AMSER Case of the Month August 2026

19-month-old female with an enlarged left fourth digit

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Patient Presentation

- HPI: a 19-month-old girl presents to a well child visit with a one-week history of a “swollen” left ring finger. Her mother is concerned about a possible injury to the area.
- PMHx: None. All developmental milestones on time.
- PE:
  - General: Irritable with exam but easily consolable
  - Upper extremities: Moderate enlargement of proximal and middle phalanx of left fourth digit. No erythema or ecchymosis. Full range of motion.
  - Skin: Fading purple, flat lesion on her abdomen that blanches and crosses midline. Two < 0.5 cm cafe au-lait spots on buttocks. Mild pigmentation of bilateral axillae.
- Labs: none ordered

What Imaging Should Be Ordered?

Select the applicable ACR Appropriateness Criteria

This imaging modality was ordered by the pediatrician

Variant: 1 **Acute blunt or penetrating trauma to the hand or wrist. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
Radiography area of interest	Usually Appropriate	Varies
US area of interest	Usually Not Appropriate	O
MRI area of interest without and with IV contrast	Usually Not Appropriate	O
MRI area of interest without IV contrast	Usually Not Appropriate	O
Bone scan area of interest	Usually Not Appropriate	☢☢☢
CT area of interest with IV contrast	Usually Not Appropriate	Varies
CT area of interest without and with IV contrast	Usually Not Appropriate	Varies
CT area of interest without IV contrast	Usually Not Appropriate	Varies

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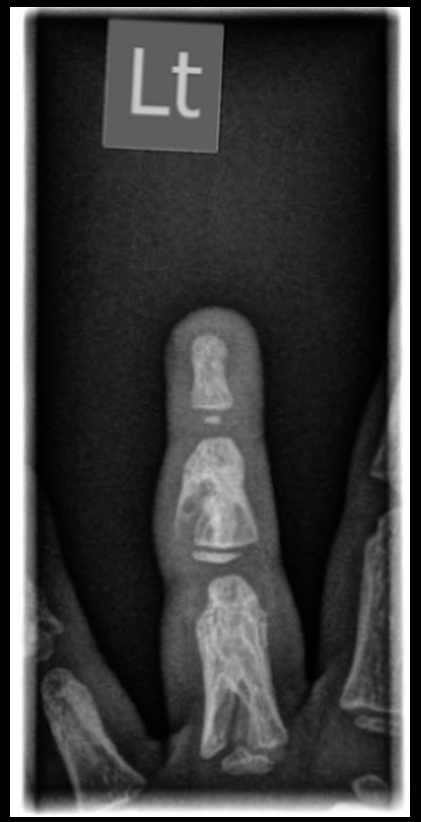
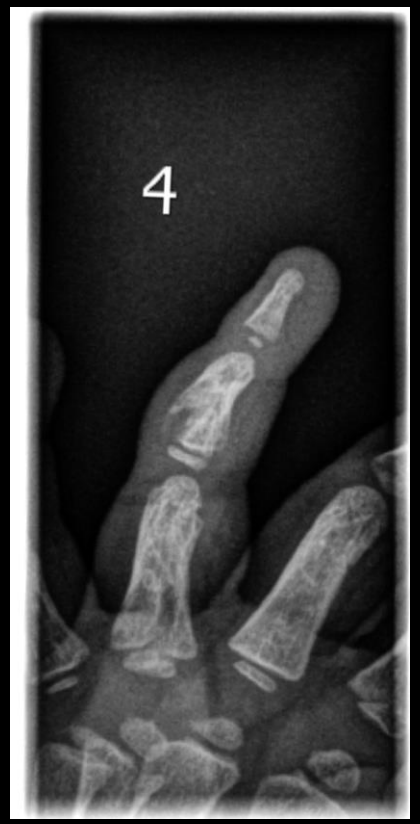
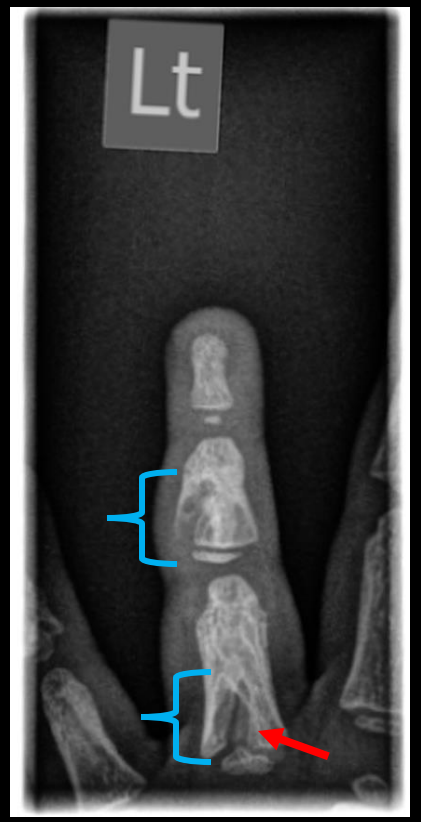
Findings (unlabeled)

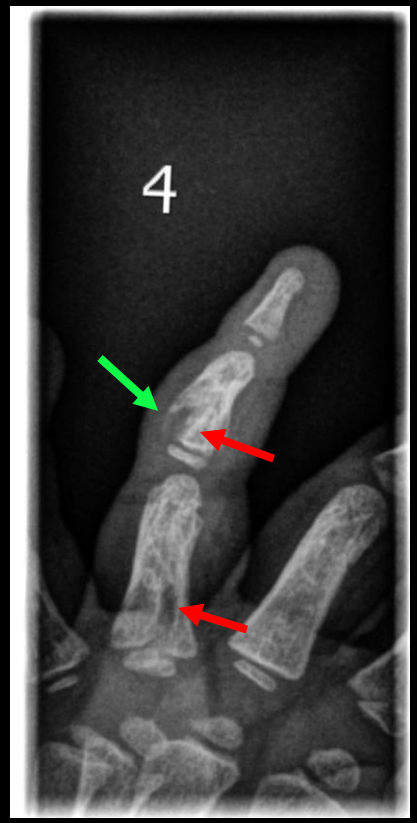


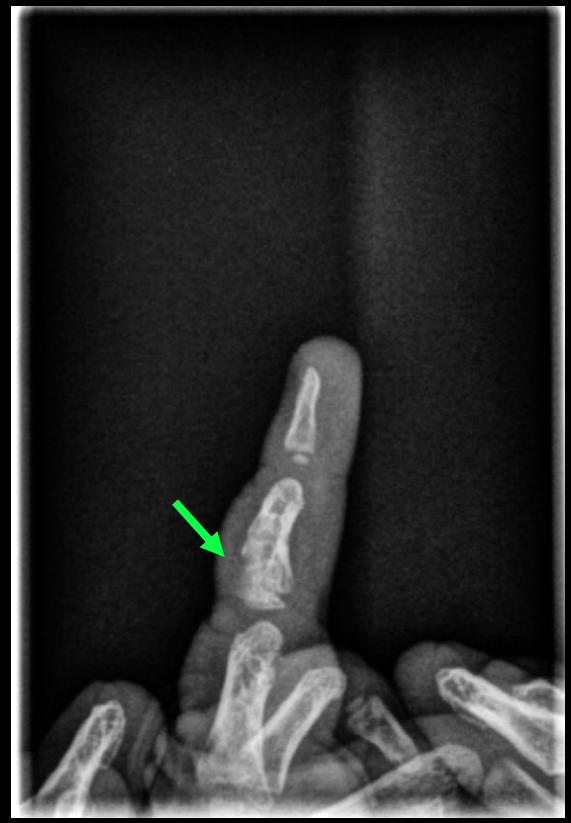


PA, oblique, and lateral views of left fourth digit

Findings (labeled)







