

## Case Report

### A 34-Year-Old Man with Porphyria Cutanea Tarda

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Rec date: Feb 22, 2014 Acc date: Jun 10, 2014 Pub date: Jun 13, 2014

#### Abstract

**Introduction:** Porphyria cutanea tarda is the most common form of porphyria. This hereditary or acquired metabolic disorder is due to decreased activity of the uroporphyrinogen decarboxylase enzyme. Typical features this scarcely reported entity are photosensitivity, skin fragility to minor trauma, bullae formation, and hyperpigmentation, predominantly on the dorsa of the hands and the arms. **Clinical case:** A 34-year-old Brazilian man presented with erosions and small bullae that had appeared on the dorsum of his hands during a 3-week period. The hypothesis of porphyria cutanea tarda was considered with base on typical lesions of photodermatitis associated with consumption of ethanol, and cell-poor subepidermal bullae with festooning of the dermal papillae. The diagnosis was further established by the classical data of the urinary porphyrins. With use of photo protectors and abstinence from alcohol, the patient remains without clinical manifestations. **Conclusion:** The diagnosis of porphyria cutanea tarda is based in clinical manifestations and can be supplemented by laboratory tests. Skin biopsy contributes to discard differential hypotheses. Detection of rare entities in primary care services often requires high degree of clinical suspicion.

#### Key words:

Hyperpigmentation; Photodermatitis; Porphyria; Porphyria cutanea tarda

#### Introduction

The eight major porphyrias can be briefly distributed in three groups: acute hepatic porphyrias (four), hepatic cutaneous porphyria PCT (one), and erythropoietic cutaneous porphyrias (three) [1]. Porphyria cutanea tarda (PCT) is the most common form of this group of metabolic disorders, and involves a partial deficiency of the enzyme uroporphyrinogen III decarboxylase (UROD) [1-6]. The gene described as UROD, RP11-69J16.2, LOC7389, OTTHUMP00000010503, and fifth enzyme of the heme biosynthetic pathway, encodes protein number 4.1.1.37. PCT is classified as: type I (OMIM 176090) or sporadic type, with near 50% level of UROD in the

liver; and type II or familial type that is an autosomal dominant disorder with low penetrance and constitutes about 20% of cases. The type II is characterized by 50% deficient activity of UROD in many tissues. PCT has variable clinical manifestations and is considered a photodermatitis characterized by recurrent vesicles and bullous lesions in areas exposed to sunlight [1-6]. The identification and elimination of the causal factors may improve the clinical and liver changes, but phlebotomy, chloroquine, or iron chelation can be necessary to the effective control of the symptoms [1-5]. The objective of this case study is to report a typical case of PCT, which is a relatively uncommon dermatological condition, and may be successfully managed by adequate preventive measures. Authors believe that case studies increase the suspicion index about PCT in primary health care.

#### Case report

A 34 year-old-man sought our service complaining of hand injuries since three weeks before. He had vesicobullous lesions, some with scabs, on the back of the hands (Figure 1), but remained asymptomatic. He was an alcohol consumer, and denied smoking or any comorbidity. There were neither neurological features, nor occurrence of similar cases amongst members of his family. Blood tests showed gamma glutamyl transferase 170 (normal: 9-85) UI/l, ALT 90 (normal: 30-40) UI/l, AST 95 (normal: 30-40) UI/l, iron 210 (normal: 30-160) mcg/dl, ferritin 400 (normal: 30-300) mcg/l, transferrin 3.2 (normal: 0.57-2.8) ng/l. Worthy of mention, the urine samples were red to brown in natural light, and revealed a pink-to-orange fluorescence with the Wood's lamp. The tests for excessive porphyrinuria were positive - marked elevation of uroporphyrin (1,200 mcg/24h), the uroporphyrin/ coproporphyrin relationship higher than 3:1, and elevation of heptacarboxilate; but porphobilinogen (0.75 mg/24 h) and ALA (3.5 mg/24 h) were normal [5]. Studies of the biopsy specimens disclosed subepidermal bullous dermatitis associated with sparse lymphocytic infiltrates; the dermal papillae were extending irregularly from the floor of the bulla into the bulla cavity. Therefore, the diagnosis of PCT was established. Abstinence from alcohol and photoprotection was the first option, and resulted in improvement of the active skin lesions. After the initial management, he was referred to longstanding surveillance at Dermatology Division, in order to perform further laboratorial tests and genetic complementary investigations.

