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PAPILLON-LEFEVRE SYNDROME – A CASE REPORT

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ABSTRACT
Papillon-Lefevre Syndrome (PLS) is a rare autosomal recessive disorder characterized by the alert of palmoplanter hyperkeratosis with precocious progressive periodontal disease that results in premature exfoliation of primary and permanent dentition. Dentist play a significant role in the diagnosis and management of PLS as there are characteristic manifestations like periodontal destruction at an early age and an early eruption of permanent teeth. This paper presents a clinical presentation of a 12 year old male diagnosed with Papillon-Lefevre Syndrome.

KEYWORDS: Hyperkeratosis, Periodontitis, Papillon-Lefevre Syndrome.

INTRODUCTION
The Papillon-Lefevre Syndrome was first described by two French physicians Papillon and Lefevre in 1924. 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. Calcification of the fax cerebri and choroid plexus and retardation of somatic development is often an associated feature. It has been suggested that 20-25% of patients show an increased susceptibility to infections of which Otitis Media is a common example.[1]

It usually manifests during the first 4 years of life with sharply demarcated hyperkeratosis, more pronounced on the soles of feet and possibly extending to the dorsa of the hands and feet. Erythromatous hyperkeratotic plaques may also be present at the elbows, knees and trunk. The second major feature of PLS is severe periodontitis which starts at the age of 3 or 4 years and affects both the deciduous and permanent teeth.[2]

In PLS the eruption of teeth occurs at the expected ages and in the normal sequence with the teeth being of normal form and structure. Eruption of primary dentition into the oral cavity is accompanied by severe gingival inflammation and a generalised aggressive periodontitis, resulting in tooth mobility. By the age of 4 or 5 years, the primary teeth frequently become loose and exfoliate. As the permanent teeth erupt the same sequence of events occur and without intervention, most of the permanent teeth are lost by around 15years of age. Severe resorption of alveolar bone gives the teeth in a "Floating-in-air" appearance on dental radiographs. This paper presents a case report of a 12 year old male patient diagnosed with Papillon-Lefevre Syndrome.[3]

CASE REPORT
An 12 year old male patient reported to the Department of Pedodontics, with the complaint of loose teeth and discomfort of chewing. On general examination, patient had overall normal physical and mental development. Patient medical history is non-contributory and the familial history revealed consanguineous marriage of the parents. Pregnancy and delivery were normal. Extraoral examination revealed hyperkeratosis of the palms, soles and the knees of both the limbs and affected skin. Intraoral examination reveals, the permanent dentition with severe gingival inflammation, deep periodontal pockets, loss of upper anterior teeth and lower left central incisor with severe mobility affecting all the remaining teeth with deposits of calculus and plaque and halitosis were present. On orthopantomograph examination showed severe generalised destruction of alveolar bone. In view of the above findings the case was diagnosed as Papillon-Lefevre Syndrome. Treatment by extraction of all teeth, followed by complete denture fabrication was planned to restore masticatory function.

Fig-1: Palmer hyperkeratotic lesion.

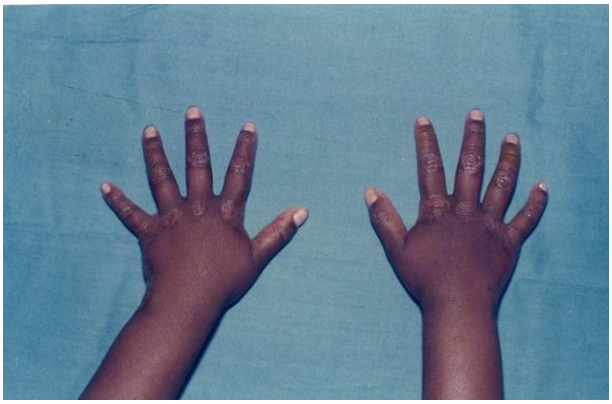

Fig-2: Hyperkeratosis on the dorsal surface of both the hand.

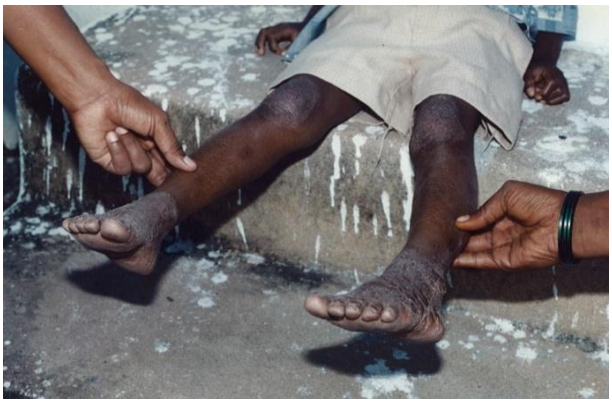

Fig-3: Planter hyperkeratotic lesion.

