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KERATOSIS FOLLICULARIS SPINULOSA DECALVANS IN A FAMILY FROM NORTHERN FINLAND

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Abstract. A northern Finnish family from Haapajärvi is presented in which keratosis follicularis spinulosa decalvans occurred in three successive generations. Of the cases described, three were females and one male. The female carriers showed both skin and ocular manifestations. Sex-linked inheritance seemed possible. A palmoplantar hyperkeratosis of peculiar type occurred as an associated genodermatosis in the same family.

Keratosis follicularis spinulosa decalvans was described in a Bavarian family by Siemens in 1925, and he also gave the disease its name (10). As early as 1905 Rochat & Lameris had found a family in the Netherlands with the same disease. Both these pedigrees were traced backwards over several generations. Study of the former German pedigree was continued by Thelen (12) and, in the latter case, further data were published by Holthuis (5) and Jonkers (6). Isolated additional cases have been reported by Leven (8), by Franceschetti et al. (2, 3, 4) and by Adler & Nyhan (1).

The disease is characterised by follicular hyperkeratosis, especially in the region of the face and neck. Scarring of the follicles is associated with partial loosening of the hair, eyelashes and eyebrows (9, 11). The most typical eye symptoms are photophobia and inflammation of the cornea and conjunctiva (2, 4). The disease is not congenital; the symptoms usually appear during the first few weeks or months of life. The clinical picture is more distinct in males than in females, who frequently have only mild abortive forms. The women who transmit the disease may also have healthy skin.

CASE REPORTS

In Fig. 4 is seen the pedigree of a northern Finnish family from Haapajärvi with keratosis follicularis spinulosa decalvans occurring in a total of five members during three successive generations. Three female subjects (cases 2, 3 and 5) and one male (case 1) were available for study. According to the family history, the grandfather was also affected though his skin changes were somewhat milder than those of the grandson (case 1). A male family member (case 4) with palmar and plantar hyperkeratosis was studied, in addition.

Case 1

The patient was a boy aged 13, whose eyes had been found sensitive to light since the age of 1 year. When he was 4 years, hard keratin plugs began to form on his face, and the skin of the neck and extremities became coarse. Alopecia of the eyebrows gradually increased. The palmar and plantar skin began to thicken at 10 years of age. The skin changes were most marked in early spring.

Dermatological status. The follicular openings on the facial skin are widened and hard keratin plugs are seen projecting. The change is most distinct on the areas of the nose and cheeks, where yellowish keratin is observed in great amounts in the follicular openings and surrounding skin (Fig. 2). Spinulosis is of slighter degree on the neck. Follicular hyperkeratosis also occurs on the extensor surfaces of the extremities.

The palmar and plantar skin is slightly hyperkeratotic throughout. Hyperkeratosis appears in addition as narrow cords on the volar surfaces of the fingers. The eyebrows are almost totally absent and show follicular plugging. The light-coloured, short and sparse eyelashes protrude in all directions. At the top of the head there is a hairless area measuring 5 by 3 cm and covered by overlapping scaly plaques. Facial lanugo is absent in the most severely affected areas.

The lips are dry and cracked, the furrows round the mouth accentuated. The oral mucosa is of lighter colour and thicker than normal and the papillae of the tongue are large.

In the conjunctivae there are small light-coloured protuberances on both sides of the corneas. Epithelial defects also occur on the corneas. Vision in right eye is normal, the left eye shows slight myopia and astigmatism.

Laboratory studies. Hb 12.3 g%, white blood cell count 4 900, sitol test negative, cholesterol 204 mg%, phospholipids 225 mg%, triglycerides 58 mg%, vitamin A

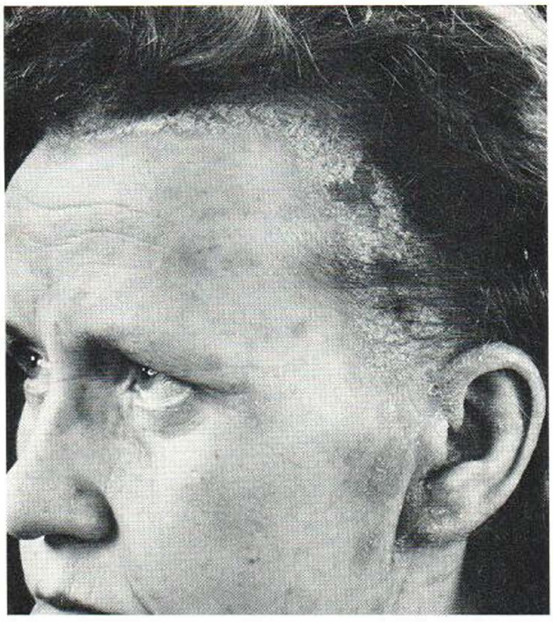

Fig. 1. Keratosis follicularis spinulosa decalvans. Case 2. Eczema seborrhoicum and follicular plugging.