PA, oblique, and lateral views of left fourth digit

- Expansile, intramedullary, lytic lesions in the proximal and middle phalanges of the left fourth digit
- Cortical discontinuity with overlying soft tissue swelling most prominent in the middle phalanx
- Narrow zone of transition visualized in the proximal and middle phalanges

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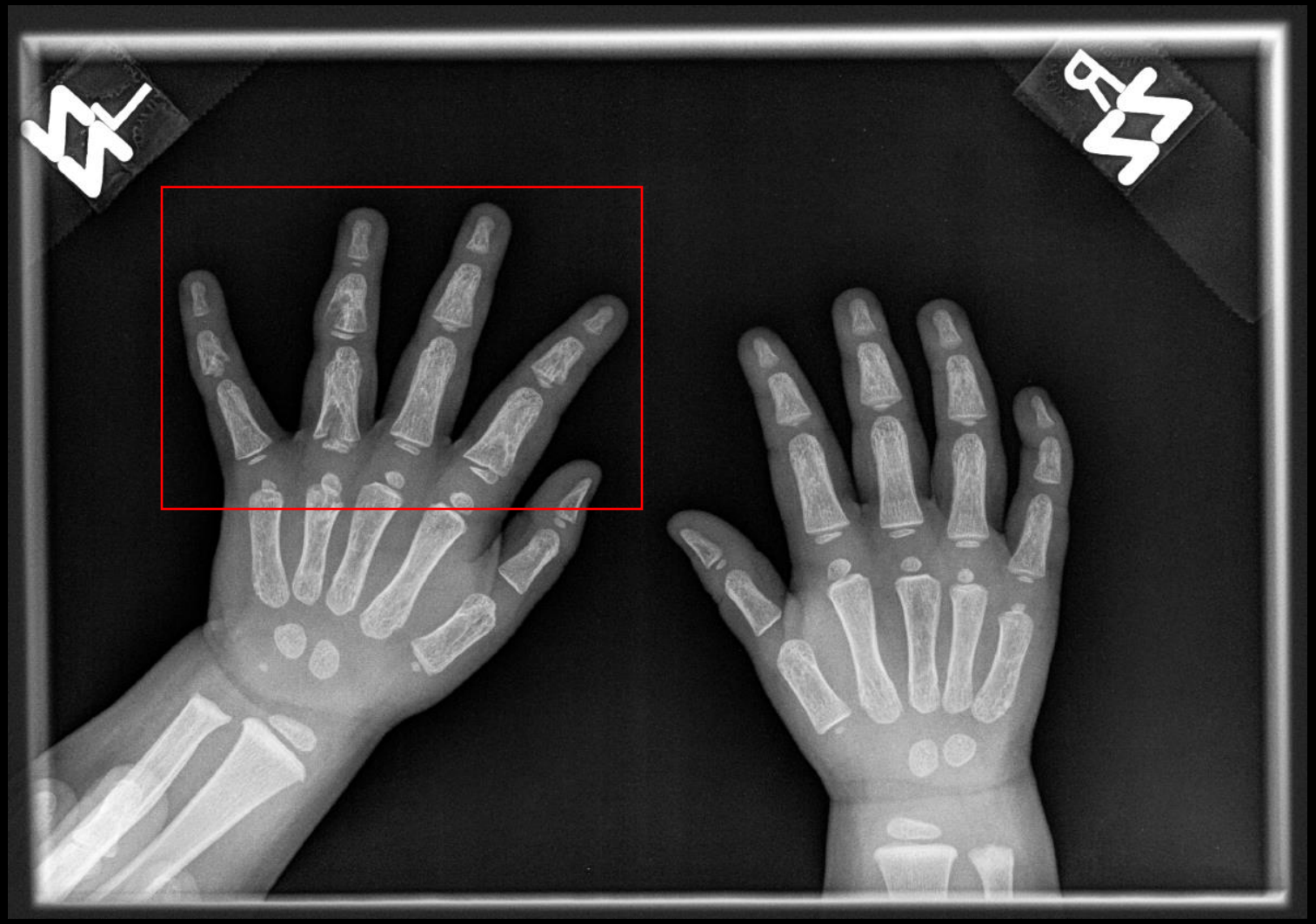
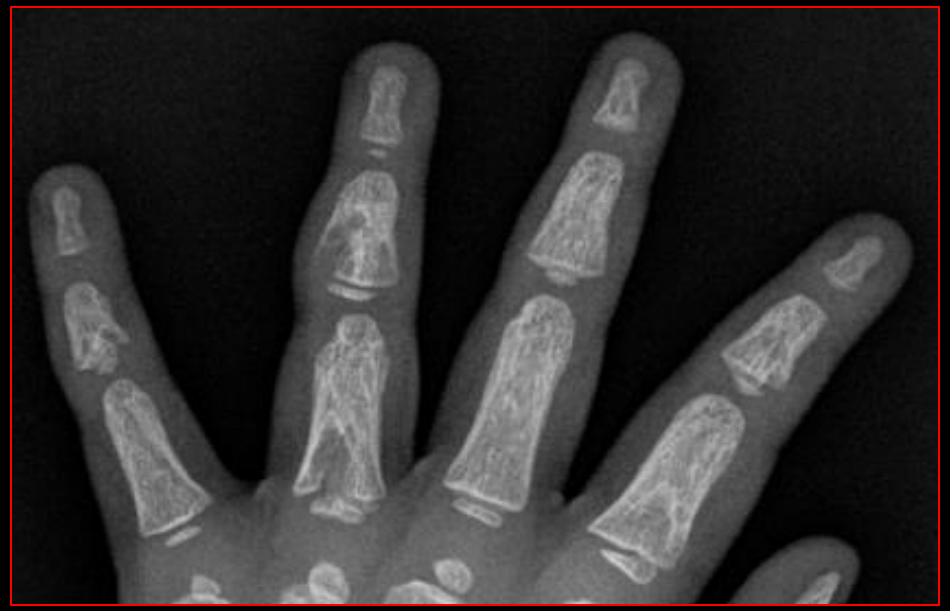
Clinical Course

- Patient was referred to orthopedics, endocrinology, and genetics
- A skeletal survey was ordered as part of further workup

Findings (unlabeled)

