PAPILLON - LEFEVRE SYNDROME
(Report of two siblings)
Case Report

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Summary

Two siblings with Papillon-Lefevre syndrome are described. Apart from the usual features, both had symmetrical progressive hyperkeratosis and dystrophy of all the nails.

KEY WORDS: Papillon-Lefevre Syndrome, Siblings, Progressive hyper keratosis.

In 1924, Papillon and Lefevre[1] described two siblings with a syndrome of hyperkeratosis of the palms and soles with premature loss of the deciduous and permanent teeth. Principal manifestation is palmoplantar hyperkeratosis usually appearing at 1 to 4 years of age with simultaneous periodontal involvement of deciduous dentition. Stein[2] observed that the degree of hyperkeratosis is most severe at the time of periodontal involvement. Gorlin et al[3] had reviewed most of the reported cases and found that this syndrome resulted from homozygosity for autosomal recessive genes. They also observed that the parents of affected persons did not manifest the disease but showed a high rate of consanguinity. Recently Handa et al[4] reported a case of this rare syndrome.

Report of cases

Case 1

A 17 years old male, attended the out-patient section of Dermatology and STD department of Medical College Hospital, Alleppey in July 1976. His complaint was progressive thickening of skin of both upper limbs and lower limbs, including that of palms and soles, and premature loss of teeth. The thickening of skin had first appeared on palms and soles, a few days after birth. Palms and soles were abnormally red and scaly at that time. Gradually the thickening of skin extended in an almost symmetrical manner to the outer aspects of fingers and toes. By the age of 5 years both hands and feet had become thick and scaly. During this period all his twenty nails had become dystrophic. The skin thickening had slowly progressed upto the elbows and knees, without leaving any normal skin in the involved areas. At about the age of thirteen years patient noticed a circular thickened and scaly area on the sacral region. This also slowly increased in size. Patient's developmental milestones were reported to be normal. Teeth erupted normally; but within a few months gums became red and swollen with pus coming out of the root of teeth. Gradually the teeth loosened and fell off one by one. Permanent teeth also erupted at the usual

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age, but were lost in the same way as the deciduous. Patient had difficulty in walking particularly during winter months because of the fissures at the margins of soles. He noticed slight improvement of the skin during the warm humid months, but had intolerable sweating at that time. Many times he had suffered from abscesses and ulcers on the thighs and legs. These used to heal only slowly.

This patient was the eldest among five children born of non consanguinous parents. His seven years old brother had similar complaints.

Systemic examination did not reveal any abnormality.

Diffuse hyperkeratosis with scaling was present distally on both upper limbs upto the elbows and on both lower limbs upto the knees. The involvement was strikingly symmetrical and stopped abruptly at the elbows and knees with well defined raised margins. The normal skin markings were exaggerated on the hyperkeratotic area. Scaling was most prominent at the margins and on the dorsal aspects of palms and soles (Fig. 1). Painful fissures were present at the edges of both heels. The soles were severely affected with marked hyperkeratosis, fissuring and had a foetid odour. There was a well defined large plaque on the back extending upto the sacral region with same features as that of lesions on other areas (Fig. 2). All the nails were onychogryphotic with Beau's lines. The distal ends of nail plates were curved downwards (Fig. 1). Nail folds, especially of the fingers were

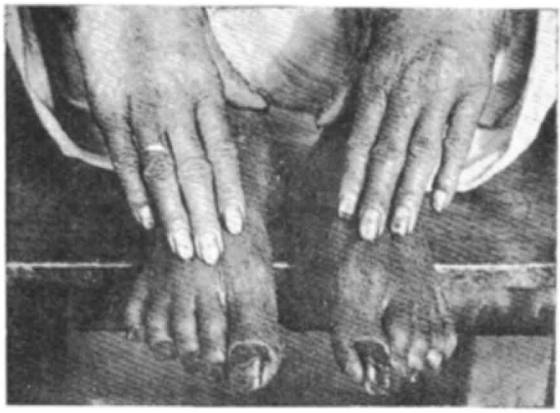

Fig. 1
Exaggerated skin markings, scaling and curved nail plates.

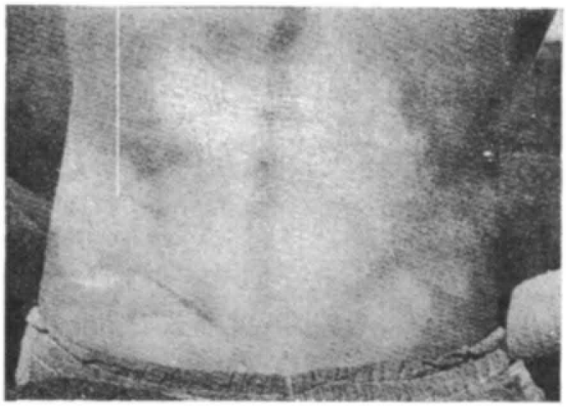

Fig. 2
Well defined scaly plaque on the back.

