Papillon-Lefevre syndrome: A Rare Case Report

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Abstract:

Papillon-Lefevre syndrome is an extremely rare autosomal recessive genetic disorder characterized by hyperkeratosis of palms soles and premature loss of both deciduous and permanent teeth. Recent theories suggest that this condition is linked to a mutation of the Cathepsin-C gene. Here present two cases of the Papillon-Lefevre syndrome from two distinct families, both of which exhibit all the recognizable symptoms. Both patients presented with persistent thickening of the skin of palms and soles. Severe generalized periodontal destruction and aggressive periodontitis with mobility of teeth showed as intraoral features and orthopantomography showed the generalized alveolar and arc-shaped bone loss in both patients.

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Introduction: Papillon Lefevre syndrome is a rare autosomal recessive disorder characterized by hyperkeratosis of palms; soles and also severe destructive periodontal disease affecting both primary and permanent teeth. [1-4]

Two doctors from France Papillon and Lefevre documented a brother and sister who had a disorder known as palmoplantar hyperkeratosis associated with severe early-onset periodontitis in 1924. [2;5;6-8]

The etiology of PLS is multifactorial with genetic immunological and microbial factors all playing an important role in the etiopathogenesis of this syndrome. [2-4]Although the PLS are not well understood and the exact etiology is still unknown. [4] It has been reported that mutation of the cathepsin-c gene (CTSC) located on chromosome 11q14-q21 is responsible for PLS syndrome. [2,3,7] The cathepsin-c gene is primarily expressed in the epithelial region such as palms, soles, knees, fingers and keratinized gingiva. These are the most commonly affected areas by PLS. [3,4,6]

The PLS syndrome typically has its onset between the ages of 1 to 4 years. [1-2,4]Its prevalence is estimated to be 1 to 4 per million in the general population. [2,4,6-7] Consanguineous marriage has been linked to an increased expression of the autosomal recessive disorder in 20 to 40% of PLS patients. [2,4,6-7]

There is no racial predominance and no difference in the effects on men and women. [2,4] Sometimes extending on the dorsal surface of hands and feet. The symptoms may worsen in winter and also be associated with painful fissures. [1-2,6]

Oral manifestation involves extensive severe aggressive periodontitis leading to painful destruction of alveolar bone involving drifting and mobility and exfoliation of both deciduous and permanent teeth without any signs of root resorption. Firstly, gingiva gets inflamed with the eruption of primary teeth, but it appears standard after exfoliation of primary dentition. [4,8] Both maxillary and mandibular ridges are highly resorbed, leading to reduced vertical dimensions. [2] Premature exfoliation of permanent teeth by

age of 13-16 years. Gorlin et al have added the third feature of Dura calcification. [3,8-9]

CASE REPORT (CASE-1): A 17-year-old Indian male patient reported to the Department of Oral Medicine and Radiology, Faculty of Dental Science DDU, Nadiad with a chief complaint of loosening of teeth in the upper right front region & lower right back region for 1 week & discomfort while chewing food. The patient had no relevant medical history.

On General Examination, the patient was moderately built with a steady gait. Cognitive development was also normal. On cutaneous examination of the palms, the palmar surface of both hands showed dryness & keratosis with variable macules. The patient also gives H/O of cracked palms. Examination of the plantar surface shows mild keratosis. Elbows & knees were unaffected. Hair & nails were normal.

Past Dental History: The guardian informed that the deciduous teeth had erupted normally and complete shedding of all deciduous teeth by the age of 6 years was noted followed by normal eruption of permanent teeth.

Intraoral Examination: Upon intraoral assessment, the teeth that showed Grade I mobility were 14,11,24,25,31,41, and 45. Whereas the ones with grade II mobility were in 12 and 15 along with grade III mobility in 13 and 47. In addition, the guardian gave H/O of the child having masochistic activity of using force to remove the tooth, and having Grade III mobility 13, for aesthetic concern. The overall hygiene of the mouth was poor with heavy deposits of plaque and halitosis was also present with deep periodontal pockets & bleeding on probing. Gingiva around the teeth 12, 13, and 14 appeared erythematous and swollen showing suppuration. Grade IV recession of gingiva was observed around 47.

Family history does not reveal consanguineous marriage and other siblings were not affected. Intraoral periapical radiograph of right maxillary Canine shows radiolucency around the apical one-third of root with little bone support and periapical x-ray view of maxillary right 1st and 2nd premolar shows vertical bone loss upto an apical third of the root.

