Osteopoikilosis – the importance of recognizing a rare but benign bone disorder

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Osteopoikilosis (OPK) is a rare, benign, hereditary and usually asymptomatic osteodysplastic disease characterized by multiple well-defined sclerotic bone lesions.

We report the case of a 36-year-old female patient sent to the Rheumatology out-patient clinic due to pain in her third right proximal interphalangeal joint lasting for several months, without stiffness, swelling or associated trauma. She had no other symptoms and her medical and family histories were unremarkable. On physical examination, only tenderness in the aforementioned joint was noted. X-rays were ordered and several round, well-defined, juxtaarticular, symmetrical and sclerotic bone lesions were found not only in her hand (Figure 1), but also in her shoulder, knee and pelvis (Figures 2-4). A PET-scan was requested and showed no abnormal isotope uptake. These radiological findings were suggestive of OPK. Pain was managed through rest and non-steroidal anti-inflammatories.

OPK was first described in 1915 by Albers- Schönberg and has an estimated prevalence of 1 in 50,000 individuals[1-4]. It associates with a heterozygous mutation in the LEMD3 gene (chromosome 12) and it can be transmitted through autosomal dominant inheritance with variable penetrance[1-4]. No gender predominance is known[2,3,4]. Pathophysiology is still poorly understood, but a defect in endochondral ossification resulting in focal densities within trabecular bone has been suggested[2,4]. Diagnosis is mostly made by incidental radiographic findings of multiple small, round, well-defined juxtaarticular radiopacities ranging from a few millimeters to a few centimeters[1,2,4]. They are symmetrical and can be found in the phalanges, carpal bones, metacarpals, tarsal bones and pelvis[1,4]. Skull, ribs, collarbones and vertebrae are rarely affected[1]. Most patients are asymptomatic, but some may complain of mild joint pain and swelling[2,3,4]. In approximately 25% of cases, OPK associates with dermatological manifestations, such as dermatofibrosis lenticularis disseminate (the Buschke-Ollendorff syndrome)[2-5]. It can also associate with other conditions such as dacryocystitis, cardiac and renal malformations and endocrine diseases[2]. The main differential diagnosis includes osteoblastic metastasis, mastocytosis, tuberous sclerosis, melorheostosis and osteoma[1,2,4,5]. The symmetric distribution, lack of bone destruction, and juxtaarticular location helps to differentiate OPK from them[2,4,5]. Bone and PET-scans are unremarkable in OPK and bone biopsy should be reserved for cases of diagnostic uncertainty[2,4]. As it is a benign disease, no routine follow-up visit or studies are usually necessary. When needed, pain is managed through non-steroidal anti-inflammatories and opioid analgesics[1,2,4].

In conclusion, OPK is a rare but benign bone condition that is found incidentally in imaging studies performed for other reasons. The present case is atypical in the sense that joint pain is not a common feature. OPK should be recognized by physicians to avoid unnecessary and potentially harmful invasive procedures.

Figure 1. Hand AP radiography showing small, round, well-defined, juxtaarticular sclerotic lesions typical of OPK.

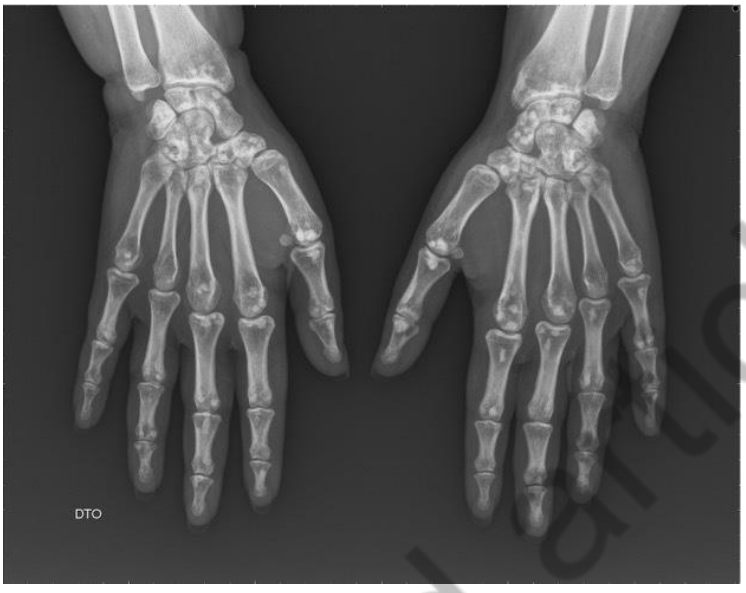


Figure 2. Left shoulder external rotation radiography.

