AMSER Case of the Month December 2020

3 yo with inability to bear weight and spiral femur fracture concerning for non-accidental trauma

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Patient Presentation

- HPI: 3 yo M who presented with 1 day of inability to bear weight on left lower extremity (LLE) after a low-level fall after he slipped on a scarf. Spiral fracture was found on initial hip/femur X-rays, initially concerning for non-accidental trauma. LLE was splinted. Per mother, patient did not notice any other symptoms, such as joint tenderness, effusions, redness over LLE.
- ROS: negative
- PMHx: none, no prior trauma, falls or fractures
- PSHx: none
- FamilyHx: no osteogenesis imperfecta, no Hx of recurrent fx in family

Pertinent Labs

- BMP
  - Slight hypocalcemia 8.9, slight hypophos 5.5
  - LDH, uric acid, ESR, CRP within normal limits
- CBC
  - WBC, RBC within normal limits
  - 1% bands
- 25-OH-Vitamin D - low at 17.7

What Imaging Should We Order?

ACR Appropriateness Criteria

Variant 1: **Acute hip pain. Fall or minor trauma. Suspect fracture. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
Radiography hip	Usually Appropriate	☢☢☢
Radiography pelvis	Usually Appropriate	☢☢
Radiography pelvis and hips	Usually Appropriate	☢☢☢
CT pelvis and hips with IV contrast	Usually Not Appropriate	☢☢☢
CT pelvis and hips without and with IV contrast	Usually Not Appropriate	☢☢☢☢
CT pelvis and hips without IV contrast	Usually Not Appropriate	☢☢☢
MRI pelvis and affected hip without and with IV contrast	Usually Not Appropriate	O
MRI pelvis and affected hip without IV contrast	Usually Not Appropriate	O
Bone scan hips	Usually Not Appropriate	☢☢☢
US hip	Usually Not Appropriate	O

X-ray femur and hip were the modalities ordered by ER physician

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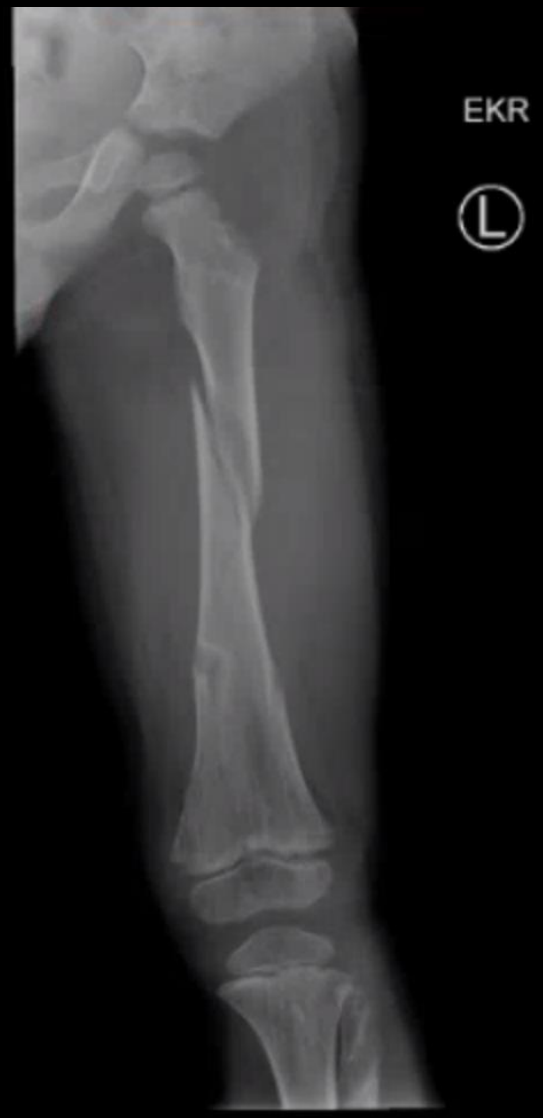
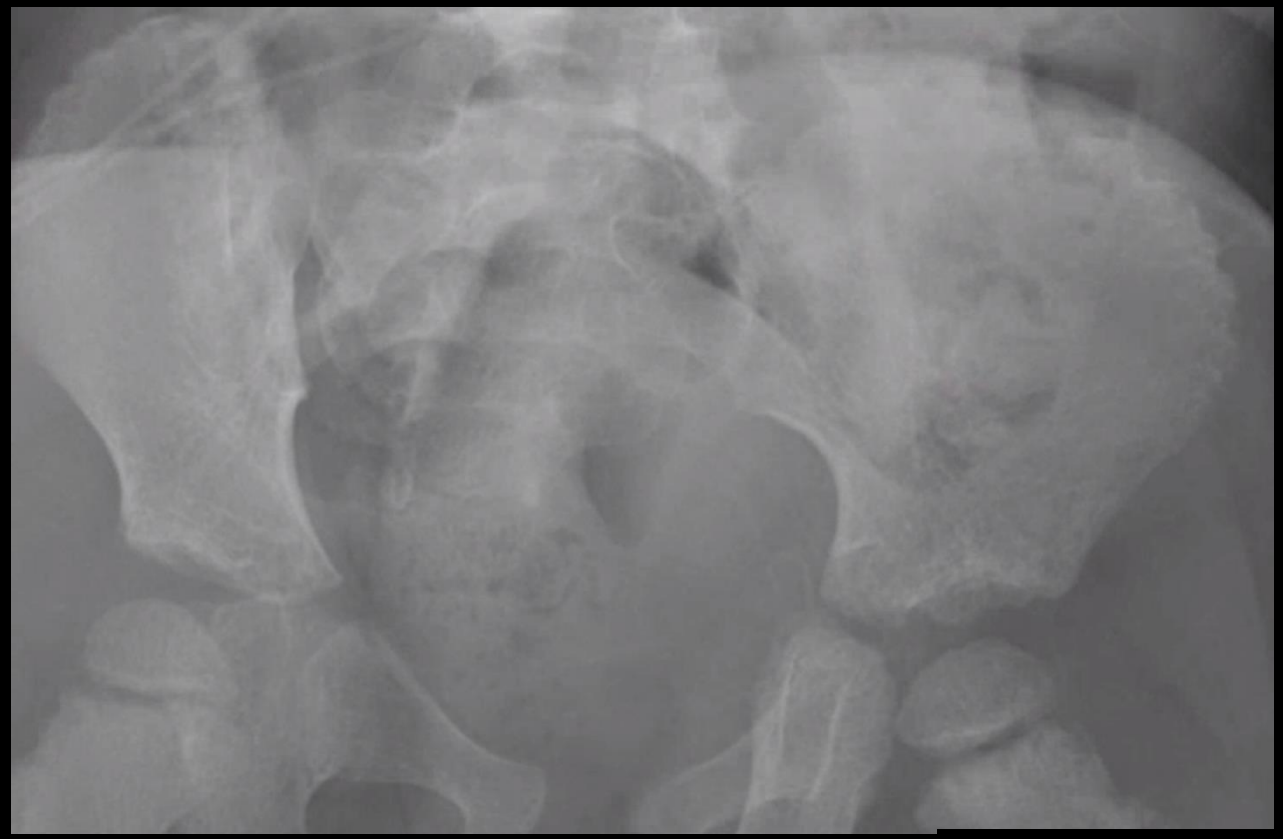
X-ray femur and hips, left (unlabeled)

