KERATOSIS PALMARIS ET PLANTARIS CUM DEGENERATIONE GRANULOSA VÖRNER

M. Berčič, S. Vrečko, J. Miljković, D. Rems

ABSTRACT

This is a report on three cases of keratosis palmaris et plantaris cum degeneratione granulosa Vörner.

The first case is a 40-year-old female patient who has palmoplantar keratoses since her first year of life. The diagnosis of Vörner's disease was made in 1983, in spite of negative family data. It was confirmed on the basis of the clinical picture, the course of the disease and especially on the basis of rare histological symptom, epidermolytic hyperkeratosis. The final confirmation is represented by the patient's son, our second case, born in August 1991 with palmaplantar keratotic lesions already at birth.

The third case, an 18-year-old girl, is unrelated with the other two and comes from a different region of the country. Her family case history is negative.

KEY WORDS

hereditary palmoplantar keratoses, epidermolytic hyperkeratosis, Vörner's disease

INTRODUCTION

Palmoplantar keratoses are a heterogeneous group of rare genodermatoses which can sometimes be associated with other symptoms, or only keratodermic lesions are presented on palms and soles.

A special variant of palmoplantar keratosis, which Vörner described in 1901, has identical clinical symptoms with Unna-Thost disease. The lesions are symmetrical and consist of diffuse thickening of the skin on palms and soles. Manual dexterity may be reduced severely. A restriction of movement is present. There are no associated symptoms.

Both diseases are transmitted as autosomal dominant traits: they begin at birth or during the first or second year of life. Vörner's disease (syn.: keratosis palmaris et plantaris cum degeneratione granulosa; epidermolytic PKK) can be excluded only histologically. Whereas in Unna-Thost disease the only sign is a massive thickening of the stratum corneum, in Vörner's disease the histological picture shows the specific symptom, i.e. epidermolytic hyperkeratosis.

OUR CASES

Case 1. V. M., a 40-year-old female patient, a teacher by profession. We first met her in 1983, during a random study

of a group of acquired keratodermic palmoplantar dermatoses.

Anamnestic data: The disease first appeared during her first year of life. The initial symptom was a severe itching of the palmoplantar skin . Later on, the skin of the palms and soles became thick and hard. In the course of several years these conditions were worsening steadily, but at present the disease no longer progresses. Because of the keratotic lesions, the movement of the hands is restricted. The temporary scaling and painful fissures are much more disturbing to our patient. As she is not a manual worker, she does not suffer too much and can cope with her problems. Her treatment consists of a permanent symptomatic topical therapy.

Clinical findings: Diffuse thickening of the palmoplantar skin, including the volar side of the fingers, of the palms and the region of the Achilles tendon of the feet. The symmetry of keratotic lesions is evident. The thickened skin is waxy and yellow, with exaggerated markings. There are some fissures. The volar and plantar hyperkeratoses are sharply defined against the normal skin by a pinkish red margin (Fig. 1). There are no other associated abnormalities of the skin, hair, nails or teeth. Otherwise her state of health is satisfactory. The routine laboratory findings are within normal limits.

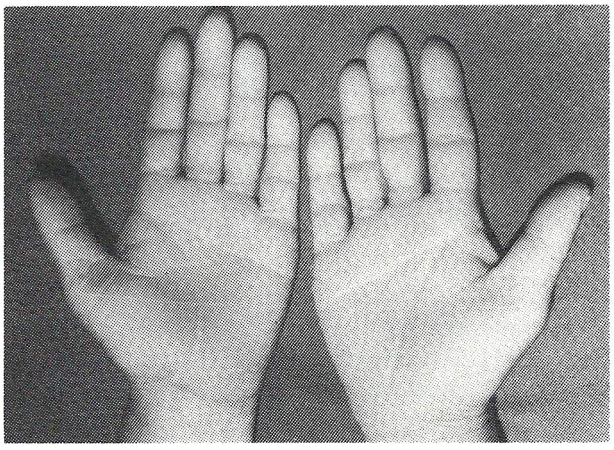

Figure. 1.
The clinical picture of epidermolytic PPK. Diffuse thickening of the skin of palms and of the volar sides of fingers. (Case 1.)

