graphic shows both thymopharyngeal duct tracts extending from the lateral hypopharyngeal area to the location of the normal lobes of the thymus in the superior mediastinum. The thymopharyngeal tracts are remnants of the 3rd branchial pouch.* **(Right)** *Coronal CECT in an infant with respiratory distress shows a cystic mass inferior to the right thyroid lobe, deviating & compressing the trachea.*





(Left) *Transverse ultrasound in a 7 year old demonstrates a mixed cystic & solid thymic remnant splaying the carotid sheath vessels. The lateral component demonstrates the "dot & dash" echogenicity typical of thymus, while the medial component is cystic (showing mobile intraluminal echoes on real-time imaging).* **(Right)** *Coronal T2 FS MR in the same patient shows a mixed solid & cystic thymic remnant, the solid portion of which was isointense to intrathoracic thymus on all sequences (not shown).*





(Left) *Oblique graphic of the neck shows the tract of a 4th BAA extending from the hypopharynx to the location of the left thyroid lobe. This explains why this lesion often presents with thyroiditis/abscess.* **(Right)** *Axial CECT in a child with a recurrent draining 4th BAA fistula was obtained after a barium swallow study. Air is in the proximal fistulous tract just below the left pyriform sinus. Barium is in the tract extending anteriorly through the strap muscles. Barium was also noted at a skin opening (not shown).*

Acute Parotitis

KEY FACTS

TERMINOLOGY

- Acute inflammation of parotid gland
  - Bacterial: Localized bacterial infection ± abscess
  - Viral: Usually from systemic viral infection
  - Calculus induced: Ductal obstruction by sialolith
  - Autoimmune: Acute episode of chronic disease
  - Juvenile recurrent parotitis (JRP): Intermittent idiopathic episodes of parotid inflammation

IMAGING

- Typical appearance: ↑ size & enhancement of parotid ± stranding of surrounding fat; parotid retains normal shape
- Bacterial parotitis: Typically unilateral
  - Periparotid cellulitis/stranding common
  - Intra- or periparotid abscess may occur
- Calculus-induced parotitis: Typically unilateral
  - Large duct with intraluminal stone
- Viral parotitis: Typically bilateral
  - Clinical diagnosis; imaging rarely required
- Autoimmune parotitis: Typically bilateral
  - Diagnosed with serum markers
  - Sialography for chronic complications
- JRP: Often unilateral clinically with bilateral imaging abnormalities
  - Normal main duct with dilated intraglandular branches (sialectasis)

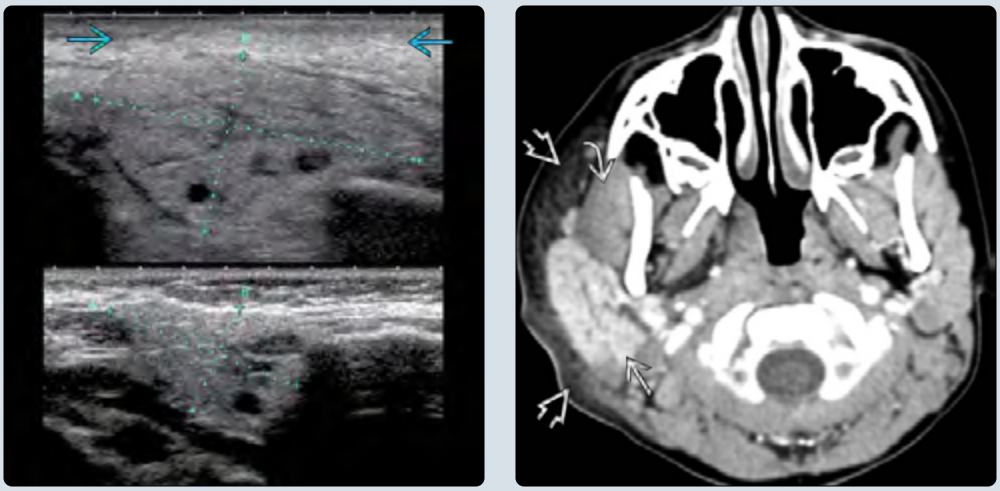
TOP DIFFERENTIAL DIAGNOSES

- Infected 1st branchial cleft anomaly
- Parotid infantile hemangioma
- Salivary gland neoplasms
- Parotid sialosis
- Benign lymphoepithelial lesions of HIV

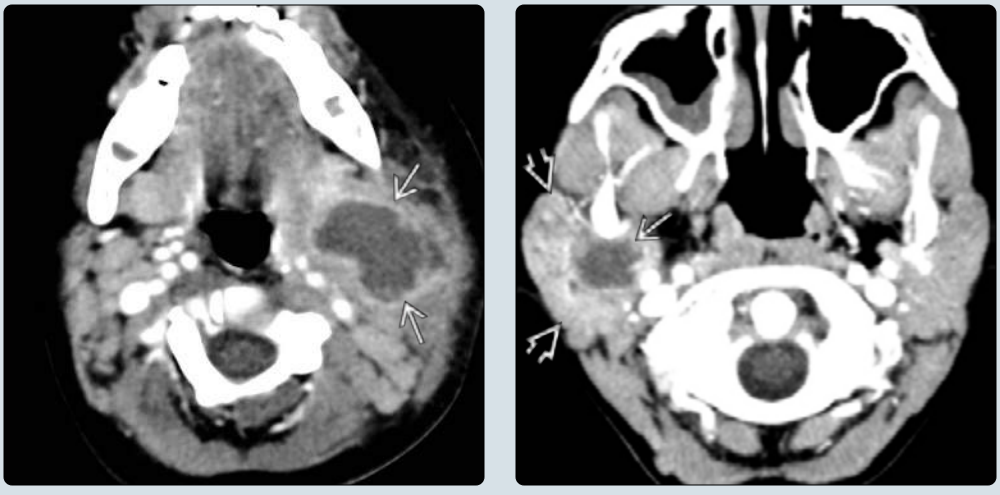
DIAGNOSTIC CHECKLIST

- Re-image if mass persists after resolution of infection (to exclude underlying branchial cyst or malignancy)
- Sialography for recurrent disease to assess complications

(Left) *Longitudinal US of the right (upper) & left (lower) parotid glands (denoted by calipers) in a toddler with right facial swelling shows normal size & echogenicity of the left parotid gland & overlying soft tissues. The right parotid gland is enlarged & mildly heterogeneous with induration of the overlying soft tissues ⇨, typical of parotitis.* **(Right)** *Axial CECT shows diffuse asymmetric enlargement & enhancement of the right parotid gland ➡ with associated facial cellulitis ➡ & myositis ➡ due to acute bacterial parotitis.*



(Left) *Axial CECT shows a low-density, rim-enhancing collection ➡ replacing the left parotid gland. There is substantial surrounding fat stranding. These findings are consistent with an abscess complicating acute bacterial parotitis.* **(Right)** *Axial CECT shows an irregular collection ➡ deep within the enlarged, asymmetrically enhancing right parotid gland ➡. This collection, which could represent phlegmon or abscess, was treated as the former & resolved without drainage on IV antibiotics.*



TERMINOLOGY

Synonyms

- Acute sialadenitis

Definitions

- Acute inflammation of parotid gland
  - Bacterial: Localized infection ± abscess
  - Viral: Usually from systemic viral infection
  - Calculus induced: Ductal obstruction by sialolith
  - Autoimmune: Acute episodes of chronic disease
  - Juvenile recurrent parotitis (JRP) or chronic recurrent parotitis of childhood: Intermittent idiopathic episodes of parotid inflammation

IMAGING

General Features

- Best diagnostic clue
  - Enlarged parotid(s) with surrounding fat stranding
- Location
  - Bacterial: Usually unilateral
  - Viral: 75% bilateral; ± submandibular &/or sublingual gland involvement
  - Calculus induced: Unilateral
  - Autoimmune: Usually bilateral
  - Juvenile recurrent: Unilateral or asymmetric clinical presentations with bilateral imaging findings
- Morphology
  - Entire gland usually involved (but can be focal)
  - Parotid retains normal triangular configuration

CT Findings

- CECT
  - Bacterial: Enlarged, diffusely enhancing parotid
    - Inflammatory stranding of surrounding fat
    - Rim enhancement of low-density abscess (if present)
  - Viral: Enlarged parotids with mild enhancement
  - Calculus induced: Calcified stone; dilated parotid duct with enhancing walls; otherwise like bacterial
  - Autoimmune: Less stranding of surrounding fat
    - May have ductal dilation if longstanding disease

MR Findings

- T2WI
  - Diffuse moderately high signal ± focal areas of fluid signal (microabscesses or dilated ducts)
- T1WI C+
  - Enlarged parotid gland with diffuse, moderate enhancement
  - Abscesses: Rim-enhancing fluid collections
- MR sialography (3D SSFP)
  - Exquisite for demonstrating cysts or dilated ducts
  - JRP: Normal main duct with dilated intraglandular branches (sialectasis)

Ultrasonographic Findings

- Enlarged, hypoechoic, heterogeneous gland
- Numerous small, hypoechoic lesions bilaterally in JRP
- Calculi echogenic with posterior acoustic shadowing ± dilated, hypoechoic duct
- Focal hypoechoic collection suggests abscess formation

Imaging Recommendations

- Best imaging tool
  - Bacterial & calculus-induced inflammation: CECT best for detection of stone or abscess
  - Viral: Typically diagnosed clinically
    - Imaging rarely required
  - Autoimmune disease: Diagnosed with serum markers
    - Sialography for chronic complications
  - JRP: MR sialography

DIFFERENTIAL DIAGNOSIS

Infected 1st Branchial Cleft Anomaly

- 1st branchial cleft cyst in or adjacent to parotid gland
- Superinfection presents as parotid abscess

Parotid Infantile Hemangioma

- Diffusely expanded, hypervascular, & homogeneously hyperenhancing parotid; normal surrounding soft tissues
- Presents with swelling in first few weeks to months of life

Salivary Gland Neoplasms

- Firm focal or infiltrative mass without hyperenhancement
  - Benign: Pleomorphic adenoma
  - Malignant: Adenoid cystic carcinoma, mucoepidermoid carcinoma, acinic cell carcinoma

Parotid Sialosis

- Bilateral prolonged, painless, soft parotid (& occasionally submandibular) gland enlargement

Benign Lymphoepithelial Lesions of HIV

- Bilateral heterogeneous glands, ± cystic & solid lesions
- Prominent Waldeyer ring & cervical nodes
- Affects parotid only, not other salivary glands
- May be found prior to HIV seroconversion

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Bacterial: Sudden onset of parotid pain & swelling
  - Viral: Prodromal symptoms of headaches, malaise, myalgia followed by parotid pain, earache, trismus
  - Calculus induced: Recurrent episodes of swollen, painful gland, usually related to eating
  - Autoimmune: Recurrent episodes + dry mouth
  - JRP: Recurrent episodes ± fever, malaise

Demographics

- Age
  - Bacterial: > 50 yr or neonates
  - Viral: Most < 15 yr; peak age 5-9 yr
  - JRP: Usually begins by 5 yr; resolves by age 10-15

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Infantile Hemangioma, Cervicofacial

KEY FACTS

TERMINOLOGY

- Benign vascular neoplasm (not malformation) of proliferating endothelial cells
- Most common soft tissue tumor of infancy

IMAGING

- Doppler US (including spectral): Characteristic flow patterns
  - High vessel density (> 5/cm²) with low-resistance arterial waveforms but no arteriovenous shunting
- Contrast-enhanced CT/MR
  - Well-defined mass with diffuse & intense enhancement during proliferative phase
  - High flow vessels in/adjacent to mass during proliferation
  - ↓ size with ↑ fatty replacement during involuting phase

