HPI: 43 y/o F presents with acute on chronic knee pain

Jonathan Gan, MS4
Case Western Reserve University School of Medicine

Sofija Conic, MD, PGY-2
Cleveland Clinic Foundation

Deborah Brahee, MD
Cleveland Clinic Foundation
Patient Presentation

- **HPI**: 43 y/o female presents to clinic with bilateral 5/10 shooting knee pain for past 2-3 weeks. Has had on-and-off pain for 30+ years, but recently unable to bear weight and perform ADLs.
- **ROS**: Numbness and tingling in knees. No fevers, chills, or rashes.
- **PMHx**: IBS, chronic pain syndrome, asthma, fibromyalgia, vitamin D deficiency
- **PSHx**: knee and ankle “bone spurs” s/p excision, osteosarcoma of right rib s/p resection (no known follow-up), cholecystectomy, hysterectomy w/ bilateral salpingectomy
- **FHx**: diabetes, HTN, COPD, breast and ovarian cancer (maternal aunt and grandmother)
Patient Presentation

Pertinent physical exam findings:

- General: A&O x3, no acute distress
- Cardiac: RRR, normal S1 S2
- Musculoskeletal:
  - Mild TTP in right posterior calf
  - No knee point tenderness or effusion
  - Full ROM, negative Lachman, posterior drawer, and varus/valgus laxity in bilateral knees
  - Lower extremity motor, sensation, and reflexes intact bilaterally
  - Negative straight-leg raise bilaterally
  - DP and PT pulses 2+ bilaterally
  - Full hip ROM bilaterally
What Imaging Should We Order?
Select the applicable ACR Appropriateness Criteria

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Radiography knee</td>
<td>Usually Appropriate</td>
<td></td>
</tr>
<tr>
<td>Image-guided aspiration knee</td>
<td>Usually Not Appropriate</td>
<td>Varies</td>
</tr>
<tr>
<td>CT arthrography knee</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
<tr>
<td>CT knee with IV contrast</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
<tr>
<td>CT knee without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
<tr>
<td>CT knee without IV contrast</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
<tr>
<td>MR arthrography knee</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
<tr>
<td>MRI knee without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
<tr>
<td>MRI knee without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
<tr>
<td>Bone scan knee</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
<tr>
<td>US knee</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
<tr>
<td>Radiography hip ipsilateral</td>
<td>Usually Not Appropriate</td>
<td></td>
</tr>
</tbody>
</table>

Frontal, lateral, and merchant views were ordered.

Patient was referred to orthopedic surgery.
Findings (unlabeled)
Findings (labeled)

Undertubulation of metadiaphyses

Exostoses
Final Dx:

Multiple Hereditary Exostoses
Case Discussion

Overview:

- Characterized by 2+ skeletal exostoses
  - Common locations include distal femur and proximal humerus
- Autosomal dominant
  - $EXT1$ and $EXT2$ tumor suppressor gene mutations
  - Incomplete penetrance
  - Males > females
- Typically present in 2$^{nd}$ decade
Case Discussion

Imaging findings:

- X-ray
  - Multiple osteochondromas with variable skeletal distribution
  - Broadened shaft (undertubulation) at ends of long bones
  - Malignant transformation (lifelong risk as high as 25%)
    - New cortical irregularity
    - Continued growth after skeletal maturity has been reached
    - Bony destruction
    - Large soft tissue component

- MRI
  - Can be considered as follow-up imaging in cases of soft tissue impingement and concern for malignant transformation
Case Discussion

Exostoses:

- Benign growth of bone extending outwards from surface
- With cartilage cap = osteochondroma
- Growth patterns
  - Sessile or pedunculated
  - At the metaphysis and projecting away from epiphysis
  - Broadening of metaphysis from which it arises
  - Variable appearance of cartilage cap (thin/thick, regular/irregular)
Case Discussion

**Tubulation:**

- Normal adult long bones should have **diaphyseal narrowing**
- This process occurs due to (1) periosteal bone resorption and (2) endosteal bone formation at the metaphysis during longitudinal bone growth

<table>
<thead>
<tr>
<th>Undertubulation</th>
<th>Overtubulation</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Wide, broad appearance of metadiaphyses (Erlenmeyer flask deformity)</td>
<td>• Narrow, gracile appearance of metadiaphyses</td>
</tr>
<tr>
<td>• Can be seen in:</td>
<td>• Can be seen in:</td>
</tr>
<tr>
<td>• Gaucher’s disease</td>
<td>• Neurofibromatosis</td>
</tr>
<tr>
<td>• Thalassemia</td>
<td>• Osteogenesis imperfecta</td>
</tr>
<tr>
<td>• Multiple hereditary exostoses</td>
<td>• Immobilization</td>
</tr>
<tr>
<td></td>
<td>• Muscular dystrophy</td>
</tr>
</tbody>
</table>
Case Discussion

Management and prognosis:

• Monitoring for malignant transformation
  • Spine, scapula, pelvis, and proximal femur are more commonly associated with malignant transformation

• Surgical excision can be considered in cases of:
  • Pain/discomfort
  • Deformity or growth disruption
  • Cosmetics
  • Malignant transformation

• Good prognosis and generally static disease in adulthood
References


