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KERATODERMIA PALMARIS ET PLANTARIS WITH CLUBBING AND SKELETAL DEFORMITY OF THE TERMINAL PHALANGES OF THE HANDS AND FEET

Report of Findings in Two Sisters

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Abstract. Two sisters are presented whose parents were cousins. The subjects had a triad of symptoms, keratodermia, digital clubbing, and osseous changes in the distal phalanges. The mode of inheritance and possible mechanism behind the skeletal changes are briefly discussed.

Keratodermia of the hands and feet is a hereditary abnormality not infrequently seen together with other congenital stigmata. Clubbing of the fingers is most commonly seen as an acquired symptom in pulmonary or cardiac disease but which may at times be encountered as a hereditary phenomenon with or without accompanying disease.

This paper reports the observations on two sisters who presented the unique picture of keratodermia, clubbing of fingers and toes together with unusual skeletal changes in the terminal phalanges.

CASE REPORTS

Case 1. L. F., born 1938. The disease began in childhood with a symmetrical keratosis of the palms and soles. At school she was greatly troubled by pronounced hyperhidrosis of the palms and soles and at the age of 20 bilateral cervical sympathectomy was performed. She improved after operation. In 1968 an X-ray of her skull was taken because of a slight concussion. The sella turcica seemed somewhat enlarged, and she came to the medical department for further investigation. She had a typical keratodermia (Fig. 1), and in addition, clubbing of fingers and toes (Fig. 2). X-ray investigation revealed a peculiar deformity of the terminal phalanges which had a "spatulated" appearance. The distal end seemed splayed out and showed marginal defects suggesting atrophy (Figs. 3, 4). X-ray of her ancles showed the bones to be coarsely structured with some periosteal outgrowths but no apparent cortical thickening. The investigations revealed no signs of pulmonary, cardiac or endocrine disorder.

Case 2. B. F., born 1940. Her disease began at the same age as her sister's. Because of troublesome hyperhidrosis she underwent both cervical and lumbar symphathectomy. After operation she was afflicted with orthostatic hypotension. On examination in 1969 she showed the same clinical picture as the elder sister, though the signs were less pronounced. X-ray revealed no sellar enlargement. The X-ray findings of the terminal phalanges were similar to those of her sister. An X-ray of the hands taken in 1961 showed that there had been no change within the last 8 years. This patient had light and irregular menstruations, but had no other signs of endocrine disorder. The

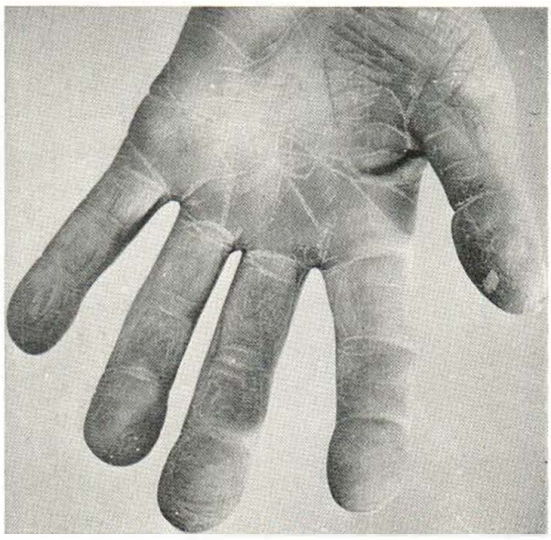

Fig. 1. Case 1. Keratodermic changes.

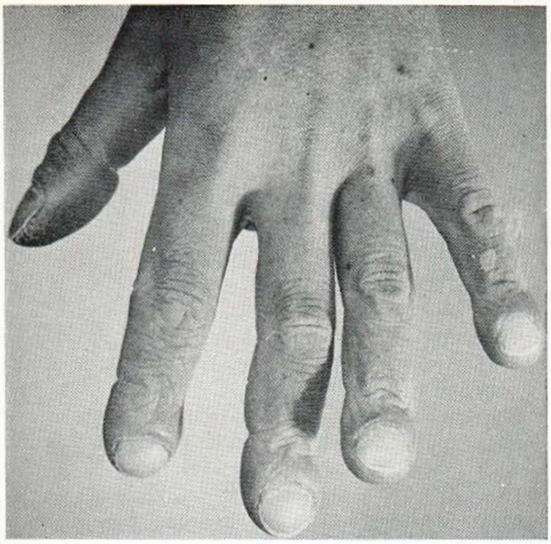

Fig. 2. Case 1. Clubbing of fingers.

parents of the sisters were cousins. Neither parent nor any known relative showed any of the changes described. There were two other siblings, a sister and a brother, who are completely normal.

