Papillon-Lefevre Syndrome-3 Years Follow up: A Case Report*

*Filiz ACUN KAYA[1**], Zelal SEYFIOGLU POLAT[2], Esma AKUZUM BARAN[3], Gulucag Giray TEKIN[4]*

[1] *Assistant Professor, Dicle University, Faculty of Dentistry Department of Periodontology, Diyarbakir, TURKEY.*
[2] *Assistant Professor, Dicle University, Faculty of Dentistry Department of Prosthodontics, Diyarbakir, TURKEY.*
[3] *MsC, PhD, Şehitlik Ana Çocuk Sağlığı, Diyarbakir, TURKEY.*
[4] *Research Assistant, Dicle University, Faculty of Dentistry Department of Periodontology, Diyarbakir, TURKEY.*

Abstract

This syndrome appears in childhood or in early periods of descent ages. It is characterised by palmar plantar hyperkeratosis, tendence to getting dry and chopping of skin, thin and sparse hair and early-onset periodontitis. Not all of them, but there is a tendency of familial transition. In some patients the teeth looses begins at the ages of 2-4 years and after loosing all primary dentition it concluses by loss of permanent dentition at the ages of 15-16. This case report describes the clinical periodontal findings, medical treatment and prosthodontic treatment of a 5-year-old male patient with PLS. The patient provided informed consent, and the study was conducted in accordance with the Helsinki Declaration of 1975, as revised in 2000.

Upon initial presentation, a full periodontal examination was completed. Conventional probing depths (PD), gingival index (GI), and plaque index (PI) were measured prior to medical treatment, which involved oral hygiene instruction. And after these a mobile prosthetic aparatus was constructed for the patient.

Because of severe periodontal destruction in the present teeth it was decided to extract all of them. The patient was send to prosthetic construction department and a child prosthesis was planned. The patient follow ups were performed in 3 months periods. A new prosthetic aparatus was planned with the beginning of eruption of permanent teeth. The patients long term follow up is stil going on.

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Introduction

Papillon–Lefèvre syndrome (PLS) is characterized by hyperkeratosis of hands and feet and by a generalized aggressive periodontitis in both the primary and the permanent dentition[1]. The syndrome is a rare autosomal recessive trait with an incidence of between one and four persons per million. Parental consanguinity is demonstrated in between 20% and 40% of the cases[1]. Calcification of the falx cerebri and the choroid plexus, and retardation of somatic development is often an associated feature[2-4]. It has been suggested that 20–25% of patients show an increased susceptibility to infections[1,3,5], of which otitis media is a common example[6].

The recently identified genetic defect in PLS has been mapped to chromosome 11q14–q21, which involves mutations of cathepsin C[7,8]. Studies in PLS patients have shown more than 90% reduction in cathepsin C activity[8,9]. Despite these advances in characterizing the genetic basis of the syndrome, the pathogenic mechanisms leading to the periodontal involvement remain elusive. An impaired chemotatic and phagocytic function of polymorphonuclear leukocytes (PMNs) has been described in many reports[10-15]. In contrast to the above studies[16-17], however, reported normal PMN chemotaxis. Few reports have addressed lymphocyte function in PLS.

Periodontal effects appear almost immediately after tooth eruption when gingiva become erythematous and oedematous. Plaque accumulates in the deep crevices and halitosis can ensue. The primary incisors are usually affected first and can display marked mobility by the age of 3 years. By the age of 4 or 5 years, all the primary teeth may have exfoliated[1,4]. Treatment with oral hygiene instructions, scaling and root planing has been reported unsuccessful[18-20]. Non-surgical

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****Corresponding author:***
Assist. Prof. Dr. Filiz ACUN KAYA
Dicle University, Faculty Of Dentistry,
Department of Periodontology,
21280 Diyarbakir, TURKEY.

