PAPILLON-LEFEVRE SYNDROME: A CASE REPORT

Sanchita Kundu[1] Shabnam Zahir[2]

[1]*Department of Oral and Maxillofacial Pathology.* [2]*Department of Pedodontics and Preventive Dentistry, Guru Nanak Institute of Dental Sciences and Research, Panihati, Kolkata.*

Corresponding Author: Sanchita Kundu, Associate Professor, Dept.of Oral and Maxillofacial Pathology, Guru Nanak Institute of Dental Sciences and Research,Panihati,Kolkata-700114. E-mail : sanchita_op@rediffmail.com.

Abstract

Papillon-Lefevre Syndrome is a very rare syndrome of autosomal recessive inheritance characterized by palmer-planter hyperkeratosis and early onset of a severe destructive periodontitis., leading to premature loss of both primary and permanent dentitions.Genetic studies have shown that mutations in the major gene locus of chromosome 11q14 with loss of function of cathepsin C gene are responsible for this syndrome. An early diagnosis and intervention can help to preserve teeth and prevent their premature exfoliation. A clinical case of Papillon- Lefevre syndrome is discussed herewith having all of the characteristic features.

Key Words: Papillon-Lefevre syndrome, palmoplanter hyperkeratosis, periodontitis, cathepsin c gene.

Introduction

Papillon Lefevre syndrome(PLS) is a rare autosomal recessive disorder characterized by the development of palmoplanter hyperkeratosis with precocious progressive periodontal disease that results in premature exfoliation of primary and permanent dentitions.[1,3,4,7,8] The disease was first described by two French physicians, Papillon and Lefevre in 1924. It has a prevalence of 1-4 cases per million persons and both males and females are equally affected with no racial predominance.[2,3,4]

The pathogenesis of PLS is not well understood. It has been reported that loss of functional mutation effecting both the alleles of Cathepsin C gene, located on chromosome 11 q 14-q 21 is associated with PLS.[2] This gene is important in the structural growth and development of the skin and appropriate immune response of myeloid and lymphoid cells.[3] The cause of the periodontal disease and increased susceptibility to infection in PLS is due to decreased neutrophil phagocytosis, bacterial infection and impaired reactivity to T and B cell mitogens.[1,2,7,8]

In PLS the palmoplanter keratoderma typically has its onset between the ages of 1 and 4 years. The sharply demarcated, erythematous keratotic plaques involve the entire surface of the palms and soles, sometimes extending onto the dorsal surface of the hands and feet.The lesions are punctate and diffuse, with dry scaly skin and vary in thickness from one to several millimeters. There may be associated hyperhidrosis of the palms and soles, which may cause a foul smell. These plaques may also be seen on the elbows and knees. The symptoms may worsen in winter and associated with painful fissures. Eyelids, cheeks, labial commissures, legs, thighs and axillae are also involved. The nails in advanced cases, may show transverse grooving and fissuring.[1] Periodontitis in PLS affects both the deciduous and the permanent dentition and develops soon after the eruption of the teeth.[3] The development and eruption of the deciduous teeth proceeds normally, but their eruption is associated with gingival inflammation and subsequent rapid destruction of the periodontium. The resulting periodontitis characteristically

unresponsive to traditional treatment modalities and the primary dentition is usually exfoliated prematurely by the age of 4 years.[2] After exfoliation, the inflammation subsides and the gingiva appears healthy. With the eruption of permanent dentition the whole process of gingivitis and periodontitis is repeated and there is subsequent premature exfoliation of the permanent teeth by the age of 13-16 years.[1,2] Severe generalized resorption of alveolar bone gives the teeth a "floating-in-air" appearance on dental radiograph.[1,4]

A multidisciplinary approach is important for the management of patients with PLS. The skin manifestations are usually treated with emollients and oral retinoids. The periodontal disease may be arrested by improving oral hygiene, extraction of severely decayed teeth, scaling, proper restoration, systemic antibiotics, construction of complete and partial dentures and long-term antimicrobial irrigation.[1]

Based on above clinic-pathological and radiological knowledge, a case of PLS is discussed herewith along with an overview of its treatment.

Case Report

A male child named Sayan Mazumder, aged 11 years, was reported to the department of Oral and Maxillofacial Pathology of Guru Nanak Institute of Dental Sciences and Research, Panihati, Kolkata with chief complain of premature exfoliation and mobility of permanent teeth associated with presence of scaly lesions of the hands and toes for the last 7 years.[Fig-1] The past dental history revealed that his deciduous teeth had erupted normally but exfoliated gradually by the age of 5-7 years. Permanent teeth were erupted normally but exfoliations of lower left central incisor along with mobility of multiple teeth were noted by the age of 9 years. Parents had noticed scaly lesions on the planter and palmer surfaces of the hands and feet by the age of 4-5 years. No significant family history was there regarding the same complaint. Intraoral examination revealed the presence of missing/exfoliated lower left central incisor along with mobile lower right and left lateral incisors, right central incisor and all 1st molars. Gingiva was red and soft with presence of deep periodontal pockets. The mucosa in other areas of the oral cavity appears to be normal.[Fig-2] Dermatological examination revealed the presence of symmetrical , well demarcated, yellowish keratotic and confluent plaques affecting the skin of the palms and soles, extending up to the dorsal surfaces of hands and feet.[Fig-3,4,5]

OPG of the patient showed severe loss of alveolar bone in relation to all the existing teeth, giving the teeth a 'floating-in-air" appearance.[Fig-6]

After considering the clinical and radiological features, a diagnosis of PLS was made and the patient was referred to the Dept. of Pedodontics and Preventive Dentistry for further treatment. Skin manifestations are usually treated by dermatologists with emollients and oral retinoids.

