RADY 403 Case Presentation

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UNC SCHOOL OF MEDICINE
Radiology

Focused patient history and workup

A 13 year old female with a known history of hereditary multiple exostoses (HME), acne, and myopia presented to the orthopedic clinic, after referral from her primary care physician, for evaluation of an enlarging and symptomatic known osteochondroma located at the left distal femur. She reported that, recently, the lesion had significantly increased in size and was more apparent. She denied any skin changes in the area, numbness, or tingling. She reported feeling discomfort in the region of the distal femur upon kneeling, but otherwise had not experienced pain in the area. She is aware of having multiple other osteochondromas of her extremities. The patient's family history is negative for known HME.

Focused patient history and workup

Physical Exam

- Well appearing and in no acute distress
- Easily palpable bony growth at the left distal medial femur. No overlying bruising or skin changes.
- Large growth palpated at the left proximal and medial humerus without TTP or skin changes.
- 2+ Patellar and Achilles reflexes bilaterally
- Normal physiologic valgus of the knees.

Review of Systems

- Reports discomfort around the knee when kneeling.
- Denies numbness/tingling
- Denies any bruising or skin changes.
- Denies any recent fevers

List of pertinent imaging studies

- AP & Lateral Femur XR
- AP & Lateral Tibia/Fibula XR
- AP & Lateral Humerus XR

Last review date: 2013

American College of Radiology
ACR Appropriateness Criteria®

Clinical Condition: **Primary Bone Tumors**

Variant 1: **Screening. First study.**

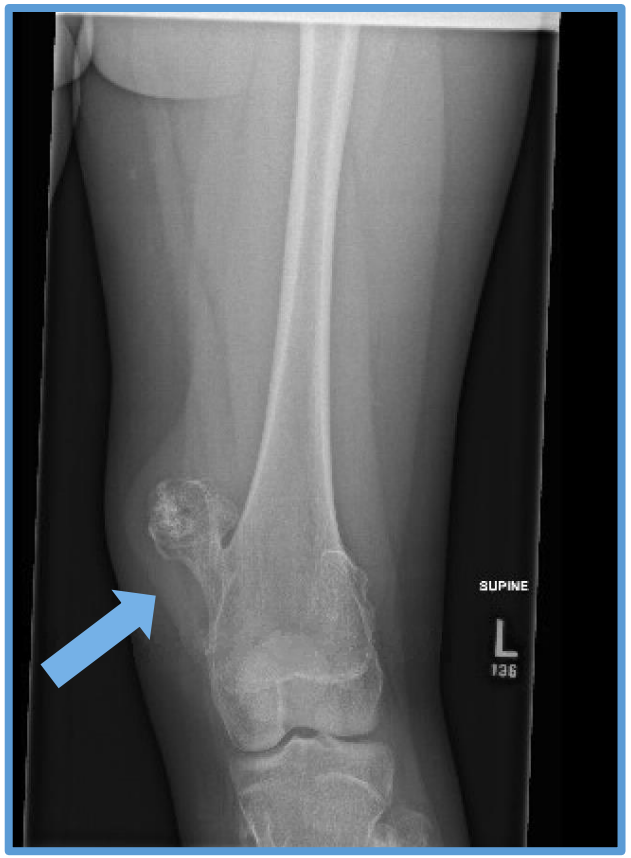
Radiologic Procedure	Rating	Comments	RRL*
X-ray area of interest	9	This procedure is absolutely required in a patient with suspected bone lesion.	Varies
US area of interest	1		O
MRI area of interest without and with IV contrast	1		O
MRI area of interest without IV contrast	1		O
Tc-99m bone scan whole body	1		☢☢☢
CT area of interest without IV contrast	1		Varies
CT area of interest with IV contrast	1		Varies
CT area of interest without and with IV contrast	1		Varies
FDG-PET/CT whole body	1		☢☢☢☢

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

*Relative Radiation Level

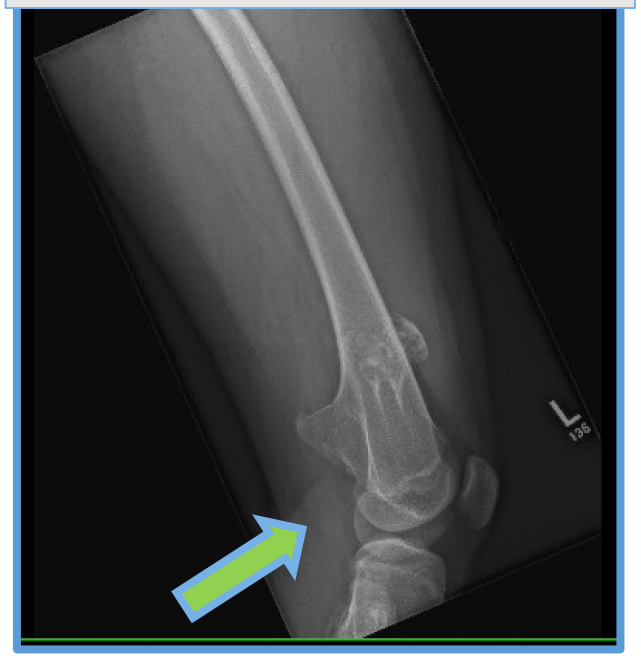
Image from: https://acsearch.acr.org/docs/69421/Narrative/

