

PAPILLON – LEFEVRE SYNDROME – EVIDENCE BASED REPORT

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ABSTRACT

An interesting case of Papillon-Lefevre Syndrome with persistent thickening, flaking and scaling of the skin of palms and soles associated with recurrently swollen and friable gums since the age of 4 has been presented. Papillon-Lefevre Syndrome is an extremely rare genodermatosis inherited as an autosomal recessive trait that is mainly ascertained by dentists because of the severe periodontitis that afflicts patients. This is an interesting case report, which presents the typical features of PLS.

Key words: Papillon-Lefevre Syndrome, Anodontia, Keratoderma, premature exfoliation of teeth, neutrophil defect.

INTRODUCTION

Papillon – Lefevre Syndrome [PLS], first described by 2 French physicians, Papillon and Lefevre in 1924, is an extremely rare genodermatosis inherited as an autosomal recessive trait that is mainly ascertained by dentists because of the severe periodontitis that afflicts patients¹.

PLS varies from mild psoriasiform scaly skin to overt hyperkeratosis, typically develops within the first 3 years of life. Most patients display both periodontitis and hyperkeratosis².

The PLS locus has been mapped to chromosome 11q14-q21. Its prevalence is estimated to be 1-4 per million persons in the general population with a carrier rate of 2 – 4 per 1000. The possible mechanisms relating genetics and periodontal disease include virulent infection, immune response and underlying tissue pathology³.

CASE REPORT

A 12-year old boy presented with persistent thickening, flaking and scaling of the skin of his palms and soles associated with recurrently swollen and friable gums since the age of 4. He had premature shedding of his deciduous teeth and had lost most of his permanent teeth.

His physical examination revealed symmetric, well demarcated keratotic confluent plaques affecting the skin of his palms and soles and extending onto the dorsal surfaces. His nails show dystrophy and transverse grooving.

Intraoral examination revealed swollen and bleeding gums. The involved gingiva was bright red and there was loss of gingival stippling. There was loss of most of his teeth except for upper central incisors, upper & lower canines. There was grade I mobility in relation to upper central incisors.

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Roentgenographic examination of teeth revealed unerupted lower molars. The alveolar bone around the mobile teeth was devoid of definable lamina dura. An extensive alveolar bone loss was noted giving the teeth a “floating in air” appearance.

His laboratory examination included complete blood count, liver function transaminase levels, total bilirubin, alkaline phosphatase, c- reactive protein – all of which were within normal limits. Oral prophylaxis was done and artificial dentures were advised.

DISCUSSION

PLS is a manifestation of homozygosity of autosomal recessive genes with consanguinity as a contributing factor⁴. Males and females are equally affected and there is no racial predominance⁵. 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 1-4 years of age⁶. The sharply demarcated erythematous keratotic plaques may occur focally, but usually involve the entire surface of the palms and soles, sometimes extending onto the dorsal surfaces of the hands and feet^{1&7}. Often there is associated hyperhydrosis of the palms and soles resulting in a foul smelling odor¹. These findings have a tendency for worsening in winter.

Periodontal disease is a major feature of PLS, which starts at the age of 3 -4 years^{1&7}. The development and eruption of the deciduous teeth proceed normally, but their eruption is associated with gingival inflammation and subsequent rapid destruction of the periodontium¹. The primary dentition is usually exfoliated prematurely by the age of 4 years. After exfoliation, the inflammation subsides and the gingiva appears healthy. However, with the eruption of the permanent dentition, there is recurrence of gingivitis and periodontitis and there is premature exfoliation of the permanent teeth^{1,8 &9}.

Microbiologic studies on the oral microflora of patients with PLS revealed predominantly gram-negative anaerobic bacilli on periodontal regions. In most of the PLS cases, *actinobacillus actinomycetemcomitans* was reported. Severe periodontal disease differentiates PLS from other palmoplantar dermatosis. The degree of dermatologic involvement may not be related to the level of periodontal infection^{3,10 & 11}.

The patient in this report showed classic events of gingivitis, periodontitis, normal development and eruption of deciduous teeth but early loss of both deciduous teeth [3-4years] and most of the permanent teeth [12 years]. Extra-oral examination revealed sharply demarcated keratotic plaques in his palms and soles, which are characteristic of PLS.

