Case Report

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Striate Palmoplantar Keratoderma (Brunauer-Fohs-Siemens Syndrome)

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Abstract

A 24-year-old man presented with linear hyperkeratotic plaques on the volar fingers, which he had since childhood. That was exacerbated by manual labour. No family member was affected with similar condition. Histopathologic and electron microscopic results were consistent with striate palmoplantar keratoderma. Striate palmoplantar keratoderma or Brunauer-Fohs-Siemens syndrome is an autosomal dominant condition that presents with linear hyperkeratosis on the palms and fingers and focal plaques on the plantar aspects of the feet. Histopathologic features include hyperkeratosis, hypergranulosis, and acanthosis with no epidermolysis. Electron microscopic examination shows diminished desmosomes, clumped keratin filaments, and enlarged keratohyalin granules. The syndrome has been linked to mutations in desmoglein 1, desmoplakin, and keratin 1. Keratolytics are the mainstay of therapy. Use of topical or oral retinoids and surgical debridement has also been described.

Keywords: Striate; Palmoplantar keratoderma; Brunauer-Fohs-Siemens syndrome

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Introduction

Striated palmoplantar keratoderma (Brunauer-Fohs-Siemens syndrome) is a very rare, focal, nonepidermolytic PPK with an autosomal dominant pattern of inheritance. Striated PPK has a well-characterized clinical presentation that differentiates it from other genodermatoses. There is a focal linear hyperkeratosis of the palms that extending from the palms into the volar surface of fingers and focal plaques on the plantar aspects of the feet. Striate PPK is a genetically heterogeneous disorder of keratinization, several mutations in desmoglein 1, desmoplakin, and keratin 1 have been identified. We report in this paper a rare case of Brunauer-Fohs-Siemens syndrome, who presented with linear hyperkeratotic plaques on the volar fingers, which he had since childhood.

Case Report

A 24-year-old male patient, not known to have any medical illness, presented to dermatology clinic with asymptomatic lesion over the palmar surface of both hands since childhood that was exacerbated by manual labour. Slight improvement with topical keratolytic medications but never disappear. Recently, it interferes with his daily activity. Systemic review was unremarkable and no family members had similar lesions.

Physical examination

He had normal weight, length, and vital signs. No dysmorphic features. Cardiovascular system, Respiratory system, Gastrointestinal system and neurological examination were normal.

Skin examination

Figure 1 Over the palmar surface of the left hand, there are linear hyperkeratotic plaques extending the length of the volar fourth fingers and focally on the third and fifth fingers. Plantar surfaces of both feet showed focal hyperkeratotic plaques. Hair, teeth, nail, mucous membrane, and non-palmoplantar skin were normal.

Several laboratory investigations were performed for the patient; his complete blood count, electrolytes, liver and renal function tests were within normal limits. Echocardiography revealed normal findings.

Skin biopsy **Figure 2** showed marked hyperkeratosis, hypergranulosis and irregular acanthosis overlying mild perivascular chronic inflammatory infiltrate. Electron microscopy examination showed abnormal tonofilaments with clumps of keratin.

Figure 1 Over the palmar surface of the left hand, there are linear hyperkeratotic plaques extending the length of the volar fourth fingers and focally on the third and fifth fingers.

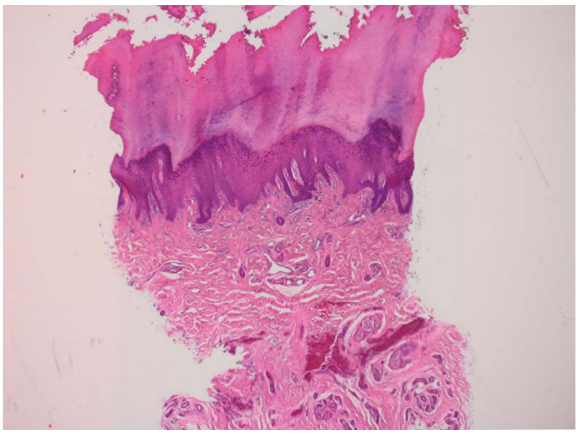

Figure 2 (Hematoxylin and eosin stain x4) showed marked hyperkeratosis, hypergranulosis and irregular acanthosis overlying mild perivascular chronic inflammatory infiltrate.

