RESEARCH PAPER

Inherited Diffuse Palmoplantar Keratoderma with Transgradient Extension: A Case Report

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ABSTRACT

Background: Palmoplantar keratoderma (PPK) represents a heterogeneous group of disorders characterized by abnormal thickening of the skin over the palms and soles. These disorders can be acquired or inherited, and autosomal dominant or recessive patterns are frequently used in the transmission of hereditary variants. Usually appearing early in infancy, diffuse forms can show gradual thickening and, in certain variations, transgradient extension past the palmoplantar surfaces. **Case Presentation:** We report the case of a 26-year-old male presenting with progressive thickening of the skin over both soles since early childhood. The disorder started out as minor roughness and then developed into diffuse hyperkeratosis, which became more noticeable in puberty. Prolonged standing and walking aggravated the patient's severe fissuring over weight-bearing regions, especially the heels. Blistering and related systemic abnormalities, such as hearing loss, dental problems, or abnormalities of the hair, were not seen. Similar participation was found in his father's family history, indicating autosomal dominant inheritance. Upon cutaneous inspection, the whole plantar area was affected by bilateral, symmetrical, diffuse yellowish hyperkeratosis with deep fissures and transgradient spread along the lateral boundaries. There was no pseudoainhum, and the mucosa, hair, and nails were all normal. **Conclusion:** This case highlights a classical presentation of inherited diffuse palmoplantar keratoderma with transgradient extension. Recognition of clinical features and family history is essential for diagnosis, differentiation from syndromic variants, and appropriate management. Early intervention can help reduce morbidity associated with fissuring and functional impairment.

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INTRODUCTION

Palmoplantar keratoderma (PPK) represents a heterogeneous group of dermatological disorders characterized by abnormal thickening of the stratum corneum over the palms and soles [1]. These disorders may be generally categorized into acquired and inherited types. inherited variations are caused by mutations that impact skin barrier function and epidermal keratinization [2]. Both autosomal dominant and autosomal recessive inheritance patterns are possible; autosomal dominant variants usually exhibit vertical transmission across generations [1, 3]. Based on the distribution of hyperkeratosis, PPK is classified clinically into diffuse, localized, and punctate kinds [4]. The whole palmoplantar surface thickens uniformly in diffuse PPK, which often manifests in early childhood and frequently lasts a lifetime [4]. Some variations show a "transgradient" pattern, in which hyperkeratosis affects the frontal or lateral parts of the hands and feet in addition to the palms and soles [5]. This trait helps with clinical distinction and is especially typical of some hereditary variants, such as Mal de Meleda and Greither's syndrome [5,6]. Mutations in keratin genes (KRT1, KRT9) and other structural proteins involved in epidermal integrity and differentiation are linked to the pathophysiology of hereditary PPK [2,7]. These genetic anomalies cause excessive keratin buildup and poor desquamation by interfering with keratinocyte maturation [7]. Clinically, patients exhibit waxy, yellowish, thickened skin that is prone to fissuring, particularly over weight-bearing regions, causing discomfort and functional impairment [1]. Syndromic types of PPK may occasionally be indicated by concomitant characteristics including nail dystrophy, hyperhidrosis, sensorineural hearing loss, or dental abnormalities [6,8]. Since some variations of PPK are

linked to systemic symptoms and long-term problems, it is crucial to accurately distinguish between the different forms of PPK [6]. Establishing the diagnosis requires a thorough clinical assessment, a thorough family history, and supporting tests including histopathology and genetic research [2]. We describe the clinical characteristics and diagnostic importance of a case with hereditary widespread palmoplantar keratoderma with transgradient extension.

CASE PRESENTATION

A 26-year-old male presented with progressive thickening of the skin over both soles since early childhood. The condition initially manifested within the first few years of life as mild roughness over the plantar surfaces, gradually increasing in thickness and extent. During adolescence, the hyperkeratosis worsened and continued until maturity. The patient complained of frequent periods of excruciating fissuring over the heels, which were greatly interfering with everyday tasks and were especially made worse by extended standing and walking. Blistering did not occur at the beginning of the illness or as it progressed. Alopecia, hyperhidrosis, dental anomalies, hearing loss, and other related systemic symptoms were rejected by the patient. No evidence of pseudoainhum or restrictive digital bands were present. His father's palmoplantar thickening was noteworthy in the family history, suggesting an autosomal dominant inheritance pattern. Consanguinity did not exist in the past.

Upon dermatological examination, the whole plantar area showed bilateral symmetrical diffuse yellowish hyperkeratosis. The lesions had a rough, hyperkeratotic texture and a noticeable thickening. Over weight-bearing regions, especially the heels, deep cracks were seen. The hyperkeratosis showed a transgradient pattern, extending across the plantar surfaces along the lateral margins of the feet.

There was little involvement in the dorsal portions of the feet, and there were no signs of inflammation or erythema. An examination of the teeth, hair, nails, and mucosa revealed nothing unusual. Digital constrictions and pseudoainhum were not present. Inherited frequent palmoplantar keratoderma with transgradient extension was diagnosed based on clinical symptoms, early start, progressive course, and a favorable family history.