Tel: +90 412 248 81 01-3430
Fax: +90 412 248 81 00
e-mail: facunkaya@dicle.edu.tr

treatment combined with use of systemic antibiotics [21-24] and additional periodontal surgery[23,25] has also been reported to fail. Following such tooth loss, the gingival appearance resolves and may well return to health only for the process to be repeated as the permanent dentition starts to erupt[1]. The majority of the teeth are lost by the age of 14–15 years[1,4,5]. There is dramatic alveolar bone destruction, often leaving atrophied jaws[5]. Patients are often edentulous at an early age.

CASE

A 5 year old male patient appointed to Dicle University Faculty of Dentistry Department of Periodontology with the complaint of primary teeth mobility and early losses.

In the history of the patient there was no systemic problem, but palmar plantar hyperkeratosis was observed (fig. 1,2). The 2. degree of relativeness of his parents was determined. By the intraoral examination it was determined that except the canines and primary 2. molars all the primary dentition were lost early in both upper and lower jaw (fig. 3). In addition to third degree mobility, alveolar bone loss, severe gingivitis and plaques and pus formation was observed in the vicinity of the present teeth. In the radiologic examination by panoramic graphy severe alveolar bone loss was observed (fig. 4). By the periodontal examination and index scores; the existance of 5 to 10 mm probing depths (CD=7.87 mm), gingival scores as GI=2 (26), and plaque scores as PI=2 (27)was established. In the light of these findings Papillon- Léfevre Syndrome was diagnosed in the patient. This diagnosis was confirmed by the dermatology clinic as palmar plantar hyperkeratosis. First of all, the patient was prescribed an antibiotic of amoxicillin (250 mg, 2x1, one week) and metranidazol (250 mg, 2x1, one week) and a moth rinse of 0.2% chlorhexidine gluconate (2x1, one week), and educated for oral hygiene. Patient was followed up two week later. But, because of severe periodontal destruction in the present teeth, all of them were extracted.

The patient was send to prosthetic construction department and a child prosthesis was planned (fig. 5). But the patient refused to use his mobile prosthetic application. The patients follow up trials lasted untill the permanent dentition eruption. After this period a new prosthetic application was constructed and the patient was persuated to use (fig. 6). The patients long term follow up is stil going on.

