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Familial Pachydermodactyly

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Pachydermodactyly is a rare form of digital fibromatosis. There are only 15 cases reported in the literature, all with a negative family history. Only one of the previously reported cases was a woman. We now report 2 female cases, mother and daughter, who, to the best of our knowledge, are the first patients with familial pachydermodactyly. The histological evolution of this disease over the years had not been previously reported. ***Key word: digital fibromatosis.***

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Pachydermodactyly is a distinct form of digital fibromatosis, characterized by asymptomatic, bulbous, soft-tissue swelling around the proximal phalanges and/or proximal interphalangeal joints of the hands. We report 2 new cases, mother and daughter, who to the best of our knowledge are the first patients with familial pachydermodactyly.

CASE REPORTS

Case 1

A 28-year-old woman consulted us because of progressive swelling of the proximal interphalangeal area of the fingers of both hands since the age of 11. There was no history of significant trauma, tenderness or movement limitation of the fingers. Physical examination revealed swelling of the proximal phalanges and proximal interphalangeal joints of the ring and little fingers of her right hand and the index and middle fingers of her left hand. The changes were along the back and sides of the involved areas, with extension up to the sides of the palms. The skin was freely movable but thickened and scaly with pachydermic appearance. A general clinical examination was otherwise unremarkable, and, in particular, her toes were normal.

Routine laboratory findings showed no abnormality, and roentgenograms of the hands revealed only swelling of the soft tissue without bony involvement. A skin biopsy specimen showed hyperkeratosis, slight acanthosis overlying a dermis with collagen deposits in irregular thick bundles. Masson trichrome staining revealed an increased number of abnormal collagen fibers arranged in a haphazard array. Orcein staining showed sparse, elongated and thinned elastic fibers, and alcian-blue-PAS staining revealed increased deposits of acid mucins. A biopsy performed 15 earlier years earlier had shown similar changes, although deposits of collagen and mucin were less marked.

Intralesional triamcinolone was slightly effective in improving the clinical appearance of the lesions.

Case 2

A 61-year-old woman, the mother of case 1, had a long history of barely perceptible swelling of the proximal phalanges of both hands. The back of the little finger of her right hand showed a firm intradermal, subcutaneous nodule similar to the swellings of her daughter (Fig. 1). Over the years, the lesions had remained stable and the patient did not agree to biopsy or treatment.

DISCUSSION

Pachydermodactyly was first described by Verbov (1) in 1975, who considered it to be a form of atypical knuckle pad. Fourteen similar cases have been reported since then (2–9), and they have been regarded by most authors as a form of digital fibromatosis. All cases presented asymptomatic thickness of tissues on the back and sides of the proximal phalanges or proximal interphalangeal joints of the hands. Because the cutaneous thickness in a patient extended to the metacarpophalangeal areas, it was defined as pachydermodactyly transgrediens (9). Apart from one case (5), all affected patients were males. Pachydermodactyly is usually seen after adolescence, although one case has been reported in a congenital form as a part of tuberous sclerosis (8). Apart from the association in another case with carpal tunnel syndrome (3), all affected patients were otherwise normal. The cause of these swellings remains unknown. There was no family history in the previously reported cases, and only in 2 patients (5, 7) was a traumatic origin shown. Histological findings included hyperkeratosis, acanthosis and a thickened dermis with collagen deposition in irregular bundles, mucin, sparse and cytologically benign fibroblasts and a decreased number of abnormal elastic fibers. Biochemical evaluation showed an increased amount of type III and a small amount of type V collagen. Ultrastructural studies revealed a marked reduction in collagen-fiber diameter in the reticular dermis. Subcutaneous resections and intralesional triamcinolone have resulted in considerable improvement in some cases. The suspension of trauma led to the regression of the lesions in the cases of traumatic origin.

