

Letter to Editor

## Pachyonychia congenita type 1: Poor response to acitretin

Durga Prasad Dubey, MD., DVL<sup>1</sup>, Swale Iftikhar, MD., DVL<sup>1</sup>, Raj Kumar, MD., DVL<sup>1</sup>

<sup>1</sup>Department of Dermatology, Venereology and Leprology, Baba Raghav Das Medical College, Gorakhpur, Uttar Pradesh, India.

Dear Editor,

Pachyonychia congenita (PC) is a rare form of ectodermal dysplasia marked by keratin abnormalities and characterised by thick and discoloured nails. The primary cause is mutation in genes responsible for encoding specialised keratins such as K6a, K6b, K6c, K16 or K17.<sup>[1]</sup> Clinically, individuals with this condition often present with painful palmoplantar keratoderma, oral leukokeratosis, hypertrophic nail dystrophy, follicular papules and epidermal cysts.<sup>[2]</sup>

A 9-year-old boy presented with a history of progressive thickening and discolouration of nails on both hands and feet, accompanied by painful raised lesions on his palms and soles, persisting for 6 years. He was born to parents who had a consanguineous marriage, with no reported family history of similar symptoms. There were no signs of mental impairment, eye abnormalities, excessive sweating of the palms and soles or previous mucosal lesions. The child experienced significant emotional distress, including shame, embarrassment and low self-esteem, due to being bullied at school for his appearance, which deeply affected both him and his parents.

During examination, we noted hyperkeratotic plaques on the soles of feet, which were tender upon touch [Figure 1a]. Similar thick, hyperkeratotic plaques were observed on both palms, which were also tender upon touch [Figure 1b]. The lesions on the soles caused pain and discomfort while walking. Moreover, all fingernails and toenails exhibited brown discolouration and dystrophy, with evident thickening. Subungual hyperkeratosis resulted in the nails being elevated distally [Figure 1c]. Onychoscopy revealed a pincer-shaped nail plate with compact yellowish-white keratosis adherent to both the underside of the nail plate and the nail bed [Figure 1d]. Foul-smelling, macerated, white, hyperkeratotic plaques were found on the forehead and around the mouth [Figure 1e]. In addition, multiple follicular

hyperpigmented hyperkeratotic papules were observed on the knees, elbows, neck, buttocks and back. No abnormalities were noted in the oral mucosa, and epidermal cysts were absent. Vascular, muscular and neurological examinations revealed no abnormalities.

Potassium hydroxide microscopy examination and culture of the nail clippings did not detect any fungal elements. All routine investigations, including a lipid profile, were within normal range. Due to financial constraints, genetic testing could not be performed. A skin biopsy from a hyperkeratotic lesion on the elbow revealed orthokeratosis and acanthosis [Figure 2].

Based on the clinical features, histopathological findings and the patient's medical history, a diagnosis of type 1 PC was made. The treatment plan involved administering acitretin orally at a nightly dose of 25 mg, along with the use of an emollient containing urea, lactic acid and salicylic acid. After 8 weeks of therapy, significant improvement was noted, including a reduction in the thickness of the palms and soles [Figure 3a and b], considerable pain relief, and a decrease in keratotic lesions throughout the body [Figure 3c]. However, no noticeable change was observed in the nails. After 8 weeks of treatment, the dose of acitretin was gradually tapered to 25 mg on alternate nights for the next 4 weeks and subsequently discontinued. The patient was then maintained on topical therapy alone. Monthly follow-up was conducted for 4 months, during which the clinical improvement was sustained. No side effects were noted during follow-up. The patient was subsequently lost to follow-up. Following the initiation of acitretin therapy, the skin lesions showed marked improvement, boosting the child's and his parents' confidence and emotional well-being.