Histopathological findings: Regular acanthosis of the epidermis. Massive, compact hyperkeratosis. A thickened granular layer containing an increased number of small or large and also irregularly shaped keratohyalin granules and homogeneous eosinophilic bodies. Within the epidermal cells of the stratum granulosum et stratum spinosum there are perinuclear clear areas, the cells seem vacuolised. The basal and suprabasal layer are both normal. The superficial dermis is devoid of significant inflammatory cell infiltrates (Fig. 2).

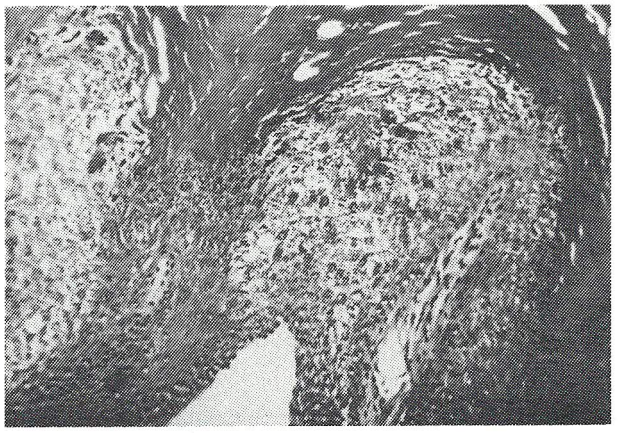

Figure 2.
Epidermolytic PPK. Hyperkeratosis, very broad granular zone with granules of various size and shape. HE - 160X.(Case 1.)

The course of the disease in our first patient is steady, the disorder does not progress.

Treatment. As the patient has no major difficulties, we did not decide on treatment with medicaments such as etretinate. The symptomatic local therapy is quite sufficient.

Case 2. V. M., an one-year-old boy, whose mother is our first female patient. In spite of her knowledge of her own hereditary disorder, she wanted the child. In the third month of pregnancy a karyogram was made, but it showed no pathological signs. As in the Maternity Center the termination of pregnancy was not strictly advised, she decided to give a birth to her child. The boy was born normally at term, after an uneventful pregnancy. Already at birth he had palmoplantar lesions. But otherwise he was a normal child, without any other abnormalities.

We first saw him when he was ten months old. Except for the skin manifestations, he was a healthy child and doing well.

The keratotic lesions on palms and soles were almost identical with his mother's, but the symptoms were not as exaggerated (Fig. 3, 4).

The histological findings were also the same. The histological picture showed the typical epidermolytic hyperkeratosis, but with less expressed histological symptoms (Fig.5, 6, 7)

Ultrastructural findings.The material was sent for electro-microspic analysis.

The course of the disease. For the time being, the disease

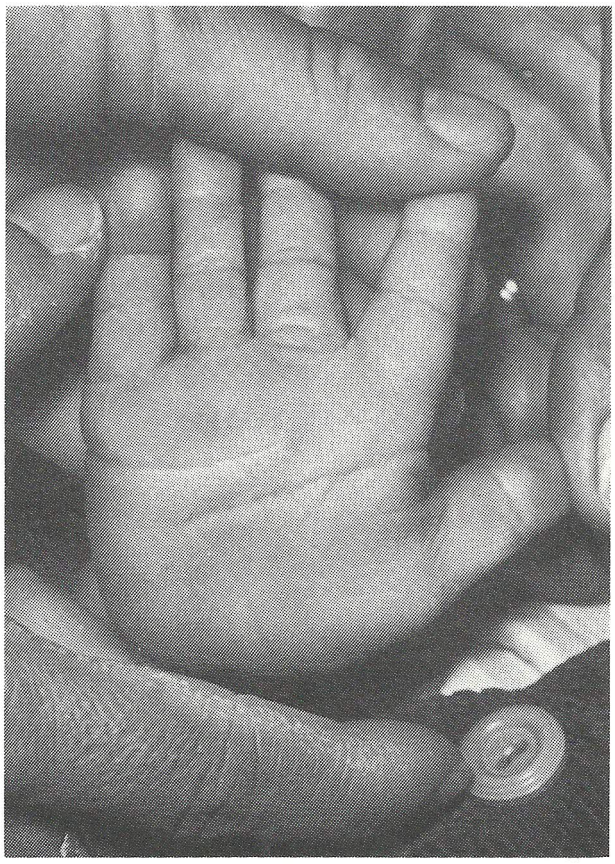

Figure 3.
Epidermolytic PPK in an 1-year-old boy. Involvement of the palm. (Case 2).

