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Journal Of Dermatology and Venereology

Case Report

# Porphyria Cutanea Tarda in A Young Female Patient, Without Comorbidities or Alcoholism: Single Case-Report.

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**Article Info**

**Received:** March 30, 2025
**Accepted:** April 01, 2025
**Published:** April 14, 2025

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**Citation:** Vitória Radichewski, Iago Westphal, Gabriella Di Giunta Funchal, David Cavalcanti Ferreira and Gustavo Moreira Amorim. (2025) "Porphyria Cutanea Tarda in A Young Female Patient, Without Comorbidities or Alcoholism: Single Case-Report." Journal of Dermatology and Venereology, 3(1); DOI: DOI: 10.61148/JDV/015.



## Abstract:

**Background:** Porphyria Cutanea Tarda (PCT) is the most prevalent form of porphyria, resulting from a deficiency in uroporphyrinogen decarboxylase (UROD), leading to porphyrin accumulation and consequent cutaneous lesions, photosensitivity and hepatic issues.
**Case Presentation:** A 35-year-old white female patient, previously healthy, experienced recurrent bullous lesions and the hands, after minimal trauma or sun-exposure. Physical examination revealed tense vesicular lesions on the hands, milium scars. Laboratory findings showed elevated urinary porphyrins and high ferritin levels, while skin biopsy indicated superficial ulceration and perivascular hyalinization with PAS-positive deposits.
**Treatment and Outcome:** Periodic phlebotomies were implemented, resulting in notable clinical improvement.
**Discussion:** This case is atypical due to the patient's young age and the rarity of PCT in women compared to the typical male demographic between their fourth and sixth decades of life.
**Conclusion:** The diagnosis of PCT was challenging, taking six years from symptom onset. This case highlights the importance of considering PCT in differential diagnoses of cutaneous photosensitivity and emphasizes the role of phlebotomy as an effective treatment strategy.

**Keywords:** porphyrias; porphyria cutanea tarda; phlebotomy

## Introduction:

Porphyria Cutanea Tarda (PCT) is the most common form of porphyria, characterized by an enzymatic defect in uroporphyrinogen decarboxylase (UROD).[1] This defect leads to the accumulation of porphyrins, resulting in cutaneous photosensitivity and hepatic manifestations. Some risk factors such as alcohol, estrogens, hepatitis C, and iron overload can precipitate the disease. [1-3] This case report describes the presentation, diagnosis, and treatment of a patient with PCT, highlighting the clinical challenges and therapeutic options.

### History and Clinical Manifestations:

A 35-year-old white female patient from South Brazil, previously healthy, presented with recurrent bullous lesions in the dorsal aspects of her hands, after minimal trauma and after sun exposure. Lesions presented in a chronic remittent course, for at least 6 years. She reported using oral contraceptives and only sporadic alcohol consumption (mainly wine, not more than a couple

of doses per week. She denied a family history of dermatological diseases. On physical examination, blisters and vesicles were observed on the hands, along with many atrophic and millium scars. Very discreet hypertrichosis, identified in the pre auricular region, bilaterally. When asked, she said she needed to shave her upper lip more frequently in recent years.

**Laboratory Tests:**
The suspicion of PCT was confirmed by measuring urinary porphyrins in a 24-hour sample, revealing high levels of uroporphyrins (uroporphyrin I 3204 - VR 4.1 to 22.4 - uroporphyrin II 699 - VR 0.7 to 7.4) and heptacarboxyporphyrins 814.5 - VR 0 to 3.3). Skin biopsy showed superficial ulceration of the skin with keratinocytes in the epidermis, dermal fibrosis and perivascular hyalinization in PAS-positive material, which in immunofluorescence showed deposits of immunoglobulins and complement. Tests for hepatitis B and C were negative, and ferritin levels were high.

**Therapeutics and Results:**
She didn't had any kind of improvement with low-dose hydroxychloroquine. The treatment with periodic phlebotomies was than chosen. The patient showed progressive improvement of the blisters after the first phlebotomy, with complete remission of the lesions and skin friability after the sixth treatment session.

## Discussion/Conclusion:

This report describes a rare case of PCT in a young, white, 35-year-old female patient, which differs from the most common age and gender profile in the literature. PCT generally affects men between the fourth and sixth decades of life, associated with European descent. [5-8] There are three subtypes of PCT: type 1 (sporadic), type 2 (mutations in the UROD gene), and type 3 (positive family history without detectable mutations). 2,3 Without genetic testing, it was not possible to classify the patient's subtype, but type 3 was ruled out due to the absence of a family history.

Risk factors such as smoking, alcoholism, HCV infection, iron accumulation, and use of exogenous estrogens are common in patients with PCT. [2,5] The clinical manifestations are skin lesions in areas exposed to the sun, with vesicles, blisters, skin friability, areas of hyperchromia and hypochromia, and milium. In the case presented, alcohol consumption and use of oral contraceptives were the main risk factors identified, and the patient's symptoms, including lesions on the arms, hands, legs, and feet, and hypertrichosis, are consistent with the literature description.

Porphyrin levels are essential for the diagnosis of PCT, confirmed by determination of urinary porphyrins in a 24-hour sample. Treatment consists of removal of the causative factors and periodic phlebotomies or use of low doses of 4-aminoquinolines.[7]

**Justification for Presentation:**
This case highlights the diagnostic challenges of PCT, a rare condition that took six years to be diagnosed. The presentation aims to discuss the clinical and therapeutic challenges of PCT, contributing to the understanding of the disease and earlier diagnoses.

