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Papillon-Lefevre Syndrome: Report of two cases in the same family

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ABSTRACT
Papillon-Lefevre syndrome (PLS) is a rare autosomal recessive disorder characterized by the association of palmoplantar hyperkeratosis and premature loss of both deciduous and permanent teeth. Two siblings (29-years-old female and 36-years-old male) with PLS who complained about early teeth loss, esthetic problems and difficulty during eating and speech refered to our clinic. After intraoral and radiologic examination we planned total prosthesis to first case's upper and lower jaw and partial prosthesis to second patient's upper and lower jaw. Patients' financial problem affected the treatment planning. This case report presents prosthodontic rehabilitation of two patients with PLS in the same family.
Keywords: Papillon-Lefevre syndrome, Palmoplantar keratoderma, loss of dentition

INTRODUCTION

The Papillon-Lefevre syndrome (PLS) was first described by two French physicians, Papillon and Lefevre in 1924. It is a rare autosomal recessive disorder characterized by a diffuse palmoplantar hyperkeratosis and periodontal destruction of both primary and permanent dentition.[1] The prevalence of PLS is one to four per million individuals and >300 cases have been reported worldwide.[2] Males and females are equally affected with no racial predominance.[3] It seems that the incidence in the Saudi Arabia is higher compared with other parts of the world, and this may be attributed to cluster marriages (consanguineous marriages).[3]

Exact etiology and pathogenesis is unknown, but intensive work during last decade has revealed some important factors in the development of the disorder. In recent years, genetic, immunologic and microbial factors are suggested as responsible for the initiation and progression of PLS.[1] Several mutations have been reported in the cathepsin C gene in individuals with PLS from diverse ethnic groups.[4] The cathepsin C gene is expressed in the 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. This discovery will certainly lead to better understanding of the signs and symptoms associated with PLS.[1,5]

Clinically, hyperkeratotic palms and soles with rapidly progressive periodontitis are hallmarks of this condition. Other findings associated with but not considered to define PLS, may include psoriasiform plaques of the elbows and knees, dystrophic nails, intracranial ectopic calcifications, recurrent pyogenic infection of the skin, retardation, arachnodactyly, decreased neutrophil, lymphocyte or monocyte functions and pyogenic liver abscess due to impairment of the immune system[6-8]. The degree of dermatologic involvement may not be related to the level

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of periodontal infection.[4,9] The cause of the periodontal disease and increased susceptibility to infection is attributed to decreased neutrophil phagocytosis, bacterial infection, and impaired reactivity to T and B cell mitogens.[4,10]

The aim of this report was to present prosthodontic rehabilitation of two cases of PLS in the same family.

CASES

A 29-years-old female (case 1) and her brother, a 36-years-old male (case 2) referred to the Gazi University, Faculty of Dentistry (Ankara, Turkey) the complaint of early teeth loss, esthetic problems and difficulty during eating and speech. Also, they complained of difficulty pronouncing words during speech. Medical history revealed that the patients were diagnosed with PLS during childhood. Vaseline pomade and oral retinoid were prescribed for the disease by a dermatologist, because of adverse effects of oral retinoid they have given up using them. They were using only Vaseline pomade at the time of application.

Parents were consanguineous marriage. Mother had a full term, normal, uneventful pregnancy. The mother had noticed skin lesions on the palms and soles of the children when they were 5-6 months old. On physical examination, both of the patients were slightly slim and had overall normal mental development. Routine blood investigations and liver function tests were within the normal limits in both of the cases.

Dental history revealed that their deciduous teeth had erupted normally at 8-9 months of age, which started loosening at three years and were all eventually lost by four years of age. Patients were not sure about the time of eruption of permanent teeth, but described gingival bleeding during brushing and eating, after the eruption of permanent teeth.

Case 1

Extraoral examination revealed dry and rough skin with normal development of hair. Horizontal lines, dystrophic deformity and pitting were seen in the toenails. There were symmetric, well-demarcated, keratotic and confluent plaques affecting the skin of her palms and soles, also extending onto the dorsal surfaces of hands and feet (Figure 1).

