AMSER Case of the Month
June 2023

A 57 y/o male presented for living liver donor evaluation and was found to have abnormal imaging

HPI: 57 y.o. male presenting for living transplant evaluation with no concerns

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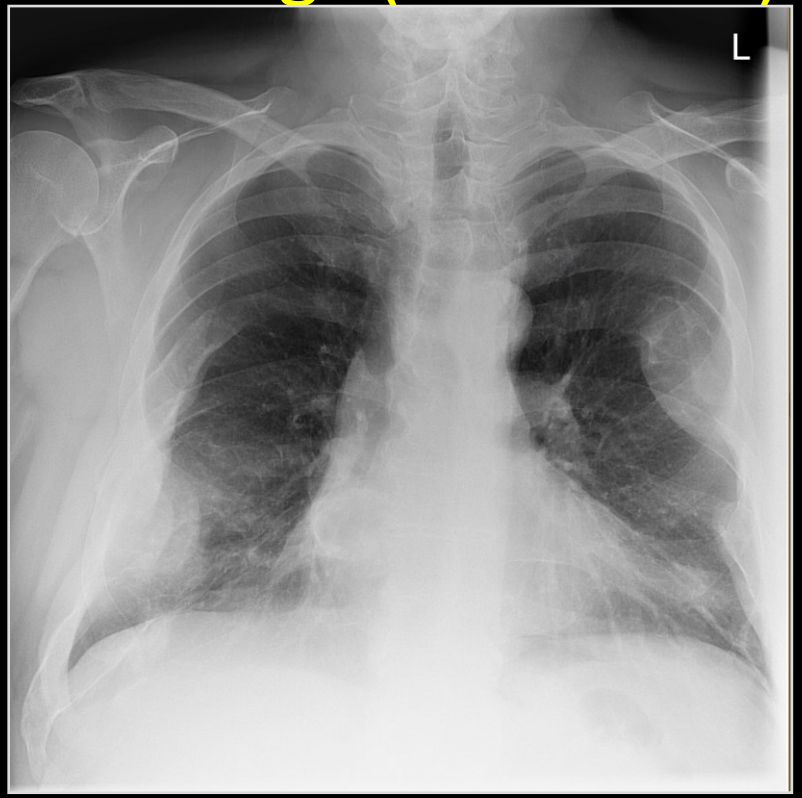
Patient Presentation

- HPI: patient presents for routine living liver donor evaluation and workup. He had no concerns at the time and reported being healthy his whole life
- Past medical history: Hypertension, GERD, ADHD
- Social History: Never a smoker and has no history of alcohol abuse
- Medications: lisinopril 40 mg, atomoxetine 25 mg
- Physical Exam: Unremarkable

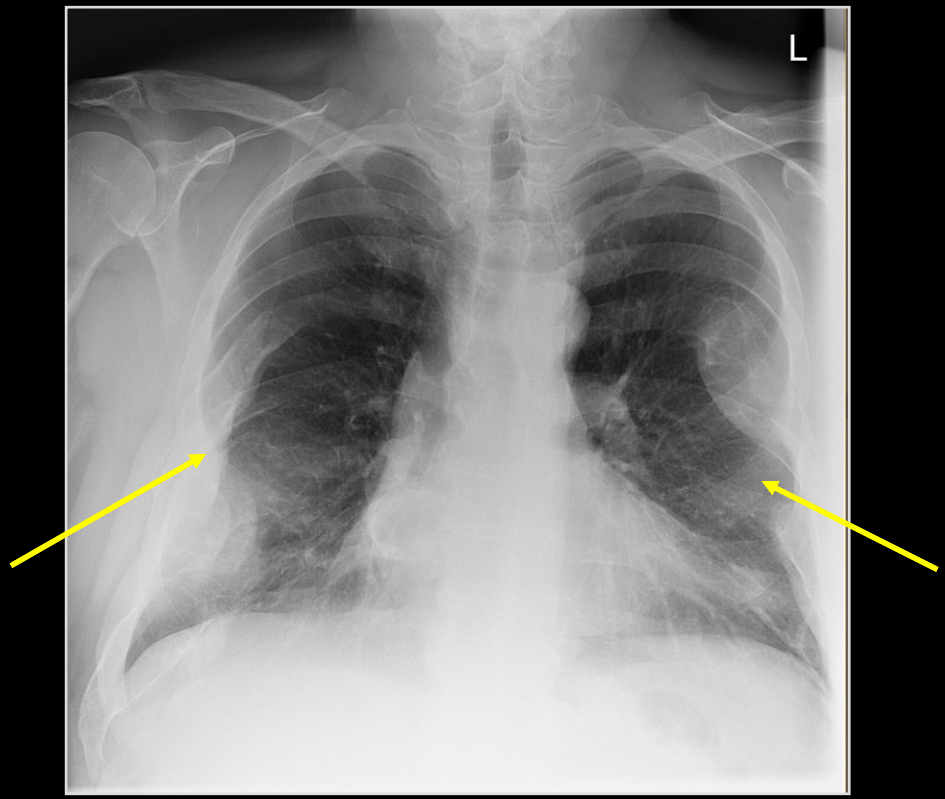
Chest X-ray was ordered as part of routine transplant evaluation