TOP DIFFERENTIAL DIAGNOSES

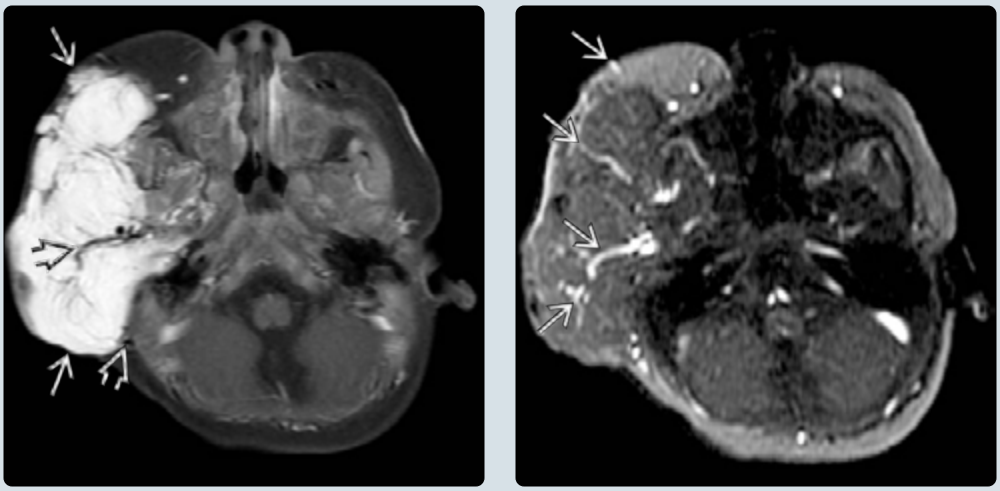
- Congenital hemangioma
- Venous malformation
- Soft tissue sarcoma
- Plexiform neurofibroma
- Arteriovenous malformation

PATHOLOGY

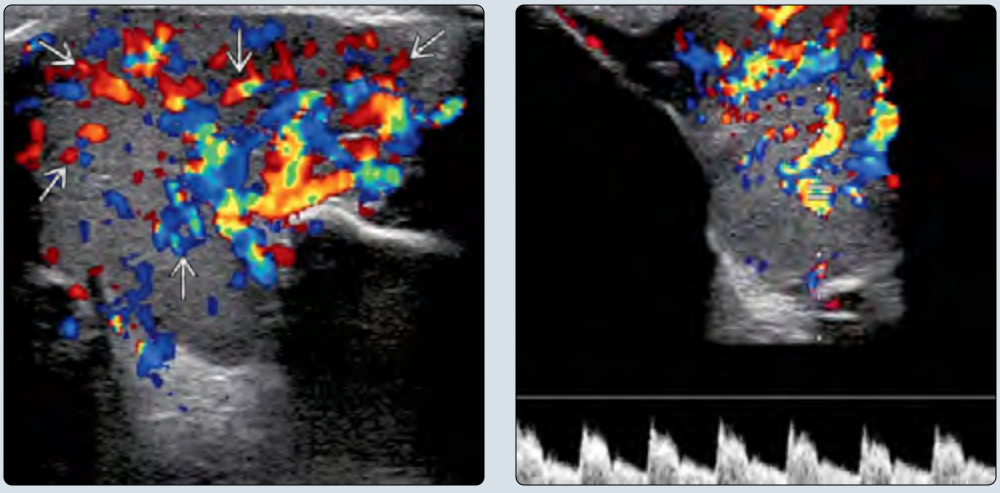
- GLUT1 immunohistochemical marker positive in all phases of growth & regression
  - In contrast to GLUT1 negative congenital hemangiomas

CLINICAL ISSUES

- Typically inapparent at birth → appears in first few weeks of life → grows rapidly for months → spontaneously involutes over years
  - Typically warm, soft, raised reddish or strawberry-like cutaneous lesion
- Majority do not require treatment; propranolol 1st-line therapy in setting of ulceration or vital structure compromise (e.g., airway or orbit)
- If age, clinical & imaging appearance, or growth history atypical for IH, biopsy recommended



(Left) *Axial T1 C+ FS MR in a 5 month old shows a large, lobulated, intensely enhancing mass ➡ infiltrating the massively enlarged right parotid gland. Notice the prominent intralesional ⇨ & perilesional ➡ flow voids typical of an infantile hemangioma (IH).* **(Right)** *Axial 2D SPGR flow-sensitive MR sequence in the same patient shows multiple high-flow vessels within & adjacent to the primary parotid IH ➡.*



(Left) *Transverse color Doppler ultrasound in a 1 month old shows a lobular lesion replacing & expanding the parotid gland. Note the high vessel density ➡ typical of a proliferating IH.* **(Right)** *Transverse color Doppler spectral tracing through the lesion in the same patient demonstrates low-resistance arterial waveforms typical of a proliferating IH. The waveforms will develop a high-resistance pattern during involution.*

TERMINOLOGY

Abbreviations

- Infantile hemangioma (IH)

Synonyms

- Capillary hemangioma
- Widespread misuse of term "hemangioma" in literature; IH different entity from
  - Congenital hemangioma, spindle cell hemangioma, epithelioid hemangioma, lobular capillary hemangioma (pyogenic granuloma): All benign but different vascular neoplasms
  - Hemangioendothelioma: Higher grade vascular neoplasm
  - Cavernous hemangioma, vertebral body hemangioma, & synovial hemangioma: Venous malformations

Definitions

- Infantile hemangioma (IH): Benign vascular neoplasm (not malformation) of proliferating endothelial cells
  - Most common soft tissue tumor of infancy
- 2014 revised classification by International Society for the Study of Vascular Anomalies (ISSVA) retains 2 main categories
  - Vascular tumors (true neoplasms with cellular proliferation; generally grow out of proportion to patient)
  - Vascular malformations (congenital errors of vessel development; generally grow commensurate with patient)

IMAGING

General Features

- Best diagnostic clue
  - During proliferative phase (PP): Lobular, well-defined mass with intense, diffuse enhancement + high flow vessels in/adjacent to mass
  - During involuting phase (IP): ↓ size, vascularity, & enhancement with progressive fatty replacement
- Location
  - 60% occur in head & neck
    - Essentially any space of face or neck, including parotid, orbit, nasal cavity, subglottic airway; rarely intracranial
      - When intracranial &/or multiple, consider PHACES association
    - Typically not intramuscular
- Size
  - Depends on phase
    - Proliferative phase: Rapid growth beginning few weeks after birth & continuing 6-24 months
    - Involuting phase: Gradual regression over next several years
    - Involuted phase: Inactive relatively small residual lesion
- Morphology
  - Majority: Isolated, focal, well-circumscribed lobulated lesions in subcutaneous tissues
    - Tend to displace/efface rather than encase normal structures
  - Occasionally multiple, transspatial, or deep
    - May be part of PHACES association
      - **P**osterior fossa malformations (cerebellar hypoplasia most common)
      - **H**emangioma (infantile) of face & neck, typically segmental or midline
      - **A**rterial stenosis, occlusion, aneurysm, hypoplasia, agenesis, or aberrant origin
      - **C**ardiovascular defects (aortic coarctation/aneurysm/dysplasia, aberrant subclavian artery ± vascular ring, VSD)
      - **E**ye abnormalities (PHPV, coloboma, morning glory disc anomaly, optic nerve hypoplasia, peripapillary staphyloma, microphthalmia, cataract, sclerocornea)
      - **S**upraumbilical raphe or sternal clefts/defects
- CECT
  - Well-circumscribed lobulated mass with diffuse & intense contrast enhancement in PP
    - Prominent vessels in/adjacent to mass
    - No internal Ca^{2+} or surrounding edema
  - Progressive fatty infiltration of mass + ↓ size in IP
- MR
  - T1: Isointense to muscle in PP; hyperintense from fatty replacement during IP
  - T2: Mildly hyperintense relative to muscle
  - T2 FS/STIR: At least moderately hyperintense relative to muscle (but not fluid signal intensity) during PP; hypointense to muscle (follows fat) during IP
  - T1 C+ FS: Intense contrast enhancement in PP
  - GRE: Hyperintense high flow vessels in/adjacent to mass in PP
    - Corresponding serpiginous flow voids in/adjacent to mass on SE/FSE sequences
  - MRA: Stenosis, occlusion, agenesis, or aneurysm of craniocervical vessels (in PHACES association)
- Ultrasonographic findings
  - Grayscale
    - Soft tissue mass with variable echogenicity & few macroscopic vessels
      - ↑ echogenicity during IP
  - Color/spectral Doppler
    - High vessel density (> 5 vessels/cm^2), high systolic Doppler shift (> 2 kHz), & low resistive index arterial vessels without arterialized veins (to suggest shunting) during PP
      - Mean venous peak velocities not ↑ (unlike AVM)
    - ↓ vessel density, ↑ resistive index in IP

Imaging Recommendations

- No imaging necessary in majority of patients
  - Due to characteristic cutaneous appearance & timeframe
- Best imaging tool depends on indications
  - To establish diagnosis with atypical history, appearance, or clinical behavior of lesion: US with spectral Doppler if relatively superficial
    - Deeper lesions may not have classic cutaneous appearance
  - To define deep extension of lesion with implications for compromise of vital structures (e.g., orbit & airway): MR
  - To plan/evaluate therapy before & after treatment (if considering medical or surgical/laser therapy): MR

- To evaluate for suspected PHACES association (e.g., large segmental facial IH): MR
- Protocol advice
  - Pulsed/spectral color Doppler US to document characteristic flow throughout lesion
  - MR imaging should include flow-sensitive, fluid-sensitive, & T1 C+ FS sequences
    - Parotid hemangiomas: Evaluate location of CNVII

DIFFERENTIAL DIAGNOSIS

Congenital Hemangioma (RICH/NICH)

- Present at birth or on prenatal imaging; does not proliferate after birth
  - Rapidly involuting congenital hemangioma (RICH): Involutes by 3-14 months
  - Noninvoluting congenital hemangioma (NICH)
- Solid, heterogeneous, less well-defined mass ± Ca^{2+}, hemorrhage, necrosis
- GLUT1 negative on histology

Venous Malformation

- Congenital slow flow vascular malformation composed of large venous lakes
- Fluid signal intensity throughout mass; ± fluid-fluid levels, phleboliths
- Gradual patchy fill-in with contrast

Soft Tissue Sarcoma

- Solid or mixed cystic/solid mass, typically firm
- Mild to moderate enhancement ± osseous erosion
- Internal vascularity present but typically < < IH

Plexiform Neurofibroma

- Infiltrative lobulated masses with target appearance in cross section
- Transspatial involvement ± poorly defined margins
- Additional stigmata of neurofibromatosis type 1

Arteriovenous Malformation

- Congenital high flow vascular malformation
- Arteriovenous shunting through tangle of feeding arteries & large draining veins
  - ± other soft tissue components

PATHOLOGY

General Features

- Etiology
  - Proposed theory: Clonal expansion of angioblasts with high expression of basic fibroblast growth factors & other angiogenesis markers
- Genetics
  - Majority sporadic

Microscopic Features

- Prominent endothelial cells forming small vascular channels (PP); flat endothelial cells + fibrofatty replacement (IP)

Immunohistochemical Features

- Glucose transporter 1 (GLUT1) positive during all phases of proliferation & regression

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Growing superficial soft tissue mass in young infant (PP); typically with warm, soft, raised reddish or strawberry-like cutaneous discoloration
    - Spontaneous involution over next several years
  - Occasionally deeper lesions show bluish skin discoloration secondary to prominent draining veins
- Other signs/symptoms
  - Ulceration of overlying skin
  - Airway obstruction
  - Proptosis from orbital lesion
  - Features of PHACES association

Demographics

- Age
  - Median age at presentation: 2 weeks; majority present by 1-3 months
    - Typically inapparent at birth
    - Up to 1/3 nascent at birth (i.e., pale or erythematous macule, telangiectasia, pseudoecchymotic patch or red spot)
- Gender
  - F > M (1.5-4:1)
- Epidemiology
  - Most common head & neck tumor in infants
  - Incidence of 1-2% in neonates; 12% by age 1 year
    - ↑ in preterm & low birth weight infants
      - Up to 30% of infants weighing < 1 kg
- Ethnicity
  - Most frequent in Caucasians