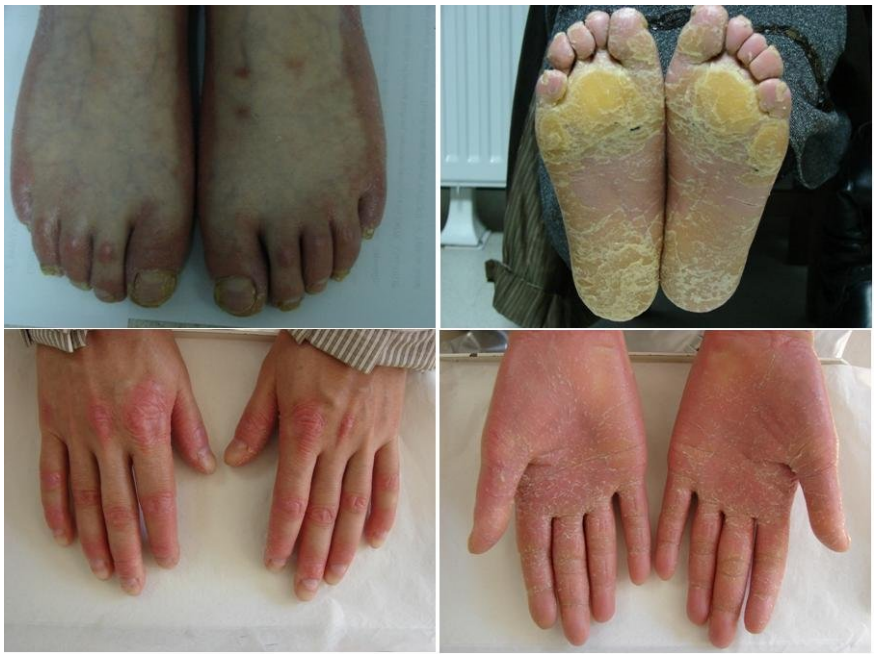

Figure 1. Case 1's hands and feet.

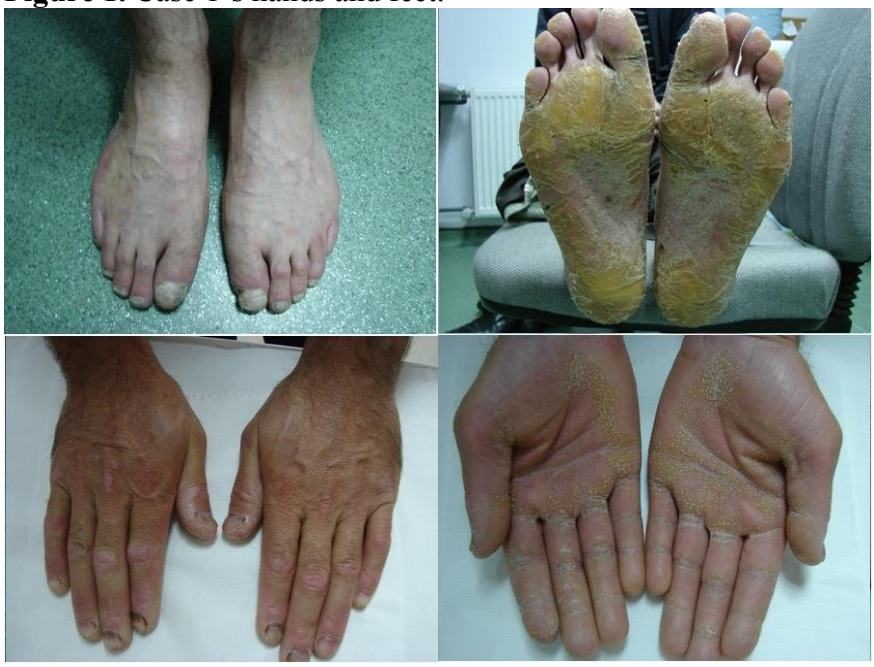

Fig. 2. Case 2's hands and feet.