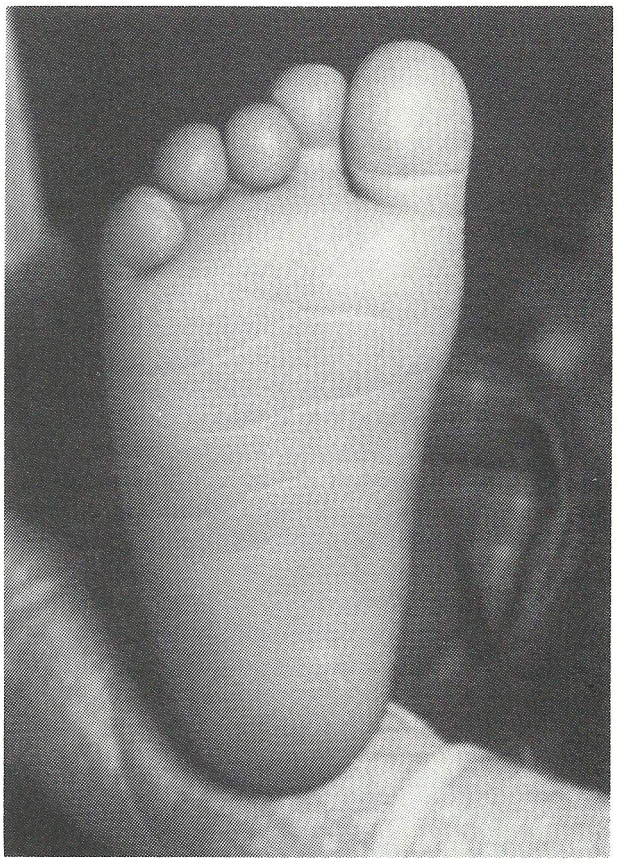

Figure 4.
Epidermolytic PPK in an 1-year-old boy. Involvement of the sole. (Case 2.)

is not changing for the worse.
Treatment. It is in the form of symptomatic topical therapy with cold cream alternating with an ointment containing salicylic acid.

Case 3. S. M., an 18-year-old girl, a student. In 1991 she had to be hospitalised because of painful symmetrical palmoplantar keratotic lesions. She declared that she had the condition since birth and that nobody in her whole family had similar difficulties.

The histological picture of epidermolytic hyperkeratosis revealed the diagnosis of Vörner's disease.

The clinical picture was identical with that in the other two patients, i. e. there were symmetrical, sharply demarcated palmoplantar keratotic lesions with a pinkish red rim.

Treatment. As the long-term therapy with etretinate orally was not satisfactory, the treatment now consist of a permanent symptomatic local application of ointments containing salicylic acid. The exaggerated keratotic masses are also removed with a special file. The patient does relatively well.

DISCUSSION

The epidermolytic PPK is considered to be a rare hereditary disorder. According to the statement by Hamm et al. (1), since the first description of this disease by Vörner in 1901, further cases were not published before 1926. The family observation by Hahn in 1911 (2) represents an exception. In the last decades, however, the number of reports has increased rapidly. In the paper published in 1988, Hamm et al. (1) gave a list of previous reports on 42 family observations and 11 sporadic cases of Vörner's disease, including their own twelve histologically confirmed cases (1, 2, 3, 4, 5, 6, 7).

