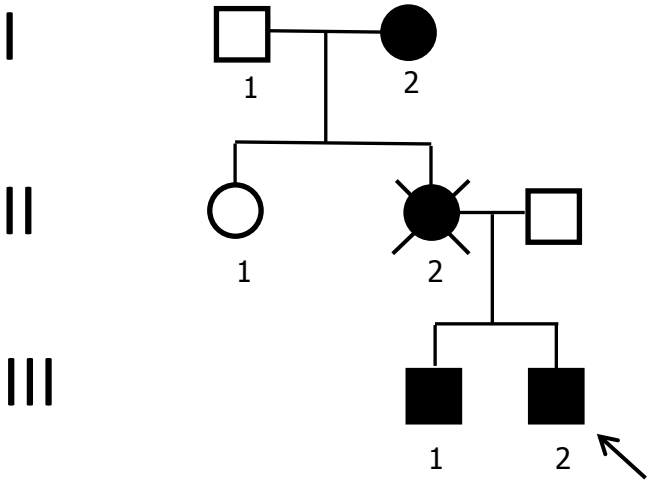
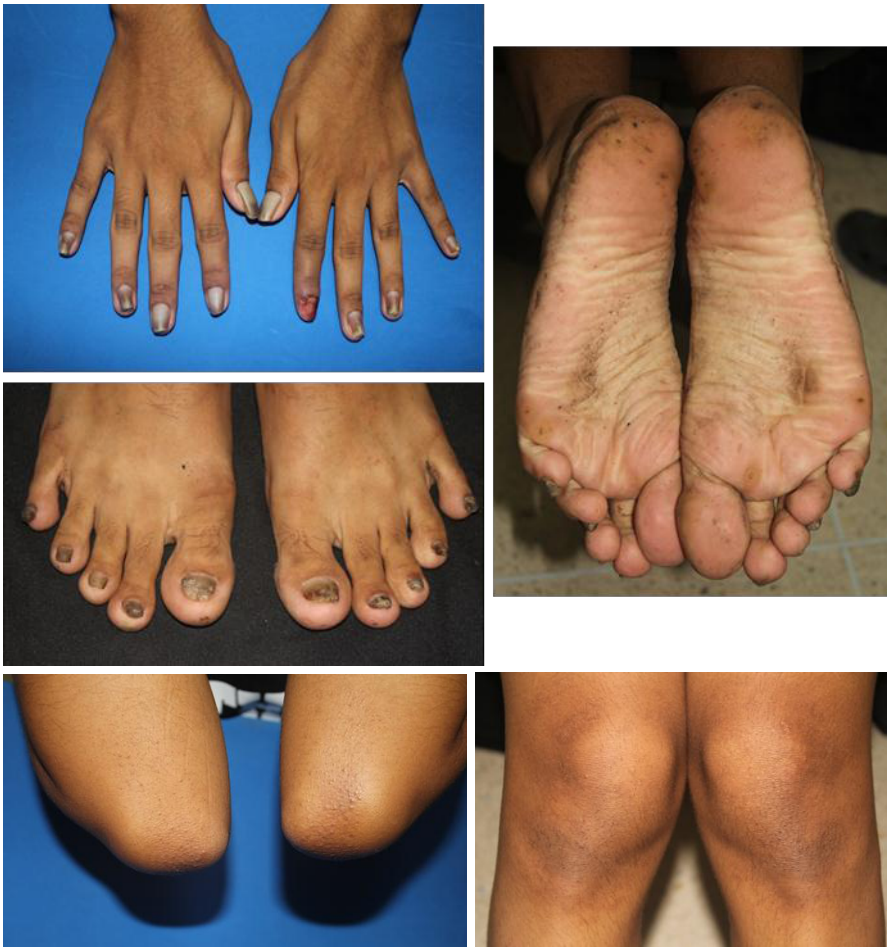
Case 7
A 16-year-old Thai boy from Samut Prakan.
Chief complaint: Nail thickening since childhood memory.
Present illness The patient noticed his nail thickening since childhood. He has increased perspiration on hands and feet. His mother and brother are suffering with the same condition.
Past history: No known underlying disease.

Family tree



Physical examination
Multiple follicular papules at both elbows and knees are noticed.
There is yellowish thicken nail plate with increase transverse nail curvature with distal nail bed hyperkeratosis involving all toes and fingernails.
There is keratotic plaques at weight bearing area of plantar surface both feet.
No bushing eyebrow, no cyst and no teeth abnormality.



Diagnosis: Pachyonychia congenita type l
Treatment:
Acetretin (10) 1 tab OD, 5% salicylic acid apply on nail hs, calcipotriol ointment apply periungual area bid.
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Discussion

Pachyonychia congenita (PC) is an autosomal dominant disorder characterized by hypertrophic nail dystrophy, painful palmoplantar keratoderma, oral leukokeratosis, pilosebaceous cysts (including steatocystoma and vellus hair cysts) and follicular keratoses.

PC-1 (Jadassohn–Lewandowskyis) is typically associated mutations in the keratin-16 gene (KRT16) on chromosome 17 or in the KRT6a gene on chromosome 12. These genes encode for structural proteins expressed by epithelial cells.[1] Clinically it is characterized by nail abnormalities, hyperkeratosis of the palms, soles, knees and elbows, tiny cutaneous horns in many areas, and oral leukokeratosis. Hyperhidrosis of the hands and feet is usually present.[2] Patients with PC-1 who have KRT6a and KRT16 mutations display distinct phenotypic differences. Patients with PC-K6a experience earlier onset, more extensive nail disease and more substantial disease outside palms and soles.[3]

The rarer PC-2 (Jackson–Lawler) is typically associated mutations in KRT17 or KRT6b. The clinical presentations of PC-2 are distinguished by multiple pilosebaceous cysts, teeth present at birth, and hair changes such as protuberant eyebrows. Extensive and infected exural, vulval or scrotal cysts can present as hidradenitis suppurativa.[4]

PC is diagnosed by clinical findings and by molecular genetic testing.

The current treatment modalities primarily center on symptomatic relief, hygienic grooming practices, and treatment of secondary infection when indicated. Painful plantar keratoderma can be reduced by limiting the friction and trauma to the feet, reducing hydration of the stratum corneum by using wicking socks and ventilated footwear, selecting shoes that are comfortable and maintaining an ideal body weight. Foot care includes paring down hyperkeratotic areas and topical therapies for hyperkeratosis

(emollients and lotions containing keratolyics). Care of thickened nails often requires the use of surgical or razor blades or sanders such as a Dremel® tool.[1] None of the currently available therapeutic options are ideal, although they provide some relief, with mechanical/surgical options being preferred over medical therapies. These results emphasize the need for more efficient and targeted therapies.[5]

References

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3. Spaunhurst KM, Hogendorf AM, Smith FJ, Lingala B, Schwartz ME, Cywinska-Bernas A et al. Pachyonychia congenita patients with mutations in KRT6A have more extensive disease compared with patients who have mutations in KRT16. Br J Dermatol. 2012;166:875-8.
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5. Goldberg I, Fruchter D, Meilick A, Schwartz ME , Sprecher E. Best treatment practices for pachyonychia congenita. J Eur Acad Dermatol Venereol. 2013.