Case Report

Prosthodontic Management of Papillon-Lefevre syndrome with special focus on its characterization and diagnosis: A Clinical Report

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Abstract:
Papillon-Lefevre syndrome is a rare autosomal recessive disorder and is characterized by the diffuse palmoplantar hyperkeratosis with rapid destruction of the periodontal support of both the primary and secondary dentition. Patient is often completely or partially edentulous at the stage of primary (4-5 yrs) and permanent dentition (13-15 yrs). Though, exact etiopathogenesis is unknown; three major factors have been identified as accountable for its initiation and progression. These are hereditary, immunological and microbial factors. Total rehabilitation in such cases is often difficult for health care professionals. However, correct diagnosis, perfect treatment planning and appropriate prosthodontic rehabilitation of such unfortunate individuals can improve their oral functioning, esthetics and overall social life. Here is a clinical report of such a rare condition with special focus on its diagnosis, characterization and rehabilitation by removable prosthodontics.
Keywords: Papillon-Lefevre syndrome, Palmoplantar keratoderma, Intracranial calcification, Prosthetic rehabilitation.

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Introduction

The Papillon-Lefevre syndrome was first described in 1924 as a rare autosomal recessive genetic disorder by two French physicians Papillon and Lefevre.[1] Characteristically it is a diffuse hyperkeratosis of palm and soles with severe periodontitis. Prevalence of 1-4 cases per million and carrier frequency of 2-4 per 1000 population is reported with equal male and female predilection and no racial predominance.[2] In literature various terminologies have been used for PLS; Palmar-plantar hyperkeratosis with severe periodontal dystruction involving both primary and permanent dentition, Palmoplantar keratoderma with periodontitis, Keratoris palmoplantaris with periodontopathia, Hyperkeratosis palmoplantaris with periodontitis.[3]

In 1964 Gorlin et al added the feature of calcification of falx cerebri as its third dimension, converting it into a triad.[4] Moreover, numerous erythematous keratotic plaque may be found on palms and soles with foul smelling pus discharge.[5] Oral manifestations of PLS include severe gingivitis, periodontitis,

halitosis and high risk of superadded infections because of decreased neutrophils and lymphocytes or monocytic function. One classical finding in PLS is that both the dentitions are affected. The primary dentition is lost by 5 years & permanent dentition is lost by 15 years.[6] This means that after 14 years, the patient is totally edentulous & is rendered helpless & totally handicapped.[7-8] The development and eruption of primary dentition is frequently associated with gingivitis and periodontitis. The classic treatment regime for such periodontal condition is seems to be unresponsive and often leads to the premature exfoliation of primary dentition. The gingival and periodontal pathologies usually disappear until the permanent teeth erupt and typically being reappeared with the eruption of permanent dentition resulting into complete or partial edentulousness.[9] Generally the patient consults dentist first because of premature teeth loss and associated problems. This clinical report describes such a rare condition with special attention on its diagnostic characterization, various treatment options and Prosthodontic rehabilitation.

Etiopathogenesis

Although the exact etiology and pathogenesis of this syndrome remains relatively difficult to understand; three major bases (immunologic, microbiologic, and genetic) have been suggested in the literature.

a) **Hereditary**: The loss of functional mutations of the lysosomal protease cathepsin C gene, located on chromosome 11q14.1-q14.3 is recognized in the disease progression of PLS.[10,17-18] The cathepsin C gene is expressed in the integument system such as palms, soles, knees, and keratinized oral mucosa, and is also expressed in various immune cells, including polymorphonuclear leukocytes (neutrophils), macrophages, and their precursors.[11] In addition, every patient of PLS is reported to be having homozygous alleles for the identical cathepsin C gene mutation inherited from a particular ancestor. However there are case reports regarding the delayed onset of PLS without causal cathepsin C gene mutation.[12]

b) **Immunological:** Reduced immunologic response and associated increase susceptibility to infection may be attributed to the decreased phagocytosis, and impaired reactivity to T and B cell mitogens.[10]

c) **Microbiological:** Gram-negative microbes are known to be primary factors in the etiopathogenesis of periodontitis. The elevated antibody titers to Actinobacillus actinomycetemcomitans and Capnocytophaga in PLS patients proves the involvement of this bacterium in periodontal pathologies associated with PLS.[10,13]

Clinical Report

A male patient aged 40 years, reported to the dental office with the chief complaint of multiple missing teeth in the upper and lower jaw and inability to eat food properly. Hence the patient showed enthusiastic desire for artificial replacement of lost teeth.

