

## Adult-Onset Epidermolysis Bullosa Pruriginosa in a 52-Year-Old Filipino Female: A Case Report

Brice Patangan Serquiña<sup>1</sup>, Niña Aala Gabaton<sup>1</sup>, Yu-Chen Lin<sup>2,3</sup>, Han-Tang Wang<sup>2</sup>,  
Chao-Kai Hsu<sup>2</sup>, Bryan Edgar Ko Guevara<sup>1</sup>

1 Department of Dermatology, Southern Philippines Medical Center, Davao City, Philippines

2 Department of Dermatology, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan

3 International Center for Wound Repair and Regeneration (iWRR), National Cheng Kung University, Tainan, Taiwan

**Citation:** Serquiña BP, Gabaton NA, Lin YC, Wang HT, Hsu CK, Guevara BEK. Adult-Onset Epidermolysis Bullosa Pruriginosa in a 52-Year-Old Filipino Female: A Case Report. *Dermatol Pract Concept*. 2023;13(3):e2023158. DOI: <https://doi.org/10.5826/dpc.1303a158>

**Accepted:** January 22, 2023; **Published:** July 2023

**Copyright:** ©2023 Serquiña et al. This is an open-access article distributed under the terms of the Creative Commons Attribution-NonCommercial License (BY-NC-4.0), <https://creativecommons.org/licenses/by-nc/4.0/>, which permits unrestricted noncommercial use, distribution, and reproduction in any medium, provided the original authors and source are credited.

**Funding:** None.

**Competing interests:** None.

**Authorship:** All authors have contributed significantly to this publication.

**Corresponding author:** Bryan Edgar K. Guevara, MD, FPDS, FDSP-PDS, Consultant, Department of Dermatology, Southern Philippines Medical Center, Davao City, Philippines. E-mail: [bry0529@yahoo.com](mailto:bry0529@yahoo.com)

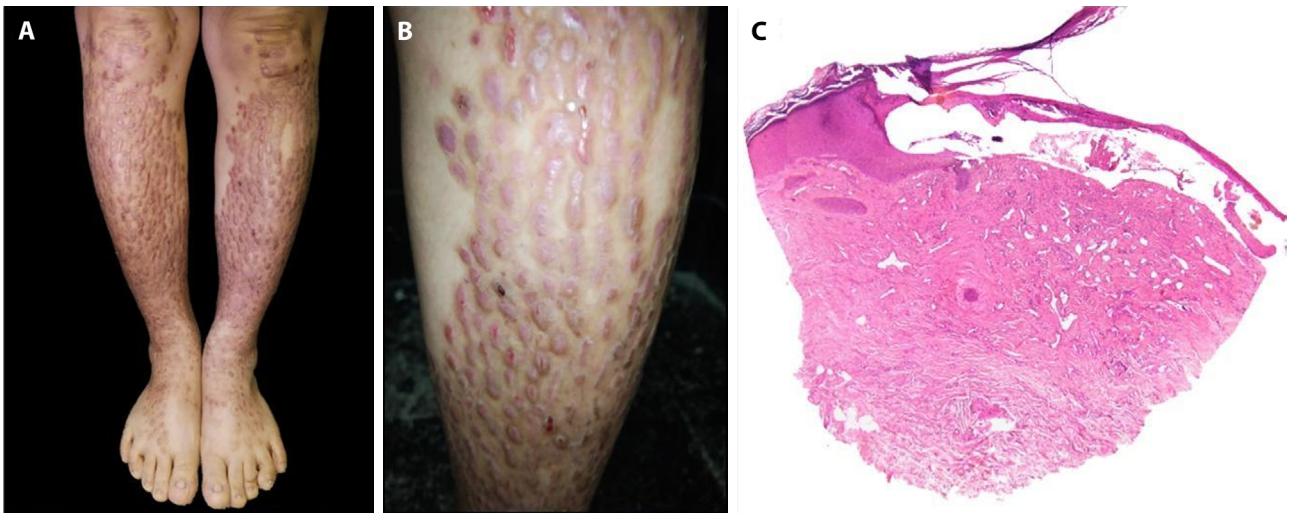
### Case Presentation

A 52-year-old Filipino female presented with a 29-year history of a recurrent pruritic erythematous blisters which evolved into coalescent nodules and plaques on the lower extremities. She was previously treated with topical and intralesional steroid injection with partial resolution. Two of her siblings were affected with similar lesions. Upon physical examination, there were multiple erythematous to hyperpigmented prurigo-like lesions with excoriations on both distal legs, dorsum of feet and forearms. Nail dystrophy was noted on all toes (Figure 1, A and B). Histopathological examination showed a hyperkeratosis overlying an acanthotic epidermis associated with a cell-poor subepidermal blister and fibrotic changes with abundant blood vessels on the superficial dermis (Figure C). Direct immunofluorescence (DIF) results showed negative findings. Genetic testing of her blood samples revealed that the proband was compound

heterozygous for a nonsense and another splice-site mutation in COL7A1. In silico splicing effect prediction of COL7A1: c.7485G>A indicates that COL7A1 protein translation may be affected. These results confirmed the diagnosis of epidermolysis bullosa (EB) pruriginosa.

### Teaching Point

EB pruriginosa manifests as prurigo-like plaques and nodules with pruritus [1,2]. Histopathology findings of a subepidermal blister, negative DIF findings, and a COL7A1 mutation confirms the diagnosis [2]. We present a rare case which can be misdiagnosed as prurigo nodularis. However, the chronicity of the lesions, partial and temporary relief to topical and intralesional steroids, affection of siblings, prompted further investigation. Histopathology, DIF studies and COL7A1 gene mutation confirmed the diagnosis of this rare and challenging case.



**Figure 1.** (A) Multiple erythematous to hyperpigmented prurigo-like lesions with linear excoriations on both distal legs and dorsum of feet associated with nail dystrophy on all toes. (B) A closer view of the prurigo-like linear lesions on the left leg. (C) Histopathology, H&E 4x. Cell-poor subepidermal blister.

## References

1. Pai VV, Sori T, Naveen KN, Athanikar SB, Rai V, Shastry DU. Epidermolysis bullosa pruriginosa: A report of two cases. *Indian Dermatol Online J.* 2014;5(1):44-47. DOI: 10.4103/2229-5178.126030. PMID: 24616855. PMCID: PMC3937487.
2. Nijhawan M, Agarwal ., Nijhawan ., Jhangra R, Goel P, Sehgal VN. Epidermolysis bullosa pruriginosa a Clinico-pathological study in an index case, highlighting its affliction in 11 of 27 member of the family. *Journal of Dermatology & Dermatologic Surgery.* 2017;21(2):100-103. DOI: 10.1016/j.jdds.2017.04.002.