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# Fracture patterns in malignant osteopetrosis (Albers-Schönberg disease)*

**N. Dahl[1], G. Holmgren[2], S. Holmberg[3], and H. Ersmark[3]**

[1]Department of Clinical Genetics, University Hospital, Uppsala, Sweden
[2]Department of Clinical Genetics, University Hospital, Umeå, Sweden
[3]Department of Orthopaedic Surgery, Samariterhemmet Hospital, Uppsala, Sweden

**Summary.** We report the occurrence and distribution of 17 fractures in four patients with malignant, autosomal recessive osteopetrosis. The frequency of the disease in the Caucasian population is in the order of 1 per 20,000, of which the vast majority suffer from a mild autosomal dominant form. The patients have been followed up for 17–22 years and have multiple handicaps. Their case histories indicate that the lower extremity is the most common site for pathological fractures. The traumata were all caused by common accidents, usually falls. Conservative treatment was successful, with normal healing time in the four cases presented.

Osteopetrosis is the result of universal failure of the bone remodelling process. The cause of the disease is impaired function of osteoclasts, which leads to excessive accumulation of bone owing to insufficient bone resorption and reformation [11].

The clinical manifestations as diagnosed by X-ray examination are a significant increase in skeletal bone mass, resulting in total or near-total obliteration of the marrow cavity. Bone growth is abormal and results in short, broad or long, thin bones. Anaemia is often observed, as the primary site of haematopoiesis in the marrow has been reduced. Pathological fractures are an important complication, as is an increased susceptibility to bone infection, especially chronic osteomyelitis of the jaw [5]. Other abnormalities that occur secondary to the loss of osteoclast activity include: skeletal abormalities that interfere with dentition, remodelling of the optic canal and auditory meatus with subsequent optic atrophy, hearing loss and neurological disorders. There is no effective treatment, although bone marrow transplantation has been curative in some cases [8].

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*Offprint requests to:* H. Ersmark M.D., Department of Orthopaedic Surgery, Samariterhemmet Hospital, Box 609, S-751 25 Uppsala, Sweden

Two forms of osteopetrosis occur. The congenital or malignant type is severe and usually fatal; a more benign form develops during adolescence and is not lethal. Inheritance is determined by autosomal recessive genes, with the exception of the benign type which is autosomal dominant. The incidence of the disease in man is low, in the order of 1 per 20,000 in the white population of which the vast majority suffer from a mild, autosomal dominant form [6].

Treatment may be difficult because of the abnormal bone structure, and a tendency to delayed healing has been reported [10].

We report fracture patterns and subsequent healing in four patients suffering from the malignant autosomal recessive form of osteopetrosis.

## Patients and methods

All patients referred to hospitals in the northern parts of Sweden (Västernorrland County and Norrbotten County) with the diagnosis osteopetrosis during the period 1970–1990 were considered for inclusion in the study. Nine patients were identified, all with the malignant form of osteopetrosis. Five of these nine patients died before the age of 15 years and are not presented here. The four surviving patients still alive, two women and two men, had fractures that were classified as pathological. None of the four patients had been subjected to bone marrow transplantation.

## Results

All the fractures in each patients are described in Fig. 1. One of the patients, a 22-year-old women, also had spondylolysis together with osteopetrosis at lumbar level 4 and 5. This has been described earlier elsewhere [12].

## Discussion

Albers-Schönberg or "marble bone" disease is a rare hereditary condition first described early in the twentieth century [2, 3]. At least 300 cases of this rare condition have been reported [1, 10].