Diagnostic considerations of Papillon-Lefevre syndrome

a) **History**

Clinical history revealed that patient had normal eruption of primary dentition at 7-8 months of age. At 3 years of age, teeth became loose, mobile and started exfoliating. Patient was completely edentulous at the age of 4.5 years. No other family member suffered from such condition. At the age of 8 months, multiple erythematous areas on the palm and soles were first noticed by his mother. Patient also observed mobility and loosening of permanent dentition at the age of 11 years,

and most of them were lost by the age of 13 years. Mother had absolutely normal and uneventful delivery.

b) Clinical signs and symptoms

The overall body built, gait and mental status of patient was normal. Hyperkeratinized skin lesions with reddening started on elbows and knees first and then gradually involved palms and soles. Dermatological examination revealed rough, scaly and dry skin. Development of hairs and nails was normal. (Figure 1)

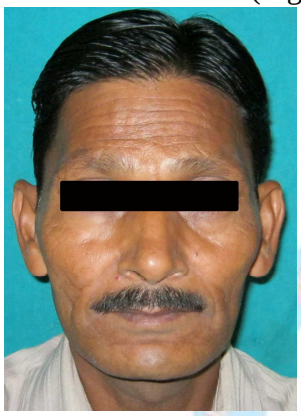

Figure 1: Pre-treatment frontal view

c) Extraoral examination

The palmoplantar lesions were mostly symmetric, grayish-white, keratinized well demarcated and consolidated patches affecting elbow, knees, palms, soles and even dorsum of hand and feet. Numerous suppuration areas with foul smelling pus discharge have further worsened the conditions especially in winters. (Figure 2, 3 and 4)

d) Intraoral examination

Early loss of most of the permanent tooth observed except maxillary left third molar, mandibular right canine, first premolar and mandibular right first premolar and they showed grade III mobility with heavy deposits of plaque and calculus. Deep, generalized periodontal pockets measuring about 5-6mm were seen. The upper and lower alveolar ridges were highly resorbed resulting into decreased occlusal vertical dimension of occlusion and so called "denture face" appearance. Gingival bleeding after brushing and halitosis was one of the most troublesome features. (Figure 5)

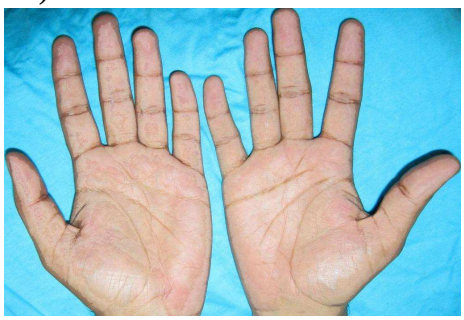

Figure 2: Hyperkeratinization with keratotic plaque on the palmer surface of hand

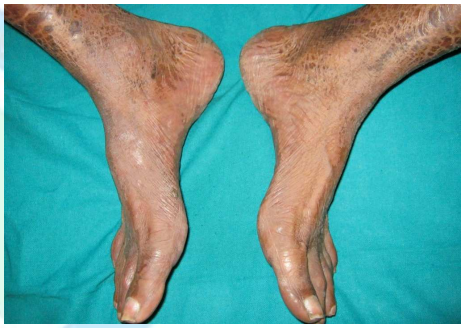

Figure 3: Obliterated arches of feet with thickening and keratinization of planter skin

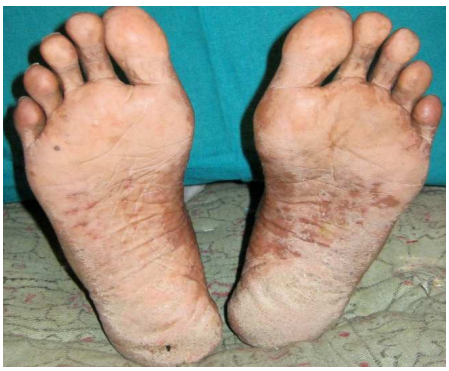

Figure 4: Keratotic plaque and hyperkeratinization of the palnter surface of feet

