Oral Health Case Reports
ISSN: 2471-8726
Tiwari, Oral health case Rep 2016,2:1
DOI: 10.4172/2471-8726.1000103
Case Report Open Access

Papillon-Lefevre Syndrome in 9 Year Old Peadatric Patient: A Rare Case Presentation with Comprehensive Review

Manjul Tiwari*
School of Dental Sciences, Sharda University, Uttar Pradesh, India

Abstract

Papillon-Lefevre syndrome is a rare autosomal recessive disorder in which the frequently observed manifestations are palm plantar keratinization and premature loss of both deciduous and permanent teeth. Here I present a rare case report of Papillon-Lefevre syndrome in 9 year old Peadatric patient along with a comprehensive review of the etiology, pathology, clinical features, differential diagnosis and management of the condition.

Keywords: Papillon-Lefevre syndrome; Palm planter Keratosis; Autosomal recessive disorder

Introduction

Papillon-Lefevre syndrome (PLS), 1st described by two French physicians Papillon and Lefebvre in 1924, is an extremely rare genodermatosis inherited as an autosomal recessive trait, affecting children between the ages 1-4 years [1].

The disorder is characterized by diffuse palmoplantar keratoderma and premature loss of both deciduous and permanent teeth [2]. The second major feature of PLS is severe periodontitis, which starts at age 3 or 4 years [2,3]. The development and eruption of the deciduous teeth proceed normally, but their eruption is associated with gingival inflammation and subsequent rapid destruction of the periodontium. After exfoliation, the inflammation subsides and the gingiva appears healthy [3]. Nail changes are apparent in advanced cases, manifested by transverse grooving and fissuring [3,4].

The cause of PLS is not well understood, 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, are associated with PLS.5 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. In PLS defect in immune cells especially PMNLs is noted.

Here I report a rare case of Papillion lefevre syndrome in a 9 year old boy. Karyotyping was carried out in the present case to check the cathepsin C defect.

Case Report

A 9-year-old boy presented with a chief complaint of spontaneous exfoliation of his deciduous & permanent dentition teeth. He also had a history of persistent thickening, flaking and scaling of the skin of his palms and soles associated with recurrently swollen and friable gums since age of [5,6]. The remainder of his past medical history was unremarkable. There was family history of ichthyosis, palmoplantar keratodermas and spontaneous exfoliation in his younger sibling (Figures 1-3).

On oral examination, his gums were edematous, friable and receding with all missing teeth except mandibular and maxillary first molars. Grade II mobility was present in these teeth (Figures 1-3). OPG showed severe bone loss in relation to existing teeth (Figure 4) while lateral cephalogram showed intracranial calcifications (Figure 5). On extra oral examination, there were symmetric, well-demarcated, yellowish, keratotic, confluent plaques affecting the skin of his palms and soles and extending onto the dorsal surfaces (Figures 6-9). No nail dystrophy was seen. Well-circumscribed, psoriasis form, erythematous, scaly plaques were present on the elbows and knees bilaterally (Figures 10-14). Routine blood investigations, liver function tests like serum transaminase levels, total bilirubin, and alkaline phosphatase levels were carried out and were found within normal range. Karyotyping showed autosomal recessive gene defect on the 19th trigon. Co-relating the clinical, radiological and laboratory findings, a final diagnosis of Papillon-Lefevre syndrome was made diagnosed as Papillon-Lefèvre syndrome (PLS). Molars were extracted and complete denture was given.

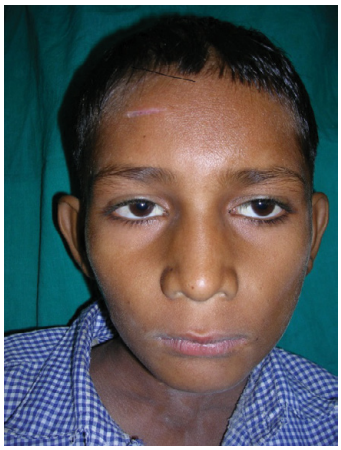
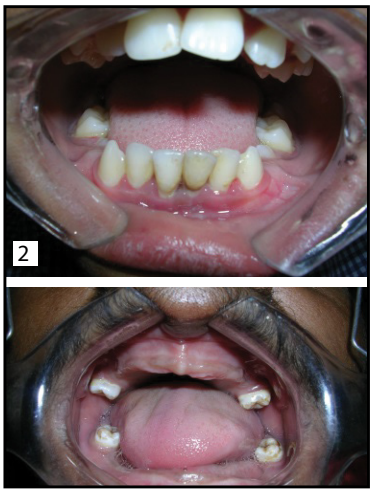


Figure 1: Shows the extra oral view.

