

Case Report

Porphyria Cutanea Tarda with Addisonian Pigmentation

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Received: 30 July 2025

Accepted: 25 October 2025

Published: 22 January 2026

DOI

10.25259/IJPGD_170_2025

Quick Response Code:



ABSTRACT

Porphyrias are a rare group of metabolic disorders due to hereditary or acquired deficiency of enzymes of the heme biosynthetic pathway. They manifest with neurovisceral and/or cutaneous symptoms depending on the defective enzyme. Porphyria cutanea tarda (PCT) is a common subtype mainly affecting the skin with blisters and easy skin fragility on photo-exposed sites with an estimated prevalence of 1 in 10000 to 1 in 25000. It results from altered activity of the uroporphyrinogen III decarboxylase (UROD) enzyme, causing accumulation of photosensitive carboxylated porphyrins such as uroporphyrinogen in the skin and liver. We report a case of a 46-year-old male farmer presenting with photosensitivity, blistering, and easy skin fragility on photo-exposed and trauma-prone sites for 2 years. He was diagnosed and treated as PCT after clinical and laboratory evaluation.

Key words:- Photosensitivity, Porphyria cutanea tarda, Porphyrin

INTRODUCTION

Porphyria cutanea tarda (PCT) is a metabolic blistering disease with photosensitivity and skin fragility. PCT is divided into three subtypes [Table 1]. Hepatic iron overload is present in nearly all cases of PCT, with elevated plasma iron in 50% cases. PCT typically presents with increased skin fragility, blisters, erosions and crusting on sun-exposed areas and areas prone to repeated trauma. Biallelic mutations in porphyria genes may be associated with Addisonian pigmentation.

CASE REPORT

A 46-year-old male farmer presented with photosensitivity, hyperpigmentation of sun-exposed skin of the face with spontaneous blistering and easy skin fragility on trauma-prone sites for 2 years. He is a known diabetic and was habituated to 30 mL of local rice beer every day for the past 5 years. The patient presented with tense blisters over the sun-exposed areas of face, trunk and limbs, rupturing in a week on manipulation, causing erosions, post-inflammatory hyperpigmented macules, scarring and milia. There were multiple erythematous skin coloured papulo-vesicular lesions on the forehead and upper face [Figure 1a]. Multiple atrophic hyperpigmented scars were seen over the neck and upper back [Figure 1b]. He had a tense bulla of size 2 cm² over the dorsal aspect of the proximal-inter-phalangeal (PIP) joint of the middle finger of the right hand [Figure 1c]. Multiple small hyperpigmented macules were present over the bilateral palms, soles and the lower lip mucosa, resembling Addisonian pigmentation [Figure 1d]. He was hypotensive with no history of intake of iron supplements, smoking or photosensitising drugs. A past history of tuberculosis was ruled out in the patient and his family members.

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Table 1: Subtypes of PCT.

Features	Type 1 PCT	Type 2 PCT	Type 3 PCT
Inheritance	Sporadic	Autosomal dominant	Autosomal dominant
UROD enzyme activity	Decreased activity only in the liver	Decreased in liver, red blood cells and fibroblasts	Normal in erythrocytes
Family history	Negative	positive	Positive
Etiology	Acquired factors precipitate PCT (e.g., alcohol, tobacco, estrogen therapy, hepatitis C virus, human immunodeficiency virus infection, iron overload, exposure to polychlorinated hydrocarbons.	Genetic mutation in <i>UROD</i> gene+environmental triggers	Unknown genetic defect (non- <i>UROD</i> -related suspected)

PCT: Porphyria cutanea tarda, UROD: Uroporphyrinogen III decarboxylase



Figure 1: (a and b) The face and back of patient showing hypopigmented and hyperpigmented macules, scarring and milia formation. (c) A tense bulla of size 2 cm² over the dorsal aspect of the PIP joint of the middle finger of the right hand. (d) Addisonian pigmentation on the palms and soles.

The patient was evaluated for PCT and epidermolysis bullosa acquisita with coexisting Addison's disease. Histopathology from a bulla revealed focal hyperkeratosis, subepidermal bulla, festooning of dermal papilla into the bulla cavity, chronic inflammatory cell infiltrate in the dermis, with periodic acid

schiff (PAS)-positive, diastase-resistant eosinophilic material around the capillary walls [Figure 2a and b].

Direct immunofluorescence (DIF) showed moderate to intense staining of immunoglobulin G (IgG), moderate staining of immunoglobulin M, C3 and fibrin along the superficial dermal vessel walls [Figure 2 c and d].

Ultrasonography (USG) abdomen/pelvis was suggestive of B/L grade 1 medical renal disease with serum creatinine at 2.16 mg/dL. His 24-h urine porphyrin level was 2465.7 µg/24 h. All other investigations were within normal limits.

The patient was evaluated for co-existing Addison's disease, as biallelic mutations in porphyria genes are associated with adrenal insufficiency.^[1] The serum cortisol level was normal with elevated serum vitamin B12 and ferritin (2022 ng/mL) and low serum iron (43.37 µg/dL). Addison's disease was ruled out by the endocrinologist.

The patient was diagnosed as PCT based on his clinical features, histopathology, DIF and 24-h urine porphyrin levels. He was advised to avoid direct sun exposure and was started on tablet hydroxychloroquine 200 mg, twice/week. The patient is on follow-up to rule out the development of Addison's in the future.