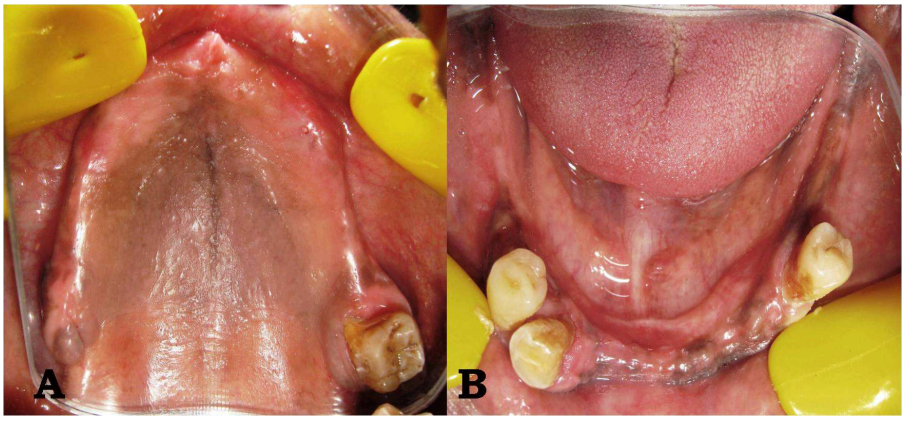

Figure 5: Intraoral view showing shrunken maxillary arch and highly resorbed mandibular arch with few remaining teeth and gingival inflammation (A and B)

e) Radiographic examination

The extraoral radiography (Panoramic and Cephalometric) confirmed generalized alveolar bone loss. The characteristic radiographic hallmark "floating in air" or "teeth in space" appearance is well appreciated and may be attributed to the extensive loss of bone support around the remaining teeth. Moreover, the evidence of intracranial calcifications on skull radiograph led down the final diagnosis of Papillon-Lefevre syndrome. Hand wrist radiograph showed that the metacarpal index was 9.3, advocating elongation of the metacarpals.[14] (Figure 6,7 and 8).

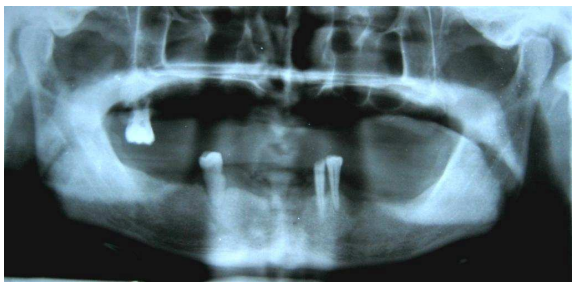

Figure 6: OPG showing multiple missing permanent teeth with generalized horizontal bone loss giving the 'floating in air' appearance

f) Laboratory investigation

Laboratory investigations were carried out to rule out the hematological and biochemical changes. The neutrophils and lymphocytes were reported to be lowered and may be correlated to the extensive periodontal tissue destruction (defective neutrophil/lymphocytic chemotaxis). Furthermore, the high serum alkaline phosphates level was indicative of increased periodontal destructive activity.

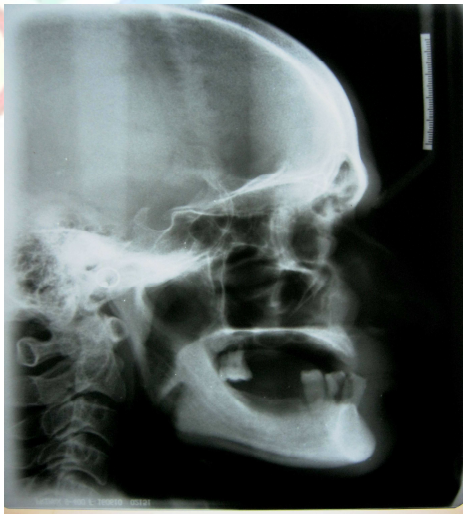

Figure 7: Lateral cephalogram showing no radiographic evidence of intracranial ossification

