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Case Report

Punctate Palmoplantar Keratoderma: Case Report

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Abstract

Punctate Palmoplantar Keratoderma Type 1 (PPKP1) is a rare genetic disorder characterized by autosomal dominant inheritance, manifesting as punctate keratotic papules on the skin of the palms and soles. This report discusses a new case of this condition to underscore the rarity of this dermatosis. Recent research has identified mutations in the AAGAB gene as the cause of PPKP1, explaining its familial patterns. Treatment typically includes surgical excision of hyperkeratotic papules, supplemented by low-dose oral retinoids and topical applications of urea and salicylic acid to reduce symptom recurrence.

Keywords: Punctate Keratotic Papules; Dermatosis; Salicylic Acid

Introduction

Punctate Palmoplantar Keratoderma Type 1 (PPKP1) is an infrequent keratinization disorder characterized by the formation of punctate keratotic papules on the palmoplantar skin. These lesions can vary in size and may merge, particularly at pressure points such as the soles, leading to discomfort. The disorder follows an autosomal dominant inheritance pattern. This report introduces a new case of the condition.

Case Report

The patient, a 33-year-old male with an unremarkable medical history, presented to our dermatology clinic with painful hyperkeratotic papules affecting both palmoplantar areas, which he has endured since childhood. This condition has notably impacted his cosmetic appearance and caused discomfort during prolonged shoe-wearing. He indicated that his brother began experiencing similar symptoms at the age of four and that he have had genetic testing for AAGAB mutations and it was positive. Skin biopsy of the papules have noticed hyperkeratosis, hypergranulosis, acanthosis and coronoid lamella-like structure lacking the dyskeratotic and vacuolar that we can find in porokeratosis. The genetic testing for heterozygous mutations of AAGAB have been done in our genetic department and they confimed the diagnosis of PPKP1 in our case. Treatment involved manual removal of the lesions and the application of a 20% urea cream to prevent the recurrence of painful hyperkeratosis (Fig. 1).