History revealed that the keratotic lesions were noted by the age of 2 years in his palms and soles.

Hair are usually normal but nails may show onchodystrophy and transverse grooving, claw-like phalanges with convex nails [arachnodactyly] and osteolysis⁴.

The patient in this case-report showed onchodystrophy, transverse grooving with arachnodactyly in the nails.

The cause of PLS is not well understood, but recently, 2 research groups have reported that loss of function is due to mutations affecting both the alleles of the cathepsin C gene, located on chromosome 11q14.1 – q14.3, were associated with PLS^{1&8}. The cathepsin gene encodes a cysteine lysosomal protease also known as dipeptidyl peptidase I, which functions to remove dipeptides from the aminoterminal of the protein substrate. The cathepsin C gene is expressed in epithelial regions commonly affected by PLS such as palms, soles, knees and keratinized oral gingiva. It is also expressed at high levels in various immune cells including PMN, macrophage and their precursors^{1 & 13}.

The exact cause of periodontal disease in PLS has not been found, but it has been attributed to decreased neutrophil phagocytosis, bacterial infection and impaired reactivity to T and B cell mitogens^{14,15&16}.

Gingival infection, abscess formation, loss of alveolar bone and destruction of periodontal ligament are probably the causative factors in shedding of teeth. Page and Baas^{4&17} were of the opinion that the defect seems to be with the periodontal component, especially the cementum and as the teeth lose their close relation to bone, the intensity of inflammation subsides. According to Preus^{4&18}, the hereditary defect perhaps lies at the epithelial surface barrier leading to reduced defense against virulent periodontopathogens.



Fig 1: Dorsal aspect of hands



Fig 3 : Keratotic plaques in soles

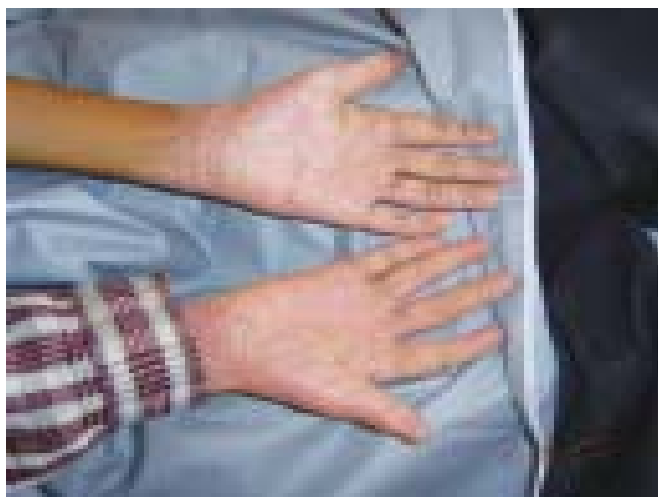


Fig 2: Keratotic plaques in palms

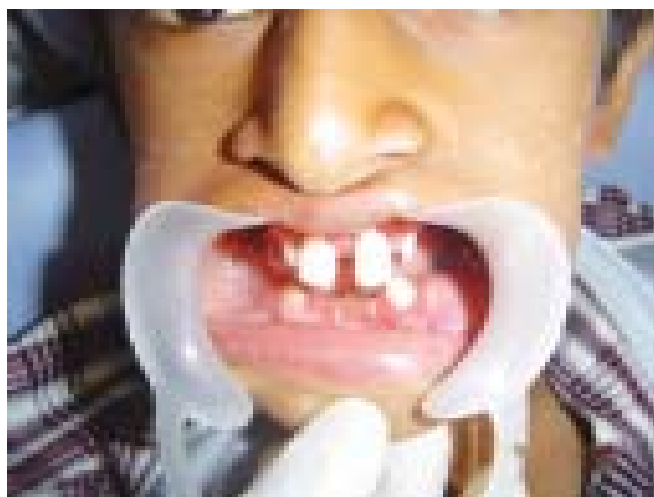


Fig 4 : Intraoral view

It is usually not necessary to treat cutaneous lesions unless they interfere with patient's activities. Frequent periodontal check ups, oral hygiene instructions and antibiotic therapy only delay the shedding of teeth. Early extraction of teeth too has been advocated to prevent bone loss. Etretinate, isotretinoin & acetretin have been successful in improving the cutaneous as well as gingival lesions.

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