Natural History & Prognosis

- Majority undergo PP followed by spontaneous regression by age 9 years
  - 90% resolve by 9 years
- Large & segmental facial hemangiomas have ↑ incidence of complications if not treated

Treatment

- Majority do not require treatment
- Treatment indications
  - Compromise of vital structures (e.g., optic nerve compression or airway obstruction)
  - Significant skin ulceration
- Treatment options
  - Propranolol (β-blocker) now primary therapy (instead of oral steroids) due to low side effect profile
  - Less common: Intralesional steroids, laser, surgical excision

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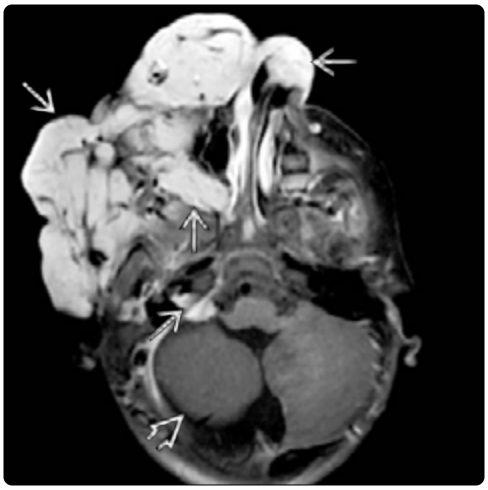
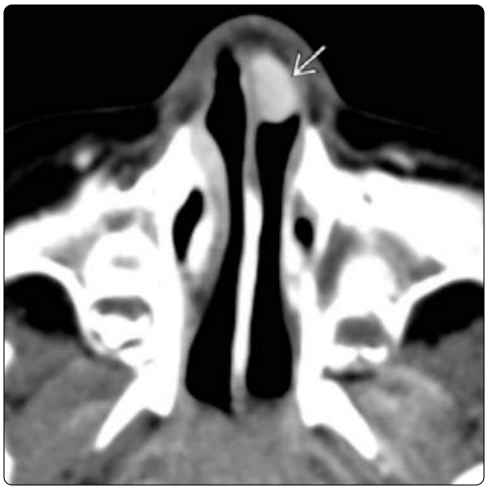
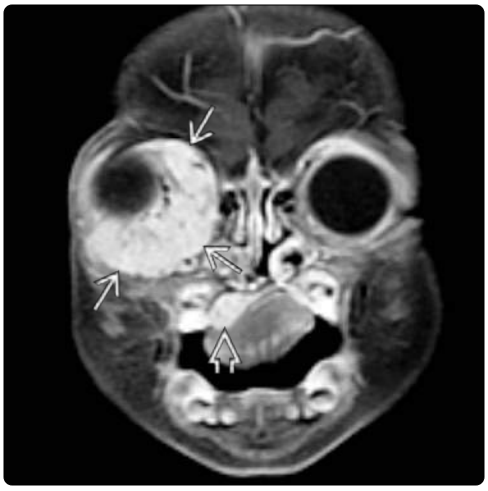
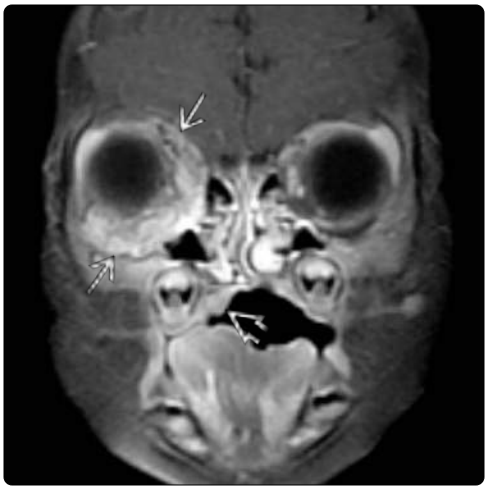


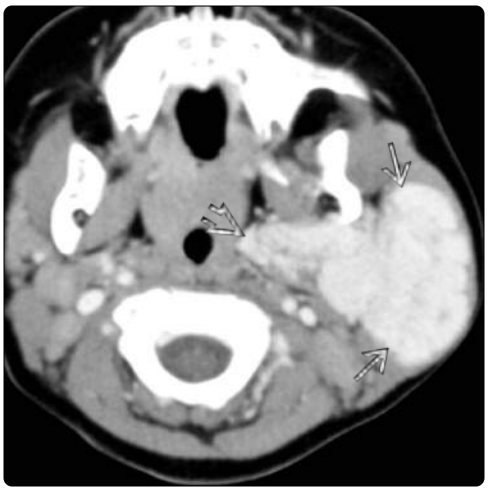


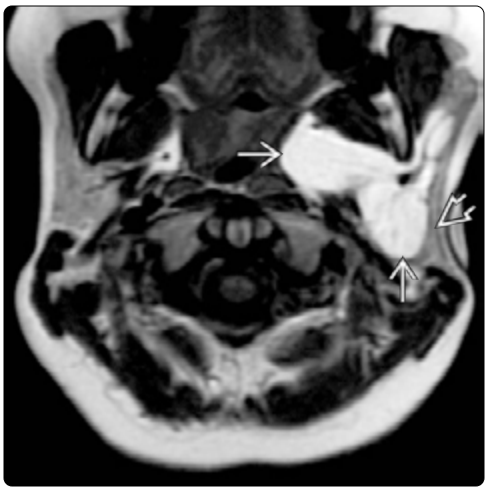
(Left) *Axial T1 C+ FS MR in a 4-month-old girl with PHACES association demonstrates multiple enhancing infantile hemangiomas in the right parotid space, right posterior-inferior orbit, right check, nose, & right IAC/CPA. Also note the ipsilateral right cerebellar hemisphere hypoplasia.* **(Right)** *Axial CECT in a 7 month old with a nasal mass demonstrates an intensely enhancing mass occluding the left nasal vestibule, typical of IH. A pyogenic granuloma could have a similar appearance.*





(Left) *Coronal T1 C+ FS MR in a 2 month old shows a large, intensely enhancing IH involving the right intraconal & extraconal orbit. There is a 2nd lesion involving the right palate.* **(Right)** *Coronal T1 C+ FS MR in the same patient after 6 months of propranolol therapy (which was instituted to prevent orbital complications) shows significant interval regression of the orbital & palatal lesions.*





(Left) *Axial CECT in a 1-year-old girl demonstrates intense homogeneous enhancement of a large IH replacing the entire superficial & deep lobes of the left parotid gland.* **(Right)** *Axial T1 MR in the same child 8 years later shows interval size decrease & diffuse fatty replacement of the left parotid IH. Note the thin sliver of overlying normal-appearing superficial parotid gland tissue.*

KEY FACTS

TERMINOLOGY

- Rhabdomyosarcoma (RMS): Malignant neoplasm of striated muscle; most common childhood soft tissue sarcoma

IMAGING

- Up to 40% occur in H&N
  - Orbit, parameningeal sites, other sites
- MR best to
  - Characterize solid soft tissue mass
    - Often intermediate signal on T2 FS or STIR
    - Often mild to moderate contrast enhancement
    - Often restrict diffusion
  - Evaluate intracranial spread (requires contrast)
- Bone CT best to look for bone destruction or remodeling
- Neck imaging for cervical metastatic adenopathy
  - PET/CT may improve staging & treatment evaluation

TOP DIFFERENTIAL DIAGNOSES

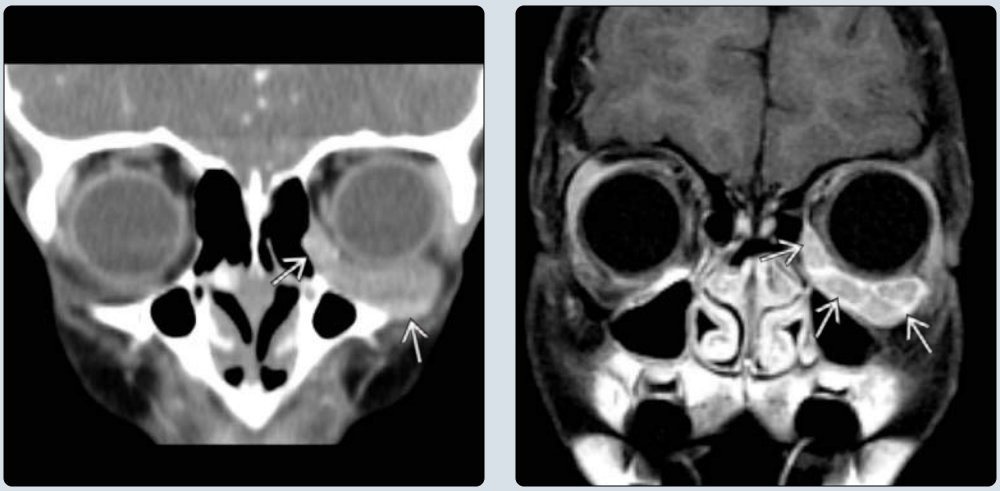
- Infantile hemangioma
- Langerhans cell histiocytosis
- Juvenile angiofibroma
- Plexiform neurofibroma
- Nasopharyngeal carcinoma
- Non-Hodgkin & Hodgkin lymphoma
- Leukemia

PATHOLOGY

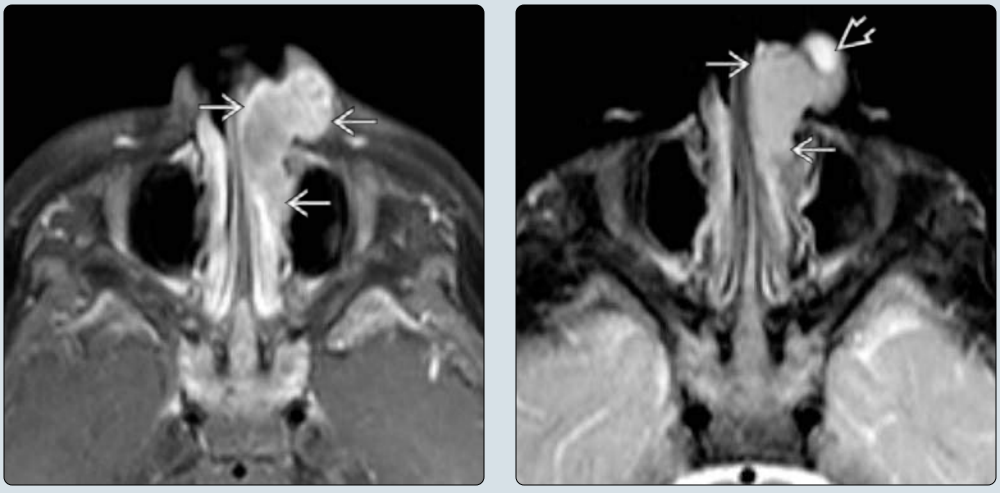
- Originates from primitive mesenchymal cells (rhabdomyoblasts) committed to skeletal muscle differentiation
- 3 histologic subtypes
  - Embryonal RMS: Most common; young children
  - Alveolar RMS: 2nd most common; 15-25 years of age
  - Pleomorphic RMS: Least common; 40-60 years of age

CLINICAL ISSUES

- Presentation: 70% < 12 years of age, 40% < 5 years
- Treatment: Surgery, chemotherapy, ± radiation therapy



(Left) *Coronal CECT in an 8 year old shows a nonspecific, mildly heterogeneous extraconal mass ➡ in the inferior & inferomedial left orbit without bone destruction.* **(Right)** *Coronal T1 C+ FS MR in the same child shows a heterogeneously enhancing mass ➡ with an atypical appearance for a hematoma, hemangioma, or vascular malformation. Biopsy confirmed an embryonal rhabdomyosarcoma (RMS). The absence of bone destruction does not exclude RMS.*



(Left) *Axial T1 C+ FS MR in a 15-month-old boy with intermittent epistaxis & swelling of the left nasal ala demonstrates a heterogeneously enhancing left intranasal/nasal alar mass ➡ obstructing the left nasal cavity.* **(Right)** *Axial STIR MR in the same patient demonstrates primarily intermediate signal intensity throughout the mass ➡ with the exception of a small cystic/necrotic region anteriorly ➡. Subsequent biopsy revealed an alveolar RMS.*