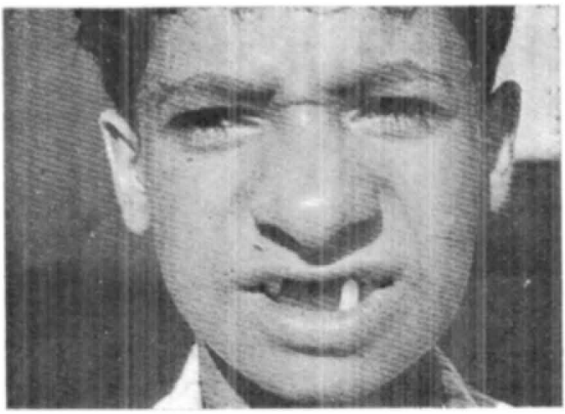

Fig. 3
Shows the premature loss of teeth,

thickened. All teeth except two canines and two premolars were absent (Fig. 3) and those present were loose with purulent exudate from the gingiva. Face was slightly scaly and erythematous. Hyperhidrosis was visible on the unaffected areas. There were multiple scars on the legs and thighs. There was an abscess on the left thigh.

The patient was examined again in July 1981 i.e., after 5 years, when he attended the hospital for multiple abscesses and ulcers. The hyperkeratosis had extended upto the shoulders and upto the hips leaving no intervening normal skin. The lesion on the back had extended anteriorly to the abdominal wall. The hyperhidrosis had become more severe on the normal skin areas. Patient had complete artificial denture.

Investigations

Routine blood and urine test results were within normal limits. Blood, VDRL, blood sugars and serum proteins were also normal. Nail clipping for fungus was negative. X-ray skull showed no calcifications.

Histopathology of the skin from the margin of the lesion showed marked hyperkeratosis and thickening of stratum granulosum. Some areas showed separation between the keratin layer and stratum malphighii. In the dermis perivascular inflammatory infiltration was seen.

Case 2

Younger of the two affected siblings was a 7 years old boy. He also had complaints of thickening of palms and soles with premature loss of teeth. Thickening of palms and soles was evident about a month after birth. The thickening of skin gradually extended and involved both hands and feet. Mile stones of development were normal. Teeth erupted at the expected time, but were lost in the same way as that of his brother.

Systemic examination did not reveal any abnormality.

He had marked hyperkeratosis of both hands and feet. On the upper limbs the process had extended upto about 4 cms above the wrists and on the lower limbs to about 7 cms above the ankles (Fig. 4). Scaling was very prominent on the dorsal aspects of feet. There were multiple fissures on the edges of both heels. All the nails were dystrophic with onychogryphosis, Beau's lines and subungual hyperkeratosis. Nail folds were thickened (Fig. 4). Lower canines and incisors were loose and discoloured. Upper

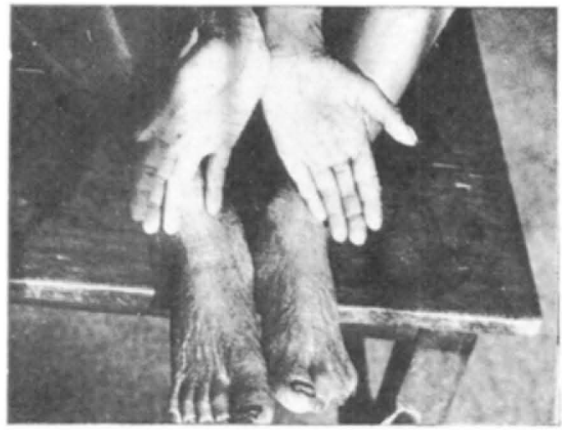

Fig. 4
Hyperkeratosis, and scaling of skin with nail dystrophy in the younger sibling.

incisors and canines were lost. Gingivae were inflamed and produced an offensive odour.

Routine blood and urine examination results were normal. Blood VDRL test was non reactive. X-ray skull did not show any calcification of tentorium.

Comment

Papillon-Lefevre syndrome is a rare condition. Usually hyperkeratosis of palms and soles appears between 1-4 years of age[4]. In the cases described here the hyperkeratosis of palms and soles appeared a few days after birth. An unusual feature in these two cases is the progressive hyperkeratosis, which is not described in the earlier reports. Even after all the teeth were lost, the hyperkeratosis kept progressing. Involvement of legs and thighs was present in the case reported by Ziprokowski et al[5]. Hyperkeratotic plaques were present on eye lids, cheeks and labial commisures in the case reported by Hawes[6]. Nail dystrophy was present in both patients. There were multiple Beau's lines and onychogryphosis of all nails with thickening of the nail folds. These changes can be explained by the hyperkeratosis of dorsal aspects of fingers and toes with periodic inflammation. Hyperhidrosis was seen on the unaffected skin. The affected areas were dry and scaly. During hot humid climate the hyperhidrosis was severe and it was less during cold months. This appears to be a compensatory hyperhidrosis for temperature regulation. Physical development was normal in both cases.

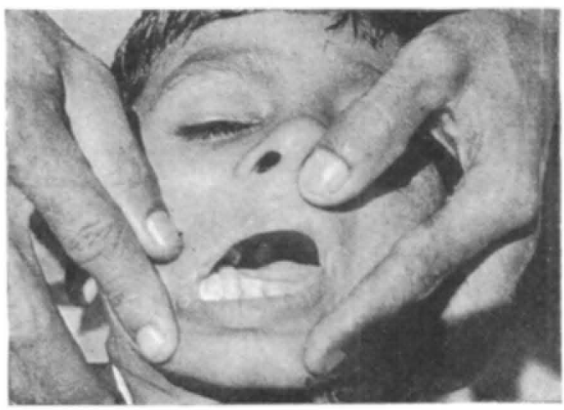

Fig. 5
Premature loss of teeth in the younger sibling.

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