Therapy was initiated with topical Salicylic acid 10% cream once daily, urea 20% cream twice daily and petrolatum ointment twice daily.

We were follow-up the patient in the out-patient dermatology clinic every two months for 2 years' duration. At two follow-up visit, there was a slight clinical improvement. No new lesion appeared during a 24 months' follow-up period until the time of writing this paper.

Discussion

Palmoplantar keratodermas (PPKs) represent a heterogeneous group of both hereditary and acquired diseases that are linked together by the characteristic phenotypic expression of epidermal thickening of the palms and soles. Hereditary PPKs tend to occur earlier in life, can be traced in family pedigrees, and may be associated with well described syndromes. Conversely, acquired PPKs occur later in life, without a positive family history, and tend to be attributable to an underlying etiology. There are different classifications of PPKs. PPK is classified clinically as diffuse, focal, striate, or punctuate type per the arrangement of hyperkeratosis. PPK can be classified into transgradient and non-transgradient forms per the distribution of lesions. Also, PPK can be classified histopathologically into epidermolytic and non-epidermolytic types. It can be isolated palmoplantar keratoderma or associated with other features such as impaired hearing, cardiomyopathy, starfish keratoses, pseudoainhum, knuckle pads, atrophy, oral lesions, nail changes, blisters [1].

Striate PPK is known to be caused by heterozygous mutations in either the desmoglein 1 (type I striate PPK), desmoplakin (type II striate PPK) or keratin 1 (type III striate PPK) gene. Focal isolated striate palmoplantar keratoderma also known as Brunauer-Fohs-Siemens syndrome is a rare, simple non-epidermolytic palmoplantar keratoderma with an autosomal dominant mode of inheritance. The age of onset is usually in early childhood. It is characterised by a focal linear hyperkeratosis of the palms that extending from the palms into the volar surface of fingers. Areas of pressure on the soles are often more affected than the hands. Manual labour and repetitive trauma may exacerbate palmar hyperkeratosis. On the other hand, the patients with sedentary occupations tend to have milder changes.

Usually there is no any local or systemic association with Brunauer-Fohs-Siemens syndrome. But when we reviewed the literatures, we found one case report of Brunauer-Fohs-Siemens syndrome that associated with acral malignant melanoma [2]. The major histopathological features of striate PPK include hyperkeratosis, hypergranulosis, and acanthosis with no epidermolysis. Granular and filamentous material that represents clumped keratin filaments also may be observed [3]. Electron microscopic examination may show diminished desmosomes, clumped keratin filaments, and enlarged keratohyalin granules.

Treatment of keratoderma is symptomatic. Selecting an appropriate treatment modality of PPKs depend on many factors include the severity of symptoms, the degree of hyperkeratosis, response to treatment and the age of the patient. Keratolytics and emollients are the mainstay of therapy. Both topical and systemic retinoids have been used, with variable effects. Because it may improve the hyperkeratosis but also tends to increase the fragility of the skin. Other options include surgical debridement, and bath PUVA photochemotherapy were being described in the literature [4].

Conclusion

Striate palmoplantar keratoderma is a rare genodermatosis that is clinically characterized by linear hyperkeratosis on the volar aspect of the fingers with focal thickening of the palmoplantar skin. Histologic evaluation showed marked hyperkeratosis, hypergranulosis and acanthosis. The syndrome has been linked to mutations in desmoglein 1, desmoplakin, and keratin 1. However, additional novel mutations in desmosomal components and keratins might be detected in striate PPK. Treatment may include keratolytics, oral retinoids, and surgical debridement.

Conflict of Interest

The authors have no conflicts of interest that are directly relevant to the content of this case report. No sources of funding were used to assist in preparation of this manuscript.

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