New Lesions on the Knees With Palmoplantar Keratoderma

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A 40-year-old Latin American woman presented for evaluation with a 5-week history of redness, thickened skin, and itching of the knees, as well as persistent redness and thickened skin of the palms and soles consistent with Papillon-Lefèvre syndrome (PLS). The article discusses the clinical presentation and treatment of PLS.

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Case Report

A 40-year-old Latin American woman presented for evaluation with a 5-week history of redness, thickened skin, and itching of the knees. She also had a history of persistent redness and thickened skin of the palms and soles that started 1 week after birth. Her medical history included the loss of her permanent teeth at the age of 14 years. She reported having one sibling with similar symptoms in a family of 10 children. Her own 2 children were unaffected.

Physical examination displayed erythematous and tan thickened hyperkeratotic plaques with pitting of her palms and soles with extension to the dorsal surfaces (Figures 1 and 2). Well-circumscribed erythematous plaques were present on both knees (Figure 3). Thickening and marked dystrophy of the fingernails and toenails also were present. Oral examination revealed the presence of dentures and gingival inflammation.

The patient declined a biopsy of the affected areas. She also declined therapy with oral retinoids because of concerns regarding potential side effects. Her initial therapy involved the use of topical emollients: urea cream 40% in the morning and tazarotene gel 0.1% in the evening, with moderate improvement.

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Comment

Papillon-Lefèvre syndrome (PLS) is a rare autosomal-recessive genodermatosis characterized by keratoderma and early-onset and severe periodontitis. It initially was described by Papillon and Lefèvre in 1924.[1,2] It occurs in 1 to 4 individuals per million, equally affecting both sexes with no racial predominance.[3]

PLS is caused by a mutation in the cathepsin C (CTSC) gene on chromosome 11q14.1-14.3, encoding a cysteine-lysosomal protease known as dipeptidyl peptidase I.[4] The CTSC gene is expressed in various immune cells, including polymorphonuclear leukocytes and macrophages, and in epithelial regions on the palms, soles, knees, and keratinized oral gingiva, correlating with clinical findings.[2] In addition to the classic form of PLS, 5 cases with late-onset variation of PLS have been described.[5-8] Histopathology is nonspecific showing hyperkeratosis, occasional patches of parakeratosis, and acanthosis.[9]

Patients begin to develop indications of PLS in the first 4 years of life. Primarily, there is the development of diffuse palmoplantar keratoderma in pressure areas, which may occur focally or extend onto the dorsal surfaces of the hands and feet.[10] Periodontitis is the other major indicator of PLS.[10] Symptoms of gingival infection, abscess formation, and reabsorption of alveolar bone temporarily resolve after the loss of deciduous teeth. These features reoccur with the eruption and eventual loss of permanent teeth, necessitating dentures at a prematurely young age. The degree of periodontal infection is not related to the extent of dermatologic involvement.[11]

In a study of 47 patients with PLS, 23 patients (49%) had psoriasiform plaques on the knees and elbows. Ichthyosis was found in 2 patients (4%).[11] The 47 patients in the study developed their symptoms at an early age. Other features of PLS include palmoplantar hyperhidrosis with a foul-smelling odor and transverse grooving and fissuring of the nails.[2] Recurrent pyogenic infections of the skin can occur in approximately 20% of patients, possibly resulting from a dysfunction in