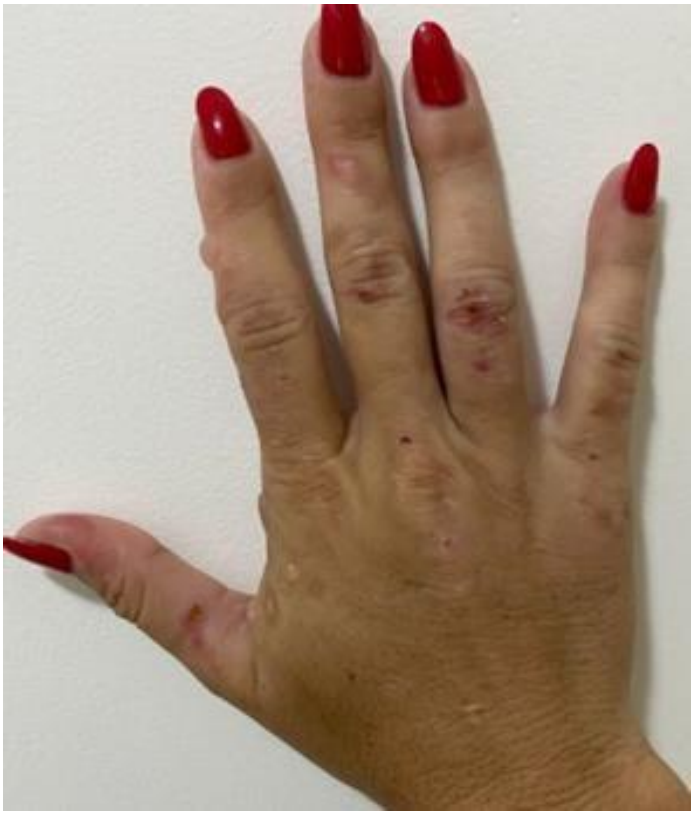

**Figure 1:** Legend: Multiple vesicles on the back of the hands, in addition to atrophic scars from old injuries and some milium scars. Pretreatment.

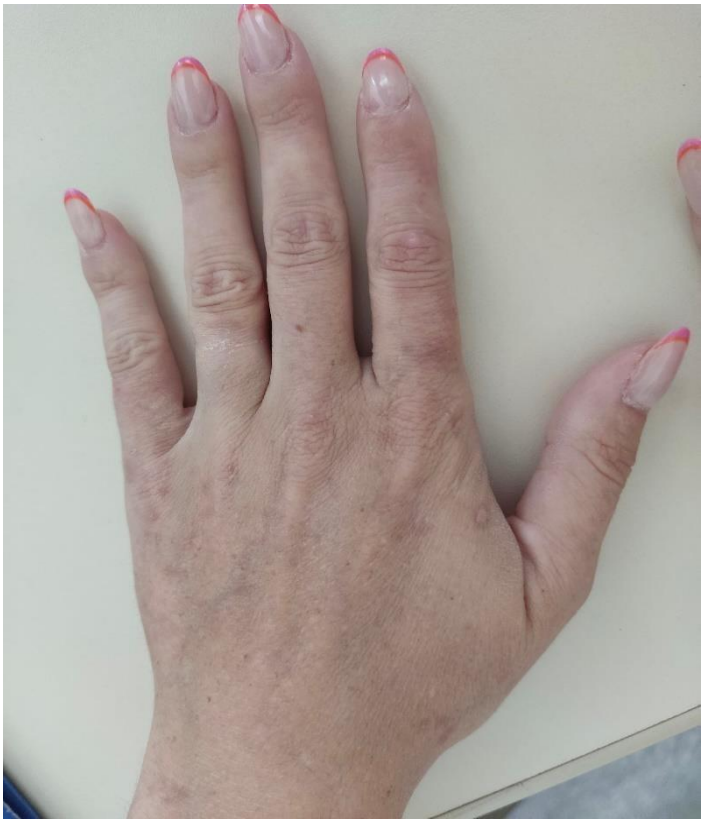

**Figure 2:** Legend: Complete clinical remission after 6 sessions of phlebotomies.

**Data Confidentiality:** The authors declare having followed the protocols in use at their working center regarding patients' data publication.
**Informed Consent:** Obtained.
**Conflicts Of Interest:** All authors report no conflict of interest.
**Financial Support**: No funding. Authors were responsible for the minimal costs involved in this article.

## References:

1. Bissell DM, Anderson KE, Bonkovsky HL. Porphyria. NEJM. 2017; 377(9):862–72.
2. Singal AK. Porphyria cutanea tarda: Recent update. Molecular Genetics and Metabolism. 2019; 128(3):271-281.
3. Stölzel U, Doss MO, Schuppan D. Clinical Guide and Update on Porphyrias. Gastroenterology. 2019;157(2):365-381.
4. Kakoullis L, Louppides S, Papachristodoulou E, Panos G. Porphyrias and photosensitivity: pathophysiology for the clinician. Postgraduate Medicine. 2018;130(8):673–86.
5. Phillips JD. Heme biosynthesis and the porphyrias. Molecular Genetics and Metabolism. 2019;128(3):164–77.
6. Almeida I, Vanzela T, Akabane A, Roselino AM. Porphyria cutanea tarda and associated factors in Brazilian casuistic over the past twenty-four years: What changed? J Am Acad Dermatol. 2017;76(6):AB201.
7. Cabezas Arteaga JE, Vieira FMJ, Silva dos Reis VM. Experience in management of porphyria cutanea tarda in a tertiary referral Brazilian hospital from 2002 to 2017. Int J Dermatol. 2019;58(8):925–32.
8. Succi IB, Pôrto LC, Domingues PMG da RP, Fonseca JCM. Genetic ancestry of patients with porphyria cutanea tarda in a country with mixed races: a cross-sectional study (Rio de Janeiro - Brazil). Anais Brasileiros de Dermatologia. 2018;93(1):148–50.