AMSER Case of the Month August 2025

50 Y.O. male with Beta-Thalassemia: Imaging the Long-Term Osseous Effects

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Patient Presentation

- 50-year-old male with history of chronic thromboembolic pulmonary hypertension (CTEPH), atrial flutter (s/p ablation), prior PE
- Presenting with dyspnea, palpitations, and tachycardia
- Known beta-thalassemia intermedia
  - Transfusion-dependent since childhood
  - On iron chelation (Deferiprone 3500 mg/day) since age 8
  - History of splenectomy
  - Hemoglobin goal maintained at ~10 g/dL
  - Receives 2 units of pRBC every 2 weeks
  - Known alloantibodies: anti-Kell and anti-e

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What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 2: **Adult. Acute respiratory illness in immunocompetent patients with positive physical examination or abnormal vital signs or organic brain disease or other risk factors for poor outcome. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
Radiography chest	Usually Appropriate	☢
US chest	Usually Not Appropriate	O
MRI chest without and with IV contrast	Usually Not Appropriate	O
MRI chest without IV contrast	Usually Not Appropriate	O
CT chest with IV contrast	Usually Not Appropriate	☢☢☢
CT chest without and with IV contrast	Usually Not Appropriate	☢☢☢
CT chest without IV contrast	Usually Not Appropriate	☢☢☢
CTA chest with IV contrast	Usually Not Appropriate	☢☢☢
V/Q scan lung	Usually Not Appropriate	☢☢☢

This imaging modality was ordered.

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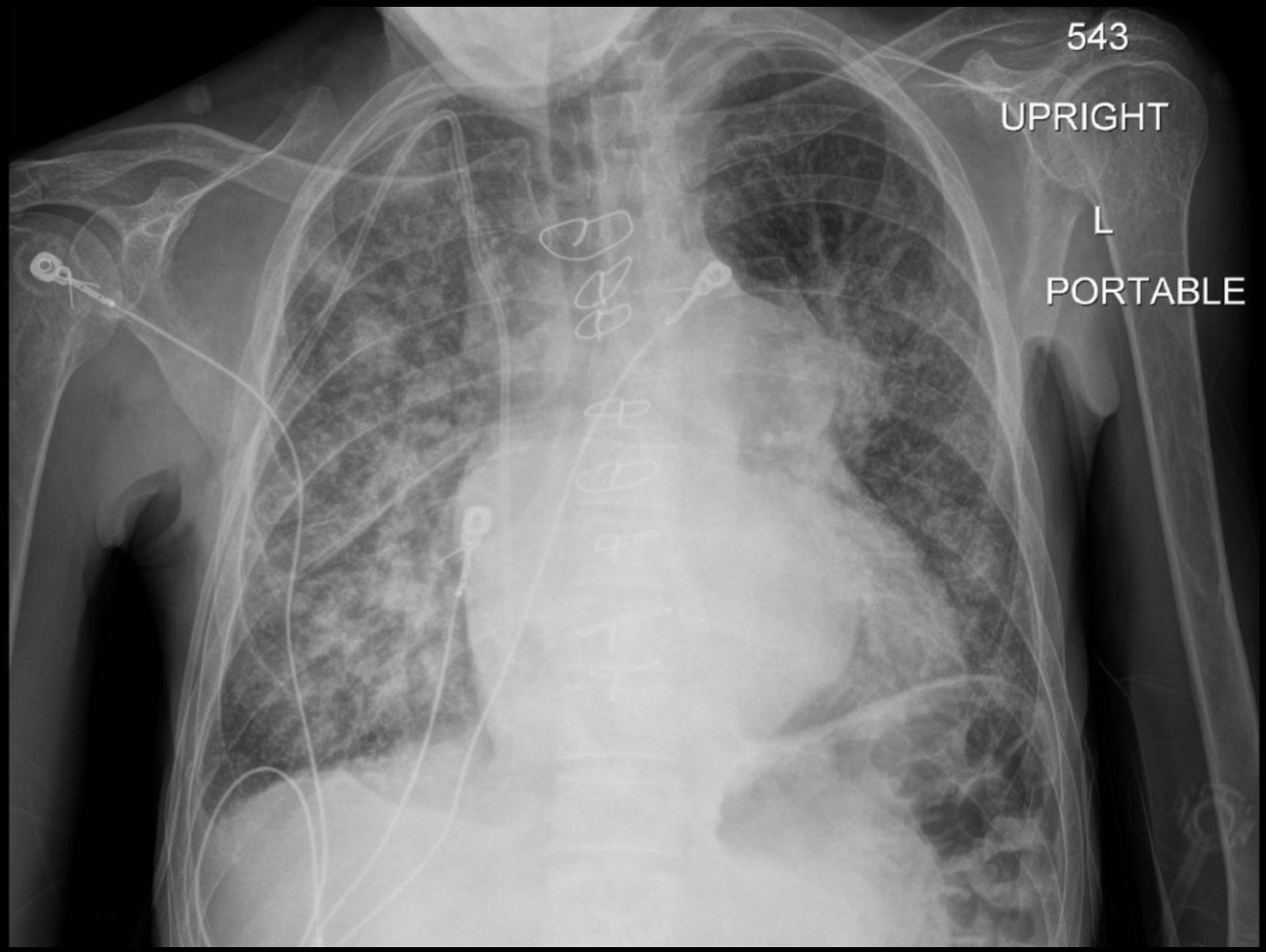
Findings: (unlabeled)

