SPONDYLOLYSIS IN OSTEOPETROSIS

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We report the occurrence of spondylolysis and/or spondylolisthesis of the lumbar vertebrae in five patients with osteopetrosis, four of them having multiple lesions. The case histories indicate that spondylolysis had developed in the pathological bone as a result of increased stress and that it is an acquired lesion.

Marble bone disease was first described early in the 20th century (Albers-Schönberg 1904; 1907) although it appears to have occurred even in prehistoric man (Regöly-Mérei 1962). About 200 to 300 cases of this rare condition have been reported (Johnston et al. 1968; Aegerter and Kirkpatrick 1975). The term osteopetrosis was first used by Karshner in 1926.

Osteopetrosis is characterised by increased density and widening of the bones. Essentially, the lesion is one which involves the failure of resorption of calcified fetal chondroid and of primary bone, which prevents replacement by mature lamellar bone. Many authors have reported its clinical features and described its histopathology (Krompecher 1940; Cohen 1951; Johnston et al. 1968). Shapiro et al. in 1980 confirmed that the cause of the disease is impaired function of osteoclasts. The exact molecular biology is not yet known.

There is no effective treatment, and pathological fractures are an important complication. Their treatment may be difficult because of the abnormal bone structure and an increased susceptibility to bone infection.

Although spondylolisthesis has been recognised for over 200 years (Andry 1741) and has an extensive literature we could find only two reports on the association of spondylolysis with osteopetrosis (Grepl 1955; Becker 1956). We report five patients suffering from this combination of disorders.

CASE REPORTS

Case 1. A 22-year-old woman with osteopetrosis sustained a fracture of the femur and was treated successfully by internal fixation. She was rather small, looking only about 17 years of age and rather like a Dresden doll. Radiographs of her lumbar spine showed spondylolysis of L4 and spondylolisthesis at L5/S1 (Figs 1 and 2). She had never complained of backache. She later suffered re-fractures of the femur, first above and then below the fixation device.

Case 2. A 13-year-old girl was investigated for back pain. Radiographs of her lumbar spine showed spondylolysis of both L3 and L4 vertebrae and confirmed that she had osteopetrosis (Fig. 3). She was treated in a corset and 15 months later it appeared that the fractures had healed (Fig. 4). This patient later sustained a fracture of the femur but this was treated elsewhere.

Case 3. The brother of Case 2 was seen at the age of 12, complaining of back pain. Radiographs of his lumbar spine showed spondylolysis of the fourth lumbar vertebra as well as osteopetrosis (Fig. 5). He was treated in a corset and has been reported by his family doctor to be doing well.

Case 4. A 20-year-old woman with osteopetrosis showed fractures of the pars interarticularis at both L4 and L5 vertebrae. She was treated elsewhere and we have no further details other than diagnostic radiographs.

Case 5. A 32-year-old woman had osteopetrosis diagnosed when she was admitted with a fracture of the right femur. At operation, no medullary cavity could be found; the fracture was stabilised by means of two pins, and a plaster spica was applied. The fracture healed with some angulation and 6 cm shortening.

The patient had had some low-back pain seven years earlier after her first child, but investigation at that time had shown no spondylolysis. After femoral fracture the patient refused a shoe-raise and continued to walk with a severe limp.

Eight years after the femoral fracture, she developed severe low-back pain and radiographs showed bilateral spondylolysis of the second, third, fourth and fifth lumbar vertebrae. Her symptoms settled with conservative treatment in a corset. Two years later, she sustained a fracture of the left femur. At operation for

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internal fixation, but re-fracture at another site occurred twice in one of them.

Until the 1950s spondylolysis was thought to be a congenital condition, but after extensive investigation of embryos (Rowe and Roche 1953) and on the basis of autopsy findings and clinical observations (Wiltsie 1962; Wiltsie, Widell and Jackson 1975; Wiltsie, Newman and Macnab 1976) spondylolysis is now considered to be due to overload stresses superimposed on a congenital predisposition. We agree with this and we report five cases, four with multiple lesions.

We found two reports in the German literature, but the authors regarded the spondylolysis as a congenital disorder (Grepl 1955; Becker 1956). It seems more likely that these defects are produced in the pathologically fragile osteopetrotic bone by repeated stress, which causes fatigue fracture at the typical sites. This is supported by the fact that in two of our cases treatment with a corset was successful. In our fifth case, stress on the spine caused by severe femoral shortening is considered to have been an important factor in producing spondylolysis at four lumbar levels. In this patient's family no relatives had spondylolysis although this would be expected in 19 to 25% of the family of a patient suffering from spondylolysis without osteopetrosis (Wiltsie 1962; Wynne-Davies 1979).

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