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Case Report

Osteopoikilosis - associated with dwarfism and presenting as painful joint effusion

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ABSTRACT

Osteopoikilosis is usually asymptomatic with only 15-20% patient having pain or joint effusion. Sclerotic dysplasia has multiple differential diagnosis including benign and malignant etiology. Characteristic features of symmetric, round to oval sclerotic lesions in multiple joints with axial skeleton sparing, no cortical erosion, normal clinical and blood investigations help differentiate osteopikilosis from critical malignant sclerotic pathology.

Keywords: Osteopoikilosis, Spotted bone disease, Sclerotic bone dysplasia

INTRODUCTION

Osteopoikilosis (spotted bone disease, osteopathia condensans disseminata) is a rare autosomal dominant osteosclerotic dysplasia first described by Albers-Schönberg in 1915.[1] The reported incidence of the condition is 1 in 50,000 with slightly higher male preponderance.[2] Osteopoikilosis is usually asymptomatic and usually diagnosed incidentally on radiographs.[3] However, osteopoikilosis can present in 15-20% patients with pain or joint effusion without any deformity or dysfunction.[4,5] It is characterised by multiple, round or ovoid sclerotic lesions in cancellous bones, particularly in epiphyses and metaphyses of long bones, scapulae, pelvis, sacrum, carpi and tarsi, while the ribs, clavicle, spine and skull are typically spared.[3,5]

Osteopoikilosis must be differentially diagnosed from other sclerotic conditions like osteoblastic metastasis, melorheostosis, mastocytosis, osteopathia striata, Ollier's disease, tuberous sclerosis, and Paget's bone disease.[5,6] Among these the most important one to differentiate is from osteoblastic metastasis which is the most common malignant bone tumours.[7] The osteopoikilosis lesions can be differentiated from metastasis by their symmetrical sclerotic lesions in epiphyses and metaphyses which are uniform in size and never inducing cortical erosion, less axial skeletal involvement, normal alkaline phosphatase levels and normal bone scan study.[3,5] Osteopoikilosis may be associated with other conditions like Buschke-Ollendorff syndrome, dermatofibrosis lenticularis, melorheostosis, dwarfism, synovial osteochondromatosis, dacryocystitis, spinal stenosis, keloid formation, scleroderma like lesions, rheumatoid arthritis and ankylosing spondylitis.[5,8-13] Most cases stay benign and asymptomatic throughout life, although there have been rare reports of osteosarcoma, chondrosarcoma and giant cell tumour developing in patients with osteopoikilosis.[14-16]

CASE REPORT

A 19-year-old female patient presented with complaints of right ankle pain and swelling. There was no history of any direct or indirect trauma. There were no complaints of fever, fatigue, loss of weight, polyarthralgia or similar complaints in the past. On local examination there was effusion in the ankle joint with normal skin and no local rise of temperature or redness. Her stature was short for her age with a height of 132 cm and demonstrating

disproportionate dwarfism. Her mother was also short statured with 130 cm height. Radiography for the ankle was planned along with blood investigations.

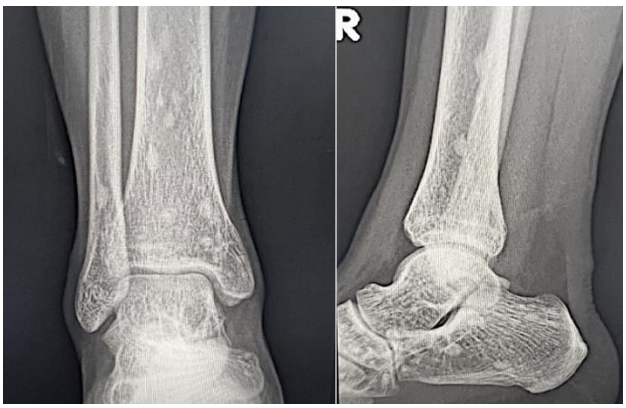

Figure 1: Right ankle radiograph.

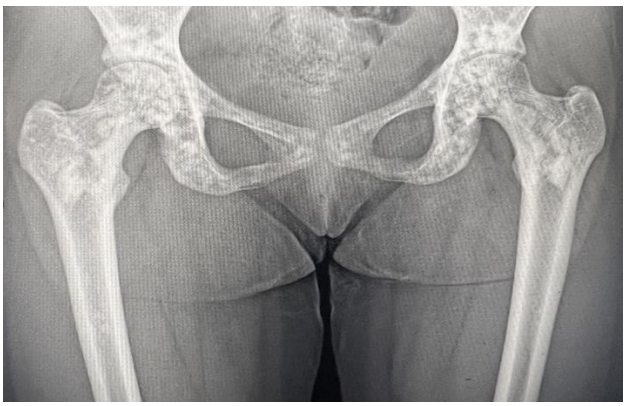

Figure 2: Pelvis with both hips radiograph.

