ISSN 2347-5579

Unique Journal of Medical and Dental Sciences

Available online: www.ujconline.net

Case Report

PAPILLON-LEFÈVRE SYNDROME IN TWO SIBLINGS: A CASE REPORT

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Received: 25-02-2014; Revised: 23-03-2014; Accepted: 22-04-2014

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ABSTRACT

Papillon-Lefèvre syndrome (PLS) is a very rare autosomal recessive disorder characterized by palmoplantar hyperkeratosis and severe early onset of destructive periodontitis leading to premature loss of both primary and permanent dentitions. Here we are presenting case report of siblings who presented with palmoplantar hyperkeratosis and aggressive periodontitis.

Keywords: Papillon-Lefèvre syndrome, Periodontitis, Palmoplantar, Hyperkeratosis, Cathepsin C.

INTRODUCTION

It was first described by two French physicians Papillon and Lefèvre in the year 1924, in a brother & sister showing severe dental alterations with palmar and plantar hyperkeratosis[1]. Papillon-Lefèvre syndrome (PLS) is an uncommon autosomal recessive type-IV palmoplantar ectodermal dysplasia. Of the palmoplantar ectodermal dysplasias, only PLS and Haim-Munk syndrome (HMS) are associated with premature periodontal destruction[2].

Since 1924, only few cases of this syndrome have been reported, its prevalence is estimated to be 1 - 4 per million in the general population with carrier rate of 2 to 4 per 1000. Consanguineous marriage was determined in 20 to 40% of patients with PLS[3].

CASE REPORT

Case 1: A 13 year old boy presented with the chief complaint of multiple missing teeth (Fig. 1a). He gave history of premature shedding of his deciduous teeth and loss of multiple permanent teeth after a minor episode of trauma.

On extra-oral examination mild thickening& scaling of the skin of his palms & soles were noted. (Fig. 1b & 1c)On intraoral examination all incisor & first permanent molars were missing and the remaining teeth were grade II or III mobile. Gingiva appeared red, swollen & edematous. OPG showed generalized severe horizontal bone loss with multiple floating teeth (Fig. 1d). Lateral skull radiograph ruled out any intra-cranial calcification (Fig. 1e).

Figure 1a: Showing missing anterior teeth with red inflamed and swollen gingivae

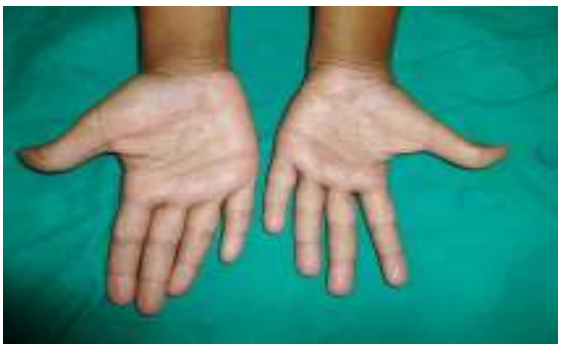

Figure 1b: Mild thickening & scaling of the skin of palms

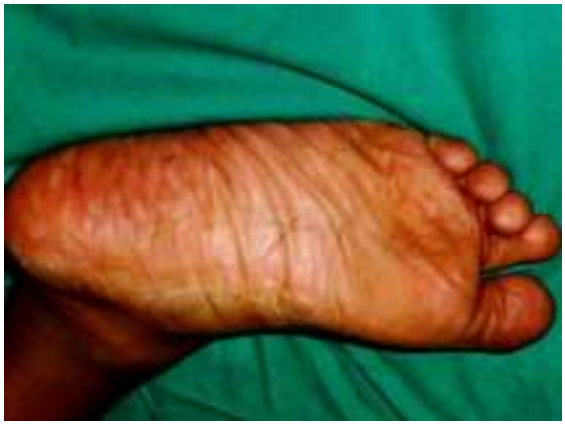

Figure 1c: Plantar keratosis

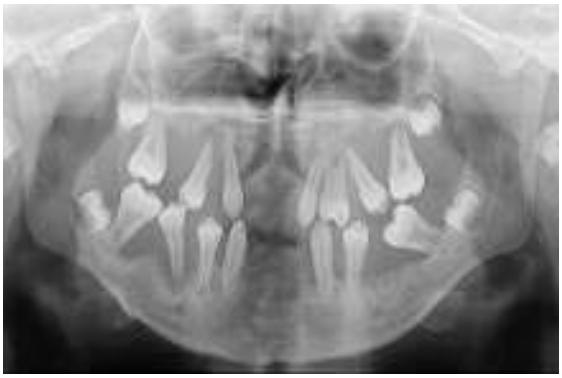

Figure 1d: OPG showing severe loss of alveolar bone and teeth appear to be "floating in air."

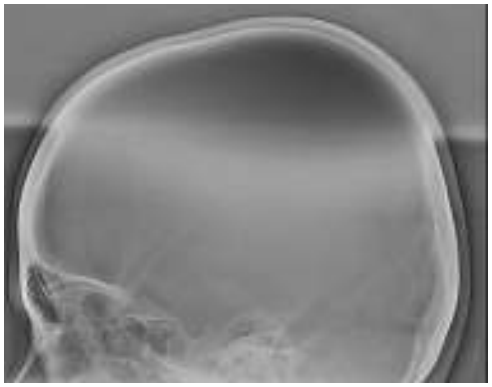

Figure 1e: Lateral skull radiograph showing no evidence of any intra-cranial calcification

Case 2:14 year old sister of theabove mentioned patient reported with the chief complaint of recurrent episodes of bleeding gums & denudation of the skin over the finger with painful fissures (Fig.2a).