tolerance normal, blood sugar tolerance normal, D-xylose tolerance normal, serum proteins 6.9 g%, electrophoretic pattern normal, immunoglobulins normal, serum concemration of electrolytes and liver function tests normal, urinary sediment normal, urine amino acid analysis normal, red cell G-6-PD normal.

Chromosomal analysis: normal male karyotype.

Histology. (1) The biopsy specimen from the cheek shows comedo-like follicular widening and plugging. (2) Skin specimen from the neck reveals a strongly grooved and hyperkeratotic epidermis. There is follicular widening and plugging (Fig. 3). (3) In a section from apparently healthy looking skin of the ankle the epidermis is hyperkeratotic and grooved. The pits are filled with keratin.

Case 2

A woman aged 37 years, mother of the boy described above. The skin of her face had been oily since puberty with follicular widening. Keratin plugs appeared occasionally. Even as a child she suffered from photophobia. Thickening of palmar and plantar skin also appeared early in life. Since the age of 35 her scalp, external ears and neck had been affected with scaling dermatitis, which sometimes spread to the trunk in the form of patches.

Dermatological status. Follicular openings on the nose and cheeks are large and filled with keratin. The forehead, near the hairline, the external ears and the surrounding skin are involved by reddish and scaling seborrhoic eczema (Fig. 1). Patchy eczema is also seen in some places on the trunk. The axillae and inguinal region are covered with larger areas of eczema.

The scalp shows scaling in large plaques. The hair is fine, dry and sparse. The eyebrows are sparse in the lateral portions. Eyelashes are short and sparse and stand out in different directions. There is partial absence of axillary and pubic hair.

Thick yellowish hyperkeratosis is present in a triangular area in the distal part of the palms, the border running from the thenar to the base of the little finger. Hyperkeratotic cords extend to the volar surfaces of the fingers. There are hyperkeratotic areas in the calcaneal and plantar regions of the soles and on the volar surfaces of the toes.

The conjunctivae show light-coloured protuberances and the corneas small spots of erosion. The patient has myopia and astigmatism.

Histology. (1) A skin specimen from the healthy-looking extensor surface of the upper arm shows a hyperkeratotic epidermis with small keratin pits. (2) A specimen from the area of palmar hyperkeratosis shows an epidermis covered by extremely thick, but mature and normal-looking keratin.

Case 3

A woman aged 31 who has a healthy 9-year-old son. Palmar and plantar hyperkeratosis appeared when she was 6 years, photophobia at the age of 10. At puberty, marked follicular widening developed on the face, occasionally accompanied by hard follicular plugging. Scaling eczema first appeared on the patient's scalp when she was

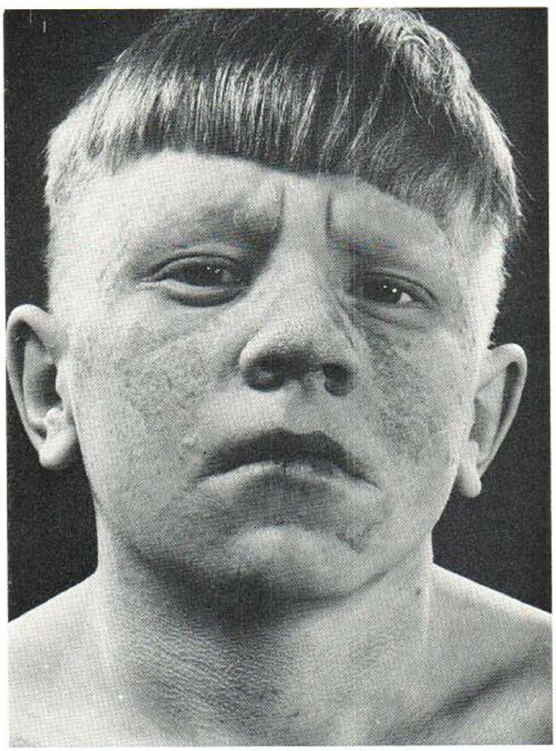

Fig. 2. Keratosis follicularis spinulosa decalvans. Case 1. Intense follicular hyperkeratosis on the facial skin.