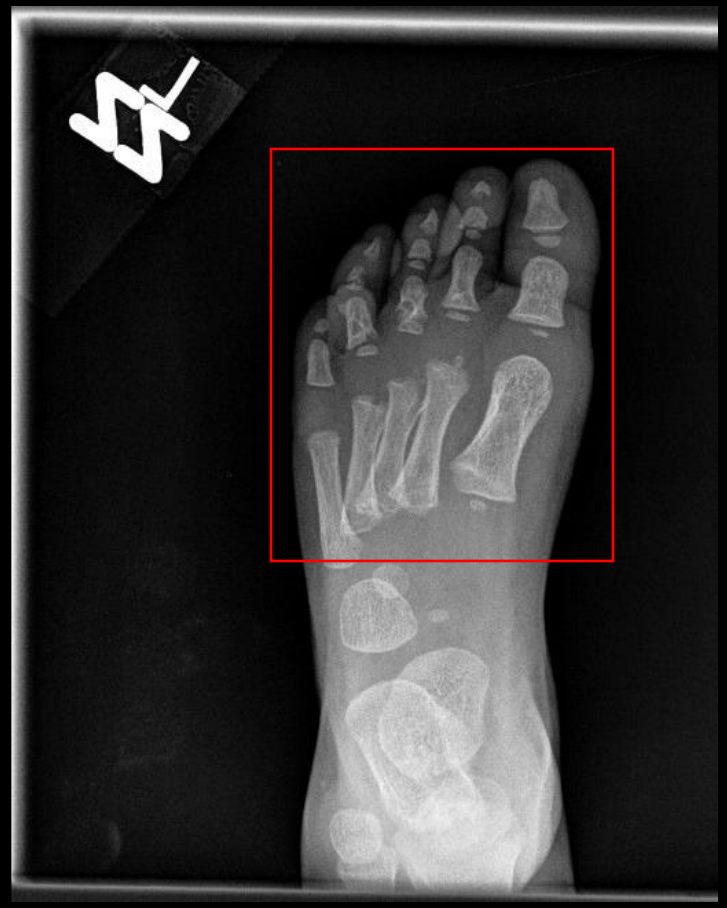


Bilateral PA view

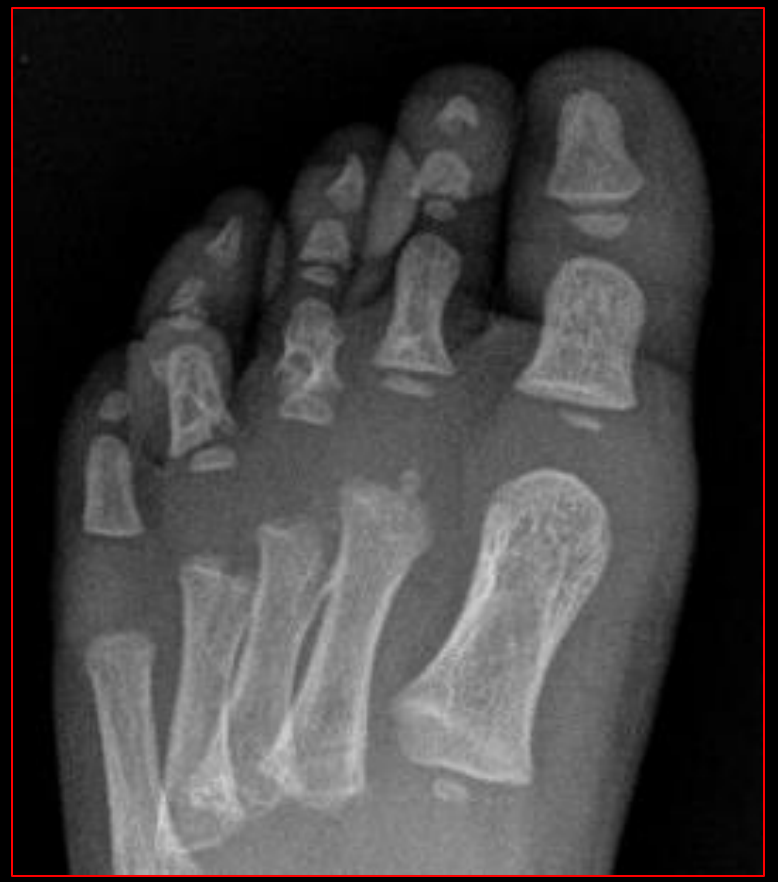


Magnified view

Findings (unlabeled)