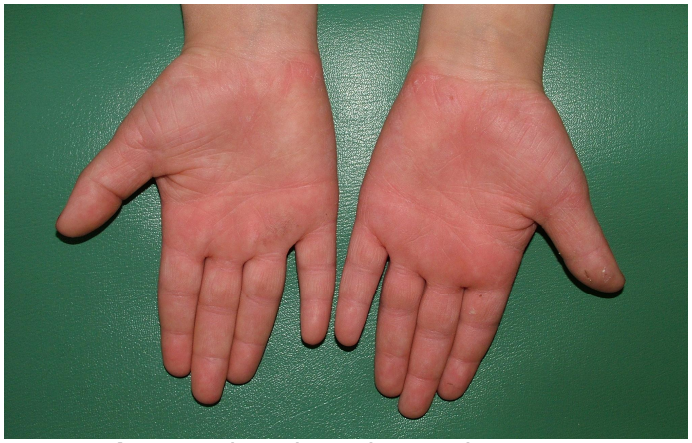

Fig.1 Palmar hyperkeratosis

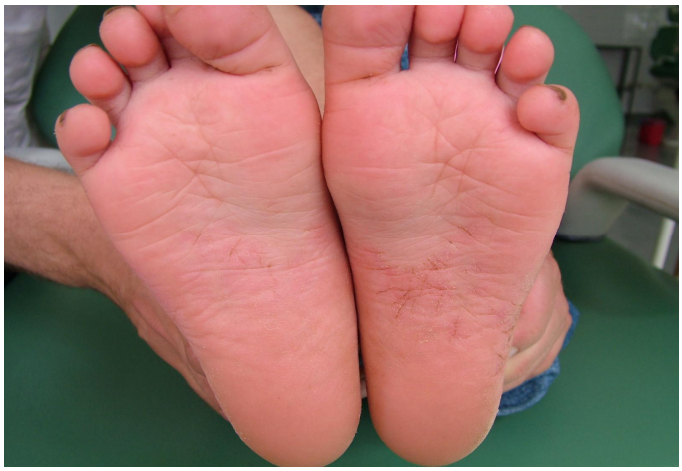

Fig. 2 Plantar hyperkeratosis

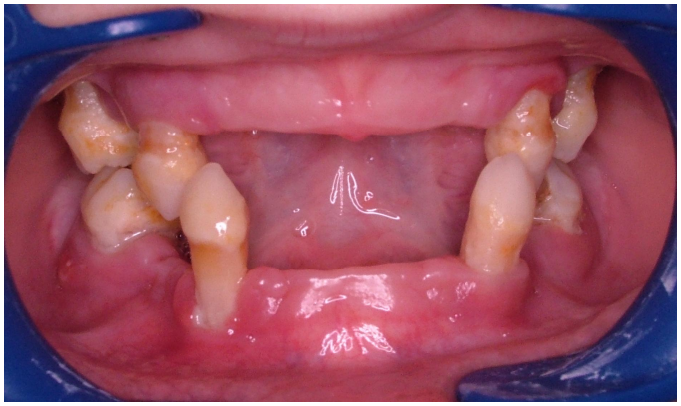

Fig. 3 Intra-oral scene

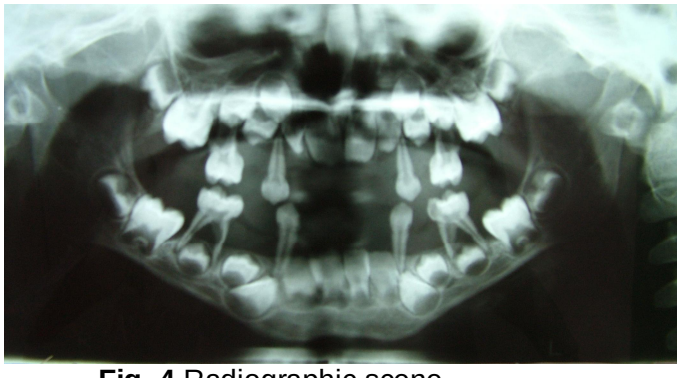

Fig. 4 Radiographic scene

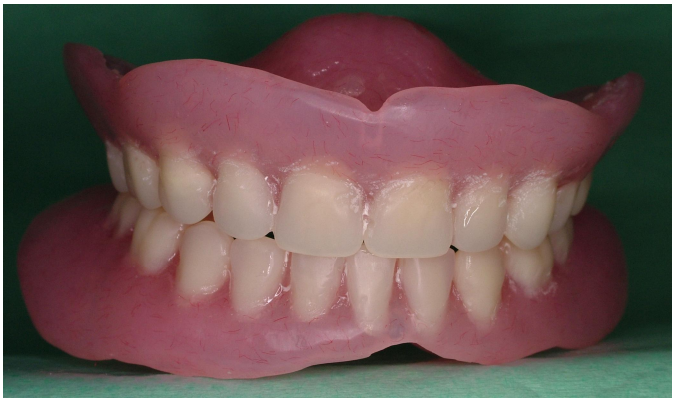

Fig. 5 Total prosthetic apparatus

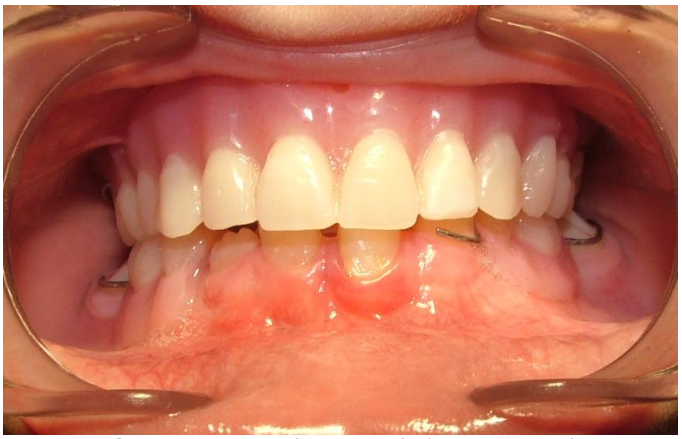

Fig. 6 Intra-oral scene (After 3 years)

