



A Rare Case of Pachyonychia Congenita and the Need for Awareness

✉ Hami Kork, ✉ Gürbüz Yıldırım, ✉ Zeynep Altan Ferhatoğlu

Department of Dermatology, Istanbul University-Cerrahpaşa, Cerrahpaşa Faculty of Medicine, İstanbul, Türkiye

I am writing to share an extraordinary case of pachyonychia congenita (PC) that recently came to our attention. PC, a group of rare keratin disorders inherited in an autosomal dominant manner, is characterized by palmoplantar keratoderma, dystrophic nail changes, and oral leukokeratosis. Distinguishing the five different keratin mutations is crucial as they result in varied symptoms.¹ This letter aims to raise awareness about the challenges faced by individuals with PC and advocate for a greater understanding of this condition within the medical community.

Our 15-year-old female patient exhibited typical nail changes and palmoplantar hyperkeratosis associated with PC. Genetic analysis confirmed the diagnosis, revealing a *KRT6A* gene mutation.² Her case underscores the importance of early detection and genetic analysis in establishing a definitive diagnosis.

PC is classified into five subgroups based on keratin gene mutations.³ Genetic testing confirmed the prevalence of the PK-K6a subtype in our patient's case. The clinical presentation included not only nail changes (Figure 1a-c) but also plantar keratosis (Figure 1d), oral leukokeratosis (Figure 1e), and follicular hyperkeratosis on the knees, elbows (Figure 1f), and face.

The challenges encountered by individuals with PC are multifaceted, encompassing physical discomfort, aesthetic concerns, and potential complications. Currently, no curative treatment is available for PC, and therapeutic options aim to alleviate symptoms. Our patient was recommended various treatments, including keratolytic agents, moisturizers, and pain management strategies.



FIG. 1. (a) Tubular thickening and discoloration of the fingernails, (b) dystrophic fingernails, (c) toenails exhibiting tubular thickening, white discoloration, and a pincer-nail appearance secondary to thickening, (d) heel subjected to compression in the plantar region, with yellow hyperkeratotic plaques following diffuse skin lines in the metatarsal region, (e) oral leukokeratosis, (f) follicular papules on the elbow surface.



Corresponding author: Hami Kork, Department of Dermatology, Istanbul University-Cerrahpaşa, Cerrahpaşa Faculty of Medicine, İstanbul, Türkiye

e-mail: hamikork7@gmail.com

Received: February 14, 2024 **Accepted:** 2024-March 21, 2024 **Available Online Date:** 04.07.2024 • **DOI:** 10.4274/balkanmedj.galenos.2024.2024-2-23

Available at www.balkanmedicaljournal.org

ORCID iDs of the authors: H.K. 0009-0006-2440-7210; G.Y. 0000-0003-4195-1825; Z.A.F. 0000-0003-3090-656X.

Cite this article as: Kork H, Yıldırım G, Altan Ferhatoğlu Z. A Rare Case of Pachyonychia Congenita and the Need for Awareness. *Balkan Med J.*; 2024; 41(4):317-8.

Copyright@Author(s) - Available online at <http://balkanmedicaljournal.org/>

Moreover, addressing the risk of misdiagnosis is crucial, particularly concerning oral leukokeratosis, which is often mistaken for *Candida albicans* infection. Accurate diagnosis and treatment are vital for the well-being of affected individuals, and collaboration with otolaryngology surgery may be necessary in cases involving the larynx.

Furthermore, ongoing research is exploring potential treatments for PC, including mammalian target of rapamycin inhibitors, short interfering RNA, statins, botulinum toxin A, and other pharmacological interventions. These developments underscore the importance of continued research to improve the quality of life for individuals with PC.⁴

In conclusion, I urge the medical community and the public to enhance their understanding of PC. Increased awareness can result in earlier diagnosis, improved management, and a more supportive environment for individuals affected by this rare genodermatosis.

Authorship Contributions: Concept- G.Y., Design- G.Y., Supervision- Z.A.F.; Data Collection or Processing- H.K., G.Y., Z.A.F.; Analysis or Interpretation- H.K., G.Y., Z.A.F.; Literature Search- H.K., G.Y., Z.A.F.; Writing- H.K.

Conflict of Interest: No conflict of interest was declared by the author.

REFERENCES

1. Samuelov L, Sarig O, Adir N, et al. Identification of clinically useful predictive genetic variants in pachyonychia congenita. *Clin Exp Dermatol.* 2021;46:867-873. [\[CrossRef\]](#)
2. Pavlovsky M, Peled A, Sarig O, et al. Coexistence of pachyonychia congenita and hidradenitis suppurativa: more than a coincidence. *Br J Dermatol.* 2022;187:392-400. [\[CrossRef\]](#)
3. Ziemann AG, Coulombe PA. Pathophysiology of pachyonychia congenita-associated palmoplantar keratoderma: new insights into skin epithelial homeostasis and avenues for treatment. *Br J Dermatol.* 2020;182:564-573. [\[CrossRef\]](#)
4. McCarthy RL, De Brito M, O'toole E. Pachyonychia Congenita: Clinical Features and Future Treatments. *Keio J Med.* 2023 Sep 28. doi: 10.2302/kjm.2023-0012-IR. [\[CrossRef\]](#)