Prior Chest XR

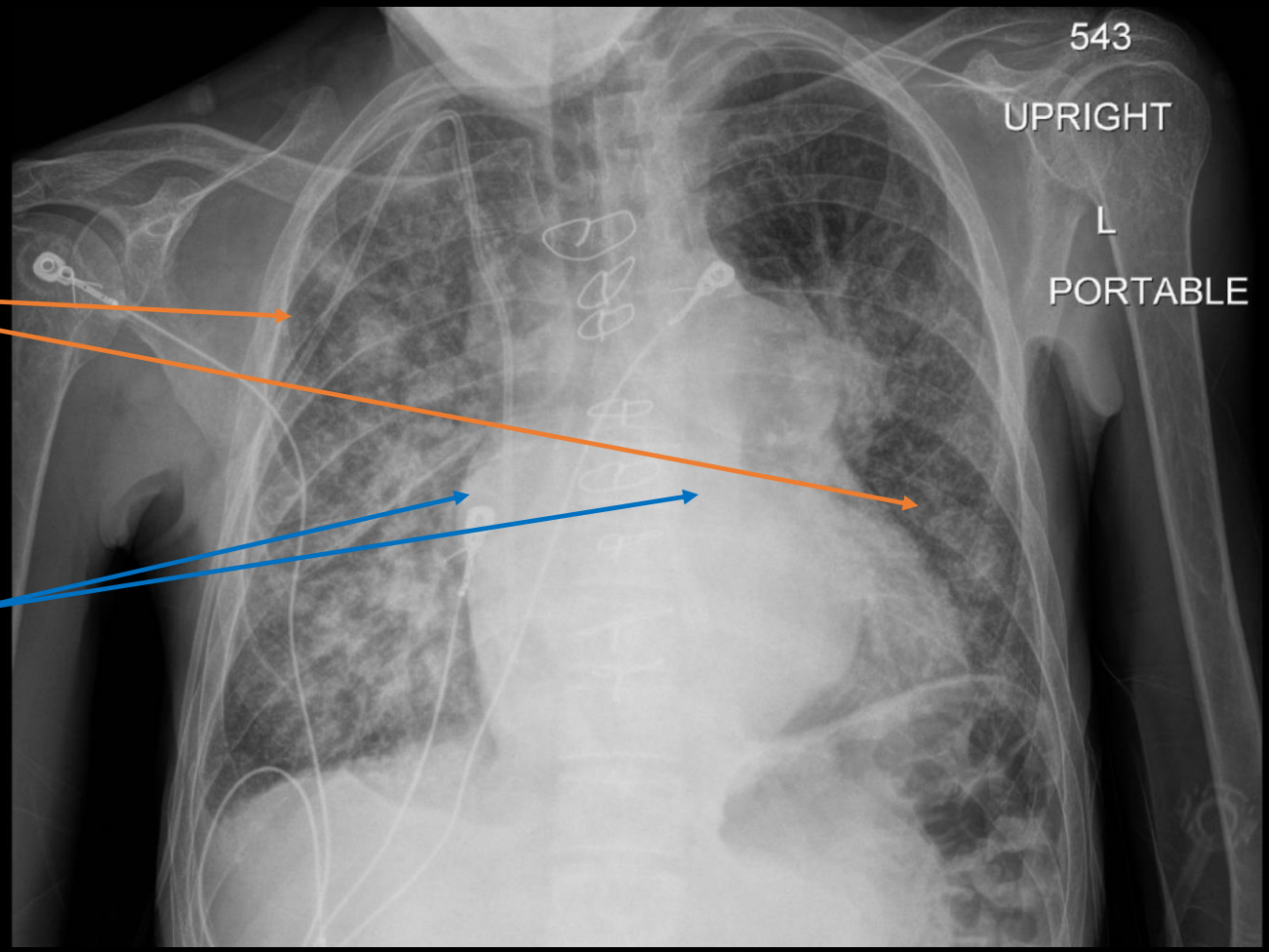


Findings: (labeled)

Prior Chest XR

Bilateral airspace opacities

Paraspinal masses



Patient Presentation

- Patient was then treated for multifocal pneumonia.
  - Initial chest radiographs revealed new consolidative airspace opacities.
  - Infectious disease team requested a CT chest to further characterize the CXR findings.
  - Patient then completed a course of antibiotics and ID recommended follow-up imaging in 20 days to confirm resolution.
- CT chest revealed incidental findings suggestive of beta thalassemia-related bony changes and extramedullary hematopoiesis.
- This led to a retrospective evaluation focused on imaging changes attributable to beta thalassemia.

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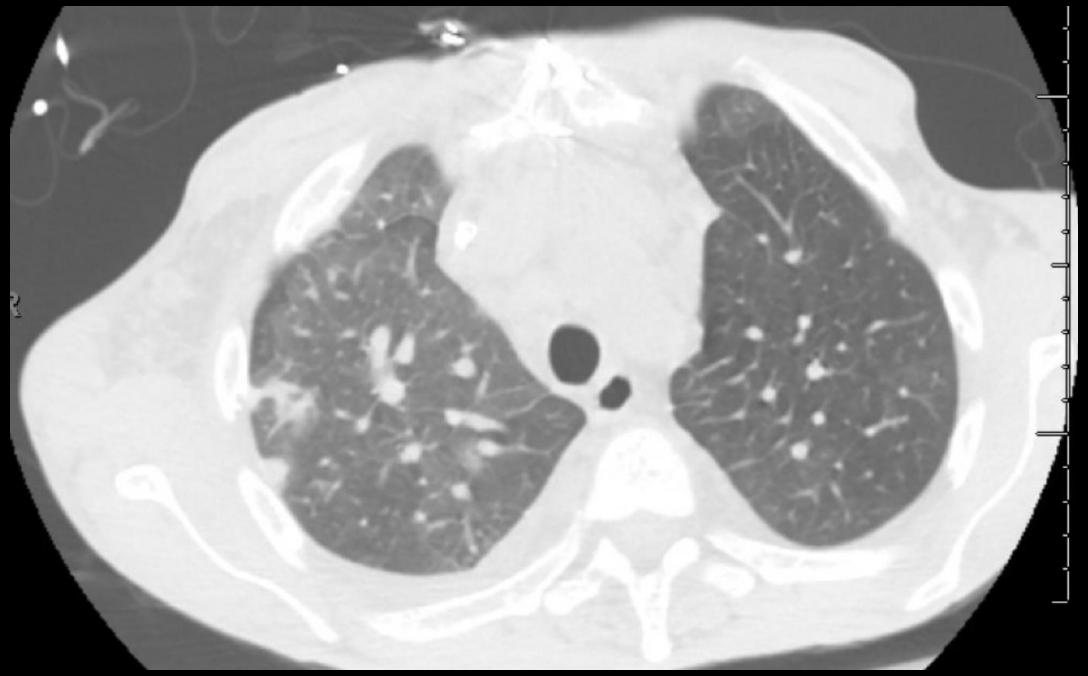
CT chest findings