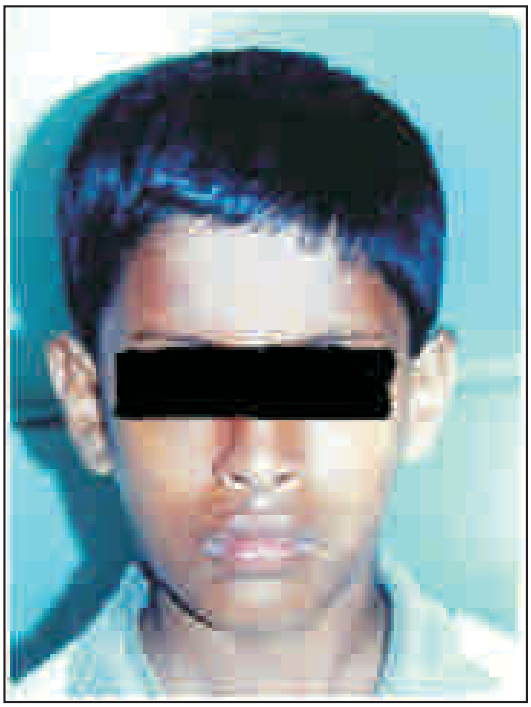

Fig 1: Extra-oral photograph of the patient.

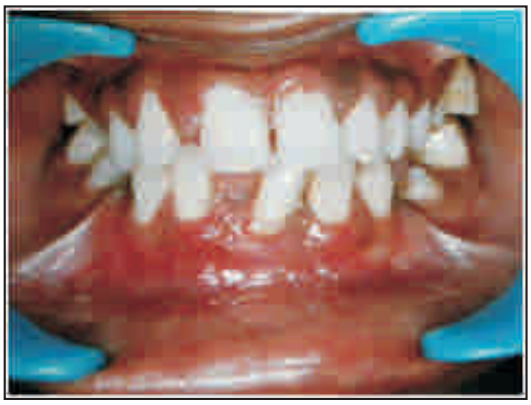

Fig 2: Intra-oral photograph showing exfoliated lower left central incisor along with presence of spacing and poor oral hygiene.

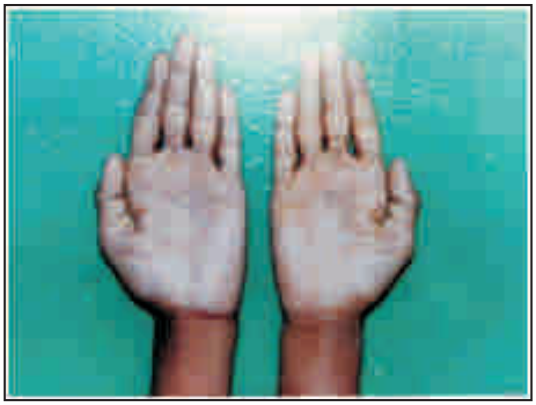

Fig 3: Extra-oral photograph showing scaly lesions over the palms

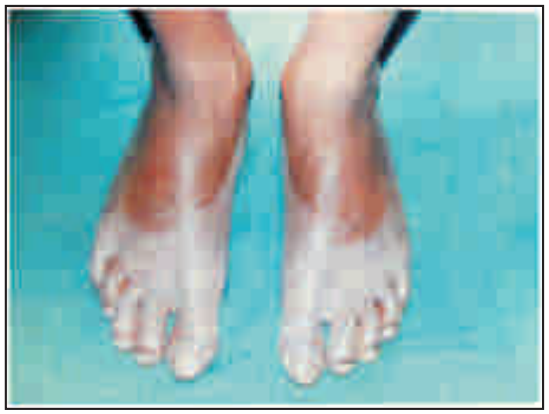

Fig 4: Extra-oral photograph showing scaly lesions over feet.

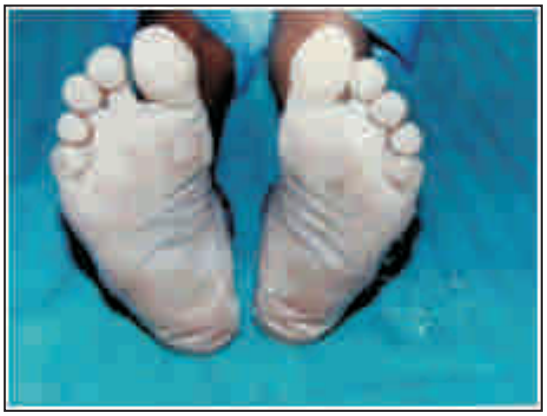

Fig 5: Extra-oral photograph showing scaly lesions over the sole.

