

Based on clinicopathological correlation, a diagnosis of dermatofibrosarcoma protuberans was made and she was referred to surgeon for deep excision with wide margins. The patient is under periodic follow-up to detect any recurrence at the earliest.

Dermatofibrosarcoma protuberans (DFSP) is an uncommon, slow growing, locally aggressive dermal tumor.¹Clinically, it is characterised by solitary or multiple violaceous to reddish blue papulonodules and plaques, usually over the trunk and proximal extremities.²Rarely, the lesions may be found on head and neck, pubic region and genitalia.^{3,4}Variants include confluent nodules forming a sclerotic plaque-like lesion, keloid-like sclerotic plaque, tumor-like and atrophic plaque. Histopathology is diagnostic. On immunohistochemistry, CD34 was positive. Histological variants include fibrosarcomatous type, pigmented type and myxoid type. There is a marked tendency for local recurrences, however, distant metastases is rare. Surgical excision with wide margins (2-3 cms) is the treatment of choice. For non-resectable cases, imatinib may be given.⁵

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Palmoplantar keratoderma progradiens et transgradiens associated with periodontitis – A case of Papillon-Lefevre syndrome

Sir, Papillon-Lefèvre syndrome (PLS) is characterized by palmoplantar keratoderma and a generalized aggressive periodontitis affecting both the primary and the permanent dentition.¹ It was first described by Papillon and Lefèvre in 1924.² It is considered to be a very rare syndrome having autosomal recessive inheritance and a prevalence of about 1-4 cases per million population. The history of parental consanguinity has been seen in 20% to 40% of the cases.³ Increased susceptibility to infections like recurrent otitis media occurs in 20-25% of the patients.¹ In this article we report an 18-year-old female with palmoplantar keratoderma associated with severe periodontitis.

An 18-year-old unmarried female visited our dermatology out patient department with chief complaints of hyperkeratotic plaques over palms and soles since infancy, which

progressed to involve dorsal aspects of hands and feet (Transgradient). Later, skin lesions had involved more proximal parts like distal parts of forearms, legs, elbows and knees (Progradient). There is aggravation of skin lesions during winters. There is history of parental consanguinity. The past dental history revealed that her primary teeth had erupted normally. These underwent early shedding and complete shedding of all deciduous teeth took place by the age of 5-6 years. Her mother reported that all of the patient's deciduous teeth were hypermobile. There had been normal eruption of all permanent teeth, but gradually the permanent teeth also had started becoming mobile and was followed by the early shedding of the permanent teeth by the age of 13 years sparing one molar on each side. After this she visited a dentist and is using artificial dentures since then. There is no history of similar complaints in the parents or any of her siblings. She didn't give any history of recurrent infections. There is no history of upper abdominal pain or fever. On examination, there were bilaterally symmetrical hyperkeratotic plaques involving palms and soles which extended on the dorsal aspects of hands and feet and distal parts of forearm and legs (**Figure 1a,1b**). There were also hyperkeratotic scaly plaques over knees and extensor aspects of elbows (**Figure 1c**). Examination of hair and nails was normal. Oral examination revealed artificial dentures (**Figure 2a**) which on removal showed loss of all teeth (**Figure 2b**) except one molar (**Figure 2c**) on each side. There was also atrophy of maxillae which clinically presented as loss of maxillary prominences. Rest of the systemic examination was unremarkable. Her routine laboratory investigations were within normal limits.



Figure 1 Bilaterally symmetrical hyperkeratotic plaques over hands and feet which have involved dorsal aspects of both hands and feet (Transgradient). The plaques have also progressed proximally to involve lower parts of legs and wrist (progradients) (a,b). Hyperkeratotic plaques over extensor aspects of elbows and knees (c).

