Diagnostic Dilemma

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Ground Rules

1. Any idea is a good one

2. We are all smart and have something to learn from each other

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PCP Visit

4 y/o healthy male, well-appearing, interactive, playful

- **normal** vital signs, no fevers

- **bruising** under L eye after wrestling cousins this past weekend

- **blood** around his lips this morning

- PCP notes **ecchymoses** to palate, posterior pharynx, and uvula. Marked hepatosplenomegaly present

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ED: Initial work-up?

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Initial Results

Labs

- WBC 10.8 ANC: 430 HGB: 9.2 Plt 26 Neutrophils 0.3
- CMP WNL alk phos low at 57
- Coags w/ elevated PT (21.6), PTT (38.8) and INR (2)
- Fibrinogen 38
- Uric acid and LDH WNL
- Peripheral blood smear without circulating blasts

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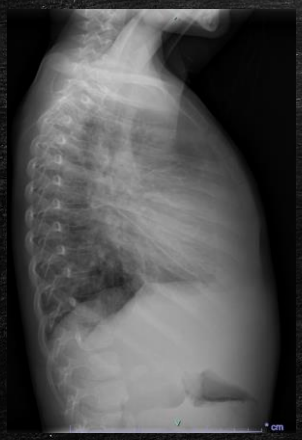
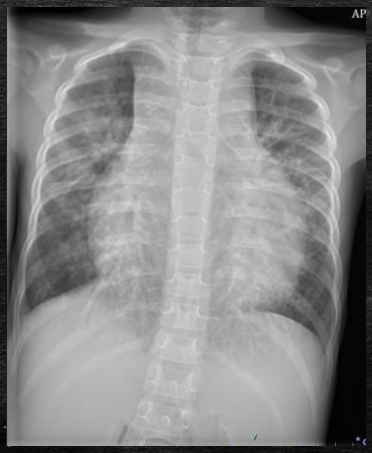
Chest x-ray





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Next steps in work-up?

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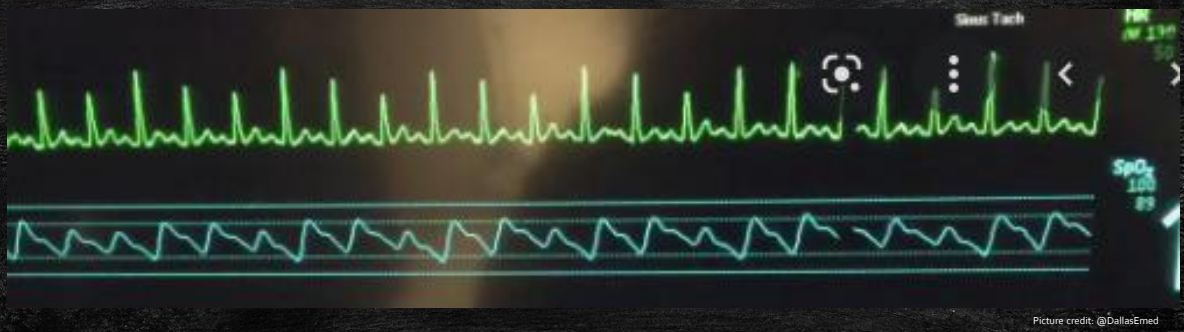
Flash quiz: What's wrong with this?



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Results come in:

Echo Results:

- large circumferential pericardial effusion echocardiographic evidence of tamponade physiology

- pericardial drain placed

Bone Marrow Results

- No blasts

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Days 2-4

- Re-accumulation of large pericardial fluid collection without tamponade
  - Drain optimized, 800 mL-1.2 L of output per day

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4 y/o previously healthy M with:

- Pancytopenia
- Coagulopathy
- Pericardial effusion with high output
- Hepatosplenomegaly

Bone marrow without blasts

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Differential diagnosis?

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Differential?

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What else do you want to order?