The well demarcated psoriasiform plaques worsening in winter were observed in the knees and in the elbows. She had upper lip retrusion, mandibular prognatia, maxillary retrognathia and decreased lower facial height (Figure 2).

Intraoral examination showed premature loss of teeth, and no other abnormality was detected in relation to soft tissues. There was loosening of permanent teeth from 10 years of age and eventually all the permanent teeth were lost by 15 years of age. Bleeding was also associated to tooth loss. Hence, she expressed a keen desire for replacement of all her lost teeth. Panoramic radiograph showed generalized loss of alveolar bone. Skeletal Class III relationship was observed and no intracranial calcification was evident on the lateral cephalogram. After clinic and radiographic examination, total denture prosthesis was planned to rehabilitate masticator and speech functions and esthetic.

Case 2

Extraoral examination revealed that the skin was dry and rough on palpation and hair was normal. Horizontal lines, dystrophic deformity and pitting were seen in the toenails. There were symmetric, well-demarcated, keratotic and confluent plaques affecting the skin of his palms and soles, extending onto the dorsal surfaces of hands and feet (Figure 3).

No psoriasiform plaques were observed anywhere of the body. He had upper lip retrusion, mandibular prognatia, maxillary retrognathia and decreased lower facial height (Figure 4).

Intraoral examination showed premature loss of teeth, and no other abnormality was detected in relation to soft tissues. He reported that all of his permanent teeth were lost by 16 years of age except for four teeth (17,27,37 and 47). Bleeding was also associated at the time of tooth loss. Hence, he expressed a keen desire for replacement of all his lost teeth.

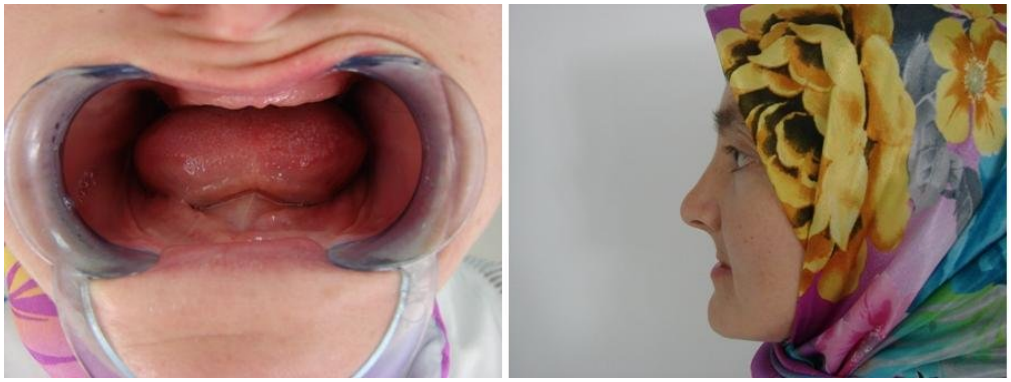

Figure 3. Case 1 before treatment.

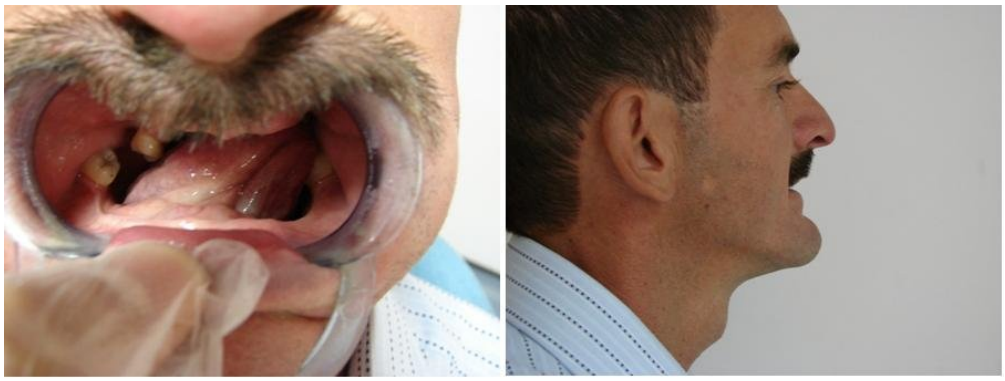

Figure 4. Case 2 before treatment.

