A Case Report of Papillon - Lefevre Syndrome

Shah P P, Shah M J**, Parikh K K**, Khan F F*****

Papillon-Lefevre syndrome is a rare autosomal recessive genetic disorder in which there is a palmoplanter keratinisation and premature loss of both deciduous and permanent teeth. Consanguinity of parents is evident in about one third of cases. Early diagnosis of the syndrome can help to preserve the teeth by early institution of treatment, using a multidisciplinary approach. Here we describe a case of Papillon-Lefevre syndrome along with the clinical features, differential diagnosis and management of this syndrome are discussed.

Keywords: Papillon-Lefevre syndrome, palmoplanter keratinisation consanguinity.

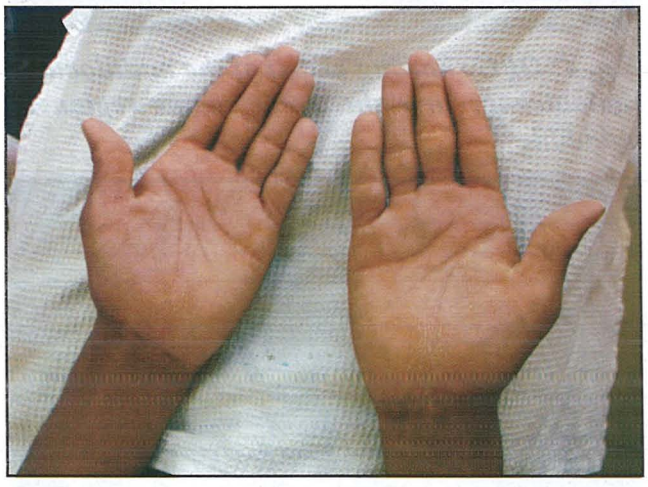


Fig. 1: keratotic plaques on palms.



Fig. 2: Keratotic plaques on soles.

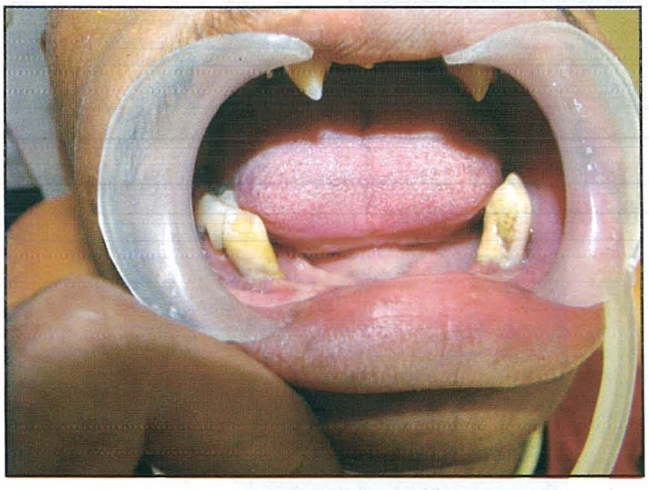


Fig. 3: Lower jaw showing severe periodontitis and exfoliated anterior teeth.

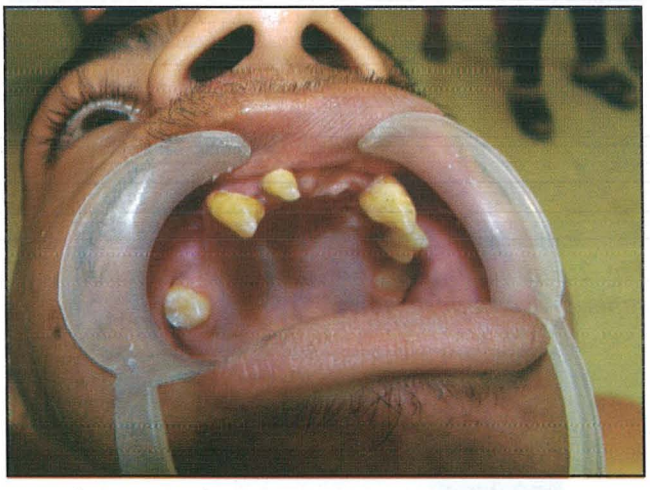


Fig. 4: Upper jaw showing severe periodontitis and exfoliated anterior teeth.

**Head of the Department and Reader.*
***Reader.*
****Senior Lecturer.*
*****Tutor, Department of Oral Medicine and Radiology, Faculty of Dental Science, Dharmsinh Desai University, Nadiad, Gujarat, India.*

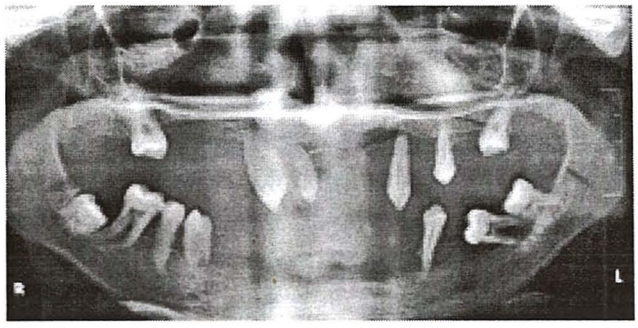


Fig. 5: OPG showing severe bone loss and loss of multiple teeth.