Her initial ankle radiography demonstrated multiple round to ovoid sclerotic lesions at the right ankle, subtalar and chopart joint. There were no cortical erosions or thinning, no periosteal reactions and no joint destruction. All blood investigations were normal including alkaline phosphatase, rheumatoid factor, serum uric acid, white cell count, hemoglobin, renal function test, liver function test and inflammatory markers (ESR and C- reactive protein).

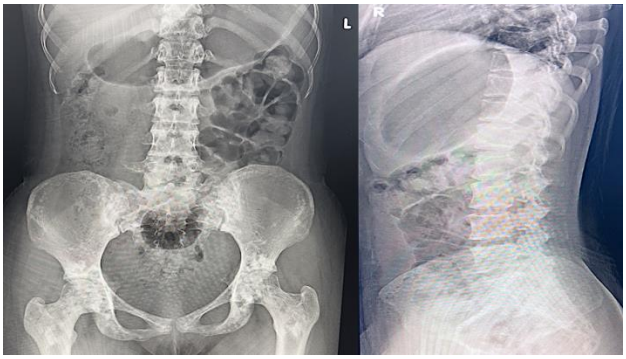

Figure 3: Lumbar spine sparing.

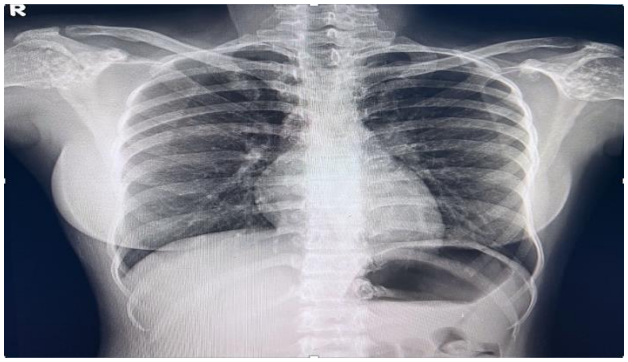

Figure 4: Bilateral shoulder joint involvement with ribs and clavicle sparing.

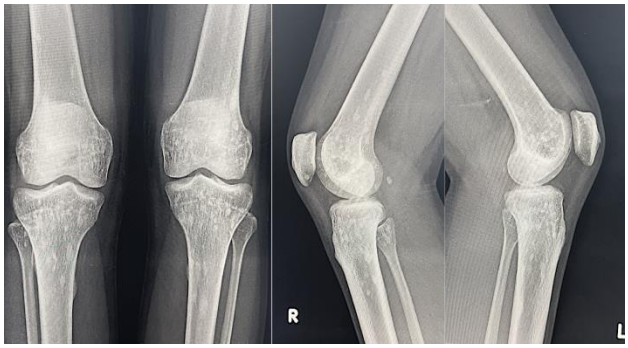

Figure 5: Bilateral knee radiograph.

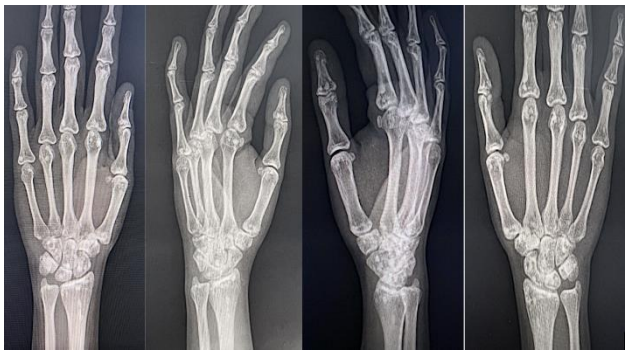

Figure 6: Involvement of bilateral wrist and small joints of hand.

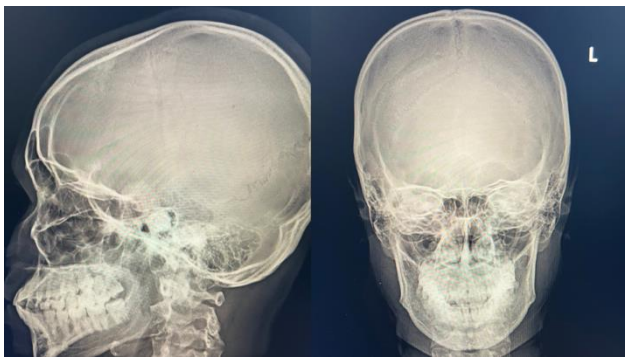

Figure 7: Sparing of the skull.

