

Bone Cysts and Spontaneous Fractures in Two Siblings with Dyskeratosis Follicularis Darier

Torkel Menné and Aksel Otkjær Nielsen

Department of Dermatology, Rigshospital, University of Copenhagen, Copenhagen, Denmark

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Abstract. Two siblings with Darier's disease, multiple bone cysts and recurrent "spontaneous" fractures are described.

Key words: Dyskeratosis follicularis Darier; Bone cysts; Spontaneous fractures

The frequency of bone cysts in patients with dyskeratosis follicularis Darier is unknown. In 6 patients, Löken (1) found one with multiple cysts and one with a solitary cyst. A further 4 patients with concomitant Darier's disease and bone cysts (2, 4, 5) have been described previously.

Our patients are remarkable by being siblings and having had recurrent spontaneous fractures.

FAMILY HISTORY

For four generations Darier's disease has occurred in the family. None except our 2 patients have had spontaneous fractures. A sister suffering from the same disease died as a child after a traffic accident.

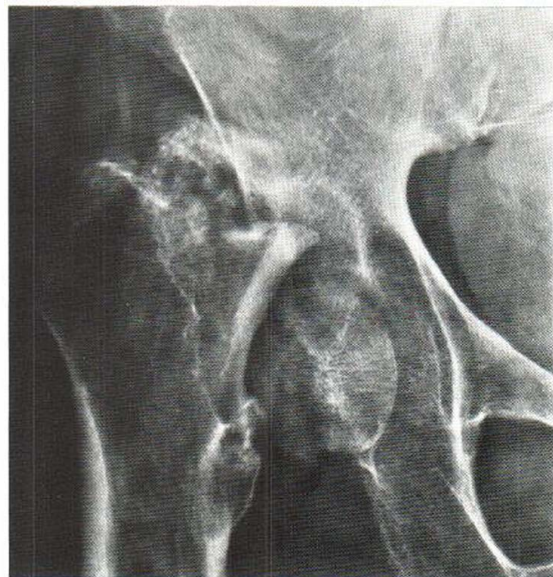


Fig. 2. Case 1. Right hip joint. Fracture of the collum femoris with displacement of the caput. In the tuberculum minus a cystic space is seen (arrow).

CASE REPORTS

Case 1

A 50-year-old woman suffered from severe Darier's disease since childhood. In the first 6 years of her life she had 6 fractures of the lower extremities and as an adult woman sustained a fracture of the right ankle after a negligible trauma. X-ray examination in another hospital revealed fractures of both femoral necks. Fig. 2 shows a fracture of the right hip joint. A fracture of the left hip joint was identical. Cystic spaces were found in both trochanteri minores. X-ray examination of the rest of the skeleton

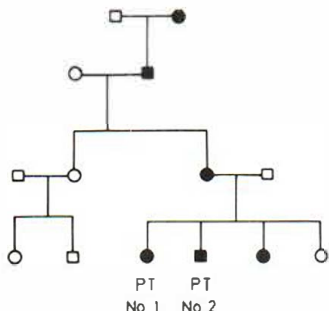


Fig. 1. Pedigree: ○=female, □=male, ●=female with Darier's disease, ■=male with Darier's disease.



Fig. 3. Case 2. Skin changes on admission to the hospital.



Fig. 4. Cyst in the caput of the left humerus (case 2).

failed to reveal any other bone cysts. The patient did not remember any trauma that could explain the severe fractures.

Laboratory investigations. Acid phosphatase, alkaline phosphatase, se-calcium, se-phosphorus, se-vitamin A and se-carotene values were normal.

Case 2

A 58-year-old man had suffered from Darier's disease since the age of 14. Since then, he developed severe, universal skin changes (Fig. 3). At 30 years of age he fractured the right collum of the femur while lying in bed. On X-ray examination cysts were found in the proximal parts of both humeri (Fig. 4), in both radii, both large trochanters (Fig. 5), as well as in metatarsal bones.

Laboratory investigations. Acid phosphatase, alkaline phosphatase, se-phosphorus, se-vitamin A and se-carotene values were normal. Se-calcium was low, 2.08 mmol/l (normal: 2.3–2.7).

DISCUSSION

In 1959, Borup Svendsen et al. (3) investigated 51 cases of Darier's disease, which, at that time, was the total number of cases known in Denmark. Both patients described in this paper were included in that study and had already sustained spontaneous fractures. No X-ray examination was done at the time. None of the other patients had experienced spontaneous bone fractures.

In the previously published papers on concomitant Darier's disease and bone cysts, pathologic



Fig. 5. Tomogram of the right hip with cystic space in the trochanter major.

fractures have not been described. We suggest that the cause of the pathologic fractures in our patients are the bone cysts. The appearance of the fractures (Fig. 2) supports this idea. The pathologic fractures are a therapeutic problem as orthopedic surgery is difficult due to the skin lesions, which constitute a source of infection.

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