Femur, AP view

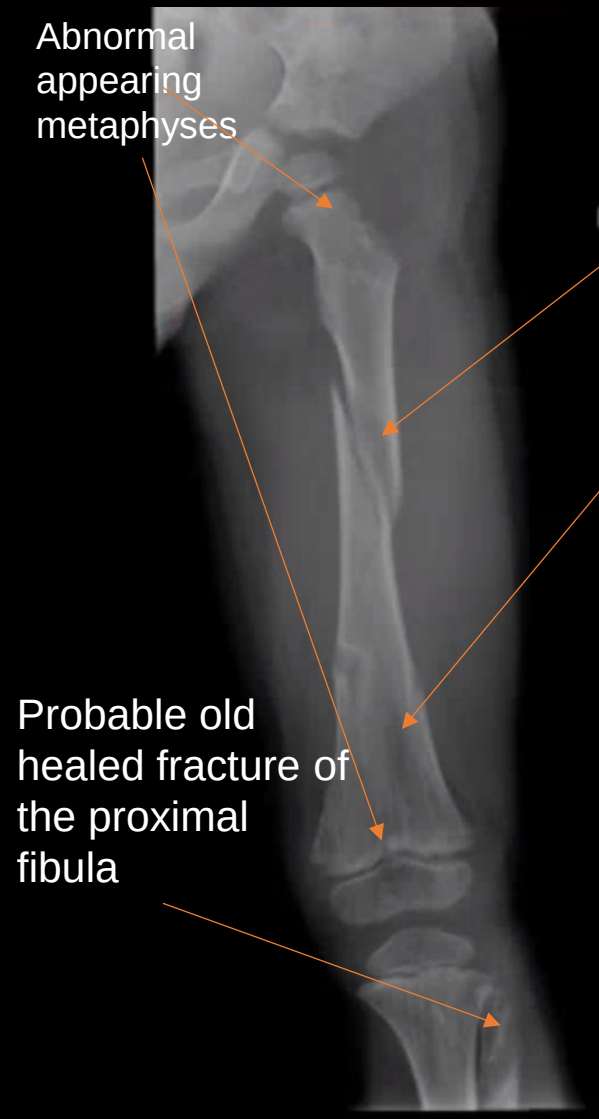


Hips, AP view



X-ray femur and hips, left (labeled)

AP view



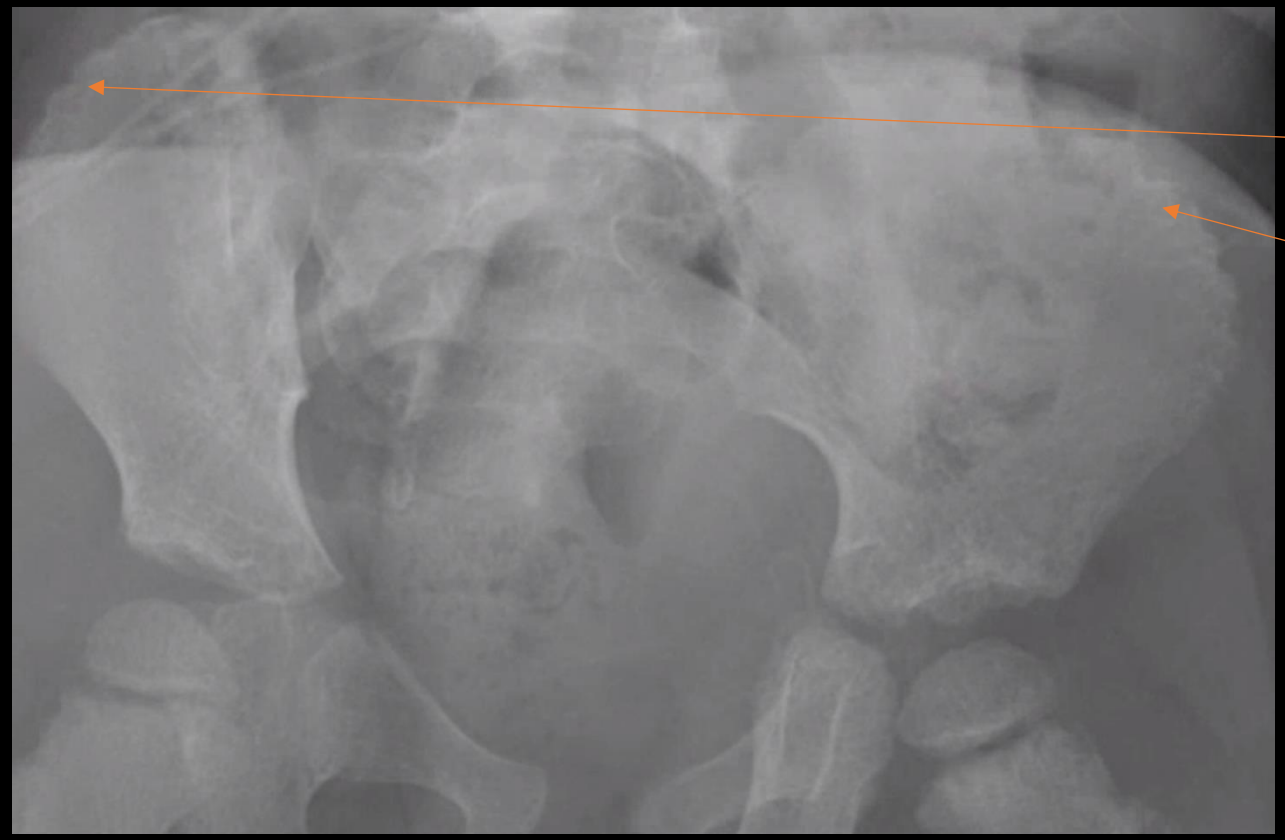
Abnormal appearing metaphyses

Spiral fracture at proximal to mid-diaphysis of left femur

Abnormally vertical lucencies in distal diaphysis

Probable old healed fracture of the proximal fibula

Hips, AP view



Unusual irregular appearing iliac crests bilaterally

Spiral fracture with old fractures and generally irregular appearing bone in femur and in iliac crests concerning for non-accidental trauma or metabolic bone disorder or cancer

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ACR Appropriateness Criteria

Chest X-ray was performed with the femur/hip X-rays and showed bony irregularities on bilateral humeri so full skeletal survey was initiated

This imaging modality was ordered by the pediatric inpatient team

Variant 3: **Child with one or more of the following: neurologic signs or symptoms, apnea, complex skull fracture, other fractures, or injuries highly suspicious for child abuse. Initial imaging evaluation.**

