CASE REPORT

Papillon-Lefèvre syndrome associated with Aggressive periodontitis: A rare case report

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ABSTRACT:

Papillon-Lefèvre Syndrome (PLS) is a rare syndrome of autosomal recessive inheritance characterized by palmoplantar hyperkeratosis and aggressive periodontitis, leading to premature loss of both primary and permanent dentitions; there are also reports of at least six cases of late-onset variation of PLS without underlying Cathepsin-C gene mutations have been documented. The etiology and pathogenesis of PLS is still unknown but recent reports suggest that the condition is linked to mutations of the cathepsin C gene. This case report describes about a case of Papillon-Lefèvre syndrome, along with a comprehensive review of etiology, pathology, clinical features, differential diagnosis and management of the condition.

Key words: Papillon-Lefèvre syndrome, palmoplantar hyperkeratosis, aggressive periodontitis.

INTRODUCTION:

Papillon-Lefèvre Syndrome (PLS) was first described by Papillon and Lefèvre in 1924 who had reported a case of siblings with palmoplantar hyperkeratosis associated with severe early-onset periodontitis and premature loss of primary and permanent teeth.[1] PLS is a rare syndrome and an autosomal recessive condition with palmoplantar hyperkeratosis and aggressive periodontitis as cardinal features, leading to premature loss of both primary and permanent dentitions. The etiology and pathogenesis of PLS is still unknown but recent reports suggest that the condition is linked to mutations of the cathepsin C gene. There are also reports of at least six cases of late-onset variation of PLS without underlying Cathepsin-C gene mutations which have been documented in literature.[2] Even though PLS is considered as a very rare condition with a prevalence of 1 per 4 million in the general population, more than 200 cases have been documented worldwide. Because of autosomal recessive inheritance pattern, the parents are not typically affected. Consanguinity between parents is noted in approximately one-third of the cases. There seems to be no predilection for either sex or racial predominance.[3]

Diagnosis of PLS is based on clinical findings of disease specific dermatologic manifestations and periodontal involvement. This case report describes the clinical diagnosis of a case of PLS as well as its management with antibiotics supplemented with meticulous surgical periodontal therapy.

CASE REPORT:

A 28-year-old male reported to the Dental out patients department of Armed Forces Medical College with a chief complaint of missing tooth in upper anterior region and mobility of multiple teeth (Fig:1). The patient was apparently normal before three years and had lost his upper tooth because of mobility two years back. On eliciting the medical history, he gave a history of skin lesions which were exacerbated in winter, though he had no history of any underlying systemic disorders. His family history revealed that he had no siblings and his parents didn't suffer from any such condition. On general examination, the dermal lesions were present as scaly plaques with fissures and hyper pigmented margins involving the instep of soles and medial aspect of feet (Fig: 2A). His lateral aspects of palms and fingers had ill defined scaly plaques (Fig: 2B). On gingival examination the color of gingiva was pink with melanin pigmentation in anterior region. There was presence of knife edge margins with exaggerated scalloping in relation to first and second quadrant (Fig: 1). Periodontal probing with University of North Calorina 15 probe showed 6-7mm of probing depth in relation to 16, 26, 36 and 46 and the upper anteriors had a generalized probing depth of 6mm. The attachment loss was inconsistent with the amount of plaque and calculus. The patients orthopantomograph showed 31 teeth with upper right canine missing. The radiographic features revealed vertical bone loss in respect to mesial and distal aspect of 16 and 26 along with horizontal bone loss in upper anterior teeth and vertical bone loss in the mesial and distal aspect of 36 and mesial aspect of 46 (Fig: 3).

The case was referred to Department of Dermatology, AFMC. The incisional biopsy of the skin lesion was taken from the plantar surface at the Department of Dermatology and the obtained specimen was submitted to the pathological laboratory of Department of Pathology, AFMC, Pune after fixing in 10% neutral buffer formalin. The histopathological features revealed hyperkeratosis with no evidence of dysplasia. He was prescribed oral Acitretin 25 mg daily for 3 months initially. Immaculate scaling, root planing was carried out at Dept of Periodontology, AFMC. During Phase I periodontal therapy the patient was prescribed 500 mg of Amoxycycline thrice daily and 400mg of Metronidazole twice daily for seven days followed by host modulation therapy with 20mg of subantimicrobial dose of Doxycycline (SDD) for six months. The case was taken up for periodontal surgery after four weeks of completion of Phase-I therapy. Periodontal flap surgeries were carried out in each quadrant with intermittent gap. Patient was advised for chlorhexidine mouth wash for 2 weeks post operatively and has been asked to maintain meticulous oral hygiene. The post-operative event has been uneventful as the patient has been on regular maintenance visits to the Department of Periodontology. The patient is still on observation and supportive periodontal therapy at regular intervals.