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Next steps in work-up

- Retic count
- Ferritin
- Inflammatory markers
- Pericardial studies including viral/fungal/bacterial etio
- Viral serologies
  - HIV, EBV, CMV, parvovirus
- Rheum
  - Haptoglobin, Coombs, ANA profile, C_3, antiphospholipid antibodies, CD_{25}
- Pan-CT

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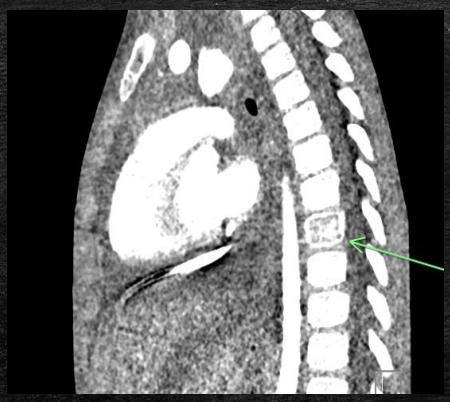
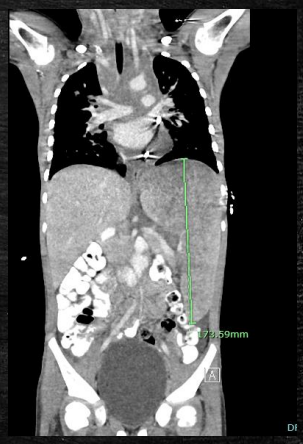
CT :



T8 vertebrae diffusely osteopenic w/out discrete lesion or pathologic fracture. Infiltrative marrow process is possible



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- Lymph node biopsy: no obvious lymph node easily accessible for biopsy
- Has lots of pancytopenias… So the team decided to get a PET scan to try to find a high yeild target

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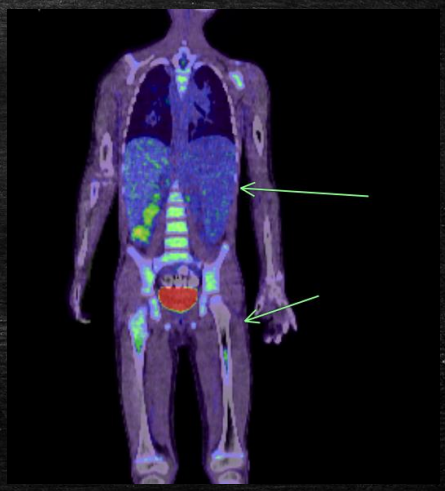
PET



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Next Steps

- Right femur bone marrow biopsy unrevealing
- Continued to have ~1L pericardial drain output per day

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Differential?

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Kaposiform Lymphangiomatosis[1]

- Newer subtype of generalized lymphatic anomaly
- "Kaposiform" hemosiderotic spindled lymphatic endothelial cells among abnormal and enlarged lymphatic channels
- Abnormal development of lymphatic system; does not appear to be genetic
- Usually presents with respiratory symptoms or hemorrhagic effusion

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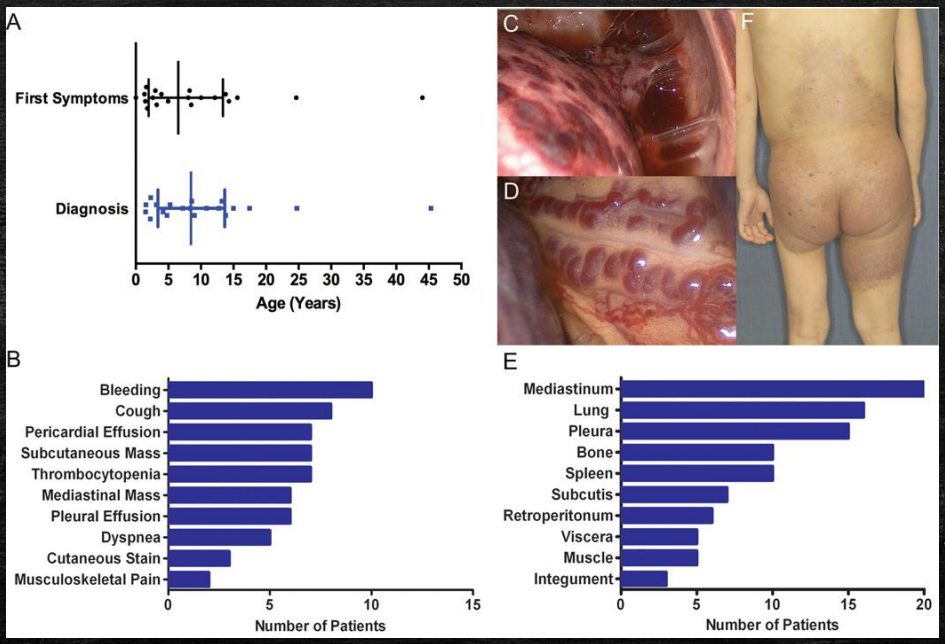

