Case Report

Papillon-Lefèvre Syndrome- A Case Report and Review of Literature

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Abstract: Papillon-Lefèvre syndrome is a very rare syndrome of autosomal recessive inheritance characterized by palmar-plantar hyperkeratosis and early onset of a severe destructive periodontitis leading to premature loss of both primary and permanent dentitions. Various etiopathogenic factors are associated with the syndrome; but a recent report has suggested that the condition is linked to mutations of the cathepsin C gene. A case of Papillon-Lefèvre syndrome of an 18 year old girl is presented, having all of the characteristic features. Her father also had premature shedding of his deciduous teeth. Severe generalized periodontal destruction with mobility of teeth was evident on intraoral examination; orthopantomograph examination showed severe generalized loss of alveolar bone in the patient. The classical presentation of this syndrome is discussed.

Key words: Hyperkeratosis, Papillon-Lefèvre syndrome, Periodontitis.

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Introduction:

Papillon-Lefèvre syndrome (PLS), first described by two French physicians Papillon and Lefèvere in 1924.[1] It is a rare autosomal recessive disorder of keratinization. Its reported incidence is 1-4 per million and both the sexes are equally affected.[2] The disorder is characterized by diffuse palmoplantar keratoderma and premature loss of both deciduous and permanent teeth. The palmoplantar keratoderma typically has its onset between the ages 1 and 4 years.[3] Another component of PLS is asymptomatic ectopic calcification in choroid plexus and tentorium. Although this has been taken as a cardinal feature, but being inconsistent it is not considered important for the diagnosis. About 20% of these patients also show an increased susceptibility to infections due to some dysfunction of lymphocytes and leukocytes.[4]

The recently identified genetic defect in PLS has been mapped to chromosome 11q14–q21, which involves mutations of cathepsin C. The cathepsin-C gene is expressed mainly in the epithelial regions such as palms, soles, knees, and keratinized oral gingiva. These are generally the areas that are most commonly affected by PLS.[5] Periodontal effects appear almost immediately after tooth eruption when gingiva becomes

erythematous and oedematous. The primary incisors are usually affected first and can display marked mobility by the age of three years. By the age of four or five years, all the primary teeth may have exfoliated.[6] We hereby present a case report of 18 year old girl Papillon-Lefèvre syndrome having all of the characteristic features of the syndrome.

Case Report:

An 18-year-old girl reported with a chief complaint of loose teeth and discomfort in chewing along with recurrently swollen and friable gums. She also complained of persistent thickening, flaking and scaling of the skin of palms and soles. There was excessive sweating from her hands and feet since 15 years along with excessive brittleness of nails and pigmentation of skin. She also had premature shedding of deciduous teeth. History revealed that her deciduous teeth had erupted normally but gradually exfoliated by the age of 4-5 years. Similarly most of her permanent teeth too were lost prematurely after erupting normally. There was history of recurrent swelling of gums and foul breath followed by loosening and exfoliation of teeth. At the age of four years, her parents also noticed a progressive thickening of palmoplantar skin.

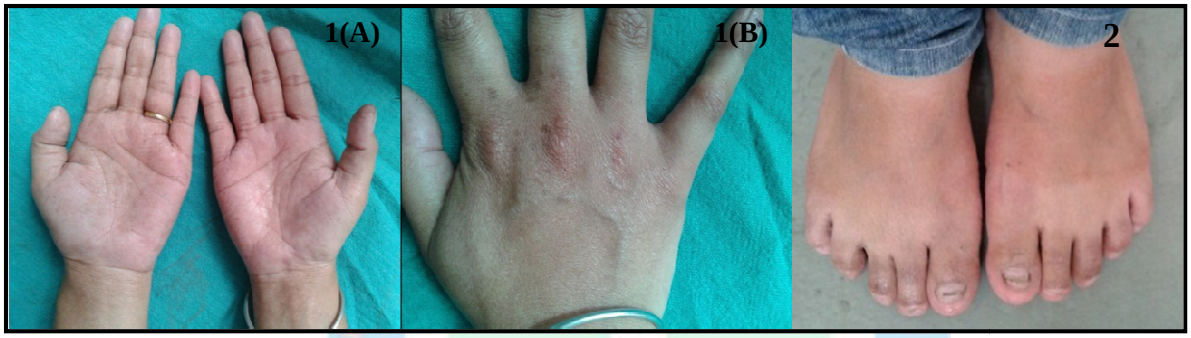

Figure 1: (A) Keratotic plaques affecting the palmar surfaces of hands; (B) Keratotic plaques affecting the dorsal surfaces of hand and

Figure 2: Keratotic plaques affecting the dorsal surface of feet

Her family history was contributory as her father also suffered from premature shedding of both deciduous and permanent dentition.

General physical and systemic examination was within normal limits. Cutaneous examination revealed symmetrical diffuse hyperkeratosis and mild erythema affecting the skin of palms and soles extending to the dorsal and palmar surface of hands (Figure 1a and1b) and feet (Figure 2). Intraoral examination revealed missing 17,16,15,11,21,25,26,27, 34,36,45,46,47 (FDI Notation) along with grossly carious 48. The gingiva was edematous and friable along with presence of fetid odour. (Figure 3) There was mobility i.r.t 35 and attrition of lower anterior teeth.