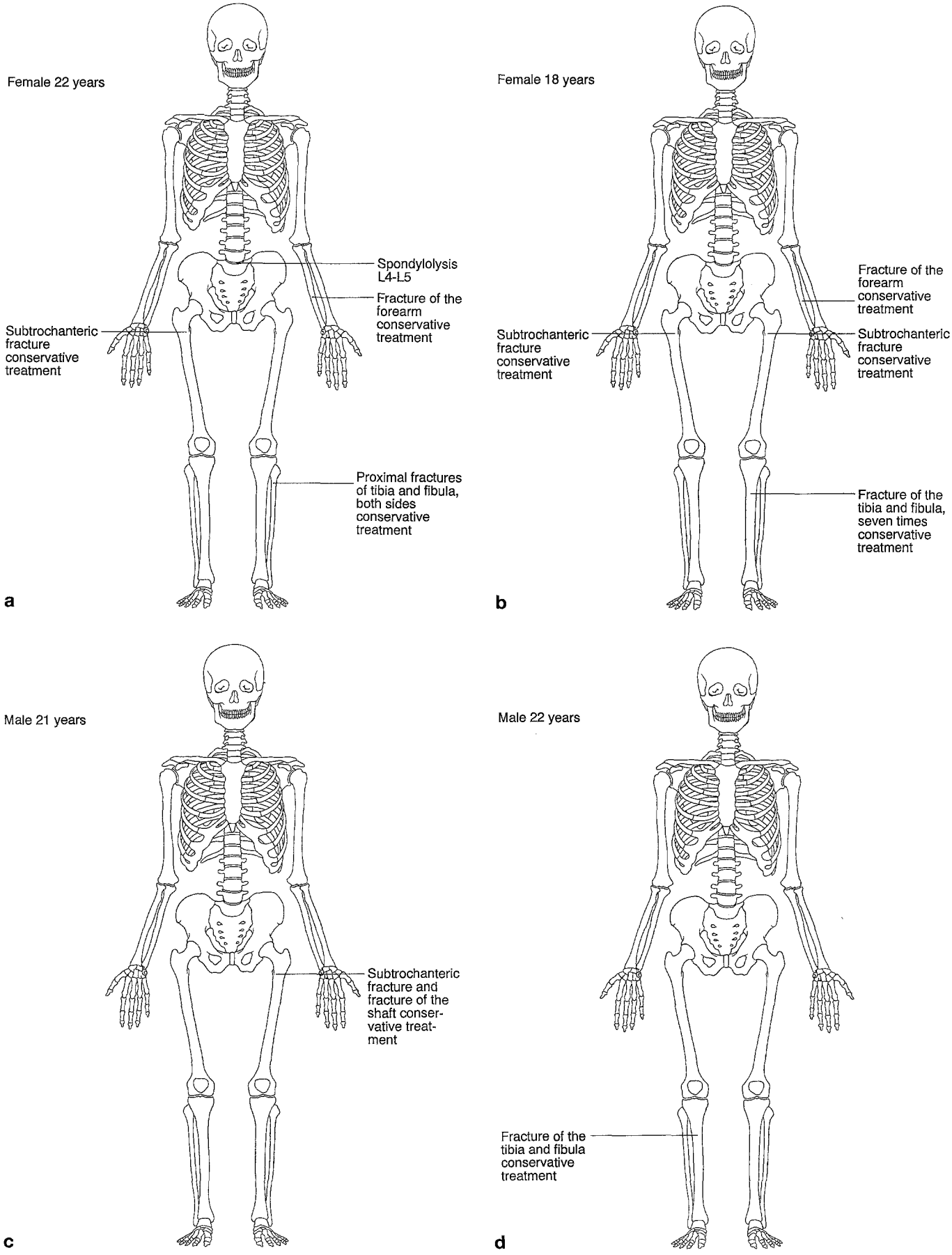


**Fig. 1a–d.** The distribution of pathological fractures in four patients with the malignant form of osteopetrosis

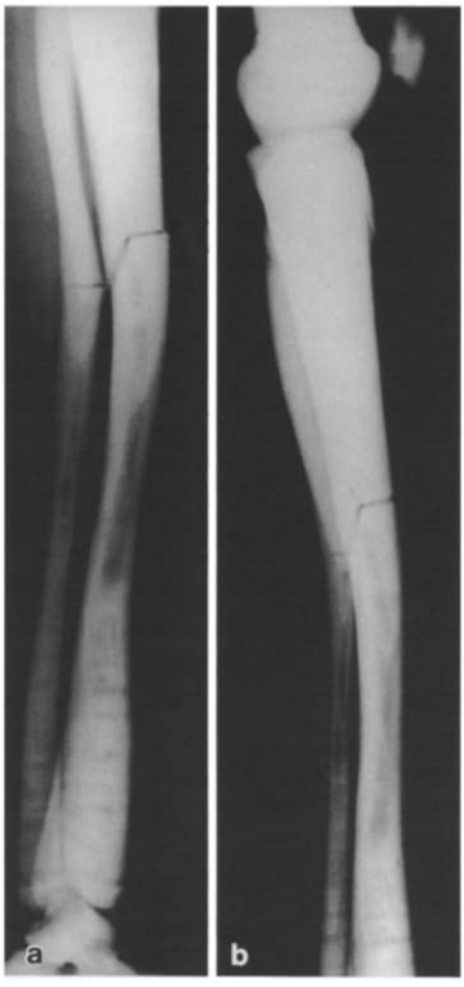


**Fig. 2a, b.** An X-ray illustration of the increased bone density in osteopetrosis, showing a fracture of the tibia and fibula in an 18-year-old girl

Spondylolysis in association with osteopetrosis as a result of increased stress in pathological bone has only been reported three times in the literature [4, 9, 12]. In this series of four patients with osteopetrosis we found one more with spondylolysis of the lumbar spine.

The four patients in this series confirm, as earlier reported [7], that osteopetrosis carries an increased risk for fractures. The number of fractures per patient ranges from one to ten. The causes of trauma were all minor; the most common were slight falls indoors, which resulted in fracture of the long bones of the lower extremities, especially fractures of the proximal femur (subtrochanteric fractures), tibia and fibula.

Pseudoarthrosis was not seen in any of the four patients. Although the fractures were classified as pathological, the fracture healing, with conservative treatment in these cases, has been classified as normal. Plaster of Paris or an orthosis therefore seem to be good choices for treatment and probably should also be recommended, as these patients are at increased risk for osteomyelitis [5]. Figure 2 illustrates the increased bone density in osteopetrosis, showing a fracture of the tibia and fibula in a case of a 18-year-old girl.

The four patients in this series had reached an unusual high age for those having symptoms of the malignant form of osteopetrosis. It is also noteworthy that no cases of the benign form were found and diagnosed during the period 1970–1990. The incidence of the disease within this region of Sweden was estimated at approximately 1 per 15000 head of population, all with the malignant form.

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