Papillon Lefevre Syndrome – A Case Report
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Abstract
Papillon Lefevre Syndrome (PLS) is a rare autosomal recessive disorder of palmoplantar keratoderma (PPK) and premature loss of both dentitions. The exact pathomechanism of these clinical events mainly remains speculative. Early recognition of this entity as well as multi-disciplinary management may help in the prognosis of these cases. This paper presents two cases of the PLS syndrome among siblings.

Key Words: Papillon lefevre syndrome; Palmoplantar keratoderma; peridontosis; premature teeth loss

Introduction
Papillion Lefevre Syndrome (PLS) is a genetic disorder that typically apparent from approximately 1-5 years of age.(1,2) It is characterized by Palmoplantar hyperkeratosis and peridontopathy.(1-5) It was first described by two French physicians Papillon and Lefevre in 1924.(3) Genetic analysis suggests that disorder may result from changes (mutation) of a gene that regulates production of enzymes known as cathepsin C. An increased prevalence of parental consanguinity has been reported in PLS patients.(2) All PLS are homozygous for the same cathepsin C mutation inherited from a common ancestor. This paper presents two cases of the PLS syndrome among siblings.

Case report
Case 1
This 19-year-old girl presented with palmoplantar hyperkerotosis since the age of four years and total loss of teeth by the age of 4 years. She was the first child born to apparently healthy -consanguineous parents after an uneventful pregnancy and birth. Her younger sibling was reportedly normal. 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 lost prematurely after erupting normally (Figure 1).

Figure 1: Edentulous due to early exfoliation of permanent teeth, Figure 2: Palmer and planter keratosis extending on dorsal surface of feet, Figure 3: Planter keratosis with deep fissuring on soles

There was history of recurrent swelling of gums and foul breath followed by loosening and exfoliation of teeth. At the age of four years, her parents also noticed a progressive thickening of palmoplantar skin. It was associated with marked aggravation of erythema, scaling and dryness during the eruption of teeth, and had improved after complete exfoliation of dentition. On clinical examination, the patient has keratinization of palmer and planter surface with spill over onto dorsal surface of hands and feet. The knees and elbows were also affected in case1. (Fig 2) Deep fissures were present on the soles of both the cases. (Figure-3)

Case 2
This 18-year-old girl had a history of pyogenic abscess and had been treated for the same. In both the cases nails and hairs were normal. On intra oral examination all the teeth were missing in both the cases, however 28 and 48 found erupting in case 2. The gingiva around the teeth was inflamed swollen and tender while edentulous area appeared to be normal.(Fig-5) Complete blood count, blood chemistry profiles were within normal limits. Panoramic radiograph obtained which revealed generalized horizontal alveolar bone loss.

Figure 4: erupting 3rd molar with inflamed surrounding gingiva
Figure 5: Planter keratosis extending on dorsal surface of feet
Figure 6:sharply demarcated keratotic plaques on both the knees

Dermatological consultation were sought and diagnosis of PLS established. Keratolytic preparations containing 20% salicylic acid were prescribed for skin lesions. Advised complete denture fabrications to restore masticatory function. In view of the above findings, the cases were diagnosed as Papillon Lefevre syndrome (PLS).

Discussion
PLS is a disorder of keratinization that is inherited as autosomal recessive trait. A major gene locus for PLS has been mapped on chromosome11q14 and inheritance of mutation of the cathepsin C gene is found in homozygote’s of PLS.(1-6) Usually parents and siblings...
heterozygous for cathepsin C mutation do not show either palmoplantar hyperkeratosis or severe early onset of periodontitis characteristic of PLS.(6,7)

The severe periodontitis starts at the age of 3 or 4 years. The development and eruption of deciduous teeth proceeds normally, but their eruption is associated with severe gingival inflammation in the absence of local etiological factors.(8) The periodontal pockets rapidly deepen with severe loss of alveolar bone and marked fetor odor. Primary dentition usually exfoliated prematurely by the age of 4 years and even there is premature exfoliation of permanent teeth by the age of 13 to 16 years.(9) Severe resorption of alveolar bone gives the teeth a “floating in air “appearance on dental radiograph.(10-14)

The palmo-plantar keratoderma (PPK) typically has its onset between age of 1 and 4 years.(12) Sharply demarcated erythematous keratotic plaques involve the entire surface of the palms and soles which sometimes extending on dorsal surface of hands and feet. Often, there is associated hyperhidrosis of the palms and soles resulting in foul odor. These well demarcated psoriasiform plaques occur on elbows and knees. These findings may worsen in winter.(13) The degree of dermatologic involvement may not be related to periodontal infection. Nail changes are apparent in advanced cases manifested by transverse grooving and fissuring.(10) A multidisciplinary approach is necessary in the management of patients with PLS.(14) Our patients showed these classical events of gingivitis, periodontitis and precocious loss of deciduous as well as permanent teeth at the age of 15 years.

Clinically both the patients had the characteristics skin lesion, sharply demarcated keratotic plaques involving entire surface of palm and soles. Case 1 had typical palmoplantar keratoderma and showed tendency towards improvement after exfoliation of all the teeth. Retinoids treatment may end up with normal dental development if started during eruption of permanent teeth. Periodontitis is usually difficult to control. It is reported that etretinate and acitretin modulates the course of periodontitis and preserve the teeth. Nails may show transverse grooving, claw like phalanges with arachnodactaly and osteolysis described in PLS.

In case 2, patient had history of pyogenic abscess. Systemic retinoid have proven to be effective in PPK of PLS as well as other PPKs. The concern that retinoid treatment in PLS may increase the risk of pyogenic abscess. Bacteremia resulting from periodontitis coupled with neutrophils dysfunction appears to be responsible for the development of liver abscess. (13,14) In case 2 history of pyogenic abscess was present and treatment was given. A course of antibiotics should be tried to control the active periodontitis and to prevent bacteremia and subsequent pyogenic liver abscesses.(13,14)

**Conclusion**

PLS can adversely affect the growing children psychologically, socially and esthetically hence early dental treatment and genetic counseling may be benefit for affected individual and their families as other treatment is symptomatic and supportive. Early recognition of this entity as well as multidisciplinary management may help in the prognosis of these cases. Research on genetic disorder and their causes is ongoing. It is hoped that this new knowledge will lead to prevention and treatment of genetic and familial disorder in the future.

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