

CASE REPORTS

Keratoderma Palmoplantaris Varians (Striata et Areata). A Form of Chronic Idiopathic Acrokeratosis Described by Degos

A Padial, V Morales, JC Armario-Hita, JL Ingunza, and JM Fernández-Vozmediano

Servicio de Dermatología, Hospital Universitario de Puerto Real, Universidad de Cádiz, Spain

Abstract. We report a case of a 15-year-old boy with hyperkeratotic lesions that were linear or striated on the palms and nummular on the soles. He was the only family member known to be affected, suggesting that the condition could be attributed to a *de novo* mutation or the recessive form of keratoderma palmoplantaris striata, described by Degos as chronic idiopathic acrokeratosis. The lesions did not improve with topical treatments (keratolytic agents, emollients, or corticosteroids) or oral retinoids. We observed that scratching of the affected areas was the main reason for deterioration of the lesions.

Key words: palmoplantar keratoderma, keratoderma striata, keratoderma *varians*, keratosis palmoplantaris *varians*, keratoderma of Wachters.

QUERATODERMIA PALMOPLANTAR VARIANS (STRIATA ET AREATA) TIPO ACROQUERATOSIS ESENCIAL CRÓNICA DE DEGOS

AResumen. Paciente varón de 15 años de edad con lesiones hiperqueratósicas con disposición lineal o estriada en las palmas y numular en plantas. Era el único miembro afecto conocido de su familia, por lo que se podría tratar de una mutación *de novo* o de la forma recesiva de queratosis palmoplantar *estriata varians*, denominada por Degos «acroqueratosis esencial crónica». Las lesiones no mejoraron con los tratamientos tópicos instaurados (queratolíticos, emolientes y corticoides) ni con retinoides orales. Se pudo constatar que el rozamiento o fricción de las zonas afectas era el principal factor de empeoramiento de las mismas.

Palabras clave: queratodermias palmoplantares, queratoderma estriada, queratoderma *varians*, queratosis *palmoplantaris varians*, queratoderma de Wachters.

Introduction

Palmoplantar keratoderma (PPK) of the *varians* type is an uncommon hereditary condition that is characterized by thickening of the skin on the palms and soles. Patients affected by this condition generally experience 2 clinical forms of hyperkeratosis simultaneously: a striate form on the palms and a nummular or areate form on the soles. However, a characteristic of this type of PPK is that there are several variants, even within the same family. The lesions do not usually improve with standard treatments and they seem to be associated with friction or rubbing in the affected areas, leading to exacerbation of the hyperkeratosis.

Case Description

The patient was a 15-year-old boy with no personal or family history of interest who consulted for hyperkeratotic lesions that had been present on the palms and soles since birth. The lesions were more severe in areas of rubbing or friction. The soles were affected by thick and compact keratoderma with a predominance on the head of the first and fifth metatarsals, hallux, and heels, where it was horseshoe-shaped. The arch of the foot was spared, and in areas of less friction the skin was parchment-like (Figure 1). On the palms, the keratoderma was linear and mainly involved pressure points and the flexor aspect of the fingers. The lesions were clearly predominant on the right hand (Figure 2) and the patient had 20-nail dystrophy (Figure 3).

Additional tests (complete blood count and biochemistry) were normal. The histopathology study was inconclusive and only revealed the presence of marked hyperkeratosis with focal parakeratosis, significant hypergranulosis, and acanthosis (Figure 4). These findings

Correspondence:
José Manuel Fernández Vozmediano
Servicio de Dermatología
Hospital Universitario de Puerto Real
Cádiz, Spain
fdezvozmediano@dermasur.com

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suggested keratosis palmoplantaris striata et areata or focal keratosis palmoplantaris varians.

Treatment was applied locally with keratolytic agents, emollients, and corticosteroids, and systemically with oral retinoids, but there was no response. The disease course was varied and was more influenced by mechanical factors than by the treatment applied.

