



# 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.

## Pachyonychia Congenita Associated with Steatocystoma Multiplex

Joong Uhk Kim, Toshitatsu Nogita, Satomi Terajima and Makoto Kawashima

### Abstract

We present an unique case of pachyonychia congenita associated with steatocystoma multiplex. A 33-year-old Japanese man had thickening and gray-brown discoloration of all nails and a large number of nodules or tumors over his entire skin. No palmar and plantar hyperkeratosis, leukokeratosis of the mucous membranes, or follicular keratosis were observed. Histology of these tumors revealed the typical features of steatocystoma multiplex.

*Key words:* pachyonychia congenita; steatocystoma multiplex

### Case Report

A 33-year-old Japanese man had numerous nodules, tumors, and thick nails. Thickening and discoloration of all his nails had been noticed since birth. The nodules developed on his chest and extremities and progressively increased in size, number, and extent at the age of six months. None of his family had similar lesions.

Physical examination revealed thickening and gray-brown discoloration of all nails (Fig. 1). Hundreds of variable-sized, skin-colored, yellow, or bluish nodules or tumors spread over his entire skin (Fig. 2). No palmar and plantar hyperkeratosis, leukokeratosis of the mucous membranes, or follicular keratosis were observed.

Biopsy specimens were obtained from a skin-colored nodule and a bluish nodule. Their histological features were similar: a cyst in the mid-dermis whose wall consisted of two or three layers of squamous epithelium and contained sebaceous lobules (Fig. 3). These cysts were diagnosed as steatocystoma multiplex.

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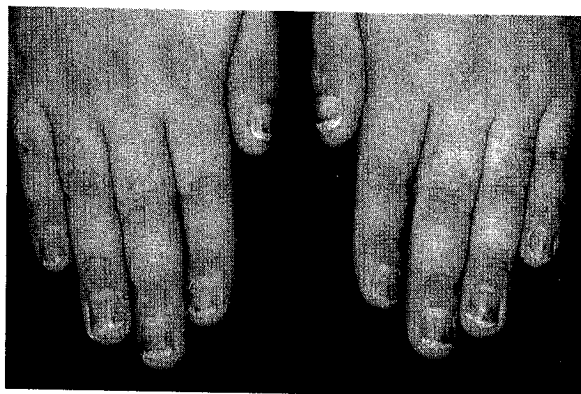


Fig. 1. Thickening and discoloration of the nails of both hands.

### Discussion

Based on the clinical features, no history of trauma, absence of varix syndrome or neurological disorder, and histological features, we diagnosed this case as pachyonychia congenita associated with steatocystoma multiplex.

Pachyonychia congenita is a rare hereditary disorder characterized by nail dystrophy, palmar and plantar hyperkeratosis, leukokeratosis of the mucous membranes, and follicular keratosis (1-3).

Akesson stated that one must find hypertrophy of all nails and dyskeratotic skin lesions to make the diagnosis of pachyonychia congenita (4). Feinstein et al. (5) comment-

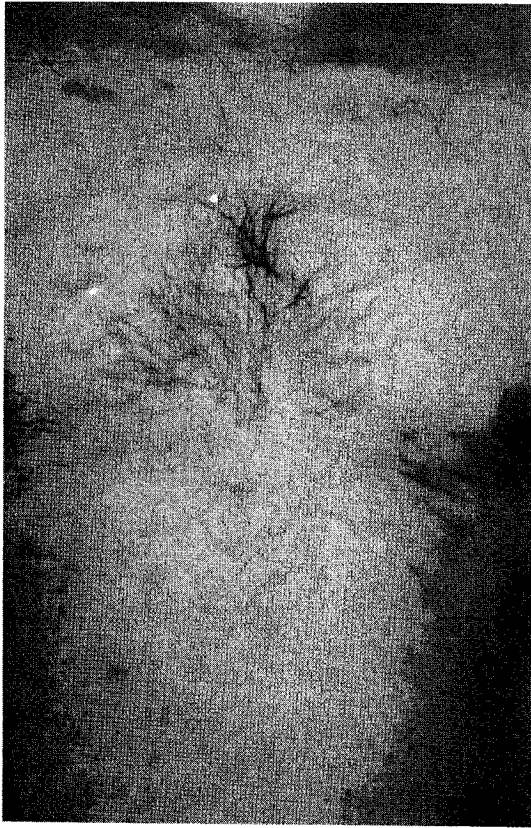


Fig. 2. Hundred of nodules and tumors on the anterior chest.

