

Case Report

Papillon-Lefèvre syndrome: A case report of two siblings and review of the literature

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Abstract Papillon-Lefèvre syndrome is a rare autosomal recessive genodermatosis which is characterised by periodontitis, palmoplantar keratoderma and predisposition to pyogenic infections and occurs due to cathepsin C gene mutation (located on chromosome 11). The loss of primary teeth usually occurs by the age of 4 years and secondary teeth by second decade. The disorder is associated with significant cosmetic and functional disability.

Key words

Papillon-Lefèvre syndrome, periodontitis, palmoplantar keratoderma.

Introduction

Papillon-Lefèvre syndrome is an extremely rare genodermatosis inherited as an autosomal recessive trait, affecting children between the ages 1-4 years. It has a prevalence of 1-4 cases per one million. Males and females are equally affected and there is no racial predominance. The disorder is characterized by periodontitis resulting in severe gingivitis in children which leads to the loss of teeth by the age of 4-5 years unless treated. Permanent teeth may be lost in the same fashion. Transient palmoplantar keratoderma is an associated feature. The treatment is supportive and consists of retinoids in combination with antibiotics and dental care lessening the gingival inflammation and saving the teeth. Here we report the case of Papillon-Lefèvre syndrome in two siblings and briefly review the literature.

Case report

A 21-year-old male, student by occupation,

referred by a dental practitioner presented with chief complaints of thickening of skin of palms and soles since birth and loosening of teeth since the age of 5 years. The skin lesions started as small, localised areas of rough, thick skin over the palms and gradually progressed to involve almost whole of the palms and similar lesions then appeared over the soles after an approximate period of 6-8 months. There was history of aggravation of these lesions in winter months with increased roughening of the skin and painful fissuring. The patient gave history of pain during walking and difficulty in performing daily routine activities with hands. The patient also gave history of loosening of teeth since the age of 5 years. However, there was no history of loss of teeth. The patient also gave history of similar skin and oral lesions in her elder sister. There was history of loss of both deciduous and permanent teeth in her, which were replaced with artificial dentures. There was history of consanguineous marriage. There was no other significant medical or surgical history.

On examination there were multiple, bilaterally symmetrical, yellowish, hyperkeratotic plaques covered with adherent scales over the palms and soles (**Figures 1 and 2**). There were few hyperkeratotic plaques over the dorsa of both

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Figure 1 Plantar keratoderma.



Figure 2 Hyperkeratotic lesions over hands and feet.



Figure 3 Loose carious teeth.

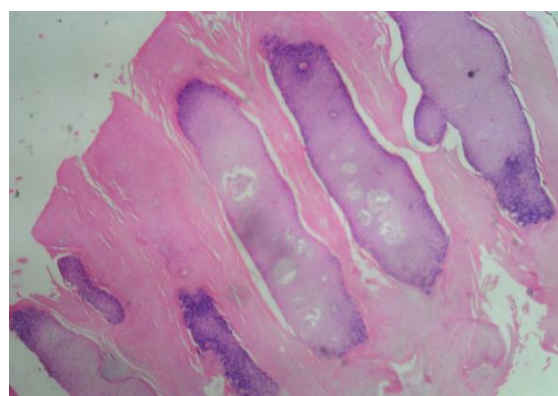


Figure 4 Gross hyperkeratosis on histopathology.

hands. Oral examination revealed multiple, loose carious teeth especially the central incisors (**Figure 3**), canines and premolars. The teeth were abnormally mobile on manipulation. The gingivae in relation to teeth were soft and erythematous. The hair and nails were normal.

All the routine investigations were within normal limits. Biopsy of the skin lesions was performed which was consistent with keratoderma (**Figure 4**).

Discussion

Papillon-Lefèvre syndrome, first described by two French physicians Papillon and Lefèvre in 1924, is an extremely rare genodermatosis inherited as an autosomal recessive trait, affecting children between the ages 1-4 years.

It has a prevalence of 1-4 cases per one million. Males and females are equally affected and there is no racial predominance.^{1,2} The disorder is characterized by periodontitis resulting in severe gingivitis in children which leads to the loss of teeth by the age of 4-5 years unless treated. Permanent teeth may be lost in the same fashion. Transient palmoplantar keratoderma preceded by erythema appears in the first year of life and spreads to the dorsal aspects and up the Achilles tendon. Psoriasiform plaques on the elbows and knees may be present, but in a survey of 47 patients cutaneous involvement was not related to the severity of periodontal disease and did not correlate with the age.^{3,4} Virulent Gram negative organisms invade the alveolar socket, usually including *Actinobacillus actinomycetemcomitans*. Frequent pyogenic infections of the skin and internal organs occur. Associated hyperhidrosis causes an unpleasant odour. The

hair is usually normal but may be sparse. Dural calcification, especially in the attachment of tentorium and choroid has been noted in some cases. Malignant melanoma has been reported as a complication.^{5,6}

Neutrophil phagocytosis and reactivity to T and B cell mitogens are impaired. Inheritance is autosomal recessive and the condition is characterised by homozygous mutation in the gene encoding the lysosomal protein cathepsin C. This finding explains the predisposition to pyogenic infections, but the mechanism of keratoderma is not established.⁷

Histopathological changes are non-specific, but shows hyperkeratosis with irregular parakeratosis and a moderate perivascular infiltrate. Electron microscopic findings include lipid-like vacuoles in the corneocytes and granulocytes, reduction in tonofilaments and irregular keratohyalin granules. The treatment consists of retinoids in combination with antibiotics and dental care lessening the gingival inflammation and saving the teeth.⁸

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