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.

2386 East Heritage Way, Suite B, Salt Lake City, Utah 84109 USA
Phone +1-877-628-7300 • Email—Info@PC-Project.org
www.PC-Project.org

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Letter to the Editor

PACHYONYCHIA CONGENITA WITH PATENT DUCTUS ARTERIOSUS

PRANESH NIGAM, R.D. MUKHIJA* AND K.K. KAPOOR**

To the Editor,

Pachyonychia congenita is an unusual genodermatosis characterized by extensive hyperkeratosis of the nails, skin, mucous membrane and hair, which may be present at birth or start in early infancy. Changes in the nails may be the only abnormality in the majority of the cases but it may involve skin, mucous membrane, hairs, teeth, palms, soles and eyes. Epidermal cysts and premature eruption of the teeth have been reported in some cases (1-3). To the best of our knowledge, to date no one has reported the association or co-existance of pachyonychia congenita with other systemic congenital disorders (CVS). The present communication reports this rare syndrome associated with an congenital heart disease.

CASE REPORT

Thirteen year old Hindu male attended the Skin O.P.D. for his thickened and deformed nails since birth and recurrent inflammation with swelling of the nail folds. He developed keratoderma of palms and soles at the age of 5 years. There was unusual increased sweating (hyperhydrosis) of palms. Extrusion of thick, hard, hyperkeratotic material from underneath the nails after every attack of inflammatory swelling of nail folds, had been a constant feature since then.

On examination there was marked wedge-like thickening of all fingers and toe nails with a prominent transverse curve and discolouration without nail loss. Marked subungual hyperkeratosis caused elevation of the free margins of the nails. Markedly tender, blister like swelling and erythema of the right middle and index finger was seen at the time of examination exhibiting a bead of purulent discharge on pressure. Keratoderma of palms and soles over pressure sites were seen. Palms showed presence of unusual wetting (hyperhidrosis). A small patch of leukoplakia was seen on left cheek and lower lip which was not noticed by either the patient or his father.

Systemic examination revealed only a typical continuous machinery murmur best heard over the second left intercostal space. Murmur was more prominent in recumbent posture and during inspiration without any splitting of the second heart sound (P_2).

All relevant investigative findings were normal except for slight prominance of the main and peripheral pulmonary arteries in the P.A. view X-ray

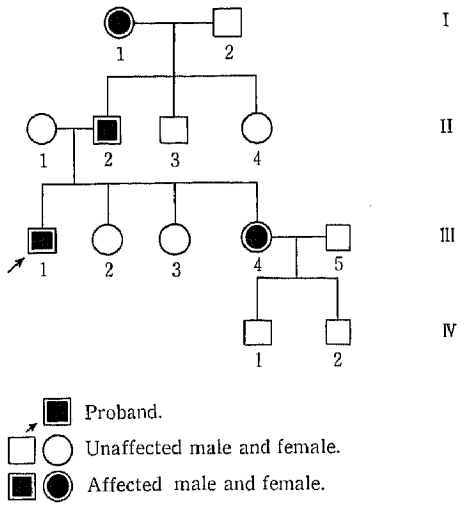

Fig. 1. Family tree showing autosomal dominant mode of transmission.

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Reader in Medicine, *Professor of Dermato-venereology, **Lecturer in Medicine, B.R.D. Medical College, Gorakhpur-273013, India.

chest. Nail and skin scrapings in 10% KOH and with Swartz stain were negative for fungal spores and hyphae.

On interrogation, the patient's father (Fig. 1, II-2) confessed that so far as he remembered his mother (I-1) had had similar nail lesions, which he also had. He had greyish white plaques (leukoplakia) on the right side of tongue in addition to nail involvement. His daughter (aged 23 years, III-4) had the same type of nail lesions but her sons were normal (IV-1 & 2). The family tree is typical of autosomal dominant trait inheritance.

DISCUSSION

Pachyonychia congenita is a rare genodermatosis, inherited as a simple Mendelian dominant trait characterized by incomplete penetrance. Sarojini et al. (2) suggested an autosomal recessive inheritance in their case report. The majority of the patients present with discolouration, variable thickening, and subungual hyperkeratosis of the nails as the only abnormality. One of the present three cases (Fig. 1, III-4) had abnormalities of the nails only. Other features such as palmo-plantar keratoderma, hyperhydrosis, bullae, ichthyosis, keratosis pilaris, steatocystoma multiplex, corneal dyskeratosis, cataract, hoarseness of voice, and caries teeth may be associated with nail changes in some cases. Lesions on the oral mucous membrane and tongue are often seen as leukoplakia, as in two cases of present three (II-2, III-1), where this disease was diffuse (4). Mucous membrane of the nose, larynx, anal canal and tympanic membrane can also be involved in a few patients. The present case (III-1) had combination of nail involvement with palmo-plantar keratoderma, hyperhydrosis and leukoplakia of the oral mucous membrane.

We did not come across any report describing this rare disease combined with congenital heart disease. Our present case can be categorized as type III as classified by Joseph (5) who also saw small patent ductus arteriosus (Crouzon Syndrome).

The family tree revealed (Fig. 1) autosomal dominant inheritance as in most other reports (3, 4). The present case had patent ductus arteriosus, whose incidence in full term infants is about 1 in 2000 live births, accounting for about 10% of all types of congenital heart disease (6). Persistent patency of the ductus arteriosus may occur in more than one member of a family, suggesting possible genetic factors in certain instances. Many genes are required in cardiac organogenesis and, if one mutant gene is going to produce cardiac maldevelopment, it should be a gene of large defect. In general, genes of large defect influence the development of more than one structure; thus the explanation of syndrome rather than a discrete cardiac anamoly. In this case, too, there might be a large defect in the gene which leads to this association of two congenital disorders. We must look for systemic anamolies in patients with Pachyonychia congenita.

SUMMARY

Three cases of Pachyonychia congenita, a rare genodermatosis affecting predominantly skin and mucous membrane in a family with various manifestations, are reported. Nails showed subungual hyperkeratosis with discolouration. Keratoderma of palms and soles at the pressure sites became evident at the age of 5 years in one case only who later on developed hyperhydrosis. Small patches of leukoplakia were present in two cases. Small patent ductus arteriosus was detected at the time of clinical examination in one patient and diagnosed as Crouzon syndrome. The family tree analysis showed an autosomal dominant mode of transmission.

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