AMSER Case of the Month January 2022

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

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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

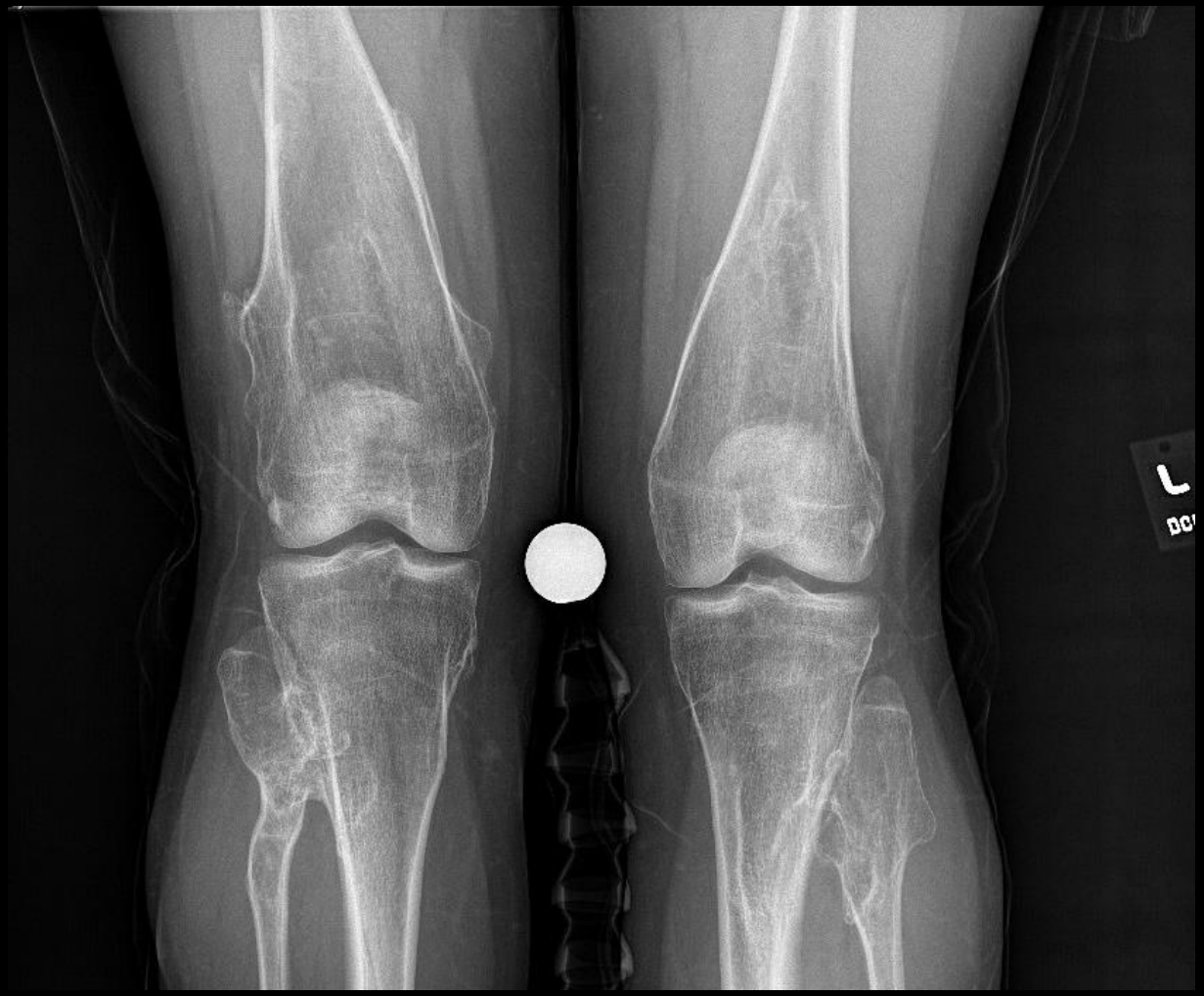
Variant 1: **Adult or child greater than or equal to 5 years of age. Chronic knee pain. Initial imaging.**