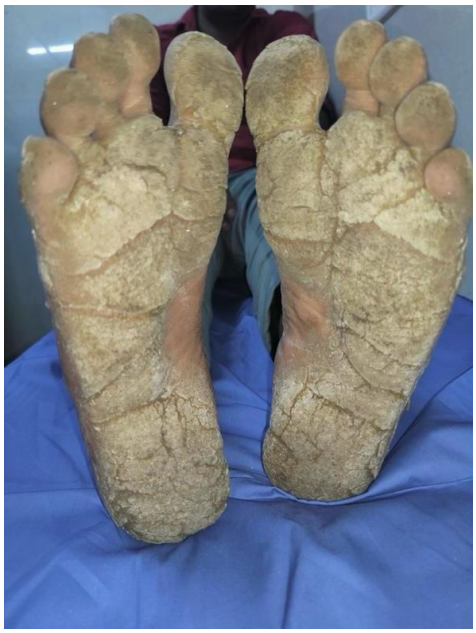

Figure 1: Diffuse plantar keratoderma with fissuring

Clinical photograph showing bilateral, symmetrical diffuse yellowish hyperkeratosis involving the entire plantar surfaces. Over the weight-bearing regions, especially the heels, there is noticeable thickening with deep cracks and noticeable scaling. The rough, hyperkeratotic texture of the lesions is indicative of hereditary widespread palmoplantar keratoderma.

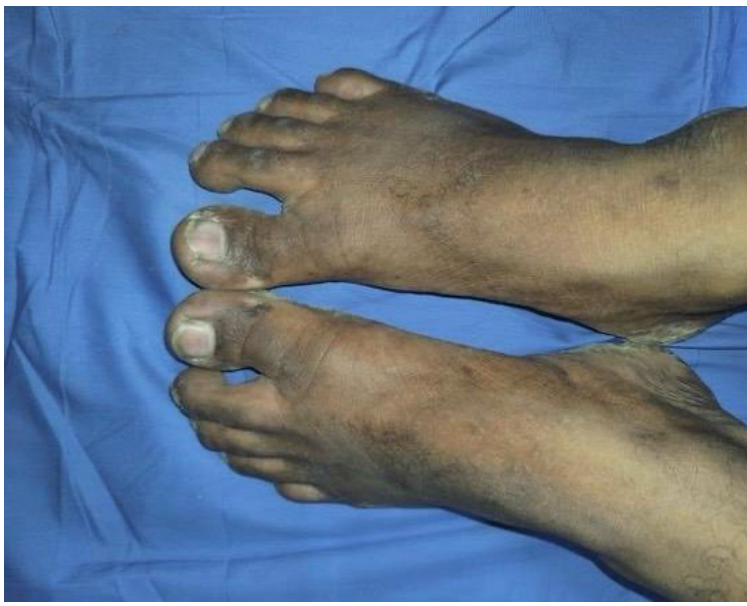

Figure 2: Transgradient extension over the dorsum of the feet

Clinical image showing extension of hyperkeratosis beyond the plantar surfaces onto the lateral and dorsal aspects of both feet (transgradient pattern). The overlying skin appears thickened with mild scaling, while the nails remain normal, supporting a non-syndromic inherited palmoplantar keratoderma.

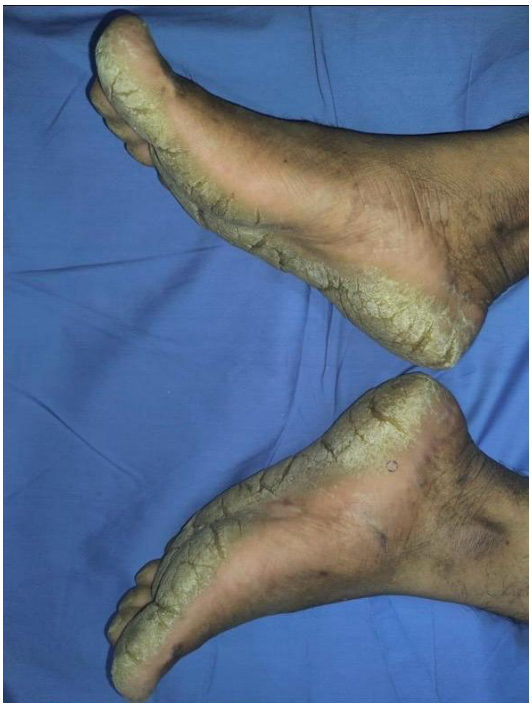

Figure 3: Lateral view showing transgradient keratoderma with fissuring

Lateral aspect of both feet demonstrating diffuse hyperkeratosis extending from the plantar surface onto the lateral borders (transgradient pattern). Inherited widespread palmoplantar keratoderma is consistent with the skin's noticeable thickening, scaling, and fissuring, particularly around the heel edges.