Panoramic radiograph showed generalized loss of alveolar bone. Alveolar bone support surrounding the teeth (17,27,37 and 47) was sufficient, but the teeth were slightly displaced toward to adjacent edentulous regions. Skeletal Class III relationship was observed and no intracranial calcification was evident on the lateral cephalogram. After clinic and radiographic examination, any treatment was not required for remaining teeth and partial denture prosthesis was planned to restore masticator and speech functions and esthetic (Figures 5 and 6). Informed and signed consent forms were obtained from the patients.

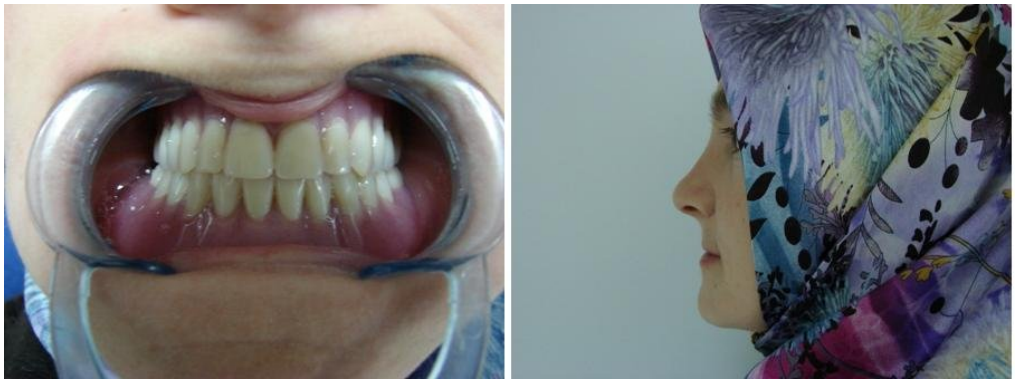

Figure 5. Case 1 after treatment.

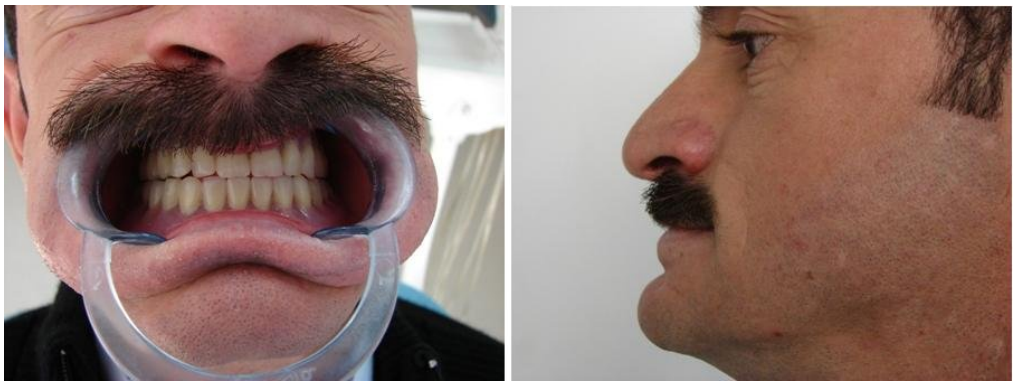

Figure 6. Case 2 after treatment.

DISCUSSION

PLS is a rare disorder which is inherited in an autosomal recessive manner. Both parents are phenotypically healthy and there is no family history of the disease other than the affected person and possibly some siblings. Both parents must carry the autosomal gene for the syndrome to appear in their offspring.[11] Therefore, incidence of PLS is higher in consanguineous marriages.[3] In this report, parents of the patients were consanguinity and phenotypically healthy.