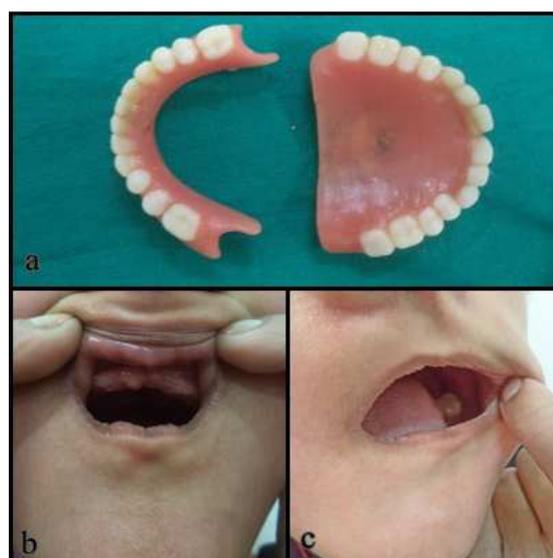


Figure 2 Artificial dentures used by the PLS patient (a). Loss of teeth with sparing of a molar on left lower side (b,c).

Though the etiology of PLS is not clear, several factors have been proposed which include genetic, immunological and microbiological factors. These patients have a genetic defect which has been mapped to chromosome 11q14-q21 involving mutations of the gene cathepsin C.^{4,5} More than 90% reduction in cathepsin C gene activity has been reported in PLS patients.⁵ This cathepsin C

gene encodes a cysteine-lysosomal protease known as dipeptidyl-peptidase I (DP-I). DP-I removes various dipeptides from the amino terminal of the protein substrate and also has an endopeptidase activity. The various epithelial regions of the body such as palms, soles, knees, and keratinized oral gingival mucosa express cathepsin-C gene which explains the localization of lesions in PLS. Various immune cells including polymorphonuclear leukocytes, macrophages, and their precursors express this gene which explains the immunological abnormalities in this syndrome. All PLS patients are homozygous for the cathepsin-C mutations while as parents and siblings, who are heterozygous for cathepsin C mutations do not show either the palmoplantar hyperkeratosis or severe early onset periodontitis characteristic of PLS.⁴ Reduced immunologic response and associated increased susceptibility to infection in PLS can be attributed to the defective phagocytosis, and impaired reactivity of B and T cells to various mitogens.⁶ Regarding microbiological aspects, Gram-negative bacteria are supposed to be the primary factors in the etiopathogenesis of periodontitis. Various reports have shown elevated antibody titers to *Actinobacillus actinomycetemcomitans* and *Capnocytophaga* in these patients which suggest the pathogenetic role of these bacteria in periodontitis.^{6,7}

The palmoplantar keratoderma usually starts between the ages one and four years. In our case it started during infancy. The sharply demarcated erythematous keratotic plaques may occur focally, but usually involve the entire surface of palms and soles. The keratoderma spreads to dorsal surfaces and up the Achilles tendon. Psoriasiform plaques may also progress proximally to involve the knees and elbows which was also seen in our case.^{8,9}

Severe periodontitis forms the second major component of PLS, which usually starts at the age of three or four years. The eruption and

development of primary teeth proceeds normally. However, their eruption is associated with severe inflammation of gums and subsequently leads to rapid destruction of the periodontium. This periodontitis does not respond to conventional periodontal treatment modalities. As a result deciduous teeth are exfoliated prematurely by 6 years. The gingival inflammation subsides after exfoliation and gums appear healthy as before. But, with eruption of the permanent teeth, the process of gingival inflammation and the subsequent periodontitis is repeated leading to exfoliation of the permanent teeth by 14-15 years. However, in some cases the third molars are spared.^{1,3,10} Apart from these basic features, other manifestations include liver abscesses, increased incidence of pyogenic infections, dural calcification and mental retardation.^{1,3}

There is no definitive treatment for this disorder. Several treatment options have been mentioned to control periodontal disease like conventional periodontal therapy, advice on oral hygiene, and use of systemic antibiotics. The viability of non-affected teeth can be prolonged by the identification of specific periodontal microbes and institution of specific antibiotic therapy together with the extraction of severely periodontally compromised teeth. Benefits from oral retinoids, such as acitretin, etretinate, and isotretinoin have been reported. Etretinate and acitretin have been reported to modulate the course of periodontitis and preserve the teeth in these patients and also improve keratoderma. At the stage of active periodontitis, a course of systemic antibiotics should be given to preserve teeth. This will also prevent bacteremia and subsequent development of pyogenic liver abscess. Any patient of PLS presenting with fever of unknown origin should be evaluated for pyogenic hepatic abscess.^{1,11,12}

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