Findings (unlabeled)



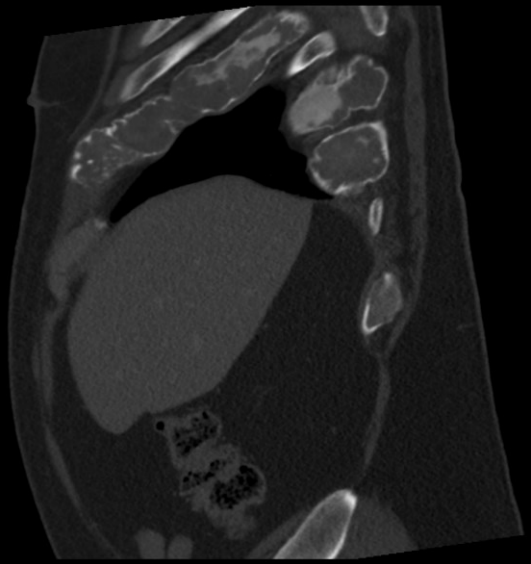
Findings: (labeled)

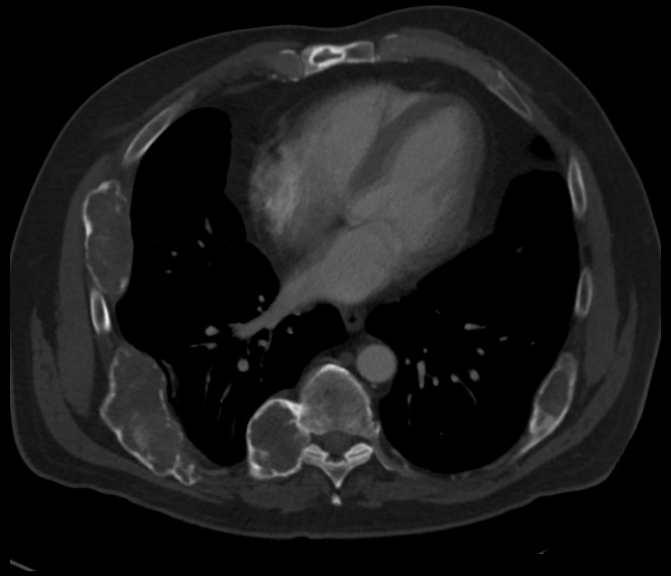
Yellow arrows: Rib deformities can be seen bilaterally.



CT abdominal angiogram was ordered as part of the liver transplant evaluation

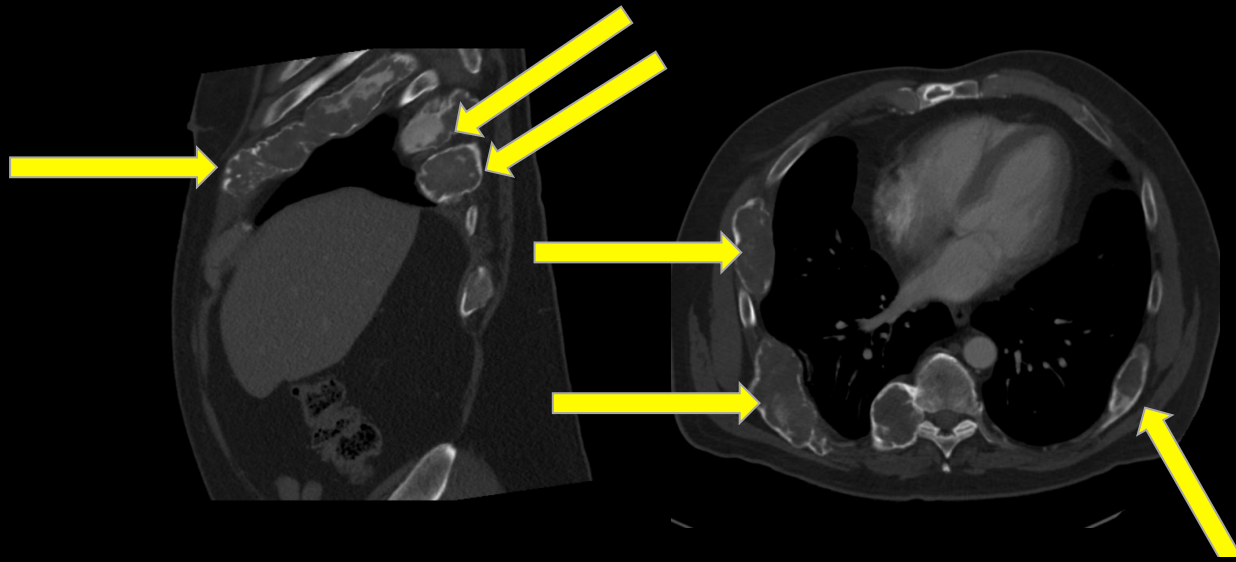
Findings (unlabeled)