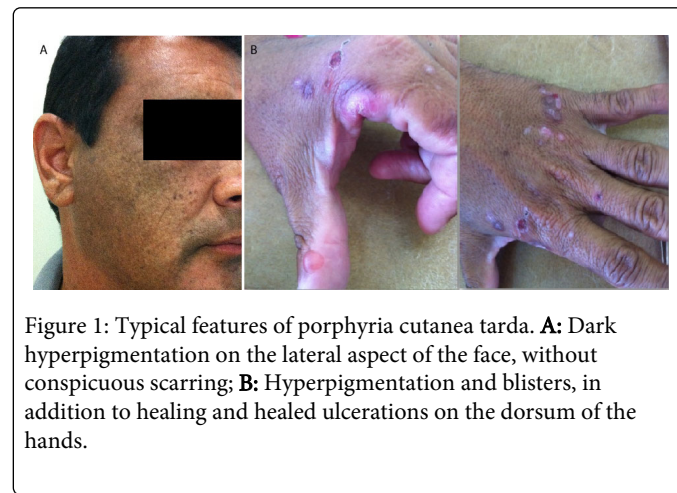


Figure 1: Typical features of porphyria cutanea tarda. **A:** Dark hyperpigmentation on the lateral aspect of the face, without conspicuous scarring; **B:** Hyperpigmentation and blisters, in addition to healing and healed ulcerations on the dorsum of the hands.

## Discussion

This young man had vesicobullous asymptomatic lesions on the dorsum of the hands during three weeks, associated with changes in the gamma glutamyl transferase, iron and transferrin levels. The Wood's lamp test was positive and biopsy data were consistent with porphyria cutanea tarda. With photo protectors and alcohol withdrawal, there was a rapid improvement of the skin lesions. PCT is an uncommon metabolic disorder, which may be either inherited or acquired, and results from a partial deficiency of the activity of UROD [1-6]; this disturbance leads to hepatic accumulation of uroporphyrin and hepta-carboxyl porphyrinogen. PCT is the most common form of porphyria, mainly among men, and is classified into subtypes. The sporadic, begins in adulthood, is restricted to the liver, and is triggered by alcohol intake, estrogens, iron, and other drugs use, hepatitis C, HIV infection, and hemodialysis. The familial type is an autosomal dominant disorder with decreased activity of the enzyme in diverse tissues, in addition to the liver; and the type III, which is hereditary but without the UROD mutation found in the type II [1-6]. Similar to sporadic type, the onset of familial PCT is observed in the fourth decade of life, and requires precipitating factors because the clinical features develop if hepatic UROD is inhibited. People with PCT frequently present increased levels of serum iron and of ferritin, and hereditary hemochromatosis can play a role in this disease [5,6]. Moreover, advanced investigations have indicated that PCT may be associated with C282Y and H63D mutations in the HFE gene [6]. Clinical characteristics of PCT are vesicles and bullae that evolve into erosions and crusts, frequently distributed in body areas that are exposed to UV radiation and to superficial trauma [1-6]. The basis of diagnosis is clinical, supplemented by tests for porphyrins in urine, blood and stool, and findings of the skin biopsy [1-6]. Differential diagnosis of PCT includes bullous lupus erythematosus, epidermolysis bullosa acquisita, hydroa vacciniforme, photoinduced bullous drug reactions, polymorphic light eruptions, pseudoporphyria, scleroderma, and solar urticaria [4]. The treatment requires identification of precipitating factors, and utilization of photoprotector and repeated phlebotomy can be alternative options, as well as chloroquine schedule at low dosage [1-6]. The precipitating factor of the PCT flare up herein

described was the alcohol consumption, which reduces enzymatic activities involved in heme metabolism, as aminolevulinic acid dehydratase, coproporphyrinogen oxidase, ferrochelatase, and uroporphyrinogen decarboxylase [1,4]. Moreover, ethanol and its metabolites can induce the enzymes ALAS1 and CYP2E1, generate reactive oxygen species, cause mitochondrial injury, reduce antioxidant defenses, activate liver inflammation, enhance the endotoxin production, and increase the amount of iron in hepatocytes [2]. As a whole, these phenomena lead to an accumulation of photoreactive porphyrin compounds, which propitiates the occurrence of repeated episodes of skin lesions in the photo-exposed areas. Without the necessary clinical suspicion, recognizing PCT can constitute a challenging task [4]; therefore, case reports might increase the suspicion index of the primary health care workers.

## References

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