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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Supernumerary digits associated with pachyonychia congenita type I

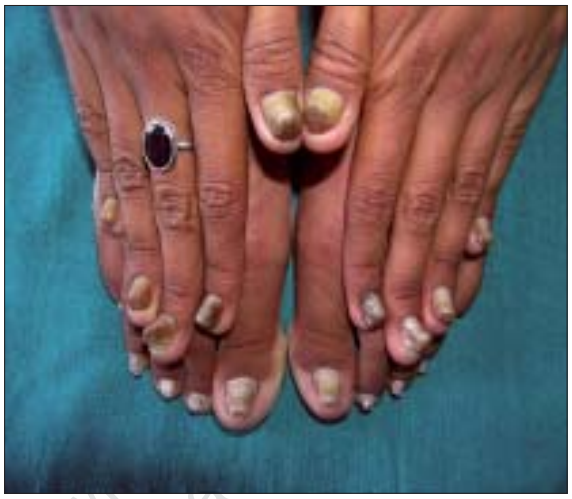

Figure 1: Thickening of nail plates, subungual hyperkeratosis, yellowish brown discoloration and distal upward growth of nail plates

Sir,

An 18 year-old girl presented with thickening and discoloration of all nails, painful callosities on soles, whitening of tongue and papules over extensors of legs and back. On enquiry, it was discovered that discoloration of nails started within a few months of birth. Gradually nails became so thick and hard that by the age of four years, it was difficult to cut the nails. Whitening of tongue, callosities on the soles and papules over the extremities and back developed later. There was no history of palmo-plantar hyperhydrosis, bullae, cysts, hoarseness of voice, teeth malformation, hair loss or any systemic complaints. No family history of similar nail changes or other complaints was present. The patient was born of nonconsanguineous parents.

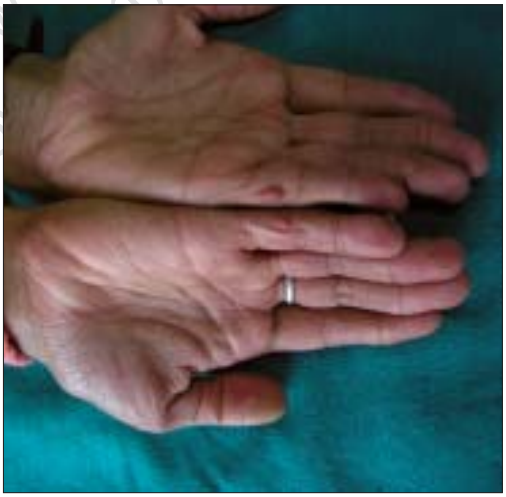

Figure 2: Supernumerary digits (postaxial) on the ulnar side of hands

Examination revealed marked thickening and hardening with yellow-brown discoloration of all nail plates; fingernails were more thickened than toenails. The nail plates were uplifted by subungual hyperkeratosis with upward growth of the distal portion [Figure 1]. Hypercurvature on the transverse axis of all nail plates was present giving a pinched shape to the free edges. Focal palmoplantar keratoderma was present at pressure areas. She had oral leukokeratosis in the form of white diffuse opaque thickening of the tongue and buccal mucosa. Apart from this, there were hyperkeratotic follicular papules over extensors of the extremities and trunk. In addition, she had supernumerary digits on the ulnar aspect of her hands [Figure 2]. X-ray of hands did not reveal any bony element in the supernumerary digits. There was no history of supernumerary digits in the family. Hair, teeth, ophthalmological and otorhinolaryngological examinations were normal. Potassium hydroxide examination and fungal culture of nail plates and tongue scraping did not show any fungal element. Nail bed biopsy was refused. She was put on topical keratolytics and oral vitamin A (50000 i.u.) due to the family's inability to afford acitretin. Hyperkeratotic papules responded without any improvement in nails and oral leukokeratosis.

Pachyonychia congenita is a rare, heritable disorder of the skin first described by Jadassohn and Lewandowsky in 1906.[1] The syndrome is the expression of a mutant autosomal gene with dominant effect but variable expression. Rarely has autosomal recessive inheritance been reported.[2] Family history in our patient was absent which may be explained as a case of incomplete penetrance within the family or as a sporadic case. The extent of mutation in the highly conserved 1A domain of K6, K16 and K17 (chromosome 17) is associated with various abnormalities associated with pachyonychia congenita.[3]

Two distinct types[4] are described under the term Pachyonychia congenita; our patient had most of the features of pachyonychia congenita type I except hyperhidrosis and blisters. In addition, she had rudimentary supernumerary digits, which may be a chance occurrence or a new association of pachyonychia congenita. To the best of our knowledge, supernumerary digits have not been described with pachyonychia congenita so far. There are a few reports of isolated nail dystrophy[5] or ectopic nail[6] associated with polydactyly. The most common form of a supernumerary digit is the postaxial polydactyly (PAP), in which the additional digit, which may be well formed (PAP-A) or rudimentary (PAP-B), arises from the ulnar border of the hand, at or near the base of the fifth digit as seen in our case. Postaxial polydactyly usually occurs sporadically but autosomal inheritance is seen frequently. Loci on chromosome 13 and 7 have been identified for postaxial polydactyly.[7] It may appear in isolation or rarely in association with other birth defects. Several syndromes have been identified that include postaxial polydactyly as associated clinical manifestations. Among these are trisomy 13, acrocallosal syndrome, Ellis van Creveld syndrome, Meckel Gruber syndrome, McKusick-Kaufman syndrome.

In a nutshell, association of supernumerary digits with pachyonychia congenita type I is being reported for the first time which may be a chance occurrence or a new association.

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