Palmoplantar keratosis and early-onset form of rapidly progressive periodontitis are two cardinal diagnostic features in PLS. Other skin lesion may appear as well-demarcated psoriasiform plaques on the elbows and knees, dystrophic nails, recurrent pyogenic infection.[6-8]

Additionally, it was reported that cheeks, eyelids, labial commissures and thighs can be affected by the keratosis, although it varies significantly.[12] In both of our cases, the skin findings were characteristic; sharply demarcated keratotic plaques involving the entire surface of the palms and soles. The lesions were diffuse, with a dry, scaly surface; they varied in thickness and were rough on palpation. There were horizontal lines, dystrophic deformity and pitting in the toenails. In case 1, the well demarcated psoriasiform plaques were observed in the knees and elbows. The skin lesions in these patients appeared in the first and second decades of life. The patients were diagnosed during childhood with PLS by a dermatologist. The dental history was also in accordance with the known symptoms of the syndrome, i.e, early exfoliation of permanent teeth. Case 1 had totally edentulous jaws and case 2 had partially edentulous jaws. No other abnormality was detected associated with soft tissues in intraoral examination.

Bindayel et al.[13] investigated the craniofacial characteristics of patients with PLS and they reported that these patients have a Class III skeletal pattern. Patients with PLS generally have the following features additionally: Class III skeletal relationship, mainly due to a retrognathic and hypoplastic maxilla; decreased lower facial height, retroclination of the mandibular incisors as a compensation for maxillary prognatism and upper lip retrusion.[13] In addition, it was reported that intracranial calcifications may be present in PLS.[5,6,8-10] In both of our cases, craniofacial features including Class III skeletal relationship, upper lip retrusion, mandibular prognatia, maxillary retrognathia and decreased lower facial height were found in accordance with the findings of Bindayel et al.[13] Panoramic radiographs of both patients showed extensive alveolar bone loss and no intracranial calcifications were evident on the lateral cephalograms.

Like the dermatologic manifestations, periodontal disease in PLS also begins during early childhood.[2] The onset of disease usually coincides with the eruption of primary teeth. Following the eruption, the gingival become inflamed. This is generally followed by a rapid destruction of periodontium and most affected children experience premature loss of primary teeth. Gingiva seems to resume normal appearance after exfoliation of the primary dentition. The aggressive inflammatory periodontal process then repeats itself after the eruption of the permanent teeth, and in general all or most of the permanent dentition is lost during the teenage years.[1]

Theories on etiology of the periodontal lesions seen in PLS include anatomical defects (cemental, epithelial and salivary), microbiological factors (Actinobacillus actinomycetemcomitans) and other putative periodontal pathogens, viral agents (cytomegalovirus, Epstein-Bar type 1 virus), and host response (PMN function) and Iymphocyte functional defects as causative factors.[1] In PLS, the destruction to the periodontium is severe and commences early in life at approximately 1 to 4 years of age.[14] In our cases, laboratory examinations were within the normal limits. The patients were adult and they cannot give clear information about the loss of their primary teeth. They reported that their permanent teeth were lost by 15-16 years. Bleeding was also associated at the time of tooth loss.

Incidence of this rare entity is increasing in recent times and it involves severe irreparable destruction of dentition at an early age, so the dental professional has an important role to play and hence should be well thorough with etiological factors and effective treatment of PLS. The PLS debilitate individuals socially, psychologically and physically, so oral rehabilitation of the patients is essential. Prosthodontic replacement provides a psychological boost up to these patients not only the esthetic appearance but also the function.[15] For many years, clinicians have been aware of the prosthetic problems posed by severely atrophic thin alveolar ridges and have constantly endeavored to develop new methods of treatment. Preprosthetic surgical techniques have been introduced as aids to retention and stability of dentures. Modern dental implants are available that offer not only considerably better stability and retention of prosthesis but also improved comfort and masticatory efficiency and esthetic.[1] In our patients complained of difficulty during eating and speech because of toothlessness. We applied conventional treatments due to economical problems to rehabilitate masticator and speech functions and esthetic.

In conclusion, the dentists are usually the first member of the team to see this interesting and challenging diagnostic problem and must differentiate it from other entities on the basis of the unique dermatologic findings and the absence of other symptoms. The PLS causes physiologic, physical and social problems for an individual. Prosthodontic rehabilitation is essential for chewing, nutritition, speech and esthetic and it must be considered as very important part of treatment in patients with PLS.

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