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.

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Radiography hip</td>
<td>Usually Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
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<tr>
<td>Radiography pelvis</td>
<td>Usually Appropriate</td>
<td>⭐⭐⭐⭐</td>
</tr>
<tr>
<td>Radiography pelvis and hips</td>
<td>Usually Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
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<tr>
<td>CT pelvis and hips with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
</tr>
<tr>
<td>CT pelvis and hips without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐⭐⭐</td>
</tr>
<tr>
<td>CT pelvis and hips without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
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<tr>
<td>MRI pelvis and affected hip without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐</td>
</tr>
<tr>
<td>MRI pelvis and affected hip without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐</td>
</tr>
<tr>
<td>Bone scan hips</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
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<tr>
<td>US hip</td>
<td>Usually Not Appropriate</td>
<td>⭐⭐⭐⭐⭐</td>
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</table>

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

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

*Relative Radiation Level

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

<table>
<thead>
<tr>
<th>APPENDICULAR SKELETON</th>
</tr>
</thead>
<tbody>
<tr>
<td>Humeri (AP)</td>
</tr>
<tr>
<td>Forearms (AP)</td>
</tr>
<tr>
<td>Hands (PA)</td>
</tr>
<tr>
<td>Femurs (AP)</td>
</tr>
<tr>
<td>Lower legs (AP)</td>
</tr>
<tr>
<td>Feet (PA or AP)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>AXIAL SKELETON</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thorax (AP and lateral), to include ribs* and thoracic and upper lumbar spine</td>
</tr>
<tr>
<td>Pelvis (AP), to include the mid lumbar spine</td>
</tr>
<tr>
<td>Lumbosacral spine (lateral)</td>
</tr>
<tr>
<td>Cervical spine (AP and lateral)</td>
</tr>
<tr>
<td>Skull (frontal and lateral)</td>
</tr>
</tbody>
</table>

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

R femur x-ray, AP view

Left humerus

Right humerus
Skeletal survey (labeled)

R femur x-ray, AP view

Left humerus

Right humerus

Contour abnormality of lateral portion of proximal metaphysis

Well-demarcated longitudinally oriented lucencies in proximal metaphysis

Lucency in distal metaphysis

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

Lucent lesion in proximal diaphysis with lateral endosteal scalloping
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 hand x-ray

Multiple radiolucent lesions concerning for enchondromas

Right hand x-ray

L foot x-ray limited by cast, Skull and spine x-ray unremarkable
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
Parents concerned about deformity in Left ring and middle finger w/o any functional deficits.

Orthopedics Follow-up Images @ age 5 (labeled)

L hand X-ray AP view

R hand X-ray AP view

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

Most prominent soft tissue expansion present in L 3rd and 4th middle phalanges
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
American College of Radiology. ACR Appropriateness Criteria. Acute Hip Pain-Suspected Fracture. Revised 2018
https://radiopaedia.org/articles/enchondroma?lang=us
https://radiopaedia.org/articles/enchondromatosis?lang=us