Fracture patterns in two types of autosomal-dominant osteopetrosis

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Thirty-five individuals with autosomal-dominant osteopetrosis were interviewed and radiographs were reviewed. Twenty had the radiographic Type I osteopetrosis, characterized by diffuse, symmetric osteosclerosis and pronounced sclerosis of the skull with a thickened cranial vault. Fifteen had Type II, where the most striking findings were diffuse symmetric osteosclerosis,"Rugger Jersey Spine," and endobones (bone within a bone) in the pelvis, while the cranial vault was almost unaffected. Of the 12 probands who had had a fracture, 2/20 were Type I and 10/15 were Type II. Fracture complications were also more frequent in Type II

Within the autosomal-dominant form of osteopetrosis, two distinct and strictly family-related radiographic types have recently been described (Andersen and Bollerslev 1987). The most striking finding in Type I is pronounced sclerosis of the cranial vault (Figure 1), whereas"Rugger Jersey Spine" (Figure 2) and endobones (bone within a bone) in the pelvis are characteristic findings in Type II. Both types show radiographic signs of defective bone resorption (Bollerslev and Andersen 1988). The autosomal-dominant form is often asymptomatic, and the diagnosis may be reached by chance (Johnston et al. 1968, Bollerslev 1987). The finding of increased bone density in osteopetrosis could make one believe that the strength of the bone is increased, but an increased risk of fractures and a tendency of delayed healing have been described in association with the disorder (Johnston et al. 1968, Beighton et al. 1977, Bollerslev 1987). However, the fracture frequency has never been related to the radiographic heterogeneity of the disease.

We report the fracture patterns in the two autosomal-dominant types of osteopetrosis.

Subjects and methods

The study was based on a systematic epidemiologic survey of osteopetrosis in Fyn County (Funen), Denmark, where a total of 32 individuals with the autosomal-dominant form of osteopetrosis were found (Bollerslev 1987). Since then, 3 more individuals have been disclosed. Twenty probands had the radiographic Type I osteopetrosis (10 women, 10 men) with a median age at the time of the investigation of 38 (10–76) years, and 15 had Type II osteopetrosis (7 women, 8 men) with a median age of 30 (9–78) years. The two groups were comparable as regards age and sex.

The probands were interviewed with respect to fractures and complications in healing, and all the radiographs were reviewed. The total number of fractures in each proband was recorded, and an estimate of fracture complications was defined as a history of more than three fractures and/or a history of pathologic fracture and/or delayed healing with or without pseudarthrosis.

Differences were compared using Fisher's exact test (two-tailed) and $P < 0.05$ was chosen as the level of significance.

Results

Two of the 20 patients with radiographic Type I osteopetrosis had a history of fractures, and in both cases after an adequate trauma. None had a history of fracture complications. Ten of the 15 patients with radiographic Type II osteopetrosis had a history of fractures, and 5 had a history of fracture complications.

Thus, of the 12 probands who had had a fracture (Table 1), 2/20 had osteopetrosis Type I and 10/15 Type II ($P < 0.002$).

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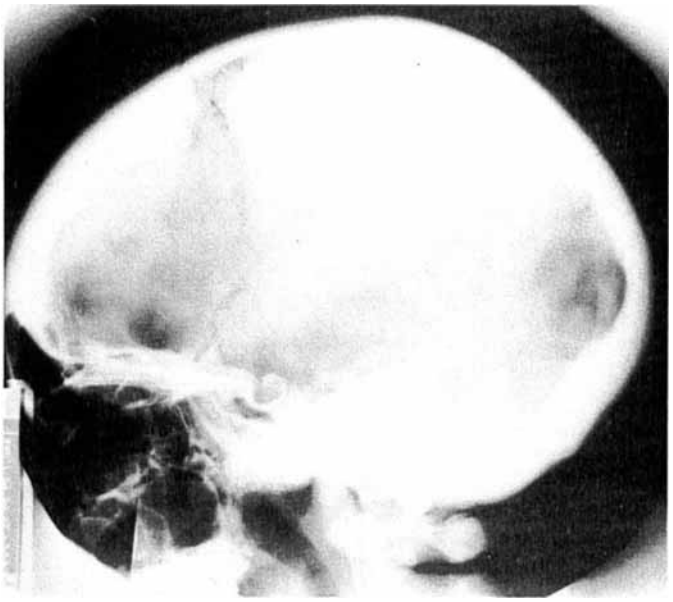

Figure 1

Figure 1. The skull in autosomal-dominant osteopetrosis Type I. Profuse sclerosis with enlarged thickness of the cranial vault.

