

Indication for the Identity of Palmoplantar Keratoderma Type Unna-Thost with Type Vörner

Thost's Family Revisited 110 Years Later

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Palmoplantar keratoderma (PPK) type Unna-Thost is known to be the most common form of a hereditary disorder of keratinization of palms and soles. The disease is clinically identical with PPK type Vörner which is histologically characterized by epidermolytic hyperkeratosis. By reinvestigation of the family originally seen by Thost in 1880, the features of epidermolytic hyperkeratosis were found histologically confirming the diagnosis PPK of Vörner. This proves the identity of PPK type Thost with PPK of Vörner. Because of the histological variability of epidermolytic hyperkeratosis, detailed light and electron microscopic studies are necessary in cases of diffuse types of PPK. Key words: Epidermolytic hyperkeratosis; Genodermatoses; Genealogy.

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In his thesis of 1880, Thost investigated a family with a diffuse, non-transgredient palmoplantar keratoderma (PPK) (1). In this family, 17 persons in four generations were affected. The mode of inheritance of the disorder was autosomal dominant. In 1883 Unna published a clinically identical PPK in a study about the nosology of keratoses (2). He observed two families also with an autosomal dominant inheritance of the PPK. Histologically the so-called PPK Unna-Thost is said to show non-specific changes with orthohyperkeratosis, acanthosis and thickening of the granular layer (3). It seems curious that we – as well as other authors (4) – are unable to find any comprehensive case reports in the literature that confirm this histological finding. Nevertheless, the disease known as PPK type Unna-Thost is believed to be the most common form of a hereditary disorder of keratinization of the palms and soles (5).

A further family with diffuse PPK clinically identical to the PPK type Unna-Thost was described by Vörner in 1901 (6). However, this disease histologically presents the characteristic feature of epidermolytic hyperkeratosis consisting of perinuclear vacuolization of the keratinocytes and large irregularly shaped keratohyalin granules. Considering the different types of diffuse PPK, the keratosis of Vörner is the only one with characteristic histology, which allows a definitive diagnosis. The PPK of Vörner is said to be a rare disease. Until 1970, except for Vörner's original publication, only four observations had been reported, but from 1970 to 1988, 25 further reports were published (7). As the result of our own histological investigations and pedigree data, evidence of the identity of PPK type Thost with type Vörner is given.

REPORT OF A CASE

A 44-year-old woman with a lifelong history of keratosis on palms and soles was seen. Fifteen years earlier the diagnosis of PPK Unna-Thost was suspected on clinical grounds. On re-examination, the patient showed diffuse yellow keratoses over the entire surface of palms and soles. The keratoses were sharply bordered with erythematous margins (Figs. 1a–b). There was no palmoplantar hyperhidrosis.

A plantar biopsy specimen disclosed massive orthohyperkeratosis, a thickened granular layer with large irregular keratohyalin granules and perinuclear vacuolization of the keratinocytes irregularly distributed in the upper part of the epidermis (Figs. 2, 3). These are the typical histological features of epidermolytic hyperkeratosis confirming the diagnosis of PPK of Vörner in this patient. For mycologic investigations, direct microscopic analyses and cultures of material from palms and soles of persons VI-3, VII-5, and VII-6 were performed, but all with negative result. Also in PAS-stained histological specimens from the propoita (VI-3), fungal elements could not be identified. The family history revealed a pedigree with seven generations dating back to 1778 (Fig. 4). Fifty-two of all registered 189 family members were affected. Because all names and birth dates of the family members were known, it could be confirmed that 5 persons of the third generation in this family (III, 1; III, 2; III, 4; III, 6; III, 8) are the same individuals originally investigated by Thost and mentioned by name and age in his thesis in 1880.

DISCUSSION

The re-examination of the family originally published by Thost demonstrated histologically the characteristic features of epidermolytic hyperkeratosis. This proves the identity of PPK type Thost with type Vörner. In his study, Unna (2) described two families with a clinically identical form of PPK corresponding to the publications of Thost and Vörner. Unna did not mention histological examinations. Neither identity nor disparity of the disorder seen by Unna with the one described by Thost can be proved in the absence of histological studies. Therefore it is impossible to show that the families seen by Unna represent a disease different from Thost's PPK. When examining patients with clinically suspected PPK of Unna-Thost histologically, we – as well as other authors – have invariably found the features of epidermolytic hyperkeratosis of Vörner's PPK. Therefore we doubt not only the high frequency of the PPK of Unna-Thost but generally the existence of two separate entities of autosomal dominant inherited diffuse PPK. Because of the similarity of the clinical features in the three reports of Thost, Unna, and Vörner (1, 2, 6) it seems most likely that all families suffer from the same disease. It is remarkable that in 1933 Cockayne in his book summarized these three descriptions under one term which he called "tylosis palmaris et plantaris" (8). Thorough light- and electron-microscopic studies of Swedish patients with diffuse PPK were unable to demonstrate signs of epidermolytic hyperkeratosis

