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Case Report

Papillion Lefevre Syndrome-A Case Series of Siblings

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Abstract: Papillon Lefèvre syndrome is a rare disease characterized by skin lesions caused by palmar-plantar hyperkeratosis, and severe periodontal destruction involving both the primary and permanent dentitions. It is transmitted as an autosomal recessive condition and consanguinity of parents is evident in about one third of cases. The palmar plantar keratoderma typically has its onset between the ages of 1 and 4 years and severe periodontitis starts at the age of 3 or 4 years. Two cases of Papillon-Lefevre syndrome in the same family (siblings) are presented. A 15 year old girl and 13 year old boy having all the characteristic features of syndrome.
Keywords: Papillion lefevre syndrome, Palmar Plantar keratoderma, severe periodontitis

INTRODUCTION

Papillon–Lefèvre syndrome (PLS) is characterized by hyperkeratosis of hands and feet and by a generalized aggressive periodontitis in both the primary and the permanent dentition[1]. 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[1]. Calcification of the falx cerebri and the choroid plexus, and retardation of somatic development is often an associated feature[2].The recently identified genetic defect in PLS has been mapped to chromosome 11q14–q21, which involves mutations of cathepsin C[3,]. Studies in PLS patients have shown more than 90% reduction in cathepsin C activity. An impaired chemotatic and phagocytic function of polymorphonuclear leukocytes (PMNs) has been described in many reports. The palmoplantar keratoderma typically has its onset between the ages one and four years.[4]The sharply demarcated erythematous keratotic plaques may occur focally, but usually involve the entire surface of palms and soles resulting in foul-smelling odor.

The second major feature of PLS is severe periodontitis, which starts at the age of three or four years. [5] The development and eruption of deciduous teeth proceeds normally, but their eruption is associated with gingival inflammation and subsequent rapid destruction of the periodontium. The resulting periodontitis characteristically is unresponsive to traditional periodontal treatment modalities and the primary dentition is usually exfoliated prematurely by the age four years. After exfoliation, the inflammation subsides and gingiva appears healthy. However, with eruption of the permanent dentition the process of gingivitis and periodontitis is usually repeated and there is subsequent premature exfoliation of the permanent teeth, although the third molars are sometimes spared. [6]

Radiographic features are characterized by generalized loss of alveolar bone. Gorlin *et al.* have added the third feature of dural calcification. Reyes also observed radiographic evidence of intracranial calcification. Histopathologic findings of affected skin consist of hyperkeratosis, occasional patches of parakeratosis, acanthosis, and slight perivascular inflammatory infiltrate. [7]

CASE NO 1

A 15-year-old female patient presented to the department of Periodontics and oral Implantology, Government dental college, Srinagar (India) with the chief complaint of multiple loss of teeth. Clinical history revealed that she had normal emergence of deciduous teeth at 8-9 months of age, which started loosening at three years and were all eventually lost by four years of age. Patient was not sure about the time of eruption of permanent teeth, but described gingival bleeding during brushing and eating, after the eruption of permanent teeth. There was loosening of permanent teeth from l0 years of age and eventually all the permanent teeth were lost by 12 years of age, except for one mobile lower third molar tooth (fig 1). Bleeding was also associated at the time of tooth loss.

Dermatological examination revealed dry skin with normal development of hair and nails. There were symmetric, well-demarcated, yellowish, keratotic, and confluent plaques affecting the skin of her palms and soles, also extending onto the dorsal surfaces of hands and feet (fig 1). In the light of these findings Papillon-Lefevre Syndrome was diagnosed in the patient. This diagnosis was confirmed by the dermatology clinic as palmar plantar hyperkeratosis.

Intraoral examination showed premature loss of all teeth, except for the left mandibular third molar

which was covered with deposits and it showed severe mobility. No other abnormality was detected in relation to soft tissues. Patient had a reduced facial height due to resorption of the alveolar ridge (fig 1)

Routine blood investigations and liver function tests were carried out and were found to be within the normative range of values