Imaging Studies: Femur XR



Left Distal Femur: AP View

Left Distal Femur: Lateral View



There is a 6.7 x 4.6 cm pedunculated mass/outgrowth with chondroid matrix projecting medially from the distal femoral metaphysis. It is lytic and sclerotic in appearance and is continuous with the medullary cavity and the cortex. There is also a 6.8 x 4.4 cm sessile mass projecting posteriorly at the distal femoral metaphysis. It is mostly lytic in appearance and also continuous with the medullary cavity and cortex. Both lesions are suggestive of osteochondroma. No pathologic fractures noted.

Normal Left Femur as reference

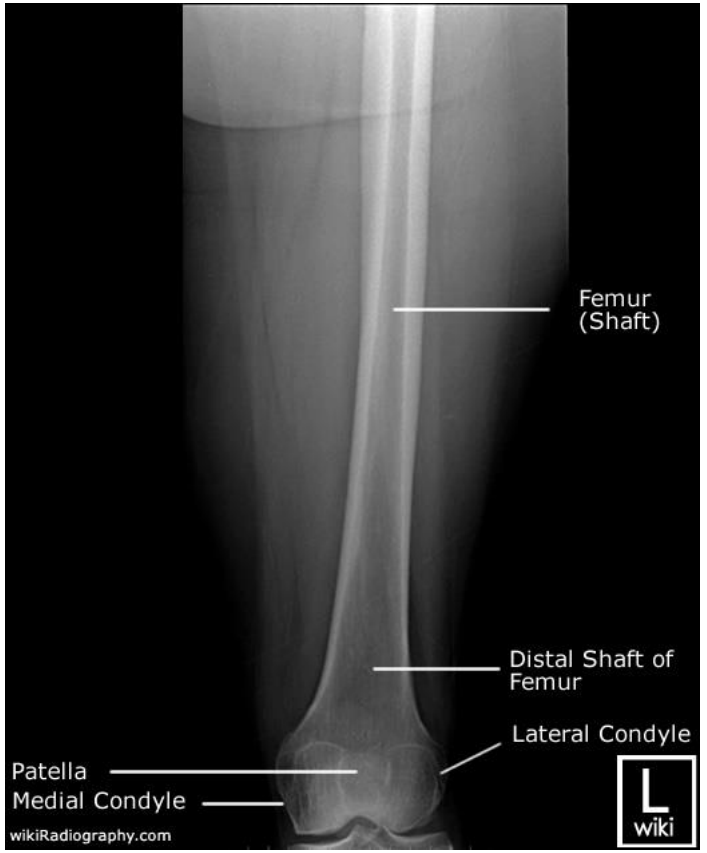


Image from: http://www.wikiradiography.net/page/Femur+Radiographic+Anatomy

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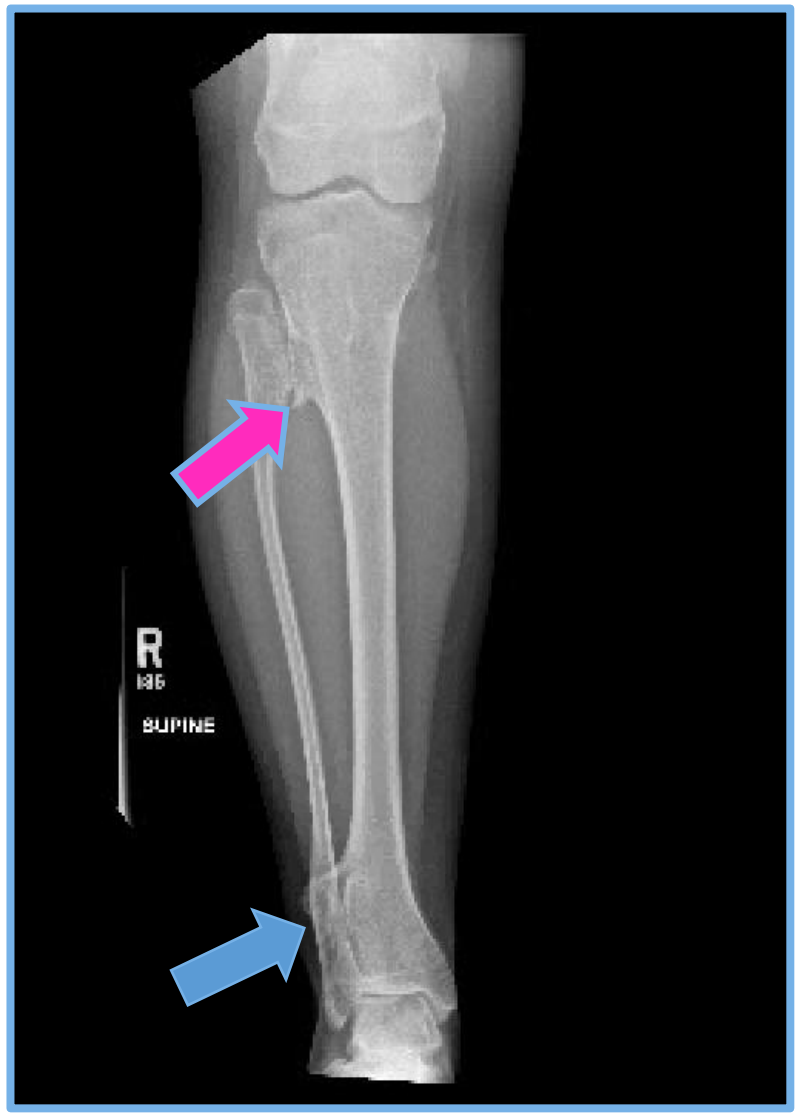
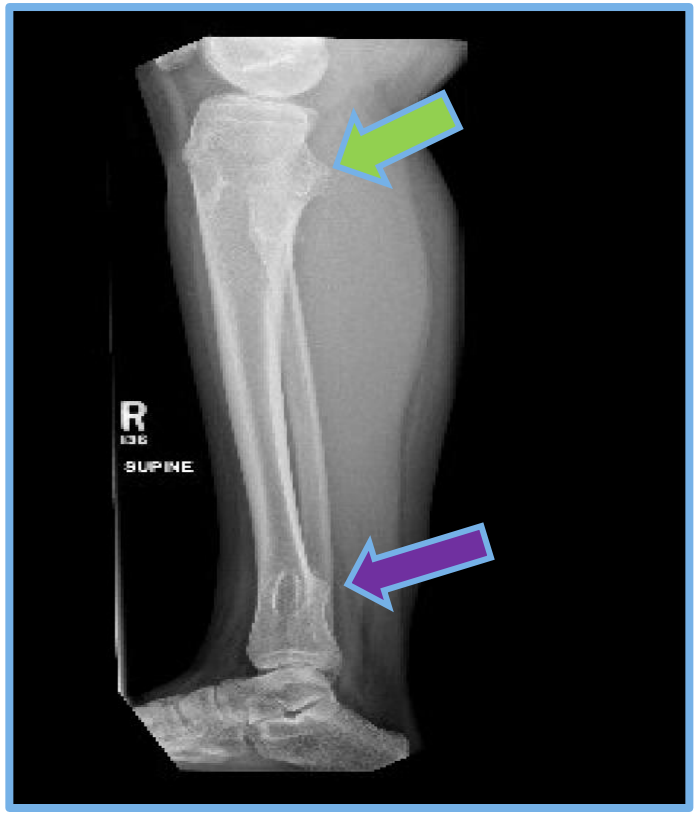
Imaging Studies: Tib Fib XR