Figure 2. The spine in autosomal-dominant osteopetrosis Type II. Note end-plate thickening ("Rugger-Jersey Spine").

Table 1. Fractures and complications of fractures in 12 patients with autosomal-dominant osteopetrosis

Case		Type	Age at investigation	Sex	Fractures	Age at fractures	*a*	*b*
1	BT	I	29	M	Tibia	14	–	–
2	HT	I	26	F	Transverse proc. lumb. spine	17	–	–
3	EC	II	45	M	Patella	32	–	–
4	FC	II	23	M	Scaphoid x 2	18/22	+	–
5	SC	II	43	M	Scaphoid x 2	27/33	+	–
					Tibia	35	–	–
					Spine		–	–
					Ribs		–	–
6	CC	II	29	M	Fibula	?	–	–
					Vertebral arcus	17	–	+
7	KJ	II	18	M	Head of radius	14	–	–
8	IN	II	44	F	Ribs	34	+	+
					Transverse proc. lumb. spine	34	–	–
					Patella	43	–	–
9	MN	II	18	M	Supracondylar	3	–	–
					Tibia	6	–	–
					Ulna	15	–	–
					Finger	16	–	–
					Metacarpal	17	–	–
					Ribs	18	–	–
					Clavicle		–	–
					Scapula		–	–
10	SN	II	36	F	Cervical spine	25	–	–
11	DR	II	78	F	Femur shaft	70	–	–
12	SD	II	9	F	Neck of femur	9	–	–

a Delayed healing and tendency to pseudarthrosis.
b Pathologic fractures.

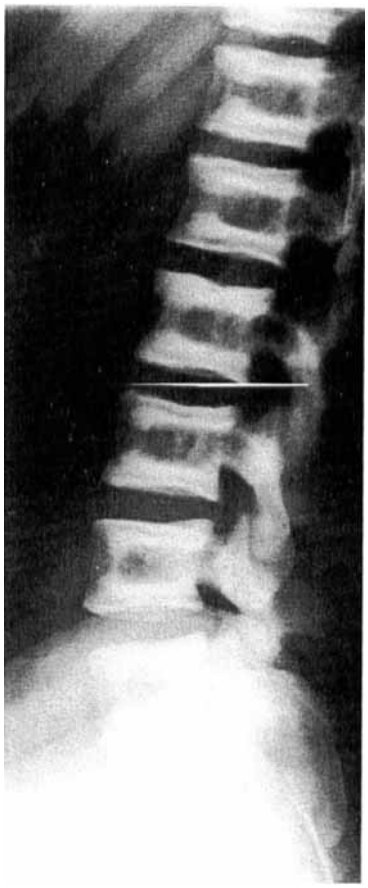

Figure 2

Discussion

Pathologic fractures and delayed healing with variable frequencies have been described in the autosomal-dominant form of osteopetrosis (Johnston et al. 1968, Bollerslev 1987). These features may reflect heterogeneity of the disorder. Type I does not seem to have an increased fracture risk, whereas the risk is highly increased in Type II, even higher than reported by Johnston et al. (1968). None of our probands had a history of osteomyelitis, which, especially in the jaw, also traditionally has been associated with autosomal-dominant osteopetrosis (Johnston et al. 1968, Beighton et al., 1977).

Radiograpy of the long bones has revealed an equally enlarged cortical thickness and a normal total subperiosteal width in the two radiographic types of autosomal-dominant osteopetrosis (Bollerslev and Andersen 1988). The distribution of peripheral osteosclerosis in the long bones did not seem to differ between the types. Differences in fracture patterns may therefore be related to the composition and structural properties of bone rather than to simple mechanical factors.

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