TERMINOLOGY

Abbreviations

- Rhabdomyosarcoma (RMS)

Definitions

- Malignant neoplasm of striated muscle
- Most common childhood soft tissue sarcoma

IMAGING

General Features

- Best diagnostic clue
  - Solid soft tissue mass with variable contrast enhancement
- Location
  - Up to 40% occur in H&N, including orbit, parameningeal sites, neck or face soft tissues, nasal cavity
    - Parameningeal sites: Middle ear, paranasal sinus, nasopharynx, masticator space, pterygopalatine fossa, parapharyngeal space
      - Intracranial extension in up to 55%
- Size
  - Variable; may present earlier in orbit due to confined space → early proptosis

CT Findings

- Invasive soft tissue mass with variable enhancement
- Osseous erosion common but not seen in all cases

MR Findings

- Isointense T1, hyperintense T2 signal relative to muscle
  - Not "fluid bright" unless necrotic/cystic components
- Variable contrast enhancement, often mild-moderate
  - Diffuse, intense enhancement atypical
- Often restricts diffusion

Nuclear Medicine Findings

- PET/CT
  - Hypermetabolic
    - May improve staging & posttreatment evaluation

Imaging Recommendations

- Best imaging tool
  - CT to evaluate osseous erosion
  - MR best for soft tissue mass characterization
  - MR to evaluate perineural & intracranial spread of parameningeal RMS
    - Thickening & enhancement of nerves, leptomeninges
  - MR to distinguish between sinonasal tumor & obstructive/inflammatory disease
- Protocol advice
  - T2 FS or STIR MR: ↑ tumor conspicuity
  - Coronal T1 C+ FS MR: Detect intracranial extension
  - Axial & coronal thin-section bone CT: Osseous erosion
  - Image neck: Rule out cervical metastatic adenopathy

DIFFERENTIAL DIAGNOSIS

Infantile Hemangioma

- Benign vascular neoplasm in infants, often with characteristic cutaneous involvement
- Intensely enhancing, round or lobulated mass with high-flow vessels during proliferative phase
- No bone destruction
- Fatty infiltration during involuting phase

Langerhans Cell Histiocytosis

- Enhancing soft tissue mass filling "punched-out" lytic bone lesion

Juvenile Angiofibroma

- Highly vascular mass causing epistaxis or nasal obstruction in adolescent males
- Intensely enhancing lesion with bone destruction & internal high-flow vessels
- Originates at sphenopalatine foramen on lateral nasal wall
- Often involves nasal cavity, nasopharynx, skull base, masticator space ± orbit, sinus, intracranial extension

Plexiform Neurofibroma

- Benign tumor of peripheral nerves in neurofibromatosis type 1
- Lobulated masses with peripherally ↑ T2 signal & centrally ↓ T2 signal (target sign)
- Bone remodeling, typically without destruction

Nasopharyngeal Carcinoma

- Nasopharyngeal mass in 2nd decade of life
- Variable contrast enhancement
- Central skull base erosion, widening of petroclival fissure, extension to pterygopalatine fossa, masticator & parapharyngeal spaces
- Unilateral or bilateral cervical & lateral retropharyngeal adenopathy

Non-Hodgkin Lymphoma

- 25% of all H&N malignancies in children
- Non-Hodgkin lymphoma (NHL) & Hodgkin lymphoma imaging findings similar
  - Difficult to differentiate
- Sinonasal, orbital, or nasopharyngeal NHL may cause osseous erosion
- Large, nonnecrotic nodes typical

Hodgkin Lymphoma

- 25% of all H&N malignancies in children
- Large, nonnecrotic nodes typical
- Extranodal sites less common

Leukemia

- Granulocytic sarcoma = chloroma
  - Rare manifestation of acute myeloid leukemia (AML) > chronic myeloid leukemia (CML)
- Soft tissue mass ± aggressive bone destruction, diffuse marrow abnormalities

Metastatic Neuroblastoma

- Cervical primary lesions rare; most cervical disease due to metastatic adenopathy
- Metastatic disease to skull frequently bilateral: Enhancing masses surround aggressive osseous erosions, demonstrating spiculated, radiating periosteal reaction

PATHOLOGY

General Features

- Etiology
  - Originates from primitive mesenchymal cells committed to skeletal muscle differentiation (rhabdomyoblasts)
- Genetics
  - ↑ incidence in children with *TP53* tumor suppressor gene mutation
  - Most embryonal RMS have loss of heterozygosity at 11p15 locus
  - Alveolar RMS: 50% have *FOX01* to *PAX3* (or *PAX7*) gene fusion
    - *PAX3-FKHR* gene fusion has better prognosis
- Associated abnormalities
  - ↑ incidence of embryonal RMS in Noonan syndrome
    - Hematologic malignancies & neuroblastoma also seen
  - Rarely associated with neurofibromatosis type 1, Li-Fraumeni, & Beckwith-Wiedemann syndromes
  - Rarely associated with hereditary retinoblastoma
  - May occur as radiation-induced 2nd primary neoplasm

Staging, Grading, & Classification

- Intergroup Rhabdomyosarcoma Study Group (IRSG)
  - Group I: Localized tumor completely resected
  - Group II: Gross total resection with microscopic residual disease
  - Group III: Incomplete resection with gross residual disease
  - Group IV: Distant metastases
- TNM: Tumor site, size, local invasion, lymph nodes, distant metastases

Microscopic Features

- Rhabdomyoblasts in varying stages of differentiation
- Immunohistochemistry: Positive for desmin, vimentin, & antibody to muscle-specific actin
- 3 histologic subtypes
  - Embryonal RMS: Most common (> 50% of all RMS)
    - Primitive cellular structure with round or elongated cells showing hyperchromic irregular nuclei & frequent mitoses
    - Occurs in younger children
    - 70-90% occur in H&N or genitourinary tract
    - Botryoid RMS: Gross appearance like cluster of grapes, typically in patients 2-5 years of age
      - 75% arise in vagina, prostate, or bladder
      - 25% in head/neck or bile ducts
  - Alveolar RMS: 2nd most common
    - Usually occurs in patients 15-25 years of age
    - Most common in extremities & trunk
  - Pleomorphic RMS: Least common
    - Usually in adults 40-60 years of age; rarely < 15 years
    - Most arise in extremities; rarely in head & neck

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Symptoms variable, depend on location
    - Orbit: Mass, proptosis, ↓ vision
    - Sinonasal: Nasal obstruction, epistaxis; may present late with soft tissue facial mass
    - Temporal bone: Postauricular or external auditory canal mass, otitis media, CN VII palsy
    - Neck: Mass, pain, rarely airway compromise

Demographics

- Age
  - 70% < 12 years of age
  - 40% < 5 years of age
- Ethnicity
  - More common in Caucasians

Natural History & Prognosis

- Variable; depends on location & cell type
  - Orbit: Best prognosis (80-90% disease-free survival)
  - Parameningeal: Worst prognosis (40-50% disease-free survival)
  - Embryonal & pleomorphic: Better prognosis than alveolar RMS
    - Alveolar RMS without gene fusion: Prognosis similar to embryonal RMS

Treatment

- Surgical debulking, chemotherapy, ± radiation therapy
- Research ongoing in proton therapy, immunotherapy, & vaccination

DIAGNOSTIC CHECKLIST

Consider

- Not always associated with bone destruction
  - Beware of enhancing soft tissue mass without bone destruction; may simulate infantile hemangioma (IH)
    - IH almost always found in 1st year of life; RMS more common beyond 12 months
    - IH enhances more intensely & homogeneously
    - IH demonstrates high density of low-resistance arterial vessels on color Doppler

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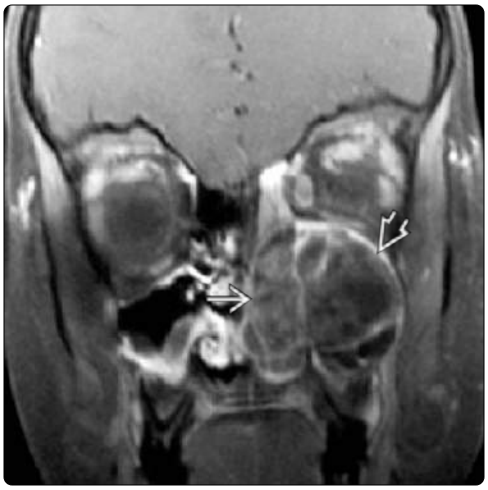
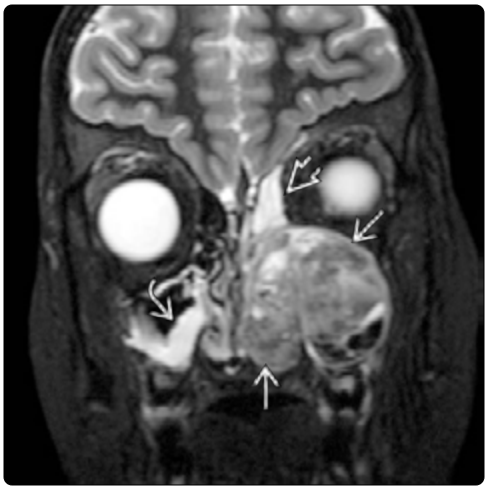
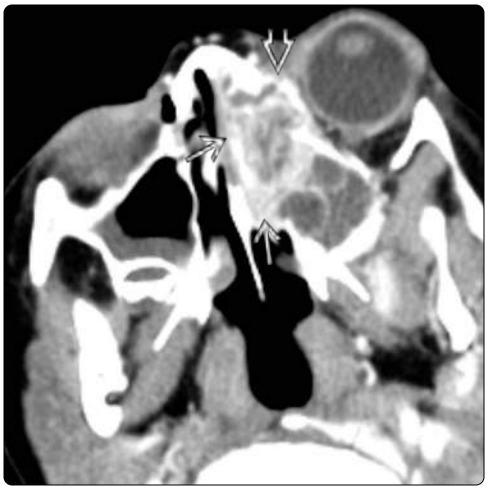
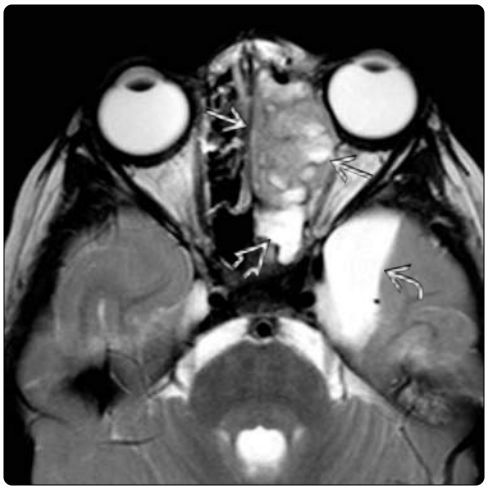


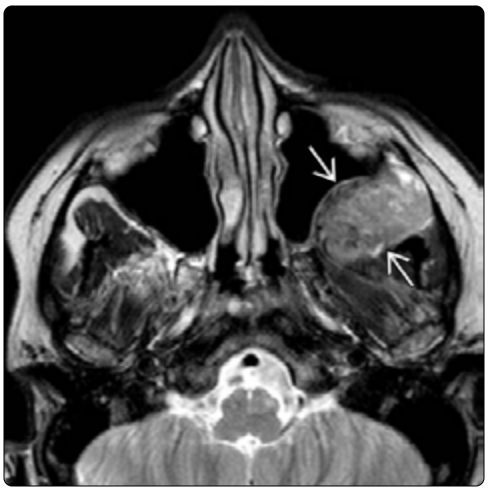


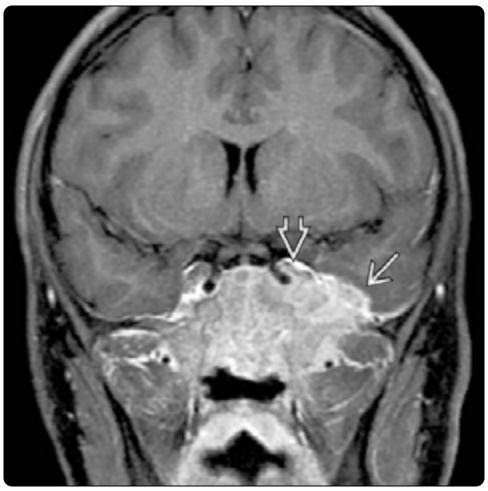
(Left) *Coronal T1 C+ FS MR shows a heterogeneously enhancing left nasal RMS with expansion of the left maxillary sinus. Some component of the disease in the left maxillary sinus could represent inflammatory disease rather than tumor.* **(Right)** *Coronal STIR MR in the same patient better differentiates tumor filling the expanded left maxillary sinus & nasal cavity from hyperintense inflammatory mucosal disease in the left ethmoid & right maxillary sinuses.*





