ABSTRACT

Introduction: Papillon-Lefevre Syndrome (PLS) is a rare autosomal recessive disorder of keratinisation, characterized by palmoplantar hyperkeratosis, periodontitis and early loss of dentition. Since these features are common to both dentistry and dermatology, members of both disciplines should be aware of the same because an early diagnosis of this condition can help to preserve the teeth by early institution of treatment, using a multidisciplinary approach.

Case Series: The authors here present two cases of Papillon-Lefevre syndrome in siblings, having all of the characteristic features, along with a comprehensive review of the etiology, pathology, clinical features, differential diagnosis and management of the condition.

Conclusion: The main priority of physician’s strategy is to refer the PLS patients at the earliest to a periodontist for the periodontal management of permanent dentition. Based on the previously reported series and our clinical experience, non-surgical periodontal therapy yielded better results when started immediately after the extraction of deciduous dentition.

Keywords: Palmoplantar hyperkeratosis, Papillon lefevre syndrome, Periodontitis

INTRODUCTION

Papillon Lefevre syndrome (PLS), also known as palmo-plantar keratoderma with periodontitis, is an inherited disorder of keratinisation. It was first described by two French physicians, Papillon and Lefevre in 1924 [1]. It is characterized by redness and thickening of the palms and soles, along with extensive loss of periodontal attachment structures accompanied by generalized, rapid destruction of the alveolar bone around both the deciduous and permanent teeth [2]. Gorlin et al. stated that the calcification of dura mater is a third component of the syndrome [3]. The prevalence of PLS is 1–4 per million individuals with both males and females being equally affected. There is no racial predominance. A genetic predisposition, with a greater frequency of occurrence in consanguineous offspring has been reported [4]. Periodontitis, the second most common feature of PLS starts at age of three or four years [5]. The eruption pattern and the development of the deciduous teeth proceed normally, but their eruption is always associated with gingival inflammation and a subsequent bone loss around those
teeth. The resulting periodontitis is characteristically unresponsive to traditional periodontal treatment modalities and the primary dentition is usually exfoliated prematurely by age of four years. The inflammation subsides immediately after the exfoliation of the primary teeth 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 [6, 7]. Severe resorption of alveolar bone gives the teeth a ‘floating-in-air’ appearance on dental panoramic radiographs [8]. The severity of dermatologic involvement may not be related to the level of periodontal infection [9]. 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 [10]. Here we report a case of siblings with PLS, one female aged 26 years and the other male aged 15 years.

**CASE SERIES**

**Case 1:** A 15-year-old male patient came to the department of periodontia, complaining of mobile anterior teeth and difficulty in eating since a year, due to the exfoliation of all the permanent teeth, excluding three mobile anterior teeth. Going through the past dental history, the patient had an early loss of primary teeth with a sequential loss of permanent teeth due to excessive mobility. The family history also revealed the fact of consanguineous marriage of the parents. He also revealed that his sister aged 26 (case 2) was also affected with the same disease. The mother had noticed skin lesions on the palms and soles of the children when they were one year old. On general examination of the patients, it was noted that they had normal overall physical and mental development.

Physical examination revealed symmetrical, well demarcated, yellow keratotic plaques on the skin of the palms and soles, which extended to the dorsal surface of the finger joints and also over the dorsal surface of the feet. These keratotic plaques on the skin were dry, scaly and rough on palpation (Figure 1). On intra-oral examination, it was found that except for the canine and the erupting lower third molar, all the other permanent tooth including the left lower canine were missing. (Figure 2). Mobility was present in all permanent canines that were present. The gingiva in relation to the existing permanent tooth was red, edematous with marked clinical attachment loss. Despite the severity of the periodontal involvement, no visible local factors were found. The mucosa of the edentulous area near the periodontally involved teeth was normal. The dental panoramic radiograph showed severe alveolar bone loss in relation to the existing permanent teeth up to the apical third of the roots, giving the teeth a ‘floating in air’ appearance (Figure 3). The lateral view of the skull radiograph showed no evidence of intracranial calcification. Since the prognosis was very poor for the remaining permanent teeth, complete extraction of the mobile teeth, with subsequent prosthetic rehabilitation was advised.
Case 2: A 26-year-old woman came to our department on the request of her brother (case 1) for examination for this study. On general examination, it was found that, she had normal physical and mental development. Physical examination revealed symmetrical, but not a well formed keratotic plaques on the skin of the palms and soles. The extension of these plaques to the dorsal surface of the finger joints and also over the dorsal surfaces of the feet was mild and not as marked as in case 1 (Figure 4). She gave a history that the keratotic plaques was marked and well demarcated, similar to her brother but later, the keratotic plaques were peeled from the skin surface after applying a medicine given to her by her dermatologist. On intra-oral examination, it was found that she was wearing a complete denture set for ten years. All her permanent tooth were lost at the age of 15, except her lower third molar, which persisted with marked inflammation around the tooth (Figure 5). The edentulous area other than the lower third molar was normal with out any inflammation. The dental panoramic radiograph showed severe bone loss in relation to the lower third molar and the resorption of the ridge was severe (Figure 6). The lateral view of the skull radiograph showed no evidence of intracranial calcification.

