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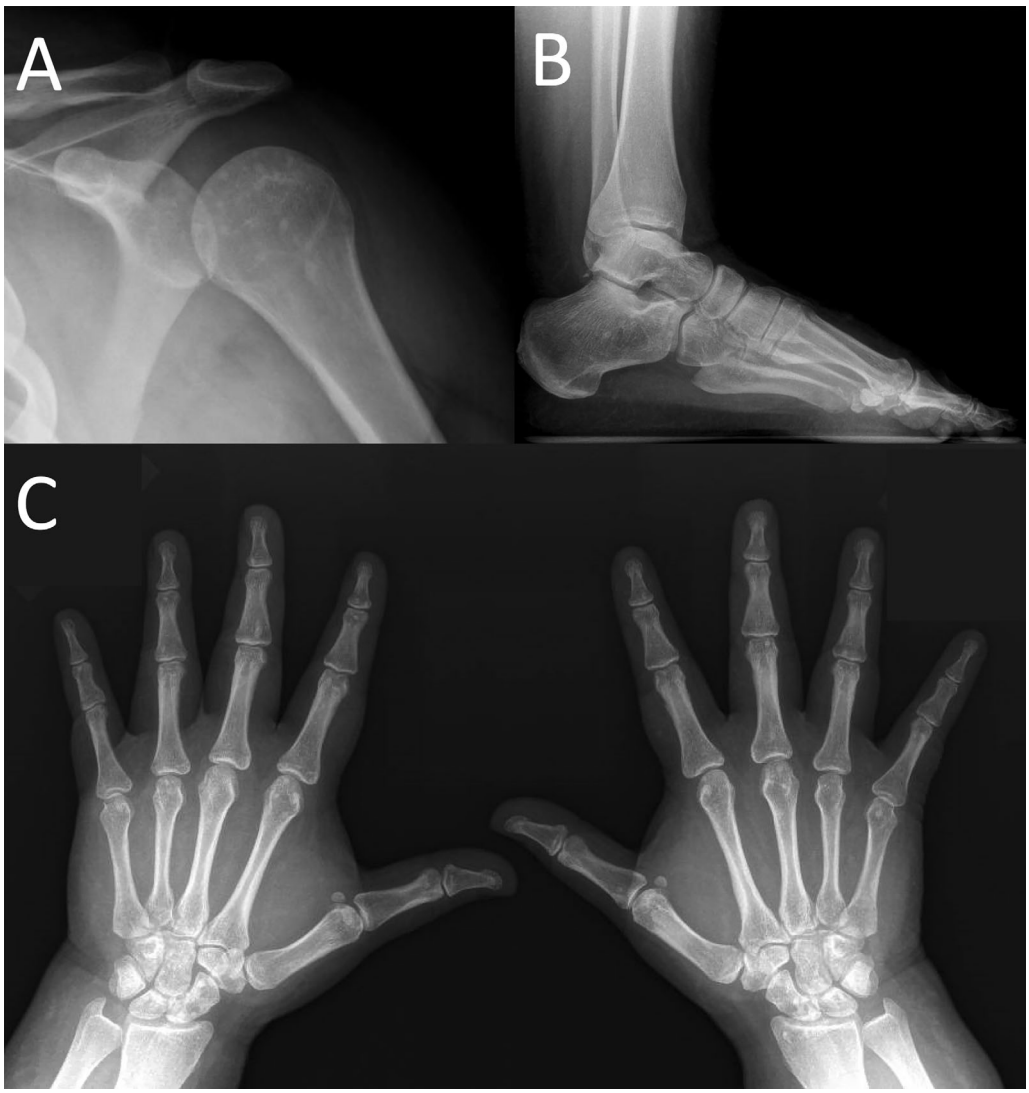
Clinical Images: Spotted bone: Incidental detection of a rare genetic disease



The patient, a 27-year-old woman with a history of Hashimoto thyroiditis, presented to the rheumatology clinic for upper and lower extremity swelling. A physical examination demonstrated hand swelling and flesh-colored lichenified papules on her sacrum, neck, and ankles. Results of laboratory workup showed weakly positive antinuclear antibodies (1:320) with a nuclear fine-speckled pattern but otherwise no autoantibodies. A skin biopsy specimen demonstrated thickened dermal collagen consistent with connective tissue nevus. We obtained anterior–posterior (AP), axial, and Grashey views on radiographs of both shoulders (**A**); AP, lateral, and oblique radiographs of both feet (**B**); and posterior–anterior, lateral, and oblique radiographs of both hands (**C**). Each view demonstrated diffusely distributed small sclerotic densities overlying the normal osseous structures consistent with scattered enostoses. The differential diagnosis included sclerosing bony dysplasias (sporadic or inherited), sclerotic metastases, osteomas, and osteosarcomas (1). Genetic testing revealed a heterozygous mutation in *LEMD3*. Her constellation of findings was consistent with Buschke-Ollendorff syndrome, an autosomal dominant condition in which diagnostic features include osteopoikilosis, connective tissue nevi, an *LEMD3* mutation, and a family history (2). Loss-of-function mutations in *LEMD3*, a gene involved in bone morphogenic protein signaling, have been previously described in cases of Buschke-Ollendorff syndrome, although the exact relationship is yet unknown (3). Imaging features that may help differentiate osteopoikilosis from other malignant disorders include its preponderance for the epiphyseal regions of short tubular bones, lesion size less than 1 cm, homogenously radiodense lesions with spiculated margins, and lack of uptake on bone scintigraphy (1,4).

Author disclosures are available at https://onlinelibrary.wiley.com/action/downloadSupplement?doi=10.1002%2Facr2.11377&file=acr211377-sup-0001-Disclosureform.pdf.

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