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Case Report

Papillion lefèvre Syndrome: Rarest of the Rare; But it is Still There

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Abstract

A rare autosomal recessives disorder, Papillon–Lefèvre syndrome (PLS) also known as palmoplantar keratoderma, anhidrosis and periodontitis was first described in 1924 by French Physician Papillon and Lefèver. The disorder is characterized by diffuse palmoplantar keratoderma and aggressive periodontitis, leading to premature loss of deciduous and permanent dentition at a very young age. Mutations in the cathepsin C gene (CTSC), located at human chromosome 11q14.1-q14.3, are the cause of PLS. A 22-year-old male boy was presented with all the characteristic feature of papillon-lefèvre syndrome. He had complaint of swollen and friable gums and discomfort in chewing the food along with persistent hyperkeratotic, flaking and scaling of the skin of palms and soles. He also had early shedding of his milk teeth with family history of consanguineous marriage of parents.

Keywords: Autosomal recessives disorder, Papillon–Lefèvre syndrome

1. Introduction

Papillon–Lefèvre syndrome (PLS) also known as palmoplantar keratoderma with periodontitis [1,2] is an autosomal recessive disorder [3]. PLS is a rare autosomal recessive trait which is characterizedby thickening of hand and feet and a periodontitis in both the milk and permanentdentition. It has been found that the prevalence of this syndrome is one to four cases per million people. In around 20-40% cases parental consanguinity has been reported. (1). Common associated features with this syndrome are calcification of and falx cerebri along with retardation of somatic development. (2-4).

About 20-25% of such patients are suggested to be more prone for infection (1,3,5,6). PLS is commonly associated with defect in chromosome 11q14–q21 with mutation in cathepsin C (7,8,9).

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Cathepsin C gene is expressed commonly on the epithelial regions such as palms, soles, knees, and keratinized oral gingiva. Cathepsin c gene encodes *dipeptidyl-peptidase* I which is a cysteine lysosomal protease which act by removing dipeptides from amino terminus of the protein substrate it also has endopeptidase activity and is also expressed in various immune cells such as leukocytes, , and their precursors.. It has been found in many studies that periodontal involvement in PLS is due to impaired chemotactic and phagocytic function of polymorphonuclear leukocytes (PMNs) and reduced activity to T- and B-cell mitogens but still the exact pathogenic mechanism leading to periodontal involvement is elusive. Periodontal effects are seen immediately after dentition when gingiva becomes erythematous and swollen. Halitosis occurs due to accumulation of plaques in the deep crevices. In this disease primary incisors are affected first and will have marked mobility by the age of 3 years and by the age of 4-5 years all the primary teeth may have exfoliated. Large number of teeth loss by the age of 13-15 years, there will be drastic alveolar bone destruction along with atrophy of the gums. We hereby report a case of papillon- lefevre syndrome having all the characteristic features of this syndrome.

2. Case report Case history

A 22-year-old malepatient reported to department of dermatology in Uttar Pradesh university of medical science, Saifai, Etawah with complaints of persistent thickening, flaking, scaling and anhidrosis of the skin of palms and soles along with lose of teeth, difficulty in chewing andrecurrently edematous and friable gums. He also had early shedding of milk teeth. The past medical history of the patient was insignificant. Family history was associated with consanguineous marriage of parents. No other family member was affected with similar kind of illness.

Fig.1 Symmetric, well demarcated, keratotic confluent plaques affecting the skin of palms and soles and extending onto dorsal surfaces

3. General and extraoral examination

Patient had overall normal physical and mental development. Extraoral examination revealed symmetric, well demarcated, keratotic confluent plaques affecting the skin of palms and soles and extending onto dorsal surfaces (Fig- 1). His nails were discolored (Fig- 2) but hairs were normal. Skin of the patient was anhidrotic, dry scaly lesions were present, eye creases were prominent. keratotic plaque affecting the dorsal surface of the feet were present.

4. Intraoral examination

On intraoral examination upper permanent central incisors (Fig-4) and lower permanent central and lateral incisors were missing (Fig-3). Severe mobility affecting all the teeth with heavy deposits of food particles and halitosis were also present.

