EPIDEMIOLOGICAL, CLINICAL AND RADIOLOGICAL
ASPECTS OF OSTEOPOIKILOSIS

I. T. BENLI, S. AKALIN, E. BOYSAN, E. F. MUMCU, M. KIŞ, D. TÜRKÖGLU

From the Social Security Hospital, Ankara

Osteopoikilosis is a rare condition showing characteristic sclerotic lesions on radiographic examination, which are diagnostic for the trait. We report four patients presenting with various complaints and 49 members of their families who later were found to have osteopoikilosis. The mean age of all 53 was 27.5 years and the male:female ratio was 33:20. Most had lesions in the small tubular bones. We studied the epidemiological, clinical and radiological features of these patients and from the pedigrees conclude that the disease is inherited as an autosomal dominant.

Osteopoikilosis, osteopathia condensans disseminata, is a rare hereditary condition. The diagnosis is usually made incidentally from radiographs which show multiple sclerotic areas in several bones of the body (Jaffe 1972; Spranger, Langer and Wiedemann 1974). We report 59 patients from four families in whom we studied the epidemiological, clinical and radiological features of the disease to evaluate its genetic transmission.

PATIENTS AND METHODS

Case 1. A 28-year-old man underwent radiographic examination for suspected fractures due to trauma. Disseminated sclerotic areas were discovered in several of his bones. We investigated his family and found 12 more cases (Fig. 1).

Case 2. The diagnosis in this patient was made from disseminated sclerotic lesions seen on a pelvic radiograph taken for congenital dislocation of the hip. It was confirmed by radiographic findings in the phalanges and metacarpal and carpal bones of both hands and feet and the right radius and ulna. Fifteen more members of his family were found to be affected with osteopoikilosis (Fig. 2).

Case 3. In this patient osteopoikilosis was diagnosed incidentally during radiographic examination of a hand injury. Further examination showed changes in the pelvic bones, radius, ulna and tarsal bones. Ten of his relatives were found to have the condition (Fig. 3).

Case 4. This patient, admitted for soft-tissue injuries, was found to have typical changes in the pelvis and in the proximal part of the femur. Of the 28 relatives of the patient, 14 were found to have osteopoikilosis (Fig. 4).

In the epidemiological study we recorded the age, male:female ratio, geographical distribution of patients and the number of consanguineous marriages. In the clinical and radiological studies, we noted any other locomotor disease, the bones most commonly affected and the size and number of the lesions in each bone.

RESULTS

We examined a total of 95 subjects from four families and found 53 to have osteopoikilosis. Their ages ranged from six to 68 years (mean 27.5); the largest group (11, 20.7%) was aged between 21 and 25 years followed by the 16- to 20-year age group (9, 16.9%); 77.4% of the patients were between six and 30 years old.

The families came from the Van, Zonguldak, Malatya, Erzincan areas in Eastern Anatolia and around the Black Sea where consanguineous marriages, and therefore hereditary diseases, are very common.

No other hereditary disease was found in the families except for the one patient who had congenital dislocation of the hip. The daughter of one patient (case 4) had psoriasis.

The diagnostic sclerotic lesions of osteopoikilosis
were most common in the phalanges of the hand (100%), followed by the carpal bones (97.4%), metacarpals (92.3%), foot phalanges (87.2%), metatarsals (84.4%), tarsal bones (84.6%), pelvis (74.4%), femur (74.4%), radius (66.7%), ulna (66.7%), sacrum (58.9%), humerus (28.2%), tibia (20.5%) and fibula (12.8%). They were usually bilateral (94.8%) but two patients had unilateral lesions, both on the right side. The number of sclerotic lesions in a single bone ranged from 1 to 1000 and increased with age, as did their radiodensity. The pelvic bones had the largest number of lesions. The smallest lesion measured 1 × 1 mm and the largest 1.2 × 1.6 cm. They were linear, ellipsoid or bullet-shaped and they were most common in the metaphyses of the long tubular bones or in their vicinity; there was no difference between proximal and distal metaphyses.

From the pedigrees, we have concluded that osteopoikilosis is an autosomal dominant disease.

DISCUSSION
Osteopoikilosis is a heritable condition, usually asymptomatic, in which osteosclerotic dysplasia of bones develops during childhood and persists throughout life.
Complications of the disease are very rare (Jaffe 1972; Spranger et al 1974; Whyte, Murphy and Siegel 1978; Lagier, Mbakop and Bigler 1984). Mindell, Northup and Douglass (1978) reported the development of an osteosarcoma in a patient who had osteopoikilosis and postulated that malignant change can occur. Ayling and Evans (1988) reported a pathological fracture through a giant-cell tumour in the distal end of an osteopoikilotic femur and Grimer et al (1989) described a chondrosarcoma in a patient with osteopoikilosis.

The radiographs showed multiple, small, variably shaped radiodensities especially in the metaphyses in which they appear in lines in the long axes of the long bones (Spranger et al 1974; Mindell et al 1978; Lagier et al 1984; Ayling and Evans 1988; Coillard et al 1988). These features differentiate the disease from melorheostosis. Histologically, the sclerotic areas are focal condensations of compact lamellar bone within the spongiosa (Figs 5 and 6).

Osteopoikilosis has been reported to occur in association with Buschke-Ollendorff syndrome which was first described in 1928 as "dermatofibrosis lenticularis disseminata" (Dahan et al 1989; Kobus, Lubbers and Coleman 1989). In the 95 patients in our study examined radiographically, we found concurrent pathology in only one, the patient with congenital dislocation of the hip who presented with secondary osteoarthritis.

Our series is the largest in the literature and provides some new data about this rare condition.

No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

REFERENCES