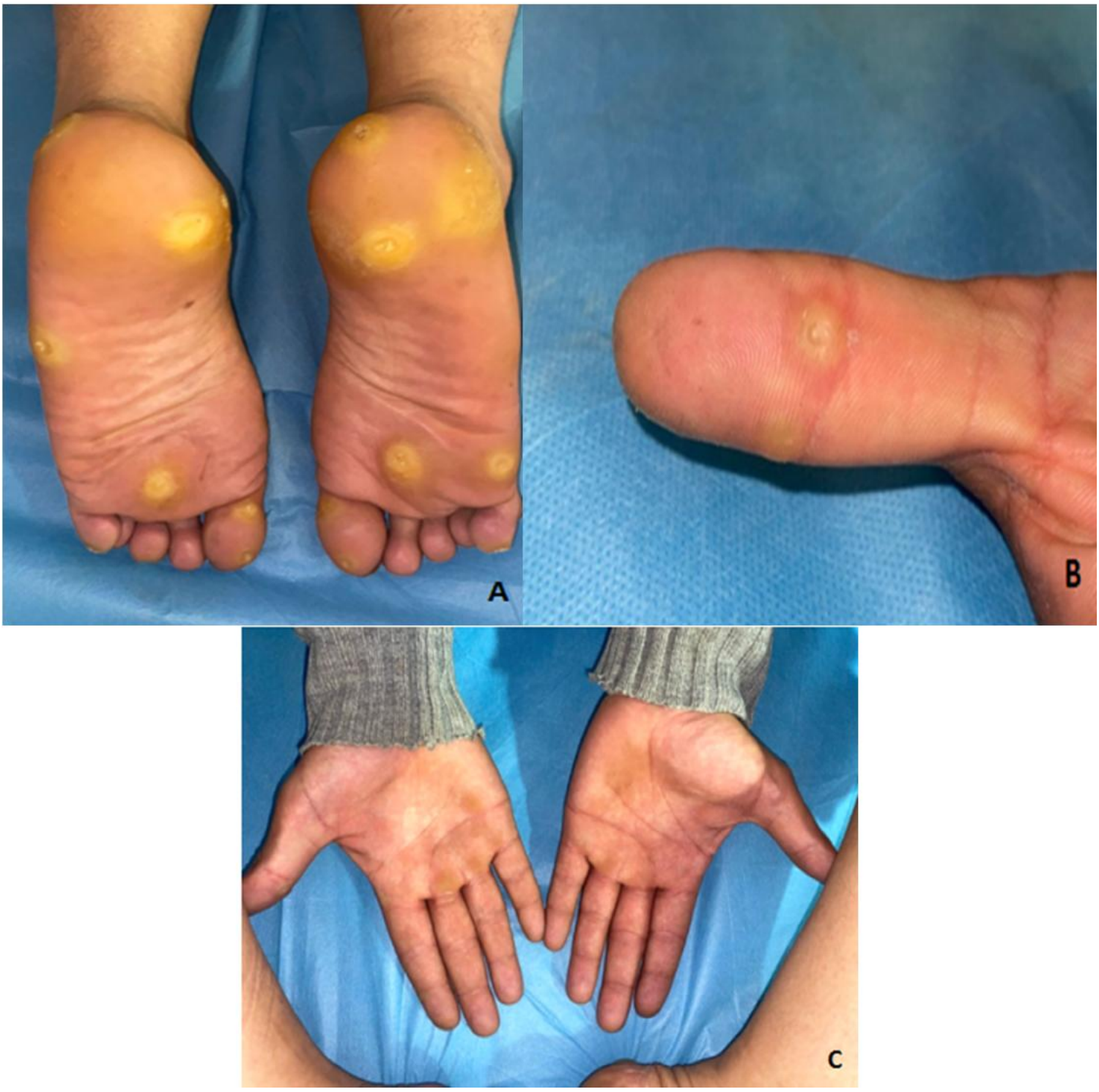

Figure 1: (A-C): Punctate palmoplantar keratoderma.

Discussion

Punctate Palmoplantar Keratoderma Type 1 (PPKP1) is a rare autosomal dominant keratinization disorder, occurring in about 1.17 per 100,000 individuals [1]. Clinical presentation typically occurs between the ages of 12 and 30, featuring multiple asymptomatic punctate hyperkeratotic papules on the palms and soles [2]. Hyperhidrosis is absent. Nail changes may include longitudinal ridging, notching, trachyonychia, onychoschizia and onychorrhexis. Associations with conditions such as ankylosing spondylitis, spastic paralysis, sebaceous hyperplasia and various malignancies have been noted [4-7]. Recent studies have linked loss-of-function mutations in the AAGAB gene to PPKP1, with one case also implicating the COL14A1 gene, associated with loci on chromosomes 8q24.13-8q24.21 and 15q22-15q24 [3]. Histopathological evaluations reveal a hyperkeratotic

epidermis without columns of parakeratosis or elastorrhexis. Electron microscopy has shown enlarged nucleoli and numerous tonofilaments in basal epidermal cells, with keratohyalin-like granules in the upper stratum spinosum [4]. Despite identifying the genetic underpinnings of PPKP1, targeted therapeutic approaches to address its pathogenesis remain elusive. Treatment options include moisturizing creams, keratolytics (such as salicylic acid, lactic acid and urea), topical retinoids, calcipotriene, topical 5-fluorouracil and oral retinoids, while surgical interventions encompass cryosurgery, mechanical debridement and excision. Topical keratolytic treatments have shown limited efficacy, whereas systemic retinoids are currently viewed as the most promising treatment for PPKP1 [5]. Patients diagnosed with PPKP1 and their relatives should undergo screening for potential associated malignancies [6]. The connection between PPKP1 and malignancy is not firmly established but some studies have reported associations between PPPK1 and certain cancers, particularly those of the gastrointestinal tract [8].

Conclusion

Punctate palmoplantar keratoderma is an uncommon genetic dermatosis characterized by painful hyperkeratotic papules that can significantly diminish quality of life. Genetic findings have identified mutations in the AAGAB and COL14A1 genes, contributing to our understanding of the condition's pathophysiology. Our patient demonstrated improvement with manual paring and 20% urea cream; however, the recurrence of symptoms upon treatment cessation profoundly affects patients. There is an urgent need for continued research, especially focused on treatment strategies, to improve outcomes for those affected.

Conflicts of Interest

The authors declare no conflict of interest in this paper.

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