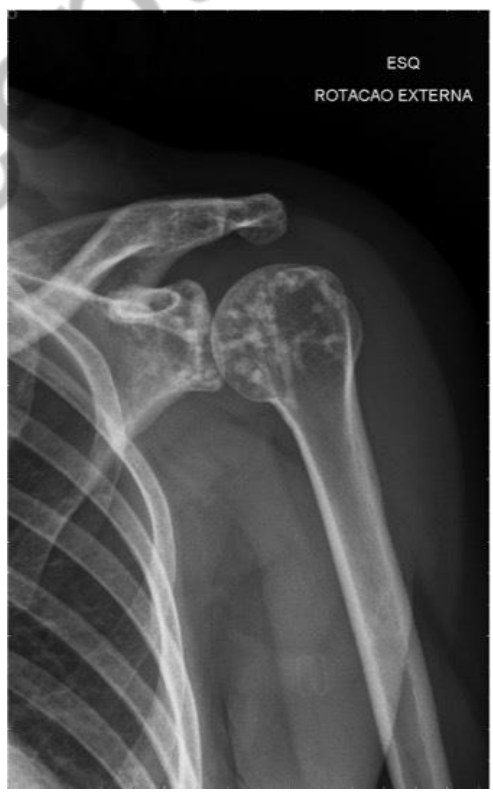


Figure 3. Bilateral knee AP radiography.

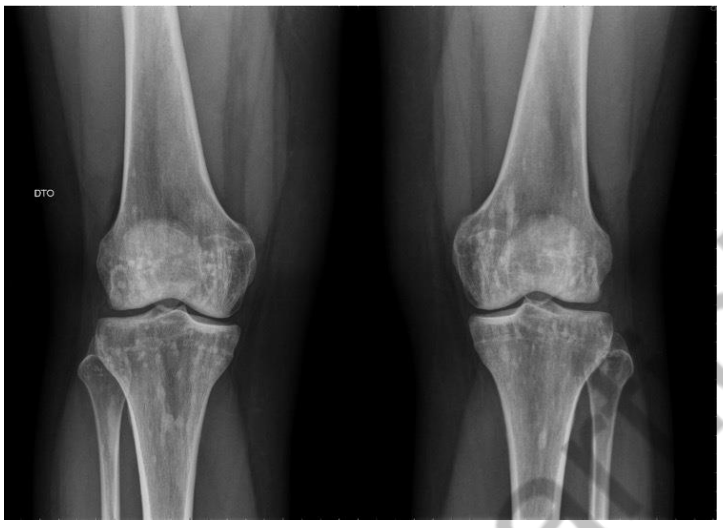
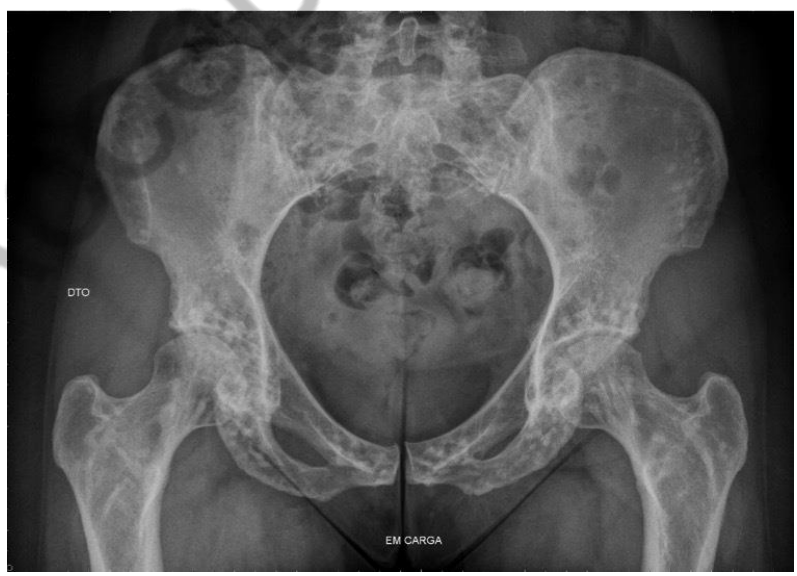


Figure 4. Pelvis AP radiography.



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