Papillon-Lefevre syndrome (here after known as PLS) is a rare autosomal recessive disorder characterized by hyperkeratosis of palms and soles and severe destructive periodontal disease affecting both the primary and permanent teeth[1]. It was first described by two French physicians (Papillon and Lefevre in 1924[2-6]. Some patients manifest generalized hyperhydrosis, very fine body hair and a peculiar dirty colored skin. These features are reminiscent of hereditary ectodermal dysplasia[3]. Other features of the syndrome which have been reported less frequently include psoariasiform plaques of the elbows and knees, nail changes, calcification of the dura and recurrent pyogenic skin infetion[6]. Pyogenic liver abscess is an uncommon presentation[7]. It has prevalence of 1-4 cases per million person[5,6,8]. Males and females are equally affected and there is no racial predominance[6,8]. An increase prevalence of parental consanguinity has been reported in PLS patients[2,6,3,9,10]. Although the causes of PLS are not well understood, it has been reported that mutation of Cathepsin C gene located on chromosome 11q14 – q21 is responsible for PLS syndrome[3,4,8,10].

The palmoplanter keratoderma typically has its onset between age of one to four years. It involves the entire surface of the palms and soles, sometimes extending on to the dorsal surface of hands and feet. The symptoms may worsen in winter and be associated with painful fissures[4,7,11].

Oral manifestations consist of advance periodontitis that is seen in both dentitions. It starts at the age of three to four years. Development and eruption of deciduous teeth precede normally, but eruption is associated with gingival inflammation and rapid destruction of periodontium. The primary dentition is usually exfoliated prematurely by the age of four years. After exfoliation, inflammation subsides and gingiva appears healthy. With the eruption of permanent dentition, process of gingivitis is reported again followed by periodontitis and there is subsequent premature exfoliation of permanent teeth by the age of 13-16 years[3,8,11,12]. Severe resorption of alveolar bone gives the teeth a 'floating in air' appearance[2,4,13]. Gorlin et al have added the third feature of dura calcification[2,3].

Defective neutrophil chemotaxis is found to be responsible for extensive periodontal tissue destruction. Bacteria responsible are Actinobacillus actinomycetemcomitans and Capnocytophaga[4,9]. Histological features are same as in chronic periodontitis and are not specific[4].

A multidisciplinary approach is necessary in the management of patients with PLS. Skin manifestations are treated with emollients. Oral retinoids including acitretin, etretinate and isotretinoin are the mainstay of the treatment of both keratoderma and periodontitis associated with PLS[8]. The periodontal disease may be arrested by improving oral hygiene, extraction of severely diseased teeth, scaling, systemic antibiotics and long term antimicrobial irrigation[12]. The use of implants could considerably enhance future therapeutic options for severely dentally compromised patients.

The case reported here is 19 years old Male patient with oral and dermatological findings which is suggestive of PLS.

CASE REPORT

A 19 years old male patient reported to the department of Oral Medicine and Radiology, College of Dental Science, D.D.University, Nadiad with the chief complaint of premature loss of some permanent teeth, mobility in remaining teeth and discomfort in chewing. The past dental history revealed that his

deciduous teeth had erupted normally but exfoliated before eruption of permanent teeth. Similarly eruption of permanent teeth was normal. At the age of 14 years, gradually his permanent teeth had started to become mobile. Some teeth exfoliated by themselves, while some were extracted due to severe mobility. Family history was very contributory. There was a history of consanguineous marriage of the parents. Both parents were healthy. Patient gave history of consanguineous marriage of his two uncles. History revealed that his 25 years old cousin brother lost his all teeth at the age of 20 years, wore denture and reported to have hyperkeratotic lesions of palms and soles. His 14 years old cousin sister had similar clinical presentation of this condition.

His parents had noticed rough and dry skin on planter surface of his feet and palmer surface of his hands at the age of 1 year for which the parents did not seek any treatment. Since then there had been gradually increasing keratinisation of palmer and planter surfaces. At present, there were symmetric, well demarcated, yellowish, keratotic and confluent plaques affecting the skin of his palm and soles. The plaques were more prominent on the soles than palms. The lesion became worse during winters, with fissuring and bleeding from the fissures.

