

Case Presentation

Osteopoikilosis: a rare case with interesting imaging

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Abstract

Background: Osteopoikilosis (OPK) is a rare osteosclerotic dysplasia. It is usually asymptomatic and diagnosis is made incidentally by radiographic findings. It has a unique radiographic presentation with multiple small, well-defined, circular, or ovoid radiodensities which are distributed symmetrically in the epiphysis and metaphysis of long bones.

Aim of the work: In this case report, a 38-year-old man with mild joint discomfort was diagnosed with OPK according to his radiographic findings and literature review.

Conclusion: It is important to diagnose OPK and to distinguish it from other medical conditions to calm the patient and to reduce unnecessary investigation.

More Information

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Introduction

Osteopoikilosis is a rare form of skeletal abnormality. The condition is a type of bone sclerosing dysplasia that is usually asymptomatic; diagnosis is made by incidental radiological findings in patients seeking treatment for other medical disorders such as fractures. Osteopoikilosis follows an autosomal dominant distribution pattern with some cases happening sporadically. The typical radiographic finding in osteopoikilosis is, small, well-defined, circular, or ovoid radiodensities which are distributed symmetrically in the epiphysis and metaphysis of long bones. In histology, these sclerotic lesions appear as bony islands in cancellous bone. Patients are usually diagnosed between the ages of 15 to 60 but there is no age or sex preponderance [1-3]. Abnormality in the endochondral bone maturation process and collagen regulation is the main pathogenesis of this condition which is caused by a mutation in the LEMD3 gene [4]. Association of connective tissue nevi (dermatofibrosis lenticularis disseminata) with typical sclerotic bone lesions of osteopoikilosis are seen in the same rare cases known as Buschke-Ollendorff syndrome [5]. The goal of diagnosing osteopoikilosis is to separate it from more serious conditions such as osteoblastic metastasis.

Case presentation

A 38-year-old man presented at the rheumatology clinic of Shariati Hospital with the chief complaint of mild discomfort in the left wrist since 6 years ago. In addition, he had a new onset of mild discomfort at both ankles and pelvic areas. He had a past medical history of mild chronic kidney disease with increased serum creatinine level and increased cortical echogenicity in ultra-sonography of both kidneys. In physical examination, there was no swelling or tenderness on both wrists, ankles, or hips. The range of motion of both wrists, ankles, and hips was normal. Examination of other joints and spinal column were normal. No other clue was found in his history and physical examination.

X-ray studies of feet, ankles, hands, wrists, and pelvis revealed the existence of multifocal clusters of small, round to oval, well-defined border, sclerotic lesions located along the proximal and distal epiphysis of joints, at bilateral tarsal joints in Lisfranc and Chopart, MTP (metatarsophalangeal), PIP (proximal interphalangeal), DIP (distal interphalangeal) of both feet, both hips, wrists, MCP (metacarpophalangeal), PIP and DIP joints of both hands. The morphology of these

lesions noticed on X-rays was identical to those seen in osteopoikilosis. All bones were free of any cortical erosion or periosteal reaction (Figures 1,2).

Three-phase bone scintigraphy showed a mild increase in radiotracer uptake in the left wrist but no focal radiotracer uptake in the left wrist and other sites of the body. Examination of other joints and spinal column were normal (Figure 3).

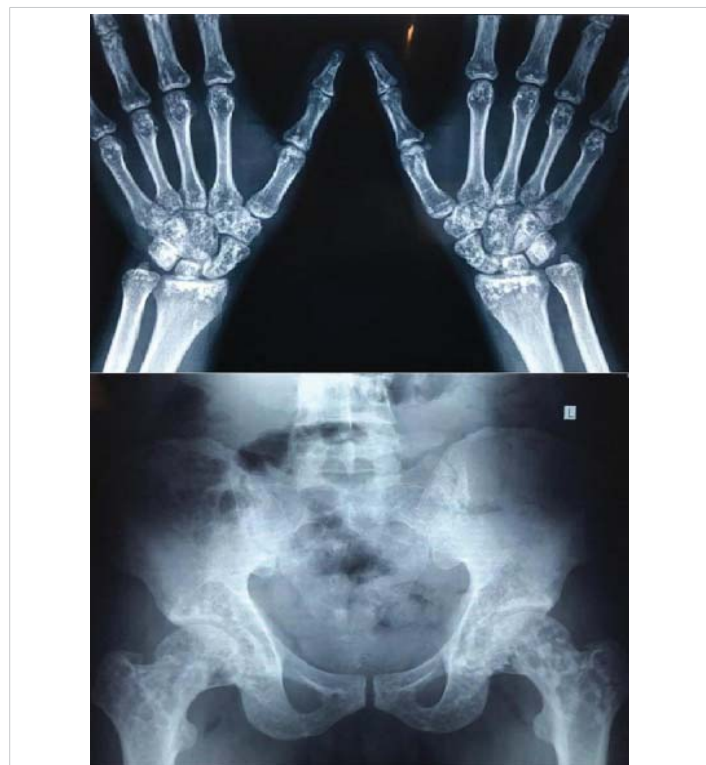


Figure 1: Anteroposterior (AP) radiograph of both hands and Anteroposterior (AP) radiography of pelvis.