(Left) *Axial CECT in a 5-year-old with left proptosis & palpable cervical adenopathy demonstrates heterogeneous enhancement in the left nasal cavity & anterior maxillary sinus with a discontinuous medial orbital wall.* **(Right)** *Axial T2 MR in the same child demonstrates a mixed-intensity mass of alveolar RMS as compared to the hyperintense sphenoid sinus disease & incidental middle cranial fossa arachnoid cyst.*





(Left) *Axial T2 MR in a 21 year old with an intraoral embryonal RMS shows anterior bowing of the posterior wall of the left maxillary sinus by a heterogeneous, mildly hypointense mass, consistent with the highly cellular RMS.* **(Right)** *Coronal T1 C+ FS MR shows an extensive left skull base RMS with left middle cranial fossa extension (displacing the left temporal lobe) & invasion of the left cavernous sinus.*

Fibromatosis Colli

KEY FACTS

TERMINOLOGY

- Most common cervical "mass" of infancy
- Benign fibrosis of sternocleidomastoid (SCM) muscle
- Postulated to be muscular response to birth trauma or peripartum injury

IMAGING

- Often diagnosed clinically without imaging
- Ultrasound modality of choice when imaging required
- Process entirely intramuscular (contained within SCM), without local invasion or inflammatory changes
- Fusiform expansion of central SCM muscle
  - Thick & short compared to contralateral SCM
- Variable heterogeneity of lesion with loss of normal muscle architecture
  - Ranges from nearly homogeneous to markedly heterogeneous on US/CECT/MR
- Variable internal vascularity within/about mass
- Fascial planes surrounding SCM preserved
- No associated adenopathy, edema, or fluid collection

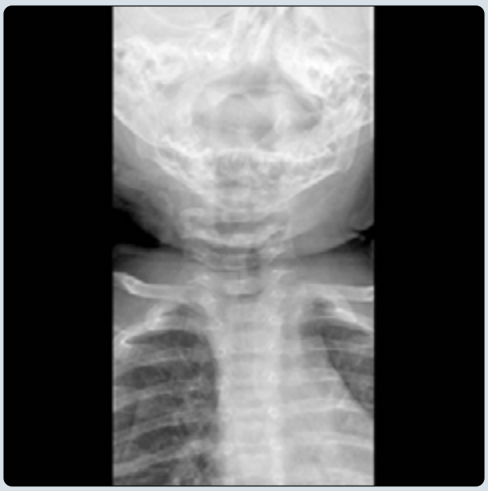
TOP DIFFERENTIAL DIAGNOSES

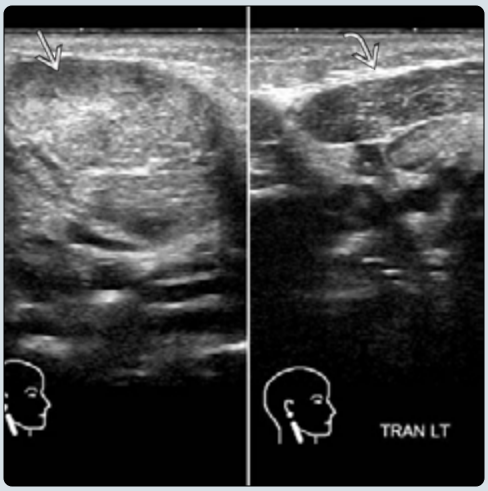
- Cervical lymphadenopathy
- Infantile hemangioma
- Cervical teratoma
- Congenital neuroblastoma
- Rhabdomyosarcoma
- Branchial cleft anomalies
- Lymphatic malformation
- Cervical extension of mediastinal thymus
- Spinal fusion anomalies

CLINICAL ISSUES

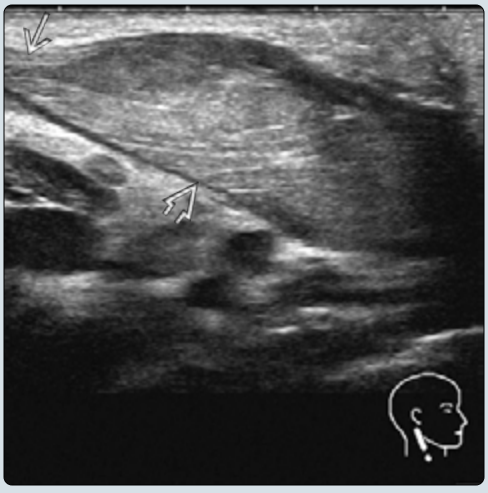
- Painless palpable mass with torticollis
- Contralateral occipital flattening (plagiocephaly) common
- Peak presentation at 24 days old
- Self-limited, usually resolves completely by 6 months of age with conservative treatment/physical therapy

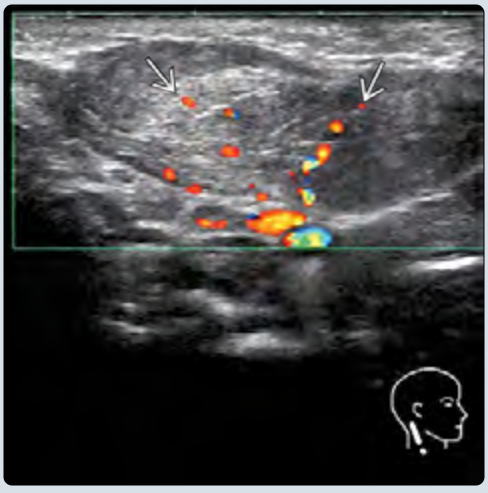
(Left) *AP radiograph of the neck shows mild torticollis with the left ear closer to the left shoulder & the chin slightly turned away. No bony anomalies are seen to explain this neck positioning in this infant with fibromatosis colli.* **(Right)** *Split screen transverse US images in a 3-week-old girl with torticollis show a homogeneously enlarged right sternocleidomastoid muscle (SCM) ⇨ compared to the left ⇨, typical of fibromatosis colli.*





(Left) *Orienting the US transducer along the long axis of the SCM optimally shows the mildly heterogeneous enlargement/expansion of the muscle belly with normal SCM tapering ends ⇨ & sharp, distinct borders ⇨ with adjacent structures.* **(Right)** *Longitudinal US with color Doppler shows that the enlarged muscle has mildly increased blood flow ⇨ & slightly increased heterogeneous echotexture centrally.*





TERMINOLOGY

Synonyms

- Sternocleidomastoid (SCM) pseudotumor, SCM tumor of infancy, congenital muscular torticollis, neonatal torticollis

Definitions

- Most common cervical "mass" of infancy
- Benign fibrosis of SCM muscle
- Postulated to be due to birth trauma or peripartum injury
- Torticollis ("wryneck"): Persistent twisting of neck such that ear on affected side is positioned lower & more midline than normal
  - When bony causes of torticollis have been excluded, consider fibrosis of SCM muscle

IMAGING

General Features

- Best diagnostic clue
  - Focal thickening & fibrosis of sternal or clavicular head of SCM muscle
  - Process entirely intramuscular without local invasion or inflammatory changes
- Location
  - Middle or lower 1/3 of SCM muscle belly
  - Sternal & clavicular muscle bundles affected equally often
  - Right side affected more often than left
- Size
  - 1-3 cm in length
  - May ↑ in size in first 2-3 months of life
- Morphology
  - Focal heterogeneous to homogeneous expansion of SCM muscle, distorting normal muscle architecture
  - SCM tapers to normal thickness proximal & distal to lesion
  - Affected SCM mildly shorter than contralateral side
  - No surrounding inflammatory changes or adenopathy

Radiographic Findings

- Cervical spine films may be obtained to exclude bony abnormality causing torticollis
  - Hemivertebrae or omovertebral bones: Congenital bony anomalies not related to fibromatosis colli
- May see nonspecific unilateral soft tissue fullness or "mass"
- Virtually never calcifies
  - If Ca^{2+} seen, consider neuroblastoma & teratoma

Ultrasonographic Findings

- Grayscale ultrasound
  - Focal mass mildly or moderately expanding SCM muscle belly
  - Loss of normal muscle architecture
  - Variable echogenicity of mass; may be homogeneous or heterogeneous
  - Affected SCM shorter & thicker than contralateral side
    - Comparison with asymptomatic side useful
    - Extended field of view imaging to show entire length of SCM useful
  - Fascial planes surrounding SCM preserved
  - No associated adenopathy, edema, or fluid collection
  - Lack of extramuscular involvement excludes other differential diagnoses
- Color Doppler
  - Variable hyperemia in acute phase
  - May see diminished blood flow in quiescent/fibrotic phase

CT Findings

- Focal thickening/expansion of unilateral SCM muscle
  - Variable enhancement & heterogeneity
- No associated adenopathy or regional inflammatory changes

MR Findings

- T1WI
  - Fusiform enlargement of affected SCM
  - Variable signal intensity
    - Usually isointense to hypointense compared to normal muscle
- T2WI FS/STIR
  - Heterogeneous intramuscular mass; affected SCM overall hyperintense to other muscles
    - Hypointense zones probably due to evolving fibrosis
- T1 WI C+ FS
  - Affected muscle enhances heterogeneously

Imaging Recommendations

- Best imaging tool
  - Often diagnosed clinically without imaging
  - Ultrasound modality of choice when imaging required
    - No ionizing radiation or sedation
    - Clearly localizes mass within SCM muscle
  - MR reserved for atypical cases
    - Best for determining extent of alternate malignant diagnoses
      - Intracranial extension (in rhabdomyosarcoma)
      - Intraspinal extension (in neuroblastoma)
  - CT best modality for showing cortical bony abnormalities of alternate diagnoses
    - Erosion, destruction, remodeling, vertebral segmentation anomalies, or congenital scoliosis
- Protocol advice
  - Regardless of imaging modality, none of these findings should be seen with fibromatosis colli
    - Involvement of tissues outside SCM muscle
    - Lymphadenopathy
    - Airway compression
    - Vascular encasement
    - Bone involvement
    - Intracranial/intraspinal extension
    - Other neck masses

DIFFERENTIAL DIAGNOSIS

Cervical Lymphadenopathy

- Nodes typically discrete & easily recognizable, though may appear mass-like when enlarged & confluent
- Adenopathy more likely to have lobulated contour than smooth, spindle shape of SCM

Infantile Hemangioma

- Rapidly growing benign vascular neoplasm presenting weeks to months after birth, ultimately regressing over years
- Characteristic Doppler US appearance: > 5 vessels per cm^2, shows numerous low resistance arterial waveforms

Cervical Teratoma

- Congenital mixed solid & cystic mass, often large
- Typically contains Ca^{2+}
- May present with airway or feeding difficulties

Congenital Neuroblastoma

- Originates along paraspinal sympathetic ganglia outside SCM
- May displace &/or encase vessels
- Look for Ca^{2+}, intraspinal extension, & bony erosion

Rhabdomyosarcoma

- Solid mass outside SCM, typically in older children
- Variable enhancement & vascularity

Branchial Cleft Anomalies

- Unilocular cyst adjacent to SCM

Lymphatic Malformation

- Compressible multicystic congenital mass, often with deep infiltration & extension across midline

Cervical Extension of Mediastinal Thymus

- Look for contiguity with mediastinal thymic tissue
- Characteristic dot & dash appearance on US

Spinal Fusion Anomalies

- Torticollis with cervical scoliosis due to hemivertebra, fused vertebra, bony bar, omovertebral body, etc.