ed that two conditions should be differentiated from pachyonychia congenita: 1) thickening of nails because of trauma and 2) congenital onychogryphosis. On the basis of 168 cases studied, they proposed a new classification: type I, hyperkeratosis of nails, palmoplantar keratosis, follicular keratosis, and oral leukokeratosis; type II, clinical findings of type I plus bullae of palms and soles, palmar and plantar hyperhidrosis, natal or neonatal teeth, and steatocystoma multiplex; type III, clinical findings of type I and II plus angular cheilosis, corneal dyskeratosis, and cataracts; and type IV, clinical findings of types I, II, and III plus laryngeal lesions, hoarseness, mental retardation, hair anomalies, and alopecia. According to their classification, our case can be classified into type II.

Pryce and Verbor (6) reported a family with pachyonychia congenita in which affected individuals showed nail involvement only. They suggested that the observed interfamilial variation was due to genetic heterogeneity based on different mutations at the same locus or mutations at different genetic

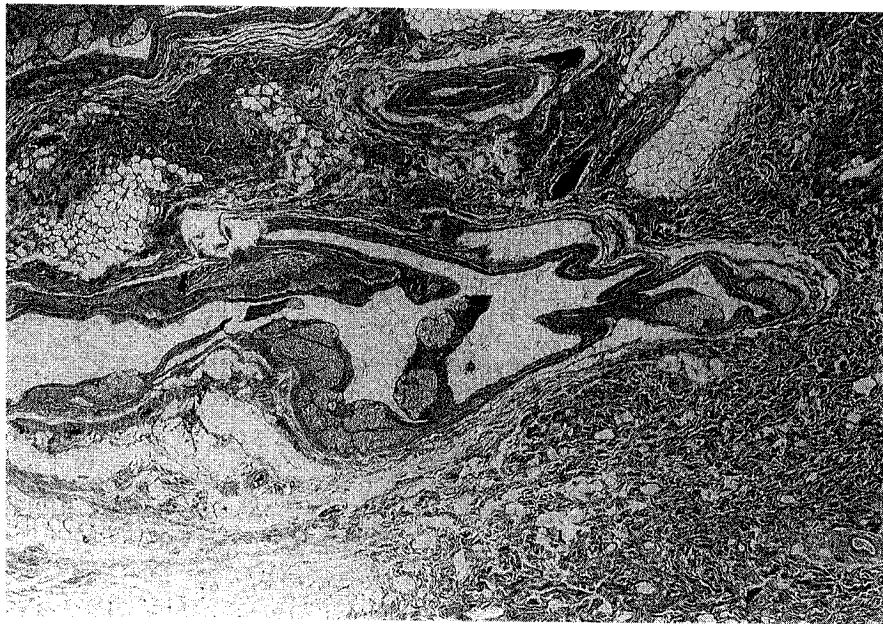


Fig. 3. Infolded cyst wall consisting of several layers of squamous epithelium. Cyst contains sebaceous elements and prominent eosinophilic cuticle (hematoxylin-eosin,  $\times 66$ ).



loci, and that their case may have pachyonychia congenita localized to the nails (16).

Our case is unique in that the patient had classical nail changes associated with multiple cutaneous cysts, but no dyskeratotic cutaneous lesions, or leukokeratosis of the mucous membranes.

Recently, Smith et al. (7) reported heterozygous k17 missense mutations in the same conserved protein motif in five Jackson-Lawlerform (PC-2) (8), pachyonychia is accompanied by multiple pilosebaceous cysts, natal teeth, and hair abnormalities, families, confirming that mutations in this gene are a common cause of PC-2, and also showed heterozygous missense mutations in K17 in two families diagnosed as steatocystoma multiplex. They described that analogous mutations in K17 might lead to milder phenotypes related to PC-2 or steatocystoma multiplex.

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