Discussion

The first reports of this condition were by Brünauer (1923),¹ Fuhs (1924),² and Siemens (1929),³ who described different hyperkeratotic processes on the palms and soles in several families. Siemens named the condition keratosis palmoplantaris striata et areata. Brünauer had reported that, in adults, the lesions followed a pattern of striate keratosis on the palms that was associated with keratotic bands affecting the eponychium and cracked keratotic paronychia, whereas on the soles, the lesions were distributed in islands on the pressure points, and on the heels, they took the form of a horseshoe, as also described by Siemens, thus constituting keratosis palmoplantaris striata et areata. However, the denomination keratosis palmoplantaris varians was first suggested by Wachters,⁴ who presented a doctoral thesis in 1963 after studying the members of 3 families and reviewing all previously published cases. He observed a high degree of clinical variability between one patient and another, even within the same



Figure 1. Compact plantar keratoderma that is predominant on the head of the first and fifth metatarsals, hallux, and heels, where it is horseshoe-shaped.

family. However, Wachters did not actually define any new condition, but merely included previously described entities under the term *varians*.

The different classifications of PPK currently lack uniformity owing to the large number of names and synonyms for each clinical form.⁵ In the Lucker classification,⁶ the disease is included in the keratosis palmoplantaris areata et striata group and is defined as hereditary nummular PPK, which is autosomal dominant

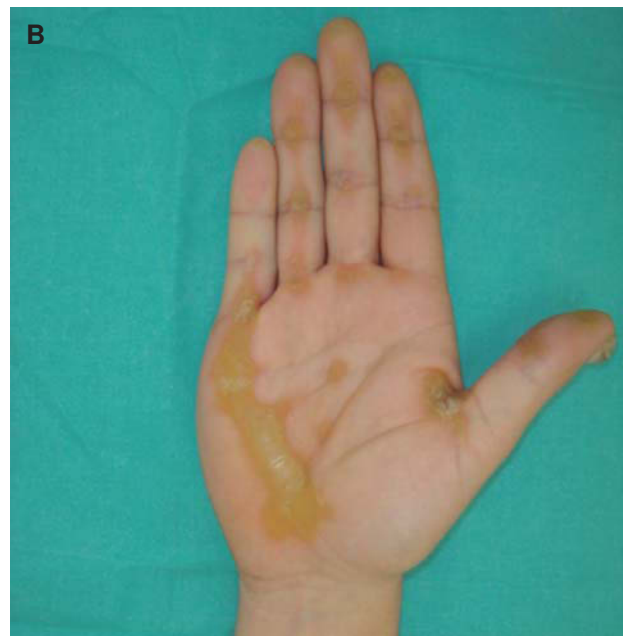


Figure 2. Linear arrangement on the palms with predominant involvement of the pressure points and flexor surface of the fingers (A). The lesions are clearly more predominant on the right hand (B).



Figure 3. Twenty-nail dystrophy

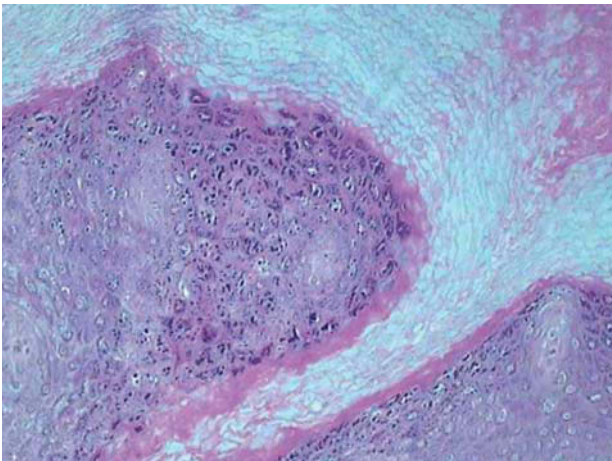


Figure 4. Marked orthokeratotic hyperkeratosis with marked hypergranulosis and acanthosis. Hematoxylin-eosin, $\times 100$.

and involves no other conditions. This classification does not separate Wachters syndrome from the well-known Brünauer-Fuhs-Siemens syndrome, although it points to clinical variability as the main characteristic of this condition. This process has also been referred to as Siemens syndrome, striate PPK, and acral PPK.

