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Case Report of Important Oro-Facial Consequences Resulting from a Late Diagnosis of Papillon-Lefevre Syndrome

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ABSTRACT

Papillon-Lefevre syndrome is a rare genodermatosis characterized by palmoplantar keratoderma and severe destructive periodontal disease. Clinically, it is characterized by hyperkeratosis on soles of the feet and sometimes on palms of hands and includes a transgradient pattern with scattered psoriasiform lesions. Following the exfoliation of primary teeth, periodontal destruction severely affects the alveolar bone in which the permanent dentition is retained, resulting in severe atrophy of the alveolar ridges. As a result, patients developed edentulism early in adolescent years. Other symptoms may include hyperhidrosis, intracranial calcification, arachnodactyly, susceptibility to infections and mental retardation. We here in report a rare case presentation of a 15 year old male Ecuadorian patient who suffers from sharply demarcated erythematous and scaly plaques in palms, forearms, knees and low back since childhood. Furthermore, the patient was partially edentulous as evidence of aggressive periodontitis was present. As a result of a skin biopsy and dental findings, the diagnosis of Papillon-Lefevre syndrome was determined. In Ecuador, etretinate or acitretin is not available, so treatment with isotretinoin 0.5 mg/kg was given to the patient. The reported case shows a late diagnosis of Papillon-Lefevre syndrome due to similar lesions that were mistaken for psoriasis. Early intervention in Papillon-Lefevre syndrome cases is important to avoid aggressive periodontitis that can be devastating. Because of so few studies on this rare disorder, the etiology of Papillon-Lefevre syndrome is not completely understood. When palmoplantar keratoderma is observed with aggressive periodontitis, one should always include Papillon-Lefevre syndrome as a differential diagnosis. We want to report this case in order to alert dermatologists and dentist to watch for these signs and assist the proper diagnosis and treatment of this rare syndrome.

Keywords: Case report, Palmoplantar keratoderma, Papillon-Lefevre, Periodontitis, Tooth

INTRODUCTION

Papillon-Lefevre syndrome (PLS) is a rare, autosomal recessive genodermatosis characterized by palmoplantar keratoderma (PPK) and severe destructive periodontal disease [1]. It develops within the first years of life with signs of transgradient hyperkeratosis of palms and/or soles. The affected gene is the CTSC gene that encodes cathepsin C, which plays an important role in the inflammatory process and in epithelial differentiation [2,3].

CASE REPORT

A 15 year old Ecuadorian patient with a negative family history of skin diseases came to our clinic complaining about skin thickening on soles since he was 6 months old. The dermatosis progressed with sharply demarcated erythematous and scaly plaques in palms, forearms, knees and low back when he was 6 years old. Physical examination showed erythema, fissuring and severe diffuse thickening of skin on the palms and soles with paronychia. The lesions extended dorsally to the extensor surfaces of the hands and feet, and over the elbows, Achilles tendons, and knees with a transgradient pattern. A scaly lesion was noted on the intergluteal cleft, as well **(Figure 1)**. The patient was partially edentulous, but inflammatory signs were not found in the gums **(Figure 2a)**.

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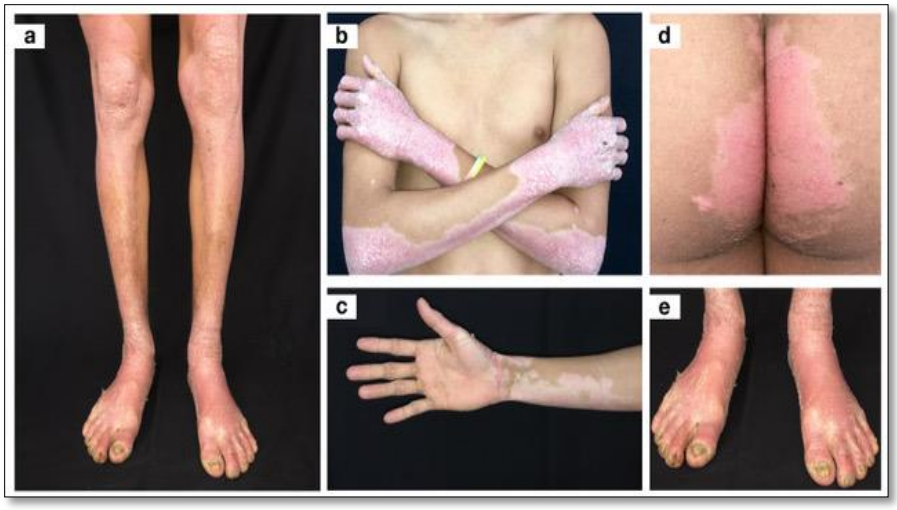

Figure 1. Extension of erythematous and scaly plaques to the dorsal surfaces of the hands and feet and over the elbows, achilles tendons and knees showing a transgradient pattern. The patient shows erythema, fissuring and diffuse thickening of skin on the palms, and on the soles with paronychia. Observe a psoriasiform lesion on the intergluteal cleft.