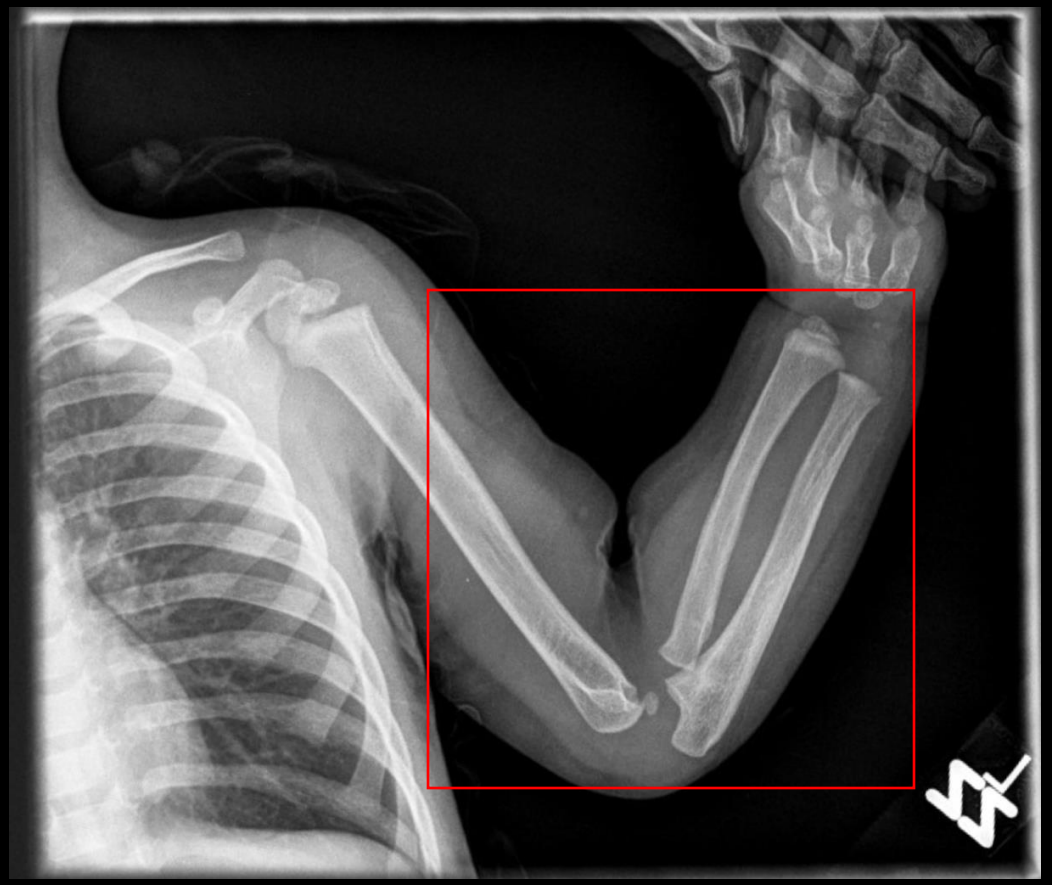


Dorsoplantar view, left foot

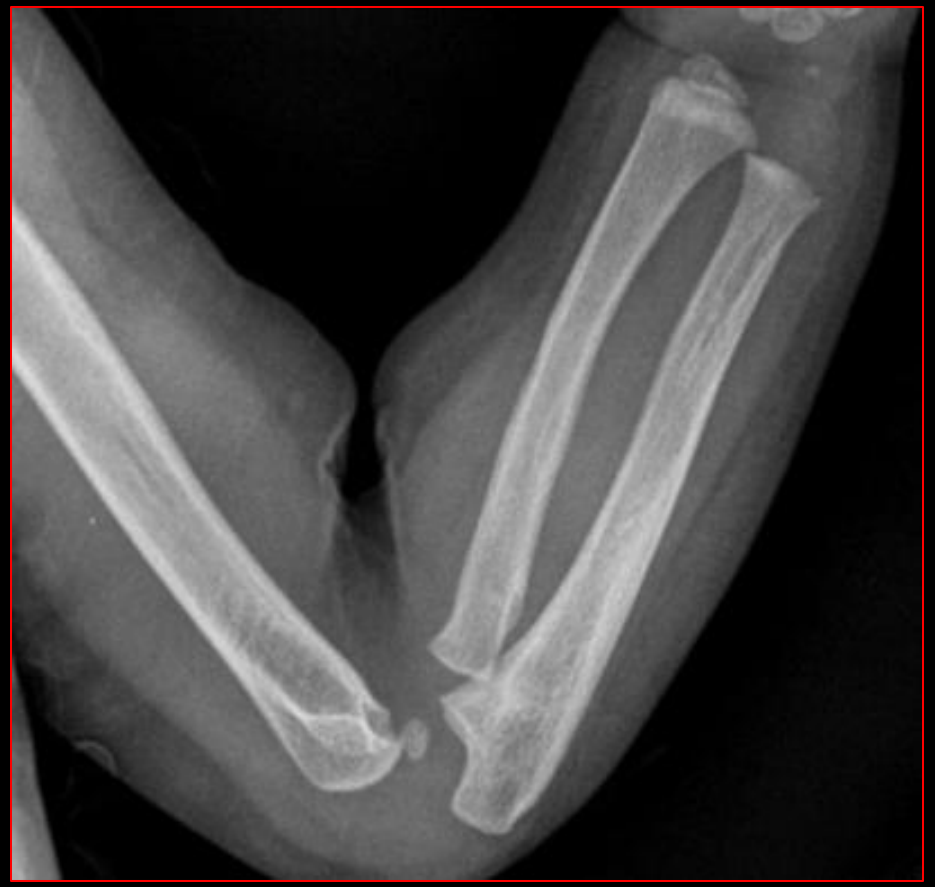


Magnified view

Findings (unlabeled)

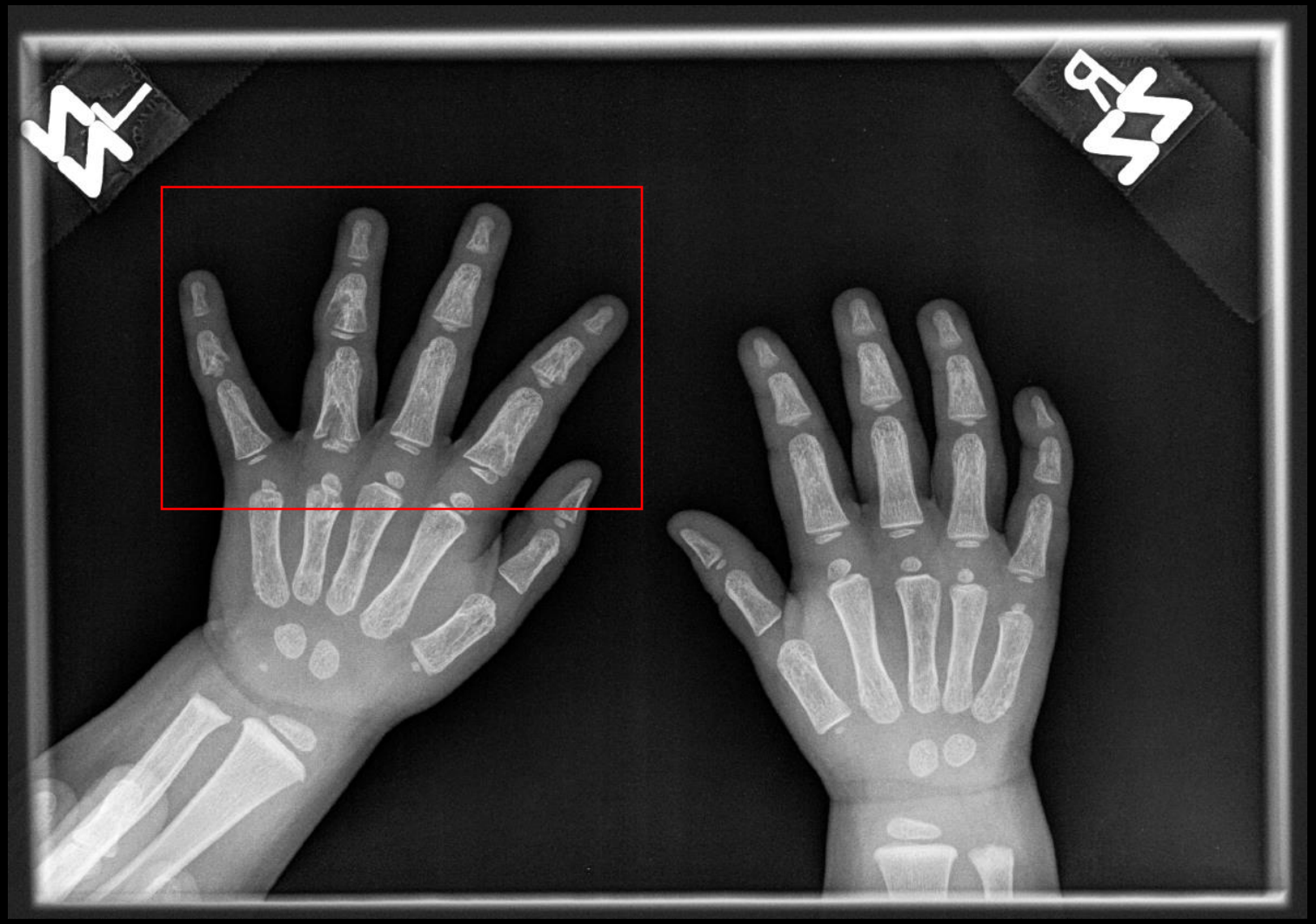


AP view, left upper extremity

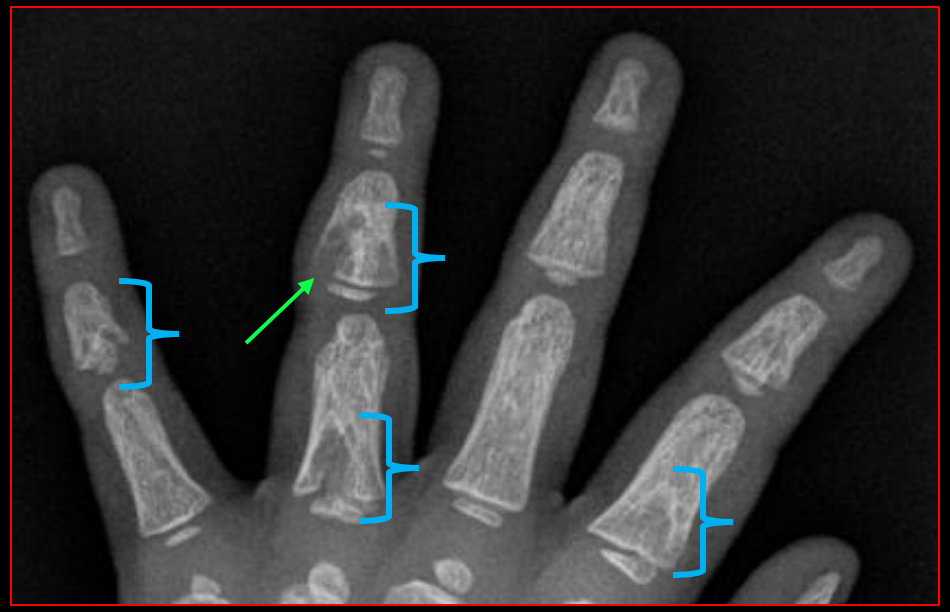


Magnified view

Findings (labeled)