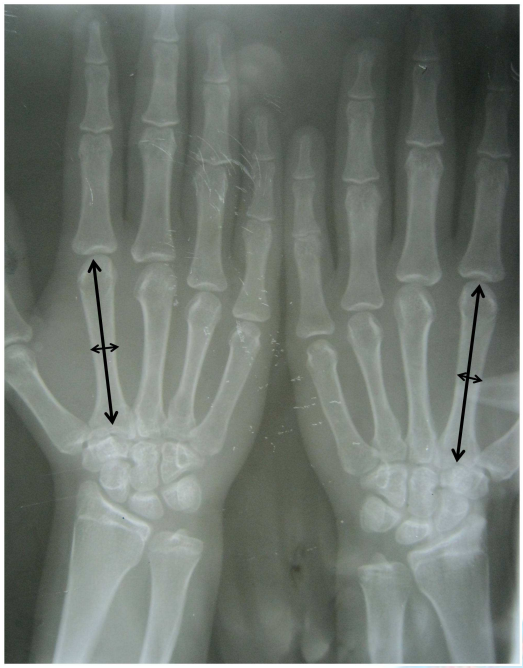

Figure 8: Hand wrist projection confirming elongations of the metacarpals

g) Histopathological examination

Superficial plantar incisional biopsy showed marked hyperkeratinization with localized parakeratosis and acanthosis. (Figure 9)

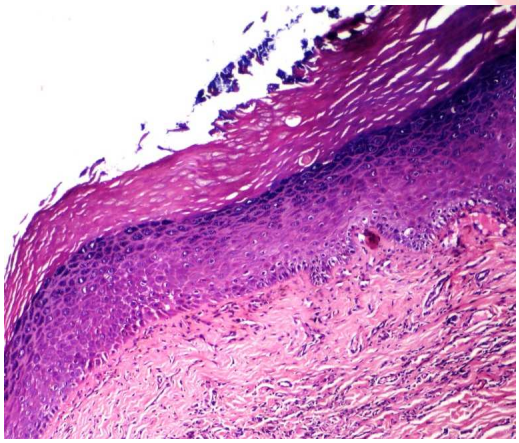

Figure 9: Histological examination of palmer skin confirms hyperkeratinization

Management

A multidisciplinary treatment approach involving the dermatologist, periodontist, and prosthodontist is necessary for the overall care of patients with PLS.

A) Symptomatic treatment

Maintenance of oral hygiene with concomitant use of prophylactic antibiotics may be acclimatized to minimize the risk periodontitis and subsequent loss of teeth. Eradication of A. actinomycetemcomitans with Trimethoprim and sulfamethoxazole has been shown as a successful treatment regime.[10] For the treatment of dermal keratosis, administration of anti-inflammatory emollients and keratolytic agents, such as topical steroids and salicylic acid are very effective. The use of Oral retinoids and Etretinate (an ethyl ester of acitretin) has also been reported to be effectual in patients with PLS.[15,16]

B) Prosthodontic rehabilitative procedures

Considering the overall scenario, it was deemed necessary to fulfill his esthetic and functional demands as a priority. Early rehabilitation affords the patient to develop normal social life, improved esthetic and function and facial support. Various treatment modalities for restoring the missing tooth were thought viz; Dental Implant supported/retained complete denture, Implants and natural teeth supported complete denture, Telescopic dentures, Over dentures with and without metal capping with minimal invasive reduction of remaining teeth, Extraction of all teeth followed by Complete Denture or Extraction of mobile teeth followed by Removable Partial Denture (in hopeless situations). Implant supported/retained complete denture and Telescopic dentures were ruled out as the patient could not bear the expenses. Considering the overall state of affairs, rehabilitation with maxillary and mandibular overdentures (with metal

