PAPILLON - LEFEVRE SYNDROME WITH MENTAL RETARDATION

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A 9 - year - old boy, born of consanguineous parents, presented with keratoderma of palms and soles associated with severe enamel hypoplasia and mental retardation. A clinical diagnosis of Papillon - Lefevre was supported by histopathology.

Key words : Keratoderma, Papillon - Lefevre syndrome, Mental retardation, Enamel hypoplasia

Introduction

Papillon - Lefevre syndrome is a rare disorder of keratinisation characterised by keratoderma of palms and soles, associated with periodontosis and tendency to pyogenic skin infections.[1] Mental retardation, ectopic intracranial calcifications and acro- osteolysis are rarely described associations.[2-5] We report here, a case of this rare syndrome with associated mental retardation.

Case Report

A 9-year-old boy presented with gradually progressive keratoderma of palms and soles with severe fissuring. The thickening of soles had first been noticed by the parents when he was 2 year old, followed by involvement of palms. The parents also complained of impaired mental functions in the child and this had prevented him from attending school. Examination revealed an intellectually impaired boy who could follow verbal commands with difficulty. Cutaneous examination revealed thickening of palms and soles with fissuring encroaching to the dorsa of toes and feet, with mild localised thickening of elbows and knees. In addition, the patient had severe enamel hypoplasia with upper central in-

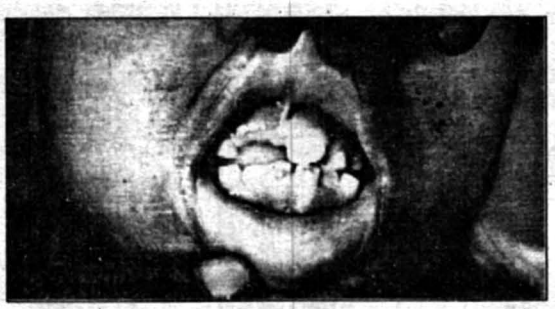

Fig 1. Teeth with severe enamel hypoplasia, gingivitis and serrated margins.

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cisors showing serrated and irregular surfaces (Fig1). X-ray of maxilla (Water's view) and central incisors revealed no abnormality of the roots, and x-ray skull did not show any evidence of intracranial calcification. The child was born to consanguineous parents. No other family member had similar cutaneous manifestations.

A skin biopsy from sole showed hyperkeratosis and acanthosis, features consistent with diagnosis of keratoderma.

Discussion

Palmoplantar keratoderma is a group of inherited dermatoses characterised by thickening of palms and soles, associated with other systemic abnormalities. The most satisfying criterion for classifying the group is to separate it into two distinct groups - a dominant and a recessive one. Further subclassification must consider the morphological aspects and other associations. Histopathology is essentially similar in all subgroups. Papillon-Lefevre syndrome is an autosomal recessive type characterised by association of periodontosis resulting severe gingivitis and recurrent pyogenic infections of the skin. Our patient, in addition, to severe enamel hypoplasia and gingivitis also had mental retardation which is one of the uncommon associations described in literature. Local therapy in the form of emollients with keratolytic was instituted with satisfactory results.

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