Pachyonychia Congenita Project

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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.

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Pachyonychia congenita (A case Report)

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Abstract

A case of "pachyonychia congenita" rare ectodermal dysplasia is presented. Literature is reviewed and discussed.

Introduction

Pachyonychia congenita is a rare ectodermal dysplasia in which nails are conspicously deformed and may also involve the skin, oral mucosa, hair, eyes and teeth. It was first described by Jadassohn and Lewandowsky'. Its association with other hereditary dyskeratosis, such as planter and palmer callosities, bullae, keratosis pilaris, icthyosis, hyperhydrosis, leukeratosis oris and steatocystoma multiplex has been well documented. Murray[3] first noted the correlation between the presence of erupted teeth at birth and pachyonychia congenita.

Case Report

A 12 years old male child attended the hospital with complaints of thickened and abnormal nails. His birth order was 8th of 10 full term normal delivery, had 4 teeth at birth which fell down by 3 months of age. There was no similar deformity in family.

Physical examination revealed small leucoplekic patches in the centre of tongue of variable size. Patch near the base of tongue was large as compared to other two small patches. There was uniform keratosis pilaris more over the covered parts of the body with tiny cutaneous horny excrescences, this was more marked on lower half of the abdomen and upper part of the thighs anteriorly as well as on the back (Fig 1). Few callosities were seen on the sole but no generalized hyperkeratosis of sole was observed All the nails were thickened and compressed from side to side (Fig 2).

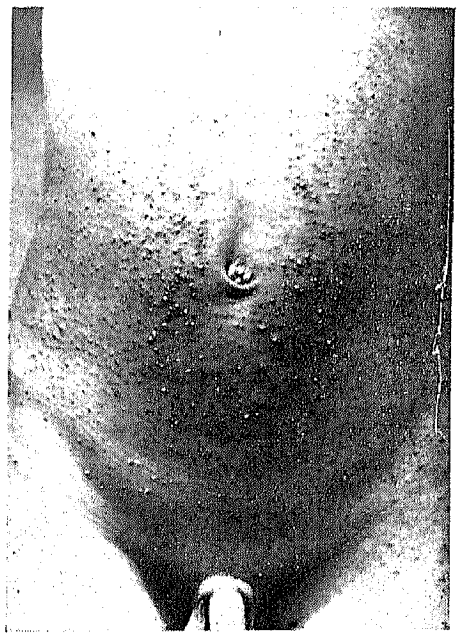

Fig. 1.

Hairs were normal. There was no history of bullous eruption of skin. Ear examination did not reveal tympanic membrane thickening but left drum was retracted and associated with deviated nasal septum on the same side. Ophthalmic examination with slit lamp showed persistant pupillary membrane and band at 10 O'clock position with thread like extension upto 2.0' clock position in the left eye but no thickening of cornea or lens opacity was observed. (Histological examination from the skin, mucous membrane and nails were refused by the parents).

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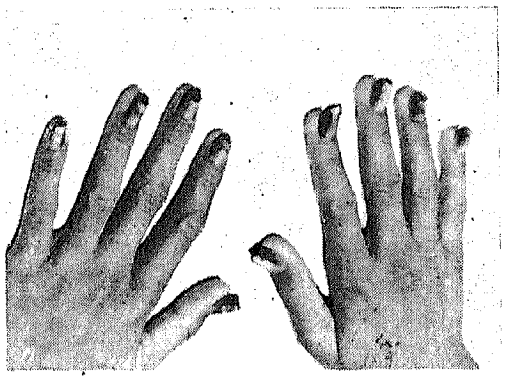

Fig. 2.

Comments

Vineyard et al[4] reported 8 cases in four generations along with stearocystoma multiplex with pachyonychia congenita and considered to be transmitted as a simple mendelian dominant with incomplete penetrance. Joseph et al[5] reported five cases in three generations and concluded histologically that the thickening was due to abnormal keratinization in terminal matrix. The mode of inheritance was autosomal dominant. But in none of the cases skin lesions including hyperkeratosis pilaris with tiny cutaneous horny excrescences were observed. Soderquist et al[6] reported 3 cases of pachyonychia congenita in 3 generations of same family with epidermal cyst and other congenital dyskeratosis. They observed presence of hyperhydrosis, callosites and frequent blistering of feet but other dyskeratosis involving mucous membrane and eyes were absent. Similarly a case has been reported by Wright et al[7] in which father and son, both were affected, father was a son of unaffected parents and this was explained on the basis of new mutation. Our case being a single isolated case in whole family, could be explained on the basis of fresh mutation and it differs from other cases reported, having typical skin lesion and oral leukokeratic patch over tongue, absence of hyperhydrosis and bullous eruptions.

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