

Papillon-Lefevre Syndrome

Marie A Congiusta, Donghyun Koo, Bapanaiah Penugonda, Arthur D Goren, Angela R Kamer

ABSTRACT

Papillon-Lefevre syndrome (PLS) is a genetically inherited disorder characterized by palmoplantar hyperkeratosis and severe early onset periodontitis. Oral symptoms begin to appear when primary teeth are in the process of erupting. A defect in the cathepsin C gene causes a dysfunction of the immune response occurring 1 to 3 times in 1 million births. Varying degrees of severity have been reported, ranging from complete edentulism at a young age to complete retention of teeth with a good prognosis.

Early intervention and proper treatment has been shown to improve prognosis. The pathogenesis of PLS is not clearly defined and has been associated with microbiologic, immunologic and genetic factors. Treatment includes conventional periodontal therapies, strict recall and maintenance, as well as antibiotics, metronidazole and chlorhexidine prescriptions as needed. Concurrently, patients are typically under the care of a dermatologist and taking retinoid medications to control their skin condition. This case report discusses the ongoing care of a patient who first presented to NYU College of Dentistry in the pediatric clinic at age 4. It is suggested that periodontal treatment initiated early in life may significantly change the prognosis for the better.

Keywords: Papillon-Lefevre syndrome, Cathepsin C gene, Periodontitis, Hyperkeratosis, *Actinobacillus actinomycetemcomitans*.

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INTRODUCTION

Papillon-Lefevre syndrome (PLS) is an autosomal, recessive genetic disease that occurs very rarely and is most often seen in offspring of consanguineous unions. This syndrome was first discovered in 1924 and described by two French physicians Papillon and Lefevre. The disease is characterized by severe early onset periodontitis and hyperkeratosis of the palms and soles. The causes attributed to this syndrome are multifaceted and have been associated with a mutation of the cathepsin C gene, which is directly related to immune cell function. Another factor that may also play a role in the initiation and progression of the periodontal aspect of the disease is the presence of certain anaerobic bacteria, in particular *Actinobacillus actinomycetemcomitans*, as has been reported in the literature.^{1,2} Many cases that have been reported discuss the importance of ongoing care in the management of the periodontal condition in order to preserve the permanent

dentition. Patients with PLS may begin to experience abscesses in the oral cavity as early as 1 to 4 months of age. Due to the severe inflammation, erythema, and mobility associated with this condition, these children suffer the loss of all their primary dentition. In cases left untreated or neglected, the loss of permanent teeth is likely to occur.

The psychological and social ramifications are rarely reported but cause a great deal of distress for the patient and their families. This report discusses the long-term care of an 18-year-old male patient of Indian descent who has been under continuous care at NYU College of Dentistry (NYUCD) since the early age of 4.

CASE REPORT/BACKGROUND

An 18-year-old male patient of Indian descent presented to NYUCD after a period of 2.5 years since his last visit. He was treated at NYUCD from the age of 4 and had been monitored throughout the years until 2009. Due to hardship and the loss of a parent, the patient did not return for treatment again until 2012 (Fig. 3D). When he returned in January 2012, his chief complaint was sensitivity of the lower anterior teeth (Figs 1 and 2). Aside from an established and confirmed diagnosis of PLS, the patients' medical history was unremarkable.

It was at the early age of 4 when the patient was referred to NYUCD by his private dentist. The chief complaint at that time was swelling and bleeding of the gums and loose teeth. Concurrently, he was under the care of a dermatologist and taking a retinoid medication via oral administration. Continual monitoring of liver enzyme functions via routine blood tests was reported, a precaution taken by

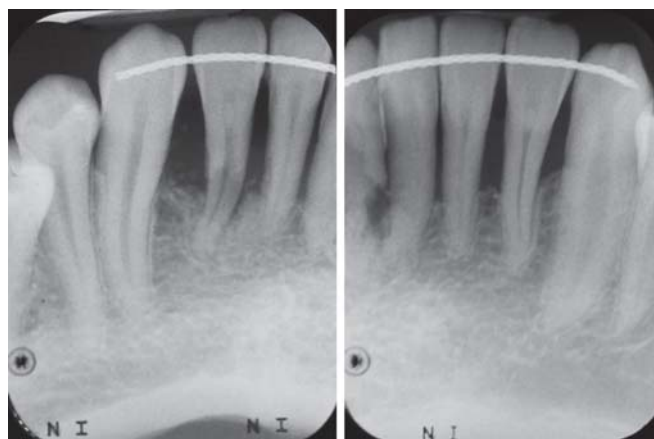


Fig. 1: Periapical X-ray of lower anterior teeth as patient returns to NYUCD after 2.5 to 3 years lapse in care (2012)



