Papillon–Lefevre Syndrome: A Case Report with Review of Literature
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ABSTRACT
Papillon–Lefevre Syndrome (PLS) is a very rare syndrome of autosomal recessive inheritance characterised by palmar plantar hyperkeratosis and early onset of a severe destructive periodontitis, leading to premature loss of both primary and permanent dentitions. The palmar plantar keratoderma typically has its onset between the ages of 1 and 4 years and severe periodontitis starts at the age of 3 or 4 years. The exact pathogenesis of these clinical events remains mainly speculative. An early diagnosis of the syndrome can help preserve the teeth by early institution of treatment, using a multidisciplinary approach. We report a case of 17-year-old female patient with PLS.

Keywords: Autosomal recessive disorder, Palmoplantar keratosis, Periodontitis, Premature teeth loss, Keratoderma.

INTRODUCTION
PLS, first described by two French physicians Papillon and Lefevre in 1924,[1] is an extremely rare genodermatosis inherited as an autosomal recessive trait, affecting children between the ages of 1–4 years[2,3]. It is characterized by severe destruction of alveolar bone involving both the deciduous and permanent dentition. Inflammatory gingival enlargement, gingival ulcerations and formation of deep pockets are frequently present. The characteristic skin lesions associated with the changes consist of keratotic lesions of the palmar and plantar surfaces[4]. Well-demarcated psoriasis form plaques occur on the elbows and knees. Often, there is associated hyperhidrosis of the palms and soles resulting in a foul-smelling odour. Another feature of PLS may be radiographic evidence of intracranial calcification[5].

CASE REPORT
A 17-year-old female patient presented with the chief complaint of exfoliation of teeth since 4–5 years. History revealed that her deciduous teeth had erupted normally but exfoliated gradually by the age of 4–5 years. Similarly, her permanent teeth too were exfoliated prematurely after erupting normally. Family history reveals that the patient’s brother is also suffering from the same condition. She also gives history of the parents’ consanguinity. The patient was conscious, well-versed with time, place and person. Gait was
normal, build was thin and height was 5’3”. On skin examination hyperkeratotic lesions were seen on palms and plantar surfaces of hands with dryness of skin. The lesions became worse during winters. The skin was dry and rough on palpation keratotic plaques were also seen on the dorsal surface of feet (Fig.2). Nails appeared dysplastic. Intra oral examination revealed multiple missing teeth and the overlying mucosa was normal on the edentulous ridges (Fig. 3). The report was compatible with PFS.

DISCUSSION

PLS is an uncommon autosomal recessive type-IV palmoplantar ectodermal dysplasia[1]. It usually manifests itself between the ages of 6 months to 4 years, coinciding with the eruption of primary teeth[6]. The two cardinal diagnostic features of the syndrome are palmoplantar keratosis and an early onset form of aggressive periodontitis.

The palmoplantar keratoderma typically has its onset between the ages 1 and 4 years. The sharply demarcated
erythematous keratotic plaques may occur focally, but usually involve the entire surface of the palms and soles, sometimes extending onto the dorsal surfaces of the hands and feet. Often, there is associated hyperhidrosis of the palms and soles resulting in a foul-smelling odour. In addition, psoriasiform plaques may be seen on the elbows and knees. The findings may worsen in winter and be associated with painful fissures. Other sites that may be affected include the eyelids, labial commissures, legs, thighs and axillae. The hair is usually normal, but the nails in advanced cases may show transverse grooving and fissuring[1,7].

The major feature of PLS is severe periodontitis, which starts at the age of 3 or 4 years. The developmental 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 the age of 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[1]. The degree of dermatologic involvement may not be related to the level of periodontal infection[8].

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 in approximately 25% of PLS patients[6]. Pyogenic liver abscess is increasingly recognized as a complication of PLS associated with impairment of the immune system[9].

Another component of PLS may be radiographic evidence of intracranial calcification in choroid plexus and tentorium. Although this has been taken as cardinal feature, being inconsistent it is not considered important for the diagnosis. Histopathological examination reveals non-specific hyperkeratosis, acanthosis, focal parakeratosis, psoriasiform hyperplasia, torturous capillaries in dermal papillae and superficial lymphocytic infiltration[7,10].

The cause of PLS is not well understood, but recently, two research groups have reported that loss of function mutations affecting both the alleles of the cathepsin-C gene, located on chromosome 11q14.1–q14.3, were associated with PLS[11]. 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[12]. The exact cause of periodontal disease in PLS has not been found but it has been attributed to decrease neutrophil phagocytosis bacterial infection and impaired reactivity to T and B cell mitogens. The exact mechanism of the increased susceptibility to infections is also unknown, but some investigators have demonstrated a dysfunction in neutrophil motility and bactericidal function[1].

A multidisciplinary approach is important for the care of patients with PLS. The skin manifestations of PLS are usually treated with emollients. Salicylic acid and urea may be added to enhance their affects. Oral retinoids including acitretin, etretinate and isotretinoin are the mainstay of the treatment of both the keratoderma and periodontitis associated with PLS. Treatment may be beneficial if it is started during the eruption and maintained during the development of the permanent teeth. 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. A course of antibiotics should be tried to prevent bacteremia and subsequent pyogenic liver abscess. The risk of pyogenic liver abscess should be kept in mind in evaluating these patients when they present fever with unknown origin[1].

**CONCLUSION**

PLS can adversely affect growing children psychologically, socially and aesthetically; therefore awareness of this syndrome is essential. Early
dental evaluation and parental counselling as a part of preventive dental treatment is essential, a multidisciplinary approach may improve the prognosis and quality of life of these children.

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REFERENCES