PAPILLON - LEFEVRE SYNDROME

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Five cases of Papillon-Lefevre syndrome occurring in 2 families were seen to have variable clinical features. Two of these cases treated with etretinate showed a good response.

Key words : Papillon-Lefevre syndrome, Etretinate.

Papillon-Lefevre syndrome (PLS) is a rare syndrome; about 50 cases having been reported in the world literature.[1] This syndrome was first described by Papillon-Lefevre in 1924. The first Indian case was reported by Sardari Lal et al in 1971.[2] There is a high incidence of consanguinity and an autosomal recessive pattern of inheritance has been assumed.[1] The principal manifestations are keratosis palmaris et plantaris, premature peri-odontoclasia and calcification of dura. Palmo-plantar keratoderma usually starts between the first and the fourth year of life. Keratoderma involves not only the palms and soles, but also the back of heels, external malleoli, tibial tuberosities, elbows, dorsa of hands and feet. The maximum degree of hyperkeratosis coincides with active peri-odontal disease. Calcified deposits occur in the tentorium and falx, but their age of onset is unknown. Deciduous teeth erupt and develop at their normal time and in normal sequence, but with the development of palmo-plantar hyper keratosis, peri-odontal abscesses appear and destruction occurs and deciduous teeth are lost. The permanent dentition follows a similar process. Characteristically, the patients having PLS are completely edentulous by the age of 16 years.

Reports in the literature indicate occasional association with hypertension, hyperglycemia, systolic murmurs, retardation of skeletal maturation, osteoporosis and thyroid enlargement.[1]

We are reporting five cases from two families to demonstrate the successful management of two cases with retinoids, the most promising drugs so far in the management of disorders of keratinization.[3]

Case Reports

Out of the five cases, three belonged to one family as shown in the pedigree chart (Fig. 1).

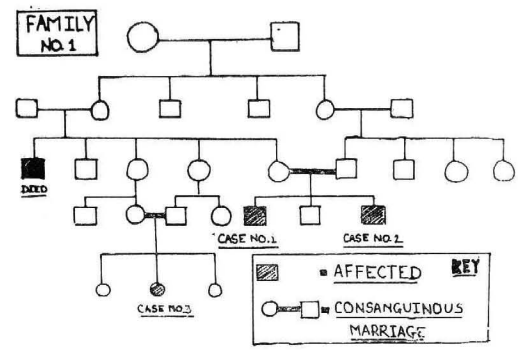

Fig. 1. Pedigree chart of the first family.

Case 1

A 27-year-old male had palmo-plantar keratoderma and hyperkeratotic scaly lesions covering both extremities. The palmo-plantar hyperkeratosis was first noticed when the patient was 6-month-old. Soon afterwards, hyperkeratotic plaques appeared on the dorsa of hands and feet, which gradually spread to involve both the forearms and legs, and at the time of diagnosis had reached upto the mid-arms and mid-thighs.

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The patient had a normal eruption of both deciduous and permanent teeth. Both dentitions evolved into peri-odontal inflammation and abscesses with subsequent loss of all teeth. He was totally edentulous by the age of 17 years.

The palms and soles had deep fissuring at places especially on the soles. All the nails were grossly deformed and destroyed. General and systemic examination revealed no abnormalities.

Case 2

A 23-year-old male, brother of case 1 also had a similar massive and crippling palmo-plantar keratoderma on the dorsa of hands, extending upto the distal portion of the forearm. Plaques were present on the elbows and the entire lower extremity from the dorsa of the feet upto the mid-thighs. Loss of teeth occurred in a similar fashion as in patient 1, leaving him totally edentulous by the age of 16 years. All the nails were grossly deformed and destroyed.

Case 3

A 13-year-old female child related to cases 1 and 2, also had a comparatively milder palmo-plantar keratoderma which was slowly progressive since it was first noticed at the age of 8 months. Hyperkeratotic scaly lesions extended only upto the dorsa of hands and feet. Rest of the extremities were devoid of any lesions. Nails were not involved. She had had a normal dentition of permanent teeth, but subsequently, these had started falling, leaving her with only two intact teeth.

The remaining two cases belonged to the other family (Fig. 2).

Case 4

A 23-year-old male had palmo-plantar keratoderma, along with hyperkeratotic scaly plaques on the dorsa of hands and feet and elbows and knees. He had started losing his teeth in young age, but 3 incisors were still present at the age of 23 years. He was also suffering from lupus vulgaris for which he was

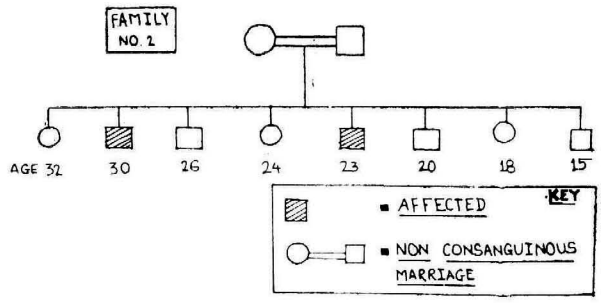

Fig. 2. Pedigree chart of the second family.

taking treatment. X-ray skull showed dural calcification.

Case 5

A 27-year-old male, brother of case 4, had similar skin lesions, but had total loss of teeth.

Routine investigations of all the patients were within normal limits. Cases 1 and 4 had minimal calcification of the falx but there was no evidence of intra-cerebral calcification in patients 2, 3 and 5. The lipid profile and LFT of patients 1 and 2 were normal.

Patient 1 was given etretinate 50 mg daily (0.75 mg/kg). He showed dramatic improvement by the end of three weeks. Patient 2 was also given the same treatment with a similar dramatic response. In both the cases, the dose was reduced to 0.5 mg/kg daily and then to 0.5 mg/kg on alternate days without any recurrence.

Comments

Various clinical variations noted in our patients included : (1) Glove and stocking distribution of hyperkeratosis which extensively involved both the extremities in case 1, and extensive hyperkeratosis of only the lower extremities in case 2 are rather unusual. Generally, only localized plaques of hyperkeratosis, restricted to the dorsa of hands and feet, knees and elbows are seen. Such a localized pattern was typically seen on the upper extremities of cases 2, 4 and 5; and on the lower extremities in cases 4 and 5. Case 3 showed no

lesions on the elbows and knees. (2) None of our patients had exacerbations of hyperkeratosis coinciding with active peri-odontal disease, as is classically described. Their hyperkeratosis was relentlessly progressive, irrespective of any factors. Case 4 had three incisors left even after the age of 23 years. (3) Dural calcification was not seen in cases 2, 3 and 5. (4) A dramatic response to etretinate therapy was seen in both the cases.

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