Papillon-Lefèvre Syndrome: Report of Two Cases in a Family

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Abstract

This report presents two cases of Papillon-Lefèvre syndrome (PLS) affecting two girls among five siblings belonging to a south Indian Muslim family. The patients were 12 and 14 years old. The patients presented with palmar-plantar hyperkeratosis which started around the age of two years. The young patient had severe destructive periodontitis with multiple periodontal abscesses and loose teeth.

Keywords: Papillon-Lefèvre syndrome, palmar-plantar hyperkeratosis, early onset periodontitis.

INTRODUCTION

Papillon-Lefèvre syndrome (PLS) is a rare disease associated with the early onset of periodontitis in primary and permanent teeth.1-5 PLS is a syndrome with a prevalence of 1 to 4 per million in the general population with no racial or sexual predominance.2 This syndrome usually affects children and is rarely seen in adults.1-6 The typical clinical presentation of PLS is characterized by diffuse or localized palmar-plantar hyperkeratosis and severe destructive periodontitis leading to premature loss of primary and permanent teeth. The etiology is unknown, but the disease is considered to be transmitted as an autosomal recessive trait with a frequency of 0.001.4 The factor suggested as the cause of PLS is an immunologic disorder in neutrophil chemotactic and phagocytic function, as a result of mutation in the cathepsin C gene located on chromosome 11q14. Consanguinity between parents was apparent in one third of cases studied.5 PLS is the only syndrome associated with early onset periodontitis among at least 19 syndromes with palmoplantar hyperkeratosis.

Palmar-plantar hyperkeratosis may appear most commonly between 6 months to 4 years of age. Hyperkeratosis of the dorsal surface of the fingers and toes, elbows, legs, thighs and trunk are observed in few PLS patients.7,8 The skin lesions appear as white, light yellow, brown, or red plaques and patches, which undergo crustation, cracking, and deep fissuring. Infection may superimpose the defective skin resulting in the formation of abscesses.9,10 The skin and oral manifestations of PLS occur simultaneously. The primary teeth erupt at normal chronological age and sequence without any structural deformities, although microdontia, root resorption, and incomplete root formation were reported in a few cases.11,12 Oral manifestation of PLS is characterized by generalized severe early onset periodontitis affecting primary and permanent teeth. All the primary teeth are usually exfoliated by 4 to 5 years of age, which leads to early eruption of permanent teeth. All the permanent teeth except the unerupted third molars are lost due to severe destructive periodontitis by the age of 13 to 15 years. Presently periodontitis observed in PLS has been classified under the category of periodontitis as a manifestation of systemic disease along with other genetic and hematologic disorders associated with early onset of periodontitis such as cyclic neutropenia, congenital neutropenia (Kostmann syndrome), Chediak-Higashi syndrome, leukocyte adhesion deficiency syndrome (type I and II), lazy leukocyte syndrome, Down’s syndrome, hereditary hypophosphatasia.13 Other features reported in many PLS patient include calcification of falx cerebri and choroid plexus and increased susceptibility to other infections.4

This report describes two cases of PLS occurring in a south Indian Muslim family with total of 5 children. This is one of the few reports which describe more than one case in the same family. The patients were presented to a private dental clinic, in Hyderabad.

CASE 1

A 12-year-old girl presented with a chief complaint of swollen gums, loose teeth and difficulty in eating. Family history revealed that the patient was the third of five siblings belonging to a south Indian Muslim family. She has one sister (age 14 years), a brother (age 16 years), and two younger brothers (age 10 years and 7 years). The elder sister was affected by similar condition. The parents, other members of the family and their close relatives were free from similar manifestations. Pregnancy, labor, and delivery were normal. The patient’s height and weight were in normal range for her age. The skin lesions started to appear after two and half years of age. There was no history of other serious illness or susceptibility to infection in areas other than the oral cavity. Dental history revealed eruption of primary
teeth in normal chronological order. At around 3 years of age, the gums were red, swollen and painful with pus discharge. Soon the teeth became mobile and were exfoliated. The child was edentulous by the age of four and half years.

Intraoral examination showed generalized severe periodontitis manifested by redness of gingiva, generalized inflammatory gingival enlargement, and multiple periodontal abscesses (Fig. 1). Deep periodontal pockets (> 6 mm) with purulent exudate were present around all permanent teeth. The teeth were mobile, drifted, and extruded. The patient had difficulty in chewing. Severe halitosis was present. The teeth were normal in size and shape.

Dermatological examination showed hyperkeratosis of palms and soles in the form of well-demarcated plaques (Figs 2 and 3). The hyperkeratosis was exacerbated during the winter months. Radiographic examination revealed a severe alveolar bone loss of all erupted teeth with less than one third of alveolar bone remaining giving the teeth a ‘floating in air’ (Fig. 4). The unerupted third molars were in different stages of crown development and were not associated with any bone changes.

CASE 2
A 14-year-old patient, elder sister of the first patient, reported to our hospital complaining of difficulty in eating due to loss of teeth. The patient's height and weight were in normal range for her age. Pregnancy, labor, and delivery were normal. Dental
history revealed eruption of all the primary teeth by the age of three years. At around 3 years of age the gums were red, swollen and painful with pus discharge. Soon the teeth became mobile and were exfoliated. The child was edentulous by the age of four years. The permanent teeth erupted at normal chronological age and affected by severe periodontitis. Consequently the patient lost all her teeth by the age of 12 years.

Dermatological examination revealed a dry and scaly skin, and a bilateral and symmetrical hyperkeratosis of the palms and soles and dystrophy of the nails of the feet (Fig. 5). The skin lesions were hyperkeratotic, yellowish, scaly and fissured, and prone to periods of remission and exacerbation. The soles were more seriously affected than the palms. The hyperkeratosis was exacerbated during the winter months. Intraoral examination revealed completely edentulous maxillary and mandibular arches (Fig. 6). No abnormality was detected in the oral mucosa including that covering the edentulous ridges. Radiographic examination revealed generalized atrophy of maxillary and mandibular alveolar ridges. The unerupted third molars except the maxillary right third molar were present in different stages of crown development and were not associated with any bone changes.

DISCUSSION

The present cases exhibited the typical clinical features of PLS: hyperkeratosis of palms and soles, and generalized early onset periodontitis accompanied by early loss of both primary and permanent teeth. The two reported cases had essentially similar cutaneous lesions in the form of hyperkeratotic plaques. The cutaneous lesions were aggravated during cold weather. The skin of the soles were more severely affected than that of the palms. Elder sister had dystrophy of nails of the feet. The two reported cases had essentially similar periodontitis accompanied by early loss of both primary and permanent teeth. The child was edentulous by the age of four years. The permanent teeth erupted at normal chronological age and affected by severe periodontitis. Consequently the patient lost all her teeth by the age of 12 years.

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PLS was differentiated from other genetic diseases associated with early onset periodontitis by the presence of the palmar-plantar hyperkeratosis. PLS was also distinguished from keratoderma palmo-plantaris of Unna-Thost and mal de Meleda as these diseases are not associated with dental problems. PLS also differs from Haim-Munk syndrome as the later presents with digital abnormalities along with palmar-plantar hyperkeratosis and early onset periodontitis involving primary and permanent dentition.

REFERENCES