AP view of the right tibia and fibula



Lateral View of the right Tibia and Fibula

Normal Tibia/Fibula XR as reference

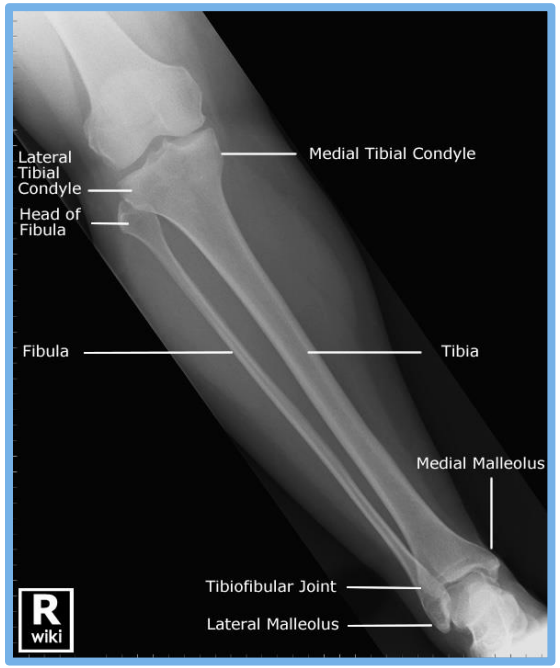
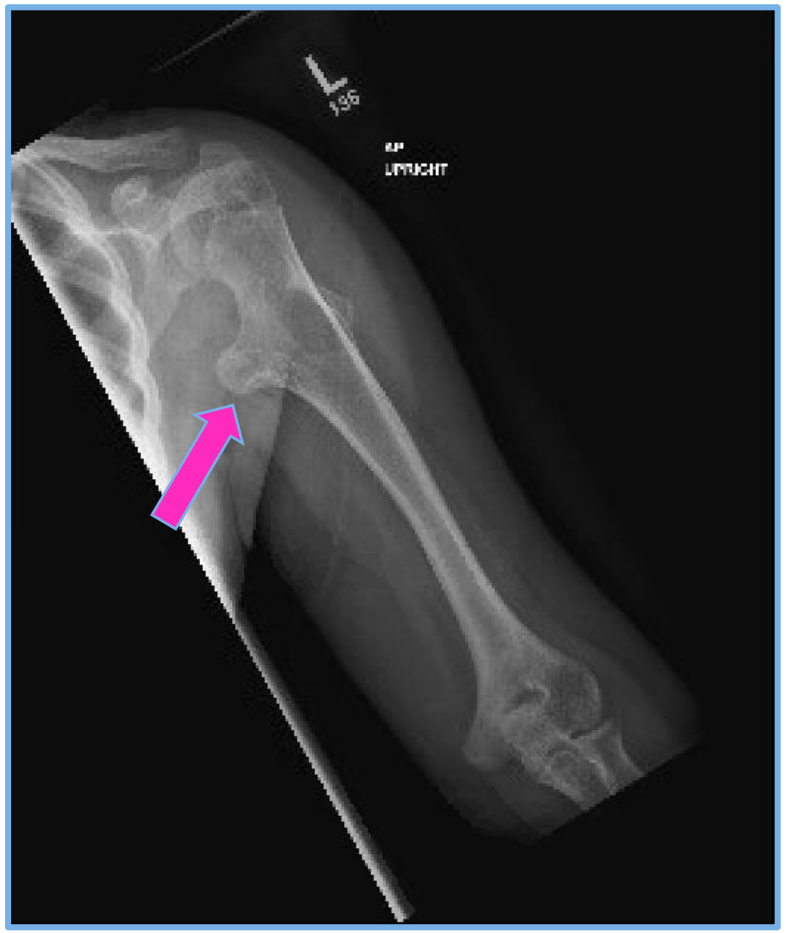


Image from:
http://www.wikiradiography.net/page/Tib%2FFib+Radiographic+Anatomy

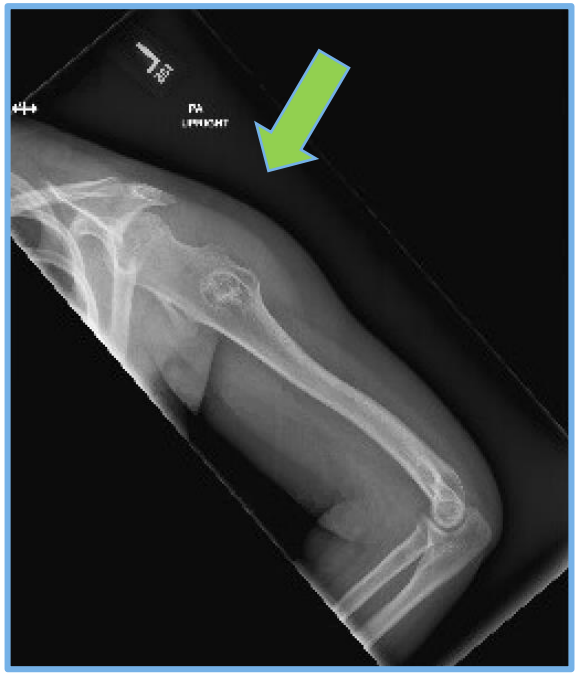
There is a 2.3 x 4.3 cm mass projecting laterally and posteriorly from the proximal tibia with involvement of the proximal fibula as well. The lesion appears to be lytic and sclerotic. A small projecting mass/outgrowth is also noted projecting from the distal tibia. Masses are continuous with the medullary cavity and cortex and are consistent with osteochondroma. No pathologic fractures noted.

Imaging Studies: Humerus XR



Left Humerus: AP view

There is a 3.5 x 2.8 cm pedunculated mass projecting medially towards the axilla from the proximal humeral metadiaphysis. There is also an adjacent projecting mass at the proximal Humerus. Both lesions are mixed lytic and sclerotic in appearance, continuous with the medullary cavity and cortex, and consistent with osteochondromas.



Left Humerus: Lateral View

Normal RT humerus XR as reference.

*** Adult XR indicated by fused growth plates.

Image from:
http://www.wikiradiography.net/page/Humerus+Radiographic+Anatomy

Patient Treatment/Outcome

- Upon review of radiographs and a thorough history and physical, the orthopedic surgeon had a discussion with the patient about how to best move forward. The patient requested removal of the left distal femur osteochondroma due to its large size and prominence as well as the discomfort that was present upon kneeling. Due to the rapid increase in size as well as the appearance on imaging, the physician agreed to perform surgical removal of the osteochondroma and made plans to send the specimen to pathology for review. The risks and benefits of surgery were discussed with the patient. Surgical consent was obtained. Excision/Curettage of the Distal Left Femur osteochondroma was scheduled.

