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 tarda

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Pachyonychia congenita is a rare genetic disorder classified in clinical subtypes. Late onset of the disease has recently been described and designated as pachyonychia congenita tarda. A patient in whom typical manifestations of pachyonychia congenita appeared at the age of 39 years is described. This report substantiates previous observations on the phenomenon of late-onset pachyonychia congenita. (J Am Acad Dermatol 1996;35:334-5.)

Pachyonychia congenita is an inheritable ectodermal dysplasia featuring pathognomonic nail abnormalities, including thickening, hardening, and pinched-up deformity of the nail plates, subungual hyperkeratosis, and distal discoloration.[1-4] These changes are usually associated with heterogeneous cutaneous and extracutaneous features.[1,2] As implied by the name, these abnormalities are present at birth or develop shortly thereafter.

A group of patients with a delayed onset of the disorder has recently been described.[5] For this rare subset, the name *pachyonychia congenita tarda* has been proposed. We describe an additional case of pachyonychia congenita tarda.

CASE REPORT

A 41-year-old French woman had nail dystrophy and palmoplantar keratoderma. She first noted progressive bilateral changes in her fingernails and toenails associated with palmoplantar skin thickening at the age of 39. There was no history of similar disorders or consanguineous marriages in her family.

Examination revealed marked thickening and hardening with yellow-gray discoloration of all nail plates. The plates were uplifted by a subungual hyperkeratosis with upward growth of the distal portion (Fig. 1). Hypercurvature on the transverse axis of the nail plates was present, giving a pinched shape to the free edges (Fig. 2). All nails were affected; fingernails were more thickened than toenails. A diffuse fissured hyperkeratosis of both palms and soles was also observed.

Fig. 1. Distal upward growth and grayish discoloration of fingernail.

Fig. 2. Thickening of nail plates, subungual hyperkeratosis, and pinched shape of free edges of right fingernails.

Direct microscopy and cultures of keratinous material from nails and palmar skin were negative. Nail plate biopsy was refused. Keratosis pilaris, palmoplantar blistering, hyperhidrosis, leukokeratosis, alopecia, dental malformation, and epidermoid cysts were absent. Ophthalmologic examination revealed blue sclerae but no corneal abnormalities. The patient was otherwise well except for amenorrhea with anovulation.

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DISCUSSION

On the basis of the clinical features accompanying its characteristic nail dystrophy, pachyonychia congenita has been divided into different subtypes.[1] In 1991 Paller et al.[5] described five patients in whom the disorder started in the second and third decades of life. The existence of a new, genetically distinct subtype of pachyonychia congenita distinguishable by its late onset was suggested, and the term pachyonychia congenita tarda was proposed. Subsequently Iraci et al.[6] described a patient in whom the pachyonychia developed in the third decade of life whereas other signs of the disorder were present since childhood. These authors suggested that pachyonychia congenita with late onset of nail dystrophy represents an additional type of pachyonychia congenita, possibly because of expression of a different allele.

In our patient both nail dystrophy and palmoplantar keratoderma developed when she was 39 years old. The diagnoses of onychogryphosis, psoriasis, reaction to chronic trauma, and subungual onychomycosis were excluded by the typical appearance of the affected nails, symmetric and synchronous onset of the disorder, negative mycologic test findings, and absence of personal history of psoriasis or chronic nail trauma. Although she did not show any other signs of the disorder, we believe our patient represents a sporadic case of pachyonychia congenita tarda.

To our knowledge, this is the latest reported onset of the disorder. It is yet unclear whether variability in the age at onset of pachyonychia congenita directly implies the existence of a distinct entity or a new subtype in its classification.[1]

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