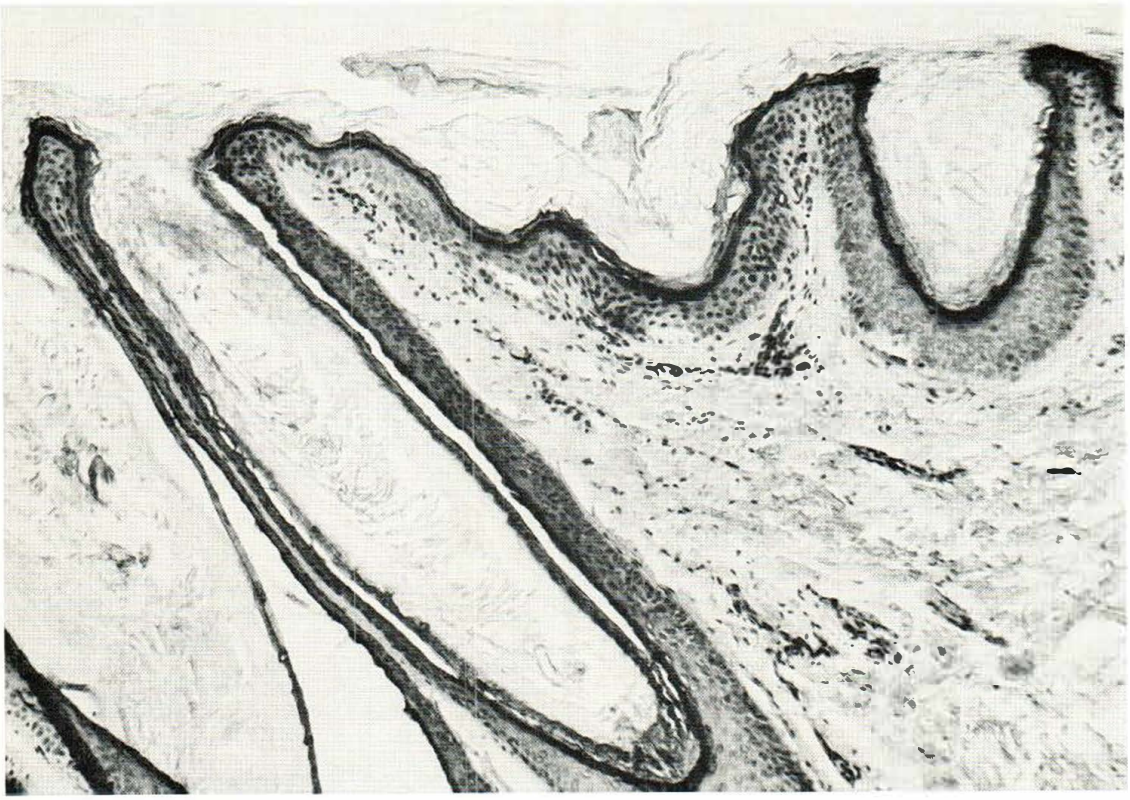

Fig. 3. Keratosis follicularis spinulosa decalvans. Deep follicular plugging in the skin of the neck. Case 1. (H-E, 1 : 100).

20. The face and trunk were sometimes involved by dry patches of this eczema.

Dermatological status. There are keratin-filled follicular openings on the bridge of the nose and the cheeks. The scalp, external ears and their surroundings are affected in places by seborrhoeic eczema, which is dry and scaling. Patches of eczema are also seen on the trunk, especially under the breasts, in the axillae and inguinal areas. The external surfaces of the extremities show follicular hyperkeratosis, which also occurs as a mild spinulosis on the neck and the upper parts of the chest and back.

Palmar hyperkeratosis is seen in a triangular area in the distal part of the palm; it extends as cords along the fingers, as in the preceding case. On the soles, hyperkeratosis is present on the heels, and under the toes.

The eyebrows are short (2–3 mm), sparse and protruding in all directions. The hair is dry and brittle. There is almost complete absence of axillary hair, and pubic hair is of less amount than usual. Lanugo hair is sparse elsewhere too.

