Differential Diagnosis for Findings on Skeletal Survey vs. Child Abuse

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Objectives

• Review features of entities that have findings resembling child abuse on skeletal survey
Topics

- Normal variants
- Osteogenesis Imperfecta
- Skeletal dysplasias
  - Metaphyseal chondrodysplasia, Schmid type
  - Spondylometaphyseal dysplasia, Sutcliffe/corner fracture type

- Menkes Syndrome
- Birth injury/iatrogenic
- Congenital syphilis
- Copper deficiency
- Scurvy
- Rickets (next talk!)
Normal Variants

- Contour changes of the metaphysis:
  - Step-off, Beak, Spur
    - Distinguishable from metaphyseal abuse injuries
    - No radiolucent separation between fragment and metaphysis

- Proximal Tibial Cortical Irregularity

Normal

Bone bark
= perichondral collar
= subperiosteal bone collar
= ring of Lacroix

Primary spongiosa = mineralized cartilage in the developing metaphysis.
The adjacent metaphyseal margin is formed by the trabeculae
of the primary spongiosa, which have been exposed by osteoclastic
resorption (curved arrow).

Normal variant: Metaphyseal step-off

- Indistinct cortical margin and focal paucity of cortical bone due to subperiosteal bone collar
- 90 degree cortical angulation at metaphysis near physis
Normal variants: Metaphyseal step-off

14 mo

9 mo
Normal variant: Beak

- Medial projection of metaphysis
  - Proximal humerus (16%)
  - Proximal tibia (1%)
  - Bilateral (77%)
Normal variant: Beak

Normal variant: Beak

14 mo
Normal variant: Spur

- Discrete linear projection of bone
- Extension of bone bark around unossified physis
- Continuous with the cortex
- Extends beyond metaphyseal margin beneath perichondrial ring
Normal variant: Spur

3 mo

Normal variant: Proximal Tibial Cortical Irregularity

• Medial projection of metaphysis
  – 4%
  – Bilateral 25%

• May be associated with physiologic periosteal reaction
Normal variant: Proximal Tibial Cortical Irregularity

3 mo
Osteogenesis Imperfecta
Osteogenesis Imperfecta

- Heterogeneous group of genetic mutations controlling collagen synthesis
- Most common disease causing fracture predisposition
- Variable presentation and severity, mild to perinatal lethal
  - Blue sclera
  - Hearing loss
  - Dentinogenesis imperfecta
  - Kyphoscoliosis → respiratory complications
  - In utero fractures
- COL1A1, COL1A2, and IFITM5 gene testing
# Osteogenesis Imperfecta

<table>
<thead>
<tr>
<th>OI type</th>
<th>Defective gene</th>
<th>Defective protein</th>
<th>Defective mechanism</th>
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<tr>
<td>I</td>
<td>COL1A1</td>
<td>α1(I) collagen</td>
<td>Collagen quantity</td>
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<td>Collagen structure</td>
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<td>V</td>
<td>IFITM5</td>
<td>BRIL</td>
<td>Matrix mineralization</td>
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<td>VI</td>
<td>SERPINF1</td>
<td>PEDF</td>
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<td>VII</td>
<td>CRTAP</td>
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<td>Collagen 3-hydroxylation</td>
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<td>LEPRE1</td>
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<td>PPIB</td>
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<td>BMP1/mTLD</td>
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<td>SP7/OSX</td>
<td>SP7/OSTERIX</td>
<td>Osteoblast development</td>
</tr>
</tbody>
</table>

**Autosomal dominant**

**Autosomal recessive**

Mild, nondeforming
Perinatal lethal
Severe, progressively deforming

Osteogenesis Imperfecta
Osteogenesis Imperfecta

9 mo
Irritability with diaper change,
Older sister with unexplained fractures.
Osteogenesis Imperfecta

2 mos later
Return to ED
with “neck pain”
Osteogenesis Imperfecta

2 mos later
Return to ED with “neck pain”

From prior skeletal survey
Osteogenesis Imperfecta

2 mos later
Return to ED
with “neck pain”

From prior skeletal survey
Osteogenesis Imperfecta

2 mos later
Return to ED with “neck pain”

OI type VI
SERPINF1
Osteogenesis Imperfecta

OI type III
Severe,
progressively deforming
COL1A2
Dysplasias
Dysplasias

• Most dysplasias with metaphyseal irregularities present with notable clinical, radiologic or laboratory findings

• Exceptions
  – Metaphyseal chondrodysplasia, Schmid type
  – Spondylometaphyseal dysplasia, Sutcliffe/corner fracture type

• Follow-up skeletal survey will not change
Dysplasias – Metaphyseal chondrodysplasia, Schmid type

- Autosomal dominant
- Non-familial cases present > 1yo or later
- Enlarged capital femoral epiphysis
- Pelvis almost always normal
- Coxa vara
- Distal ≥ proximal femoral metaphysis involved
- Anterior rib changes
- Normal spine, metacarpals, phalanges
Dysplasias – Metaphyseal chondrodysplasia, Schmid type

- 11 mo
- Enlarged femoral heads
- Normal pelvis

Dysplasias – Metaphyseal chondrodysplasia, Schmid type

- 3 yo
- Enlarged femoral heads
- coxa vara
- distal ≥ proximal femur
- Femoral bowing

