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Giant cell tumor in a patient with osteopoikilosis

Ruth M. Ayling and Paul E. L. Evans

A giant cell tumor developed in the distal femur of a 26-year-old man with preexisting osteopoikilosis. The authors are not aware of previous similar reports. Both conditions are rare, but a causal relationship between them remains unconfirmed.

Osteopoikilosis (osteopathia condensans disseminata or spotted bones) is a rare hereditary disorder characterized radiographically by multiple small dense lesions in the epiphyses and metaphyses of long bones, as well as in some flat bones, notably the pelvis. Histologically, these areas consist of condensations of cancellous bone. The disorder is usually symptomless and is detected during radiographic examination for unrelated injury. Mindell et al. (1978) reported the development of an osteosarcoma in a patient with osteopoikilosis and postulated that the condition may predispose to malignant transformation. We report a case of giant cell tumor in a patient with osteopoikilosis – to our knowledge this association has not been previously recorded.

Case report

A 26-year-old man was admitted as an emergency with pain in his left knee. The pain had been present for 10 months, but had become much more severe during the previous 24 hours. There was no history of injury. The patient, his brother, and mother were all known to have osteopoikilosis, but the past medical history was otherwise unremarkable. The left knee was warm, swollen, and tender, with movement limited by pain. Radiography of both knees 3 months after the onset of symptoms (Figure 1) was reported as showing lesions of osteopoikilosis in both femora and tibiae. Further radiographs on admission showed a pathologic fracture through a large lytic lesion in the distal end of the femur. With the benefit of hindsight, the lesion was faintly visible on the earlier films. The following investigations were normal: hemoglobin, hematocrit, white cell count, serum sodium, potassium, calcium, phosphorus, and alkaline phosphatase. Chest radiograph was also normal. Open biopsy was performed and histologic studies (Figure 2) showed a giant cell tumor. The lesion was treated by curettage and bone grafting; further histologic specimens confirmed the biopsy findings. Six months later, there was no evidence of recurrence of the tumor.

Discussion

The etiology of osteopoikilosis is not known, but, as with our patient, a hereditary tendency has been documented. The condition was formerly considered to be asymptomatic, but recent reports (Resnick and Niwayama 1971, Szabo 1971) have suggested an association with several other anomalies, including stunted growth, hydrocephalus, exostoses, palatal and dental anomalies, coarctation of the aorta, palmar and plantar keratomas, keloid formation, and urogenital defects. A papular skin eruption – dermatofibrosis lenticularis disseminata – occurs in some 10–15 per cent of cases.

Department of Orthopedics, Guy's Hospital, London, England

Correspondence: Ruth M. Ayling BSc, MB, BS, Department of Anatomy, University of Bristol Medical School, University Walk, Bristol, BS8 1TD, Great Britain

Figure 1. A 25-year-old man with pain in his left knee.
A. 3 months after the onset of pain. Both femora and both tibiae show osteopoikilosis. With the benefit of hindsight the lower end of the left femur shows an area of radiolucency.
B. 10 months after the onset of symptoms the lesion in the left femur is now clearly visible and a pathological fracture is present.

According to Szabo (1971) the histologic appearances in osteopoikilosis were first described by Schmorl. The condensations of cancellous bone consist of a peripheral area of trabeculae in which osteocytes are scant, and there are no osteoblasts or osteoclasts, together with a central core of irregular trabeculae in which both osteoblasts and osteoclasts are present. The lesions appear to be metabolically active; in the immature skeleton they become denser with growth, but later they may change their size or even disappear altogether. The precise origin of these abnormal areas remain debatable, but they appear to represent foci of deranged differentiation in cancellous bone.

The frequent localization of the lesions of osteopoikilosis to the metaphyseal and epiphyseal regions of long bones led Jancu (1971) to speculate that the disorder arose from failure of normal trabecular bone formation along lines of stress. Mindell et al. (1978) reporting a single case of osteosarcoma, took this concept one stage further by postulating that malignant transformation might occur in these lesions as a response to chronic abnormal remodeling in lines of stress.

Osteopoikilosis is a rare disorder – Jancu (1971) estimated a prevalence of 1:50,000. Giant cell tumor is also uncommon; and from this single case report, no conclusions can be drawn about a causal connection between the two conditions. There is, however, increasing evidence that osteopoikilosis is not simply an incidental radiographic finding as previously believed, but that it is associated with other skeletal and visceral abnormalities.

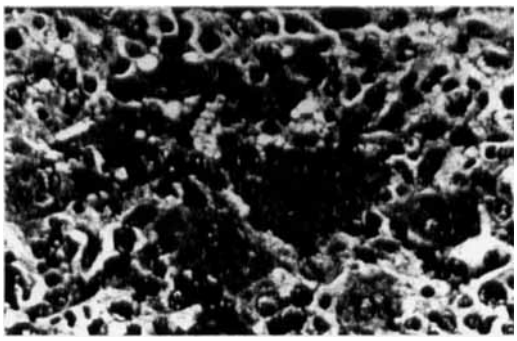

Figure 2. Photomicrograph of the giant cell tumor of bone showing both mononuclear and multinucleate neoplastic cells. HE, x 460.

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