Discussion: What is an Osteochondroma?

- Osteochondromas can also be referred to as “osteocartilaginous exostosis.”
- Osteochondromas are benign bone tumors/outgrowths that usually present as slow-growing, painless masses. They usually occur in the second decade of life and have with rare potential for malignant transformation in adulthood.
- They are outgrowths from the normal bone and are continuous with both the medullary cavity and the cortex of the normal bone. These outgrowths are covered by a hyaline cartilage cap which serves as the source of growth and can be either sessile or pedunculated.
- They usually occur at the metaphysis and tend to arise near tendon attachment sites.
- Osteochondromas make up about 30% of all benign bone tumors.

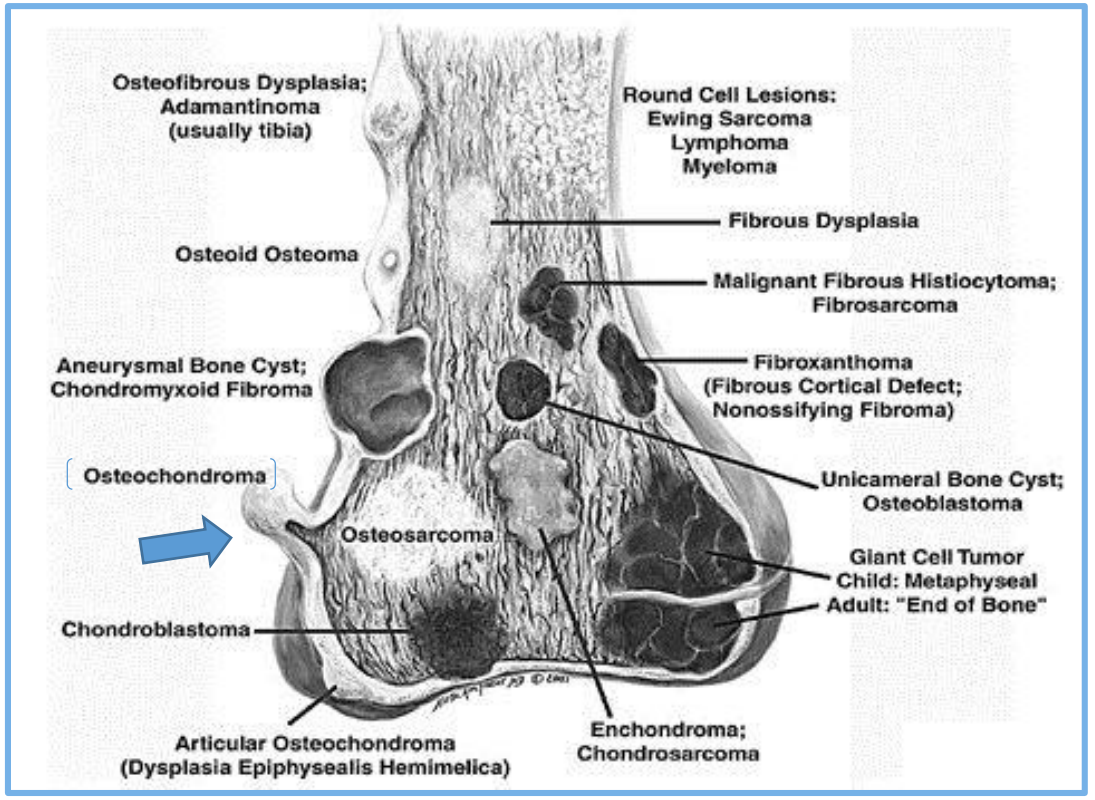


Image from: https://www.semanticscholar.org/paper/Systematic-approach-to-musculoskeletal-benign-Umer-Hasan/f8a0f9da9d90265c9c4639b00634408ba5ecaf9d

Discussion: What is Hereditary Multiple Exostosis?

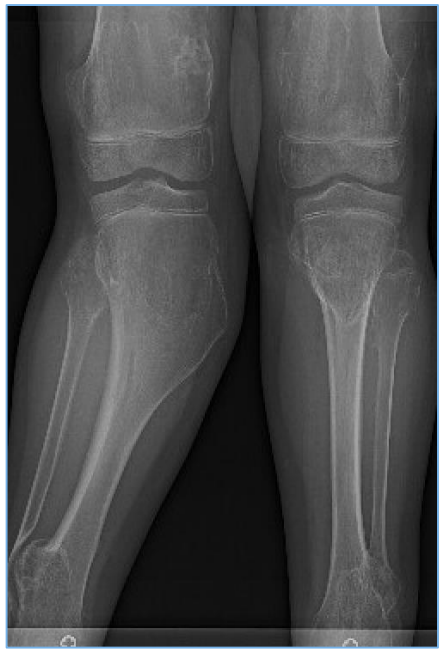
- Hereditary Multiple Exostosis (HME) is also referred to as Hereditary Multiple Osteochondromas (HMO).
- HME is diagnosed when two or more exostoses, or osteochondromas, are present in the axial and/or appendicular skeleton.
- HME occurs in about 1 in 50,000 individuals.
- Although HME can occur spontaneously or after radiation, it is usually inherited in an autosomal dominant manner and is caused by a germline mutation in the EXT1 and EXT2 tumor suppressor genes.
- Osteochondromas can occur along the appendicular skeleton and axial skeleton including the vertebral bodies.

