Papillon-Lefevre syndrome: A case report

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Abstract
Papillon–Lefèvre syndrome (PLS) is a rare autosomal recessive disorder which is characterized by diffuse palmoplantar keratoderma and precocious aggressive periodontitis, resulting in premature loss of deciduous and permanent dentition at a very early age. Several etiopathogenic factors are responsible for the syndrome, like immunologic alterations, genetic mutations, and also the role of microorganisms. Dentists play a major 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. Here, we are presenting an elaborate review of PLS, its etiopathogenesis, clinical presentation, and management options.

Keywords: Periodontitis, Modified Complete Dentures, Primary and Permanent dentition, Palmoplantar Keratoderma.

Introduction
Papillon-Lefevere syndrome (PLS) or palmoplantar keratoderma with aggressive periodontitis is an autosomal recessive genetic disorder casued by deficiency in cathepsin C.(1) Two French physician Papillon & lefevre were the first one to characterize this disease in a siblings suffering from palmoplantar hyperkeratosis associated with early onset periodontitis & premature loss of decideous & permanent dentition. (P-L 1924).(1) The third component that is dural calcification in this disease was added by Gorlin et al 1964 (Gorlin 1964).(2) Other symptoms include hyperhidrosis, arachnodactyly, increased susceptibility infection & mental retardation. (Hattab FN 1995).(3)

It has a prevalence rate of 1-4 cases per million. PLS is inherited as an autosomal recessive disorder. There is a 25% chances of risk for children to be affected if both parents are carrier of the defective gene. (Hart TC 1994).(4) The rate of prevalence is more in consanguineous offsprings. (Khan FY 2012).(5)

Alteration in ectodermal & mesodermal components leads to the skin lesion in this syndrome, however there's no such explaination concerning rapid loss of primary as well as permanent teeth. (Kothiwale SV 2008).(6)

PLS might cause cellular immune defect with decrease chemotaxis & phagocytic function of neutrophils & granulocytes.(7) Causation of periodontal problems in PLS may be related with the presence of pathogens such as Actinobacillus actinomycetemcomitans, Capnocytophaga gingivalis, Porphyromonas gingivalis, Peptostreptococcus micros, Fusobacterium nucleatum & Spirochetes.(8)

Palmoplantar keratosis appears as well defined demarcated keratotic plaques over the palmar & plantar surfaces (Fig. 1). Hyperhidrosis present may be seen with the keratosis giving a foul odor.(9)

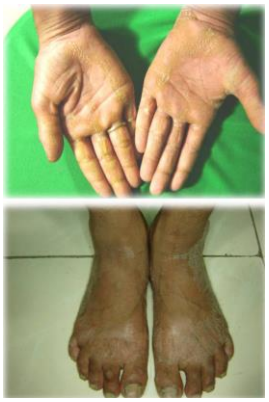

Fig. 1: Photograph showing hyperkeratosis of the palms and soles

At the age of 3-4 years early onset of periodontitis starts. Gingiva then appears inflammed & bleeds on provocation. The periodontitis has rapid progression & doesn't respond to the conventional periodontal treatment.(10)

There is premature shedding of deciduous teeth mostly by 3-4 years of age. Even though the development & eruption of it occurs at a normal chronological age. Gingiva reverts back to normal after deciduous teeth gets shed but the gingiva again gets inflammed as soon as permanent teeth erupts.(11) At the age of 13-16 years there is complete shedding of permanent teeth except third molar which gets shed later.(12)

Case Report
A 13 year old male child reported to the Department of Pedodontics & Preventive Dentistry at Dental College, with the chief complaint of loss of multiple teeth. While taking the past dental history it was revealed that his deciduous teeth had erupted

normally but were eventually lost at the age of 5 years approximately.

After eruption of the permanent teeth there was bleeding from the gingival while brushing & eating. As per patient he began loosing all his upper & lower incisors by the age of 10 years, & all the remaining tooth were Grade-III mobile when checked clinically (Fig. 2).

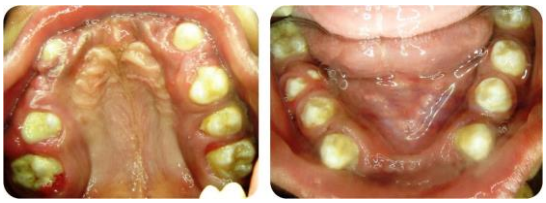

Fig. 2: Intraoral photograph

There was no relevant medical history and parents were not of consanguineous marriage, there was no significant prenatal history given by the mother.