PATHOLOGY

General Features

- Etiology
  - Uncertain: Probably response to perinatal injury, partial muscle tear, or intramuscular hematoma
- Genetics
  - No genetic predisposition

Gross Pathologic & Surgical Features

- Seldom resected
- Fine-needle aspirates more common than excisional specimens
- Firm, spindle-shaped thickening of muscle, without transmuscular inflammation

Microscopic Features

- Cytologic features of fibromatosis: Bland-appearing fibroblasts & degenerative atrophic skeletal muscle in clean background
- Collagen always present
- Occasional muscle giant cells & parallel clusters of fibroblasts

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painless palpable mass with torticollis
  - Contralateral occipital flattening (plagiocephaly) common
  - History of breech presentation & "difficult" vaginal birth common but not mandatory
- Other signs/symptoms
  - If severe or persistent, may result in limited range of motion at neck &/or facial asymmetry

Demographics

- Age
  - Newborn infants
  - Peak presentation at 24 days old
  - Classically noticed < 8 weeks of age but may worsen in first 2-3 months of life
- Gender
  - Males affected slightly more often than females
- Epidemiology
  - Occurs with 0.4% of live births
  - Breech presentation associated
  - Birth dystocia

Natural History & Prognosis

- Self-limited, usually resolves completely by 6 months of age
- Occasional cases recur or flare during periods of rapid somatic growth

Treatment

- Physical therapy to encourage full range of motion, "stretch" SCM
- 90% full recovery with conservative treatment, physiotherapy
- Surgery only indicated in unusual cases when craniofacial asymmetry or refractory torticollis persists after 1 year

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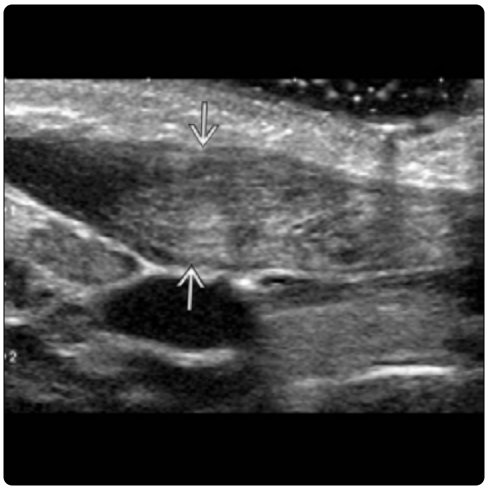
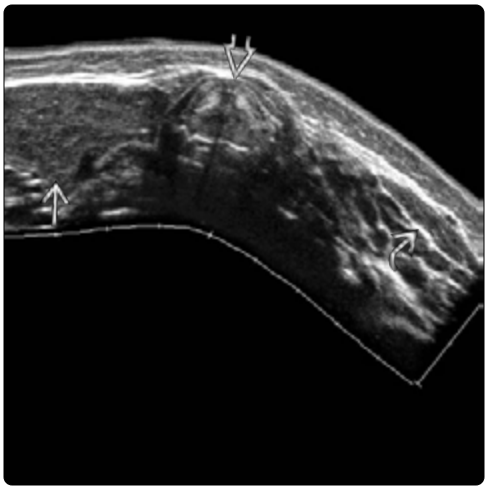
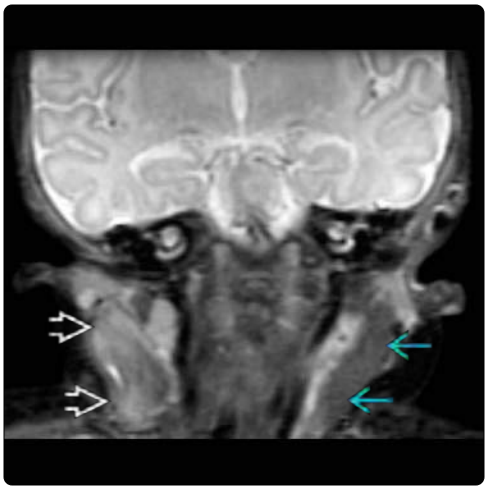
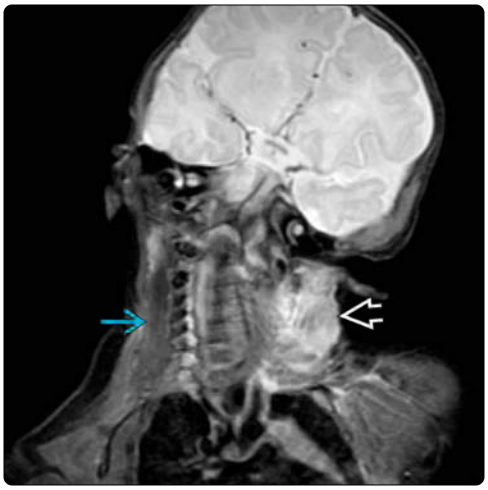


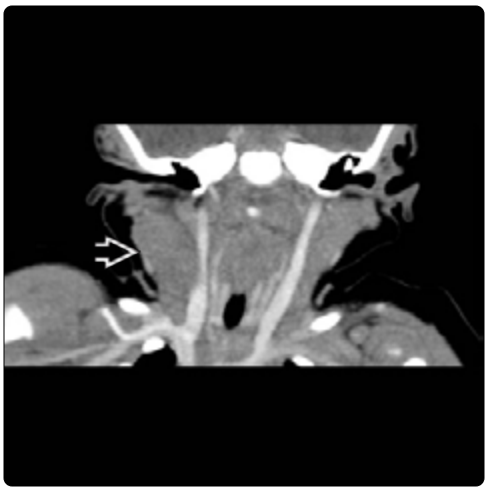


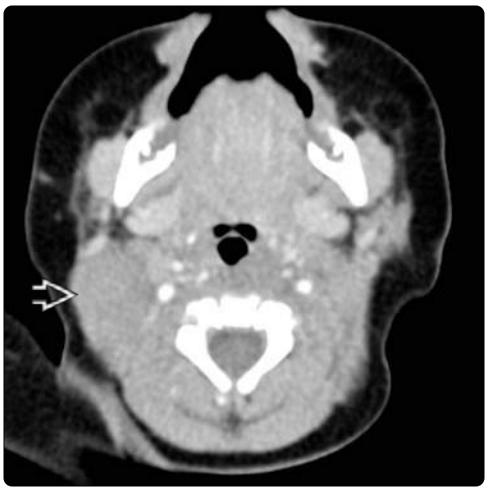
(Left) *Longitudinal US in a 1-month-old infant shows increased size & echotexture of the SCM, consistent with fibromatosis colli.* **(Right)** *Transverse extended field of view image across the neck in an infant with torticollis illustrates the enlargement of the right SCM as compared to the left. The trachea is seen in the midline at the level of the vocal cords.*





(Left) *Coronal STIR MR shows increased signal intensity & size of the right SCM as compared to the left, consistent with fibromatosis colli. MR is useful to exclude extension of the abnormality beyond the muscle sheath, adjacent adenopathy, & underlying vertebral/spinal anomalies.* **(Right)** *Coronal STIR MR in an infant with obvious torticollis shows heterogeneously increased signal intensity & thickness of the left SCM in a patient with fibromatosis colli. Note the normal right SCM.*





(Left) *Coronal CECT reconstruction in an infant with facial anomalies shows thickening of the right SCM, which lies just superficial to the jugular & carotid vessels.* **(Right)** *Axial CECT in the same patient shows the rounded mass-like SCM of fibromatosis colli. Note the lack of adenopathy, inflammatory stranding, or mass effect. The expanded SCM of fibromatosis colli can be much more heterogeneous than this case.*

Suppurative Adenitis

KEY FACTS

TERMINOLOGY

- Pus formation within nodes from bacterial infection
- Synonyms: Adenitis, lymphadenitis, intranodal abscess

IMAGING

- Enlarged node(s) with internal fluid & surrounding inflammation (cellulitis)
  - Most often jugulodigastric, submandibular, or retropharyngeal
- Loss of normal nodal architecture & internal vascularity/enhancement
- Conglomeration of necrotic nodes progressing to abscess shows marked heterogeneity of irregular collection
  - Well-defined enhancing/hyperemic wall
  - Complex hypoechoic/nonenhancing center
- US useful to confirm true liquefaction with drainable pus
  - Swirling internal debris upon compression
- CECT best defines deep extent & complications
  - Lemierre syndrome (venous thrombophlebitis), internal carotid artery spasm or pseudoaneurysm, airway compression, mediastinal extension

TOP DIFFERENTIAL DIAGNOSES

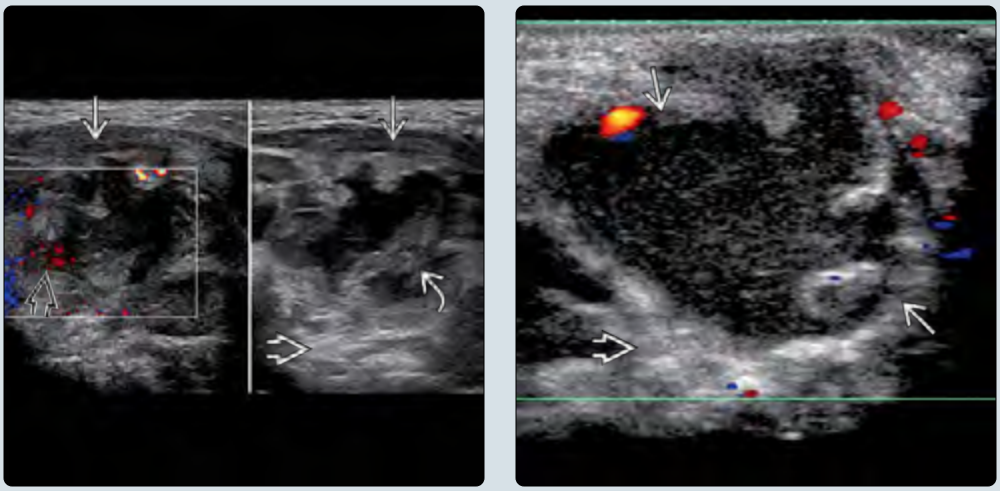
- Nontuberculous *Mycobacterium* adenopathy
- Tuberculous adenopathy
- 2nd branchial cleft anomaly
- Rhabdomyosarcoma
- Lymphoma
- Lymphatic malformation
- Fibromatosis colli

PATHOLOGY

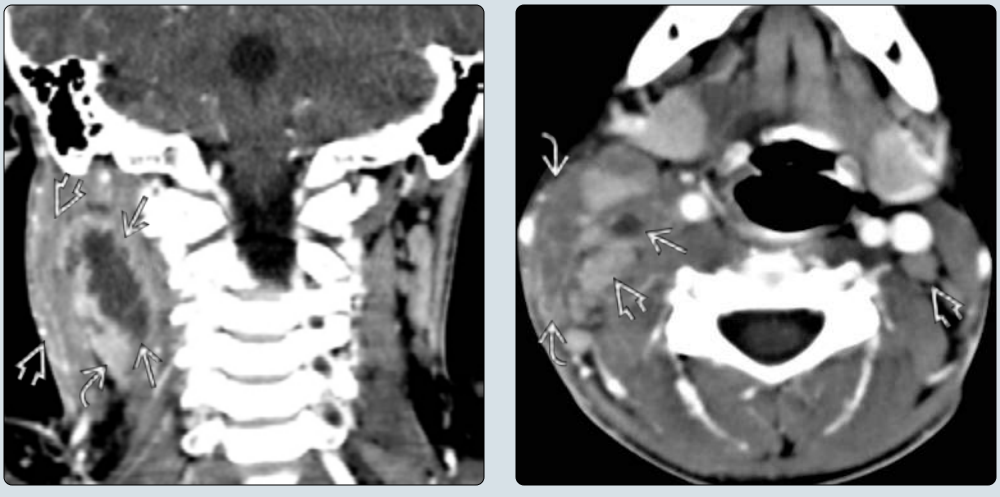
- *Staphylococcus* & *Streptococcus* most frequent organisms