Radiologic Procedure	Rating	Comments	RRL*
X-ray skeletal survey	9		☢☢☢
CT head without IV contrast	9	Use this procedure in the emergent setting.	☢☢☢
MRI head without IV contrast	8	This procedure typically performed in the nonemergent setting.	O
MRI cervical spine without IV contrast	8	Consider this procedure at the time of MRI brain imaging.	O
MRI complete spine without IV contrast	5		O
Tc-99m bone scan whole body	5	This procedure is used as a problem-solving study rather than first-line.	☢☢☢☢
MRI head without and with IV contrast	3		O
MRI cervical spine without and with IV contrast	2		O
MRI complete spine without and with IV contrast	2		O
CT head with IV contrast	1		☢☢☢
CT head without and with IV contrast	1		☢☢☢☢

Rating Scale: 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate

*Relative Radiation Level

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Complete Skeletal Survey

APPENDICULAR SKELETON
Humeri (AP)
Forearms (AP)
Hands (PA)
Femurs (AP)
Lower legs (AP)
Feet (PA or AP)

AXIAL SKELETON
Thorax (AP and lateral), to include ribs* and thoracic and upper lumbar spine
Pelvis (AP), to include the mid lumbar spine
Lumbosacral spine (lateral)
Cervical spine (AP and lateral)
Skull (frontal and lateral)

*The addition of both oblique projections to the anteroposterior (AP) view of the rib cage may increase the yield of rib fractures.

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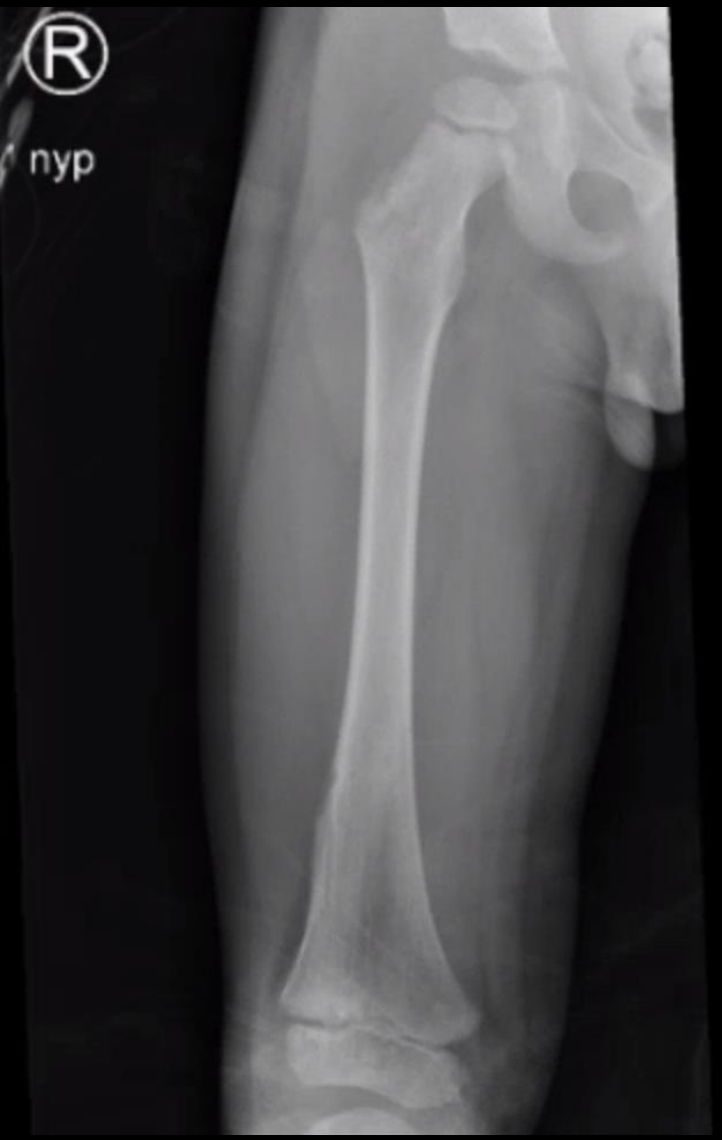
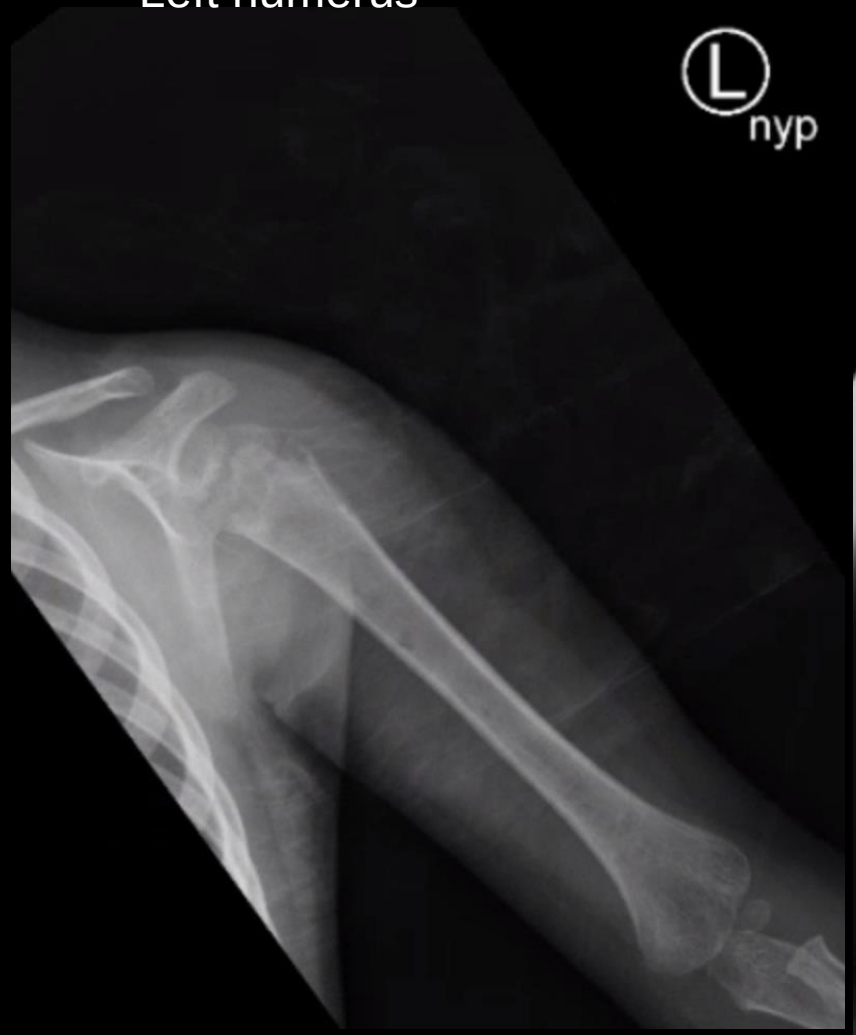
Skeletal survey (unlabeled)

