

## CASE REPORT

# PAPILLON-LEFÈVRE SYNDROME

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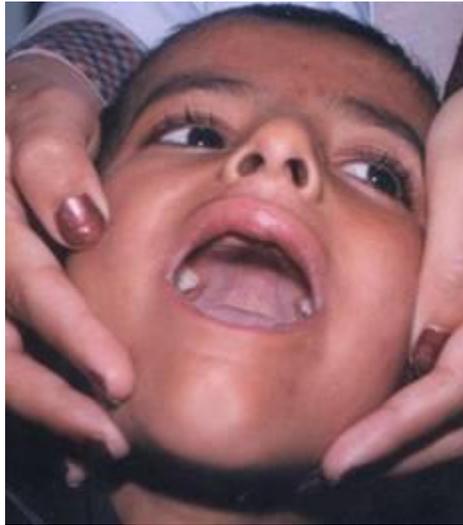
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A 9 years old male presented with keratotic plaques over the skin of his palms and soles extending onto the dorsal surface and swollen gums since the age of 4 with subsequent loss of most of his permanent dentition. These findings are consistent with Papillon-Lefèvre syndrome.

**Key words:** Papillon-Lefevre syndrome (PLS), palmar-plantar keratoderma, periodontitis, premature teeth loss

## INTRODUCTION

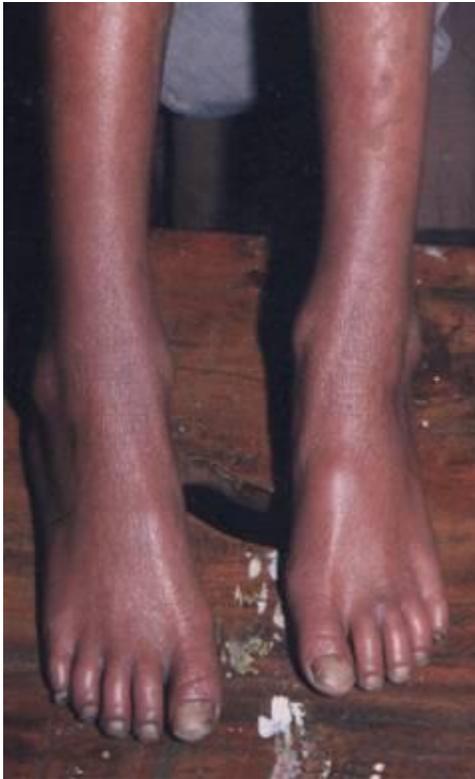
Papillon-Lefèvre syndrome (PLS) is a rare autosomal recessive disorder characterized by hyperkeratosis of the palms and soles and severe destructive periodontal disease affecting both the primary and permanent teeth. This case report describes a case of PLS classic clinical features and brief review the relevant literature.



**Figure-1: Absence of upper and lower incisor teeth.**



**Figure-2: Yellow colored hyperkeratotic areas on soles**



**Figure 3**



**Figure-4**



**Figure 5**

A six year old male child born of consanguineous parents presented to us with redness and peeling off of skin of hands and feet, cough, redness and discharge from eyes and ear, pain for last 20 days. He had exacerbations and remissions of the skin lesions and multiple infections since early childhood. He had normal outcome at birth and had normal developmental milestones till age of three. During 3<sup>rd</sup> year of his life he started developing fissures in skin of palms and soles that resulted in peeling off of skin leaving red thin skin underneath. He repeatedly contracted systemic infections. He had cough for last one month that was dry initially and later on became productive. He used to get bouts of cough and diagnosed as having pertussis. He complained of earache that was diagnosed as otitis media. Similarly he had bilateral purulent conjunctivitis. On examination of oral cavity his upper incisors, upper lower canines, upper and lower molars were absent (Figure 1). His deciduous teeth started falling off at age of three years and only his lower incisors erupted again that too became loose. Skin of both palms and soles was peeling off and underlying skin was red and shiny suggestive of keratoderma (figures 2,3,4,5). The fingers were pointed and gave a clawed appearance. On eye examination he had bilateral congested conjunctivae with purulent discharge. Ear examination revealed bulging red tympanic membrane suggestive of bilateral acute otitis media. On chest auscultation he had bilateral rhonchi. Rest of the systemic examination was normal. On family history there were no similar disorders in family.