Bilateral PA view



Magnified view

- Multiple well-marginated, expansile intramedullary lytic lesions in the second, fourth, and fifth digits of the left hand
- Endosteal scalloping and cortical thinning most prominent in the middle phalanx of the fourth digit

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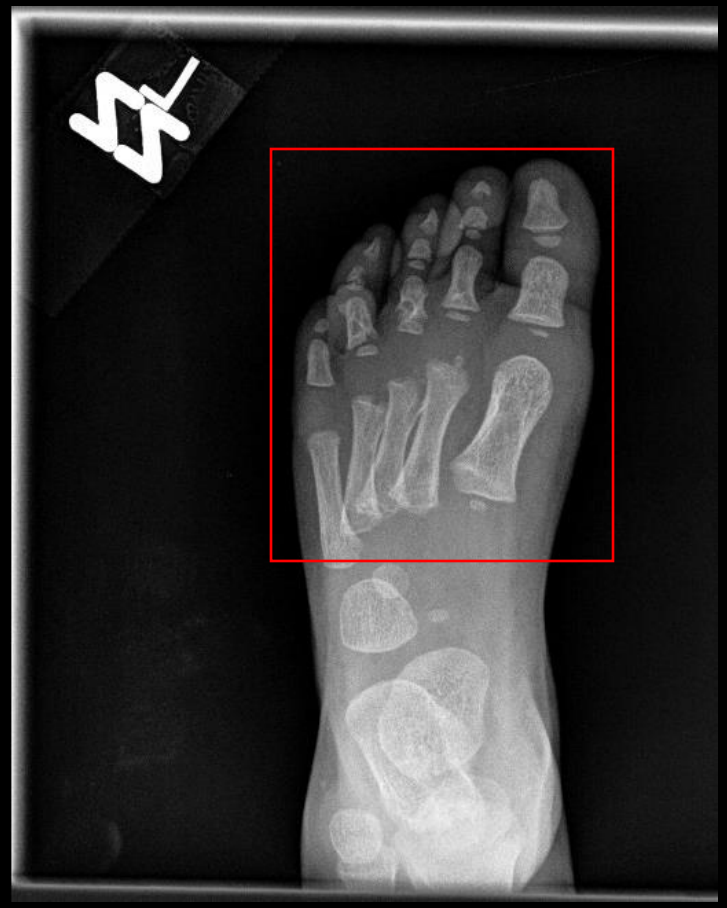
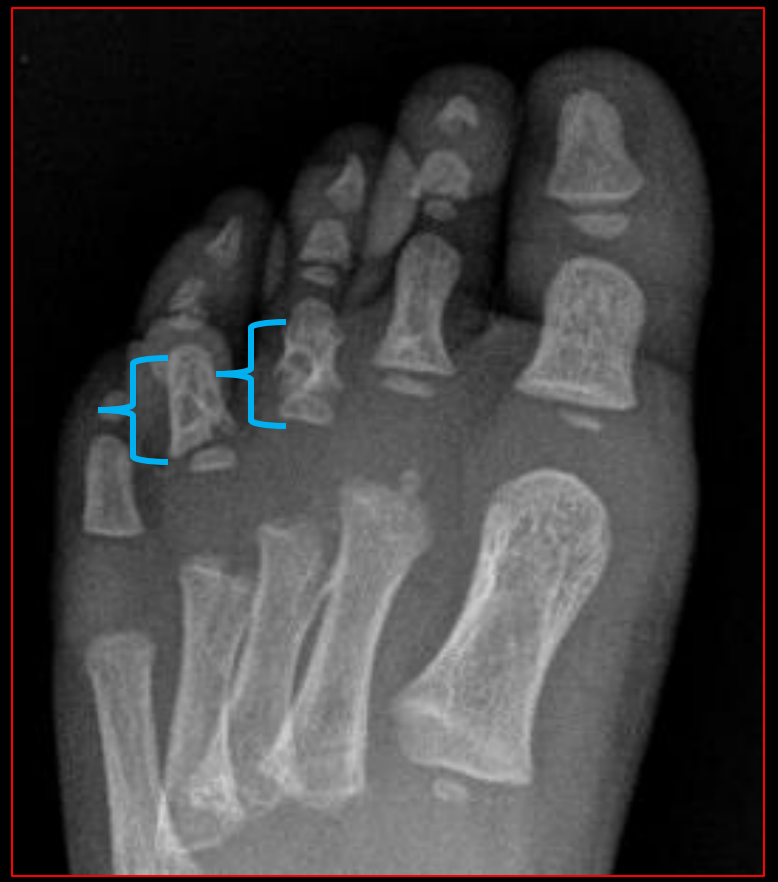
Findings (labeled)