DISCUSSION:

The disorder is characterized by diffuse palmoplantar keratoderma and premature loss of both deciduous and permanent teeth; though, reports of late onset PLS have also been documented. The case reported here also gives features of late onset aggressive periodontitis similar to the report documented by Pilger et al.[2] It is a case of a variant of PLS without any history of premature loss of deciduous teeth but with sudden onset of aggressive periodontitis leading to mobility of multiple teeth and bone loss.

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 hands and feet as seen in our case and also been reported by Siragusa.[4] The PLS skin lesions vary greatly in color, texture, and manifestations and may appear as white, light yellow, brown or red plaques and patches which undergo ulceration, cracking, and deep fissuring like the reported case. Psoriasiform plaques may also be seen on the elbows and knees though in our case, only knees were involved. The typical dermatological symptoms tend to worsen in winter and are associated with painful fissures necessitating the need for systemic therapy like in our case and the same has been also documented by Motamedi et al.[1]

In addition to the oral and dermatologic findings, patients may have decreased function of neutrophils, lymphocytes, or monocytes and an increased susceptibility to bacterial infection, leading to recurrent pyogenic infections of the skin.[1] Pyogenic liver abscess is a complication of this syndrome and often associated with impairment of the immune system.[5] Gorlin et al. have suggested

that calcification of the duramater is a third component of the syndrome[6] though it was absent in our case. Recently, the gene for PLS was mapped to chromosome 11q14q21. This major gene locus comprises of six genes including cathepsin C, which is a lysosomal cysteine proteinase that plays an important role in the intracellular degradation of proteins and also processes and activates several leukocyte and mast cell granule serine proteinases critical to immune and inflammatory responses.[7] The presence of virulent gram-negative anaerobic periodonto pathogens, such as *P. gingivalis, Capnocytophaga, spirochaetes and Aggregatibacter actinomycetemcomitans* at the site have been seen with more than 50% of total colony forming units of *Aggregatibacter actinomycetemcomitans*.[8]

The skin manifestations are usually treated topically with emollients, keratolytics including salicylic acid and urea. There are reports that oral retinoids, such as etretinate, acitretin and isotretinoin, have been beneficial in treating keratoderma seen in PLS; although retinoids are not well tolerated in general. On part of the dentist, rigorous oral hygiene, chlorhexidine mouth-rinses, frequent professional prophylaxis, and periodic appropriate antibiotic therapy are to be implemented and are necessary for long term maintenance of periodontal problems.[9] Although, PLS is rare but other syndromes those exhibit hyperkeratosis without periodontal breakdown, include Melada's Disease, palmoplantar hyperkeratosis of Unna Thost, Greither's syndrome, Howel-Evans syndrome and with periodontal involvement such as Haim-Munk syndrome (HMS) are must be considered in the differential diagnosis of Papillon-Lefèvre syndrome.

CONCLUSION:

Since PLS involves extensive irrepairable destruction, interdisciplinary approach specially involving dental professionals and dermatologists is required for both initial and long term management of this disease. PLS threatens children if it affects at an early stage along with their parents with the prospect of edentulism. Hence, early diagnosis and intervention is essential. The skin manifestations of PLS are usually treated with oral retinoids like etretinate, acitretin and isotretinoin. A course of antibiotics should also be prescribed to control the active periodontitis in an effort to preserve the teeth and to prevent bacteremia and subsequently pyogenic liver abscess. Further studies in the field of microbiology and genetics with larger samples of larger duration are required to diagnose the exact cause of periodontal destruction in such patients, so that best possible treatment could be administered.

CONFLICTS OF INTEREST: NIL

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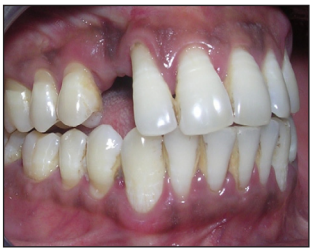

Fig 1: Intra-oral Picture of the patient

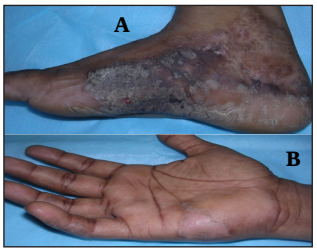

Fig 2 (A &B) : Palmo-Plantar hyperkeratosis

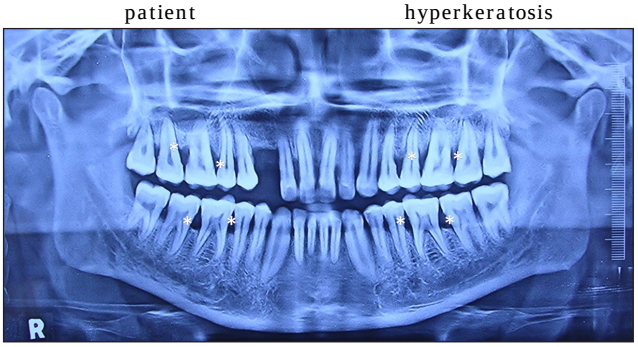

Fig 4: OPG of the patient