Palmoplantar keratoderma with dental abnormalities

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Access this article online
Website: www.idoj.in
DOI: 10.4103/2229-5178.131152
Quick Response Code:

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A 6-year-old boy presented to us with a history of scaling, thickening and fissuring of the skin over his palms and soles of more than 3 years duration. The lesions were asymptomatic except for occasional pain over the fissures. There was no history of any other significant skin or mucosal lesions. He was the first child of a non-consanguineous marriage. The child had associated dental complaints including premature loss of primary teeth, for which he was referred to a dentist. The child also had a history of hypothyroidism detected at 45 days after birth for which he had been on thyroxine supplementation for 3 years. The child was otherwise healthy with no history of any other significant skin or systemic problems. The child was good in his studies and had a normal motor and psychosocial development history. There was no significant history of recurrent skin or systemic infections. There was also no history of any other significant and relevant skin or systemic illness in the family.

On examination yellowish hyperkeratotic plaques with fissuring and some crusting was seen over the palms and soles. Both the soles were completely, symmetrically and uniformly involved, while the palms showed only localized involvement [Figures 1 and 2]. The hair, scalp, and nails were normal. No abnormalities were detected on ophthalmic examination. No skeletal abnormalities were seen. Dental examination showed periodontitis with oedematous and friable gums and loss of primary teeth. The patient was advised oral prophylaxis, including maintenance of proper dental hygiene and further oral rehabilitation as needed in the future [Figure 3]. No other systemic abnormalities were seen.

A clinical possibility of Papillon-Lefèvre syndrome (PLS) was considered taking into account the palmoplantar keratoderma associated with dental defects.

A skin biopsy from the plantar lesions showed marked hyperkeratosis with parakeratosis, a slightly thickened granular layer and moderate acanthosis with psoriasiform hyperplasia [Figures 4 and 5]. The histopathology was consistent with the diagnosis of PLS. Other investigations including liver function tests, renal function tests, and echocardiography were within normal limits.

DISCUSSION

PLS is a rare autosomal recessive genetic disorder characterized by palmoplantar keratoderma associated with periodontitis. The onset of palmoplantar keratoderma in PLS may occur within the first 3 months of life, but usually, palmoplantar hyperkeratosis and severe periodontitis start simultaneously between the ages of 1 and 4 years.[1] The two forms of hereditary keratoderma which can be associated with severe periodontitis are PLS and Haim-Munk syndrome.[2] Haim-Munk syndrome is, however, associated with other characteristics features like onychogryphosis, pes planus, arachnodactyly, and acro-osteolysis.[3]

Clinically the keratoderma of PLS is characterized by sharply demarcated hyperkeratotic plaques usually involving the entire surface of palms and soles, and sometimes extending to the dorsa of the hand and feet. Other sites like the elbows, knees, and trunk, can be involved especially in older patients. Hyperhidrosis and secondary infection can be associated with the skin lesions. Periodontitis can affect both the deciduous and permanent teeth. Initially there tends to be gingival inflammation, which can be followed by destruction of periodontium, which in turn may lead to premature loss of primary teeth. Other rare associations reported with PLS are liver abscesses and recurrent pyodermas.[1]

