CASE REPORT

Papillon–Lefevre syndrome: A case report

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Abstract Papillon–Lefevre syndrome (PLS) is a rare autosomal recessive disorder of keratinization, characterized by palmoplantar hyperkeratosis, periodontal involvement and precocious loss of dentition. The purpose of this report was to describe the case of an 18-year-old girl who presented to the outpatient department of Navodaya Dental College and Hospital, Raichur, Karnataka, India, with the chief complaint of multiple loss of teeth. Her gingiva appeared erythematous, edematous and bled readily on probing, and the teeth were mobile. Hyperkeratosis of palms and soles were found. These findings are consistent with Papillon–Lefevre syndrome. The clinical presentation, differential diagnosis, complications and management of this syndrome are discussed.

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1. Introduction

Papillon–Lefevre syndrome (PLS) is a rare autosomal recessive disorder of keratinization, characterized by palmoplantar hyperkeratosis, periodontal involvement and precocious loss of dentition (Jain et al., 2005). It was first described in 1924 by two French physicians, Papillon and Lefevre. It has a prevalence of 1–4 cases per million persons and both males and females are equally affected with no racial predominance (Gorlin et al., 1964; Cury et al., 2002).

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 one and four years (Bach and Levan, 1968). The sharply demarcated erythematous keratotic plaques may occur focally, but usually involve the entire surface of palms and soles resulting in foul-smelling odor (Gorlin et al., 1964). Well-demarcated psoriasiform plaques occur on elbows and knees (Siragusa et al., 2000). This may worsen in winter and be associated with painful fissures.

The keratosis of the plantar surface extends to the edges of the soles and occasionally onto the skin overlying the Achilles tendon and the external malleoli. Other sites that may be affected include the eyelids, cheeks, labial commissures, legs, thighs and axillae. The hair is usually normal but the nails, in advanced cases, may show transverse grooving and fissuring.

The second major feature of PLS is severe periodontitis, which starts at the age of three or four years (Yagmur et al., 2004). The development and eruption of the deciduous teeth...
proceeds normally, but their eruption is associated with severe gingival inflammation in the absence of any local etiologic factor. The gingiva is bright red, edematous and bleeds easily. The periodontal pockets rapidly deepen, with severe loss of alveolar bone and marked fetor exorius. Although gingival inflammation and alveolar resorption is usually so severe that the alveolar process is completely destroyed, even during the most active phase of periodontal destruction, the rest of the oral mucous membrane is reported to be completely normal. Primary dentition is usually exfoliated prematurely by the age of 4 years. After exfoliation, the inflammation subsides and gingiva appears healthy. With the eruption of permanent dentition, the whole process of gingivitis and periodontitis is repeated and there is subsequent premature exfoliation of the permanent teeth by the age of 13–16 years. Later, the third molars also undergo the same fate. Severe resorption of alveolar bone gives the teeth a ‘floating-in-air’ appearance on dental radiographs (Janjua and Khachemoune, 2004; Galanter and Bradford, 1969; Mahajan et al., 2003).

The degree of dermatologic involvement may not be related to the level of periodontal infection (Ullbro et al., 2003). Nail changes such as transverse grooving and fissuring are apparent in advanced cases. In addition to the dermatologic and oral findings, patients may have decreased neutrophil, lymphocyte or monocyte functions and an increased susceptibility to bacterial infection, leading to recurrent pyogenic infections of the skin. Pyogenic liver abscess is a complication of PLS and is associated with impairment of the immune system (Janjua and Khachemoune, 2004).

Radiographic features are characterized by generalized loss of alveolar bone. Gorlin et al. have added the third feature of dural calcification (Newman, 2003). Reyes also observed radiographic evidence of intracranial calcification (Reys et al., 1998). Histopathologic findings of affected skin consist of hyperkeratosis, occasional patches of parakeratosis, acanthosis, and slight perivascular inflammatory infiltrate (Angel et al., 2002).

An increased prevalence of parental consanguinity has been reported in PLS patients (Gorlin et al., 1964). All PLS patients are homozygous for the same cathepsin-C gene mutation inherited from a common ancestor. It would be pertinent to mention that there are reports of at least six cases of late onset variation of PLS without underlying cathepsin-C gene mutation (Ittni, 1992). This case report describes a case of PLS with clinical features and brief review of literature.

2. Case report

An 18-year-old female patient presented to the out patient department of Navodaya Dental College and Hospital, Raichur, Karnataka, India, with the chief complaint of multiple loss of teeth. Her history revealed that this condition, together with thickening of the skin, palms and soles was noticed by her family at the age of five.

Intraoral examination showed premature loss of upper and lower incisor teeth (except one central incisor), upper and lower first molars. Gingiva appeared erythematous, edematous and bled readily on probing (Fig. 1). Her deciduous teeth started exfoliating at age of three years and only her lower incisors erupted again that too became mobile soon. There was loosening of permanent teeth from 12 years of age and eventually many of the permanent teeth were lost by 15 years of age.

Bleeding was also associated at the time of tooth loss. Hence, the patient expressed a keen desire for replacement of all her lost teeth.

Panaromic view of the radiograph showed generalized loss of alveolar bone, complete loss of bone support around all premolars and second molars. Third molars were found to be erupting in three quadrants (Fig. 2).