We report 2 new cases of pachydermodactyly, mother and

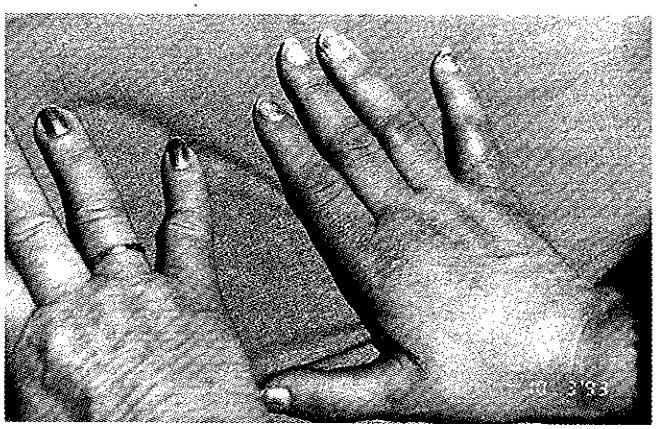

Fig. 1. The right hand of Case 1 (to the right) and the right hand of her mother (to the left). Note the swellings on the back of the ring and little fingers of the daughter, and the swelling on the back of the little finger of her mother.

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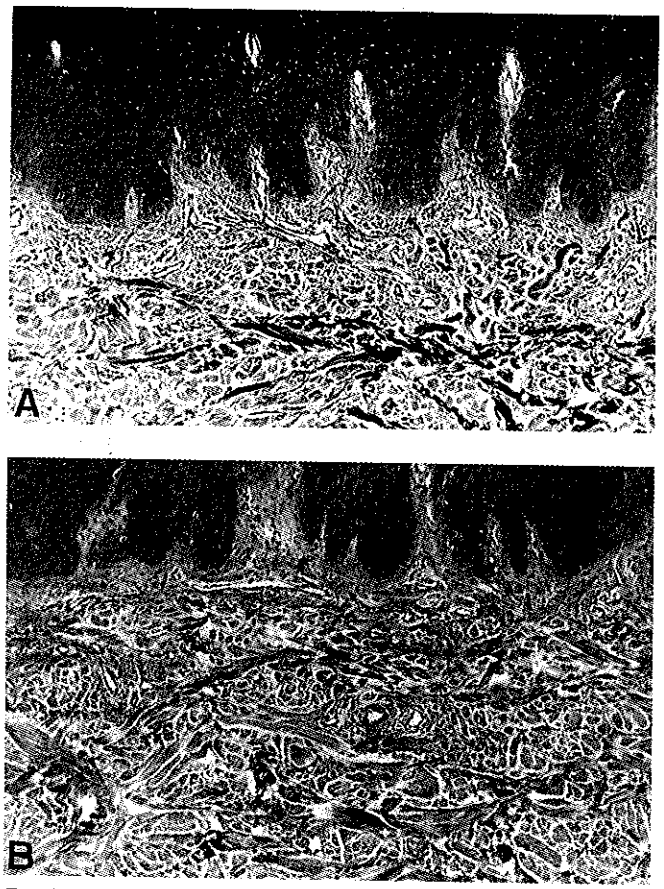

Fig. 2. Masson trichrome staining of a biopsy specimen. (**A**) The first biopsy shows a thickened dermis with irregular bundles of collagen. (**B**) A second biopsy, 15 years later, reveals more marked deposits of collagen.

daughter; to the best of our knowledge, these are the first patients with familial pachydermodactyly. Although there were no other affected members in this family, the dominant pattern of inheritance is the most probable. Besides the case reported by Draluck et al. (5), these 2 female cases have shown that the disease is not only found in males. Sola et al. have reported a case of pachydermodactyly transgrediens (9). Our case 1 had pachydermic lesions with extension up to the sides of the palms. This the second patient with this exceptional form of pachydermodactyly. Two biopsies of the lesions of our case 1 were performed with 15 years in between. The second revealed changes similar to the first, although deposits of collagen (Fig. 2) and mucin (Fig. 3) were more marked. The abnormality of elastic fibers remained unchanged. The histological evolution of this disease over the years has not been previously reported, to the best of our knowledge.

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