On extra-oral examination, skin of the soles showed well demarcated thickened hyperkeratotic plaques (Fig. 2b &2c). Intraoral examination showed mal-aligned teeth with chronic periodontitis. OPG showed generalized moderate bone loss which was more pronounced around first molars & incisors (Fig 2 d).

In both cases based on the history, clinical examination, and radiographic examination, a provisional diagnosis of Papillon Lefèvre syndrome was made.

Figure 2a: Photograph showing periodontitis with mal-alignment of teeth

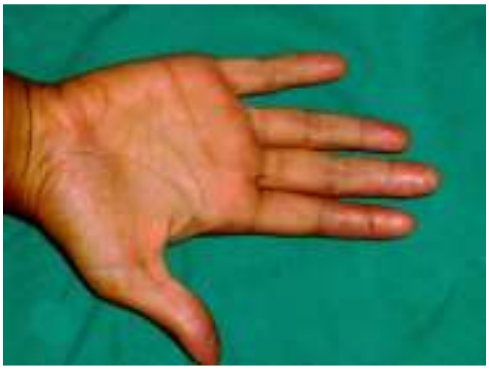

Figure 2b: Denudation of the skin of the fingers with fissuring

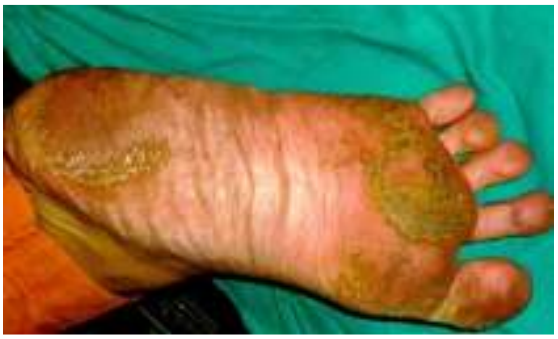

Figure 2c: Well demarcated thick keratotic plaques on the sole

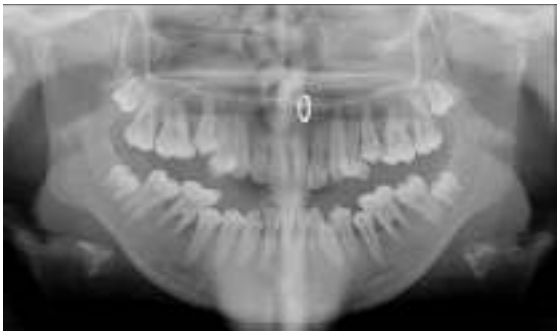

Figure 2d: OPG showing generalized moderate horizontal bone loss

DISCUSSION

Clinical presentation: The disorder is characterized by diffuse palmoplantar keratoderma and premature loss of both deciduous and permanent teeth.

1. Palmoplantar keratoderma: 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 [4]. Often, there is associated hyperhidrosis of the palms and soles resulting in a foul-smelling odor[5]. Well-demarcated psoriasiform plaques occur on the elbows and knees [4]. The findings may worsen in winter and be associated with painful fissures, as was seen in our second case.

2. Severe periodontitis: The development and eruption of the deciduous teeth proceed 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 age 4 years. After exfoliation, the inflammation subsides and the gingiva appears healthy. However, with the 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 [6,7].

Age & gender: PLS usually affects children between the ages 1-4 years[8]. Males and females are equally affected and there is no racial predominance[9].

Etiology: Few research groups have reported that loss-of-function mutations of the lysosomal protease cathepsin C gene are associated with PLS as well as related conditions. The cathepsin C gene is expressed in the epithelial regions commonly affected by PLS, such as palms, soles, knees, and keratinized oral gingiva[10].

Radiological feature: Radiographic examination of advanced cases reveals severe loss of the alveolar bone, and teeth appear to be "floating in air"[10]. In case 1, OPG shows severe alveolar bone loss and the characteristic "floating in air" teeth appearance. (Fig. 1d) In case 2, OPG shows bone loss which is more pronounced around mandibular first molars. But bone destruction is not as severe as seen in male patient (Fig. 2d).

Asymptomatic calcification of the falxcerebri and choroid plexus have been noted in many PLS patients[10]. However lateral skull radiograph didn't show any evidence of intracranial calcification in our case (Fig. 1e).

Histopathological feature: PLS periodontitis shows a severe inflammatory infiltration of the subepithelial connective tissue, leaving only a few collagen fibres[11,12]. The dense infiltration mainly consists of plasma cells dominating the entire gingiva in certain area. Electron microscopy has also revealed degenerative alterations of plasma cells[13].Both of our patient declined to undergo a biopsy.

Differential diagnosis:Hart et al. identified a mutation of cathepsin C affecting a highly conserved amino-acid residue in Haim-Munk syndrome, demonstrating that PLS and Haim-Munk syndromes are allelic disorders[14].As PLS, Haim-Munk syndrome (HMS) and prepubertal periodontitis have been described as allelic variants; hence it is appropriate to consider only HMS and premature periodontitis in the differential diagnosis of PLS.

MANAGEMENT

Because of the involvement of several structures a multidisciplinary approach is required in the management of patients with LPS. Patients were referred to the dermatologist for cutaneous lesion. For periodontitis, both the patients were prescribed appropriate antibiotics with 0.2% chlorhexidine gluconate mouth rinse and the patients were educated about importance of oral hygiene. Extraction of the mobile teeth with severe periodontitis was advised and the patients were referred to the prosthetic department for prosthesis fabrication. Presently patients are undergoing planned dental extractions with periodontal therapy & are kept on regular follow-ups.

CONCLUSION

Combined cooperation from dermatologists, pediatrician, periodontists, and prosthodontists is critical for the overall care of patients suffering from PLS, which although a very rare condition, can lead to long-lasting psychological and social trauma to the growing patient.

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Source of support: Nil, Conflict of interest: None Declared