Striate palmoplantar keratoderma in a 12q13.13 microdeletion syndrome

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Highlights

A patient with a known 12q13.13 microdeletion syndrome presented with striate palmoplantar keratoderma, which was suspected to be related to the deletion of *KRT1*, a type II cytokeratin gene. Recognition of this association may facilitate early diagnosis and management, ultimately improving patients' quality of life.

Introduction

Palmoplantar keratodermas (PPK) present with hyperkeratosis of the palms and soles. There are a variety of phenotypes with various genotypes and presentations.[1,2] We present a case of striate PPK in a patient with a known microdeletion syndrome that includes the keratin 1 (*KRT1*) gene.

Case Presentation

A 16-year-old man with multiple congenital anomalies, including complete agenesis of the corpus callosum, atrial septal defect, rocker bottom feet, microcephaly, hand contractures, esotropia, and development delay, presented to the dermatology clinic for evaluation of chronic palmar skin changes and recurrent boils on his buttocks. He has a known 12q13.13 microdeletion syndrome, which was previously confirmed with an array-based comparative genomic hybridization. Whole exome sequencing performed by pediatric genetics revealed a 2.05 Mb deletion at 12q13.13, identifying both a pathogenetic variant in *EVC* (c1887-5_1904del) and a variant of uncertain significance (c. 1500G>A).

The palmar skin lesions had been present for several years and were associated with significant discomfort and picking. Previous treatments, including topical steroids and vinegar soaks, were ineffective. The patient also had a history of chronic perirectal abscesses since birth, for which he was followed by pediatric infectious disease. Despite a recurrent history of ear infections, his immunologic workup showed normal immunoglobin, complement, and lymphocyte levels. He had a history of methicillin-resistant *Staphylococcus aureus* infections and had undergone multiple mupirocin decolonization cycles.

Physical examination revealed striated, linear hyperkeratotic plaques on the bilateral palmar surfaces with overlying scale extending along the length of each finger (Figure 1) consistent with striate PPK. Multiple inflammatory papules and nodules were also present in the gluteal cleft, suggestive of Hurley stage 1 hidradenitis suppurativa (HS).

For his PPK, he was started on a course of low-dose acitretin at 10 mg/day (0.25 mg/kg). Owing to the limited improvement at his 3-month follow-up appointment, the dose was increased to 25 mg/day (0.625 mg/kg). For his HS, he was started on a 3-month course of cefadroxil (1,000 mg/day) and topical 1% clindamycin solution.



Figure 1. Clinical examination of the patient. Striated, linear, hyperkeratotic plaques extending across each finger are present on both palms. Multiple finger contractures are also present, including flexion contractures at the proximal interphalangeal joints of the right fourth and fifth digits.

Discussion

The diagnosis of PPK primarily relies on the patient's clinical presentation. PPK is characterized by hyperkeratosis in 4 distinct patterns: (1) diffuse (generalized involvement), (2) punctate (hyperkeratotic papules), (3) focal (lesions on pressure points), and (4) striate (linear hyperkeratotic lesions).[3,4] Although histological findings of hyperkeratosis, acanthosis, and focal epidermolytic changes can support the clinical assessment, a biopsy is not required to make the diagnosis.[3,5,6]

Striate PPK is classified into 3 genetic subtypes, each associated with mutations in specific genes: (1) type 1 (desmoglein 1, DSG1), (2) type 2 (desmoplakin, DSP), and (3) type 3 (keratin 1, *KRT1*).[3] Notably, numerous type II cytokeratins, including *KRT1*,

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are clustered on chromosome 12q12-q13, aligning with the location of the patient's known microdeletion.[7]

Mutations in *KRT1* can result in defective keratin production, which may disrupt the intermediate filament cytoskeleton in the epidermis.[8] This structural instability can lead to a compensatory hyperkeratosis, which is observed in both PPK and epidermolytic hyperkeratosis.[3,9]

Thus, there was a strong clinical suspicion that the patient's striate PPK was related to his inability to produce functional *KRT1* proteins. PPK has been briefly reported in association with another microdeletion syndrome, involving a patient with a 15q22.32–q23 microdeletion who had a pathogenic AAGAB variant and subsequently developed punctate PPK.[10] To our knowledge, this is the first reported case of striate PPK in a patient with a 12q13.13 microdeletion syndrome. The contribution of this microdeletion to the patient's HS remains unclear.

The treatment of PPK includes topical keratolytics and emollients, with escalation to systemic retinoids in severe cases.[11] Specifically, oral retinoid dosages of 0.5–1.0 mg/kg have been recognized in being effective in treating the hyperkeratosis; however, owing to the risk of recurrence, patients may need long-term, low-dose maintenance therapy.[11] Although PPK is a benign condition, it may be associated with pain or discomfort, thereby warranting treatment to prevent significant impacts on quality of life.[4]

In patients with known chromosome mutations at 12q12-13, the clustering of type II cytokeratin genes may predispose them to hyperkeratosis disorders such as PPK. Awareness of this association can aid in more timely diagnosis and management.

Potential Conflicts of Interest: The authors declare no conflicts of interest.

Patient Consent: The authors attest that consent was obtained from the patient's legal guardian. A copy of the written consent form is retained by the authors.

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