***Corresponding author:** Manjul Tiwari, Assistant professor, Department of Oral Pathology and Microbiology, School of Dental Sciences, Sharda University, Uttar Pradesh, India, E-mail: manjultiw@gmail.com

Received: January 04, 2016; **Accepted:** January 14, 2016; **Published**: January 24, 2016

Citation: Tiwari M (2016) Papillon-Lefevre Syndrome in 9 Year Old Peadatric Patient: A Rare Case Presentation with Comprehensive Review. Oral health case Rep 2:103. doi:10.4172/2471-8726.1000103



Figures 2 and 3: Intra oral view showing premature loss of teeth.

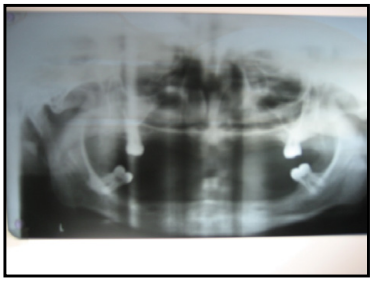


Figure 4: Orthopantomograph showing severe bone loss.

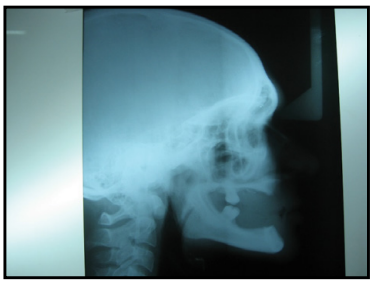


Figure 5: Lateral cephalogram showing intracranial calcifications.

Discussion

PL syndrome is inherited as an autosomal recessive disease in which both the parents are phenotypically healthy. Only the affected person and possibly some siblings have the disease, rest of the family gives no history of the disease [7].

The etiology of PL syndrome is still unknown. Factors suggested as responsible for disease includes (1) Impairment of neutrophil chemotaxis, phagocytosis and bactericidal activity. (2) Actinobacillus actinomycetemcomitans virulent gram negative bacteria found in periodontal pockets may act as triggering factor (3) defect in immune mediated mechanisms [8]. Laass et al conducted a genetic study for PL syndrome and found that chromosome 11q14-q21 is responsible for the disease where the cathepsin C gene is located. This gene encodes a cysteine - lysosomal protease which removes the dipeptides from the amino terminus of protein substrates. Cathepsin C is expressed in epithelial regions like palms, knees, soles and keratinized oral mucosa. Total loss of cathepsin C activity was seen in PL syndrome patients [9].

Histopathological findings in oral tissues have not been well described in literature but skin lesions consist of hyperkeratosis, occasional patches of parakeratosis, acanthosis and slight perivascular inflammatory infiltrate [10,11]. Cathepsin C gene mutations also results in two other closely related conditions: the Haim munk syndrome and prepubertal periodontitis which must be included in differential diagnosis of PL syndrome [11].

Haim munk syndrome is an autosomal recessive genodermatosis characterized by congenital palm planter keratoderma and progressive early onset periodontitis. In contrast to PLS, the cutaneous findings in HMS are more severe and extensive and the periodontium is less severely affected. Other clinical findings in HMS are acro-osteolysis, atrophic changes in nails, acachnodactyly, and peculiar radiographic deformity of the fingers consisting of tapered pointed phalangeal ends [12].

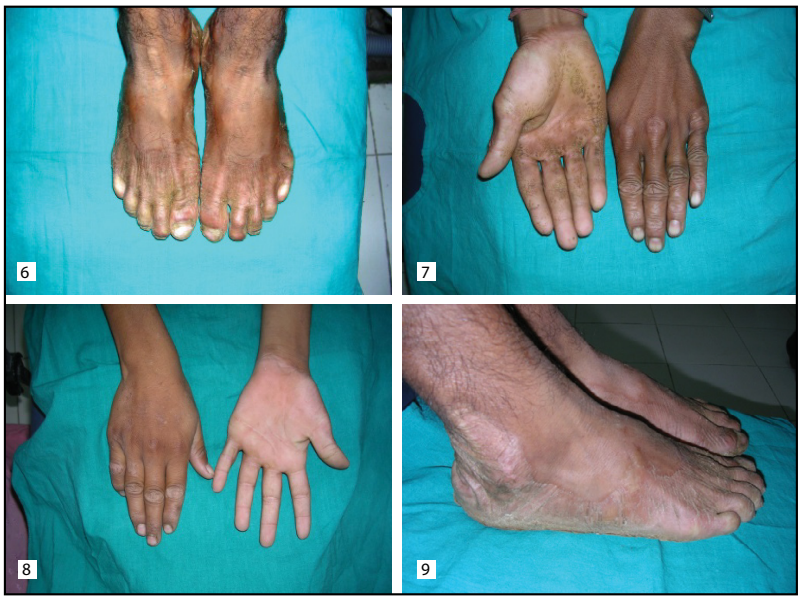
Prepubertal periodontitis is another rare genodermatosis caused by cathepsin C mutation and characterized by rapidly progressive early onset periodontitis with destruction of both deciduous and permanent teeth but it can be differentiated from PLS by absence of associated palm plantar keratoderma [13].

