OSTEOMESOPYCNOSIS: THE FIRST REPORT FROM JAPAN
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We describe three sisters in a family who presented with a clinical form of osteomesopycnosis which is a rare, benign osteosclerotic bone disorder limited to the axial skeleton. It must be distinguished from other osteosclerotic conditions which carry a worse prognosis. This is the first report of the condition in a Japanese family, and we believe it to be the first to be identified in Asian races.

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Osteomesopycnosis was first described by Simon et al (1979) and named by Maroteaux (1980). It is characterised by osteosclerosis localised to the axial spine, pelvis and the proximal parts of the long bones and is most often discovered incidentally in a young adult usually suffering from back pain, the most common complaint of the disease. The diagnosis is essentially based on radiological changes. Patients have a normal life expectancy and laboratory measurements are within normal values (Schmidt et al 1989). Because of its good prognosis, it is important to distinguish the condition from other types of bone sclerosis.

CASE REPORTS
We report a family of six individuals in which three sisters showed a clinical form of osteomesopycnosis.

Case 1. The eldest sister aged 14 years was seen complaining of mild low back pain. She was the product of a normal pregnancy, labour and delivery. There was no history of fractures, and she was of normal stature. There was no evidence of lesions of the cranial nerves or of abnormal neurology in her limbs. She was not anaemic.

Radiological examination revealed increased density of the axial bones. The vertebral end plates showed bands of sclerosis similar to the typical ‘rugger-jersey’ configuration (Fig. 1, D1). She had a spondylolysis at L5 and the ilium showed a ‘bone within a bone’ appearance (Fig. 2). In spite of the typical findings of osteopetrosis in these axial bones, the peripheral bones and skull did not show typical osteosclerosis or cortical thickening. There were, however, osteosclerotic areas in the upper thirds of the humerus and femur and in the distal femur, proximal tibia and proximal fibula (Fig. 3); at the last three sites there was no metaphyseal flaring or enlargement. There was no osteosclerosis around the elbow or the ankle.

Case 2. The second sister, aged 11 years, was also the product of a normal pregnancy, labour and delivery. She had a normal stature, no history of fractures, no evidence of lesions of the cranial nerve and no anaemia. Radiology showed changes similar to those found in case 1 (Fig. 1, D2).

Case 3. The youngest sister in the family was aged seven years and was born after a normal pregnancy and labour. She was larger in stature than a normal seven-year-old Japanese girl, but as in her sisters, there were no other abnormalities. Radiological examination showed increased density of axial bones (Fig. 1, D3), but there were no osteosclerotic changes in the proximal long bones or around the knee.

The parents (Fig. 1, M and F) were healthy and non-consanguineous, being aged 42 and 39 years. There were no signs of systemic poisoning, infection or metabolic disease. A five-year-old brother (Fig. 1, S), who also resulted from a normal pregnancy, labour and delivery, had no history of any fractures.

A skeletal survey of the three affected children did not show increased bone density at other sites. We were unable to obtain any more details of the family history.

DISCUSSION
We have described three cases of osteomesopycnosis. As far as we are aware, these are the first three cases to be reported in Asia.

Recognition of this disease may avoid confusion with superficially similar sclerosing bone conditions such as osteopetrosis, pyknodysostosis, renal osteodystrophy and atypical axial osteomalacia (Griffith, Fitzgerald and Cochlin 1988). Its radiological appearance is so characteristic that it is difficult to confuse it with any other bone disorder (Kozlowski and Neale 1991).
Fig. 1

Lateral radiographs of the spinal column. The three affected daughters (D1, aged 14 years; D2, aged 11 years; D3, aged seven years) show osteosclerosis at the end-plate, giving a ‘rugger-jersey’ appearance. The other members of the family (F, father, aged 42 years; M, mother, aged 39 years; S, son, aged 5 years) have no osteosclerotic changes.

Fig. 2

Figure 2 – AP radiograph of the pelvis and proximal femora of the eldest sister. The ilium shows the ‘bone within a bone’ appearance and sclerotic areas are seen in the proximal femur. The diaphysis of the femur is not affected. Figure 3 – AP radiograph of the knee of the eldest sister. Osteosclerotic changes were observed in the metaphysis of the distal femur, proximal tibia and proximal fibula. The diaphysis is normal.
The interesting points in our patients are the age of the youngest sister and the sites of the osteosclerotic changes around the knees.

It is not known when the skeletal manifestations first appear (Stoll, Collin and Dreyfus 1981). The youngest patients reported in the literature have been ten years old (Schmidt et al 1989; Kozlowski and Neale 1991) and therefore our seven-year-old patient is the youngest ever recorded. She shows sclerosed bone which is sufficient for diagnosis. Kozlowski and Neale (1991) claim that the condition does not appear to be progressive. A comparison of the radiographs of our three patients indicates that there is a tendency to progression with time.

To our knowledge our cases 1 and 2 are the first to show osteosclerosis in the metaphysis of the distal femora and proximal tibiae and fibulae. Osteomesopycnosis is characterised by osteosclerosis localised to the axial spine, the pelvis and the proximal parts of the long bones (Stoll et al 1981), but Kozlowski et al (1994) observed osteosclerotic areas in the proximal femoral metaphyses.

Most authors (Proschek et al 1985; Delcambre et al 1989; Renowden, Cole and Hall 1992) agree that osteomesopycnosis is an inherited disease with an autosomal dominant trait. Maroteaux (1980) and Kozlowski and Neale (1991), however, have reported cases without radiological changes in the parents of the propositae. Thus, osteomesopycnosis may be a mild form of hereditary osteosclerosis. Stoll et al (1981) tried to estimate the range of penetrance and expressivity, but the small numbers of patients have not allowed a precise answer. Observation of affected siblings of unaffected parents suggests autosomal recessive inheritance. This may be caused by germinal mosaicism of the parents.

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REFERENCES