Visual Vignette

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Case presentation: A 31-year-old man presented with stiffness of the left lower limb for 5 years, and pain over the left leg for the last 2 months following a fall. There was no history of pain in the other limbs or elsewhere over the body. There was no history of fever or weight loss. There was no associated muscle weakness, and no history to suggest connective tissue disorders. He was not known to have other comorbid conditions. On examination, there was thickening and knobby irregularity palpable along the entire length of the bones of the left leg and thigh. Other sites were not involved. On investigation, bone biochemical parameters were normal. X-rays of the left femur (Fig. 1) and left tibia (Fig. 2) were performed. **What is the diagnosis?**

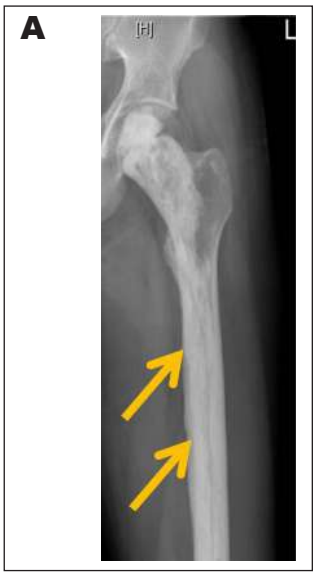

Fig. 1.

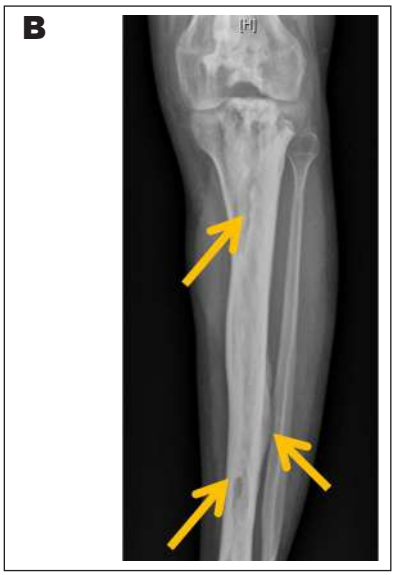

Fig. 2.

Answer: Melorheostosis. Radiology of the left femur and the left tibia showed thickening of bones with cortical and endomedullary hyperostosis diagnostic of melorheostosis. Although the patient did not complain of pain in the left hip at presentation, X-ray revealed degenerative changes involving the left hip joint. Melorheostosis (Leri disease, candle bone disease, melting wax syndrome) is a benign sclerosing bone dysplasia, characterized by the mesodermal dysplasia of the bone. Melorheostosis is derived from the Greek word (*melos* = limb, *rheos* = flow) due to the classic radiologic appearance of flowing hyperostosis resembling hardened wax, dripping down the side of a candle (1). Men and women are equally affected and no hereditary pattern has been described. Symptoms usually begin with pain of the affected limb, as a result of subperiosteal new bone formation. Restriction of movement and joint deformities develops gradually (2). Melorheostosis usually affects long bones of the upper and lower limbs, as well as the small bones of the hands and feet. Axial skeletal involvement is rare. Although the monomelic variant is the most common form, it may also present as the monostotic or polyostotic form. The etiology of the disease is not completely understood. One possible etiology is the loss of function mutation of the LEM domain-containing protein 3 gene (11q12-12q14.2), a protein involved in bone morphogenic protein and transforming growth factor beta signaling (1). Medical management with bisphosphonates reduces bone pain and slows progression of the disease. Surgical treatment includes tendon lengthening, excision of hyperostotic bone, sympathectomy, and rarely amputation (3). Lesions are likely to recur after a surgical procedure. Although melorheostosis does not shorten a patient's lifespan, it may cause considerable morbidity, and treatment is mostly symptomatic, with analgesics and physiotherapy of the involved limb.

DISCLOSURE

The authors have no multiplicity of interest to disclose.

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