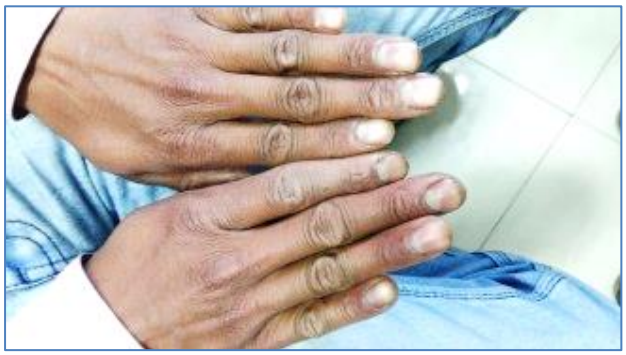

Fig.2 Discolored and dystrophic nails

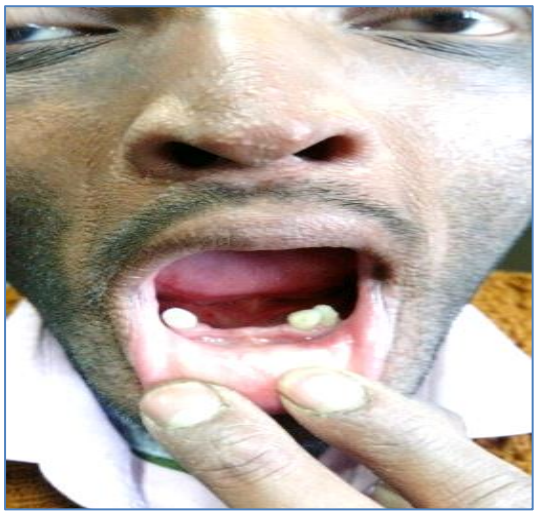

Fig.3 lower permanent central and lateral incisors are missing.

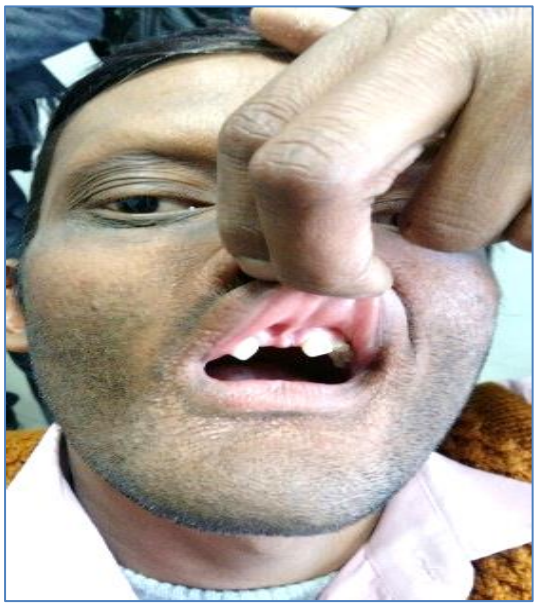

Fig.4. Upper central incisors are missing

5. Laboratory investigation

Laboratory investigation was carried out, which included hematological and biochemical assessment.

The results of these investigations were within normal limits.

6. Diagnosis

In view of above findings, the case was diagnosed as papillon lefevre syndrome.

7. Discussion

PLS is a disease which can affect the children as well as their parents psychologically, socially and esthetically. A multidisciplinary approach is required for improving the quality of life of affected children along with an early teeth examination and parental counselling for providing complete psychosocial rehabilitation of affected children. Though the exact pathogenic mechanism of this syndrome is still elusivebut many immunologic, microbiological and geneticstudies have been proposed. Microbiological studieshave suggested the involvement of many pathogens in the disease process out of which microbes like *Actinobacillus actinomycetemcomitans,* are commonly involved. Some case studies have reported that *A actinomycetemcomitans* has asignificant role in pathogenesis of PLS. According to recent researches, abnormality in cutaneous development and dental disease progression is suggestive to be associated with inactivation of cathepsin C gene (10). Due to involvement of various etio-pathogenic factor in this disease, successful treatment of periodontal destruction is still a challenging problem. In our case, due to poor financial condition of the patient, genetic testing could not be carried out but dermatological, periodontal and radiological features are strongly suggestive of PLS. The differential diagnosis of this syndrome includes Haim-Munk syndrome and hypophosphatasia.

Haim-munk syndrome is an autosomal recessive genodermatosis characterized by palmoplantar keratoderma and e periodontitis. The patient of Haim-Munk syndrome also present with thinning of nails, arachnodactyly, a rooster lysis and deformity of phalanges of hands. None of these features were found in this case hence Haim-Munk syndrome could be ruled out in this case. Another differential diagnosis is hypophosphatasia, in which deficiency of alkaline phosphatase is seen, but in this case the values are within normal range hence hypophosphatasia is also ruled out. Although the definitive treatment strategy has not yet been established, however by using conventional periodontal therapy, systemic antibiotics and oral hygiene instructions, periodontal destruction could be prevented. (11). It is important to identify the specific periodontal pathogen and start the antibiotic therapy appropriately along with removal of periodontally diseased teeth to prolong the longevity of normal teeth.Various new treatment options have been proposed which include use of oral retinoids like oral acitretin, and isotretinoin. These oral retinoids change the course of inflammation of teeth and preserve it. It is advised to start the appropriate antibiotic therapy for the active periodontitis to prevent the further complications like bacterial infection and pyogenic liver abscess. In the future innovative stem cell therapy could be proved beneficial which can open the ways to save smiles of these children.

8. Conclusion

Early diagnosis and treatment of PLS is essential as it has a great psychological and social impact on the developing children. Oral prosthesis is required for edentulous patients which includes partial or complete denture replacement. Osseo-integrated implants could be used in the near future which can be proved beneficial for restoring the esthetics as well as function. In the identification of rare syndromes like PLS, periodontist is the first person to diagnosis and intervene, therefore it is important to have greater awareness for such syndrome which would be helpful in knowing more cases for further studies.

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