Panoramic radiograph illustrated arc-like bone loss around the mandibular right 2nd molar. The tooth appears to be floating with generalized horizontal bone loss.

Routine hematological examination revealed the following: Hb% = 9.40, Total RBC = 3.41 mil/cm, PCV = 31.80, MCHC = 29.55, RDW = 20.90, Neutrophils = 51%, LFT [SGPT] = 45 Based on clinical findings and radiographic evaluation, the patient was diagnosed with Papillon-Lefevre Syndrome. The patient was referred to the Department of Periodontics for needful treatment.

CASE REPORT (CASE-2): A 17-year-old female patient reported to the department of Oral Medicine and Radiology, Faculty of Dental Science, DDU, Nadiad 4 years ago presenting a chief complaint of mobility of teeth of the upper and lower right and left back tooth region for the last 3 years and was diagnosed with PapillonLefevre Syndrome also started the treatment but due to pandemic of COVID-19 virus and lockdown she was not able to continue the treatment. Then she came after 4 years seeking another treatment.

Medical history: Not relevant

General Examination / Extra oral examination: Upon general examination, the patient was moderately built with a steady gait. Cognitive development was also normal. On cutaneous examination of palms and palmer surface of both the hands shows dryness and keratosis. The patient gave a history of skin layer peeling off the palm and foot

Intraoral examination: In 2018 intraoral examination showed that all the teeth were mobile. It indicated generalized aggressive periodontitis. At present, (2022) all teeth are fully erupted along with the 3rd molars are present and firm.

Panoramic radiograph showed several floating teeth with severe generalized horizontal bone loss and arc-shaped bone loss at the lower left and right first molar region.

Routine hematological examination (CBC and blood chemistry profile) and LFT (liver function test) was normal.

Based on clinical findings and radiographic evaluation, the patient was diagnosed with Papillon Lefevre Syndrome

and referred to the Department of Periodontics for needful treatment.

Fig-1 Showing clinical findings of case-1 [A] Generalised Inflammation of gingiva [B] Pathologic migration of central incisor because of aggressive periodontitis [C] Dyskeratotic lesion on palmer surface of hands

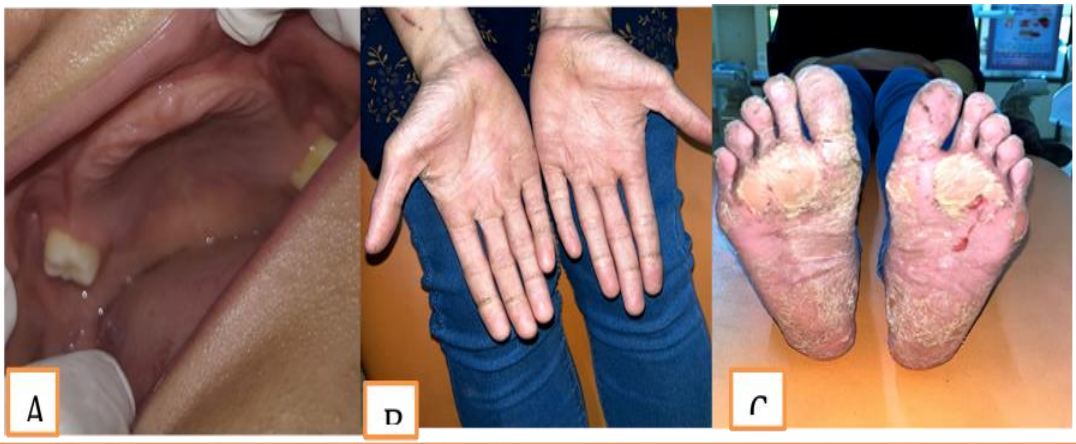

Fig 2 showing clinical findings of case-2 [A] Intra oral findings- exfoliation of multiple permanent teeth [B &C] dyskeratotic lesion present on palmer and planter surface of hands and feet