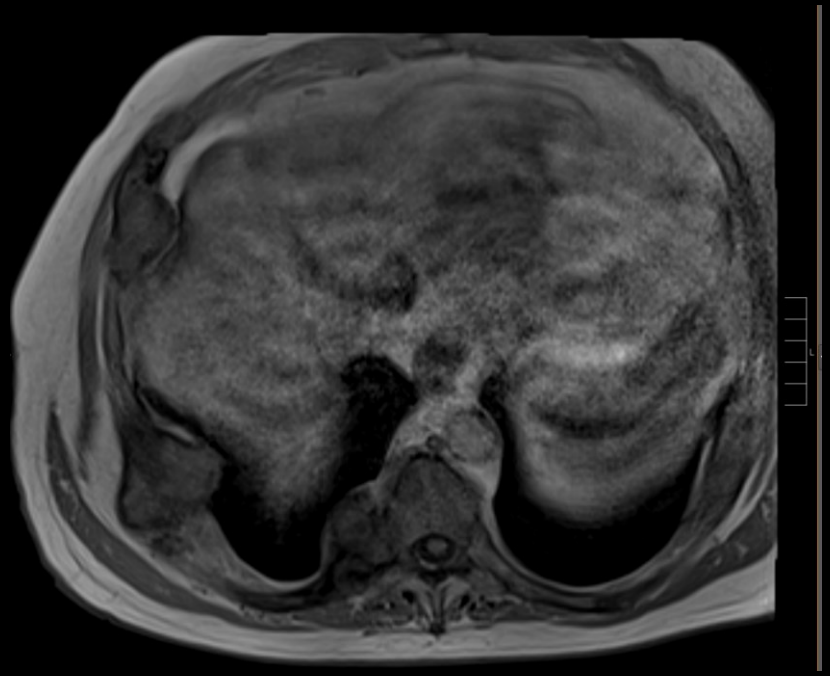
Findings: (labeled)

Yellow arrows:
Multiple bilateral expansile rib lesions with ground-glass matrix were seen.



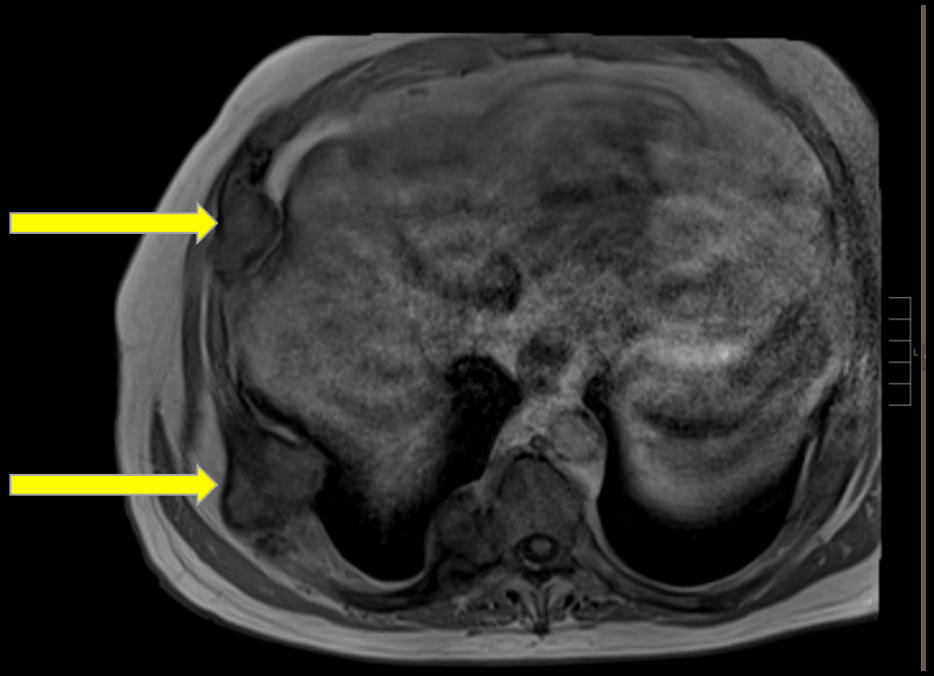
MRI cholangiopancreatography without contrast was performed as part of the liver donor evaluation

