

Visual Vignette

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Case Presentation: A 32-year-old female presented with history of pain in left hip and knee joints of 2 months duration preceded by a fall. There was no history of swelling, limitation of movements, or fracture. There was no previous history of systemic illness or long-term medication use. There was no history of fever, weight loss, previous fragility fractures, or bone pains. There was no associated muscle weakness or any history to suggest connective tissue disorders. On examination, there were no cutaneous lesions or restriction of the movements, and systemic examination was normal. Bone mineral biochemistry was unremarkable. Radiography of the hips and knees was performed (Fig. 1 and 2). **What is the diagnosis?**



Fig. 1.



Fig. 2.

Answer: Osteopoikilosis (osteopathia condensans disseminate, spotted bones). The X-ray hip and knee showed multiple small foci of dense spotty areas, diagnostic of osteopoikilosis. This is a rare, autosomal dominant, benign sclerosing disease of bone which is incidentally detected; the incidence is reported to be 1/50,000 (1). It was first described by a German radiologist, Albers-Schönberg, in 1915. Osteopoikilosis may present at any age and is commonly seen in meta-epiphyses of the long bones, pelvis, and extremities (2). It may be associated with nontender connective tissue naevi (3). Although the exact etiology is not known, it is postulated to be due to a loss-of-function mutation in the *LEMD3* gene located on chromosome 12. It is a differential diagnosis for other bone disorders with focal sclerosis, including sclerotic metastases and other sclerosing dysplasias. Bone scintigraphy helps differentiate it from other metastatic bone disorders (1). In our patient, the bone scan was negative. This disease is usually nonprogressive and does not warrant any treatment apart from pain-relieving measures, if symptomatic (2). Our patient was prescribed acetaminophen for pain relief and advised screening of family members.

DISCLOSURE

The authors have no multiplicity of interest to disclose.

REFERENCES

1. Di Primio G. Benign spotted bones: a diagnostic dilemma. *CMAJ*. 2011;183:456-459.
2. Carpintero P, Abad J, Serrano P, Serrano J, Rodríguez P, Castro L. Clinical features of ten cases of osteopoikilosis. *Clin Rheumatol*. 2004;23:505-508.
3. Pope V, Dupuis L, Kannu P, et al. Buschke-Ollendorff syndrome: a novel case series and systematic review. *Br J Dermatol*. 2016;174:723-729.

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