It has been established that this is an autosomal dominant form stemming from mutations in the desmosomal proteins affecting desmoglein 1 on chromosomes 18q12.1-q12.2 and 12q13 and desmoplakin,⁷⁻⁹ located at 6p21,⁸⁻¹⁰ although other authors locate it at 18q.^{8,11,12} It has also been linked to a mutation in the V2 domain of type 1 keratin. This genetic heterogeneity could be responsible for the clinical variability that characterizes this form of PPK.¹³ A greater tendency to develop lesions after prolonged injury has been reported in patients who express HLA B18.¹⁴

Recent publications have established 2 forms of striate PPK^{8,15}:

1. First, the so-called type 1 or Brünauer-Fuhs-Siemens syndrome, with lesions distributed linearly or in islands on the palms and soles and associated deafness in some cases. This form is considered to be autosomal dominant with mutations at loci 18q12.1-q12.2 and 12q13 of desmoglein 1 and desmoplakin, which are constituents of keratin 1.
2. Second, type 2 is present from infancy and is also considered autosomal dominant and linked only to locus 12q13 of the keratin 1 gene.

With our patient, we were unable to demonstrate the existence of previous cases; therefore, ours may be one of those isolated cases, with no family history, that Degos termed chronic idiopathic acrokeratosis,¹⁶ and which, for some authors, would be the result of a de novo or autosomal recessive mutation.¹⁴

The lesions are unnoticeable during the first months of life, develop gradually during childhood as multiple hyperkeratotic lesions on the hands, and in the following years take on a diffuse form or a parchment-like appearance on the palms. Lesions on the soles appear earlier and occur as hyperkeratosis with a nummular or island distribution. They later progress to diffuse keratoderma. On the palms, the lesions take on a linear or striate pattern, although some authors believe that this can be variable and linear, nummular, membranous, or fissured in appearance.¹⁷ The membranous or parchment-like appearance of the palms is very characteristic in people who are not physically active. The thickness of the hyperkeratosis is considerable and can lead to a certain degree of disability due to the development of painful fissures and cracks. Patients can also present other clinical manifestations, such as subungual hyperkeratosis and transverse unguis grooves, although these findings are not constant. The elbows and knees are less frequently involved and some patients present mild hyperhidrosis. There are reports of an association with woolly hair.

Histology of the skin is nonspecific and reveals orthokeratotic hyperkeratosis with foci of parakeratosis and hypergranulosis, a certain degree of acanthosis and papillomatosis, with no evidence of epidermolysis. Therefore, diagnosis is clinical depending on morphology and course. Advances in molecular biology will one day provide us with a genetic diagnosis by determining the specific mutation, and may even enable prenatal screening.¹⁸

Treatment is not very effective and only relieves the symptoms. There is no definitive cure.¹⁹ Some authors have used topical keratolytics, corticosteroids, calcipotriol, and systemic treatment with retinoids,^{20,21} although few results are available.

Conclusions

Ours was a case of PPK *varians* or *striata et areata* in the presentation described by Degos as chronic idiopathic acrokeratosis, since the patient had no family history of a similar condition; therefore, our case involved either a *de novo* or autosomal recessive mutation. Currently, there are no clear criteria for defining striate PPK as an independent entity or as synonymous with the Brünauer-Fuhs-Siemens type, except for genetic testing, which is not always readily available. Clinically, these entities are one and the same process, although molecular biology and genetic studies appear to have established at least 2 different forms. Future research is expected to provide a more precise classification.

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