To our knowledge, since 1988 three further cases were

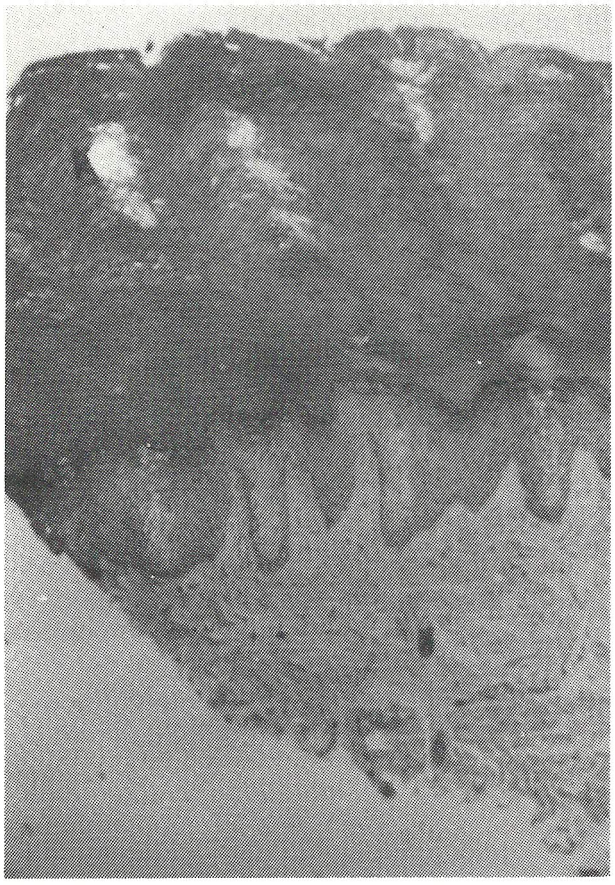

Figure 5.
Epidermolytic PPK. Hyperkeratosis, hypergranulosis, vacuolisation of the cells in stratum spinosum. HE 40X. (an 1-year-old boy case 2.)

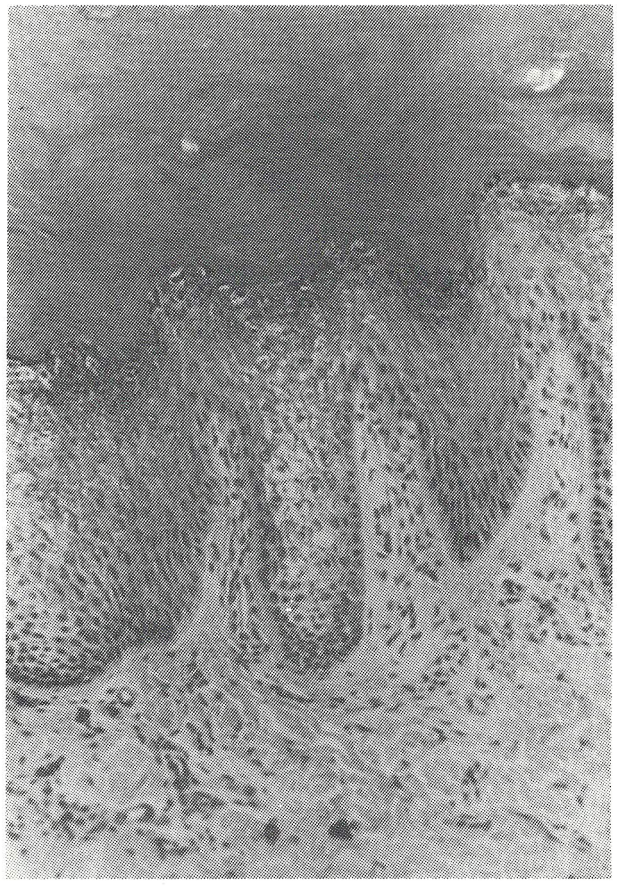

Figure 6.
Epidermolytic PPK. Higher magnification of the fig. 5. Hyperkeratosis, broad granular zone with a great number of granules, clear spaces in the cells of the stratum spinosum, HE 140X

published in literature (8, 9). To the total number we now add our three cases.

The increasing number of case reports of Vörner's disease is obviously due to the awareness of the histological pattern of epidermolytic hyperkeratosis in this disease. Within the group of diffuse PPK, Vörner's epidermolytic PPK is so far considered to be the only showing this distinctive histological traith. If this holds true, the presence of epidermolytic hyperkeratosis should allow a definite diagnosis. Consequently, a correct classification of diffuse PPK is impossible without histological investigation. For this reason, the frequency of Vörner's epidermolytic PPK was certainly underestimated in the past (1).

On the other hand, within the large group of all other hereditary keratotic disorders, Vörner's disease is not the only condition with the specific histologic and ultrastructural findings of epidermolytic hyperkeratosis. The same histological changes are also found in other disorders such as congenital bullous ichthyosiform erythroderma and nevus hystricoides, including some linear and systematised nevi.