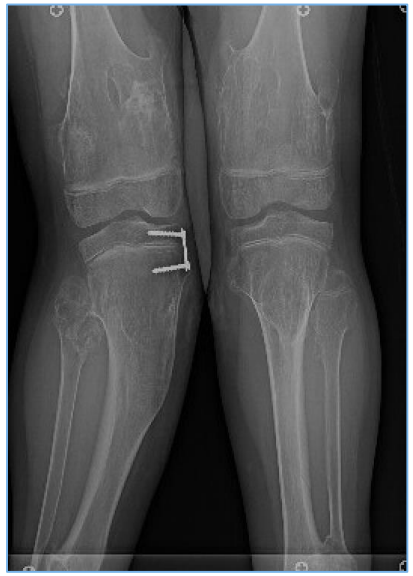
Discussion: What are complications of MHE?

- There is a small risk for malignant transformation of osteochondromas into chondrosarcoma in adulthood. This occurs in about 5% of patients with osteochondromas.
- There is risk for nerve impingement, pain, numbness, and tingling when osteochondromas grow to larger sizes and have mass effect on surrounding tissues.
- Painful fracture of the osteochondromas may occur.
- Patients with MHE may have short stature/angular deformities due to effect on nearby growth plates.
- Patients with inward projecting axial/rib involvement may develop a pneumothorax.
- Spinal impingement may occur in the presence of vertebral body involvement. MRI can be used to assess the spine for compression or abnormalities.

Discussion: What are complications of MHE?

Companion case





Osteochondromas of the distal and proximal femur, tibia, and fibula bilaterally causing **angular deformity.** Surgical screws placed for correction.

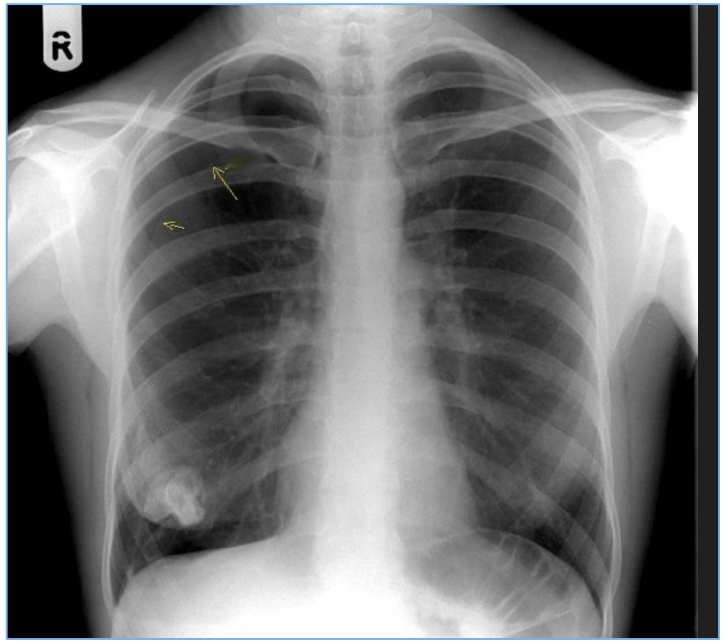
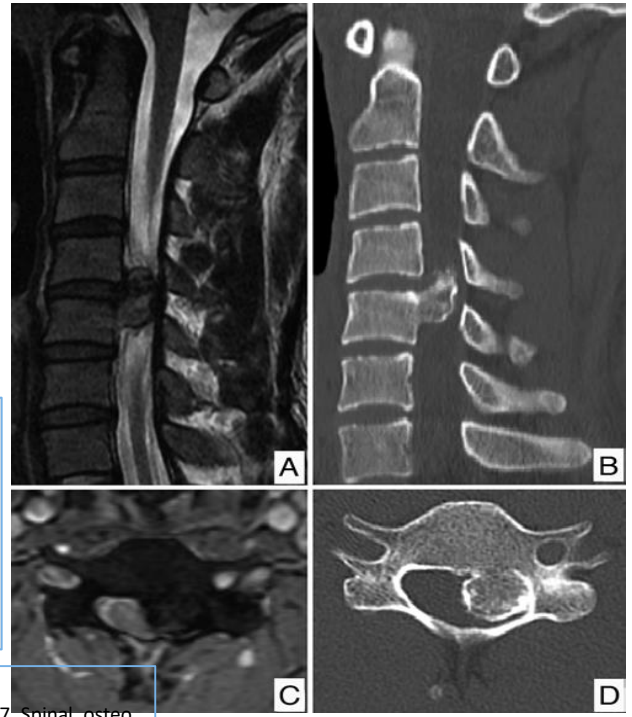


Image from: https://www.eurorad.org/case/1799

Osteochondroma present at the 6th anterior rib projecting into the right side of the thoracic cavity. **Pneumothorax** present on the right side.