In intraoral examination it was found that only 12 teeth were remaining, which were 12, 13, 17, 23, 25, 27, 34, 37, 38, 44, 45, and 46. Grade III mobility was present in all the teeth except 17, 27 and 38. Gingiva in relation to the still existing teeth was red, soft and edematous, with deep periodontal pockets and bleeding on probing. Generalized recession was found. There was pathological migration of 25 and 37. Despite of all these features no local factors like plaque and calculus were very prominent. Oral hygiene was quite poor with halitosis. Mucosa over dentulous ridge was normal.

The Orthopantograph showed severe horizontal alveolar bone loss except in relation with 17, 27, 38 where bone destruction was less. The alveolar bone around the mobile teeth was devoid of definable lamina dura. Unerupted 47 was seen. An extensive alveolar bone loss up to the apical third region of roots gave the teeth a "Floating in air" appearance. Based on the history, clinical and radiological findings, diagnosis of Papillon Lefevre Syndrome was made. Consultation of a Dermatologist was taken for confirmation of diagnosis and treatment of skin lesions. Skin lesions were treated by emollients and Salicylic acid and capsule of Vitamin A and D was given. Extraction of teeth that had severe mobility with subsequent prosthesis construction was advised.

An interdepartmental approach of Department of Oral Surgery, Periodontics, and Prosthodontics was used for the rehabilitation of the patient.

DISCUSSION

Papillon Lefevre syndrome is a rare disorder of keratinization that is inherited as an autosomal recessive manner, that is both parents are phenotypically healthy[6]. In the presented case dermatological, periodontal and radiological features strongly suggested the diagnosis of PLS. Moreover consanguineous descent has been described for this syndrome[2, 12] and case reported here was associated with consanguinity of the parents. Phenotypically, the parents were healthy suggesting an autosomal recessive pattern of inheritance. Clinically patients had the characteristic skin lesions, sharply demarcated keratotic plaques involving palms and soles. The lesions were diffused, dry and rough on palpation, which worsens during winter. Intraoral findings were remarkable with the characteristic features of periodontitis, as has been described for PLS. Patient had multiple missing permanent teeth. Remaining permanent tooth exhibited marked mobility with all signs and symptoms of gingival inflammation. OPG of the patient showed extensive alveolar bone loss in almost all the teeth. All these dental findings and skin lesions are consistent with the features of PLS.

Multiple factors have been suggested as being responsible for the initiation and progression of PLS. An impairment of neutrophil chemotaxis, phagocytosis and bactericidal activities accompanied by decrease in cell migration was considered as one

of the etiological factors[6,11]. Present microbiological studies have demonstrated Actinobacillus actinomycetemcomitans in periodontal plaque and pocket. So it has been directly related to the periodontal destruction[2,4,6,10]

An increased susceptibility to bacterial infection, leads to recurrent pyogenic infection of skin and sometimes pyogenic liver abscess occur as a complication

It is caused by mutation and defect in Cathepsin C gene. This gene is important in the structural growth and development of skin and is critical for appropriate immune response of myeloid and lymphoid cells. Altered gene also affects the integrity of the junctional epithelium surrounding the tooth[4,6,12].

Gingival infection, abscess formation, loss of alveolar bone and destruction of the periodontal ligament are probably the etiologic factors responsible for the shedding of teeth[13,17].

Mutation in Cathepsin C gene also results in two other closely related conditions, the Haim-Munk syndrome and prepubertal periodontitis. Common clinical manifestation in all these syndromes is early onset of severe periodontitis[1,3,6,8].

Periodontium in HMS may be less severely affected than PLS. Although the palmoplanter findings and periodontitis are suggestive of HMS, the absence of other clinical features, like nail deformities and arachnodactyly prevented as from diagnosing it as HMS. Prepubertal periodontitis is differentiated from PLS by the absence of associated palmoplanter keratosis[3,8].

A definite treatment regime is not yet reported. Oral retinoids including acitretin, etretinate and isotretinoin are the mainstay of the treatment of both keratoderma and periodontitis associated with PLS. Treatment may be more beneficial if it is started during eruption and maintained during development of permanent teeth, but periodontitis in PLS is difficult to control[1]. To control periodontal destruction, several treatment modalities such as conventional periodontal therapy, oral hygiene instructions, systemic antibiotics, extraction of severely periodontally compromised teeth, can prolong the viability of unaffected teeth. In the future stem cell therapy can be expected to open up new vistas in the dental treatment of such patients[2]. Early extraction of teeth too has been advocated to prevent bone loss. This allows solid base for subsequent use of artificial dentures[13]. These dentures have to be frequently modified as the growth of the jaws take place. Implant supported prosthesis can be opted when the growth period is over[9]. Thus both the dermatologist and dental professional can help to save the permanent dentition if they diagnose this disease during childhood.

