Hereditary keratosis palmoplantaris varians of Wachters (keratosis palmoplantaris striata et areata)

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SUMMARY

Keratosis palmoplantaris varians of Wachters is a rare autosomal dominant disorder. A 17-year-old boy with characteristic clinical and histopathologic findings of 10 years' duration is described. He displayed yellowish patchy and linear hyperkeratoses of the palms and nummular keratoses on the soles that were symmetrical and often confluent, specifically on the pressure points. Because there was no family history of a similar pattern, he probably represented an isolated case in his family. The typical clinical presentation, differential diagnosis, and classification of hereditary palmoplantar keratoses are discussed.

KEY WORDS

hereditary palmoplantar keratoses, keratosis palmoplantaris varians of Wachters, keratosis palmoplantaris striata et areata, striate palmoplantar keratoderma, genodermatoses, autosomal dominant

Introduction

Hereditary palmoplantar keratoses (HPPKs) are a heterogeneous group of disorders of keratinization characterized by marked localized or diffuse thickening of the epidermis on the palms and soles. They are classified by specific morphology, severity of keratoderma, distribution of the hyperkeratosis ("progrediens" or "transgrediens"), associated symptoms, mode of inheritance, age of onset, and histological findings (1–7). According to an updated classification, keratosis palmoplantaris varians of Wachters is among the hereditary isolated primary localized forms of nummular-linear (striate or insular) morphology without associated features (4, 6, 7). It is also known as keratosis palmoplantaris striata et areata or striate palmoplantar keratoderma (OMIM no. 148700) (4). We describe a patient with this rare disease and discuss the classification of HPPKs (Table 1).

Case report

A 17-year-old boy, the son of non-consanguineous parents, was seen for evaluation of palmoplantar hyperkeratosis of 10 years' duration. He had displayed keratotic plaques in the palmoplantar region from age seven. They progressed slowly and remained asymptomatic until the development of transverse plantar fissures 3 months prior to evaluation, which made extensive walking painful. The keratoderma was not associated with other cutaneous or systemic abnormalities. No one in the family was known to have similar findings. The family history was otherwise non-contributory.

Table 1. Hereditary palmoplantar keratoses.

Clinical patterns	Inheritance	No associated features	Associated features
Diffuse	AD	Unna-Thost (PPK diffusa circumscripta) Greither (PPK transgrediens et progrediens) Vörner (PPK cum degeneratione granulosa) Sybert (PPK transgrediens)	Vohwinkel (PPK mutilans) Howel-Evans (PPK with carcinoma of the esophagus) Huriez (PPK with sclerodactyly) Clouston (Hidrotic ectodermal dysplasia) PPK and sensorineural deafness Olmsted (Mutilating PPK with periorificial keratotic plaques)
	AR	Mal de Meleda (keratosis extremitatum hereditaria transgrediens et progrediens) Gamborg Nielsen ("Nagashima-type–like") Acral keratoderma (PPK transgrediens)	Papillon-Lefèvre (PPK with periodontosis) Bureau-Barrière-Thomas (PPK with clubbing of the fingers and toes and skeletal deformity)
Nummular/linear	AD	Wachters (PPK varians, Brünauer-Fohs-Siemens syndrome, Siemens syndrome) PPK nummularis ("hereditary painful callosities")	Richner-Hanhart (Tyrosinemia type II) Pachyonychia congenita (Jadassohn-Lewandowski syndrome)
	AR		Focal palmoplantar and oral mucosa hyperkeratosis syndrome (Fred syndrome) Pachyonychia congenita Keratosis palmoplantaris papillomatosa et verrucosa (Jakac-Wolf syndrome) Hanhat (PPK with lipomata)
Papular	AD	PPK punctata (Davies-Colley syndrome, Buschke-Fischer-Brauer syndrome) Acrokeratoelastoidosis (Costa syndrome) Focal acral hyperkeratosis	
	AR		Schöpf-Schulz-Passarge syndrome (PPK with cystic eyelids, hypodontia, and hypotrichosis)

From Lucker et al (6).