Discussion

Haneke[28] used the following three criteria to classify a case as PLS: (a) palmoplantar hyperkeratosis; (b) loss of primary and permanent teeth; and (c) autosomal recessive inheritance. The population prevalence of PLS is reported to be one case in 1–4 million people[29]. With both parents as recessive carriers, there is a 25% chance of producing offspring with PLS[30].

Evidence has suggested that PLS patients have decreased chemotatic and phagocytic functions of neutrophil leucocytes, or a cellular immune defect involving decreased phyto-haemaglutinin response by T lymphocytes. Products of the Gramnegative organisms isolated from PLS patients' periodontal pockets may directly or indirectly contribute to leucocyte dysfunction, and there may be genetic component in the white cell dysfunction[31].

It has been suggested that the presence of periodontal pathogens alone is not sufficient for the expression of PLS, and other factors, such as host response, play an important role in the pathogenesis of the disease process[12]. Several authors have suggested an abnormal neutrophil dysfunction with PLS[3,32,33] to explain the pathogenesis, whereas others have reported cases where they appeared within normal limits[3].

It has been suggested that the development of periodontal disease in PLS patients might be associated with a specific profile of suspected subgingival pathogens coupled with some still unknown nature of altered and reduced immune defence. In a number of case reports, Actinobacillus actinomycetemcomitans has been observed in subgingival plaque samples from periodontal pockets in cases with PLS[4,12,19,24,25,32-35]. Other putative periopathogens including Porphyromonas gingivalis, Fusobacterium nucleatum, Bacteroides forsythus, Treponema denticola and Prevotella intermedia have also been implicated to play a role in PLS periodontal pathogenesis[36,37].

Severe periodontal and alveolar bone destruction in children necessitates that a diagnosis should be reached to exclude any life-threatening disorders. These include leukaemia and neutropenias, where loosening of the teeth is an associated feature, along with extensive gingivitis, haemorrhage and ulceration[22]. Other disorders where premature loss of primary and/or permanent teeth occur include; hypophosphatasia, Langerhan's cell histiocytosis, Chediak–Higashi syndrome, acrodynia and acatalasia[1,22]. The patients discussed in this paper presented with prepubertal periodontal destruction with concomitant palmar-plantar hyperkeratosis diagnosed as PLS.

Early case reports on periodontal treatment in PLS patients describe unsuccessful outcome and tooth loss leading to edentulism as an unavoidable part of this syndrome[27]. Treatment with oral hygiene instructions and scaling and root planing has been reported unsuccessful[18-20]. Non-surgical treatment combined with use of systemic antibiotics[21-24] and additional periodontal surgery[23,25] has also been reported to fail.

PLS patients are reported to complain about loose teeth, halitosis, swollen gums, food impaction and pain during chewing[28]. Multiple periodontal abscesses are common[26]. Progressing periodontal disease leading to tooth loss is a major trauma in these children. Extensive and repeated orthodontic and prosthodontic treatment may become necessary to provide the children with a functional dentition during the growth period of their jaws. Edentulousness and placement of full dentures that need to be renewed at short intervals is an equally unappealing option.

In case reports it is established that only mechanic treatment or together with antibiotic treatment do not succesfuly result[38]. Also in our study extraction of all the primary dentition could not be prevented, and there was no chance to apply mechanic treatment. We believe that the parents natural behavior for early teeth losses and late

application have also a role in this result. By the prosthetic treatment the patients function, foundation, aesthetic and psychologic needs were tried to be encountered. The patients long term follow up trials are going to be continued and the findings will be shared with our association.

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