A woman with hypertrichosis

Introduction

Porphyria cutanea tarda is the most common subtype of porphyria, which often presents with skin manifestations later in life. The disorder results from low levels of the enzyme responsible for haem production. Although blisters are the most common skin manifestations of porphyria cutanea tarda, other skin manifestations like hyperpigmentation and hypertrichosis also occur (Chan and Lin, 2011). Some patients' clinical features can be a guide to diagnosis. The authors describe a case of porphyria and the management options.

Discussion

Porphyria cutanea tarda is the most common human porphyria, and occurs as a result of decreased activity of the uroporphyrinogen decarboxylase enzyme (Chan and Lin, 2011). The prevalence of symptomatic disease has been estimated at approximately 1 in 25 000 in the United States where 80% of cases are acquired and 20% are familial (Phillips et al, 2007). It has been associated with excess alcohol consumption, oestrogen use, hepatic siderosis, hepatitis C infection, HIV infection and smoking (Phillips et al, 2007).

Porphyrins accumulate in the liver and disseminate in the plasma to other organs including the skin where photoexcited porphyrins cause oxidative damage to biomolecular targets. As a result of increased mechanical fragility after sunlight exposure, erosions and blisters form painful indolent sores that heal with milia, dyspigmentation and scarring. This may progress to 'pseudoscleroderma', with contraction and calcification, resembling cutaneous findings in systemic scleroderma (Grossman et al, 1979).

Other manifestations include hypertrichosis (especially on the cheeks and forearms), scleroderma-like plaques that may develop dystrophic calcification, and excretion of discoloured urine that resembles port wine or tea, which is caused by the presence of porphyrin pigments (Grossman et al, 1979). Liver involvement is usually manifested by increased liver enzymes but there is a long-term increased risk of cirrhosis and hepatocellular carcinoma (Linet et al, 1999).

Treatment consists of repeated phlebotomy, whether or not there is evidence of iron overload, and low-dose hydroxychloroquine if phlebotomies cannot be tolerated (Thunell and Harper, 2000). Remission can be achieved within a few months, but until then patients should wear long-sleeved clothes to avoid exposure to sunlight. BJHM

Figure 1. Facial hypertrichosis and scleroderma-like scarring.

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LEARNING POINTS

- Porphyria cutanea tarda is the most common human porphyria.
- Porphyrins accumulate in the liver and disseminate in the plasma to other organs including the skin.
- Porphyria cutanea tarda causes mechanical fragility after sunlight exposure, erosions and blisters, hypertrichosis, scleroderma-like plaques and excretion of tea-coloured urine.
- Treatment involves repeated phlebotomy.
- Until remission is achieved, patients should avoid exposure to sunlight.

Case Report

A 19-year-old woman presented to the emergency department with a 1-year history of intermittent darkening of her urine. She also gave a history of blistering on her fingers and knuckles. The familial history was insignificant and she had not taken any medication including contraceptive pills.

On physical examination, she looked older than her age with hypertrichosis about the cheeks and temples, eyebrows, ears and arms. She had linear depressed scars on her face (particularly around the mouth) (*Figure 1*). Laboratory tests showed an increase in aspartate aminotransferase, alanine aminotransferase and ferritin with normal creatinine and complete blood count. Tests for hepatitis C and HIV were negative. A fresh sample of her urine was exposed to ultraviolet light (wavelength 400 nm) and an orange-red fluorescence was seen. Subsequent investigation showed very high levels of uroporphyrins in her urine. The diagnosis of porphyria cutanea tarda was made. Subsequently, she underwent phlebotomy.