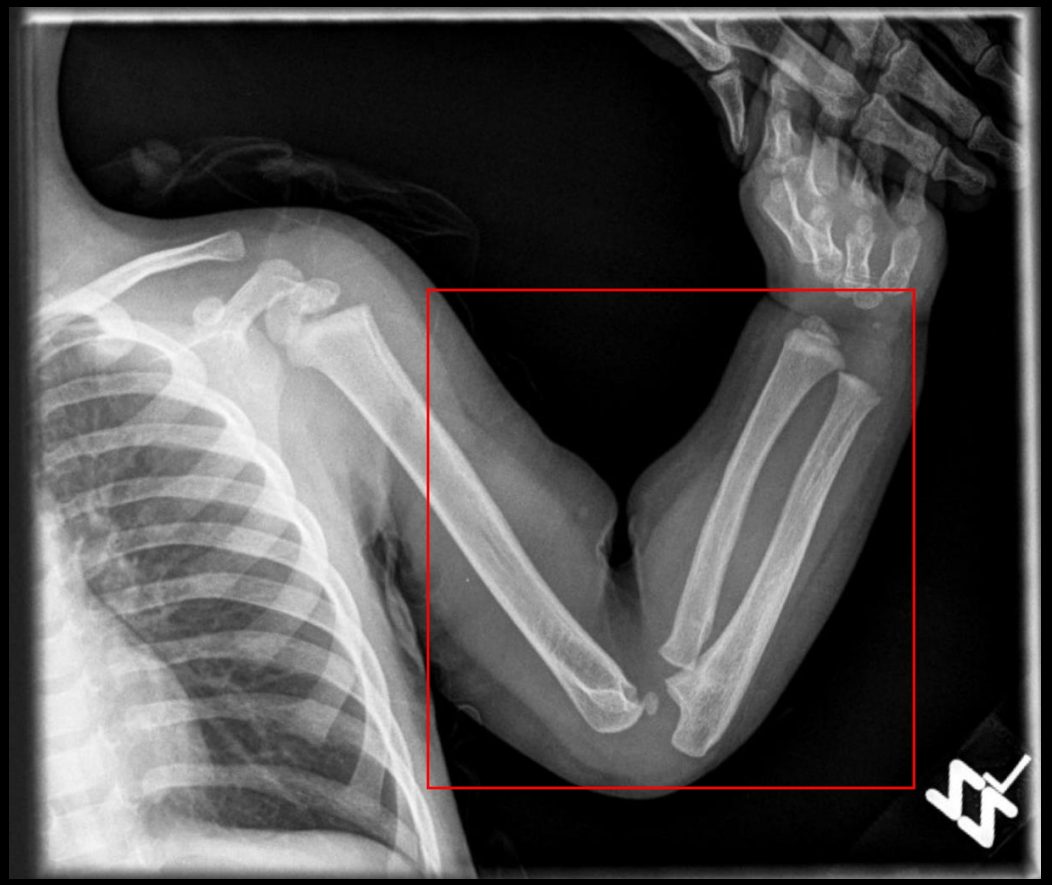
Dorsoplantar view, left foot



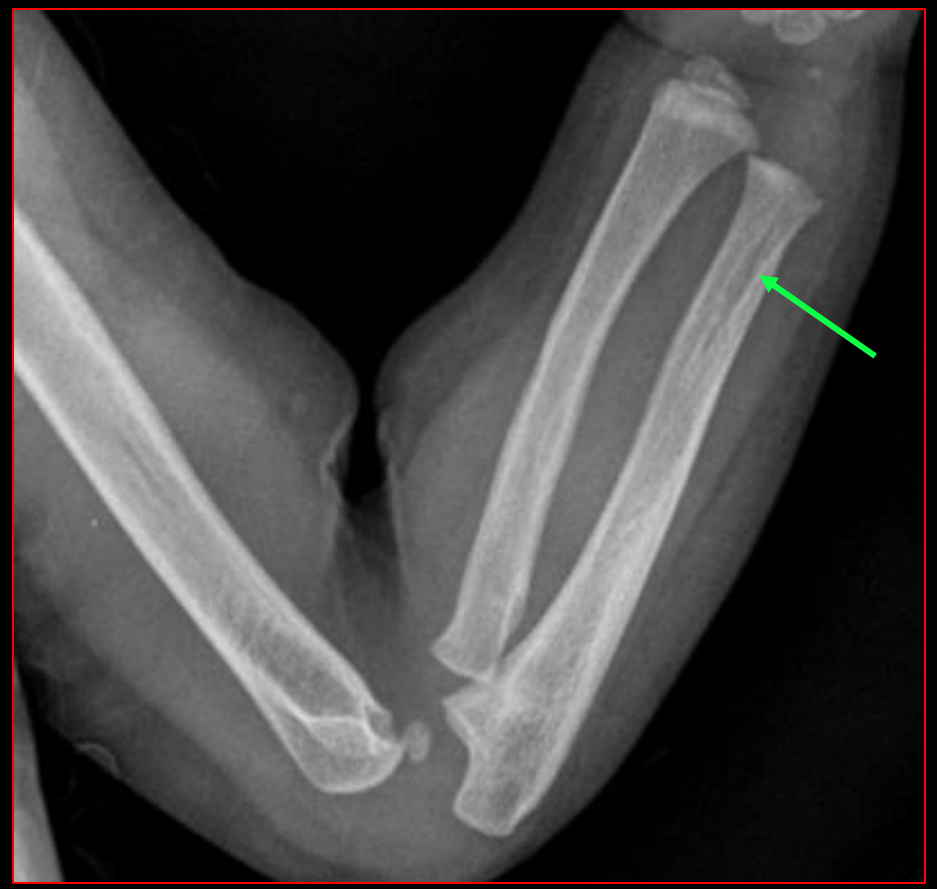
Magnified view

- Well-marginated, mildly expansile lytic regions in the proximal phalanges of the third and fourth digits of the left foot

Findings (labeled)



AP view, left upper extremity



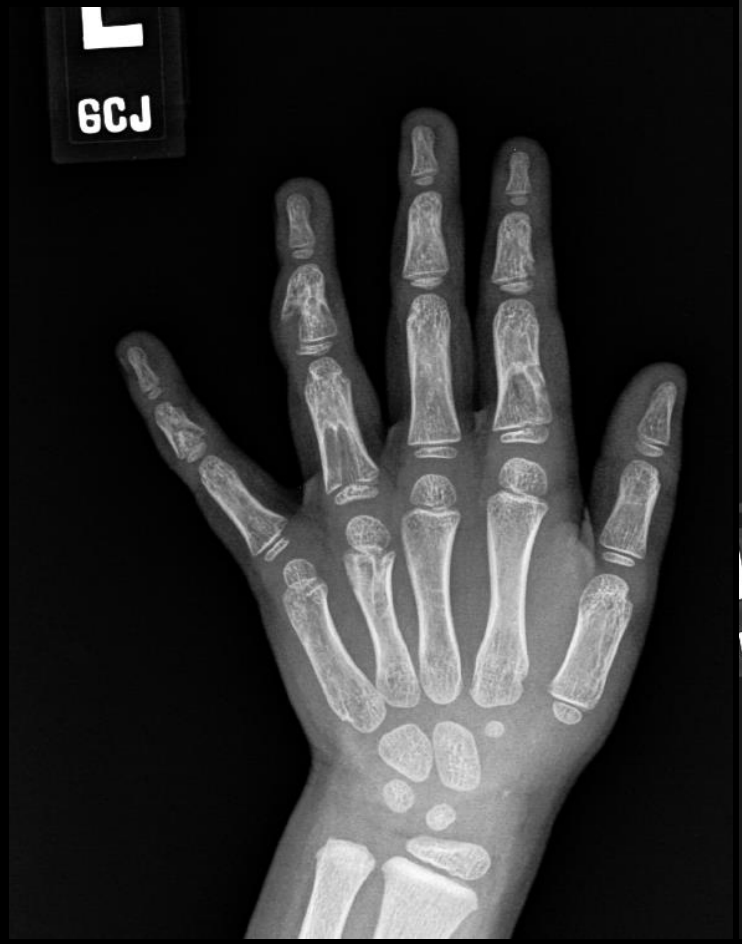
Magnified view

- Linear radiolucency in left distal ulna
- Findings overall most likely represent multiple unilateral enchondromas

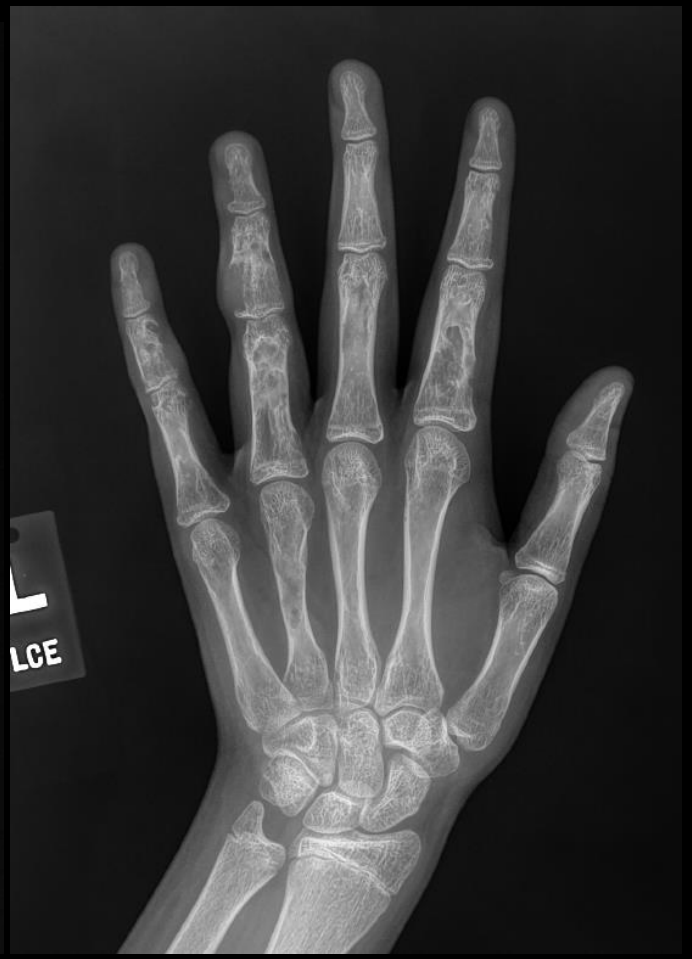
Final Dx:

Ollier Disease

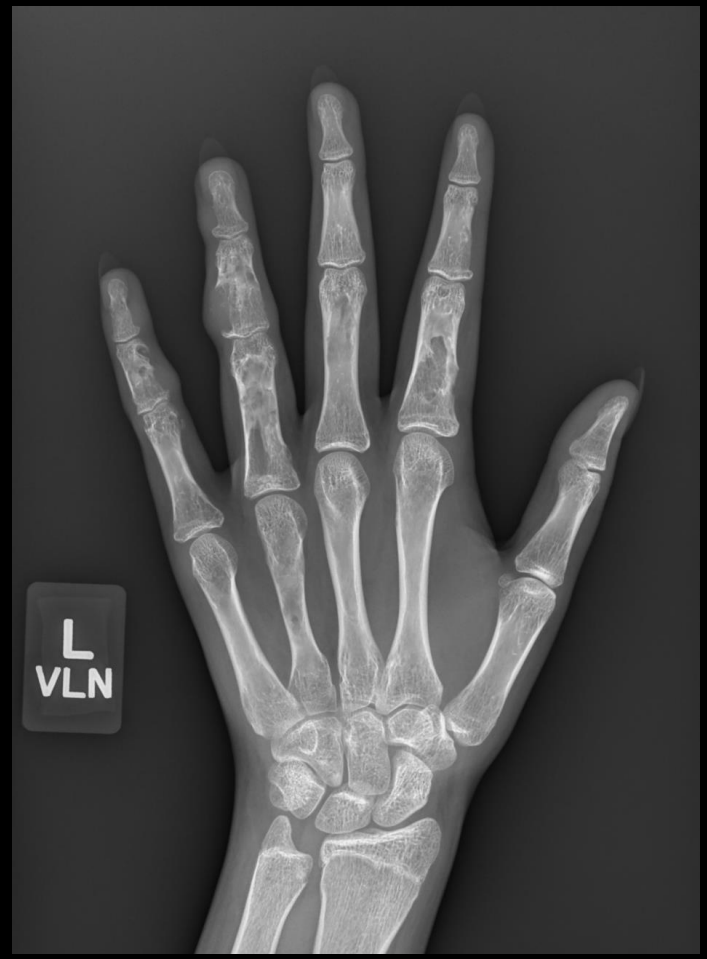
Further Surveillance



Age 3



Age 15



Age 17

Stable appearance over time.

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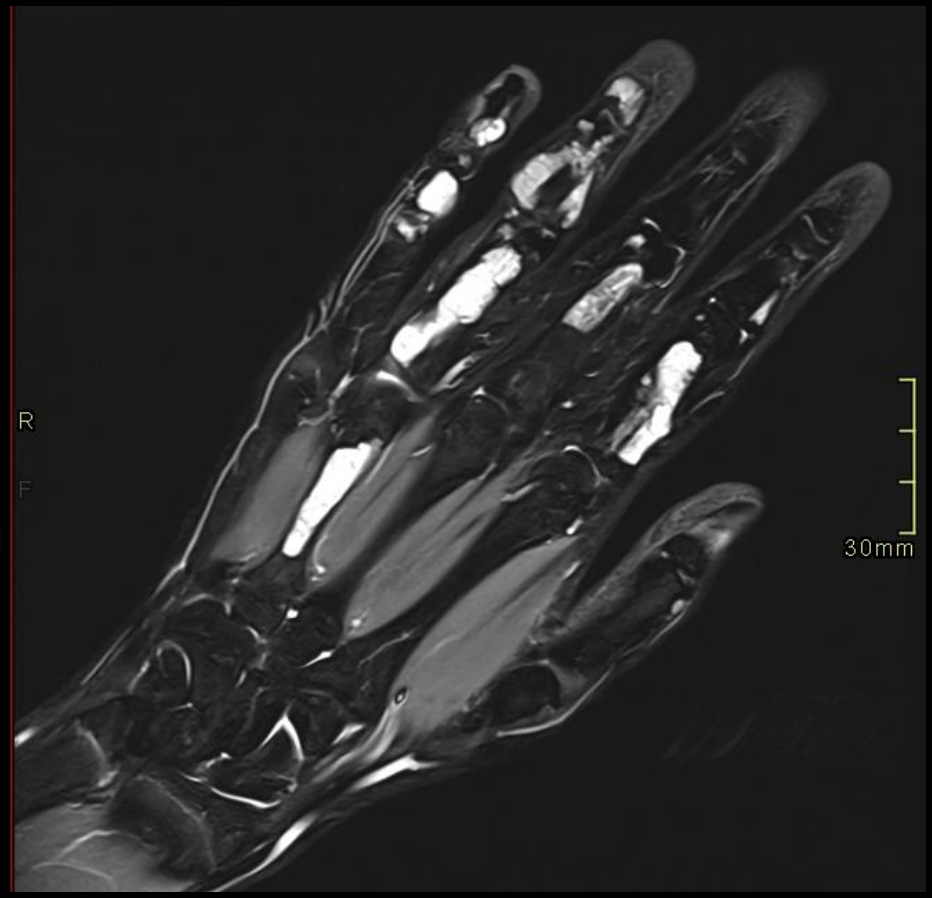
Preoperative MRI (unlabeled)

T1



STIR

Patient opted for operative repair given cosmetic concerns and mild but worsening functional limitation (inability to make a complete fist)