Routine hematological examination revealed Hb% of 10.0 g/dl, total leukocyte count of 9200 and ESR was 20 mm/hour. Urine routine examination showed 15-20 pus cells and 3-4 red blood cells suggestive of urinary tract infection. Rest of the biochemical investigations were within normal limits.

## **DISCUSSION**

Papillon-Lefèvre syndrome, or keratosis palmoplantaris with periodontopathia (PLS, MIM 245000), is inherited as an autosomal recessive trait, affecting children between the ages 1-4 years.<sup>1,2</sup> It has a prevalence of 1-4 cases per million persons.<sup>2</sup> Males and females are equally affected and there is no racial predominance.<sup>3</sup> PLS is characterized by palmoplantar keratodermas, psoriasiform plaques of the elbows and knees, periodontal disease with resultant

premature loss of deciduous and permanent teeth, and intracranial calcifications.<sup>4</sup> These keratotic plaques may occur focally, but more often involve the entire surface of the palms and soles.<sup>5</sup> Palmoplantar keratosis, varying from mild psoriasiform scaly skin to overt hyperkeratosis, typically develops within the first three years of life.<sup>6</sup> Often, they are associated hyperhidrosis of the palms and soles resulting in a foul-smelling odor.<sup>2</sup> The findings may worsen in winter and be associated with painful fissures.<sup>7</sup> The development and eruption of the deciduous teeth proceeds normally, but their eruption is associated with gingival inflammation and subsequent rapid destruction of the periodontium. The resulting periodontitis characteristically is unresponsive to traditional periodontal treatment modalities and the primary dentition is usually exfoliated prematurely by age 4 years. After exfoliation, the inflammation subsides and the gingiva appears healthy. However, with the eruption of the permanent dentition the process of gingivitis and periodontitis is usually repeated and there is subsequent premature exfoliation of the permanent teeth, although the third molars are sometimes spared.<sup>8,9</sup> The etiology of periodontal disease was explained with juvenile periodontitis due to infection with *Actinobacillus actinomycetemcomitans* rather than immunologic dysfunction or anatomic defects.<sup>5</sup> Both the deciduous and permanent dentitions are affected; resulting in premature tooth loss. Most PLS patients display both periodontitis and hyperkeratosis. Some patients have only palmoplantar keratosis or periodontitis, and in rare individuals the periodontitis is mild and of late onset.<sup>6</sup> In addition to the skin and oral findings, patients may have decreased neutrophil, lymphocyte or monocyte functions and an increased susceptibility to bacteria, associated with recurrent pyogenic infections of the skin.<sup>10</sup>

Histologically, the skin lesions of PLS have not been well-characterized in the literature. Reported findings have consisted of hyperkeratosis, occasional patches of parakeratosis, acanthosis, and as light perivascular inflammatory infiltrate<sup>4</sup>.

The PLS locus has been mapped to chromosome 11q14–q21 with an almost total loss of cathepsin C activity in PLS patients and reduced activity in obligate carriers<sup>11-13</sup>. The cathepsin C gene encodes a cysteine-lysosomal protease also known as dipeptidyl-peptidase I, which functions to remove dipeptides from the amino terminus of the protein substrate. It also has endopeptidase activity. The cathepsin-C gene is expressed in epithelial regions commonly affected by PLS such as palms, soles, knees, and keratinized oral gingiva. It is also expressed at high levels in various immune cells including polymorphonuclear leukocytes, macrophages, and their precursors.<sup>14,15</sup> All PLS patients are homozygous for the same cathepsin-C mutations inherited from a common ancestor. Parents and siblings, heterozygous for cathepsin C mutations do not show either the palmoplantar hyperkeratosis or severe early onset periodontitis characteristic of PLS<sup>8</sup>.

A multidisciplinary approach is important for the care of patients with PLS. The skin manifestations of PPK are usually treated with emollients<sup>7</sup>. Oral retinoids such as acitretin, etretinate, and isotretinoin were reported to be beneficial for both dental and cutaneous lesions of PLS. Retinoid treatment may end up with normal dental development if started during eruption of permanent teeth.<sup>16</sup> The periodontitis in PLS is usually difficult to control. Effective treatment for the periodontitis includes extraction of the primary teeth combined with oral antibiotics and professional teeth cleaning. It is reported that etretinate and acitretin modulate the course of periodontitis and preserve the teeth. A course of antibiotics should be tried to control the active periodontitis in an effort to preserve the teeth and to prevent bacteremia and subsequently pyogenic liver abscess.<sup>17</sup>

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