- Almost complete interval resolution of the previously seen likely infectious multifocal nodular opacities and right upper lobe cavity with minimal residual scarring.
- Stable pattern of bone demineralization with cortical thinning secondary to thalassemia. The paraspinal soft tissue density masses extending from T9 to T12 are unchanged and compatible with extramedullary hematopoiesis in the context of thalassemia.

Findings: (unlabeled)

PRIOR



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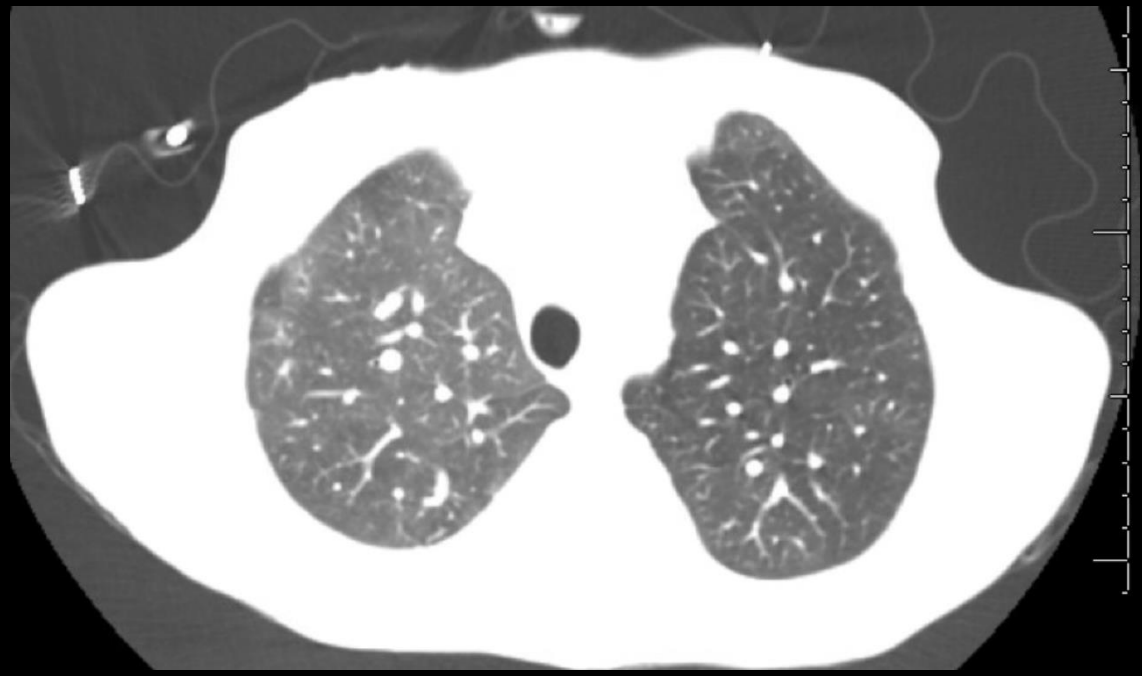
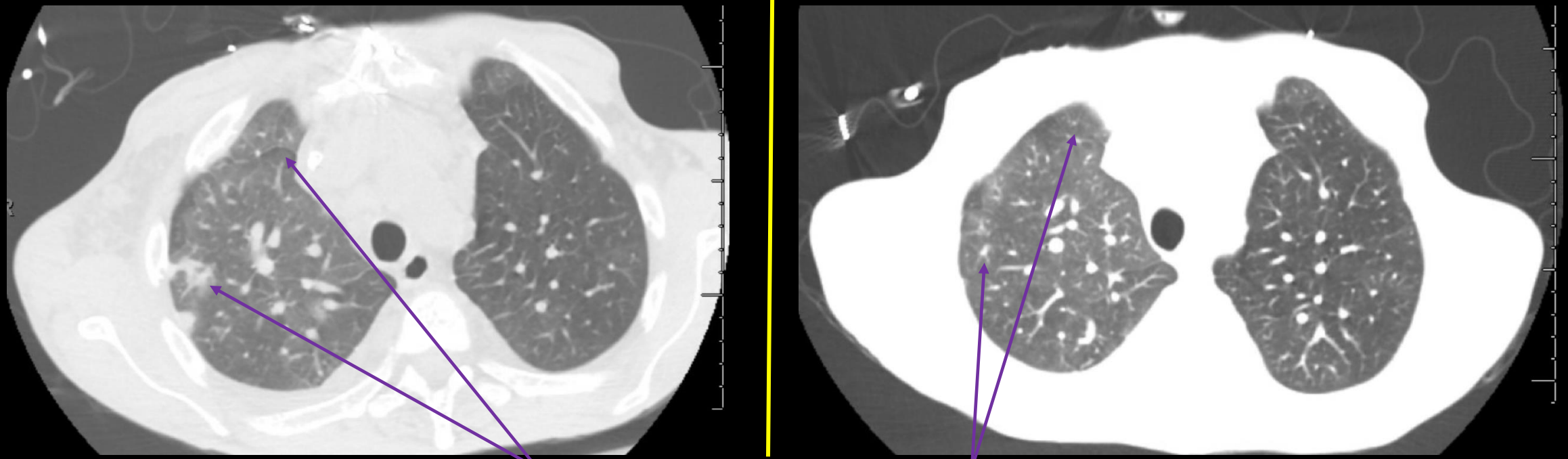


