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PHOTO QUIZ

Multiple spots on bone: diagnostic challenge or spot diagnosis?

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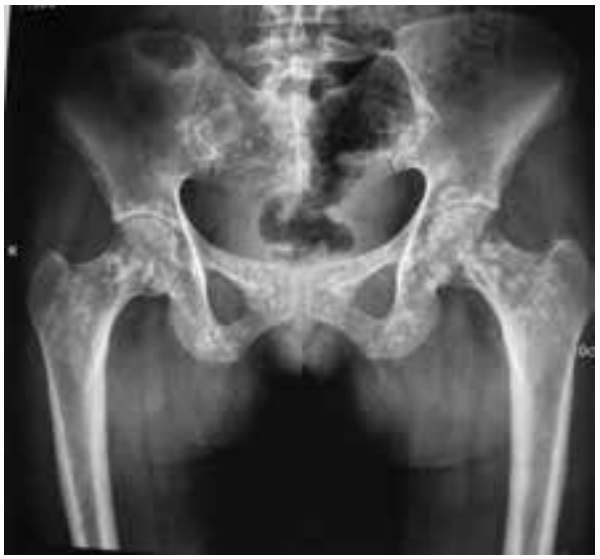
CASE REPORT

A 25-year-old female presented to us with complaints of pain over her right hip for one day following a fall from standing height. On examination there was no swelling/effusion/tenderness over the hip joint. Range of motion was full and painless. Radiographs showed no evidence of fracture or joint effusion, but multiple scattered lesions of variable size were seen over the whole pelvis and proximal femur *(figure 1)*.

WHAT IS YOUR DIAGNOSIS?

See page 376 for the answer to this photo quiz.

Figure 1. *Anteroposterior radiographs of the pelvis showing multiple sclerotic foci of variable sizes in the ileum, acetabulam, femoral head and proximal femur without narrowing of the joint spaces*



DIAGNOSIS

Osteopoikilosis was first described in 1915 by Albers-Schönberg as a sclerosing bone dysplasia of unknown cause.[1] It is also referred to as spotted bones or osteopathia condensans disseminata. A diagnosis of exclusion, cases may be under-reported. Prevalence in the general population is unknown, but an older retrospective review reported an estimated prevalence of 1 in 50,000.[2] The lesions have been described in all age groups, and although prevalence studies have shown a higher frequency among men, the apparently unequal sex distribution may be a result of referral bias in the literature (men are more likely than women to present to hospital with traumatic injuries requiring radiological investigation).[2] Osteopoikilosis exists in hereditary (autosomal dominant transmission) and sporadic forms and is characterised by defective endochondral bone formation. It is associated with a heterozygous mutation in LEMD3 that encodes an inner nuclear membrane protein; the precise function of this protein remains to be elucidated.[3]

The condition is usually asymptomatic, but in 15-20% patients there may be joint pain and joint effusions.[4] In our patient, the pain was relieved by oral analgesics (diclofenac sodium) for one week. Most reported cases of osteopoikilosis are identified during the investigation of unrelated problems in which there is no clinical history suggestive of either malignant or systemic disease. In such situations, no further workup is necessary. The characteristic radiological feature is multiple, punctate, sclerotic, round or oval foci symmetrically distributed in a predominantly periarticular fashion within the epiphyseal and metaphyseal regions. The lesions are noted in a fairly symmetric distribution, especially around the knee and shoulder, along with the pelvis, carpal and tarsal bones. The lesions are less common in the skull, ribs, vertebral bodies and mandible. Although further investigation is unnecessary in typical osteopoikilosis, when radionuclide bone scans are performed, their results are negative. The microscopic features of the lesion are identical to those encountered in bone islands. In clinical and radiological follow-up of osteopoikilosis, the lesions remain stable.

Osteopoikilosis is typically an asymptomatic incidental finding, but it can be associated with other diseases. The major differential diagnoses are osteoblastic metastases, and tuberous sclerosis. The symmetric distribution, the propensity for epiphyseal and metaphyseal involvement, and the uniform size of the foci are features that suggest osteopoikilosis, a diagnosis that is supported by a normally appearing bone scan. Skeletal metastasis and tuberous sclerosis are characterised by asymmetric distribution, common involvement of axial skeleton, including spine, osseous destruction, variation in size, and positive scintigraphic findings. In these situations bone scintigraphy plays an important role in distinguishing them from osteopoikilosis.

In conclusion, although benign, osteopoikilosis may sometimes be difficult to diagnose. The characteristic imaging features and bone scintigraphy may help in reaching the diagnosis and excluding other differential diagnosis.

ACKNOWLEDGEMENT

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