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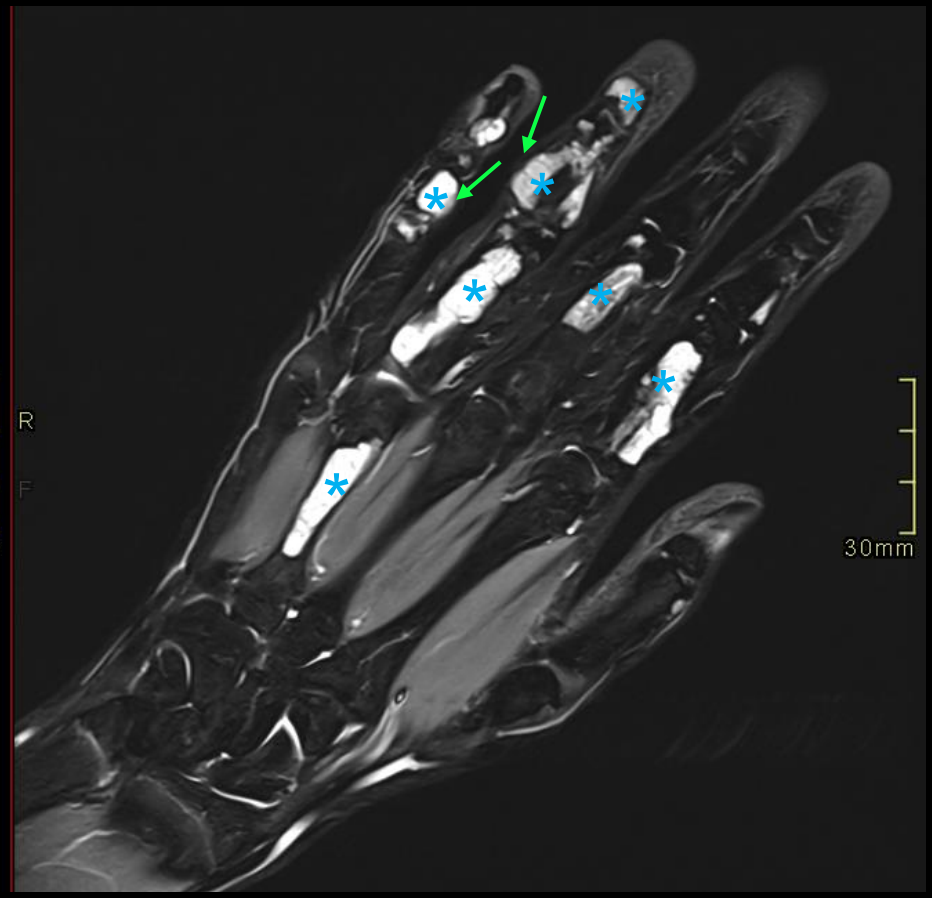
Preoperative MRI (labeled)

T1



STIR

- Lobulated T1-hypointense, T2-hyperintense intramedullary lesions involving the fourth metacarpal and multiple phalanges of the left hand
- Endosteal scalloping and cortical thinning in the middle phalanges of the fourth and fifth digits

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Case Discussion

- Enchondromas are benign intraosseous tumors arising from hyaline cartilage that are most commonly found in the short and long tubular bones of the limbs[1,2]
  - Multiple enchondromas (3+)
    - Multiple enchondromatosis: various subtypes exist within the Spranger classification system. Enchondromas are mostly symmetrically distributed[1,3]
    - Ollier disease: asymmetric distribution[3]
    - Maffucci syndrome: asymmetric enchondroma distribution + soft tissue hemangiomas[3]
- Ollier disease often manifests in the first decade of life and has a prevalence of 1/100,000[1,2]
- Linked to somatic mutations in IDH1 and IDH2 in 80% of patients[1,4]
- Malignant transformation to chondrosarcoma is seen in 30% of patients by the age of 40[1], and in 50% of patients over their lifetime[4]
  - Gliomas (2-16% of patients), granulosa cell tumors, acute myeloid leukemia also associated[4,5]

Case Discussion

- Diagnosis is based on clinical history and imaging[2]
  - Clinical manifestations vary with severity and include painless bony masses, asymmetric limb shortening or bowing, limitations in articular movement, or pathologic fracture[1,2]
  - Radiography is the initial modality of choice, and shows multiple, radiolucent, homogenous lesions with an elongated shape and well-defined, sclerotic bony margin, without periosteal reaction. Chondroid calcification may be seen. Skeletal surveys historically have been used for determining disease extent[2,3]
  - MRI of focal areas is useful for operative planning or evaluating malignant transformation[1,3]
- The role of imaging in further surveillance is under discussion given newer understandings of lifetime cancer risk[4-6]. For example, in 2023, the AACR Pediatric Cancer Working Group recommended:
  - Whole body MRI at time of diagnosis (or deferred until patient can tolerate scan without anesthesia)
  - Plain radiographs of known enchondromas every 2-3 years alongside annual physical exam
  - Annual MRI of lesions >6cm or lesions in pelvis or scapula (greater risk of malignant transformation)
  - Dedicated brain MRI with contrast in early adolescence for glioma detection
  - Whole body MRI every 1-2 years after the age of 20[4]

Case Discussion

- Treatment is conservative
  - If significant bony deformity, common interventions include osteotomy or intralesional curettage with corticoplasty[1,7]
  - Resection and radiotherapy for malignant transformation[1,2]
- Differential diagnosis
  - Polyostotic fibrous dysplasia: similar age of incidence, but presence of fibrous rather than cartilaginous tissue in the intramedullary space. Presents on radiography as well-defined, osteolytic lesions. Associated with McCune-Albright Syndrome[8]
    - This patient's café-au-lait spots were inconsistent with the larger, more irregularly shaped lesions characteristic of McCune-Albright Syndrome
  - Maffucci syndrome
    - This patient had an abdominal port wine stain, which is a capillary malformation rather than a true hemangioma
  - Hereditary multiple osteochondroma: cartilage-capped lesions lie at the bone surface and project outward[2]

References

1. Kumar, A., Jain, V. K., Bharadwaj, M., & Arya, R. K. (2015). Ollier disease: Pathogenesis, diagnosis, and management. Orthopedics, 38(6). https://doi.org/10.3928/01477447-20150603-58

2. Silve, C., & Jüppner, H. (2006). Ollier disease. Orphanet Journal of Rare Diseases, 1, 37. https://doi.org/10.1186/1750-1172-1-37

3. Weerakkody, Y., Campos, A., & Sharma, R. (2009). Enchondromatosis. In Radiopaedia.org. Radiopaedia.org. https://doi.org/10.53347/rID-6678

4. Michaeli, O., Kim, S. Y., Mitchell, S. G., Jongmans, M. C. J., Wasserman, J. D., Perrino, M. R., Das, A., MacFarland, S. P., Scollon, S. R., Greer, M.-L. C., Sobreira, N., Gallinger, B., Lupo, P. J., Malkin, D., Schneider, K. W., Schultz, K. A. P., Foulkes, W. D., Woodward, E. R., &Stewart, D. R. (2025). Update on cancer screening in children with syndromes of bone lesions, hereditary leiomyomatosis and renal cell carcinoma syndrome, and other rare syndromes. Clinical Cancer Research, 31(3), 457–465. https://doi.org/10.1158/1078-0432.CCR-24-2171

5. El Abiad, J. M., Robbins, S. M., Cohen, B., Levin, A. S., Valle, D. L., Morris, C. D., & De Macena Sobreira, N. L. (2020). Natural history of Ollier disease and Maffucci syndrome: Patient survey and review of clinical literature. American Journal of Medical Genetics Part A, 182(5),1093–1103. https://doi.org/10.1002/ajmg.a.61530

6. Deckers, C., Schreuder, B. H. W., Hannink, G., De Rooy, J. W. J., & Van Der Geest, I. C. M. (2016). Radiologic follow-up of untreated enchondroma and atypical cartilaginous tumors in the long bones. Journal of Surgical Oncology, 114(8), 987–991. https://doi.org/10.1002/jso.24465

7. Klein, C., Delcourt, T., Salon, A., Finidori, G., Glorion, C., & Pannier, S. (2018). Surgical treatment of enchondromas of the hand during childhood in ollier disease. The Journal of Hand Surgery, 43(10), 946.e1-946.e5. https://doi.org/10.1016/j.jhsa.2018.02.010

8. Singh G, Feger J, Silverstone L, et al. Fibrous dysplasia. In Radiopaedia.org https://radiopaedia.org/articles/4915

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