Single Case

Punctate Palmoplantar Keratoderma: A Case Report of Type 1 (Buschke-Fischer-Brauer Disease)

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Keywords

Palmoplantar keratoderma · Punctate palmoplantar keratoderma · Buschke-Fischer-Brauer disease · Porokeratosis punctata palmaris et plantaris

Abstract

Punctate palmoplantar keratoderma is a rare hereditary palmoplantar keratoderma. Herein we report a 59-year-old male, otherwise healthy, who presented with a 25-year history of asymptomatic persistent slowly progressing skin lesions on both hands. The parents are non-consanguineous and none of his family members had similar lesions. Skin examination revealed multiple tiny keratotic pits on both palms. Punch skin biopsy from the palmar lesion revealed epidermal depression with an overlying column of compact orthokeratosis. Based on the above clinicopathological findings, a diagnosis of punctate palmoplantar keratoderma type 1 was made. The patient was started on 40% urea and 20% salicylic acid ointment for months but with little improvement.

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Introduction

Palmoplantar keratoderma (PPK) comprises a heterogeneous group of disorders characterized by hyperkeratosis of the palms and soles [1]. It can be hereditary or acquired. Hereditary PPK can be further classified into three major categories: diffuse, focal, and punctate PPK (PPPK) [2]. These diseases can be distinguished from each other on the basis of inheritance pattern, onset, distribution, morphology, severity, histopathological findings, additional dermatological findings, and systemic manifestations [3]. The prevalence of PPPK was estimated to be 1.17/100,000 [4]. The exact etiology is little understood. It is believed that both genetic and environmental factors play some roles. PPPK displays an autosomal dominant pattern of inheritance and has been linked with two loci on chromosomes 15q22 15q24 and 8q24.13–8q24.21 [5]. Mutations in AAGAB and COL14A1 have been recognized to date [3]. This autosomal dominant condition tends to occur much later than other hereditary keratodermas. It usually appears between the age of 20 and 30 years [6]. Male patients tend to be more commonly affected [7]. Clinically, PPPK presents as multiple tiny hyperkeratotic papules on the palmoplantar surfaces, often described as “raindrop” keratosis. On palms, the lesions are usually scattered, whereas on soles, they are coalesced [8]. The lesions may evolve over time, becoming translucent, opaque, or verrucous. Some papules may form a keratotic core and detachment of the core may lead to a characteristic central depression [2]. Patients generally remain asymptomatic but rarely, pain can be caused by pressure [9]. Herein we report a rare case of type 1 PPPK on both palms.

Case Report

A 59-year-old male, otherwise healthy, presented with a 25-year history of asymptomatic persistent slowly progressing skin lesions on the hands. Past medical history and review of systems were unremarkable. The parents are non-consanguineous and none of his family had similar lesions. Skin examination revealed multiple tiny brownish keratotic pits on both palms (Fig. 1). There were no similar lesions elsewhere on his body. Hair, nails, and mucous membranes were normal. Complete blood count with differentials, peripheral blood smear, liver enzymes, urea, creatinine, electrolytes, abdomen and pelvis ultrasound, chest X-ray, and computed tomography of the chest, abdomen, and pelvis were normal. Punch skin biopsy from the lesion revealed epidermal depression with an overlying column of compact orthokeratosis (Fig. 2). Based on the above clinicopathological findings, a diagnosis of PPPK type 1 was made. The patient was reassured and started on 40% urea ointment and 20% salicylic acid ointment with little improvement.

Discussion

There are 3 types of PPPK. Table 1 shows the differences between these three types. [1, 3]. The histopathological features of our case were consistent with type 1 (epidermal depression with an overlying column of compact orthokeratosis). Important differential diagnosis includes Darier’s disease, pitted keratolysis, punctate porokeratoses, basal cell nevus syndrome, basaloid follicular hamartoma syndrome, reticulate acropigmentation of Kitamura,

Cowden syndrome, and porokeratotic adnexal ostial nevus. However, the histopathology of our case was typical for type 1 PPPK. Associated systemic involvement is rare. Studies have suggested possible associations between PPPK and lymphoma as well as malignancies of the pancreas, colon, breast, and kidney [2]. However, our patient has none of these malignancies. No standardized treatment for PPPK has yet been established [9].

Management of PPPK includes topical keratolytics, liquid nitrogen, PUVA (psoralen plus ultraviolet A), systemic or topical retinoids, systemic acitretin, etretinate or alitretinoin, and topical steroid [6]. Few numbers of reported cases demonstrated successful treatment with keratolytics and systemic acitretin. However, relapses are common [9]. Our patient was treated with topical 40% urea ointment and 20% salicylic acid ointment. At follow-up 6 months later, the lesions showed little improvement. However, the patient was lost to follow-up.

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Statement of Ethics

The authors have no ethical conflicts to disclose. Written consent has been obtained from the patient. The research was conducted ethically in accordance with the World Medical Association Declaration of Helsinki. The study protocol was approved by the institute's committee on human research. Information revealing the subject's identity is to be avoided. All patients should be identified by numbers or aliases and not by their real names.

Disclosure Statement

The authors have no conflicts of interest to declare.

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Author Contributions

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Fig. 1. Palms of the patient showing multiple tiny brownish keratotic pits.

Fig. 2. Histopathology of the lesion showing cup-shaped epidermal depression with overlying column of compact orthohyperkeratosis.

Table 1. Differences between the 3 different types of punctate palmoplantar keratoderma

Name	Punctate PPK type 1 (Buschke-Fischer-Brauer disease)	Punctate PPK type 2 (spiny keratoderma)	Punctate PPK type 3 (acro-keratoelastoidosis)
Inheritance	Autosomal dominant	Autosomal dominant	Autosomal dominant
Onset	Late childhood to adulthood	Puberty to early adulthood	Adolescence to adulthood
Morphology of the PPK	Multiple hyperkeratotic papules with central indentation; worsening of papules upon exposure to water	Early onset: multiple spiny keratosis Late onset: pits with keratotic plugs	Translucent hyperkeratotic papules, sometimes umbilicated, on lateral aspects of palms and sole
Other skin/cutaneous involvement	Nail dystrophy (uncommon)	No	Nail dystrophy (extremely rare)
Associated systemic involvement	Association with malignancies (rare)	Facial sebaceous hypoplasia in males	No
Histologic features	Epidermal depression with an overlying column of compact orthokeratosis	Epidermal depression with an overlying column of para keratosis, in contrast to porokeratosis, the granular layer is preserved	Hyperkeratosis and hypergranulosis; decreased number of fragmented elastic fibers (elastorrhexis)

PPK, palmoplantar keratoderma.