Figure 2: Anteroposterior (AP) and Oblique view radiography of both feet.



Figure 3: Three-phase bone scintigraphy of both hands.

Laboratory tests such as routine cell blood counts, erythrocyte sedimentation rate [ESR], serum electrolytes, urine analysis, thyroid function tests, alkaline phosphatase, liver function tests, rheumatoid factor [RF], antinuclear antibody [ANA] and anti-dsDNA were negative.

After confirming that there is no underlying malignancy or inflammatory disease, Osteopoikilosis was the only suitable diagnosis for him. He was discharged with acetaminophen.

Discussion

Osteopoikilosis is a rare and benign sclerosing dysplasia of bone characterized by the presence of numerous bone islands in the skeleton. Incidence is approximately 1 in 50000. Patients are usually diagnosed between the ages of 15 to 60 but there is no age or sex preponderance [1-3].

Although the pathogenesis and etiology of osteopoikilosis are still unclear, studies show that the LEMD3 gene is responsible for cases with autosomal dominant distribution pattern. LEMD3 can antagonize both the Bone morphogenetic protein (BMP) and Transforming growth factor beta (TGF-β) signaling pathway resulting in hyperostotic bone disorders [4,6]. These lesions are made of dense trabeculae of spongy bone, sometimes forming a nidus that has no communication with bone marrow [7,8].

Osteopoikilosis is usually asymptomatic. Patients usually seek medical care for conditions not directly related to Osteopoikilosis. Therefore, OP is usually considered a chance finding. Joint pain and swelling are the main clinical feature in 20% of patients [9]. Association of small yellow papules and plaques, which are connective tissue nevi in osteopoikilosis patients is known as Buschke-Ollendorff syndrome [9].

Diagnosis is based on radiologic findings. A bone scan is sometimes used to distinguish osteopoikilosis from malignant metastasis. Osteopoikilosis is described by several 2 mm - 10 mm, round to oval, radio-opaque densities in cancellous bone tissue within epiphyses and metaphyses of long bones with a symmetric distribution pattern [1-3]. The most common sites are phalanges (100%), carpal bones (97.4%), metacarpals (92.3%), foot phalanges (87.2%), metatarsal (84.4%), tarsal (84.6%), pelvis (74.4%), femur (74.4%), radius (66.7%), ulna (66.7%) and tibia (20.5%) and is less common in ribs, vertebrae and skull [3]. The pattern of these sclerotic lesions may be related intimately to mechanical strain on cancellous bone trabeculae [3,10]. In the bone scan, due to active osseous remodeling, slightly increased activity is present [9]. However, an abnormal bone scan doesn't exclude osteopoikilosis if typical radiologic findings are present [11].

Differential diagnosis includes osteoblastic metastasis, mastocytosis, synovial chondromatosis, tuberous sclerosis, Paget's disease, osteopathia striata, melorheostosis and Ollier's disease [8].

Among these disorders, osteoblastic metastasis is the most critical one to distinguish from osteopoikilosis. In Osteopoikilosis, lesions are symmetrical and uniform in size and follow a periarticular distribution. In OPK patients, bone scan is normal or slightly increased activity, and alkaline and acid phosphatase level is in the normal range. In contrast, asymmetry, axial skeleton involvement, bony destruction, and positive bone scan are the main diagnostic features of osteoblastic metastasis [1,3,10].

No specific treatment or further investigations is needed due to the benign course of the disease. Patient education and reassurance is the key to patient management [12]. Non-steroidal anti-inflammatory drugs [NSAIDs], acetaminophen, and opioids can be to relieve pain. Rare active lesions have been treated with bisphosphonate therapy, but the results are controversial [13].

Complications of osteopoikilosis are rare, but cases of osteosarcoma, chondrosarcoma, and giant cell tumor have been reported although no definite association has been established [3].

Conclusion

Osteopoikilosis is a rare disease. The importance of diagnosing this disease is to distinguish it from much more important medical conditions such as osteoblastic metastasis to prevent unnecessary investigation and decrease the level of anxiety and fear in patients.

Authors' contributions

MN and PH contributed in data gathering. LA interpreted imaging. MM and MM contributed in writing the manuscript. FT was the main physician and analysed data. The final version of manuscript was approved by all authors.

Statements

Availability of data and materials: All data generated or analyzed during this study are included in this published article.

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Patient consent statement: Written informed consent was obtained from the patient for publication of this Case report and any accompanying images. A copy of the written consent is available for review by the Editor of this journal.

Declaration: The authors of this manuscript are not representatives of the government, and this paper is not

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