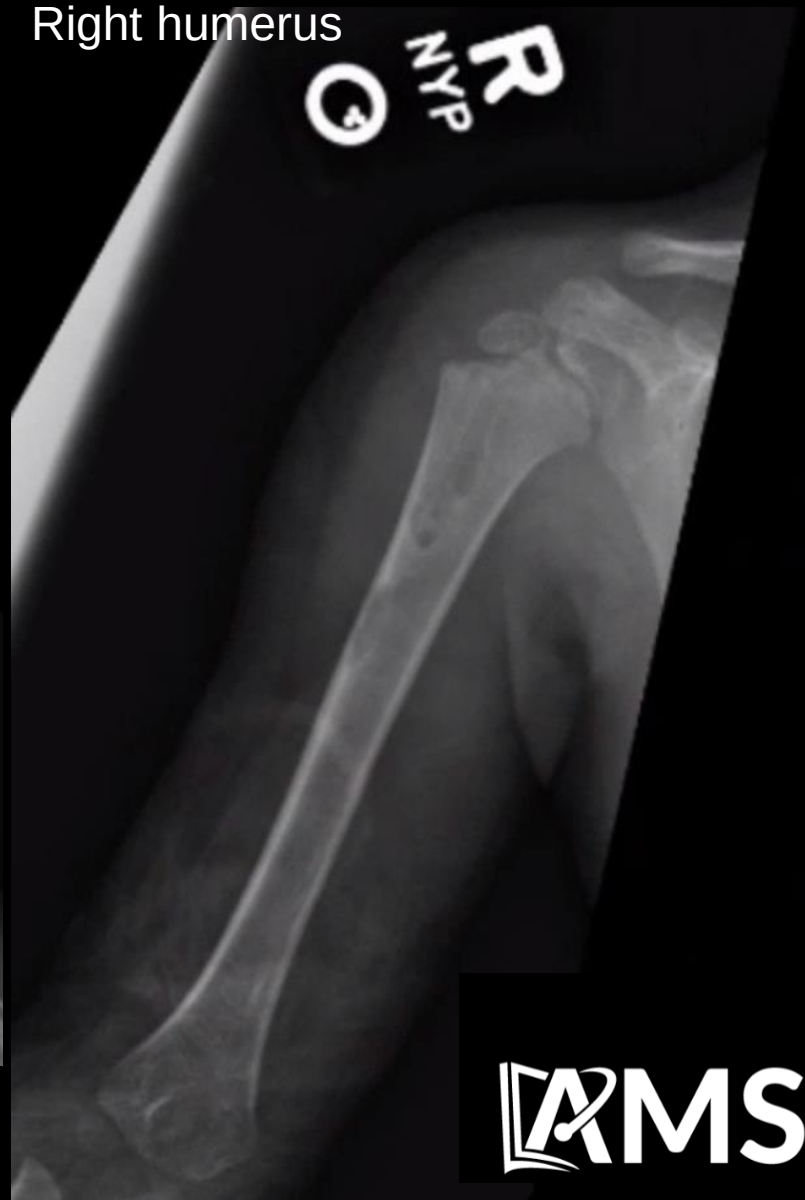
R femur x-ray, AP view



Left humerus

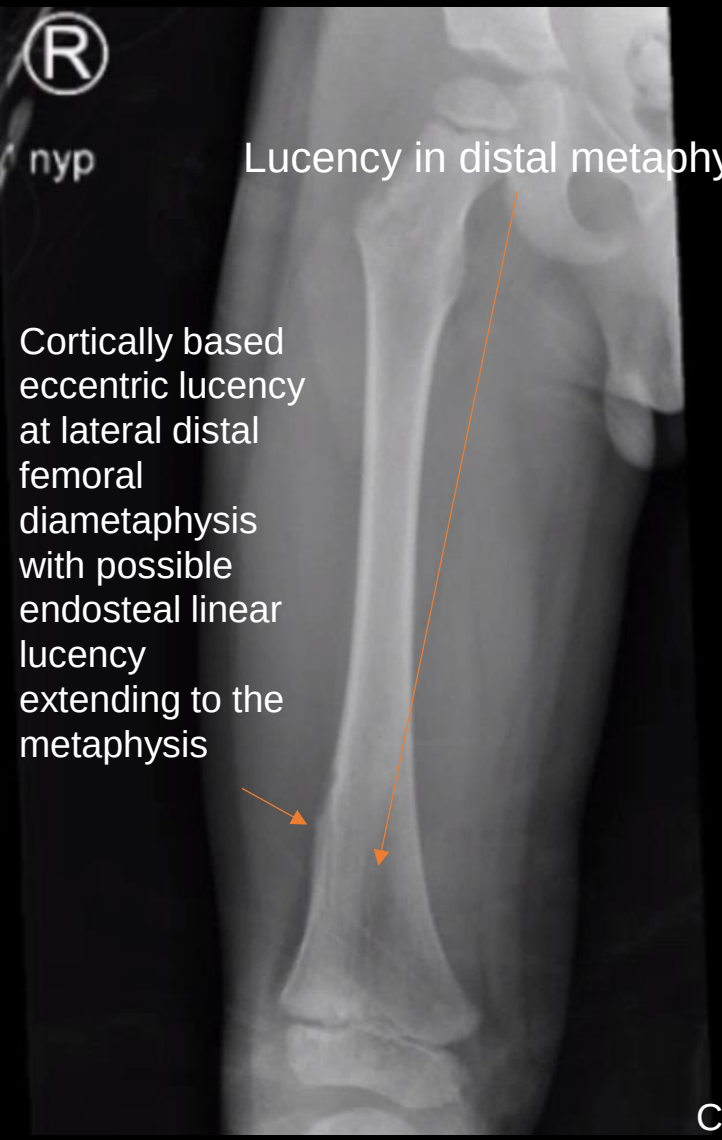


Right humerus



Skeletal survey (labeled)