DISCUSSION

Papillon Lefevre syndrome can drastically affect the psychology of children because of social and esthetical problems. Hence, early dental evaluation and parental counseling as a part of preventive dental treatment is needed. A multidisciplinary approach is needed for providing a complete psychological rehabilitation to improve the prognosis and quality of life for PLS children.

Although the etiology of PLS remains relatively obscure, Hattab et al. classified the etiology into three types: immunologic, microbiologic, and genetic factors. From an immunologic point of view, an impaired neutrophil chemotaxis, phagocytosis and bactericidal activities accompanied by a decrease in cell migration was noted [11, 12]. The immune mediated mechanisms such as lymphocytic response to pathogens, helper/suppressor T cells ratio and monocytic function were also impaired in this syndrome. Microbiologically, the presence of virulent gram negative anaerobic pathogens (Actinobacillus actinomycetemcomitans) in the periodontal pockets and periodontal plaques are noted, which might act as a trigger factors [13]. Previous case reports and studies have reported that A. actinomycetemcomitans plays a important role in the pathogenesis and progression of the rapid periodontal breakdown seen in PLS [14]. In addition to the organism, the presence of leukotoxins, collagenase, endotoxin, epitheliotoxins and fibroblast inhibiting factors suggest that PLS is mediated bacteriologically [15]. Recently genetic factors, have gained importance in the pathogenesis of PLS due to advancements in the field of genetic engineering. The inactivation of the cathepsin C gene is recently noted as the primary factor that is responsible for the abnormalities in skin.
development and periodontal disease progression [6]. An interesting point about the cathepsin C gene is that mutations in this gene also results in two closely related conditions: i) Haim-Munk syndrome, and ii) aggressive periodontitis. A common clinical manifestation in all three conditions is the severity and the early onset of periodontal destruction. Genetic testing to identify the gene mutation was not performed in our patients because of their low economic status but the periodontal, dermatological and radiological features strongly favor diagnosis of PLS. Moreover, the sibling we reported here were also associated with consanguinity of parents, which again supports our diagnosis of PLS. No family history for our cases was present, so the autosomal recessive pattern of inheritance was strongly proven. Recent studies have shown that PLS syndrome is manageable and that the permanent teeth can be saved [2]. Medications to prevent and control microbial attack and steps to conserve the periodontium should always be given priority before the disease process exceeds the clinical therapeutic limits. Mc Donald et al. have put forward the following guidelines for the successful management of PLS cases. It includes: i) extraction of all deciduous dentition, ii) construction of complete dentures three months after the removal of primary teeth, iii) prophylactic doses of tetracycline for 10 days immediately after the denture insertion, iv) adjustment of denture bases to allow for the emergence of the permanent dentition followed by another therapeutic dose of the tetracycline [16]. Tetracycline should be administered at a dosage of 250 mg four times daily for one month.

CONCLUSION

In cases of PLS, early referral to the dentist to permit proper diagnosis as well as management of patient’s oral condition is important. The periodontist and prosthodontist should be referred for the successful management of PLS. The periodontal management of the patients includes the conventional periodontal therapy, oral hygiene instructions and systemic antibiotics.

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Author Contributions

Jayachandran Dorairaj – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article or revising it critically for important intellectual content, Final approval of the version to be published

Sumantha Selvaraj – Substantial contributions to conception and design and interpretation of data, Drafting the article, Final approval of the version to be published

Mohammed Sadique – Substantial contributions to conception, and analysis, Drafting the article or revising it critically for important intellectual content, Final approval of the version to be published

Michael Shaw – Substantial contributions to design and interpretation of data, Drafting the article or revising it critically for important intellectual content, Final approval of the version to be published

Sachin Kumar Amruthal Jain – Substantial contributions to interpretation of data, Drafting the article or revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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