DIAGNOSTIC CHECKLIST

- Look for primary infectious source on images
  - Pharyngitis, dental infection, salivary gland calculi



(Left) *Transverse (left) & longitudinal (right) color Doppler & grayscale US images of an infant with suppurative adenopathy show an enlarged, heterogeneous conglomeration of level II nodes with internal debris, posterior acoustic enhancement, & peripheral hyperemia.* **(Right)** *Transverse color Doppler US in a 6 year old with fever & a tender neck mass shows an irregular hypoechoic abscess with posterior acoustic enhancement, internal echoes, & no internal vascularity.*



(Left) *Coronal CECT in a 6 year old with fever & a fluctuant neck mass shows an irregular, rim-enhancing abscess with overlying myositis/cellulitis & an adjacent nonsuppurative lymph node.* **(Right)** *More inferior image from an axial CECT in the same child shows a round focus of nonenhancement representing segmental clot in the right internal jugular vein, consistent with Lemierre syndrome. Note the right-greater-than left nonsuppurative nodes as well as the overlying myositis/cellulitis.*

TERMINOLOGY

Synonyms

- Adenitis, acute lymphadenitis, intranodal abscess

Definitions

- Pus formation within nodes from bacterial infection

IMAGING

General Features

- Best diagnostic clue
  - Enlarged node(s) with internal fluid & surrounding inflammation (cellulitis)
- Location
  - Any nodal groups of head/neck
    - Most often: Jugulodigastric, submandibular, retropharyngeal
  - May be unilateral or bilateral
- Size
  - Enlarged single node or conglomeration of nodes
    - Often in 1-4 cm range
- Morphology
  - Ovoid to round large node with central necrosis
  - Margins often poorly defined
  - Additional adjacent solid or suppurative nodes typically present

Ultrasonographic Findings

- Early adenitis may show enlarged ovoid foci with retained nodal architecture & hyperemia
- Loss of architecture & internal vascularity with necrosis
  - Central ↓ echogenicity with ↑ through transmission
- Conglomeration of necrotic nodes progressing to abscess show marked heterogeneity of irregular collection
  - Liquefied pus: Swirling internal debris with compression
  - ↑ peripheral vascularity
    - Very low-resistance & pulsatility indices

CT Findings

- CECT
  - Enhancing nodal wall with central hypoattenuation/lack of enhancement
  - Stranding/edema of surrounding tissues
  - With progression to abscess → irregular, ill-defined, peripherally enhancing, low-attenuation collection
    - Caveat: Well-defined, low-attenuation collection with rim enhancement not necessarily pathognomonic for drainable pus → consider US
      - Represents nondrainable phlegmon in ~ 20%
  - ± cellulitis, myositis, Lemierre syndrome (venous thrombophlebitis), deviation of carotid sheath vessels, internal carotid artery spasm or pseudoaneurysm, retropharyngeal soft tissue edema, mass effect on airway, mediastinal extension

MR Findings

- T1WI
  - Node with central low signal intensity
  - Dampening of normal high signal intensity of surrounding fat
- STIR/T2WI FS
  - Node with diffuse or central high signal intensity
  - Edema of surrounding soft tissues
- DWI
  - Complimentary to routine pulse sequences
  - Suppurative, necrotic nodes have reduced diffusivity (bright DWI, dark ADC signal)
    - May show higher DWI & lower ADC signal relative to metastatic necrotic nodes
      - Optimal ADC thresholds remain undefined
- T1WI C+ FS
  - Marked peripheral enhancement with poorly defined margin
  - Absent central enhancement

Nuclear Medicine Findings

- PET
  - Nonspecific ↑ FDG uptake
  - Abscess shows central ↓ FDG uptake

Imaging Recommendations

- Best imaging tool
  - Ultrasound to determine phlegmon vs. abscess & guide aspiration
    - May be difficult to determine deep relationships
  - CECT best for deep neck infections
    - Determine epicenter & total extent for aspiration or surgical planning

DIFFERENTIAL DIAGNOSIS

Nontuberculous *Mycobacterium* Adenopathy

- Asymmetric enlarged nodes with adjacent necrotic ring-enhancing masses
- Minimal or absent subcutaneous fat stranding
- Purified protein derivative (PPD) skin test weakly reactive in ~ 55%
- Usually ≤ 5 years of age

Tuberculous Adenopathy

- Painless, low jugular & posterior cervical low-density nodes
- Ca^{2+} may be present
- Strongly reactive tuberculosis (PPD) skin test, positive interferon gamma release assay
- Systemically unwell if active pulmonary infection

2nd Branchial Cleft Anomaly

- Solitary unilocular cyst, posterior to submandibular gland
- Mimics suppurative node, particularly if inflamed

Rhabdomyosarcoma

- Solid firm soft tissue neoplasm of children (typically beyond infancy) occurring in many head/neck locations

Lymphoma

- Multifocal solid enhancing nodes, often of bilateral neck & chest
- ± enlargement of tonsillar tissue

Lymphatic Malformation

- Congenital multicystic mass with thin rim & septations
- Variable internal echogenicity due to internal hemorrhage
- No significant internal vascularity
- Often infiltrative & transspatial

Fibromatosis Colli

- Self-limited benign reactive mass within sternocleidomastoid (SCM) muscle of young infants
- Expands SCM focally; ranges from homogeneous to very heterogeneous
- Likely due to birth trauma

PATHOLOGY

General Features

- Etiology
  - Primary head/neck infection: Pharyngitis, salivary gland ductal calculus, dental decay ± mandibular osteomyelitis
    - Adjacent lymph nodes enlarge 2° to pathogen: Reactive nodes
    - Intranodal exudate forms, containing protein-rich fluid with dead neutrophils (pus): Suppurative nodes
    - If untreated or incorrectly treated, suppurative nodes rupture → interstitial pus walled-off by immune system → extranodal abscess in soft tissues
  - Reactive nodes from viral pathogen may have 2° bacterial superinfection creating suppurative nodes

Gross Pathologic & Surgical Features

- Fluctuant neck mass with erythematous, warm skin

Microscopic Features

- Acute inflammatory cell infiltrate in necrotic background
  - Presence of neutrophils & macrophages
  - Negative staining for acid-fast bacilli
- *Staphylococcus* & *Streptococcus* most frequent organisms
  - ↑ incidence of MRSA
- Pediatric infections show clustering of organisms by age
  - Infants < 1 year: *Staphylococcus aureus*, group B *Streptococcus*
  - Children 1-4 years: *S. aureus,* group A β-hemolytic *Streptococcus*, atypical mycobacteria
  - 5-15 years: Anaerobic bacteria, toxoplasmosis, cat scratch disease, tuberculosis
- Dental infections: Typically polymicrobial, predominantly anaerobic

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Painful neck mass
    - Overlying skin often warm, erythematous
  - Fever, poor oral intake
  - Elevated WBC & ESR
- Other signs/symptoms
  - Symptoms referable to primary source of infection
    - Pharyngeal/laryngeal infection: Drooling, respiratory distress
    - Peritonsillar infection: Trismus
    - Retropharyngeal or paravertebral infection: Neck stiffness
- Clinical profile
  - Young patient presents with acute/subacute onset of tender neck mass & fever

Demographics

- Age
  - Most commonly seen in pediatric & young adult population
  - Odontogenic neck infections: Adults >> children

Natural History & Prognosis

- Conglomeration of suppurative nodes or rupture of node → abscess formation
  - Superficial neck abscesses: Anterior or posterior cervical space, submandibular space
  - Deep neck abscesses: Retropharyngeal or parapharyngeal space
    - Deep space abscesses can rapidly progress with airway compromise

Treatment

- Antibiotics only for small suppurative nodes & primary infection
- Incision & drainage for large suppurative nodes, abscesses, or poor response to antibiotics
  - CT & US-guided aspiration for minimally invasive management
- Nodes from atypical mycobacteria should be excised to prevent recurrence or fistula/sinus tract

DIAGNOSTIC CHECKLIST

Consider

- Nontuberculous mycobacterial adenitis if no significant inflammatory changes

Image Interpretation Pearls

- Look for primary infectious source on images
  - Pharyngitis, dental infection, salivary gland calculi
- Evaluate airway for compromise with any neck infection
  - More problematic in deep infections
- Evaluate vascular structures for thrombophlebitis

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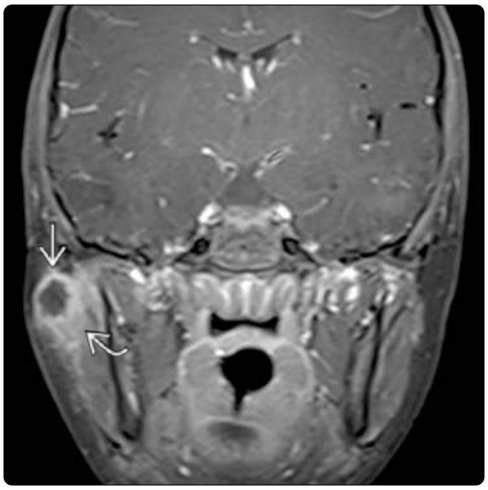
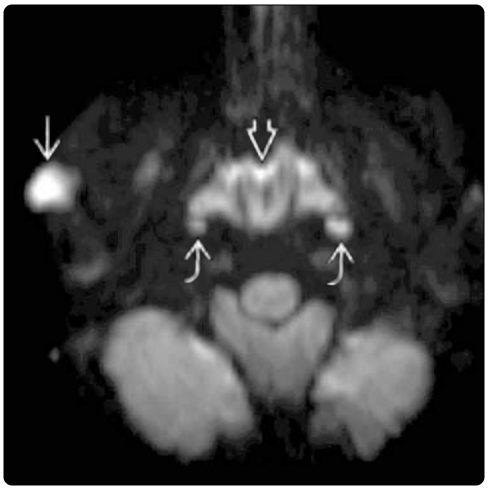
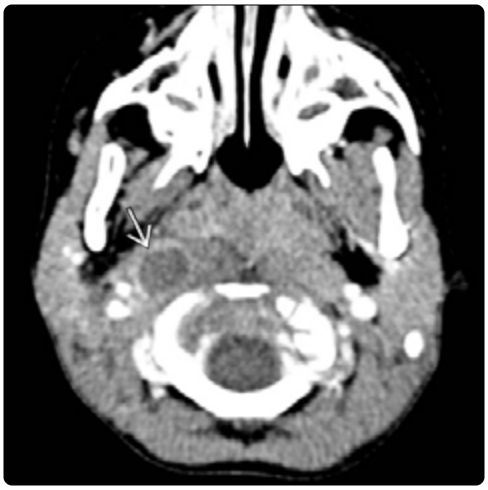
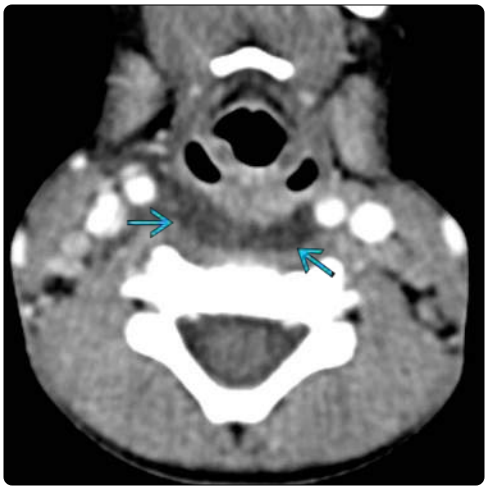