Dysplasias – Metaphyseal chondrodysplasia, Schmid type

- 8 yo
- Anterior rib changes
- Normal spine

Dysplasias – SMD Sutcliffe/Cornr-fracture type
Dysplasias – SMD Sutcliffe/Cornerr-fraction type

• Skeletal changes become more pronounced as patient becomes weight-bearing
• Predominant: metaphyses of long bones
• Subtle: vertebral bodies
Dysplasias – SMD Sutcliffe/Cornor-fracture type

- Predominant: long bone metaphyses
  - Metaphyseal irregularity and sclerosis, "fracture-like"
  - Marked bilateral coxa vara, nearly vertically oriented physes
  - Tibia vara/Blount disease-like change
Dysplasias – SMD Sutcliffe/Courner-fracture type

• Subtle:
  - vertebral bodies
    - Mild biconvex vertebral endplates
    - Very mild platyspondyly
    - Odontoid hypoplasia
Menkes Syndrome
Menkes Syndrome

- AKA Kinky hair disease
- X-linked recessive
- Prevalence 1 in 100,000-250,000
- Symptoms appear in infancy; mild forms in childhood

Erik N. Swartz CMAJ 2002;166:1442-1443
Menkes Syndrome

- Disorder of copper transport from intestinal cells
- Neurodevelopmental/growth delay
- Hypotonia
- Seizures
- Scant, stubby, coarse, yellow/white hair

Credit: Alila Medical Media/Shutterstock.com
Menkes Syndrome

- Metaphyseal widening, spurs
- Long bone fractures
- Wormian occipital bones
- Hydronephrosis
- Ureteral dilatation
- Bladder diverticula
- Tortuous intracranial arteries
- Cerebral and cerebellar atrophy
- Chronic subdural hemorrhages
Menkes Syndrome

- Metaphyseal spurs, fractures, irregularity,
- Periosteal new bone formation

3 mo with seizures
Menkes Syndrome

3 mo: atrophy
tortuous intracranial arteries
Wormian bones
Birth injury/iatrogenic
Birth injury

• Correlate with age, expected healing ~10 days
• Metaphyseal fracture, Epiphyseal separation
  – Breech, armling presentation
• Long bone shaft fracture
  – Caesarean section
• Clavicle fracture
  – High birth weight, cephalic presentation
Iatrogenic

• Club foot deformity manipulation and casting

Iatrogenic

- VACTERL
- Vertebral body anomalies
- Imperforate anus
- Renal agenesis
- Anhydramnios
- Pulmonary hypoplasia
- PROM at 20 weeks
- Born at 27 weeks
- L knee contracture
- (outside institution)
Iatrogenic
Congenital syphilis
Congenital Syphilis

- Transplacental infection by *Treponema pallidum* spirochete
- Disseminated throughout fetus
- 1/3 stillbirth; 1/3 contracts; 1/3 unaffected
- +/- symptomatic at birth
Congenital Syphilis

- Periostitis
  - Mild → severe
- Osteitis
  - Localized → diffuse
- Metaphysis
  - Dense/lucent/alternating
  - Serrations
  - Exuberant callus
  - Wimberger’s sign – focal prox tib

Congenital Syphilis

2 mo

- Periostitis
- Osteomyelitis
  - Pathologic fractures
  - Metaphysis
  - Diaphysis

Congenital Syphilis

- Early congenital syphilis
  - Prematurity
  - Hepatosplenomegaly, jaundice
  - Nasal chondritis, “runny nose”
  - Generalized lymphadenopathy
  - Maculopapular rash

- Late congenital syphilis
  - Saddle nose
  - Sabre shin
  - Frontal bossing
  - Hutchinson’s triad
    - Hutchinson’s teeth
    - Interstitial keratitis
    - CN VIII deafness
Congenital Syphilis

36 week preterm newborn, mother RPR+
Copper deficiency
Copper Deficiency

• Copper required for endochondral bone, collagen
• Rare: ~100 cases in 1987
• Full term infants body stores sufficient for 5-6 months
• Low birth weight infants have 2 months
• Modern formulas and breastfeeding should not cause
• Plasma copper < 40 μg/dl
• Ceruloplasmin < 13 mg/dl
• Other clinical findings: sideroblastic anemia, neutropenia, hypotonia, psychomotor retardation, pallor
Copper Deficiency

- 5 mo with irritability
- Bilateral metaphyseal irregularity and lucency
- Loss of cortical sharpness
- Midshaft bowing
- Metaphyseal cupping
- Metaphyseal spurs in medial distal femurs
- Osteopenia
- Increased density in zone of provisional calcification

Scurvy
Scurvy

- Hypovitaminosis C
- Bleeding tendency
- Impaired collagen synthesis
- Osteoporosis
- Impaired wound healing
Scurvy

- Osteopenia
- “pencil point” Cortical thinning
- Trummerfeld zone (“field of rubble”) metaphyseal lucencies
- Pelkan spur causing metaphyseal cupping
- Frankel line: dense zone of provisional calcification
Scurvy


- Trummerfeld zone
- Pelkan’s spur
- Frankel line
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