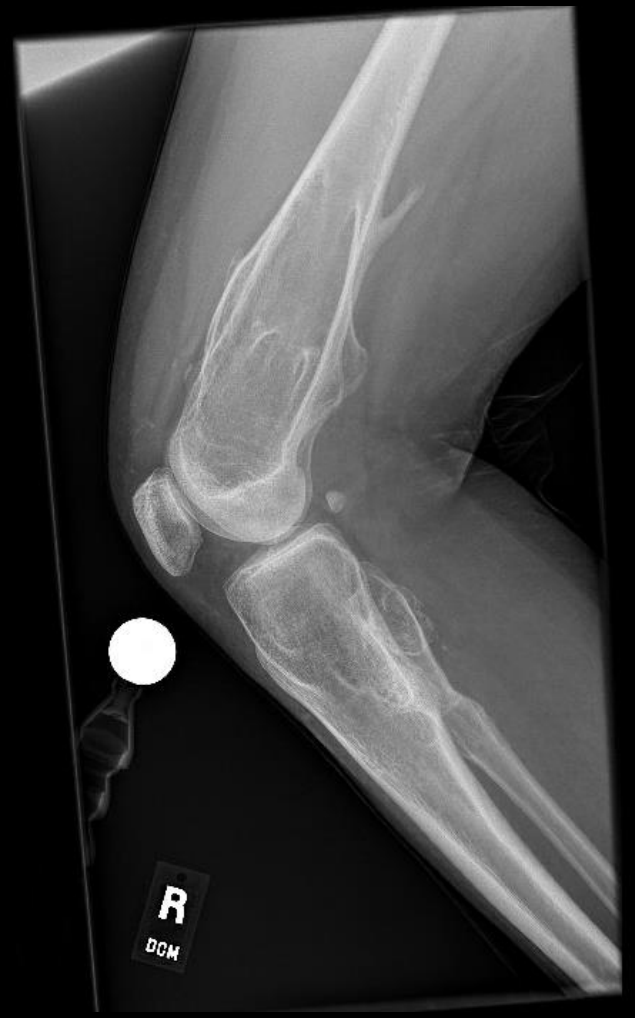
Procedure	Appropriateness Category	Relative Radiation Level
Radiography knee	Usually Appropriate	☢
Image-guided aspiration knee	Usually Not Appropriate	Varies
CT arthrography knee	Usually Not Appropriate	☢
CT knee with IV contrast	Usually Not Appropriate	☢
CT knee without and with IV contrast	Usually Not Appropriate	☢
CT knee without IV contrast	Usually Not Appropriate	☢
MR arthrography knee	Usually Not Appropriate	O
MRI knee without and with IV contrast	Usually Not Appropriate	O
MRI knee without IV contrast	Usually Not Appropriate	O
Bone scan knee	Usually Not Appropriate	☢☢☢
US knee	Usually Not Appropriate	O
Radiography hip ipsilateral	Usually Not Appropriate	☢☢☢

Frontal, lateral, and merchant views were ordered

Patient was referred to orthopedic surgery

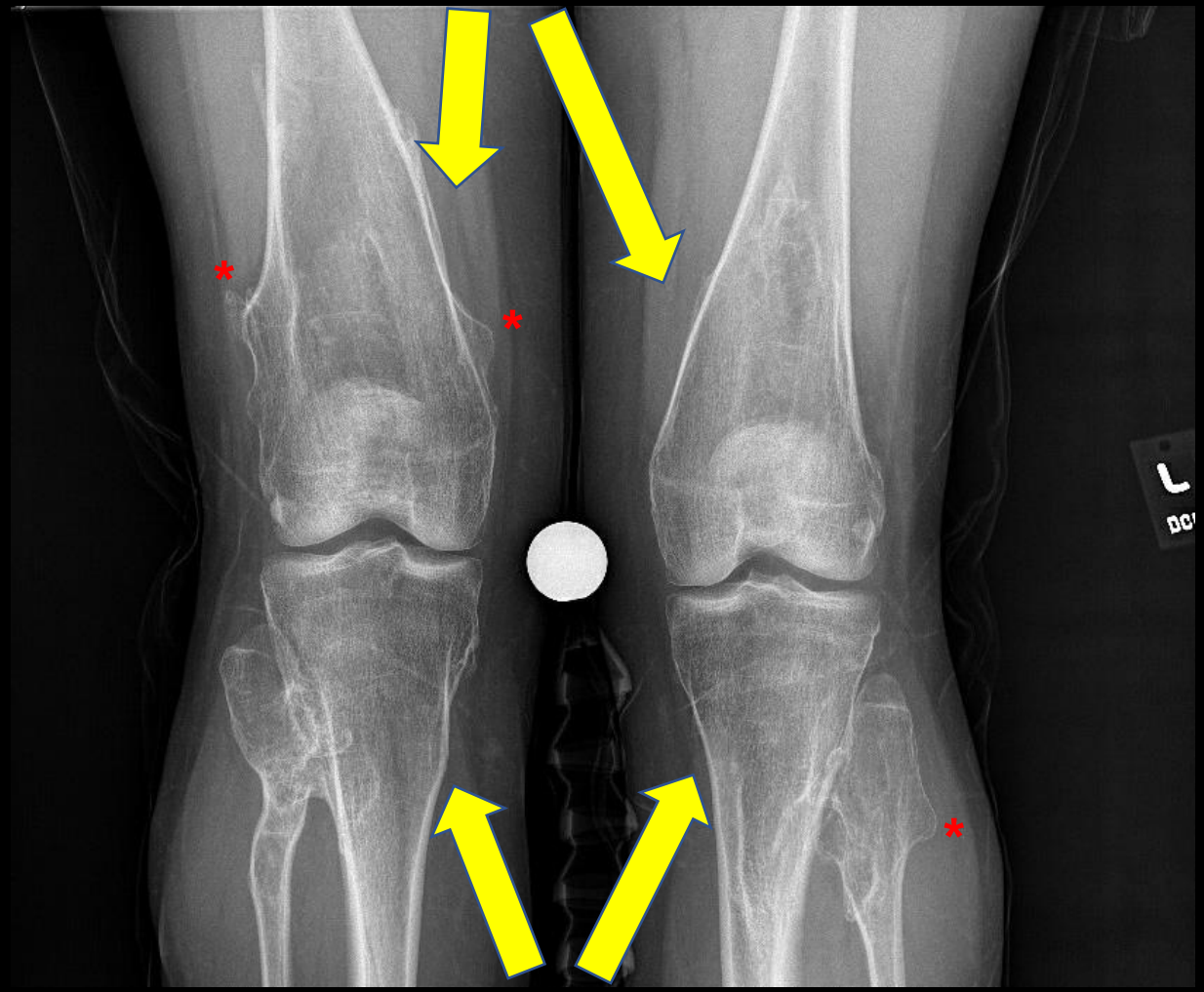
Findings (unlabeled)