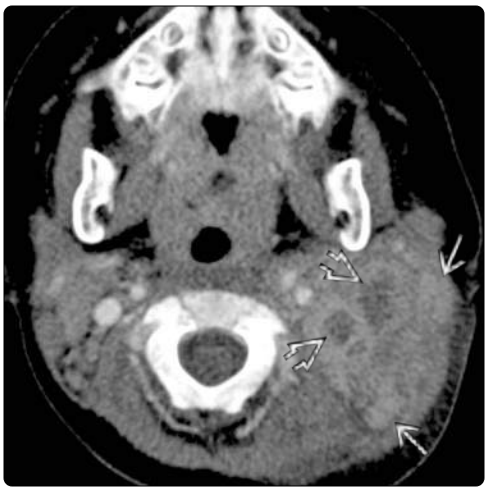


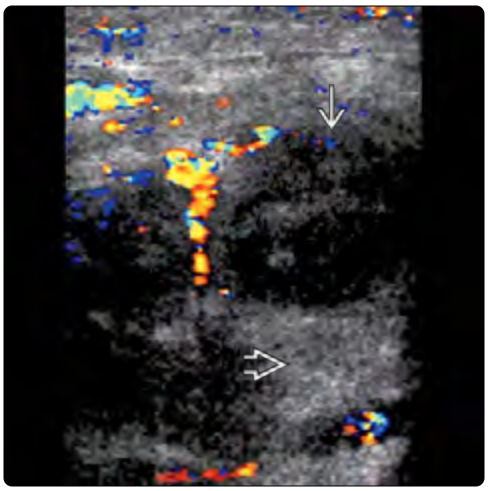
(Left) *Coronal T1 C+ FS MR in an 8-year-old boy with right cheek swelling shows a peripherally enhancing, centrally necrotic intraparotid mass with adjacent inflammatory stranding. Fine-needle aspiration revealed suppurative adenitis.* **(Right)** *Axial DWI MR in the same patient shows the parotid mass to have centrally reduced diffusivity, typical of suppurative adenitis. Note the reactive enlargement of the nasopharyngeal lymphoid tissue & reactive retropharyngeal lymph nodes.*





(Left) *Axial CECT in a child with fever & sore throat shows an enlarged low-attenuation right lateral retropharyngeal lymph node with partial rim enhancement, consistent with early purulence or phlegmon. Note the deviation of the right carotid sheath vessels, which are minimally decreased in caliber compared to the left.* **(Right)** *Axial CECT in the same child demonstrates a poorly defined prevertebral fluid collection with concave anterior margin & no rim enhancement, consistent with soft tissue edema rather than abscess.*





(Left) *Axial CECT in a young child shows a heterogeneous conglomeration of lymph nodes with focal areas of low density indicating suppurative changes. Nonsuppurative nodes are also seen in the mass.* **(Right)** *Longitudinal color Doppler ultrasound of a suppurative level IB node shows a hypoechoic rounded mass with increased through transmission & no central vascularity suggesting necrosis ± frank liquefaction.*

KEY FACTS

TERMINOLOGY

- Lymphatic malformation (LM): Subtype of slow/low flow congenital vascular malformation composed of embryonic lymphatic sacs; not neoplastic
- Composed of macrocysts > 1 cm &/or microcysts < 1 cm

IMAGING

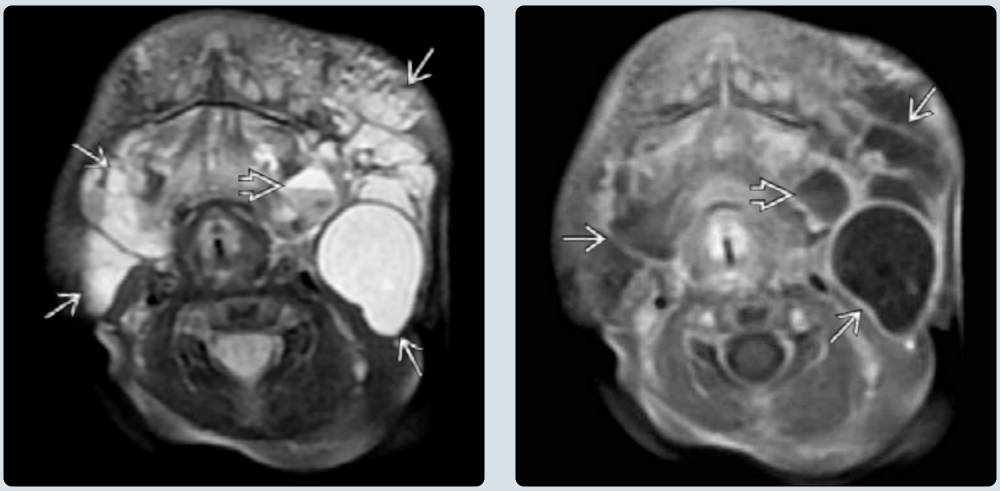
- Macrocystic LM: Multiloculated cystic neck mass with imperceptible wall, thin septations, & fluid-fluid levels
- Microcystic LM: More ill-defined, infiltrative, &/or solid appearing
- Transspatial, often crosses midline extensively
- Insinuates between vessels & other normal structures
- T2 FS/STIR MR: Hyperintense, frequent fluid-fluid levels
  - Best defines extent, relationship to airway & vessels
- T1 FS C+ MR: No significant or minimal rim enhancement
  - Must compare with precontrast T1 as hemorrhage & protein often show hyperintensity
- US: Cysts can show varying degrees of ↑ echogenicity; Doppler shows no significant internal vascularity

TOP DIFFERENTIAL DIAGNOSES

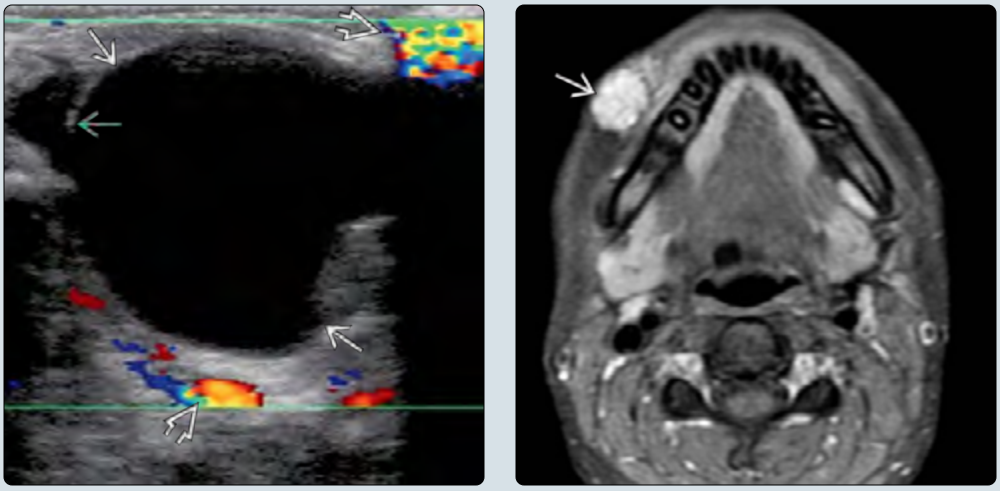
- 2nd branchial cleft anomaly
- Abscess
- Teratoma
- Neurofibroma
- Soft tissue sarcoma

CLINICAL ISSUES

- Nontender, compressible mass
  - Present since birth, grows commensurate with patient
  - May not be clinically apparent until hemorrhage, infection, or hormonal stimulation → rapid ↑ in size
- Depending on size & extent, treatment options primarily include resection, sclerotherapy (for macrocysts), & sirolimus
  - Combination therapy, often staged, may be required



(Left) *Axial T2 FS MR in a 1-week-old infant demonstrates a multiloculated, mixed micro- & macrocystic transspatial lymphatic malformation (LM) involving the left anterior neck more than the right. A single fluid-fluid level is present in a left-sided submandibular macrocyst, typical of layering blood products.* **(Right)** *Axial T1 C+ FS MR in the same child shows a typical appearance of an extensive LM. The macrocysts show only mild peripheral enhancement, & the fluid-fluid level is much more difficult to discern.*



(Left) *Transverse color Doppler US in an infant with a macrocystic LM shows the typical anechoic nature of the dominant cyst with vessels identified adjacent to, but not within, the cyst. Note the thin septation peripherally.* **(Right)** *Axial T1 C+ FS MR in a teenager with a firm, tender lump shows a lobulated, enhancing subcutaneous mass adjacent to the right mandibular body, proven to be a microcystic LM. The well-circumscribed morphology & enhancement could also be seen with a sarcoma, thus requiring excision.*

TERMINOLOGY

Synonyms

- Vascular malformation, lymphatic type; lymphatic anomaly
- Avoid incorrect terms: Cystic hygroma & lymphangioma

Definitions

- Lymphatic malformation (LM): Subtype of slow/low flow congenital vascular malformation composed of embryonic lymphatic sacs; not neoplastic
  - No communication with normal lymphatics
  - Composed of macrocysts > 1 cm &/or microcysts < 1 cm

IMAGING

General Features

- Best diagnostic clue
  - Macrocystic LM: Multiloculated cystic neck mass with imperceptible wall, thin septations, & fluid-fluid levels
  - Microcystic LM: More ill-defined, infiltrative, &/or solid appearing
  - Crosses tissue planes, insinuating between vessels & other normal structures
- Location
  - Any face/neck location (not intracranial)
  - Often in multiple contiguous spaces: Transspatial
    - Soft tissue involvement > > bone
    - Single lesion > > multiple discontinuous lesions
  - Often crosses midline extensively

CT Findings

- CECT
  - Unilocular or multilocular cystic mass with minimal rim &/or septal enhancement

MR Findings

- T1WI
  - Primarily hypointense fluid; hyperintense if prior hemorrhage or high protein content (± fluid-fluid levels)
- T2WI FS or STIR
  - Best sequence to map lesion extent: Hyperintense throughout
    - Fluid-fluid levels in multiple cysts very common
  - When transspatial, often poorly marginated
- T1WI C+ FS
  - No significant enhancement (± subtle rim enhancement)
  - Patchy enhancement suggests VLM or microcystic LM

Ultrasonographic Findings

- Unilocular vs. septated & multilocular transspatial mass
- Contents predominantly hypo- or anechoic
  - Separate compartments in multicystic mass can show varying degrees of ↑ echogenicity
    - ± swirling debris &/or layering fluid-debris levels
- No true vascular flow in cysts by Doppler
  - ± flow (from encased normal vessels) in septations

DIFFERENTIAL DIAGNOSIS

2nd Branchial Cleft Anomaly

- Ovoid, unilocular cyst at angle of mandible with characteristic displacement pattern

Abscess

- Fluid collection with thick, enhancing, irregular wall

Teratoma

- Solid & cystic components typical ± internal vascularity
- Tend to be more unilateral & focal than LM

Neurofibroma

- ± lobular, low-attenuation foci on CT without significant enhancement
- T2 MR often confirms characteristic target sign

Soft Tissue Sarcoma

- Well-defined, typically solid; rarely predominantly cystic

PATHOLOGY

General Features

- Etiology
  - Congenital error of vessel morphogenesis
- Genetics
  - Majority sporadic
  - May be part of extensive overgrowth syndrome with capillary venolymphatic malformation
  - Generalized lymphatic anomaly ("lymphangiomatosis"), Kaposiform lymphangiomatosis, & Gorham-Stout disease may have soft tissue LM in association with bone & visceral lesions

CLINICAL ISSUES

Presentation

- Most common signs/symptoms
  - Nontender, soft, compressible mass
  - Present since birth & grows commensurate with patient
    - May not be clinically apparent until hemorrhage, infection, or hormonal stimulation → rapid ↑ in size
  - Larger lesions detected prenatally
- Other signs/symptoms
  - LMs may infiltrate upper airway or cause extrinsic compression

Treatment

- Surgical resection &/or percutaneous sclerotherapy
  - Sclerotherapy primarily for macrocystic disease
  - Extensive disease often requires combined &/or numerous staged procedures
- Medical therapy with sirolimus

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