PAPILLON-LEFÈVRE SYNDROME: AN UNUSUAL CASE REPORT

Gökhan Özkan, Aydan Kanlı, Saniye Eren Halıcı

ABSTRACT

Papillon-Lefèvre syndrome (PLS) is a rare autosomal disorder characterized by palmoplantar hyperkeratosis and periodontal problems leading to premature loss of both primary and permanent dentitions. It is caused by mutation of cathepsin C gene generating deficiency in host immune response to microorganisms. Up to date, no sexual or racial predominance has been detected. This paper report the diagnosis and rehabilitation procedures for a 14-year-old male patient with PLS.

Keywords: Cathepsin C; hyperkeratosis; Papillon-Lefèvre syndrome; prosthetic rehabilitation

Introduction

Papillon-Lefèvre syndrome (PLS) is a rare autosomal recessive disorder that results from mutation of cathepsin C gene. Genetic, immunologic and microbiologic factors are influential issues regarding origin and progression of the disease. Although a. a. comitans has frequently been considered as another cause of the disease, loss-of-function mutation in the cathepsin C gene has been also shown in patients. While homozygous patients have mutation and completely lack cathepsin C activity, heterozygous patients who are carriers have low cathepsin C activity and do not show typical features of the disease. Inactivation of neutrophil serine proteases that call forth the cessation in host response to bacteria in periodontal tissues in PLS occurs as a result of deficiency at activation of cathepsin C gene in immune cells.

Papillon and Lefèvre first described the syndrome in 1924. It is characterized by palmoplantar hyperkeratosis starting in infancy. From the early onset severe periodontal destruction causing early loss of both primary and permanent teeth can be seen. Prevalence of the disease is about 1–4 cases per million. Characteristic indicators of the disease like palmoplantar hyperkeratosis and periodontal destruction were noticed between 1-4 years. A major number of teeth are exfoliated by 14–15 years of age with severe periodontal destruction. “Floating-in-air” appearance of teeth was seen on radiographs. No gender or ethnic predilection has been presented for PLS patients. Expectancy of life is normal for PLS patients.

The cathepsin C gene that is responsible for epithelial tissues such as palms, soles, knees, and keratinized oral gingiva, is affected in this syndrome. In addition to these common features infections, mental retardation, liver abscess, hyperhidrosis, calcification of the dura mater and nail changes were seen with PLS rarely. This paper report the diagnosis and rehabilitation procedures for a 14-year-old male patient with PLS.

Case Report

A 14 year-old male patient with a chief complaint of multiple missing teeth and poor chewing reported to the Department of Oral and Maxillofacial Radiology of Hacettepe University. History revealed that his parents did not have any remarkable idea when the primary teeth were lost. It was noted that when he was 4-5 years of age he started to lose permanent teeth. In extraoral examination he had ceratotic skin lesions on his hands (Figure 1) with normal feet. Medical history revealed that the patient had hyperhidrosis. No consanguinous marriage was reported in the family. Other members of the family who accompanied the patient were also examined and no similar symptoms were detected. On intraoral examination he had five mobile teeth with periodontitis, widespread rash on the alveolar mucosa and swelling on the left maxilla (Figure 2). His panoramic radiograph showed three impacted developing third molar bud and five erupted teeth with severe periodontal loss on the alveolar bone within both maxilla and mandible (Figure 3). With these signs and symptoms a provisional diagnosis of Papillon Lefevre syndrome was made and patient was referred to the dermatology and genetic clinics to confirm the condition. After confirmation of the diagnosis, the hyperkeratotic lesions were treated with retinoids by dermatology clinic. Patient was referred to periodontology and prosthodontics clinics for dental rehabilitation. Tooth 44 was retained in the mouth due to less mobility. Remaining teeth extraction and excision of reactive gingival growth on the left maxilla procedures were performed in oral and maxillofacial surgery clinic. Wound healing and periodontal health was achieved before prosthodontic treatment.

Patient had severe bone atrophy in both maxilla and mandible. Because patient doesn’t have any established occlusion maxillary complete denture and mandibular clasp-retained partial denture were planned considering his young age, insufficient facial profile and the residual alveolar ridge resorption to maintain vertical dimension to provide aesthetics, function and phonation. Following this, Maxillo-mandibular relation was achieved. After the registration of maxillo-mandibular relations, aesthetic and functional adjustments were made in try-in appointment to improve aesthetics with adequate lip fullness and inter arch relationship. The complete maxillary and partial mandibular dentures were then inserted in the patient’s mouth (Figure 4). Patient was informed to report for follow up with 3-6 months intervals.

Discussion

PLS is a type IV genodermatosis related to loss-of-function mutation of the cathepsin C gene. This leads to inactivation of neutrophil serine proteases that lead to disruption of the immune response to microorganisms. Periodontal destruction
Early and proper diagnosis of PLS has critical importance for the treatment and its prognosis. Retinoid is the first choice for PLS management by dermatologist. If the syndrome is detected at early ages, retinoid treatment aids the healing of skin lesions and development of teeth. Antibiotics are used to protect the patient from pyogenic lesions and it is also preferred for controlling the periodontal microorganisms. Tri-methoprim, sulfamethoxazole, and tetracycline have been reported as effective alternative against to bacteria that play a key role in progression of the disease. Amoxicillin plus clavulanic acid and metronidazole therapies are suggested for the prophylactic approach of periodontal problems.

Optimal environmental conditions can be achieved for the development of permanent teeth when the primary teeth were taken off from both maxillary and mandibular alveolar ridge. Only one permanent tooth was preserved for our patient to support the planned dentures because of severe periodontal destruction of other teeth. Complete elimination of the periodontal pathogens is required to provide maximum benefit from saved teeth. Professional oral prophylaxis at frequent intervals, oral hygiene practices, mouth rinses including chlorhexidine, appropriate antibiotic therapy are advised in order to control periodontal diseases. Patient has to be followed up by dermatology clinic for controlling skin lesions. Following prosthetic rehabilitation, the patient was followed up at regular intervals. Departments of dento-maxillofacial radiology, genetic and dermatology, maxillo-facial surgery, periodontology and prosthodontics had played important roles during the identification and treatment stages of this case.

Conclusion

In conclusion, if there is early loss of teeth and palmo-plantar hyperkeratosis PLS must be considered after the evaluation of panoramic radiograph. Multidisciplinary treatment approach must be performed to achieve a good prognosis.

Authors Affiliations

1. Gökhan Özkan, DDS, Ph.D., Assistant Professor, Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Adnan Menderes University, Aydın, Turkey. 2. Aydan Kanli, DDS, Ph.D., Professor, Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Hacettepe University, Aydın, Turkey. 3. Saniye Eren Halici, Ph.D., Prosthodontist, Muğla Oral and Dental Health Center, Muğla, Turkey.

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Address for Correspondence
Dr. Gökhan ÖZKAN, DDS, Ph.D., Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Adnan Menderes University, Aydın, Turkey.

Email: asgokhanozkanus@hotmail.com

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