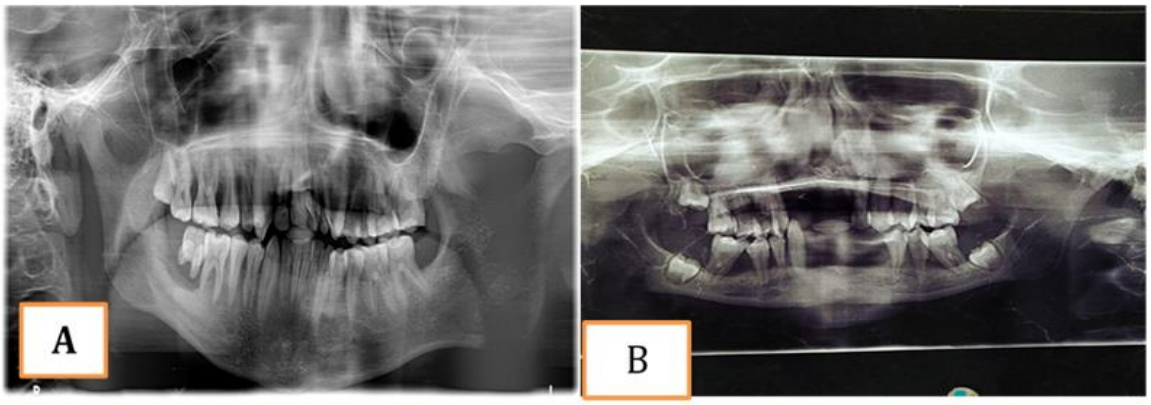

Fig-3 Showing radiographic feature of both [CASE-1; CASE-2] in generalized horizontal bone loss and arc shaped bone loss around lower 1st molar region

Discussion: Papillon Lefevre syndrome is a rare keratinization disorder inherited as an autosomal recessive trait. [1,2] This means that both parents are phenotypically healthy. Dermatological, periodontal, and radiological findings strongly suggested PLS in the presented case.[1] In this syndrome when there is parental consanguinity, the prevalence rises. [1-3,6]

Although the exact cause of this syndrome is unknown, immunological and microbiological lesions were diffuse with a dry, scaly surface that varied in thickness and was rough to the touch which worsened during winter. [1,9]

In our case- 1 - the patient had multiple missing permanent teeth and the remaining permanent teeth exhibited significant mobility, as well as all signs and symptoms of alveolar bone loss.

The patient's OPG revealed a significant alveolar bone loss in almost all the teeth except for erupting 3rd molar region.[1,9]These skin lesions and dental results are all compatible with the characteristics of PLS.

It has been hypothesized that a variety of variables contribute to the development and progression of PLS. One of the etiological factors was a reduction in neutrophils chemotaxis; phagocytosis and bactericidal activities accompanied by a decrease in cell migration.

Fusobacterium nucleatum and Treponema denticola organisms suggested that many pathogens may be implicated in disease progression.[1,3,7,9]AA(Aggregatibacter actinomycetemcomitans) is present in the plaque and pocket so it is strongly linked to the destruction of the periodontium.[1] Increased susceptibility to bacterial infection results in recurrent pyogenic liver abscesses as a complication. [1)]

The Cathepsin C gene is important for the structural growth and development of skin and the appropriate immune response of myeloid and lymphoid cells. [1-3]

The differential diagnosis would include Haim-Munk syndrome, autosomal recessive genodermatosis distinguished by congenital palmoplantar keratoderma and progressive early-onset periodontitis. Haim-Munk syndrome is also defined by arachnodactyly; acroosteolysis nail atrophy and hand phalange deformity. None of these characteristics were noticed in the current cases. A deficiency of alkaline phosphatase activity is seen in hypophosphatasia. But in our case values are within normal limits so this differential diagnosis could be excluded.

A specific treatment regime has not been reported. Nonetheless, several therapeutic techniques have been recommended to control periodontal deterioration.[1] Examples: conventional periodontal therapy, oral hygiene instructions, and systematic

antibiotics. [2]This kind of treatment may be very helpful if it is started during eruption and maintained during the development of teeth but periodontitis is hard to control in PLS. Management can be done with oral hygiene, instructions, systematic antibiotics, and extraction of severely periodontal compromised teeth. Fundamental treatment options for these patients using stem cell therapy are likely to bring new opportunities.[1]Emollient is typically used to treat dermatological symptoms, however, the effectiveness may be increased by the addition of chalice, lick acid, and a topical steroid. [3]

Conclusion: It is crucial for dental practitioners to understand PLS syndrome, as all three cases had their symptoms. 0Should be well-versed to spot the disease in its earliest stages. The dentist is typically the first member of the medical team to notice this intriguing and difficult diagnostic issue. In addition to preventing or delaying tooth loss, early diagnosis and an effective treatment strategy also reduce the risk of economic, psychological, and social shame. The parents of the patient in the current case were consanguineous spouses who arrived at the hospital as all of their siblings had dental issues even though they were ignorant of the sickness, which raises the hereditary risk to their children The disease must be made more widely known, and appropriate, comprehensive dental care must be provided.

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