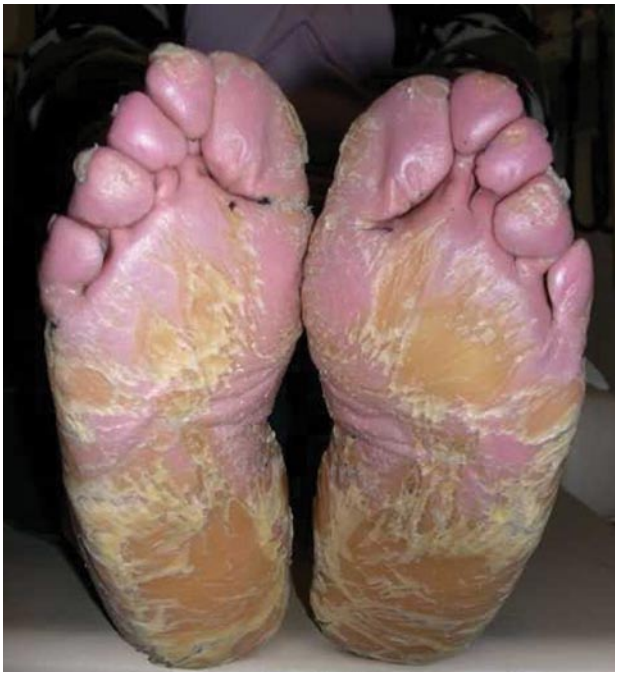

Figure 1. Bilateral hyperkeratotic tan plaques on the soles of the feet.

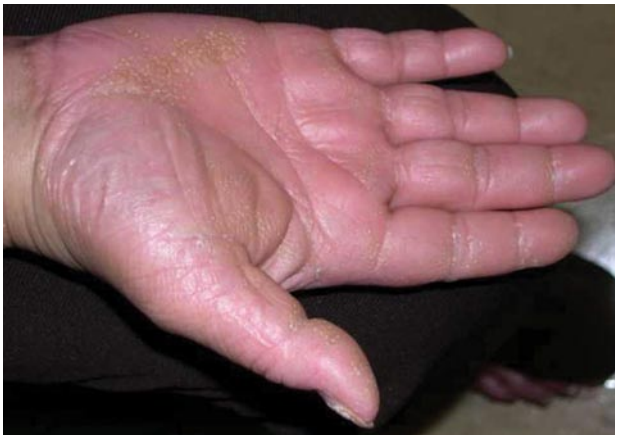

Figure 2. Hyperkeratotic plaques with pitting of the palm.

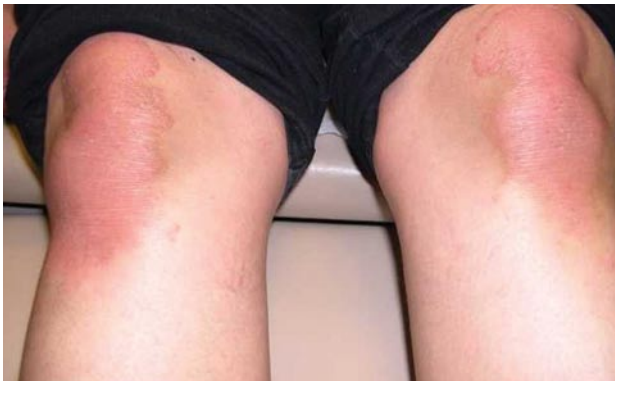

Figure 3. Bilateral, well-circumscribed, erythematous plaques on the knees.

neutrophil motility and bactericidal function.[12] Retinoids reduce hyperkeratosis and improve the keratoderma. Even though systemic retinoids are extremely useful in treating PLS, they are not curative. Their use should be balanced against short- and long-term adverse effects, especially when used in children. Additionally, cutaneous pyodermas and a tendency for liver abscess formation are recognized complications associated with this immune dysfunction. In some cases, dural calcification also has been noted.[13]

Of the palmoplantar ectodermal dysplasias, only PLS and Haim-Munk syndrome are associated with premature periodontal destruction.[2] Compared with PLS, the cutaneous findings of Haim-Munk syndrome tend to be more extensive and include clinical features such as nail deformities and arachnodactyly.[14]

Currently, oral retinoids are the mainstay treatments for both keratoderma and periodontitis in PLS.[15] Salicylic acid, urea, and emollients may enhance their effects.[12] Treatment during the development of permanent teeth is beneficial and improves keratoderma and dentition. This case represents evidence of late-onset symptom progression to the knees following early-onset palmoplantar keratoderma.

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CONTINUED ON PAGE 63

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