Symmetry of bone lesions in osteopoikilosis

Report of 4 cases

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A sclerotomic symmetry in the bony lesions was clearly demonstrated in 4 patients with osteopoikilosis. Occurrence of the lesions was highest at the C6–7 and L3–4 levels.

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Osteopoikilosis is an asymptomatic osteosclerotic dysplasia with an unclarified pathogenesis (Busch 1937). The radiographic findings are highly diagnostic (Hinkel 1957), and the familial occurrence indicates an autosomal dominant pattern of genetic transmission (Busch 1937, Szabo 1971). We have performed a semiquantitative analysis of the radiographic distribution of skeletal lesions in 4 cases of osteopoikilosis.

Patients

At our hospital, we have recently seen 4 patients (all Japanese) with osteopoikilosis: viz., a 39-year-old woman (Case 1); a 13-year-old boy (Case 2); a 47-year-old female (Case 3), mother of Case 2; and a 9-year-old boy (Case 4). The familial history confirmed the accumulation of osteopoikilosis in the family of Cases 2 and 3. The patients were of normal stature, and were mentally normal. Laboratory findings, including white and red blood cell counts, transaminases, leucine dehydrogenase, antistreptolysin-O, C-reactive protein, calcitonin, parathyroid hormone, calcium, and phosphate were uniformly within normal limits. Tumor markers including carcinoembryonic antigen, alpha-fetoprotein, CA 15-3, CA 19-9, and CA 125 were not elevated. The alkaline phosphatase level was slightly higher than normal only in Case 1. A radiographic examination showed numerous small, well-defined, homogeneous, circular-to-ovoid foci of increased radiodensity clustered in periarticular osseous regions. There was no involvement of the ribs, clavicles, or skull, typical of osteopoikilosis (Sutherland 1935). A symmetric distribution of osteopoikilosis lesions appeared in the pelvis (Figure 1).

Figure 1. Case 1. Symmetric osteopoikilosis lesions.

Anteroposterior radiographs of the whole skeleton of the patients were analyzed. Osteosclerotic foci were recorded according to sclerotomes (McCredie 1976, Murray and McCredie 1979). The osteosclerotic foci in all the sclerotomes were distributed in complete symmetry (Figure 2). No obvious lesions were identified in the C1–2 region or skull. In all 4 cases, there were numerous lesions at the C6–7 and L3–4 levels. The distribution pattern of the bone lesions was similar in the 4 cases. Especially in the mother and child, the distribution was quite similar. In Case 4, the youngest patient, there were only a small number of foci. The correlation coefficients between the number of lesions on the right side and left side in each sclerotome were 0.97–1.0.

Discussion

Since the first description of osteopoikilosis in 1915, about 300 cases have been reported (Busch 1937, Green et al. 1962, Szabo 1971), and the radiographic

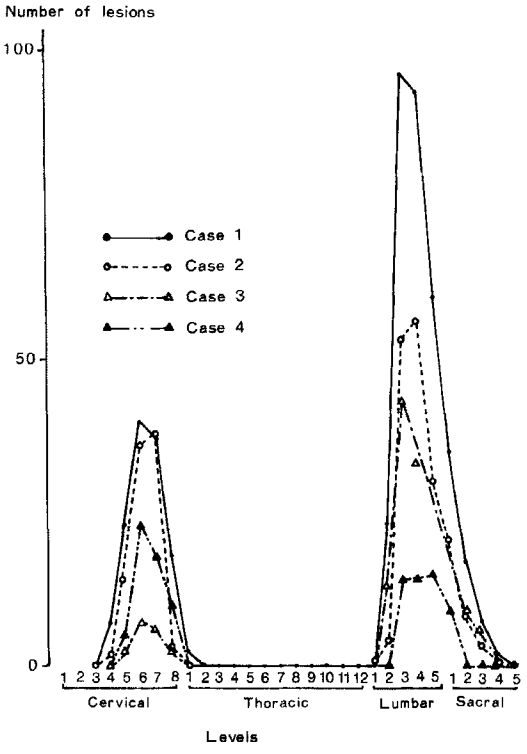

Figure 2. Sclerotomic distribution of the lesions in the 4 patients. The number of the lesions only in the right side was plotted.

diagnosis is now established (Hinkel 1957). The etiology and pathogenesis of osteopoikilosis are still obscure (Hinkel 1957). Considerable evidence suggests a relationship between this condition and other osteosclerotic disorders (Hinkel 1957), including fibrous dysplasia, osteopathia striata, and melorheostosis (Hinkel 1957, Green et al. 1962). The disorder is inherited as an autosomal dominant trait (Busch 1937, Hinson 1939). The age dependency in the appearance and disappearance of the lesions (Holly 1936) may not lend support to the inborn-error theory.

A symmetric distribution of the lesions has been observed in osteopoikilosis (Murray and McCredie 1979), although this has not been statistically analyzed. The radiopaque areas can increase or decrease in size and number, or even disappear (Holly 1936). The presence of lesions in the lower sacrum and the lower lumbar vertebrae and the absence of them in the skull and thoracic cage may suggest that bone mass is not the only essential factor determining the distribution of the lesions. We cannot explain why osteosclerotic foci appeared more frequently in the lower sclerotomes and the lower cervical levels. Embryologically, forelimb and hindlimb buds appear synchronously at the end of the fourth week of pregnancy (Hamilton and Mossman 1972). It can be hypothesized that an embryonal factor at that time expresses itself as an osteopoikilosis associated with growth.

The exact symmetry of the bony lesions in osteopoikilosis indicates that a host factor strictly controls the distribution of the lesions in the skeleton, and that this factor is related to the sclerotomes.

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