The patient was mesomorphic. His physical and mental development was normal. The patient showed hyperkeratosis papules on his hands, knees and soles.

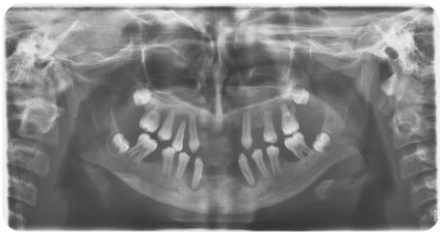

Fig. 3: Orthopantomograph showing generalised severe alveolar bone loss. Maxillary and mandibular incisors are completely lost.

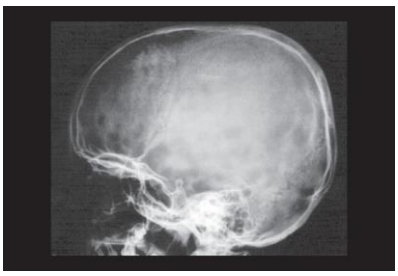

Fig. 4: Lateral ceph showing no evidence of intracranial calcification.

Radiographic Features- Orthopantamogram shows overall bone loss in respect to both maxilla and mandible & presence of teeth namely 13, 14, 15, 16, 23, 25, 26, 33, 34, 35, 36, 43, 44, 45, 46, in addition to its unerupted 17, 27, 37, 47 can be seen (Fig. 3). Loss of bone support can be seen around all the erupted teeth. Lateral cephalogram shows dural calcification (Fig. 4).

Discussion

PLS is an autosomal recessive inherited disorder. It is thought to be because of mutation in cathepsin- C. The Cathepsin-C gene is located on chromosome 11q14.1- 11q14.3 (wani a 2006) Cathepsin-C is a lysosomal exo-cysteine protease encoded by CTSC gene, central co-ordinator for activation of many serine protease in immune cells. Activation of serine protease causes degradation of extracellular matrix component leading to tissue damage & chronic inflammation. Mutation in the Cathepsin- C gene leads to prepubertal periodontitis in PLS patients.[13,14]

Differentiation should be made with certain diseases like Haim Munk syndrome, Olmsted syndrome, Huriez syndrome, Epidermolysis bullosa herpetiformis, Cole disease which shows palmar-plantar keratosis like PLS but doesn't show juvenile periodontitis. On the other hand certain diseases like hypophosphatasia, acrodynia, histiocytosis X, leukemia, cyclic neutropenia associated with periodontitis & premature loss of teeth doesn't show palmar-plantar keratosis.[13-15]

Management

Oral Management:

Periodontal therapy: Scaling & Root planning was done in the patient, followed by counseling for maintenance of his oral hygiene. Chlorhexidine mouthwash was prescribed.

Prosthetic rehabilitation: After completion of periodontal therapy patients was given removable partial dentures to replace his upper & lower anteriors (Fig. 5). Fixed partial denture was not given as the abutment teeth were mobile. Dental implants were not given because the growth of maxilla & mandible has not yet been completed & because of the socioeconomic status of the patient.

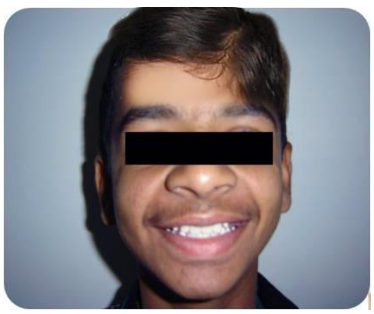

Fig. 5: Child after removable partial denture

Maintenance phase: patient was asked to report to the Department of Pedodontics & preventive dentistry every 3 months for evaluating oral hygiene status & the removable partial denture.

Dermatological Management: Patient was referred to a dermatologist who prescribed retinoids & antibiotics for his palmar-plantar keratosis.[16-18]

Conclusion

Patients with PLS are most likely to consult a dentist for their periodontal problems. As such a dentist should have sufficient knowledge about the signs & symptoms of PLS. Thus dentist should also learn to differentiate between diseases having similar signs & symptoms. An early diagnosis by the dentist can prevent the loss of permanent teeth by instituting oral retinoids during the eruption of permanent teeth. The dentist can also help those patients who comes at a later stage by administering prompt periodontal therapy to preserve those tooth which are still present.

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