Fig. 2: The view (anterior) of patient upon return to NYUCD after 2.5 to 3 years lapse in care (2012)

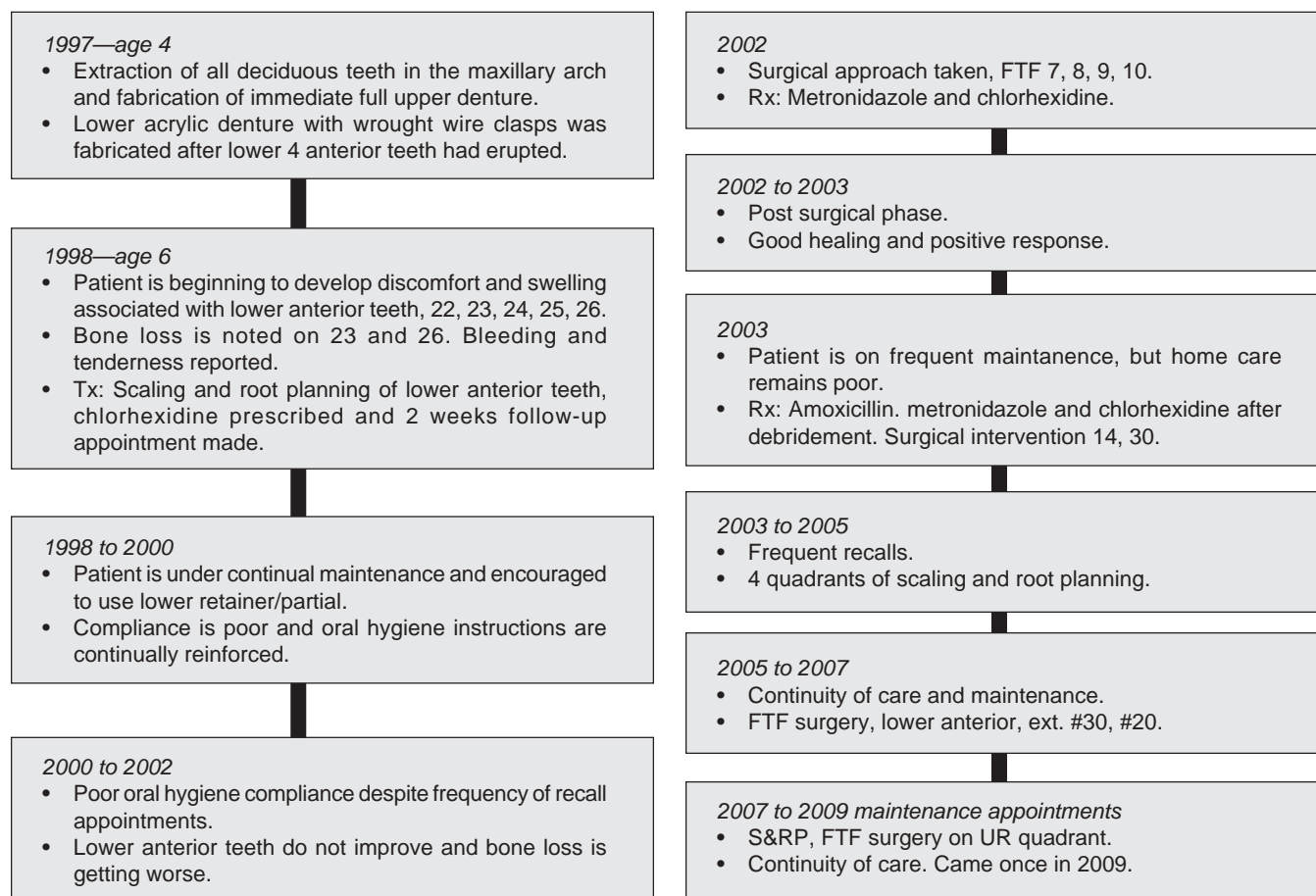
dermatologists to avoid the side effects of retinoid medication therapy. The history revealed that the palms of his hands, soles of his feet and knees had the skin manifestations of this syndrome, severe hyperkeratosis. His parents also reported an initial intraoral abscess at the age of 9 months.