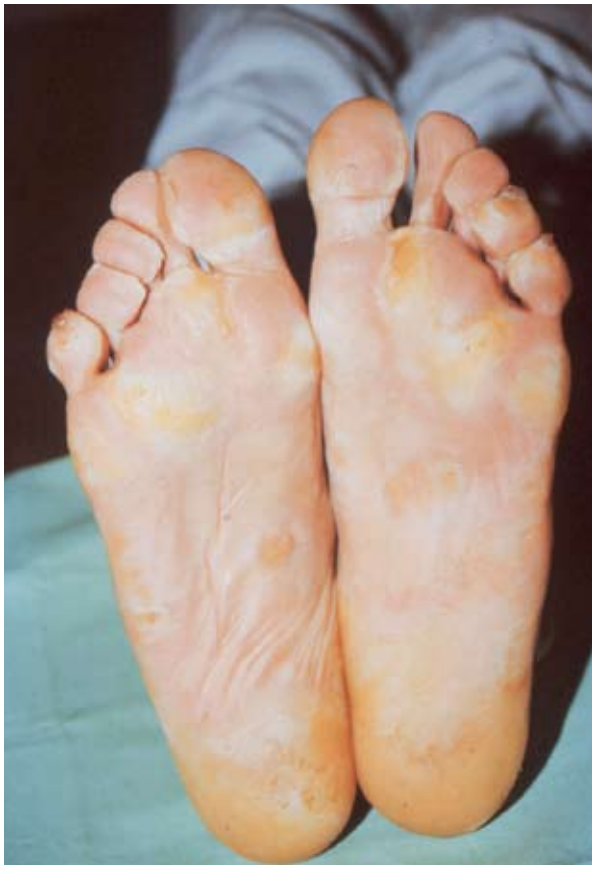

Figure 1.
Nummular hyperkeratotic plaques localized mainly at pressure points.

Physical examination showed a normal and healthy young man. Cutaneous examination revealed yellowish nummular keratoses on the soles, specifically on the pressure points (Fig. 1). Over the palms, and particularly marked on the flexor aspect of the fingers of the right hand, there were patchy hyperkeratotic plaques arranged as longitudinal bands. Numerous painful transverse fissures were also visible (Fig. 2). No other significant cutaneous findings were evident. Oral mucosae, hair, nails, and teeth were all normal.

Routine laboratory tests of blood and urine were normal. Histological examination of a skin biopsy specimen from a plantar plaque showed marked hyperkeratosis with hypergranulosis and moderate acanthosis without inflammatory cell infiltrate in the dermis (Fig. 3). No epidermolytic hyperkeratosis was found. The patient was treated with mechanical debridement and lubrication with warm soapy baths, 5 to 10% salicylic acid ointment, and 10% urea in petrolatum. Minimal improvement was attained with these measures.

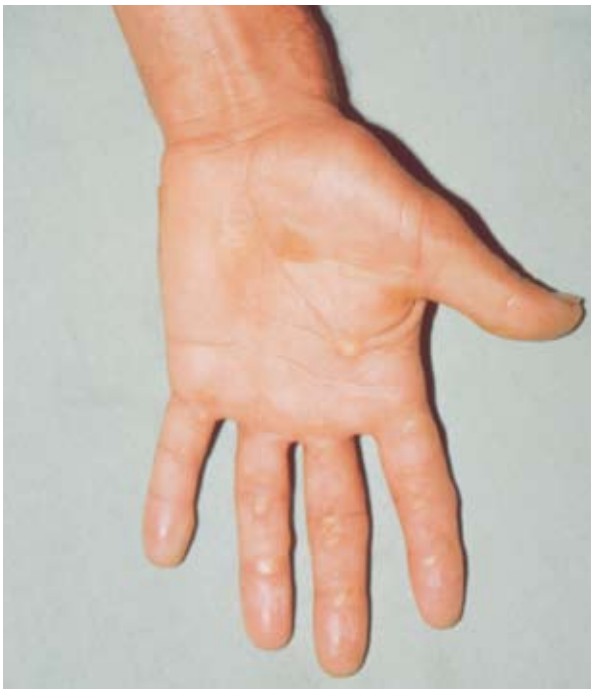

Figure 2.
Hyperkeratotic (nummular- linear) lesions on the right palm.