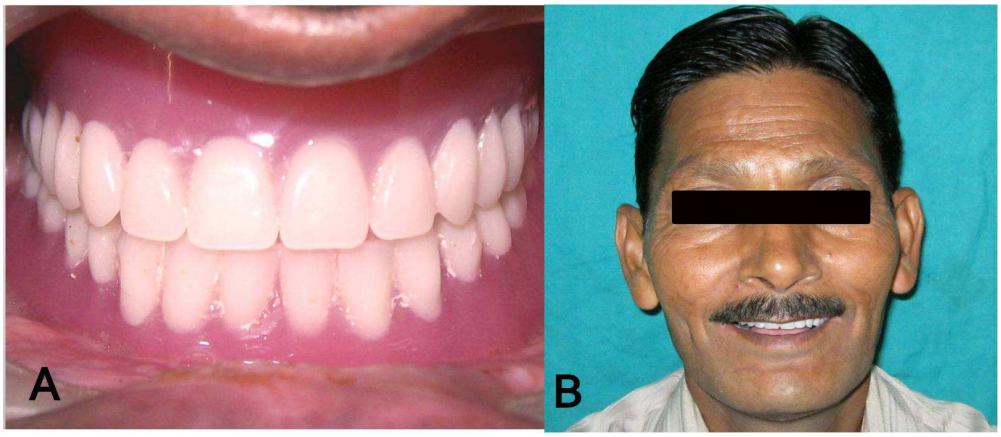

Figure 10: Complete denture in centric occlusion and post insertion extra-oral view of the patient (A and B)

coping) was planned. Pretreatment evaluation of remaining dentition and necessary preventive measures (oral prophylaxis; scaling) was completed prior to the restorative phase.

Upon root canal treatment of remaining teeth, crown preparations were completed by placing the chamfer finish line approximately at gingival level. Primary impressions were made using irreversible hydrocolloid (Alginate, Dentsply India). Special trays were fabricated on the primary casts with double thickness wax spacer. The definitive (final) impressions were made in special tray using medium viscosity elastomer (Type II Addition Silicon, Reprosil, Dentsply India). Standard clinical protocol was followed during jaw relation procedure and the casts were mounted on the semi adjustable articulator (Hanau Wide-Vue™). Teeth arrangement was done with characterization to give a refreshing new smile. Metal copings were cemented on teeth with Glass inomer cement (Luting GIC; Shofu Japan). Denture try-in was completed and dentures were fabricated in high impact heat polymerized acrylic resin. After finishing and polishing the dentures were inserted (Figure 10).

Patient was explained in detail regarding the slight discomfort he might experience in initial days due to the bulk of the denture; subsequently patient was recalled for post insertion checkups and necessary adjustments.[17]

Discussion

"Papillon Lefevre Syndrome" is one condition in which there is premature loss of the teeth affecting both the dentition. The exact etio-pathogenesis are still unknown, most likely genetic predisposition and a multifactorial etiology such as microbial infection and immunologic response are considered. There are other clinical conditions where individual presents with the similar dilemmas e.g. Haim-Munk syndrome (HMS). HMS has been described as an autosomal recessive genetic disorder characterized by congenital palmoplantar keratoderma and early onset periodontitis.[18-20] Moreover, this syndrome has been described as an allelic variant of cathepsin C gene.[21-23] In addition to Palmoplantar keratoderma with periodontitis, patients suffering from HMS experiences other symptoms that includes; recurrent pyogenic skin infections, acro-osteolysis and atrophic changes in nails. Radiographic examination of HMS patient

typically confirms the deformity of tapered, pointed phalangeal ends, claw-like volar curve, and pes planus.[24] The present clinical report showed classical features of PLS. These additional findings of HMS prevented the clinician from diagnosing the PLS as HMS. General Treatment modalities available for the management of such cases largely depend on the dentition status (primary/permanent) of the patient.[25] Considering the pattern of missing teeth and the available alveolar bone, the ideal treatment option in such case would be implant supported overdenture. Different studies have shown that Implant placement is associated with preservation of alveolar bone. However if bony atrophy progresses to the extreme in already alveolar deficient patients, implant placement may not be possible without bone grafting. The present case was belonging to a low socioeconomic group; therefore a conventional tooth supported overdenture was fabricated for functional and esthetic rehabilitation.

Conclusion

The nature of PLS has been described with special focus on its characterization, diagnosis and management of dental complications. As the PLS debilitates patient socially, psychologically and physically, the oral rehabilitation in such patients can improve the patients appearance, masticatory efficiency and minimize the onset of emotional and psychological problems often experienced by the these patients. Prosthodontic rehabilitation is a specialty job involving initial replacement with complete or partial denture with future consideration for implant-supported or retained definitive prosthesis. Considering the poor alveolar bone support and socioeconomic reasons, removable prosthetic replacement was executed as it would achieve patient's esthetic and functional demands immediately. Therefore accurate diagnosis, perfect treatment planning and suitable prosthodontic rehabilitation of such unfortunate individuals can improve their oral functioning, appearance, self confidence and minimize the onset of emotional and psychological problems.

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