DISCUSSION

PCT is a metabolic disorder due to hereditary or acquired deficiency of enzyme of the heme biosynthetic pathway. It manifests with visceral and cutaneous symptoms due to elevated tissue and plasma iron. Iron inhibits uroporphyrinogen III decarboxylase (*UROD*) through direct suppression or indirectly as an essential cofactor in generating the *UROD* inhibitor uroporphomethene.^[2] PCT typically presents with increased skin fragility, blisters, erosions and crusting on sun-exposed areas and areas prone to repeated trauma. Dark urine, non-virilising hypertrichosis, scarring alopecia, pigmented scars and milia are seen.^[3] Biallelic mutations in porphyria genes protoporphyrinogen

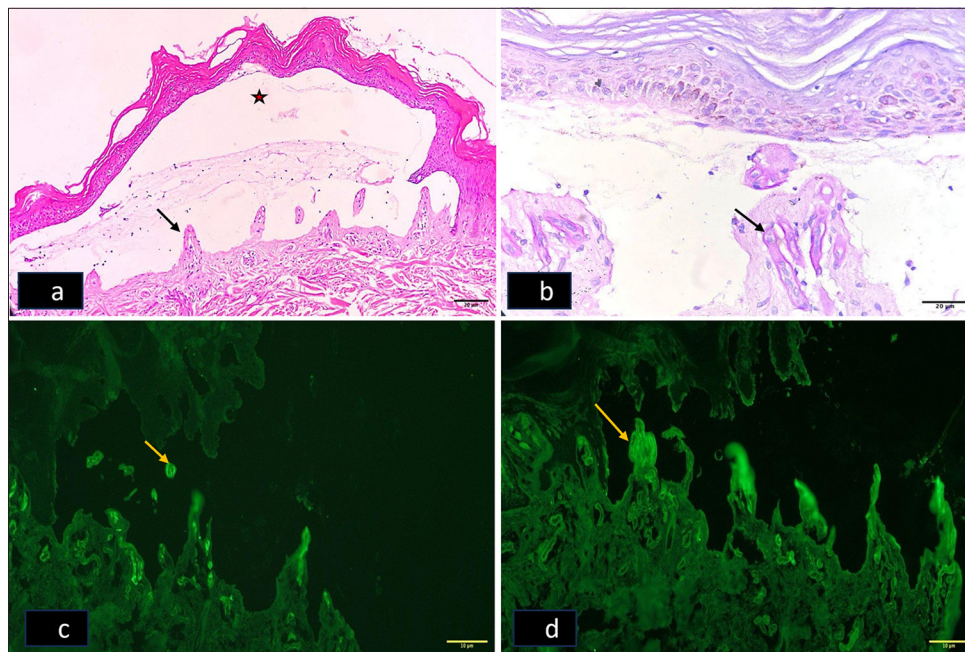


Figure 2: (a) Hyperkeratosis and subepidermal bulla (red star), festooning of dermal papilla (black arrow) into the bulla cavity, chronic inflammatory cell infiltrate in the dermis (PAS, 40x), (b) PAS-positive, diastase-resistant eosinophilic material around the capillary walls in the papillae, upper dermis and around blood vessels (black arrow) (PAS, 40x), (c and d) Moderate to intense staining of IgG, C3 along superficial dermal vessel walls (yellow arrows) (Immunofluorescence, 40x).

oxidase (PPOX), coproporphyrinogen oxidase (CPOX) may be associated with adrenal insufficiency. Reduced PPOX/CPOX activity may lead to adrenal insufficiency through three mechanisms: Inadequate haem for steroidogenic cytochrome P450 (CYP450) enzyme function, accumulation of toxic porphyrin intermediates or increased oxidative stress.^[1] Addisonian pigmentation could rarely be secondary to PCT. Characteristic histopathological findings of PCT include subepidermal blisters without inflammatory infiltrate, with preservation of dermal papillae in the lesion's floor ('festooning'). DIF shows fibrinogen, complement and immunoglobulins, particularly IgG, surrounding blood vessels in the papillary dermis and at the dermal-epidermal junction.^[4] Management of PCT includes elimination of underlying risk factors, photoprotection and specific treatment such as low-dose oral antimalarials and venesection. Low-dose antimalarial medications (hydroxychloroquine 100 mg or 200 mg twice weekly or chloroquine 125 or 250 mg twice weekly) increase urinary excretion of uroporphyrin by changing the pH of the lysosomal compartment due to the action of quinol, allowing the accumulated porphyrins to be transported out of the cell into the plasma and cleared by the kidney. Regular venesection alters iron homeostasis and creates iron depletion within hepatocytes in patients in whom antimalarial drugs are contraindicated and for those who have hemochromatosis. Erythropoietin can mobilise iron from the liver in patients developing PCT due to renal failure.^[5]

This case is reported due to the rarity of PCT and associated Addisonian pigmentation.

CONCLUSION

In this article, we report a classical case of PCT in an elderly male, along with addisonian pigmentation. This addisonian pigmentation is a rare association, which may be due to bi-allelic mutation in PPOX/CPOX gene, resulting in adrenal insufficiency and pigmentation.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship: Nil.

Conflicts of interest: Dr. Bhabani Singh is on the Editorial Board of the journal. **EQI**

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: Nidhi N, Khan AS, Samal K, Sahu K, Pattajoshi AS, Singh BS. Porphyria Cutanea Tarda with Addisonian Pigmentation. *Indian J Postgrad Dermatol.* 2026;4:96-9. doi: 10.25259/IJPGD_170_2025