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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 Affecting Only the Nails

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To the Editor:

Pachyonychia congenita (PC) is a rare heritable disorder first described by Jadassohn and Lewandowsky in 1906 (1). Nail changes that affect all the nails symmetrically are a hallmark of the disease. On the basis of the presence and prevalence of associated symptoms, numerous subdivisions of PC have been suggested. Both autosomal dominant and autosomal recessive forms have been described, reflecting heterogeneity (2). We describe a patient with nail changes in the absence of other syndrome abnormalities, indicating that the coexistence of associated symptoms is not an absolute prerequisite for the diagnosis of PC, as has been reported in the past (3,4).

A 3-month-old boy presented with a history of an enlarged head and nail disfigurement since birth. There was no history of similar nail changes in any other member of the family in two generations and the parents were nonconsanguineous. On examination, most nails had wedge-shaped thickening and discoloration of the nail plate (Fig. 1). No other abnormality was detected on mucocutaneous examination. Potassium hydroxide (KOH) examination and fungal culture were negative. Head circumference was increased, with prominent fontanelles. Computed tomography (CT) scan of the head revealed obstructive hydrocephalus due to aqueductal stenosis. Although there were no associated abnormalities as described with PC, the nail findings were characteristic of this rare disorder.

Abnormalities of the nails are the most consistent clinical findings of this syndrome. A purely clinical classification does not correlate satisfactorily with the observed phenotypic expression of all cases and recent biologic findings also throw doubt on the accuracy of such a clinical classification (5). The diagnostic clinical feature of PC is the presence of thickened wedge-shaped nails, but other clinical characteristics such as palmoplantar hyperkeratosis, localized foot blistering, follicular hyperkeratosis, mucosal leukokeratosis, and natal teeth are present in a variable number of patients (2). Wedge-shaped, thickened nails are the diagnostic clinical feature of PC. The proximal portions are smooth and normally attached to the lateral nail folds. The distal portion may increase to six times the normal thickness, producing a subungual keratinous mass that pushes the nail plate upward, arching it transversely, folding it longitudinally, and elevating it distally.

The nails are commonly shed and regrow with similar but more severe changes. Projections from the nail beds make the nails susceptible to trauma with consequent chronic paronychial infections. The extent of mutation in the highly conserved 1A domain of K6, K16, and K17 is associated with various abnormalities associated with the syndrome. The prominent nail involvement of PC reflects the
extensive expression of K6, K16, and K17 in the nail matrix (5). Isolated nail involvement in the absence of other abnormalities has been reported in the past in two families (3,4). The nail changes present in our patient are characteristic of PC, although associated findings of the syndrome are lacking. Our patient appears to represent a monosymptomatic form of PC. The variable degree of gene mutation involving K6, K16, and K17 or predominantly a particular keratin may result in nail abnormalities alone. Perhaps it may represent a different allele, resulting in a milder phenotype. The presence of congenital hydrocephalus may be another new association of pachyonychia congenita or may just be a chance occurrence.

To cite this article
doi: 10.1046/j.1525-1470.2002.0024c.x

References