The orthopantomograph showed impacted 45. There was generalised horizontal bone loss and the affected teeth lacked osseous support (Figure 4). Laboratory investigations were carried out, which included hematological and biochemical assessment. The results were within normal limits. In view of the above findings, the case was diagnosed as PLS. For treatment, a multidisciplinary approach was followed. For the skin manifestations the patient was treated with emollients. The patient was advised to start acitretin so as to modulate the course of periodontitis and preserve the teeth. Complete oral prophylaxis was advised followed by rehabilitation of the missing teeth.

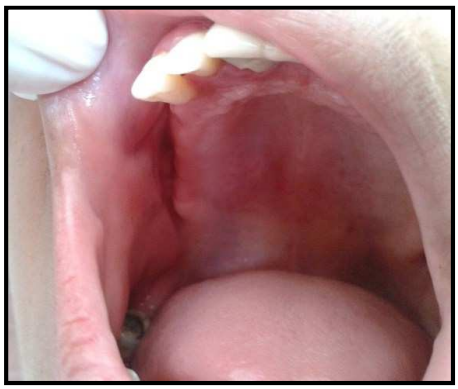

Figure 3: Intra-oral photograph revealing multiple missing teeth

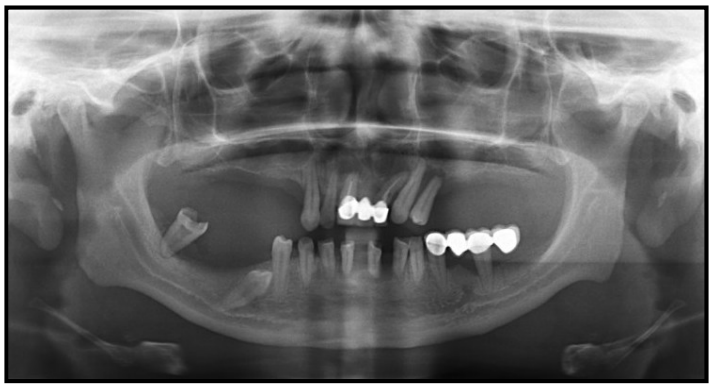

Figure 4: OPG revealing generalized horizontal bone loss.

Discussion:

PLS can psychologically, socially and aesthetically affect the growing children. Hence, an early dental evaluation and parental counselling as a part of preventive dental treatment is essential for providing complete psychosocial rehabilitation for PLS children; a multidisciplinary approach may improve the prognosis and quality of life of these children.

In the case presented, the dermatological and periodontal features strongly suggested the diagnosis of PLS. Other sites that may be affected include the eyelids, cheeks, labial commissures, legs, thighs, and axillae. The hair is usually normal, but in advanced cases the nails may show transverse grooving and fissuring. Histopathologic examination reveals nonspecific hyperkeratosis, acanthosis, focal parakeratosis, psoriasiform hyperplasia, tortuous capillaries in dermal papillae, and superficial lymphocytic infiltration.[7,8]

Papillon-Lefèvre syndrome is an extremely rare genodermatosis inherited as an autosomal recessive trait. The disease is caused most commonly by Cathepsin C gene mutations leading to the deficiency of cathepsin C enzymatic activity.[5] Characteristic clinical features are: diffuse palmoplantar keratoderma, premature loss of deciduous as well as permanent teeth and a tendency to recurrent pyogenic infections of the skin. Palmoplantar hyperkeratosis typically starts between 1-4 years of age.[9] The erythematous keratotic plaques may be focal or diffuse and are characterized by transgradient extension of keratoderma to the dorsal surface of palms and soles. Well demarcated psoriasiform plaques usually occur on the knees and elbows.[9] Repeated episodes of periodontitis and gingivitis lead to destruction of periodontium and subsequent premature loss of deciduous and permanent teeth.[9] In addition to palmoplantar hyperkeratosis and oral findings, patient may have impaired function of the immunological system, associated most probably with insufficiency of cathepsin C, which is essential for granzyme B activation and NK cell cytolytic activity.[10,11] These abnormalities are associated with increased susceptibility to recurrent pyogenic infection of skin.[9]

The other features of Papillon-Lefèvre syndrome are the intracranial calcification of choroid plexus and tentorium on radiographic examination and palmoplanter hyperhydrosis.[12] The findings of hyperhydrosis were present in case of our patient. Two close differential diagnoses of Papillon-Lefèvre syndrome are Haim Munk Syndrome[13] and prepubertal periodontitis.[14] Haims-Munk

syndrome is an allelic variant of Papillon-Lefèvre syndrome and the clinical features, in addition to palmoplanter keratoderma and loss of dentition, include arachnodactyly (claw like phalanges with convex nails), and acroosteolysis.[13] In prepubertal periodontitis, palmoplanter hyperkeratosis is absent.[14]

Oral retinoids including acitretin, etretinate and isotretinion combined with intensive antimicrobial treatment are the mainstay of treatment in Papillon-Lefèvre syndrome.[15] It should be considered to start treatment at eruption of the first teeth and maintain therapy during the development of permanent teeth. The periodontitis in PLS is usually difficult to control. Effective treatment for the periodontitis includes extraction of the primary teeth combined with oral antibiotics and professional teeth cleaning. It is reported that etretinate and acitretin modulate the course of periodontitis and preserve the teeth.[16] Early diagnosis of Papillon-Lefèvre syndrome and appropriate treatment may help to prevent of loss of dentition in these patients.

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