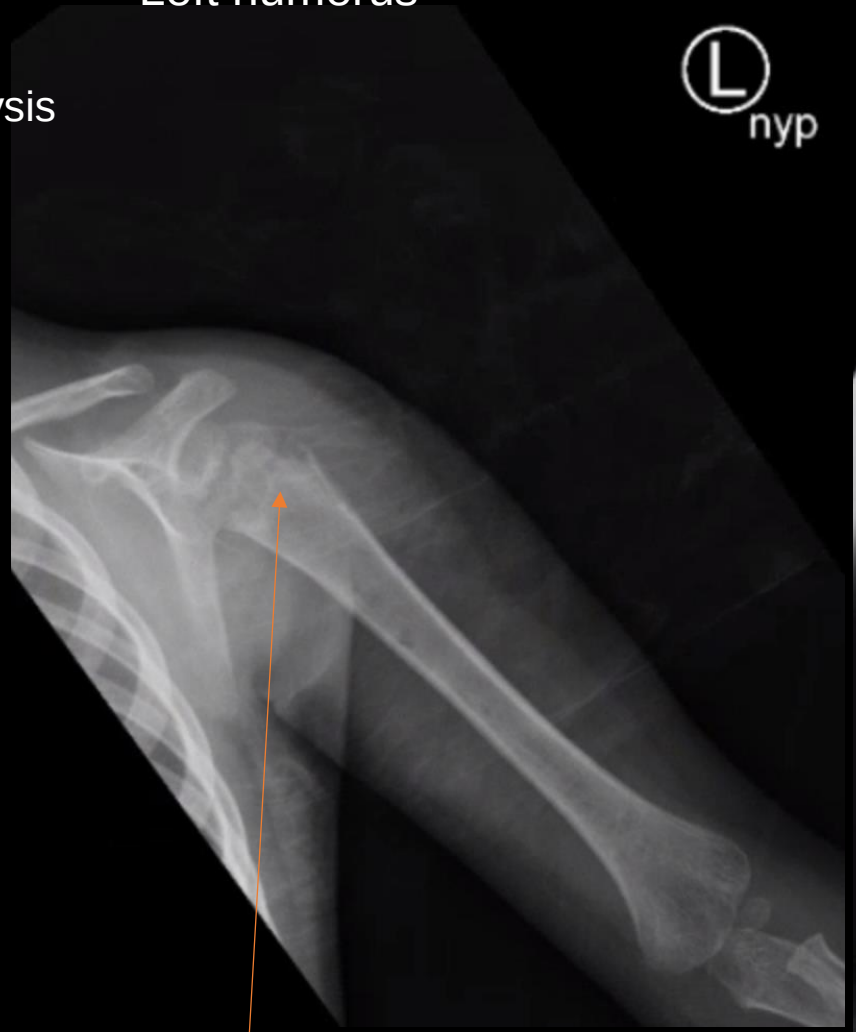
R femur x-ray, AP view



Lucency in distal metaphysis

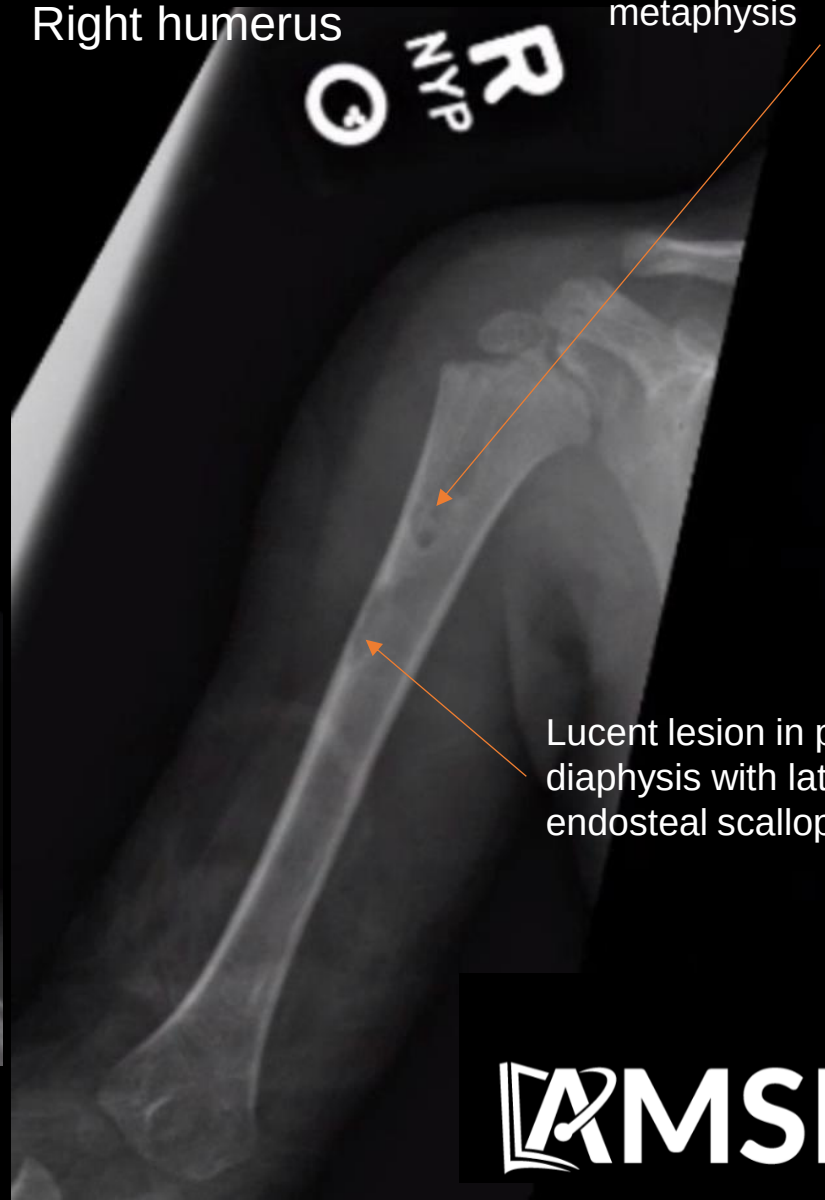
Cortically based eccentric lucency at lateral distal femoral diametaphysis with possible endosteal linear lucency extending to the metaphysis