SUMMARY

PLS debilitates individual socially, psychologically and physically. Diagnosis of the syndrome by the dentist at an early age can help to preserve the teeth by institution of early treatment and restore aesthetics, and function of such unfortunate individuals. Treatment involves initial replacement with complete and partial dentures and future consideration for an implant supported prosthesis. In present case prosthetic rehabilitation provided immediate satisfaction to the patient in terms of aesthetic and function. Individual who have PLS may meet with a genetic counsellor to discuss the risks of having children with the disease as it is autosomal recessive inherited condition.

References

1. Kiran Ganji, Seetharam Kumar D. Papillon Lefevre syndrome. Indian journal of Dental advancements 2010 April-June 2(2) 217-219.
2. NB Nagaveni, R Suma, ND Shashikiran, VV Subba Reddy. Papillon Lefevre syndrome. Report of two cases in the same family. Journal of Indian Society of Pedodontics and Preventive Dentistry 2008,Vol 26, Issue 2, 78-81.
3. P Subramaniam, S Mathew, KK Gupta. Papillon Lefevre syndrome. A case report. Journal of Indian Society of Pedodontics and Preventive Dentistry 2008 Vol 26, Issue 4, , 171-174.
4. Neville, Damm.Allen Oral and Maxillofacial pathology, 3rd edition Publisher Elseveir page: 176-178.

5. Shafer, Hine, Levi. Shafer's Textbook of Oral Pathology 5th edition, publisher Elseveir. Page: 560-561.

6. Anshu Singla, Soheyl Shaikh, Sanjeev Kumar, Jindal, Rajdeep Brar Papillon Lefevre syndrome: Bridge between Dermatologist and Dentist. JClin Exp Dent 2010:2(1)e43-6.

7. S V Dhadke, P M Kulkarani, V N Dhadke, N S Deshpande, P R Wattamuar, JAPI (Journal of association of Physicians of India) March 2006,Vol 54; 246-247.

8. Shahbaz A Janjua MD; Amor Khachemoune MD; Papillon Lefevre syndrome: Case report and review of the literature Dermatology Online Journal 2004.10(1):13

9. Pritam Shah, Dayanad Shirol, Rahul Lodaya, Chetan Bhatt, Sachin C Gugwad. Rehabilitation of three years Papillon Lefevre syndrome using Telescopic dentures- a case report. Int. Journal of Contemporary Dentistry Sept.2010,1(1)68-70.

10. Varsha J Rathod, Nilesh V Joshi. Papillon Lefevre syndrome. Areport of Two cases. Journal of Indian Society of Periodontology year 2010 Vol 14, issue 4, 257-278.

11. Jignya Shah, Shweta Goel. Indian Jouranl of Dental Research ,Vol18, Issue 4, , 210-213.

12. Motamedi M, Lotfi A, Azizi T and Moshret M, Dentistry Today. Feb 2009,28(2)102, 104-5.

13. Mahajan Vk, Thakur NS, Sharma NL, Papillon Lefevre syndrome. Indian Pediatr 2003, 40:197-200.

P.F.A. Fellowship

PFA, with its Mentor Program, Leadership Conference, Hell of Fame, Foundation Scholarships, and lecture series, is an alive, growing group of professionals in their prime who are leaders.

The honour PFA bestows is not for past accomplishments alone. The actual "honour" is to be invited to join an organization of the world's top dental leaders, and to work even harder with allies compromising the finest dental talent on this planet. PFA is not a wall plaque or a lapel pin-gathering dead symbols. But the Fellowship calls to you, of proven worth, to step higher, unite with professionals you only demand of knowing, and fall in beside them as Fellows to labour for goals you could not imagine being accomplished but by the gods themselves. If you think only to mount your certificate in the office and rest on laurels, you miss the entire excitement of the honoured bestowed. Your laurels were only the basis for invitation to Fellowship. Your dues are not in money, but in continued activities on all fronts. Your "honour" must be reproved each term, else it becomes dust and forgotten.

Think of the Academy as an opportunity. Think about the Fellowship as a legion of generals, who call you equal, and stand ready to aid your highest aim for the profession. Think about how your talents, magnified by their combined experiences and contacts, can obtain the lofties goals. Use the honour to its fullest! That honour is the cherishment of knowing you in the hearts of your Fellows-in a living honour that far outlasts our mortal beings of paper certificates.

Corrigendum

Corrigendum to Vol. 26, Issue No. 1 Page 5 March 2012 issue

Please Read the footnote for author as under:

Read *P.G. Student instead of Professor and **Professor and Head, Department of Periodontics and Oral Implantology, SGT Dental College, Hospital and Research Institute, Gurgaon, Haryana-123505 instead of Senior lecturer, Department of Oral Pathology and Microbiology, Kothiwal Dental College and Research Centre, Kanth Road, Moramustaqeem, Moradabad-244001, Uttar Pradesh.

Inconvenience is regretted.

Chief Editor.