Example of vertebral body osteochondroma with **compression of the spinal cord** on MRI

Image from: https://www.researchgate.net/publication/51395377_Spinal_osteochondroma_Spectrum_of_a_rare_disease_-_Report_of_3_cases

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Discussion: What are the risks of malignant transformation?

- As mentioned earlier, the risk of malignant transformation is very low (5% of patients with osteochondromas); however, certain signs and features raise suspicion of malignant transformation.
- Pain at the site may raise concern for malignancy as benign tend to be asymptomatic. Axial osteochondromas are more likely to transform.
- The cartilaginous cap should typically measure less than 1 cm in adulthood.
- In osteochondromas with cap >1 cm, there may be concern for malignant transformation to chondrosarcoma; in patients with a cartilaginous cap >2cm, biopsy and removal of the tumor is suggested.
- MRI is the imaging modality most often used to further assess painful or rapidly growing osteochondromas in adulthood. MRI is optimal for soft tissue and cartilaginous cap size.

Clinical Condition: **Primary Bone Tumors**

Variant 5: **Lesion on radiographs. Indeterminate for malignancy with mineralized matrix.**

Radiologic Procedure	Rating	Comments	RRL*
MRI area of interest without and with IV contrast	8		0
MRI area of interest without IV contrast	7		0
CT area of interest without IV contrast	7		Varies
Tc-99m bone scan whole body	5	This procedure may be helpful when evaluating for disease distribution or other areas of involvement.	☢☢☢
FDG-PET/CT whole body	3		☢☢☢☢
CT area of interest without and with IV contrast	2		Varies
CT area of interest with IV contrast	1		Varies
US area of interest	1		0
X-ray skeletal survey	1		☢☢☢

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

*Relative Radiation Level

Discussion: What is the typical management of HME?

- Normal management of patients with HME is to simply follow the patient with good physical exams, history, and review of systems.
- Most patients are asymptomatic, but when patients become symptomatic, a good ROS can help to determine next steps.
- One study suggests, in patients with HME, that at least one MRI of the spine during childhood/adolescence may be beneficial in assessing for potential spinal compression.
- In patients with painful or rapidly growing osteochondromas, MRI, biopsy, and surgical removal may be warranted.

Discussion: Imaging Sensitivity and Specificity

- When using a >2cm cartilaginous cap as the cutoff for identifying malignant transformation:
  - MRI has 100% sensitivity and 98% specificity.
  - CT has 100% sensitivity and 95% specificity.

Discussion: Cost

- X-Rays of the Femur or Tibia/Fibula
  - Cost: $27- $445 with “fair price” of $68
- X Rays of the Humerus
  - Cost: $32- $521 with “fair price” of $79
- Contrast enhanced MRI of the Humerus
  - Cost: $916- $4,096 with “fair price” of $1,513

Costs and “fair prices” according to healthcarebluebook.com.

Wrap Up

- Hereditary Multiple Exostoses (HME) is a condition that is often inherited in an autosomal dominant fashion and is characterized by the presence 2 or more osteochondromas.
- An osteochondroma is a benign bone tumor or outgrowth that usually develops in the second decade of life and is described as pedunculated or sessile, continuous with the medullary cavity and cortex, and covered by a thin cartilage cap.
- They are often discovered on plain film radiograph.
- Osteochondromas are usually managed by simple watching and waiting but can warrant MRI, biopsy, and/or surgery if they become symptomatic or develop features concerning for rare malignant transformation later in adulthood.