Findings: (labeled)

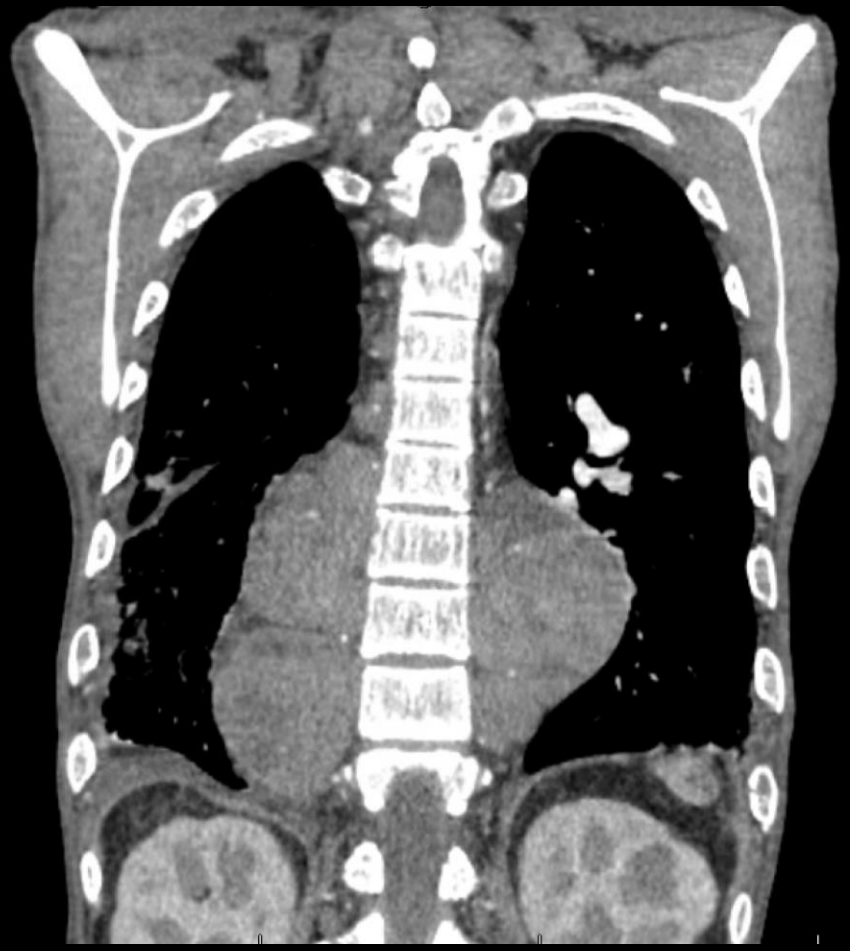
PRIOR

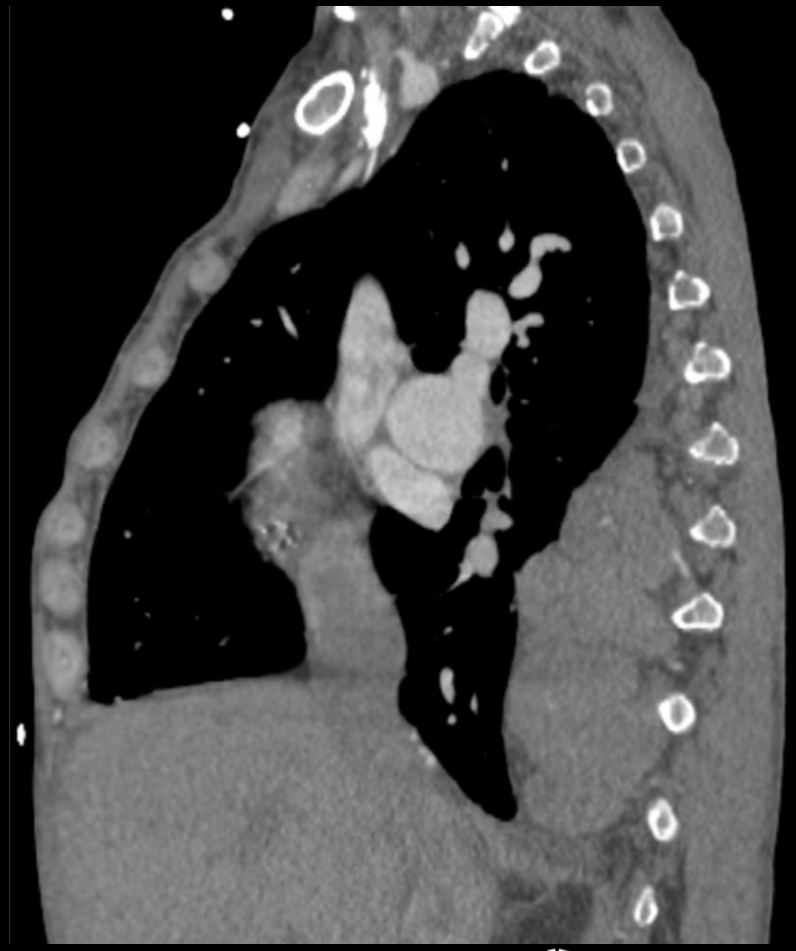
POST



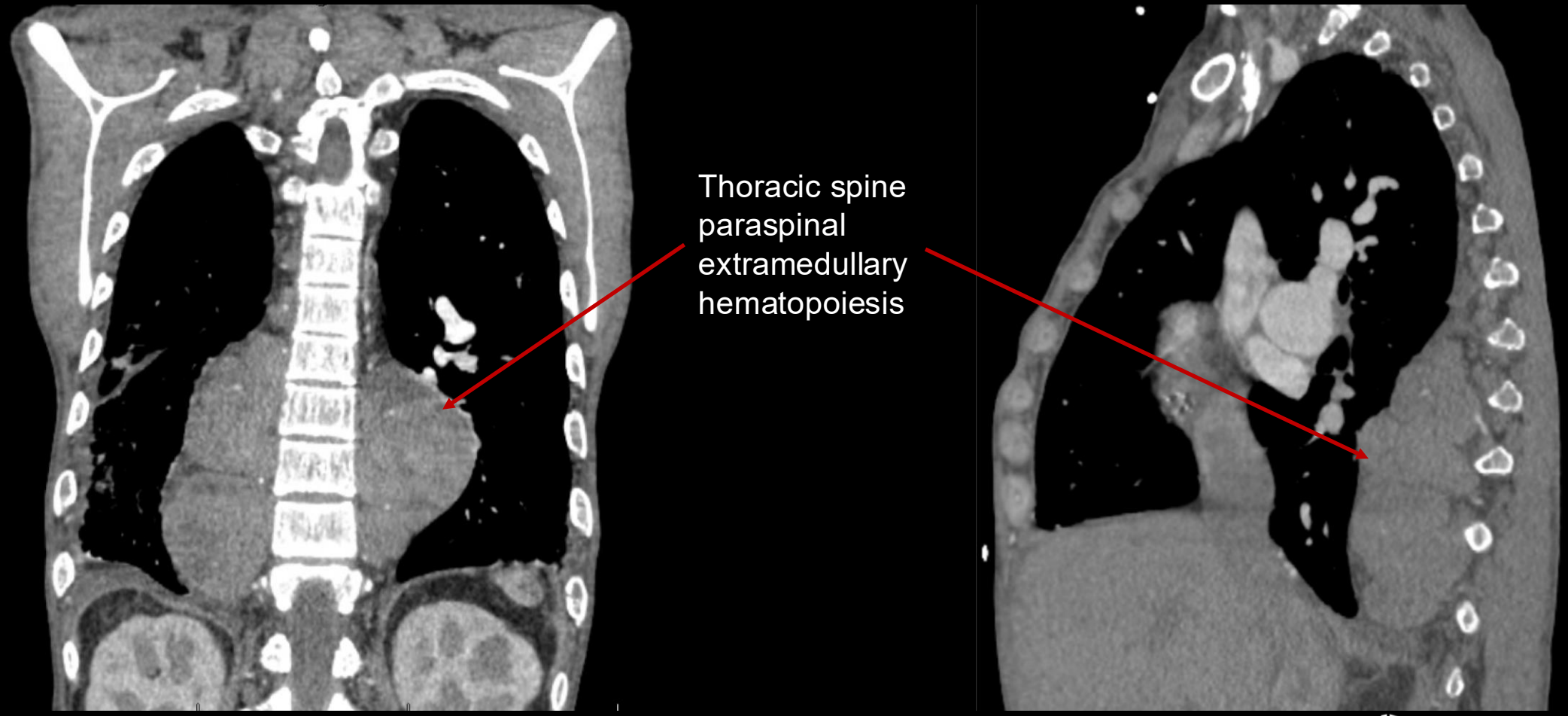