Left humerus



Contour abnormality of lateral portion of proximal metaphysis

Right humerus



Well-demarcated longitudinally oriented lucencies in proximal metaphysis

Lucent lesion in proximal diaphysis with lateral endosteal scalloping

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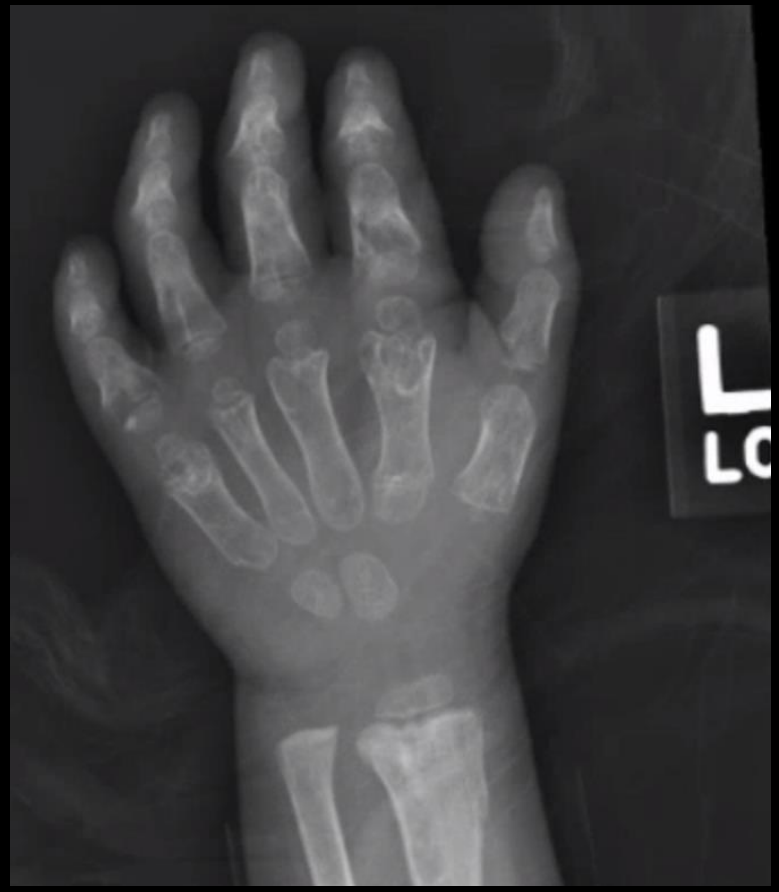
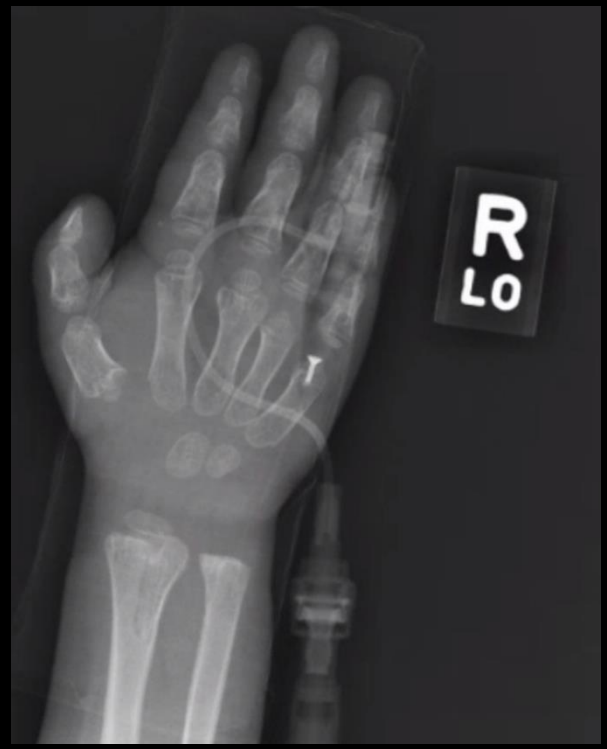
Skeletal survey continued (unlabeled)

L hand x-ray



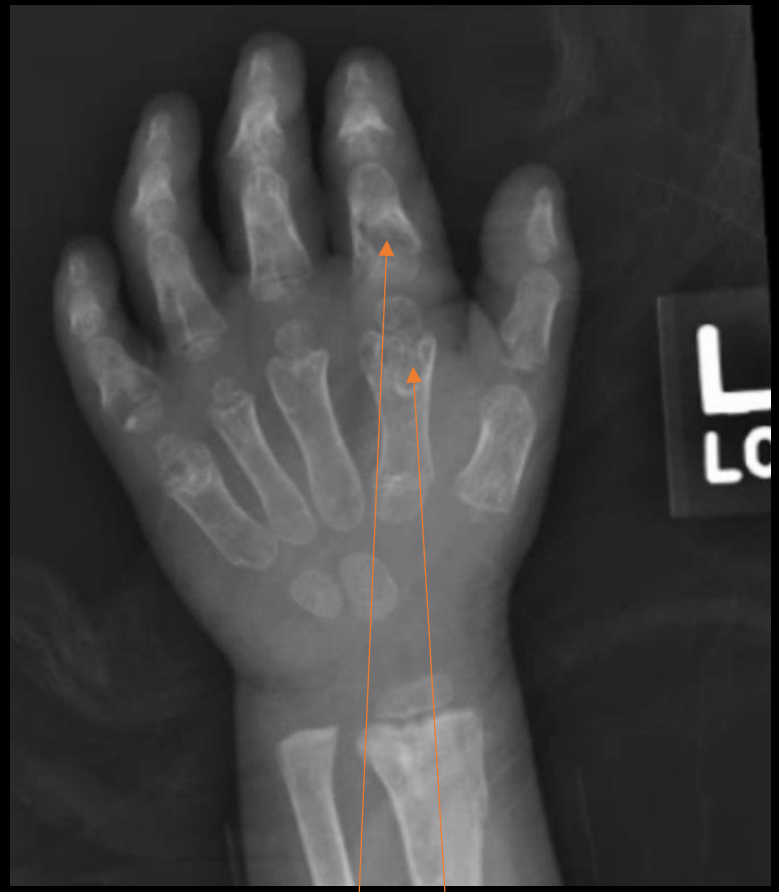
L foot x-ray limited by cast, Skull and spine x-ray unremarkable



Skeletal survey continued (labeled)

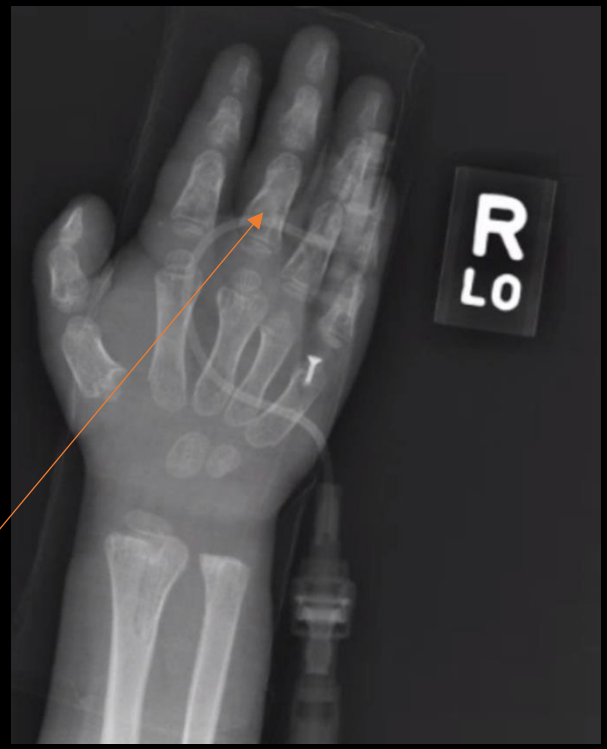
L foot x-ray limited by cast, Skull and spine x-ray unremarkable