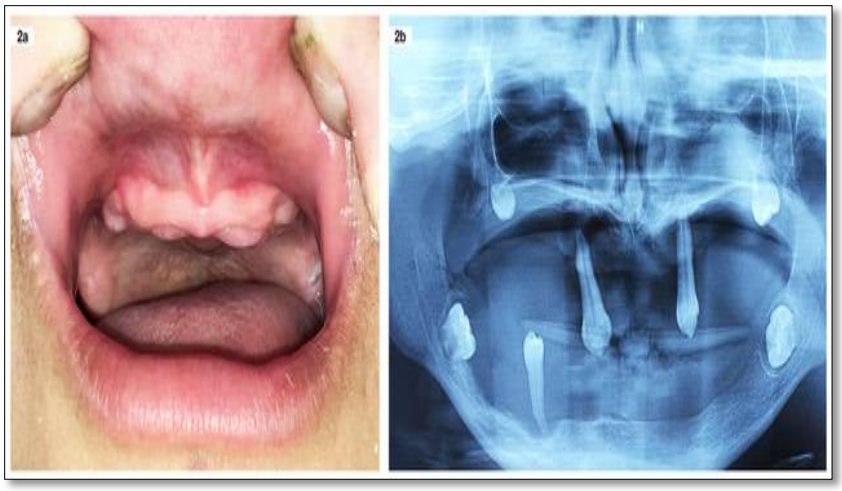

Figure 2. Oral cavity without teeth, just a molar in the back, with no inflammatory signs in the gums. Panoramic Dental X-Ray showed 3 remaining incisors teeth and 4 molars; bone loss in the mandibular arch and resorption of alveolar bone of maxilla.

The anatomopathological examination of the skin biopsy revealed lamellar hyperkeratosis, parakeratosis and focal areas of spongiosis. A panoramic dental x-ray showed 3 remaining incisors teeth and 4 molars, important bone loss in the mandibular arch and resorption of alveolar bone of maxilla **(Figure 2b)**. He was diagnosed with PLS so isotretinoin 0.5 mg/kg was prescribed for the patient. The patient is at high risk of maxilar fracture and he is a candidate for bone graft.

DISCUSSION

The Papillon-Lefevre syndrome (PLS) was first described in 1924 by Papillon and Lefevre [1,3]. This is a very rare, autosomal recessive disorder [4]. Parental consanguinity is demonstrated in 20% to 40% of the cases [2]. The patient presented above does not have consanguineous parents and also he does not have other affected sibling, altogether suggesting that the variable phenotypes of PLS can be related to the other genetic or epigenetic/environmental factors [5].

The PLS locus has been mapped to chromosome 11q14-q21. The mutation of CTSC gene, in one or both alleles, leads to the enzymatic dysfunction of Cathepsin C. This enzyme has an essential role in the activation of granule serine proteases and in epithelial differentiation. Decreased neutrophil phagocytosis and chemotaxis of leukocytes can cause the patients to be more susceptible to the infections [2,3].

Clinically, it is characterized by hyperkeratosis more on soles than on palms, with transgradient pattern and

sometimes with scattered psoriasiform lesions [2,5]. In the case reported, there was a delayed in the diagnosis because the lesions were mistaken for psoriasis. Dental alterations usually have an early-onset. The development of the primary teeth proceeds normally, but the eruption of them into the oral cavity is associated with gingival inflammation and destructive periodontitis that is unresponsive to traditional periodontal treatments. Following the exfoliation of primary teeth, periodontal destruction continues to severely affect to the alveolar bone with permanent dentition, resulting in generalized atrophy of the alveolar ridges [3]. As a result, patients developed edentulism early in teenage years. Additionally, liver abscesses, pyogenic skin infections, elevated IgE levels and cranial calcifications can be find in these patients [3,5].

The diagnosis is clinical and histopathological exam is unspecific [4]. A collaborative approach involving the dermatologist and pediatric dentist is important. Good dental care and the use of prophylactic antibiotics aim to minimize periodontitis and the loss of teeth. It has been proposed that extracting permanent teeth with periodontal disease may be a strategy to prevent the atrophy of alveolar bone [3]. Treatment of skin lesions in PLS is difficult because of recurrences. Response to topical therapy has not been satisfactory. The use of oral retinoids has been reported to be effective in patients with PLS [4] and it would diminish the inflammatory complications of the gums if it is started early [4,6,7]. In Ecuador, etretinate or acitretin is not available, thus, isotretinoin was used for our patient.

Early intervention in PLS cases is important to avoid aggressive periodontitis that can be catastrophic due to atrophy of the alveolar bone. In addition, these children may also suffer from recurrent infections. PLS lesions are very responsive to isotretinoin. Given PKK with periodontitis, one should always include PLS as a differential diagnosis.

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