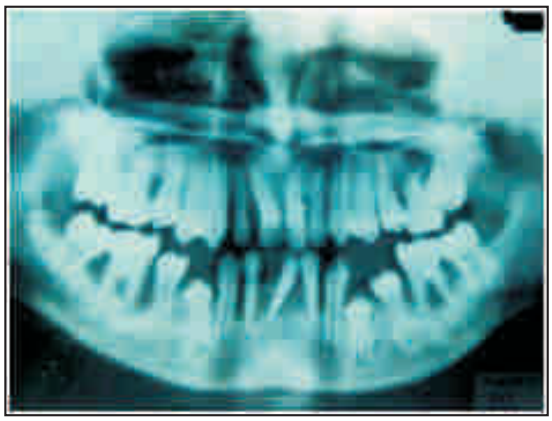

Fig 6: Panoramic radiograph showing severe loss of alveolar bone in relation to all of the existing teeth, giving a typical "floating-in-air" appearance.

Discussion

PLS is a autosomal recessive disorder, predominantly demonstrate oral and dermatologic manifestations. Because of the autosomal recessive nature, the parents typically are not affected. The predominant oral finding is accelerated periodontitis involving the entire dentitions caused by defects in neutrophil function and multiple immune-mediated mechanisms and develops soon after the eruption of the teeth.[3]This disease is associated with defective production of Cathepsin C. The gene for Cathepsin C lies on chromosome 11.[1,2] In the present case, phenotypically the parents were healthy and there was no family history of the disease, suggesting an autosomal recessive pattern though genetic testing could not be

performed due to the low socio-economic status of the parents.

The patient under discussion was a 11 year old male child, having premature exfoliation and mobility of permanent teeth associated with presence of scaly lesions on the hands and toes for the last 9 years. Early exfoliation of all deciduous teeth were started at 3 years of age and completed within 2-3 years. These clinical findings are consistent with the clinical observations of PLS reported by the earlier clinicians.[1,2,3,4]

The conventional radiographic findings usually include severe generalized resorption of alveolar bone which gives the teeth a "floating-in-air" appearance. The panoramic view of the present case was thus very characteristic of PLS.[1,4]

Treatment of PLS is more beneficial if started more earlier during the eruption of deciduous teeth and maintained during the development of the permanent teeth. Frequent oral prophylaxis, oral hygiene instructions and antibiotic therapy can, at least, only delay the shedding of teeth. Early extraction of mobile teeth has also been advocated to prevent bone loss and allow preservation of a solid base of an artificial denture.

The entire gamut of discussion involving clinical and radiological features of PLS thus explains the diagnostic, treatment and management modalities as performed in the present case.

Conclusion

The PLS debilitates individuals socially, psychologically and physically. Early diagnosis of the disease and proper intervention is very much essential to prevent edentulism of the child patient.

References

1. *Shah J, Goel Shweta. Papillon-Lefevre Syndrome: Two case reports. Indian J Dent Res 2007;18(4):210-3.*
2. *Subramaniam P, Mathew S, Gupta K K. Papillon-Lefevre Syndrome: A case report. J Indian Soc Pedod Prev Dent 2008;26:171-4.*
3. *Neville BW, Damm DD, Allen CM, Bouquot JE. Papillon-Lefevre Syndrome. Oral and Maxillofacial Pathology, 3rd edition. Noida, Elsevier Inc, 2009;176-8.*
4. *Nagaveni NB, Suma R, Shashikiran ND, SubbaReddy VV. Papillon-Lefevre syndrome:Report of two cases in the same family. J Indian Soc Pedod Prevent Dent 2008;:78-81.*
5. *Rathod VJ, Joshi NV. Papillon-Lefevre syndrome:A report of two cases. J of Indian Soc of Periodontology 2010;14:275-8.*
6. *Dhadke SV, Kulkarni PM, Dhadke VN, Despande NS, Wattamwar PR. Papillon Lefevre syndrome. JAPI 2006;54:247-7.*
7. *Mahajan V.K, Thakur N.S, Sharma NL. Papillon-Lefevre syndrome. Indian Pediatr 2003;40:1197-200.*
8. *Rathi SK. Papillon-Lefevre syndrome in two brothers. Indian J Dermatol Venerol Leprol 2002;68:155-6.*
9. *Janjua SA, Khachemoune A. Papillon-Lefevre syndrome:Case report and review of the literature. Dermatol Online J 2004;10:13.*
10. *Galanter DR, Bradford S. Case report. Hyperkeratosis palmoplantaris and periodontosis: The apillon-Lefevre syndrome. J Periodontol 1969;1:40-7.*

Source of Support - Nil

Conflict of Interest - None declared

How to cite this article:

Sanchita Kundu,Shabnam Zahir: Papillon-Lefevre Syndrome: A Case Report, Oral Max Path J, 3(2), Jul-Dec 2012: 269-272