(Crane et al, 2014)

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Kaposiform Lymphangiomatosis

- Sirolimus (~50% partial response, 25% stable disease, 17% disease progression) [2]
- Transfusion requirements improved on KLA therapies
- Angiopoietin 2- test elevated
- Whole body MRI: increased size of pericardial-mediastinal-lower neck lymphatic malformation and evidence of pulmonary lymphangiomatosis.
- Started on zoledronic acid [3], anakinra

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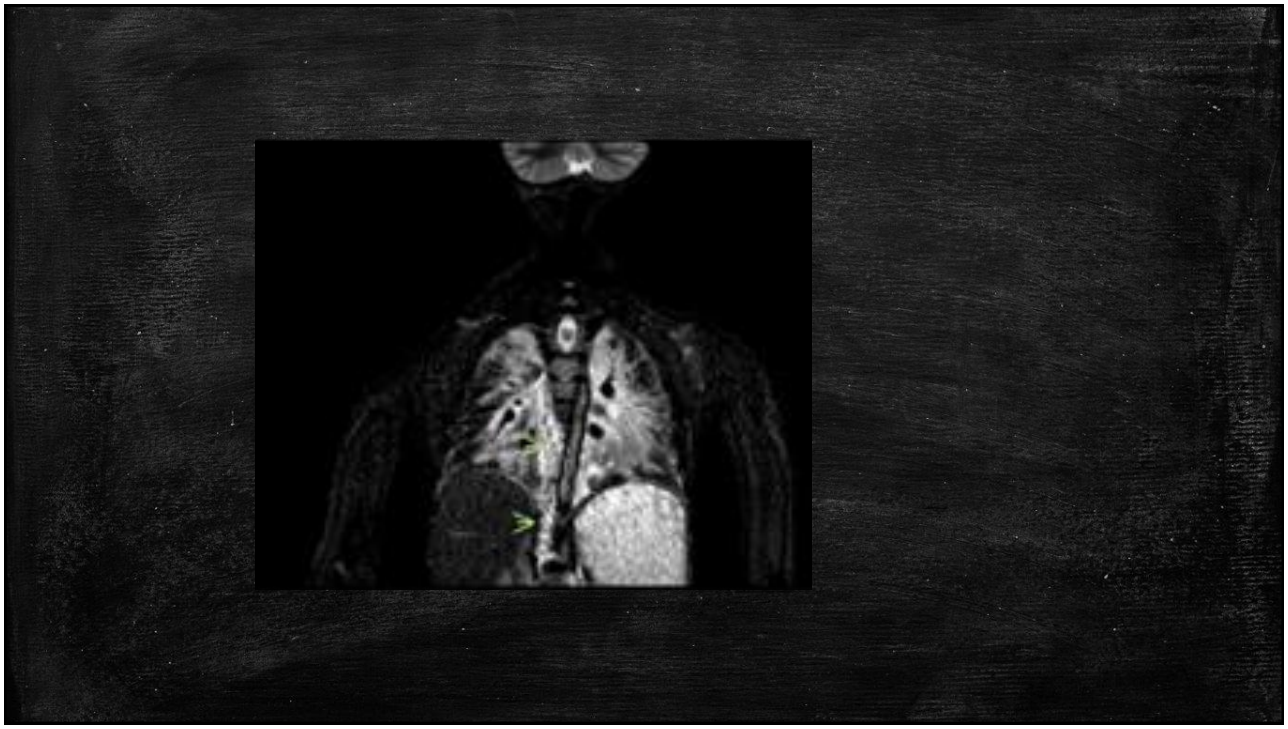

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Questions?

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References

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2. Zhou, J., Yang, K., Chen, S. *et al.* Sirolimus in the treatment of kaposiform lymphangiomatosis. *Orphanet J Rare Dis* **16**, 260 (2021). https://doi.org/10.1186/s13023-021-01893-3

3. Crane J, Manfredo J, Boscolo E, Coyan M, Takemoto C, Itkin M, Adams DM, Le Cras TD. Kaposiform lymphangiomatosis treated with multimodal therapy improves coagulopathy and reduces blood angiopoietin-2 levels. Pediatr Blood Cancer. 2020 Sep;67(9):e28529. doi: 10.1002/pbc.28529. Epub 2020 Jul 7. PMID: 32634277; PMCID: PMC8554683.

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