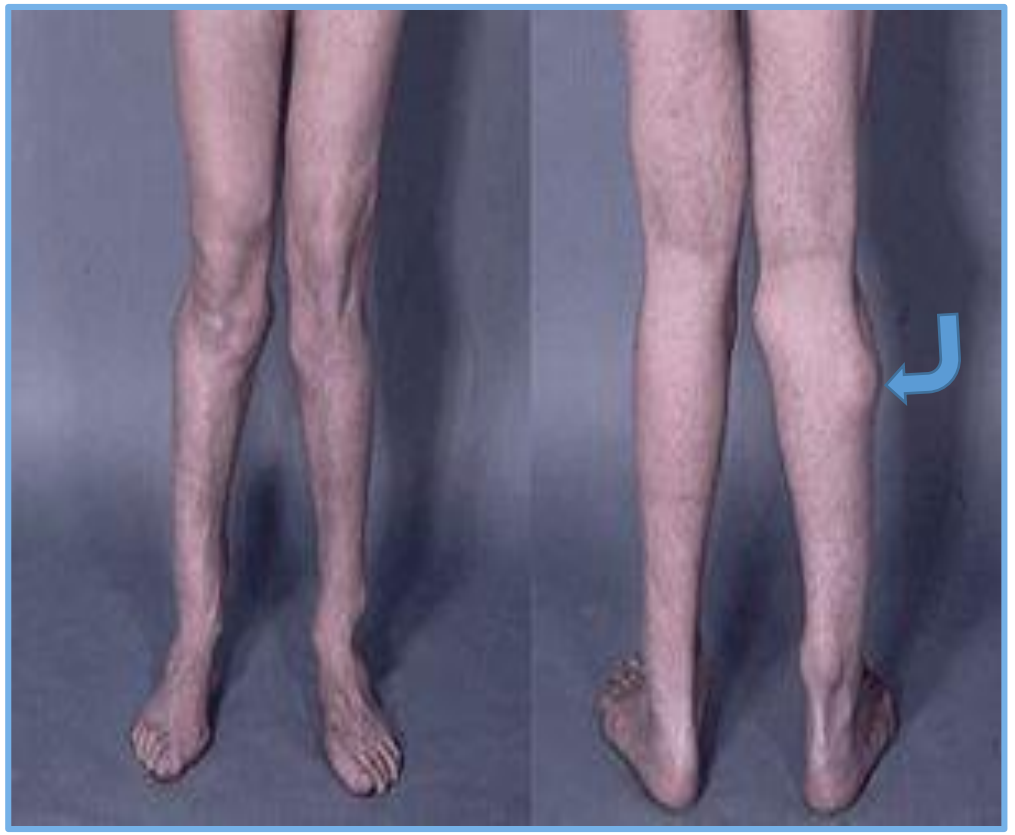

Image from: https://wikivisually.com/wiki/Hereditary_multiple_exostoses

References

1. American College of Radiology. ACR Appropriateness Criteria- Primary Bone Tumors https://acsearch.acr.org/docs/69362/narrative/. Updated 201_. Accessed June 20, 2019.
2. Challens, Andy. Femur Radiographic Anatomy. Wiki Radiography. Image from: http://www.wikiradiography.net/page/Humerus+Radiographic+Anatomy
3. Challens, Andy. Humerus Radiographic Anatomy. Wiki Radiography. Image from: http://www.wikiradiography.net/page/Humerus+Radiographic+Anatomy
4. Challens, Andy. Tib/Fib Radiographic Anatomy. Wiki Radiography. Image from: http://www.wikiradiography.net/page/Humerus+Radiographic+Anatomy
5. Fair Price Search. Healthcare Bluebook website. https://www.healthcarebluebook.com/page_SearchResults.aspx?CatID=37. Accessed June 21, 2019.
6. Modarresi, Shahla, Jude, Cecilia. "Radiologic evaluation of knee tumors in adults." *UpToDate.* Last updated Sept 19,2018. Accessed. June 19 2019. https://www.uptodate.com/contents/radiologic-evaluation-of-knee-tumors-in-adults?search=osteochondroma&source=search_result&selectedTitle=2~16&usage_type=default&display_rank=2
7. Ramesha, N, O'Dowd, M., Hogan, B. "Osteochondroma of the Rib: Case 1799." Eurorad. Published Sept 9, 2002. Accessed June 20, 2019. Image From: https://www.eurorad.org/case/1799
8. Solitary Osteochondroma. *Consultant 360*. 2018. May. 58:5. Accessed June 20, 2019. Image from: https://www.consultant360.com/articles/solitary-osteochondroma?page=1
9. Srikantha, U., Bhagavatula, I., Satyanarayana, S., Somonna, S."Spinal Osteochondroma: Spectrum of a rare disease- Report of 3 cases." Journal of neurosurgery: Spine. (2008) June; 8(6): 561-6. https://www.researchgate.net/publication/51395377_Spinal_osteochondroma_Spectrum_of_a_rare_disease_-_Report_of_3_cases
10. Tis, John E. "Nonmalignant bone lesions in children and adolescents." *UpToDate.* Last updated March 23, 2019. Accessed June 20, 2019. https://www.uptodate.com/contents/nonmalignant-bone-lesions-in-children-and-adolescents?sectionName=Osteochondroma%20and%20hereditary%20multiple%20osteochondromas&search=osteochondroma&topicRef=14916&anchor=H16&source=see_link#H16
11. Umer, M. Hasan, Obada, Noordin, Shahryar. "Systematic Approach to musculoskeletal benign tumors." *Internal Journal of Surgery: Oncology.* (2017). Nov; 2:e46. https://www.semanticscholar.org/paper/Systematic-approach-to-musculoskeletal-benign-Umer-Hasan/f8a0f9da9d90265c9c4639b00634408ba5ecaf9d