Significant resolution of multifocal nodules with residual scarring

Findings (unlabeled)





Findings: (labeled)



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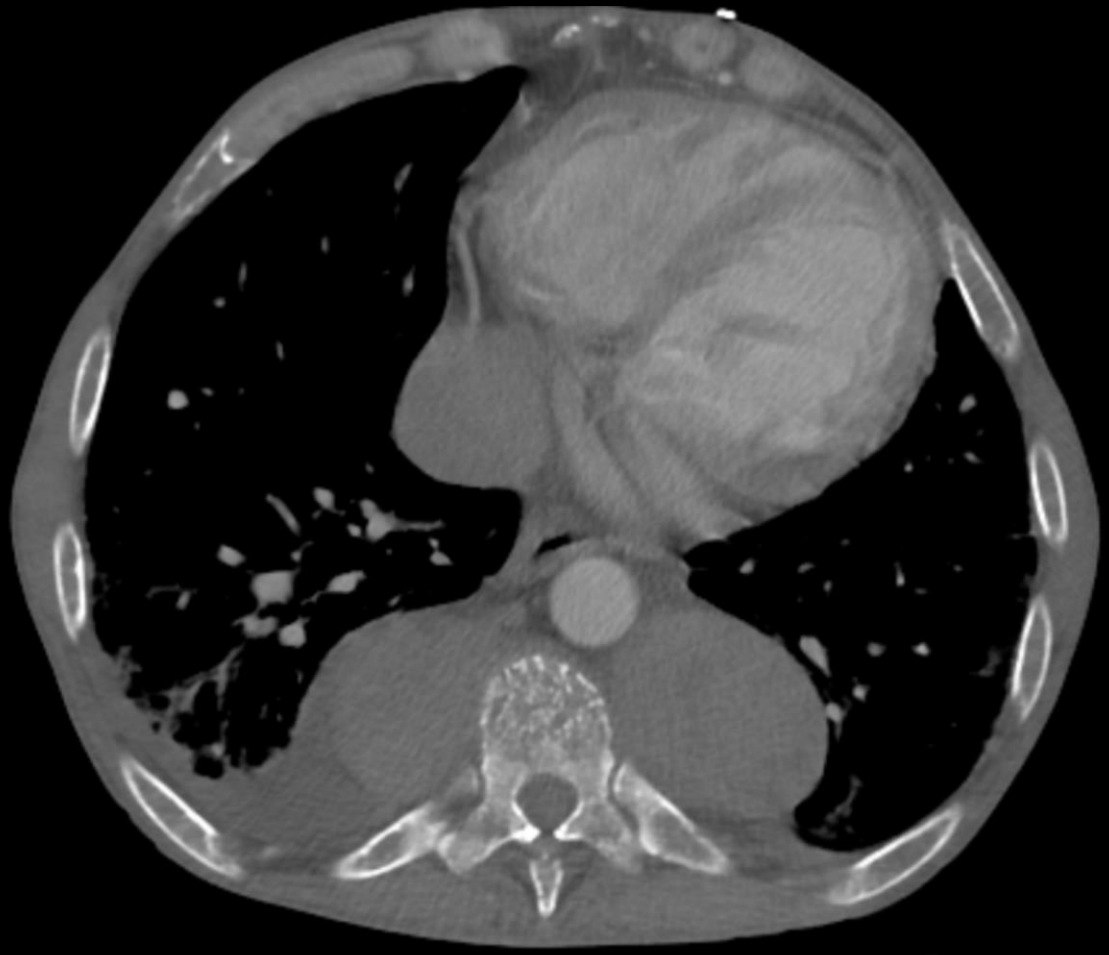

