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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
PACHYONYCHIA CONGENITA

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Summary

Pachyonychia congenita has been reported in two cases in a family—father and son. They also had other multiple epidermal defects like steatocystoma multiplex, callosities, alopecia with hypotrichia and follicular keratinosis. The younger patient had delayed eruption of the teeth.

KEY WORDS: Congenital pachyonychia, steatocystoma multiplex, alopecia, follicular keratinosis, delayed eruption of teeth.

Introduction

Pachyonychia congenita was first described in 1906 by Jadassohn and Lewandowsky. In this disorder nails are conspicuously deformed but the disorder may involve the skin, oral mucosa, hair, eyes and teeth. In the present communication two cases in a family—father and son, were found to be suffering from this rare disorder.

Case Report

Case No. 1

A 10 years old boy had thickened nails of all the toes and fingers since the age of 3 months (Fig. 1). Loss of hair was noticed in the occipital region at the age of two years. On examination, the hairs were thin and distorted. Microscopic examination of the hair revealed hypotrichia and occasional nodulation. Extensive follicular keratotic lesions (Fig. 2) were present on the scalp, neck, trunk as well as the limbs. Callosities over the pressure points of plantar and dorsal aspects of the feet had been present since the age of four years. The palms were normal. There was no history or evidence of hyperhidrosis or bullae on the soles. The teeth showed yellowish discoloration. The patient had delayed eruption of the teeth starting at the age of 17 to 18 months. This information was emphasized by the father without any direct questioning. The eyes and oral mucosa did not reveal any abnormality.

Case No. 2

A 46 years old man, father of case I, presented with thickening of all the twenty nails, loss of hair in the occipital region with callosities over the pressure points of feet. He showed evidence of keratotic follicular lesions on the scalp and neck only. He also had multiple yellowish nodules, soft in consistency, varying from 0.5 cm to 4 cm in
Discussion

Pachyonychia congenita is a rare genodermatosis transmitted as an autosomal dominant trait with incomplete penetrance. The name pachyonychia congenita denotes congenital thickening of the nails and is a misnomer, as this is not the only abnormality. The term however is being retained because of traditional use. The thickening of the nails is the predominant abnormality which is usually associated with other hereditary dyskeratoses. Jackson\(^1\) reported six cases in one family spanning three generations, with each affected member demonstrating dyskeratosis of the nails of the hands and feet and erupted teeth at birth. Three members had epidermal cysts of the face and neck. Soderquist and Reed\(^3\) also reported three patients in three generations of a family with dyskeratosis of the nails of hands and feet, epidermal cysts, callosities of the feet with blistering, hyperhidrosis, unusual hair texture and erupted teeth at birth. Other changes which have been reported are keratosis pilaris, leuko-keratosis oris and steatocystoma multiplex. Joseph\(^4\) reported five cases and especially studied the pathogenesis of nail changes in an infant afflicted with this disorder. He observed hyperkeratosis and hypertrophy of the ventral nail and demonstrated dyskeratosis of the nail root with the presence of a granular layer.

In the cases reported herein, the changes observed are thickening of a
Alopecia with extensive follicular keratotic lesions on the scalp and neck.

Fig. 2

Steatocystoma multiplex lesions on the neck and upper trunk.

Fig. 3
the twenty nails, alopecia with defective hair texture, callosities over the soles, follicular keratosis and steatocystoma multiplex. The unusual feature in our younger patient is delayed eruption of teeth which is in contradiction to the erupted teeth at birth reported by other workers\(^8\). The present cases are of interest because of the presence of multiple epidermal defects especially steatocystoma multiplex, alopecia and delayed eruption of teeth in one of them.

References