DISCUSSION

Keratodermia is a hereditary disease. It is rare in Sweden but seems to be most frequent in the north where Bergström found a prevalence of 0.55 % among school children in Norrbotten County (1). The present family had no connection with Norrbotten County. Keratodermia seems to coexist occasionally with other hereditary disorders such as periodontosis (8), arachnodactyly (7) and changes of the cornea (9). Keratodermia combined with clubbing of the fingers and toes was described in a family in 1959 (2).

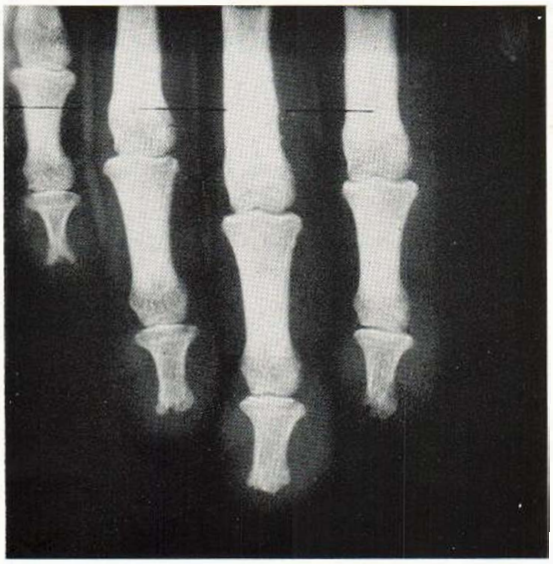

Fig. 3. Case 1. Marginal skeletal changes in end-phalanges.

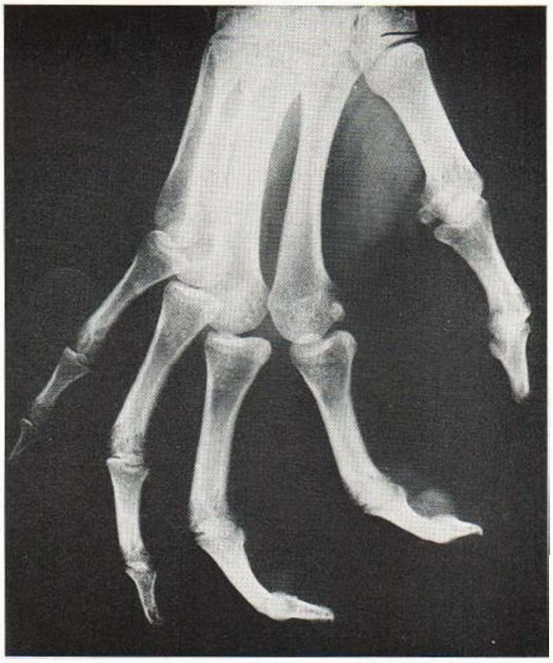

Fig. 4. Case 1. "Spatulation" of the bones in end-phalanges.

Clubbing is a well-known symptom in chronic lung disease and cyanotic congenital heart disease. Clubbing may also be present as a hereditary anomaly, and may occur as an isolated phenomenon (6). More commonly it is found together with changes in the skin and skeleton, presenting as hypertrophic osteodermatopathy (5). This syndrome is characterized by clubbing of the fingers, coarseness of the hands, feet, ankles and the forearms together with cortical thickening and periostal outgrowths of the long distal bones in the extremities. The syndrome is almost exclusively seen in men.

A few cases have been reported with clubbing of fingers and toes together with osseous changes in the distal phalanges described as spatulation and atrophy (3, 4).

Our two sisters present a triad of symptoms,

keratodermia, digital clubbing, and osseous changes in the distal phalanges. The first two symptoms are reasonably unrelated. The osseous changes might constitute a third independent hereditary trait. The possibly atrophic changes might also be secondary to the unfavourable circulatory condition in the most peripheral parts of the limbs possibly affected by the combination of keratodermia and clubbing.

Keratodermia and hereditary clubbing are both regarded as transmitted with dominant, autosomal inheritance (1). The assumption of an autosomal inheritance is also reasonable in our two cases. However, even assuming a highly variable degree of penetration, a dominant trait seems most unlikely. Considering the consanguinity of the parents, recessive inheritance seems more probable.

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