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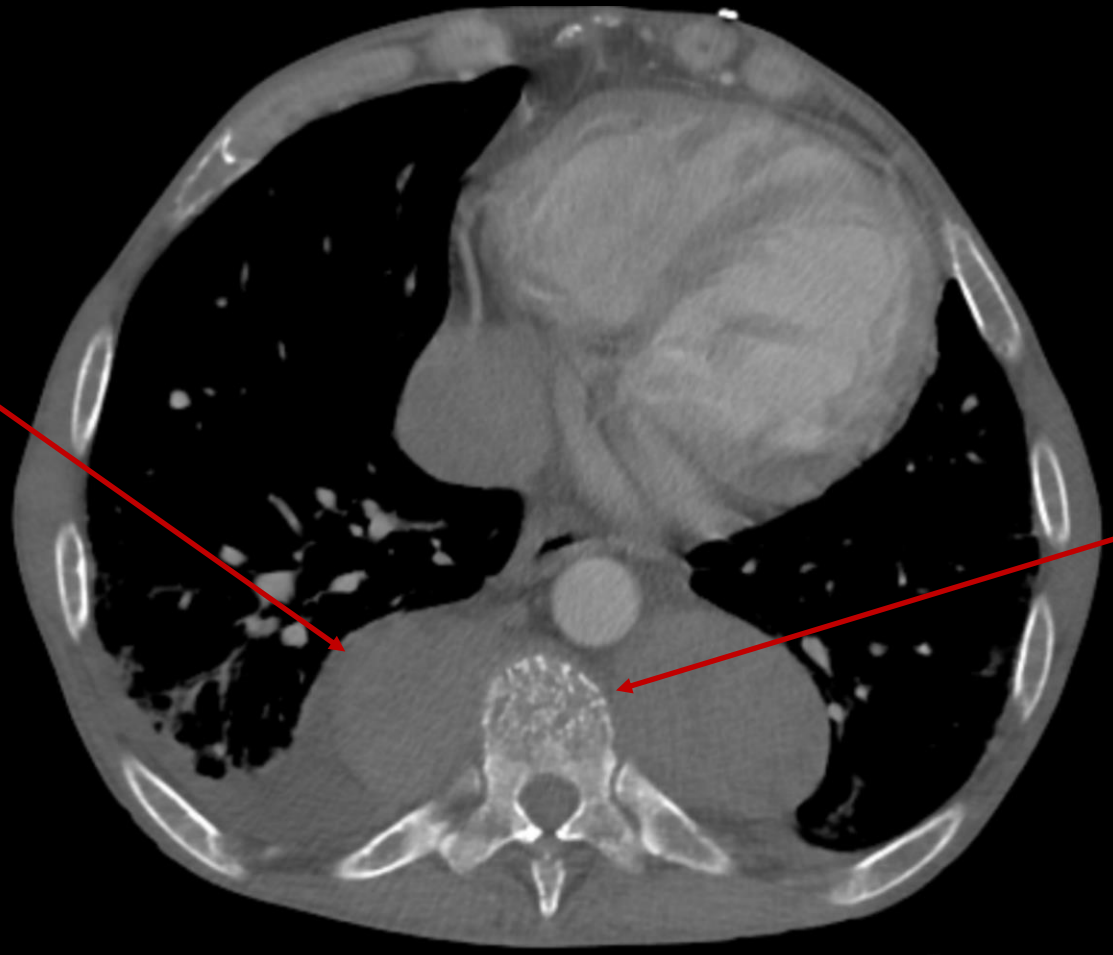