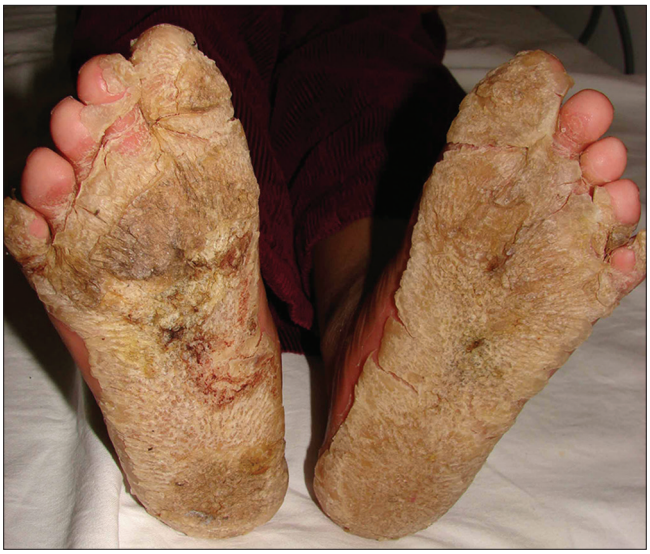

Figure 1: Hyperkeratotic plaques involving both soles symmetrically

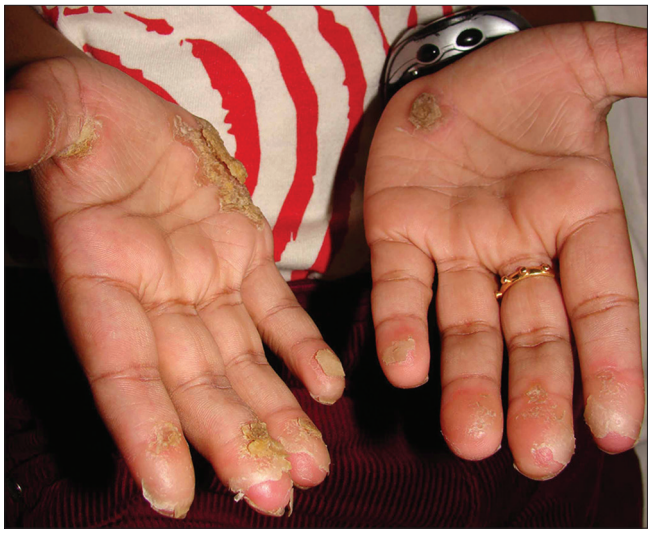

Figure 2: Hyperkeratotic plaques over both palms

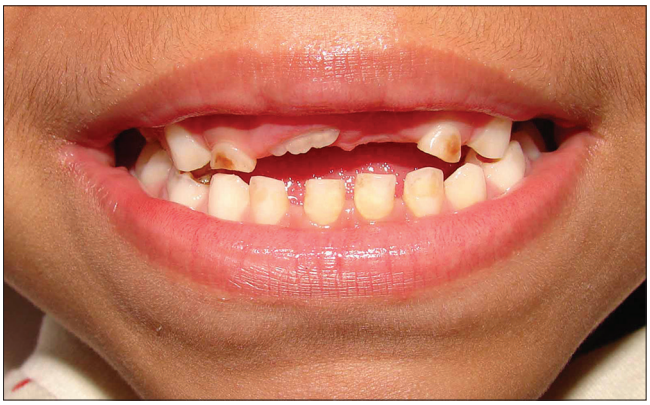

Figure 3: Dental abnormalities

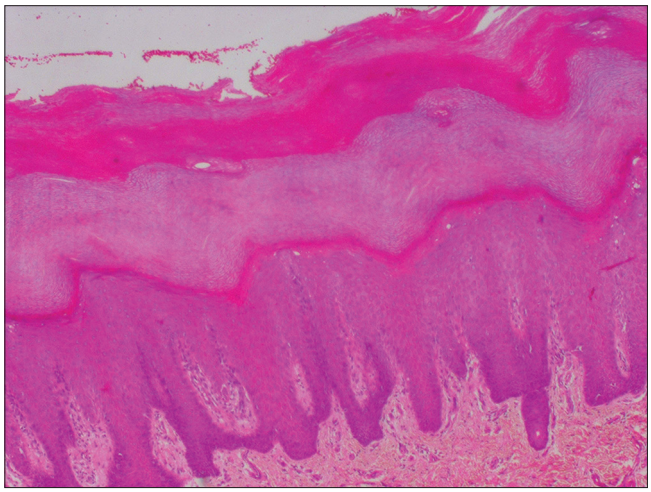

Figure 4: Marked hyperkeratosis, parakeratosis, acanthosis and psoriasiform hyperplasia (H and E, ×20)

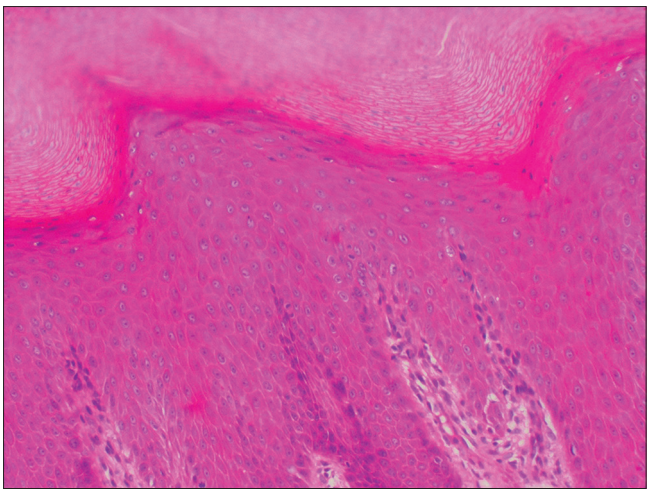

Figure 5: Acanthosis (H and E, ×40)

The exact etiopathogenesis of PLS has not been elucidated. Recent studies have suggested that loss of function mutations of the cathepsin C gene (CTSC) are associated with PLS.[4,5] However, several PLS cases exist in which mutations of the coding regions of CTSC have not been identified, although several of these families are consistent with genetic linkage to the interval of chromosome 11q14, which contains the CTSC gene locus. The exact mechanism by which defects in CTSC produce the clinical features of PLS is not clear but it has been demonstrated that CTSC is expressed in high levels in the epidermis of areas affected by PLS like palms and soles as well as in the keratinized oral gingiva. Reduced activity of CTSC can result in diminished activity of polymorphonuclear leukocyte-derived proteases like cathepsin G, which have an important role in immune responses to infections.[6]

Management of PLS requires a combined effort from the dermatologist and the dentist. Early treatment and prophylaxis is essential for the management of periodontitis. During deciduous dentition, oral hygiene instruction and prophylaxis must be given regularly. Teeth with advanced periodontal disease should be extracted. The primary drug of choice for

the treatment of keratoderma is oral retinoids. Our case was managed with oral isotretinoin (10 mg daily). The patient showed slow but definite improvement on follow-up at six months.Topical emollients, salicylic acid and urea preparations can be used as adjuncts.[1] It has been suggested that early initiation of retinoids (before the eruption of permanent teeth) can help ensure a normal dentition in cases of PLS.[7]

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Cite this article as: Kaliyadan F, Nambiar A. Palmoplantar keratoderma with dental abnormalities. Indian Dermatol Online J 2014;5:232-4.

Source of Support: Nil, **Conflict of Interest:** Nil.