As early as 1966 Schnyder and Klunker (7) were of the opinion that these three manifestations were related. This opinion is shared by other authors (6, 8). Claudine Blanchet-Bardon et al. believe that all three conditions represent the same disease in which clinical manifestations range from generalised forms (congenital bullous ichthyosiform erythroderma) to localised forms, such as Vörner's PPK and nevus hystricoides linearis. Hadlich and Ruthild Linse (8) support this opinion and confirm it with their own case of one male patient showing the clinical and histological symptoms of all three disorders.

The mode of inheritance of Vörner's disease is not absolutely

elucidated yet. Although it is considered to be a hereditary disorder, inherited in the dominant autosomal trait, there are numerous solitary cases of the disease (1). On the basis of their report on two brothers with epidermolytic PPK with negative family data, Quasem et Ahmed (9) even suggest the possibility of an autosomal recessive mode of inheritance.

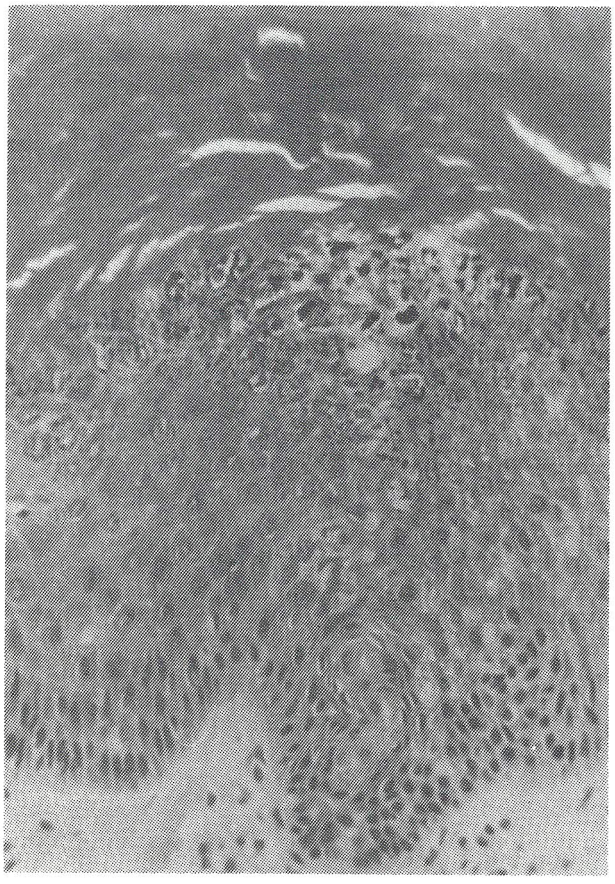

Figure 7.
Epidermolytic PPK. The typical changes in the granular layer. (Detail of fig. 6.)

Hadlich and Ruthild Linse (8) support the belief that large family investigations are difficult to carry out and they are not always precise. Therefore the number of genuine solitary cases may not be so high. We share this opinion. Our first case of Vörner's disease seemed at first to be a solitary case. But 9 years later, with the bird of the boy showing evidence of the same clinical and histological conditions, the case turned into an authentic family case (Fig. 8).

TREATMENT.

The treatment of Vörner's disease is not satisfactory.

Etretinate, or its main metabolite etretin, is effective, if at all, only in arresting the progression of the disease, but it may

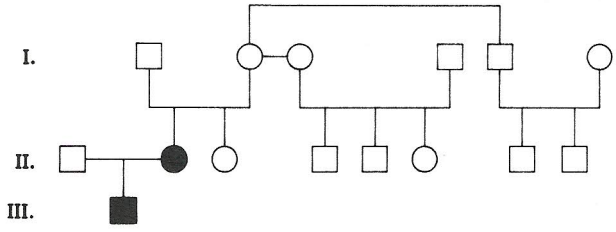

Figure 8.
The pedigree of mother and son with epidermolytic PPK. (Case 1. and 2.)

improve the working capacity of the patient. As this drug has no influence on the basic defect of disorder, it is even not indicated. Metotrexate is also not helpful. There are some reports on the relatively good effect of surgical treatment (10). So in most cases permanent symptomatic topical therapy remains as our only option.

Our patients were also treated in similar manner.

We thank M. Gajšek-Marchetti, translator, from the Medical Research Department at Maribor Teaching Hospital, for help.

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