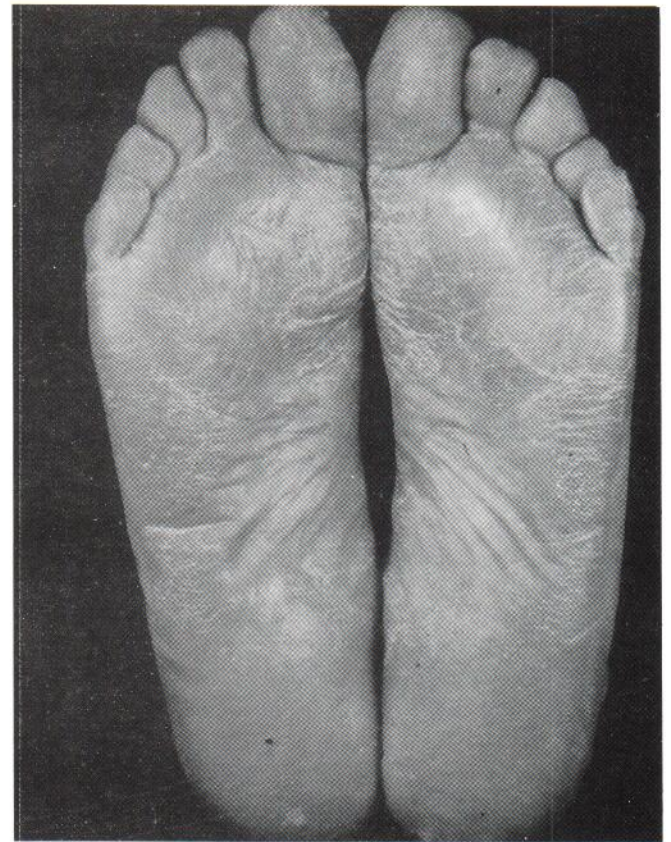
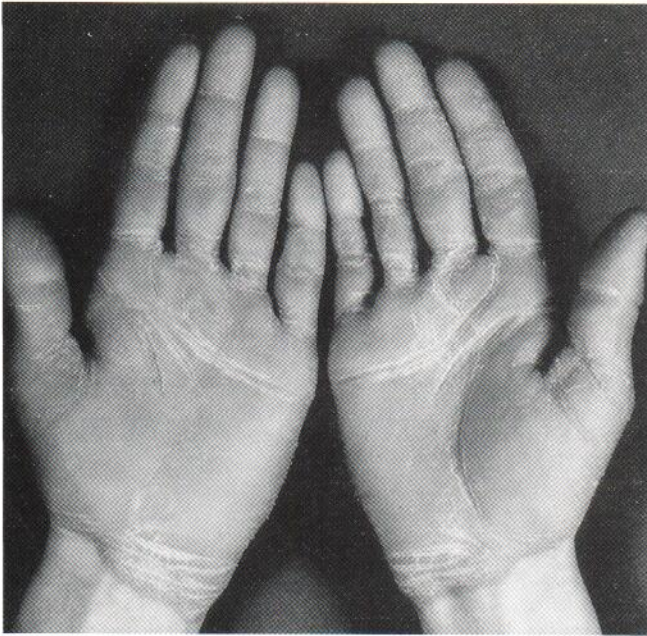


Fig. 1a-b. Diffuse symmetrical hyperkeratosis of palms and soles, with erythematous margins.

(9). But these PPK were reported with a frequency of 1 : 180 in northern Sweden (10) and may be a unique entity of a diffuse PPK.

In his histological examination, Thost (1) did not describe the characteristic signs of cellular degeneration, nor did he

mention anything that could be interpreted as epidermolytic hyperkeratosis which we found in a member of the same family 110 years later. This might be due to the wide variability of epidermolytic hyperkeratosis. Fig. 2 demonstrates the patch-like, irregular distribution of the signs of cellular degen-

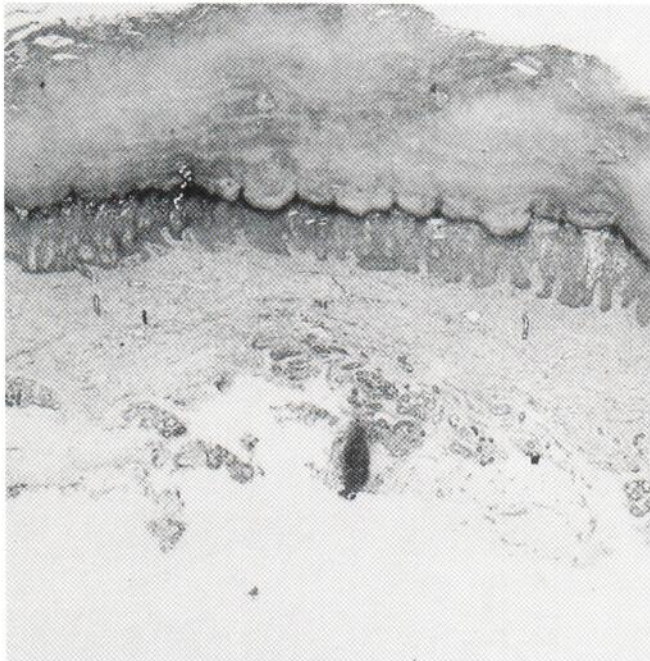


Fig. 2. Histopathology. Marked thickening of the stratum corneum, with hypergranulosis. Irregularly distributed patches of vacuolization in stratum spinosum (hematoxylin-eosin, $\times 25$).