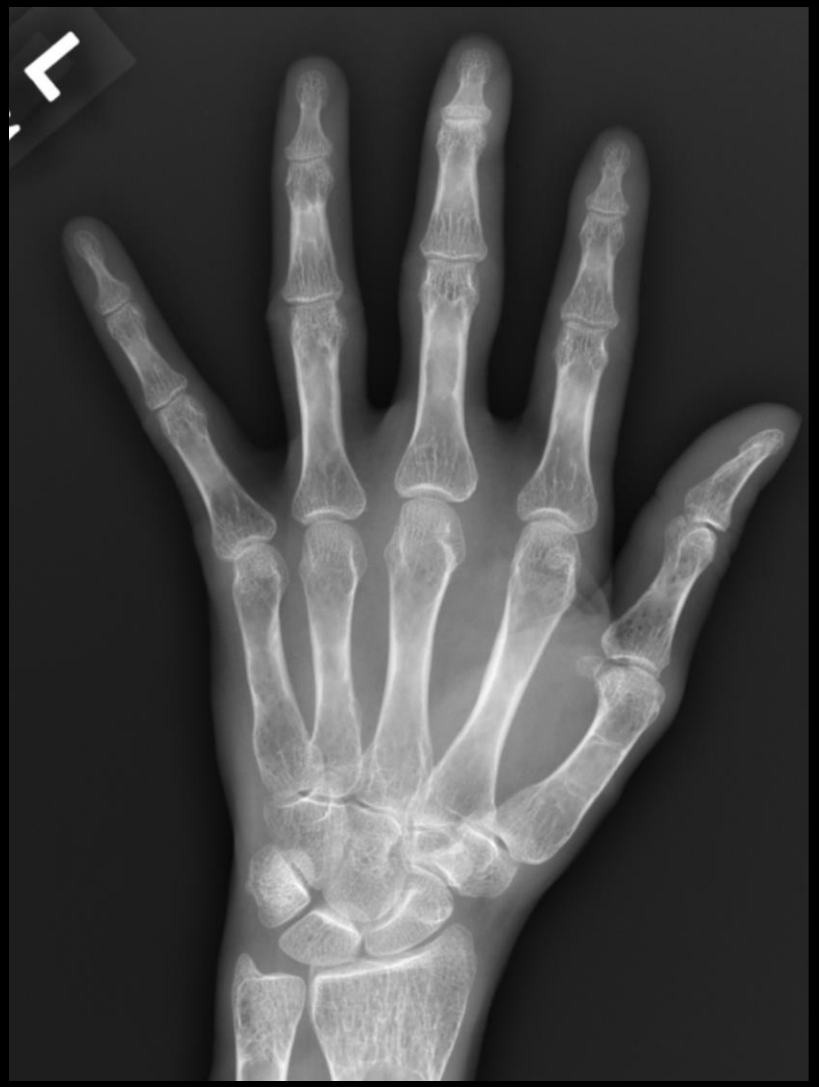
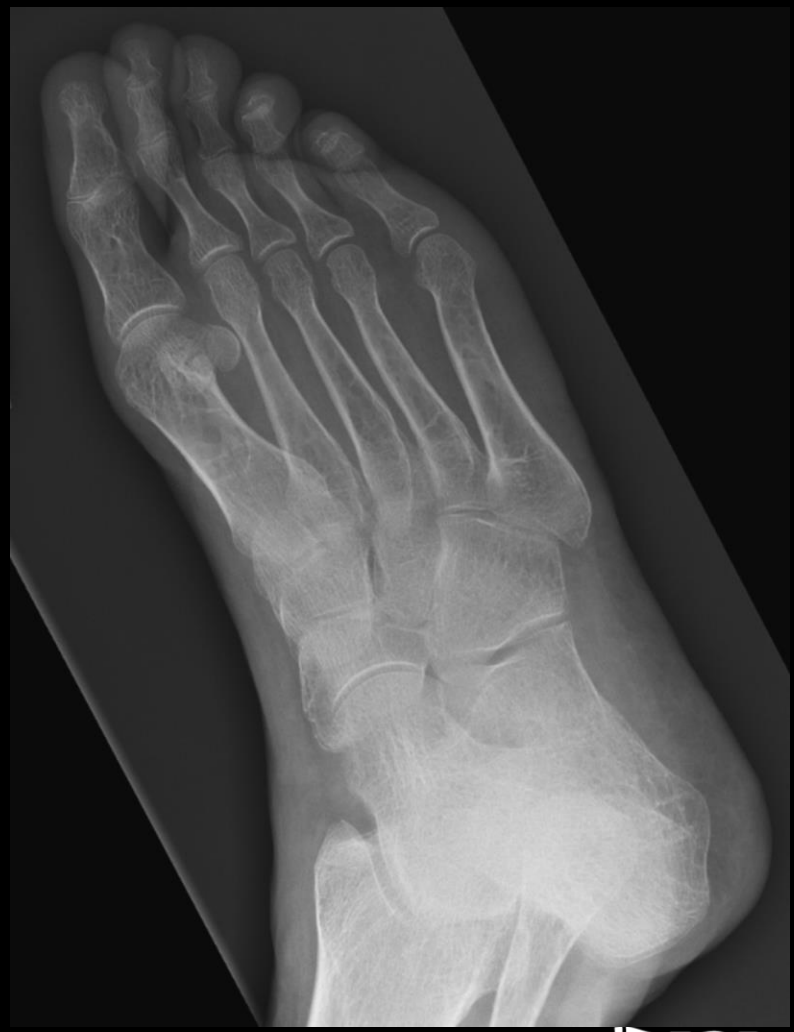
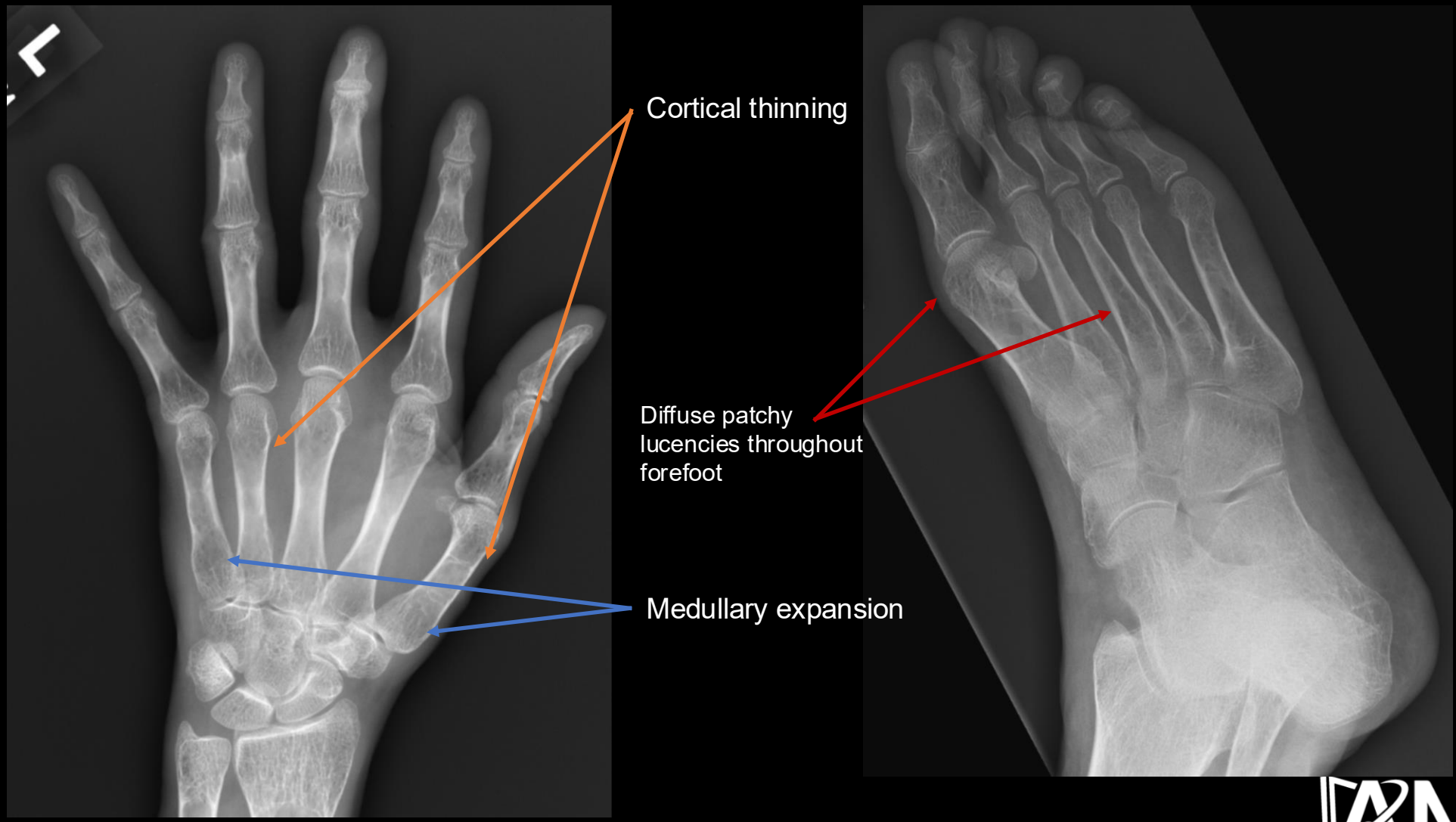