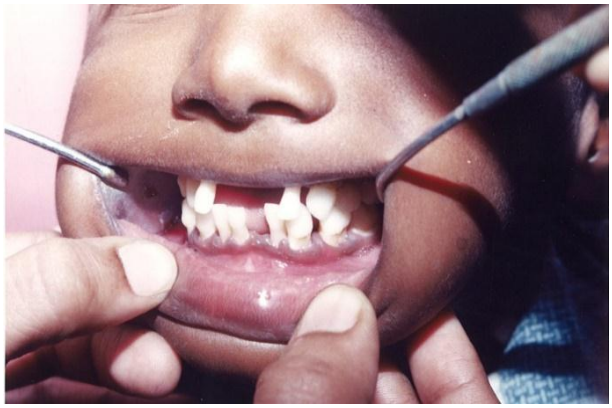

Fig-4: Premature exfoliation of Maxillary & Mandibular permanent teeth.

DISCUSSION

Papillon-Lefevre Syndrome is characterised by diffuse palmoplantal keratoderma and rapidly progressing periodontitis leading to premature loss of both primary and permanent dentition. It is an autosomal recessive inherited disorder of keratinization, characterized by redness thickening of soles and palms and severe destructive periodontal disease affects all the teeth caused by mutation in Cathepsin 'C' (CTSC) gene. Other symptoms include heperhidrosis, arachnodactyly, intracranial calcification, increased susceptibility to infections and mental retardation.[4] The pedigree study reveals the mode of inheritance of disease. PLS is inherited as an autosomal recessive disorder if both parents are carriers of the defective gene there is a 25% risk for their children to be affected. Between two and four people per thousand are heterozygous for the PLS gene and therefore they become carriers of the disorders. This results in a population prevalence of one to four per million people. Greater frequency of occurance in consanguineous off spring has been noted in approximately one third of cases.[5]

Approximately 20% to 25% of PLS cases suffer from increased susceptibility to infections other than periodontitis, most of them show predisposition to mild skin infections as furunculosis or pyodermas. Occasionally severe infections such as liver abscess or pneumonia occurs.[6]

PLS is caused by genetic defect located on chromosome 11q, which involves mutations of the CTSC gene. Various studies in PLS patients have shown more than 90% reduction in CTSC activity with resultant reduced host response against bacteria. 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 several studies.[7]

Subgingival plaque samples from periodontal pockets of PLS cases contain primarily Actinobacillus actinomycetemcomitans. Other suspected periodontal pathogens including Porphyromonas gingivalis, Fusobacterium nucleatum, Bacteroides forsythus, Treponema denticola and Prevotella inermedia have also been implicated to play a role in PLS periodontal pathogenesis.[8]

The etiology of PLS is not completely understood. Both sexes are equally affected with no racial predominance. Anatomic, microbial and viral agents as well as host response are suspected causative factors. In our case, genetic testing could not be performed to identify the gene mutation because of the low economic status of the parents consanguineous descent has been described for this syndrome and the dermatological, periodontal and radiographical features strongly suggested the diagnosis of PLS.[9]

The differential diagnosis include Hiam-Munk syndrome and Hypophosphatsia. Hiam-Munk syndrome exhibits arachnodactly acroosteolysis, atrophy of nails and deformity of phalanges in the hands. None of these features were found in the present case. In hypophosphatasia, deficiency of alkaline phosphatase activity is seen but in our case the values were within normal limits and therefore this differential diagnosis could be excluded.[10]

A definitive treatment regime is not yet reported. However to control periodontal destruction, treatment modalities like conventional periodontal therapy, oral hygiene instruction and systemic antibiotics have been suggested.[11]

Identification of specific periodontal pathogens and antibiotic therapy appropriate to these microorganisms along with extraction of severely periodontally compromised teeth can prolong viability of non-affected teeth. Newer therapeutic modalities involve the use of oral retinoids, such as acitretin and isotretinoin have proven to be beneficial in treating both the dental and cutaneous lesions. Retinoid treatment is usually started during the eruption of permanent dentition and is followed till the normal dental process is complete.[12]

CONCLUSION

Papillon-Lefevre Syndrome is a devastating disease process, which due to the associated cutaneous involvement and partial or complete edentulism, can severely affect the psychological, social and esthetic wellbeing of the patient at a very young age. Early diagnosis of the disease and proper intervention is very much essential to prevent edentulism of the clinical patient.

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