Histopathology

Histopathological examination in diffuse palmoplantar keratoderma typically shows marked orthokeratotic hyperkeratosis with a significantly thickened stratum corneum. The epidermis exhibits acanthosis, which indicates general thickening; hypergranulosis may also be present in some variations. Significant inflammatory infiltration is often absent. While nonepidermolytic variations exhibit compact hyperkeratosis without signs of epidermolysis, epidermolytic forms may exhibit distinctive vacuolar degeneration of the suprabasal keratinocytes.

Differential Diagnosis

The differential diagnosis of diffuse palmoplantar keratoderma includes several hereditary keratodermas. Similar to this instance, transgradient extension is a feature of Greither's syndrome, an autosomal dominant disorder. Hyperhidrosis and pseudoainhum are linked to Mal de Meleda, an autosomal recessive illness. Vohwinkel syndrome may be linked to sensorineural deafness and manifests as tightening digital bands. Pachyonychia congenita is characterized by painful keratoderma and nail dystrophy, whereas Papillon-Lefèvre syndrome is characterized by severe periodontitis and early tooth loss. Based on related characteristics, a thorough clinical assessment aids in the differentiation of various disorders.

Treatment and Follow-up

Management of palmoplantar keratoderma is primarily symptomatic and aimed at reducing hyperkeratosis and preventing complications. In topical treatment, keratolytic drugs including lactic acid, urea, and salicylic acid are used to soften and decrease thickened skin, while emollients are regularly used to relieve dryness and avoid fissuring. Oral retinoids such acitretin or isotretinoin may be used as systemic treatment in extreme situations. In order to improve patient comfort and mobility, supportive measures such as routine debridement, appropriate foot care, and the use of protective footwear are essential. In order to track therapy response and identify problems like increasing fissures or secondary infections, long-term follow-up is crucial.

DISCUSSION

Palmoplantar keratoderma (PPK) comprises a heterogeneous group of disorders with diverse clinical presentations and genetic backgrounds [2]. As seen in this instance, symmetrical thickening of the palmoplantar surfaces is a characteristic of diffuse hereditary PPK, which usually manifests in early childhood [3,4]. A genetic etiology is further supported by the progressive nature and durability into maturity.

An essential diagnostic characteristic is the existence of transgradient extension, in which hyperkeratosis spreads beyond the palms and soles onto nearby surfaces [5]. Greither's syndrome and Mal de Meleda are two examples of hereditary variations that are traditionally linked to this pattern [5,6]. Nonetheless, a non-syndromic type of diffuse PPK is favored in this patient due to the lack of concomitant symptoms such hyperhidrosis, nail dystrophy, or pseudoainhum [6,8].

An autosomal dominant inheritance pattern, which is frequently documented in various types of hereditary PPK, is suggested by the positive family history including the patient's father [1,3]. At the

molecular level, these conditions are often linked to mutations in keratin genes, especially KRT1 and KRT9, which result in aberrant epidermal integrity and keratinocyte differentiation [2,7]. Clinically, these abnormalities show up as thicker, hyperkeratotic skin due to excessive keratin buildup and inadequate desquamation. In diffuse PPK, fissuring is a serious clinical consequence, particularly over weight-bearing regions like the heels. Painful fissures caused by mechanical stress on the thicker, stiff skin can reduce mobility and quality of life [2]. To lower morbidity, early detection and proper treatment are crucial.

Other inherited keratodermas, such as Vohwinkel syndrome, Papillon-Lefèvre syndrome, and pachyonychia congenita, are included in the differential diagnosis. These conditions can be identified by related systemic or ectodermal characteristics [6,8]. In contrast, an isolated version of inherited PPK is supported in this instance by the lack of such traits. Reducing hyperkeratosis and avoiding fissures are the major goals of treatment, which is still mostly symptomatic. Although there is no known cure, systemic retinoids and topical keratolytics are the cornerstones of treatment [2]. In order to diagnose inherited PPK and differentiate it from syndromic variations, this case emphasizes the significance of thorough clinical assessment and family history.

CONCLUSION

Inherited diffuse palmoplantar keratoderma is a chronic genodermatosis that typically presents in early life with progressive, symmetrical thickening of the palmoplantar skin. The traditional characteristics of extensive plantar hyperkeratosis with transgradient expansion and a favorable family history indicative of autosomal dominant inheritance are highlighted in this instance. A non-syndromic variation is supported by the lack of related systemic or ectodermal abnormalities. Accurate diagnosis and separation from other hereditary keratodermas depend on the recognition of crucial clinical criteria, including early start, diffuse involvement, fissuring across weight-bearing regions, and transgradient dissemination. A comprehensive clinical assessment and a complete family history continue to be the cornerstones of diagnosis, even if histopathology and genetic tests may help confirm.

Reducing hyperkeratosis, avoiding fissures, and enhancing functional results are the main goals of symptomatic management. To reduce morbidity and improve quality of life, early intervention and routine follow-up are crucial. The significance of clinical knowledge in recognizing hereditary palmoplantar keratoderma and directing suitable treatment while differentiating it from syndromic variants with systemic consequences is highlighted by this case.

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