Findings: (unlabeled)





Findings: (labeled)



Differential Diagnosis

- Sickle Cell Disease – Causes chronic hemolysis and marrow hyperplasia, leading to bone changes and occasional extramedullary hematopoiesis (EMH).
- Hereditary Spherocytosis – Chronic hemolysis may lead to marrow expansion and splenomegaly.
- Myelofibrosis – Bone marrow failure prompts EMH, often presenting with paraspinal masses.
- Exacerbation of CTEPH–Right heart strain and heart failure symptoms, and in this patient, diagnostic evaluation was confounded by concurrent multifocal pneumonia.

Final Dx:

Extramedullary Hematopoiesis secondary to Beta Thalassemia Intermedia

Case Discussion

- Beta thalassemia is a hereditary blood disorder characterized by reduced or absent beta-globin chain production, leading to ineffective erythropoiesis and chronic anemia.
- To compensate, the body increases bone marrow activity, which can extend beyond normal medullary spaces, causing extramedullary hematopoiesis (EMH)[1].
- Bone changes result from[2]:
  - Expansion of medullary cavities due to marrow hyperplasia.
  - Thinning of cortical bone and altered trabecular patterns.
  - Skeletal deformities due to pressure effects of EMH.

Case Discussion

- Occurs when hematopoietic tissue proliferates in non-marrow sites, including paraspinal areas, liver, spleen, and bones (especially skull, ribs, vertebrae).
- Chronic anemia stimulates persistent marrow expansion.
- Leads to characteristic findings like[3]:
  - “Hair-on-end” appearance on skull X-ray.
  - Widening of diploic space, cortical thinning.
  - Rib and spine expansion (“crew-cut skull”, codfish vertebrae).
- X-ray: Detects classic changes (e.g., frontal bossing, maxillary hypertrophy).
- MRI: More sensitive for identifying EMH masses.
- CT: Evaluates bony architecture and EMH-related masses.

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Case Discussion

- Clinical Features
  - Skeletal deformities (frontal bossing, maxillary overgrowth, limb bowing)
  - Short stature, delayed bone age
  - Back pain or neurologic deficits if paraspinal EMH compresses the spinal cord[4]
- Complications
  - Pathologic fractures from bone fragility
  - Neurologic deficits due to mass effect from paraspinal EMH
- Management[5,6]
  - Regular transfusion therapy suppresses ineffective erythropoiesis and EMH
  - Hydroxyurea or hematopoietic stem cell transplant in select cases
  - Radiation therapy or surgery for symptomatic EMH masses compressing critical structures

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4. Orphanet. Extramedullary hematopoiesis. Accessed July 4, 2025. https://www.orpha.net
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6. Taher AT, Musallam KM, Cappellini MD. β-Thalassemias. *N Engl J Med*. 2021;384(8):727-743. doi:10.1056/NEJMra2021830