The conjunctivae of both eyes show light-coloured protuberances lateral to the corneas. In the right eye they occur also to the left of the cornea. In addition, the corneas show small areas of erosion. The patient has myopia and astigmatism.

Case 4

A man aged 48, who has five healthy children. Thickening of the palmar and plantar skin started in childhood.

Dermatological status. As in the cases already described, there is a localised hyperkeratotic area in the distal part of the palms, from which cords extend to the volar surfaces of the fingers. Otherwise the skin is of entirely normal appearance.

Histology. A biopsy specimen from the apparently healthy looking skin of the external surface of the upper arm reveals normal skin tissue.

Case 5

A girl aged 11 years on whose face follicular hyperkeratosis had developed in the course of a few months, involving mainly the forehead, cheeks and bridge of the nose. The follicular openings were filled with dark keratinous substance and keratin spinules projected from the skin. The rest of the skin, including palms and soles, was normal.

Histology. A specimen from the skin of the cheek shows a grooved and pitted epidermis.

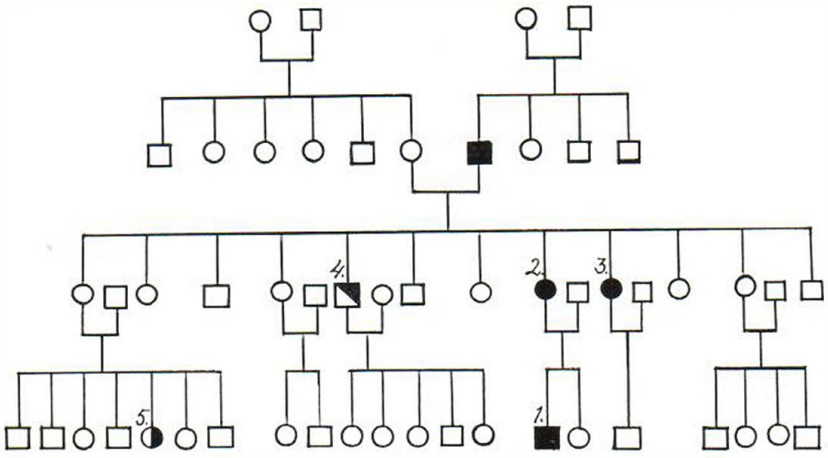

Fig. 4. Keratosis follicularis spinulosa decalvans in three successive generations in a northern Finnish family from Haapajärvi. ■ (male, ● (female), keratosis follicularis spinulosa decalvans and palmo-plantar hyperkeratosis; ◑ (female), keratosis follicularis spinulosa decalvans; ◥ (male), palmo-plantar hyperkeratosis.

DISCUSSION

Siemens (1925) defined keratosis follicularis spinulosa decalvans as a hereditary disease with sex-linked dominant type of inheritance. This implies that the most severe manifestations are found in males. Female carriers present milder forms (forme fruste). The probability of this unusual mode of inheritance has been supported by further studies of two pedigrees, extensively dealt with in the literature (5, 6, 9, 11, 12). These studies showed that the disease was always transmitted by women. The sons of affected men were healthy. Schnyder remarked, however, that the case material is too limited for the possibility of autosomal dominant inheritance to be definitely excluded (9).

In the northern Finnish family from Haapajärvi here described, the disease was inherited from father to daughter and from mother to son. This is consistent with the sex-linked type of inheritance. In the female carriers (cases 2 and 3) the disease was diagnostic, clinically as well as histologically. Each of them had both skin and eye symptoms. The symptoms, however, were slighter than those of the male patient (case 1). The carrier females in this disease proved to have fairly severe symptoms compared, for instance, with sex-linked ichthyosis, in which these women are apparently healthy-skinned or show very slight symptoms (7).

Four of the family members concerned had palmo-plantar hyperkeratosis. The palmar hyperkeratosis presented a peculiar pattern, being limited to a triangular area in the distal part of the palms and extending as cords to the volar surfaces of the fingers. Three of these patients had an associated keratosis follicularis spinulosa decalvans. In the case of one male subject (case 4), palmo-plantar hyperkeratosis was the only skin manifestation. Thus it appears obvious that palmo-plantar hyperkeratosis occurs in this family in association with another genodermatosis, viz. keratosis follicularis spinulosa decalvans.

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