Flow Chart 1 depicts the continuity of long-term care and maintenance provided to this patient at NYUCD recorded in his record. As noted in the patients dental chart, following periodontal evaluation it was determined that the

primary teeth needed to be extracted. He was provided with a full upper prosthesis and a lower acrylic partial denture that was designed around erupting lower anterior teeth. During this edentulous phase, the patient was asymptomatic and treatment was based on adjustments and relining of the dentures. At times, the patient had difficulty wearing the lower denture and was encouraged repeatedly to wear it in order to maintain arch space and form. Symptoms recurred with the eruption process of the secondary dentition (Fig. 3A).

Throughout his care, oral hygiene compliance was a struggle and constant reinforcement was necessary. Frequent recalls were needed and performed. Over time, progression of periodontitis was mostly apparent in the lower anterior teeth as well as the first molars. A review of the patients' record revealed that extractions of the lower anterior teeth had been advised by a periodontist when the patient was around 10.5 years old (2004) (Fig. 3B). However, his parents refused. Since then, these teeth remained carefully monitored and had undergone scaling and root planning, surgical intervention, antimicrobial therapy with amoxicillin, metronidazole, and chlorhexidine rinses. In order to minimize mobility, these teeth were eventually splinted.

Flow Chart 1: Exhibiting continuity of care since patient first presented to NYU college of dentistry (age 4-present)



TREATMENT AND CARE MANAGERMENTS

Treatment modalities rendered throughout this case included constant reinforcement in oral hygiene instructions, routine scaling and root planning, within a strict maintenance schedule with frequent recalls. Depending on the gingival appearance and plaque deposits, recall appointments ranged anywhere from 1 week to 4 months. A surgical approach was first used at age 10 and antimicrobial medications, with amoxicillin and metronidazole and chlorhexidine rinses were used several times at different stages of care. The periodontal condition, as well as plaque index were monitored regularly and recorded in the patient's chart. Tooth #30 and impacted tooth #20 were extracted between 2005 and 2007 (Fig. 3C).

This care continued until the lapse in care between 2009 and 2012. The patient is currently back on a strict recall schedule and is much more motivated about oral hygiene. Now that he is older and understands the severity of his condition, he is much more aware of how good oral hygiene and maintenance visits significantly impacts on the prognosis. It is our hope that this difference in commitment will positively influence the prognosis over time.

Carious teeth were immediately addressed and the patient was placed on a high caries risk protocol. Prevident 5000 plus was prescribed to use in place of his regular toothpaste. Tooth #14 had to be extracted due to endodontic and periodontal complications (Fig. 3D). The patient is undergoing thorough scaling and root planning at each recall visit and will be periodically monitored by a periodontist. A detailed demonstration of proper brushing and flossing techniques were reviewed. Homecare aids include superfloss, proxy brush and disclosing tablets to increase effectiveness of oral hygiene. A course of amoxicillin had been prescribed as well as metronidazole and chlorhexidine rinses. At this time, the patient is being seen once a week to address all his dental needs. At each visit oral hygiene is reassessed and discussed. The treatment plan includes consideration of replacement of missing teeth with several options under consideration and dependent upon the patient's financial means. To date, he has been compliant and has not missed any of his appointments.

DISCUSSION

As indicated in the literature, diagnosis is made based on the characteristic signs of PLS. Severe, early onset periodontitis and hyperkeratosis of the palms of the hands and the soles of the feet are the cardinal signs of this disease. The combined efforts of a dentist and a dermatologist are crucial to the successful outcome of a case. With the use of



Fig. 3A: Panoramic radiograph (2000)



Fig. 3B: Panoramic radiograph (2004)



Fig. 3C: Panoramic radiograph (2007)



Fig. 3D: Panoramic radiograph (2012)

retinoid medications, the skin condition improves remarkably, and patients seem to get better with age⁶ (Figs 4A and B). As per the periodontal condition, much of the literature attributes successful outcome to early diagnosis and treatment interventions.^{4,5,9} Extracting periodontally hopeless teeth in the primary and mixed dentition stage has been shown to improve future prognosis of the permanent



Fig. 4A: Hand (2012)



Fig. 4B: Foot (2012)

dentition.³ Continuous monitoring and frequent recall appointments has been shown to minimize periodontal deterioration.^{7,8} Some of the literature implies a possible benefit from the long-term use of retinoid medications used in treating the hyperkeratosis.⁶ However, further research is needed in this area.

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