Findings (unlabeled)



Findings: (labeled)

Yellow arrows: The same expansile rib lesions were seen on the MRI cholangiopancreatogram



What is the differential diagnosis?

Differential Diagnosis

1) Polyostotic fibrous dysplasia
2) Metastatic malignancy
3) Primary osseous malignancy (osteosarcomas, chondrosarcomas)
4) Ossifying fibromas
5) McCune-Albright syndrome (MAS)
6) Mazabraud syndrome

Final dx: Polyostotic Fibrous Dysplasia

Case Discussion: Fibrous Dysplasia (FD)

Epidemiology: FD equally affects male and female populations. The condition is typically diagnosed in childhood, however, mild cases often go undiagnosed. The incidence and prevalence is unknown due to many cases often going undiagnosed. There is a 4:1 ratio of the monostotic to polyostotic form[1].

Pathophysiology: Believed to be the result of developmental failure in remodeling of primitive bone to lamellar bone. This failure ultimately leads to an immature mass of trabeculae with dysplastic fibrous tissue. As a result, there is loss of mechanical strength, deformities, potential pain, and potential fractures to the affected bones[2].

Case Discussion: Fibrous Dysplasia

Presentation: FD is typically found as an incidental finding on x-rays. Monostotic is most common and typically presents as a painless swelling on a rib. Any bone can be affected, however, long bones, craniofacial bones, and ribs are most often affected. Patients can present with pain due to nerve compression or fractures of weight bearing bones, such as long bones of the legs[1].

Diagnostic Imaging: CT is the preferred study for radiologic diagnosis of FD and MRI can be used in conjunction to further support a diagnosis of FD[1,2].

Management: Treatment revolves around managing pain. Bisphosphonates have been shown to reduce bone pain. Other treatments include NSAIDs and Narcotics[3].

Case Discussion: Differentials

McCune-Albright syndrome: Can present with FD, however, typically has extraskeletal manifestations that include Café-au-lait spots, gonadal abnormalities (male), precocious puberty (females), hyperthyroidism, and Cushing's syndrome[4].

Low grade intramedullary osteosarcoma: can look similar radiographically to FD. Low grade intramedullary osteosarcoma can be differentiated from FD by a lack of reactive shell, denser mineralization, permeative borders, and has more aggressive changes over time compared to FD[2].

Ossifying Fibromas: rare lesions localized mainly to the distal third of the tibia or fibula in children under ten. Molecular analysis can be helpful in differentiating ossifying fibromas from FD[1].

References:

1)*Fibrous dysplasia - symptoms, causes, treatment: Nord*. National Organization for Rare Disorders. (2023, January 12). from https://rarediseases.org/rare-diseases/fibrous-dysplasia/#affected

2) DiCaprio, M. R., & Enneking, W. F. (2005). Fibrous dysplasia pathophysiology, evaluation, and treatment. *The Journal of Bone and Joint Surgery (American)*, *87*(8), 1848. https://doi.org/10.2106/jbjs.d.02942

3)Chapurlat, R. D., Gensburger, D., Jimenez-Andrade, J. M., Ghilardi, J. R., Kelly, M., & Mantyh, P. (2012). Pathophysiology and medical treatment of pain in fibrous dysplasia of bone. *Orphanet Journal of Rare Diseases*, *7*(Suppl 1). https://doi.org/10.1186/1750-1172-7-s1-s3

4) Collins MT, Singer FR, Eugster E. McCune-Albright syndrome and the extraskeletal manifestations of fibrous dysplasia. Orphanet J Rare Dis. 2012 May 24;7 Suppl 1(Suppl 1):S4. doi: 10.1186/1750-1172-7-S1-S4. Epub 2012 May 24. PMID: 22640971; PMCID: PMC3359955.