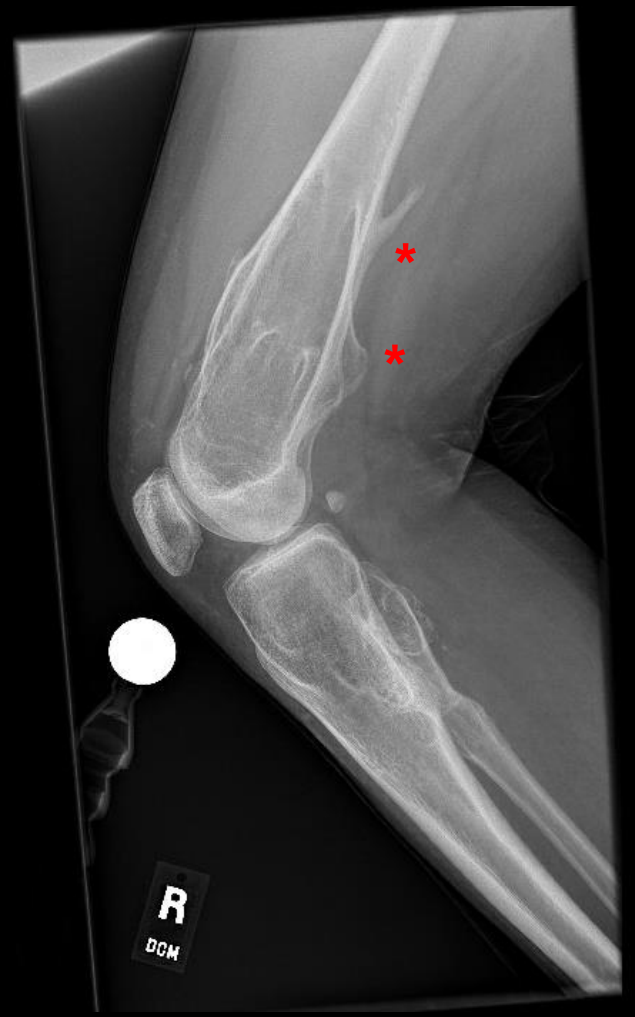


Findings (labeled)

Undertubulation of metadiaphyses



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 2nd decade

Case Discussion

Imaging findings:

- X-ray
  - Multiple osteochondromas w/ 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

Undertubulation	Overtubulation

• Wide, broad appearance of metadiaphyses (Erlenmeyer flask deformity)
• Can be seen in:
  • Gaucher's disease
  • Thalassemia
  • Multiple hereditary exostoses
	• Narrow, gracile appearance of metadiaphyses
	• Can be seen in:
	  • Neurofibromatosis
	  • Osteogenesis imperfecta
	  • Immobilization
	  • Muscular dystrophy

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

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2. D'Arienzo A, Andreani L, Sacchetti F, Colangeli S, Capanna R. Hereditary Multiple Exostoses: Current Insights. *Orthop Res Rev*. 2019;11:199-211. Published 2019 Dec 13. doi:10.2147/ORR.S183979
3. Wuyts W, Schmale GA, Chansky HA, et al. Hereditary Multiple Osteochondromas. 2000 Aug 3 [Updated 2020 Aug 6]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021.
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