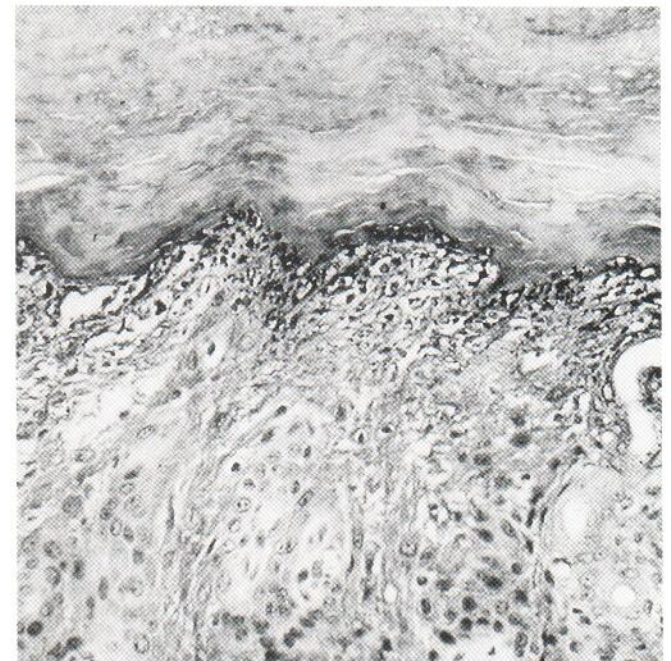


Fig. 3. Histopathology. Thickened granular layer, large irregular deposits of keratohyalin granules, perinuclear vacuolization of the keratinocytes (hematoxylin-eosin, $\times 500$).

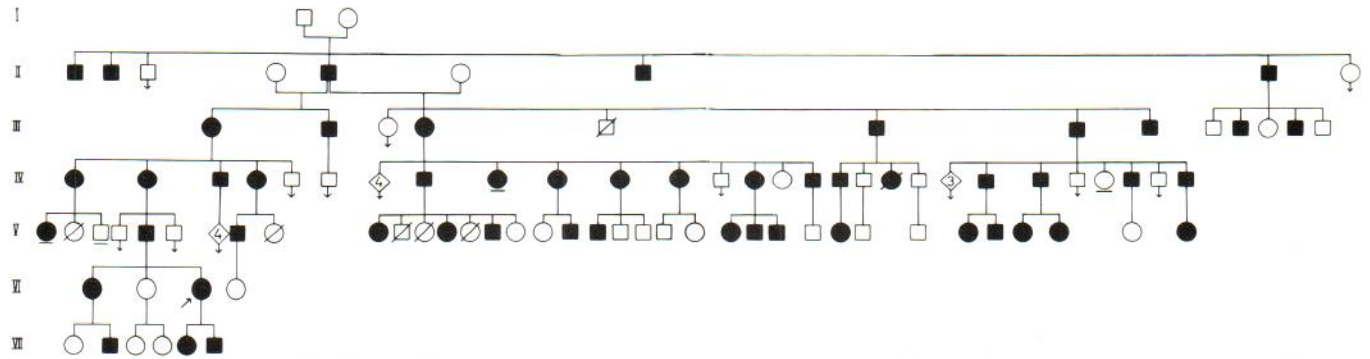


Fig. 4. Pedigree of the family. ■, □: men; ●, ○: women; ◇: individuals of unknown sex. Filled symbols represent affected persons. Crossed symbols denote death in early childhood. A crossline under a sign stands for childlessness for biological or personal reasons. Arrow pointing down represents further healthy children. Proposita is individual VI,3. The individuals III,1, III,2, III,4, III,6, and III,8 had been originally investigated by Thost.

eration. In cases of minimal expression of epidermolysis hyperkeratosis, the characteristics of this phenomenon may be easily overlooked in routine specimens, whereas in ultrastructural investigations the characteristics of epidermolysis hyperkeratosis are evident.

It is remarkable that most patients with diffuse autosomal dominant inherited PPK come from families in which the disease has been known for many generations, as in the family described here (Fig. 4). Usually the patients report that as long as they can recall, PPK is known in the family. This gives reason to assume that very few patients will suffer from PPK as the result of a new mutation. Patients with a proven negative family history by personal examination of the first-degree relatives have rarely been reported (11).

Vörner was the first to recognize the specificity of the histological feature of epidermolysis hyperkeratosis in diffuse PPK. We propose to give this autosomal dominant inherited type of diffuse PPK with the histological finding of epidermolysis hyperkeratosis demonstrated by light- or electronmicroscopy the name 'palmoplantar keratoderma Vörner'. Whether or not a clinical identical PPK without epidermolysis hyperkeratosis exists should be subject of further light- and electronmicroscopic investigations.

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