



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.

A family with pachyonychia congenita affecting the nails only

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Summary

A family with pachyonychia congenita in which affected individuals showed nail involvement only is described. Pachyonychia congenita is a rare hereditary disorder inherited in an autosomal dominant manner. Various classifications of pachyonychia congenita have been suggested but none indicates nail involvement as a solitary finding.

Pachyonychia congenita is a rare inherited autosomal dominant cutaneous disorder first described by Jadassohn and Lewandowsky in 1906.¹ Since then, a number of classifications of this condition have been proposed based on the presence or absence of clinical features other than pachyonychia.² A family with nail changes only is described.

Case reports

The pedigree of this affected family is shown in Fig. 1. Mrs L.L. (age 36; 111.2) was referred to one of us (J.V.) by her family doctor in October 1993 with chronic finger-nail infection. She gave a history of congenital thickening of finger- and toe-nails, which grew slowly. Since the age of 18 years, individual finger-nails had been tender at times with underlying debris which became infected, and affected nails were shed periodically. Here general health had always been good and in particular she had had no other problems with her skin, teeth or hair. She did not complain of cold hands. She mentioned that her daughter (IV.1) and her father (II.1) had thickened nails. Her grandfather (I.3) and his siblings (I.1, I.2) had been similarly affected. On examination, onycholysis of finger-nails with chronic paronychia, particularly affecting the left ring finger-nail, was present (Fig. 2). There was no finger-nail thickening but some overcurvature. Toe-nails were short and thickened (ingrowing big toe-nails had been removed surgically at the age of 22 years). Her oral mucosa was normal. X-rays of fingers and toes were normal. She was prescribed a 15-day course of

itraconazole, topical applications and advised to keep finger-nails short. Nail clippings grew no fungus. Inflammation had cleared and thickening of finger-nails was visible in February 1994.

Miss E.L. (age 9 years; IV.1) had normal nails at birth. Recurrent swelling around and under finger-nails followed by shedding of affected nails had begun when she was 1 month old. There had been no other problems with her skin, teeth or hair. On examination in December 1993 she showed thickened wedge-shaped finger-nails with some onycholysis but without inflammation (Fig. 3a and b). There was malalignment of thickened big toe-nails. Her oral mucosa was normal.

Mr E.W. (age 65 years; II.1) had had abnormal nails all his life but had never developed infection. He had not had other problems with his skin, teeth or hair. On examination in March 1994 there was wedge-shaped nail thickening, less marked than that of his granddaughter, and no inflammation. His oral mucosa was normal. He had recently been diagnosed as suffering from carcinoma of the bronchus.

Discussion

In this family with pachyonychia congenita the disorder is confined to the nails. There was no evidence of hair or

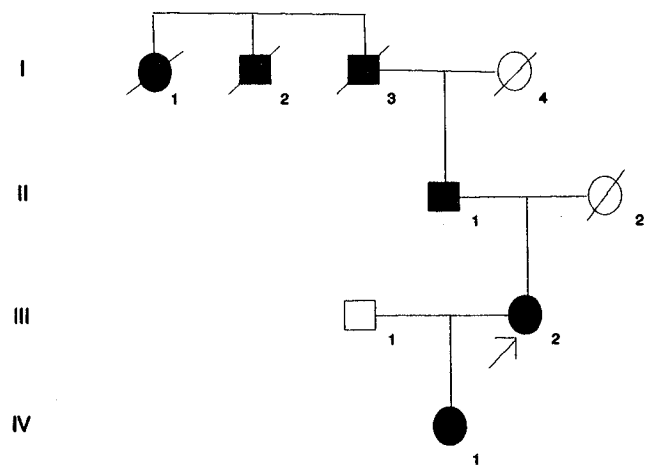


Figure 1. Pedigree of affected family.

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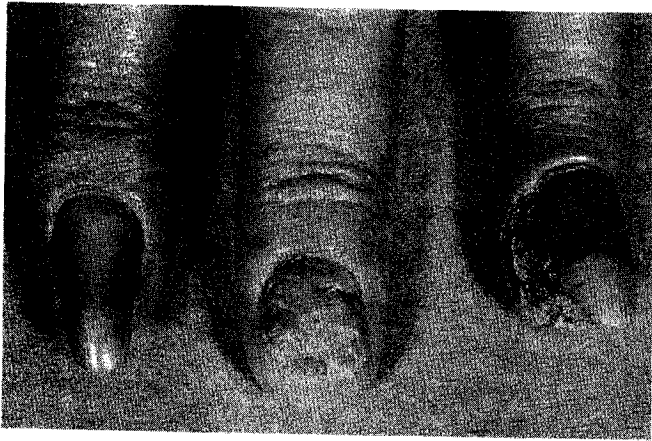


Figure 2. Family member 111.2. Fingers of left hand with posterior nail fold swelling and infected debris around ring finger-nail.

oral abnormalities, hyperkeratosis of palms or soles, blistering, hyperhidrosis, etc.³ There are similarities with a family described by Schulze as 'hereditary partial onycholysis with hard nails'.⁴ However, he excluded pachyonychia congenita in view of absent mucosal changes and he considered the distal increased curvature and partial nail thickening unlike pachyonychia congenita. It seems likely that his family also had pachyonychia congenita localized to the nails.

It is of interest to note in our family that affected individuals showed varying degrees of nail thickening at different times, and different degrees of thickening and tendency to chronic inflammation were present in different affected individuals. It is not known whether the observed interfamilial variation is due to genetic heterogeneity based on different mutations at the same locus or mutations at different genetic loci.

Acknowledgment

We are grateful to Dr Harry Wilson for translating the Schulze paper.

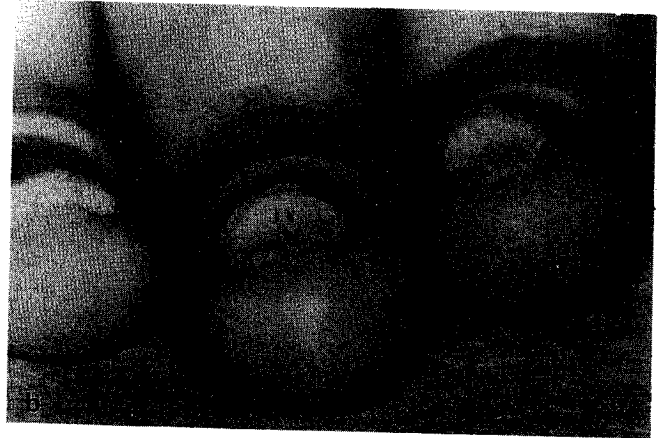


Figure 3. Family member 1V.1. Thickened wedge-shaped left fingernails.

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