L hand x-ray



Multiple radiolucent lesions concerning for enchondromas

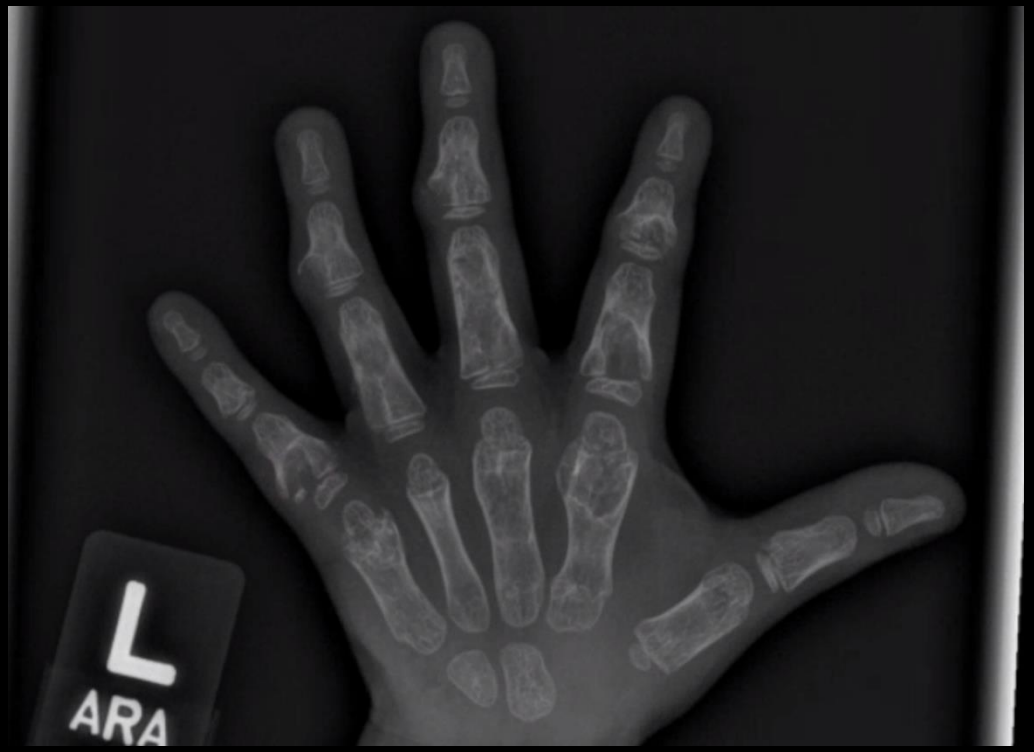
Right hand x-ray



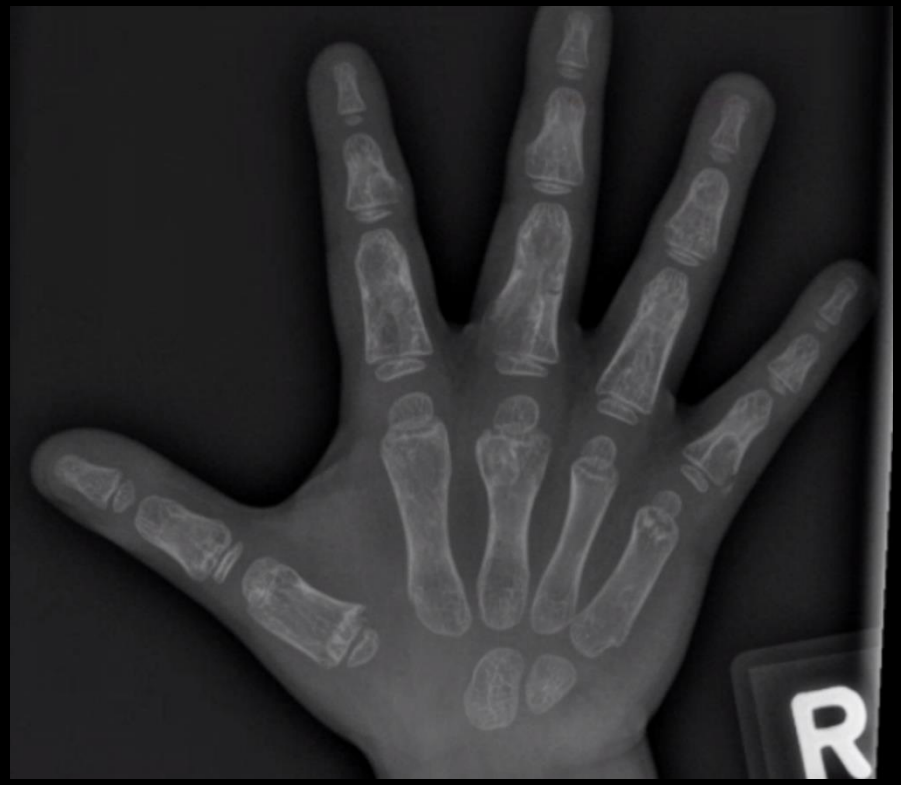
Orthopedics Follow-up Images @ age 5 (unlabeled)

Parents concerned about deformity in Left ring and middle finger w/o any functional deficits

L hand X-ray AP view



R hand X-ray AP view



Orthopedics Follow-up Images @ age 5 (labeled)

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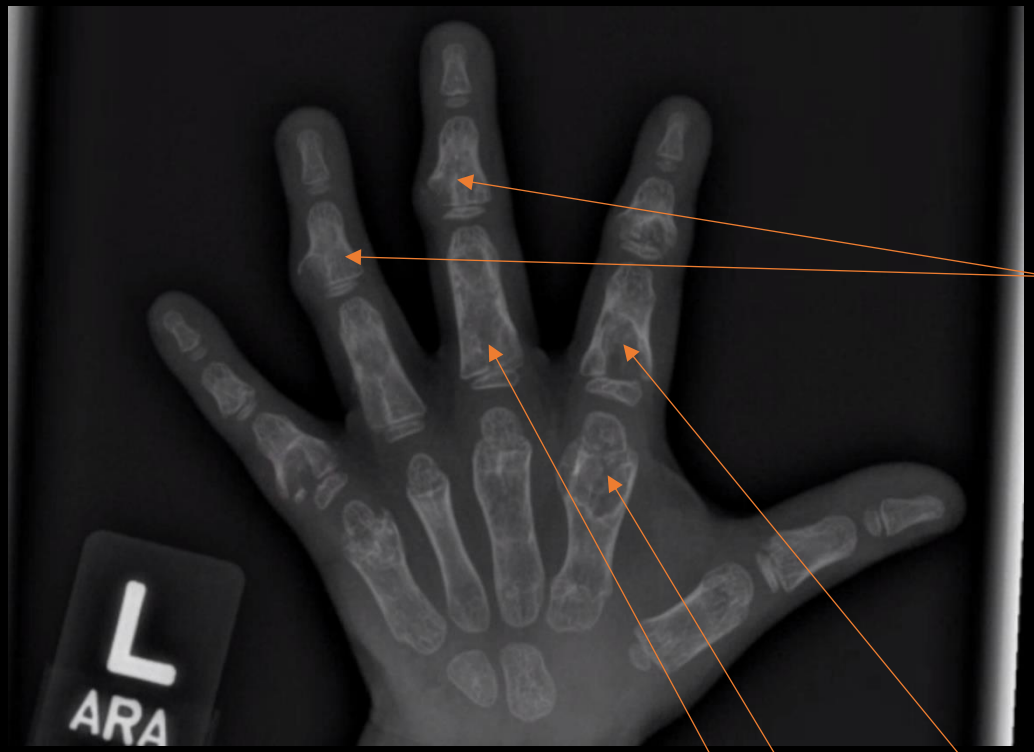
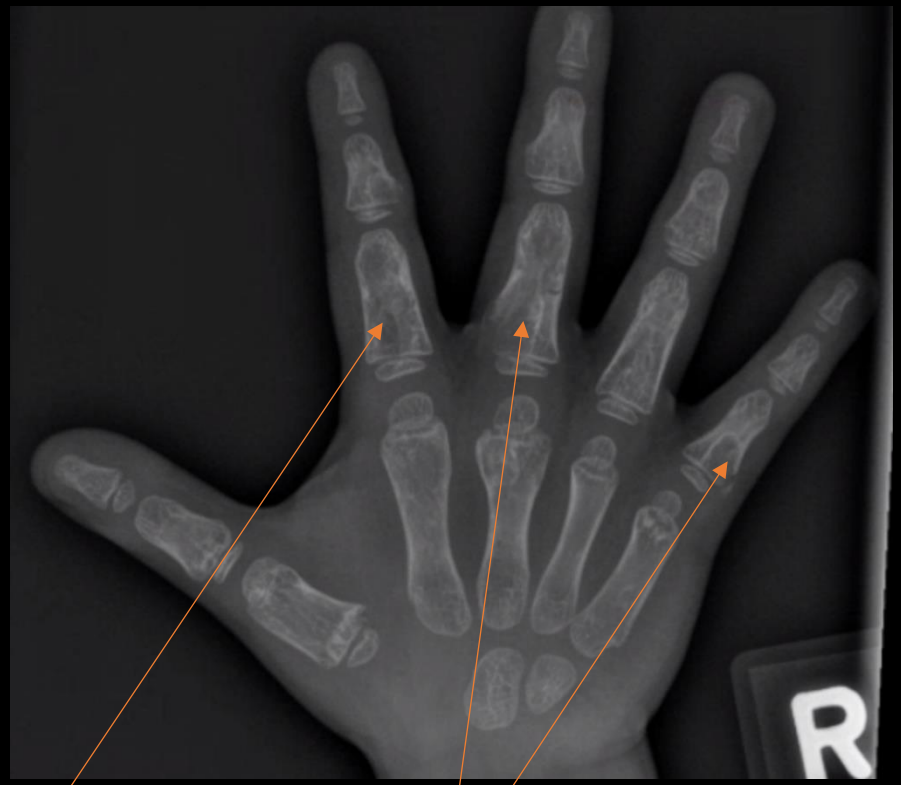
L hand X-ray AP view