In view of the radiographs she was further examined clinically and evaluated with further radiographs to rule out more grave etiology like osteoblastic metastasis. There was no clinical evidence to suggest a malignant etiology and her radiographs revealed similar symmetrical sclerotic lesions involving all other joints while sparing the ribs, clavicle, spine and skull.

Considering the widespread, symmetrical round to ovoid sclerotic dysplasia not causing cortical or joint erosions presenting at all joints except spine, ribs, clavicle and skull a diagnosis of osteopoikilosis was reached after ruling out possibility of osteoblastic metastasis clinically, radiologically and with blood investigations. The patient was reassured and informed possibility of this dysplasia being familial with her mother also having disproportionate dwarfism. Analgesic was prescribed for the right ankle pain and advised yearly follow up to watch for any complications.

DISCUSSION

Although osteopoikilosis is mostly asymptomatic and diagnosed as an incidental finding on radiographs, 15-20% patients may present with joint pain and effusion as in the present case report and other studies.[4,5] Being an autosomal dominant inheritance, family members should be assessed especially if other associated condition is prevalent within the family like disproportionate dwarfism in this patient and the mother.[5] It is paramount to rule out osteoblastic metastasis by the symmetrical oval or round sclerotic dysplasia sparing the axial skeleton with no cortical erosion, normal alkaline phosphatase and blood investigations as in this case. If in doubt bone scan may be used to differentiate metastasis from osteopoikilosis.[3,5] Reassurance is the mainstay with symptomatic treatment for any joint effusions and regular yearly follow-up to rule out any metastatic progression although very rare.[14-16]

CONCLUSION

Osteopoikilosis may present as symptomatic joint effusion with other associated conditions like dwarfism although most commonly it is an asymptomatic incidental finding and should be differentiated from metastatic osteoblastic lesions.

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REFERENCES

1. Albers-Schonberg HA. Eine seltene, bisher nicht bekannte Strukturanomalie. Fortschr Geb Rontenstr. 1915;23:174-5.
2. Szabo AD. Osteopoikilosis in a twin. Clin Orthop Relat Res. 1971;79:156-63.
3. Lagier R, Mbakop A, Bigler A. Osteopoikilosis: a radiological and pathological study. Skeletal Radiol. 1984;11(3):161-8.
4. Fairbank T. An atlas of general affections of the skeleton. An atlas of general affections of the skeleton. 1951;411.
5. Resnick D, Niwayama G. Enostosis, hyperostosis and periostitis. Diagnosis of bone and joint disorders. p. 1988;4084-8.
6. Benli IT, Akalin S, Boysan E, Mumcu EF, Kiş M, Türkoğlu D. Epidemiological, clinical and radiological aspects of osteopoikilosis. J Bone Joint Surg Br. 1992;74(4):504-6.
7. Tander B, Cengiz K, Diren B, Canturk F. A case of osteopoikilosis mimicking metastasis on MRI study. Turk J Phy Med Rehab. 2006;52:85-7.
8. Paraskevas G, Raikos A, Stavrakas M, Spanidou S, Papaziogas B. Osteopoikilosis: a case report of a symptomatic patient. J Radiol Case Rep. 2009;3(12):38-43.
9. Roberts NM, Langtry J, Branfoot A, Gleeson J, Staughton R. Osteopoikilosis and the Buschke–Ollendorff syndrome. Br J Radiol. 1993;66(785):468-70.
10. Weisz GM. Lumbar spinal canal stenosis in osteopoikilosis. Clin Orthop Relat Res. 1982;(166):89-92.
11. Günal I, Seber S, Başaran N, Artan S, Günal K, Göktürk E. Dacryocystitis associated with osteopoikilosis. Clin Genet. 1993;44(4):211-3.
12. Ureten K. Osteopoikilosis in a patient with rheumatoid arthritis complicated with dry eyes. Rheumatol Int. 2007;27(11):1079-82.
13. Sari I, Simsek I, Guvenc I, Sanal HT, Erdem H, Pay S, Dinc A. Osteopoikilosis coexistent with ankylosing spondylitis and familial Mediterranean fever. Rheumatol Int. 2009;29(3):321-3.
14. Mindell E, Northup C, Douglass H. Osteosarcoma associated with osteopoikilosis. JBJS. 1978;60(3):406-8.
15. Grimer RJ, Davies AM, Starkie CM, Sneath RS. Chondrosarcoma in a patient with osteopoikilosis. Apropos of a case. Rev Chir Orthop Reparatrice Appar Mot. 1989;75(3):188-90.
16. Ayling R, Evans P. Giant cell tumor in a patient with osteopoikilosis. Acta Orthopaedica Scandinavica. 1988;59(1):74-6.

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