PC is usually inherited in an autosomal dominant pattern with partial penetrance, though sporadic cases and autosomal recessive inheritance have also been reported.<sup>[2]</sup> Revised

\*Corresponding author: Swale Iftikhar, Department of Dermatology, Venereology and Leprology, B.R.D. Medical College, Gorakhpur, Uttar Pradesh, India. [swale123@gmail.com](mailto:swale123@gmail.com)

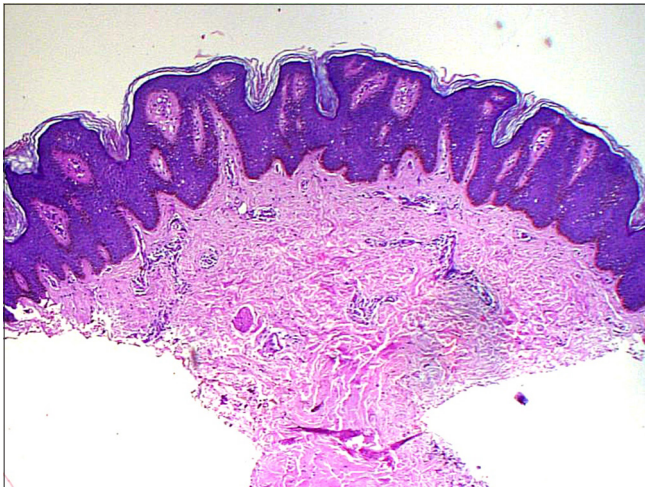
Received: 08 May 2025 Accepted: 15 July 2025 Published: 24 December 2025 DOI: 10.25259/JONS\_12\_2025

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**Figure 1:** (a) Painful plantar keratoderma. (b) Hyperkeratotic papules over the palms. (c) All nails showing thickened, dystrophic nail plates with subungual hyperkeratosis. (d) Onychoscopy revealed a pincer-shaped nail plate with compact yellowish-white keratosis adherent to both the underside of the nail plate and the nail bed (Heine delta 30,  $\times 10$ , polarised mode). (e) Thick hyperkeratotic lesions on the forehead and around the mouth.



**Figure 2:** Skin biopsy revealed orthokeratosis and acanthosis (Hematoxylin and eosin, 10x).

classification of PC proposed in 2011, is based on mutations in five specific keratin genes. The identified subtypes include PC-K6a, PC-K6b, PC-K6c, PC-K16 and PC-K17. In cases where PC is suspected but no mutation is detected, the condition is referred to as PC-U (unknown).<sup>[3]</sup> Emollients and keratolytics, such as salicylic acid, benzoic acid and propylene glycol, may be helpful for mild cases of keratoderma. Mechanical reduction of hyperkeratosis and nails can provide clinical benefit by reducing symptoms and improving comfort and functionality. Acitretin, when



**Figure 3:** (a) Marked improvement in plantar lesions after treatment. (b) Post-treatment improvement in Palmar lesions. (c) Improvement in hyperkeratotic lesions on the forehead and around the mouth after treatment.

administered at 0.5-1 mg/kg daily, may effectively enhance therapeutic efficacy and safety. However, close monitoring

is imperative during the initial 2 months of treatment, with routine checks on triglycerides, fasting serum cholesterol, and liver enzymes required every 4 weeks. Subsequently, monitoring should be conducted every 3 months. It is crucial to remain vigilant about potential long-term side effects of acitretin, such as periosteal hyperostosis and increased dermal sensitivity and fragility. Consequently, the utilisation of acitretin in managing PC may present challenges due to these complexities.<sup>[4]</sup>

In conclusion, this case highlights a rare presentation of PC Type 1 with good response to acitretin in skin lesions, but minimal to no improvement in nails. This highlights the need for effective treatment options for distressing nail thickening.

**Authors' contributions:** DPD: Data collection, draft manuscript preparation. SI: Study conception and design, literature Review, draft manuscript preparation, manuscript revision. RK: Manuscript revision, final manuscript preparation.

**Ethical approval:** Institutional review board approval is not required.

**Declaration of patient consent:** The authors certify that they have obtained all appropriate patient consent.

**Financial support and sponsorship:** Nil.

**Conflicts of interest:** There are no conflicts of interest.

**Use of artificial intelligence (AI)-assisted technology for manuscript preparation:** The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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**How to cite this article:** Dubey DP, Iftikhar S, Kumar R. Pachyonychia congenita type 1: Poor response to acitretin. *J Onychol Nail Surg.* 2025;2:118-20. doi: 10.25259/JONS\_12\_2025