Most prominent soft tissue expansion present in L 3rd and 4th middle phalanges

R hand X-ray AP view



Increased expansile lucency of bones of hand, L>R but no fractures

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Final Dx:

Multiple Enchondromatosis, specifically Ollier disease

Case Timeline

- Age 3: Initial evaluation
  - Spiral fracture is almost always pathognomonmic for non-accidental trauma (NAT)
  - Yet, given family dynamics and presence of multiple of odd bone structures and radiolucencies, NAT was deemed less likely (as family dynamics were good), metabolic bone disorder/cancer more likely
  - Then cancer was deemed less likely given normal LDH, uric acid, WBC and other inflammatory markers (ESR/CRP)
    - So metabolic bone disorder of excess production was considered most likely
  - Presence of multiple painless radiolucent lesions, involving hands/feet, causing a pathologic spiral femur fracture and lesions primarily within metaphysis → most likely diagnosis being multiple enchondromatosis, specifically Ollier disease due to lack of hemangiomas
- Age 3: Left femur spica cast application at age 3 with subsequent yearly follow-up to confirm healing of the spiral fracture
- Age 5: worsening enchondromas in hand, though no functional difficulty
- **Important point of case**: enchondromatosis should be considered even when a patient presents with pathologic fracture consistent with non-accidental trauma

Case Discussion

Enchondroma = nodule/island of cartilage tissue sequestered within bone tissue

- Cartilage arises from growth plate region

Etiology of enchondromas

- Abnormal proliferation of chondrocytes – seen in Ollier disease
- Chondrocytes growth abnormally in specific directions leading to exophytic chondromas - metachondromatosis
- Inability to reabsorb cartilage after enchondral ossification – seen in enchondrodysplasia

Radiographic findings of enchondromas – radiolucent tissue within radiopaque bone tissue

- Radiographically difficult to differentiate from low-grade chondrosarcoma (but clinically, sarcomas cause pain while enchondromas don't cause pain unless they cause a pathologic fracture)
- X-ray and CT findings: narrow zone of transition with sharp margins, no periosteal reaction or soft tissue mass
- MRI findings (used to confirm diagnosis): well-circumscribed masses that can replace the marrow. Enchondromas appear with low signal on T1, no contrast enhancement with Gad, and background high signal on T2

Location: small and large tubular bones (hands/feet, femur, tibia, humerus), rarely in axial skeleton

Ollier Disease

Ollier disease = Multiple enchondromas + mainly metaphyseal location (also diaphysis) + mainly involving hands/feet (tubular bones), long bones (femur, humerus, tibia/fibula)

Maffuci syndrome = All the same findings of Ollier disease + multiple hemangiomas

Ollier disease:

-Appear in first decade of life, painless cartilage enchondromas which may cause pathologic fractures

-Non-hereditary, spontaneous mutations in 1 of 3 genes, PTH1R, IHD1, and IDH2

-Radiologically – multiple enchondromas + vertically oriented radiolucent streaks going from metaphysis to epiphysis (column of cartilage)

Ollier Disease continued

-Malignant transformation

- -5-50% risk of malignant transformation of enchondromas in Ollier disease to become chondrosarcoma
- -If transformation suspected, biopsy needed (signs are inc pain and size or thinning cortical bone)

-Otherwise, long term follow-up required for surveillance for malignant transformation

-Increased risk of developing pancreatic neoplasms, gliomas, or juvenile ovarian granulosa cell tumors

Ollier Disease continued

-Treatment:

-if minimal involvement with no deformities, limb discrepancies or other causes of functional impairment → no surgery, follow-up with pediatrician or orthopedist

-If extensive involvement with functional impairment → corrective surgery to remove enchondromas

- Prognosis - normal lifespan though most important prognostic factor is development of pathologic fractures and malignant transformation

References:

American College of Radiology. ACR Appropriateness Criteria. Acute Hip Pain-Suspected Fracture. Revised 2018

American College of Radiology. ACR Appropriateness Criteria. Suspected Physical Abuse–Child Revised 2016.

El Abiad JM, Robbins SM, Cohen B, et al. Natural History of Ollier Disease and Maffucci Syndrome: Patient Survey and Review of Clinical Literature. *Am J Med Genetics Part A*. 2020; 182(5): 1093-1103.

Ollier disease. Retrieved September 14, 2020, from https://rarediseases.info.nih.gov/diseases/7251/ollier-disease.

https://radiopaedia.org/articles/enchondroma?lang=us

https://radiopaedia.org/articles/enchondromatosis?lang=us

Wang JP, Xu ZY, Bao ZQ, et al. Ollier disease: two case reports and a review of the literature. *Am J Transl Res*. 2018;10(11):3818-3826. Published 2018 Nov 15.