Complications in PLS include bacteremia and pyogenic liver abscess. Patients have decreased neutrophil, lymphocyte and monocyte functions and an increased susceptibility to bacterial infection, leading to recurrent pyogenic infections of the skin. Pyogenic liver abscess is a complication of PLS, is associated with impairment of the immune system [14].

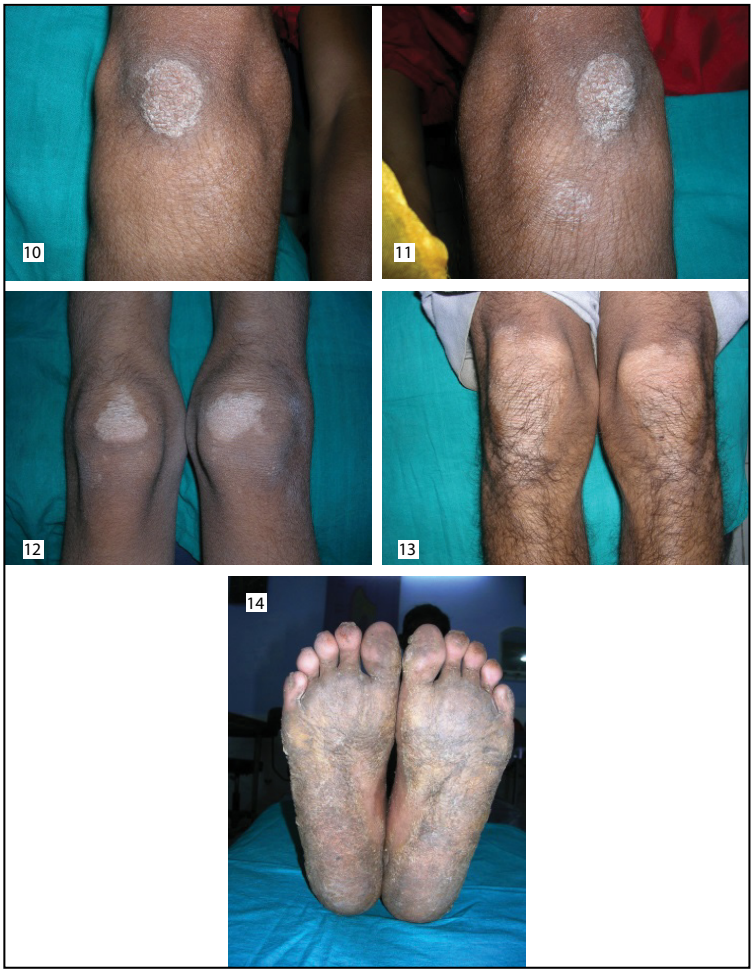
Thus, a multidisciplinary approach is important for the care of patients with PLS. The skin manifestations of palmoplantar keratoderma are usually treated with emollients. Oral retinoids including acitretin, etretinate, and isotretinoin are the mainstay of the treatment of both the keratoderma and periodontitis associated with PLS. Effective treatment for the periodontitis includes extraction of the primary teeth combined with oral antibiotics and professional teeth cleaning [15]. It is reported that etretinate and acitretin modulate the course of periodontitis and preserve the teeth [16]. PLS debilitates individuals socially, psychologically and physically. Thus, Prosthetic rehabilitation in such patients is useful. Depending on the pattern of missing teeth and the remaining available alveolar bone, the ideal treatment option in this type of case would be implant supported denture. Implants have been shown to help preserve alveolar bone, if bony atrophy progresses to the extreme in already alveolar deficient patients, implant placement may not be possible without bone grafting. There are different over denture treatment modalities based on the need for retention, stability, support and economy [16].

Conclusion

The diagnosis of Papillon-Lefevre syndrome is unique to its clinical characteristics. However, the management from a periodontal point of view appears to follow the routine phases of periodontal therapy, with special emphasis on the periodic recall review and maintenance of the individual with systemic antibiotics when deemed necessary. The syndrome can reduce the self-confidence of the patient at a very early age and thus oral rehabilitation must take the forefront. Though initial replacement is with removable partial dentures, future consideration must be given for implant-supported prostheses. The complete



Figures 6-9: Showing hyperkeratosis of hands and feet.



Figures 10-14: Shows bilateral psoriasiform plaques on knees and elbows.

rehabilitation of both periodontal health and functional esthetics is thus an interdisciplinary approach which improves the morale of the patients and parents.

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