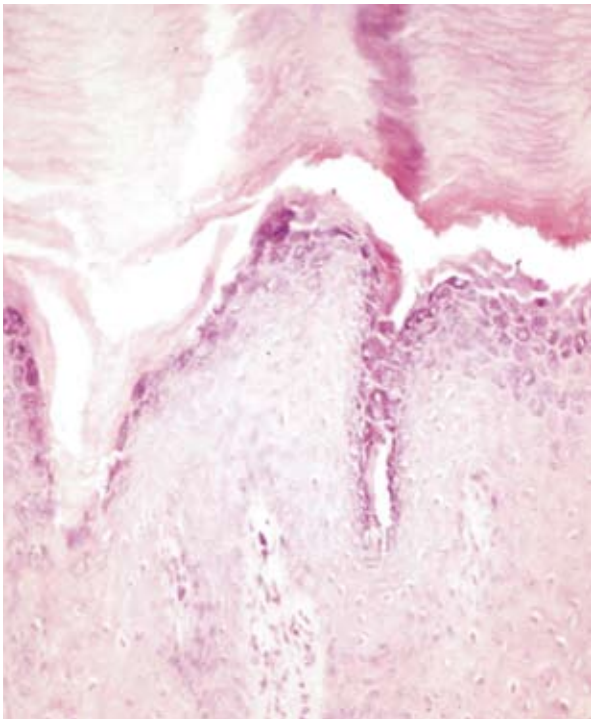

Figure 3.
Histologic examination shows hyperkeratosis, acanthosis, and hypergranulosis (Hematoxylin-eosin stain 100×).

Discussion

The hereditary palmoplantar keratoses (HPPKs) are a group of keratinization diseases characterized by thickening of the epidermis of the palms and soles and a chronic clinical course. They are classified by the morphology and distribution of the hyperkeratosis ("progrediens" or "transgrediens"); the severity, presence, or absence of associated features; the inheritance pattern; the age of onset; and histopathological findings. There are three clinical patterns: diffuse, nummular-linear (striate or insular), and papular (4, 6, 7). Keratosis palmoplantaris varians of Wachters is listed among the hereditary localized forms of nummular- linear type with no associated features. Its diagnosis in our specific case was made on the basis of the nummular-linear, non-transgrediens appearance of the keratoderma, absence of any associated features, and histologic findings lacking epidermolytic hyperkeratosis. This form of HPPK is classified in the group of uncommon nonepidermolytic HPPKs.

Keratosis palmoplantaris varians Wachters is a rare genodermatosis, more frequent in males (8), with autosomal dominant transmission and probable complete penetrance (9). However, there are also sporadic (10) and congenital cases (11). It appears in the first or second decade of life. Originally, distinct subtypes were described by Fuhs (12), Brünauer (13), and Siemens (14), because of the diversity of clinical features. All of these variants however, should be considered as one keratoderma, for which Wachters (15) in 1963 introduced the term "keratosis palmoplantaris varians" to emphasize its inter- and intra-family clinical variability, because it is characterized by varied phenotypic expression. Clinically, the palmar keratoses have a linear, nummular, membranous, fissured, or periungual configuration. The keratoses on the soles have a nummular appearance and are localized to the pressure points (6, 9, 14). In addition to the localized forms, nummular keratoses over the elbows, knees, and Achilles tendon were also described (9). Nail changes and periodontosis have been noted as associated features in a father and son (16). Nevertheless, because of the absence of associated features in other families (8, 9, 12–15), keratosis palmoplantaris varians of Wachters is classified in the group of nummular-linear HPPKs without associated features (4, 6, 17). In addition, the differential diagnosis also mandates the exclusion of Carvajal-Huerta syndrome (generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and an arrhythmogenic left ventricular cardiomyopathy) and Howel-Evans syndrome (familial focal non-epidermolytic palmoplantar keratoderma and esophageal squamous cell carcinoma) (3, 18, 19).

It is not possible to establish whether our patient's condition represents a spontaneous mutation or a rare type of sporadic recessive HPPK. Its genetic basis is not known (1), although some patients with striate keratoderma have mutations in keratin 1, desmoplakin, or desmoglein 1 (3). An immunohistochemical and ultrastructural study performed on one patient to characterize altered keratinization and maturation patterns using anti-cytokeratin antibody KL1 showed no significant difference in reaction pattern in comparison with controls, although early expression of both filaggrin and involucrin was found (10). Ultrastructurally, tightly packed tonofibrils and large masses of keratohyalin granules with abnormal configuration were evident.

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A U T H O R S ' A D D R E S S E S

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