Medical history was noncontributory. Parents were not of consanguineous marriage. Dermatological examination revealed symmetrical, well-demarcated, rough, erythematous, hyperkeratotic, scaly lesions on her knees, elbows, palms and soles (Figs. 3 and 4). Transverse grooves, which were present in most of the fingernails, were most prominent on the thumb (Fig. 5). Subsequently, there was reddening of the palms and soles at six months of age, which gradually thickened and became rough and scaly. Her general growth, hair growth, sweat glands and mental development were all normal.

Routine hematological examination revealed Hb% of 11.0 g/dl, bleeding time – 2 min, clotting time – 2 min 10 s, total leukocyte count of 9900, polymorph – 73%, lymphocyte – 23%, eosinophil – 4%, and ESR was 20 mm/h.

Summing up the clinical features and investigations diagnosis was confirmed as Papillon–Lefèvre syndrome and oral tetracycline was given for two weeks, after which clinical...
improvement was observed. For her dental condition, oral prophylaxis and a temporary removable prosthodontia appliance were suggested. The patient was referred to a dermatologist for further opinion and management.

3. Discussion

In our case, the skin and periodontal findings bore a striking resemblance to the classical descriptions of the syndrome. The dental history of the patients was also in accordance with the known symptoms of the syndrome, i.e., early exfoliation of all deciduous teeth commencing at 3 years of age, with complete exfoliation within 2–3 years.

Clinically, the patients had the characteristic skin lesions: sharply demarcated keratotic plaques involving the entire surface of the palms and soles. The lesions were diffuse, with a dry, scaly surface; they varied in thickness and were rough on palpation. The hair and nails appeared normal and transverse grooves on the fingernails were observed in our case. Intraoral findings were remarkable, with the characteristic features of periodontosis, as has been described for PLS. Patient had multiple permanent teeth missing. The permanent teeth that were present, exhibited marked mobility, with all the signs and symptoms of gingival inflammation (such as redness and edema and deepening of periodontal pockets that bled on probing) but without evidence of local factors. Radiograph (OPG) of the patients showed extensive alveolar bone loss up to the level of the apical third of the roots of almost all the permanent teeth that were present, giving the teeth a ‘floating-in-air’ appearance. All these dental findings and the skin lesions are consistent with the characteristic features of PLS.

A multitude of etiologic factors appear to be involved in the causation of PLS. It has been reported to be due to mutation of the cathepsin-C gene, which is expressed in the commonly affected epithelial regions such as the palms, soles and knees and the keratinized oral gingiva. It is also expressed at high levels in various immune cells, including polymorphonuclear leukocytes, macrophages and their precursors (Janjua and Khachemoune, 2004). The exact cause of periodontal disease in PLS has not been found, but it has been attributed to decrease neutrophil phagocytosis, bacterial infection and impaired reactivity to T- and B-cell mitogens (Rathi, 2002). Actinomyces actinomycetemcomitans, causing periodontal damage and alterations in the polymorphonuclear leukocyte functions, have been postulated as being a probable pathogenic mechanism. Gingival infection, abscess formation, loss

Figure 3 Symmetrical, well-demarcated, rough, erythematous, hyperkeratotic, scaly lesions.

Figure 4 Yellow colored hyperkeratotic areas on soles.

Figure 5 Transverse grooves, in most of the fingernails, which were prominent on the thumbnail.
of alveolar bone and destruction of periodontal ligament are probably the causative factors responsible for the shedding of teeth (Yagmur et al., 2004; Mahajan et al., 2003; Gonzalez et al., 1997; Ghaffer et al., 1999).

Other findings such as intracranial calcification, hyperhidrosis, susceptibility to infections, and mental retardation have been reported (Hacham-Zadeh and Goldberg, 1982). We could not observe any of these findings in our case.

Histological changes include hyperkeratosis, focal parakeratosis, marked acanthosis, focal parakeratosis, psoriasiform hyperplasia, tortuous capillaries in dermal papilla, and superficial perivascular lymphocytic infiltrate. Since dental changes are overlooked by dermatologists, this syndrome is often confused with other hereditary keratodermas, especially those of Unna-Thost and Mal De Meleda where no odontological finding are seen. Although the association of palmoplantar keratoderm with periodontal disease was first reported by dermatologists, most subsequent publications have appeared in the dental literature. The diagnosis of PLS may require collaboration between the dermatologist and dentist.

The skin manifestations of PLS are treated with emollients, with salicylic acid and urea added to enhance the effect. Oral retinoids including acitretin, etretinate and isotretinoin are the mainstay of treatment of both the keratoderma and the periodontitis associated with PLS. Normal dentition is observed with retinoids only when they are given before the onset of eruption of permanent teeth at 5 years of age. Treatment is more beneficial if it is started during the eruption and maintained during the development, of the 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. A course of antibiotics should be tried to control the active periodontitis in an effort to preserve the teeth and to prevent bacteremia and subsequently pyogenic liver abscess (Almuneef et al., 2003). Early extraction of teeth has also been advocated to prevent bone loss and allow preservation of a solid base for subsequent use in artificial dentures (Janjua and Khachemoune, 2004; Mahajan et al., 2003). Hence, we suggest that if parents notice keratotic plaques on the palms or plantar surfaces of the feet of their child, they should consult a dermatologist. Histological examination of the lesions will enable an early diagnosis of PLS so that early treatment can be instituted with retinoids, which can prevent the development of more skin lesions and modulate the course of periodontitis and, thus, preserve the teeth.

References