Management

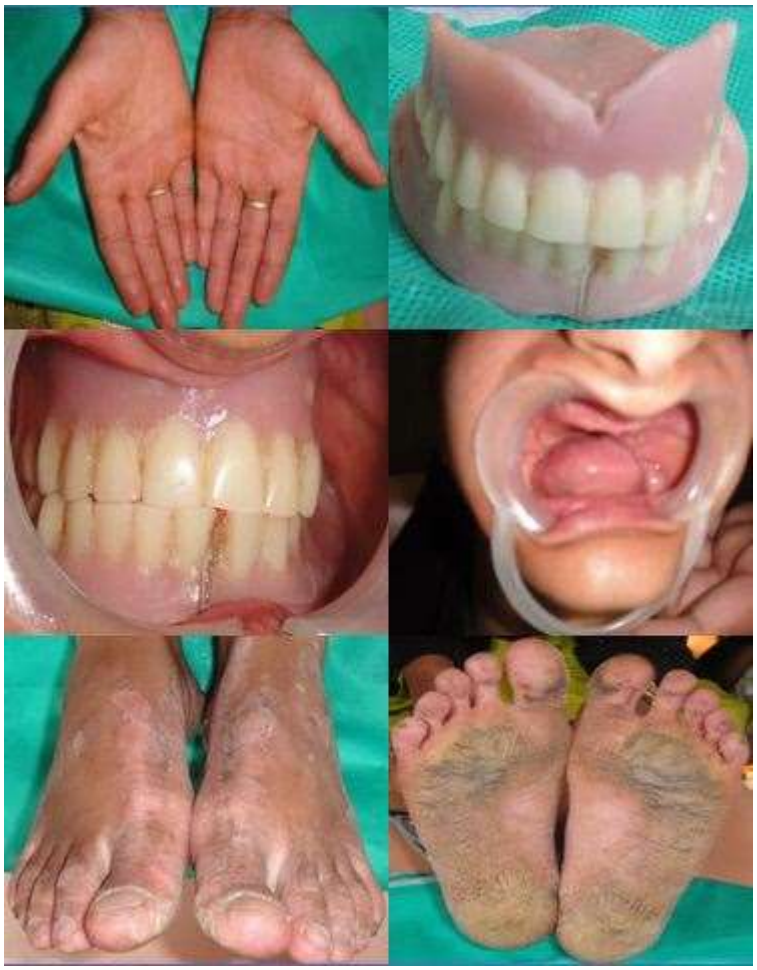
Since the patient was a young teenage girl, it was necessary to manage the esthetic needs of the patient as a priority. It was appropriate to give her complete dentures, considering her functional requirements, such as mastication and speech. Prior to the routine clinical procedure for complete dentures, the mandibular third molar was extracted. The dentures were fabricated in the laboratory and inserted into the patient's mouth (fig 1). Routine postdenture instructions were given to the patient

CASE NO 2

A 13-year-old boy patient (above patient's brother) reported to the department of periodontics and oral Implantology, Government dental college Srinagar (India) with a chief complaint of loosening of multiple teeth (fig 2)

On examination, there were symmetrical, well demarcated, keratotic and confluent plaques affecting the skin of the palms and soles which extended to the dorsal surface of the feet and hands. The skin was dry and rough on palpation.

On intraoral examination, mobility was present in all the teeth (fig 2). The gingiva was red, soft and edematous with deep periodontal pockets and bleeding on probing. The mucosa in other areas of the oral cavity appears to be normal. OPG of the patient showed severe alveolar bone loss in relation to all the teeth.(Fig 2).



Case 1 (fig 1)

Case 2 (fig 2)

Laboratory investigation was carried out, which included haematological and biochemical assessment. The results were within normal limits.

Management

First of all, the patient was prescribed an antibiotic of doxycycline 100mg OD x 3 weeks and a mouth rinse of 0.2% chlorhexidine gluconate and educated for oral hygiene to prolong the life of his dentition.

DISCUSSION

Papillon Lefèvre syndrome was first described by Papillon and Lefèvre in 1924. The disease is characterized by diffuse palmoplantar hyperkeratosis and juvenile periodontitis. Haneke used the following three criteria to classify a case as PLS: (a) palmoplantar hyperkeratosis; (b) loss of primary and permanent teeth; and (c) autosomal recessive inheritance.

Although the exact pathogenesis of this syndrome remains relatively obscure, immunologic, microbiologic, and genetic bases have been proposed. Microbiological studies have demonstrated *Actinobacillus actinomycetemcomitans* , *Porphyromonas gingivalis* , *Fusobacterium nucleatum* , and *Treponema denticola* organisms, suggesting that many pathogens may be involved in the disease process. Previous case reports and studies have reported that *A. actinomycetemcomitans* plays a significant role in the pathogenesis and progression of the rapid periodontal breakdown seen in PLS. [8]

It has been suggested that the presence of periodontal pathogens alone is not sufficient for the expression of PLS, and other factors, such as host response, play an important role in the pathogenesis of the disease process. Several authors have suggested an abnormal neutrophil dysfunction with PLS [9] to explain the pathogenesis

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

A multidisciplinary approach is important. PPK is usually treated with topical emollients. Salicylic acid and urea can be added to enhance their effect. Systemic retinoids have proven to be effective in PPK of PLS as well as in other PPKs.A definite treatment regime is not yet reported; however, to control periodontal destruction, several treatment modalities

have been suggested, e.g., conventional periodontal therapy, oral hygiene instructions, and systemic antibiotics.

Identification of specific periodontal pathogens and antibiotic therapy appropriate to these microorganisms, along with extraction of severely periodontally compromised teeth, can prolong the viability of nonaffected teeth. Newer therapeutic modalities involve the use of oral retinoids, such as acitretin and isotretinoin. In the future, stem cell therapy can be expected to open up new vistas in the dental treatment of such children.

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