A tale of nonhormonal hairs

Sir,

Porhphyria cutanea tarda (PCT) is a hepatic porphyria in which the activity of the heme synthetic enzyme uroporphyrinogen decarboxylase is deficient. It may be sporadic (80%) or familial (20%). Hypertrichosis can occur in PCT without the classical skin manifestations of blistering and thickening of the skin, making the diagnosis difficult.[1]

A 35-year-old lady presented with history of excessive growth of hair over the face and the hands for the past 10 years [Figure 1]. She was evaluated multiple times for the hormonal status and which was always normal. On detailed history, she gave history of itching and burning sensation of sun-exposed areas on exposure to sunlight. Physical examination showed thick terminal hair over the face and the forearms [Figure 2] and below the knee [Figure 3]. Hairs over the chest, abdomen, lower back, and pubic area were normal. She also had thickening of the skin over the fingers, terminal onycholysis, and absorption of the digits [Figures 4 and 5]. Based on the clinical history and physical findings, the diagnosis of porphyria was thought of and under ultraviolet light, acidified urine showed coral pink fluorescence of uroporphyrins. In view of the age of onset, absence of family history, and elevated uroporphyrins, a final diagnosis of PCT type 1 was made. The patient was managed with therapeutic phlebotomies and low-dose hydroxychloroquine and had a 75% improvement in symptoms after 1 year.

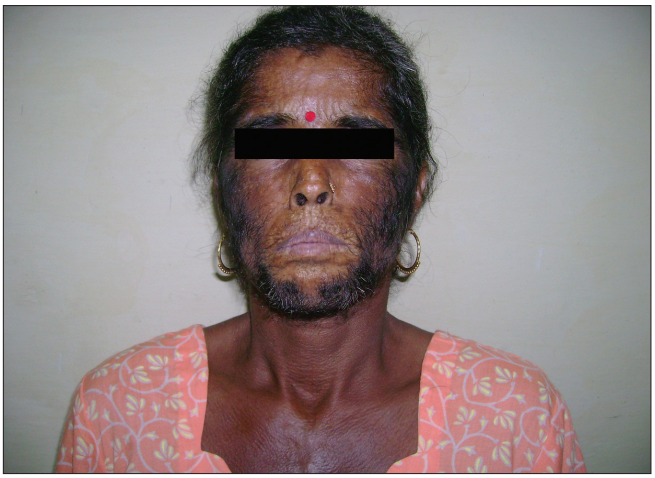

Figure 1: Hypertrichosis

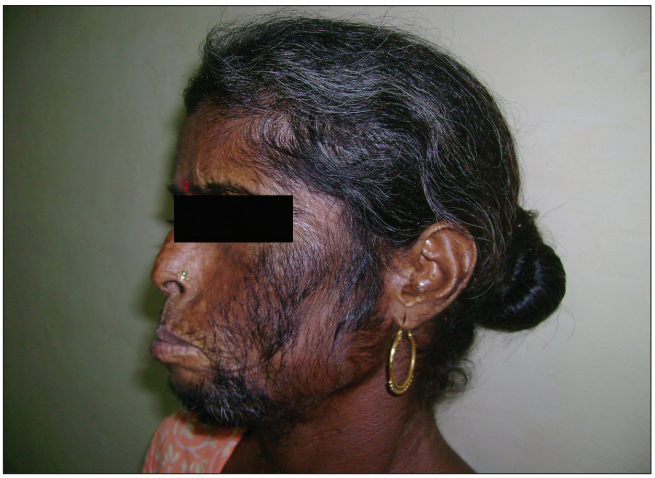

Figure 2: Hypertrichosis over sun-exposed area

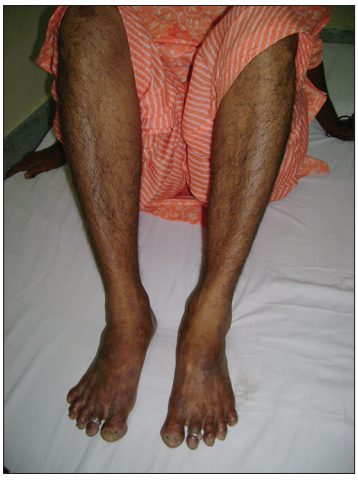

Figure 3: Hypertrichosis over the leg

Porphyrias are due to altered activity of specific enzymes of the heme biosynthetic pathway. Out of the porphyrias, X-linked protoporphyria, congenital erythropoietic

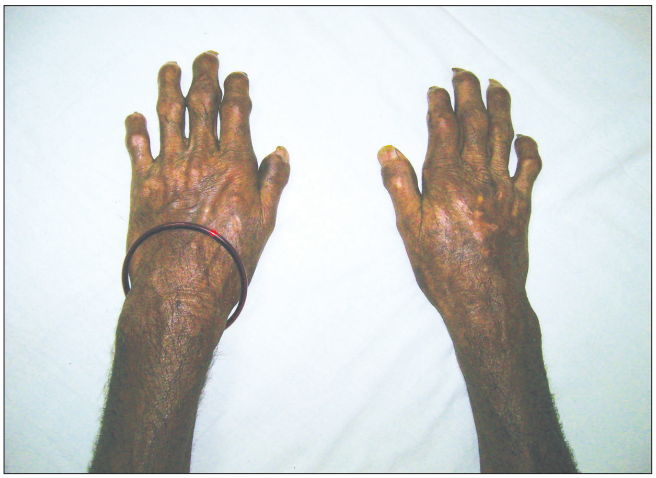

Figure 4: Blisters over the skin, reabsorption of fingers, and pseudoscleroderma

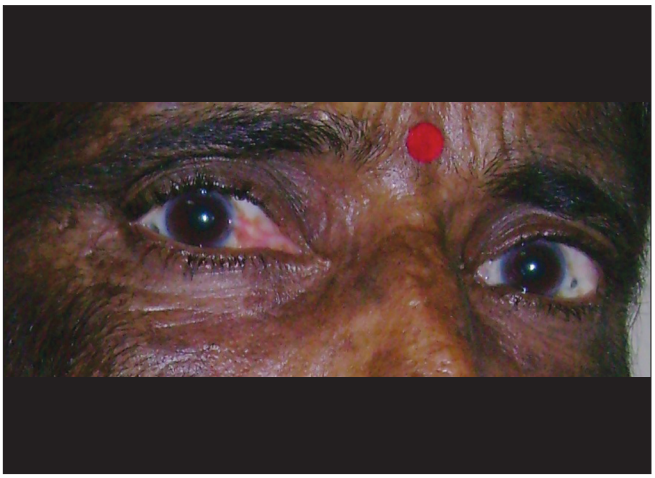

Figure 5: Cicatricial conjunctivitis

porphyria, PCT, hepatoerythropoietic porphyria, hereditary co-proporphyria, variegate porphyria (VP), and erythropoietic protoporphyria have skin manifestations, which is due to the accumulation of photoactive porphyrins in the skin.

Cutaneous photosensitivity with vesicles and bullae are common. Thickening, scarring, and calcification of skin and reabsorption of terminal parts of the digits happens, which resembles scleroderma—the pseudoscleroderma appearance.[1,2]

Excessive hair growth in porphyrias predominantly occurs over the sun-exposed areas sparing the other androgen-dependent areas, offering a diagnostic clue. The hypertrichosis can sometimes be so extreme that the affected patients are called "monkey's children."[3]

PCT is the most common porphyrias and presents usually after puberty with skin lesions.

The precipitating factors for this disease includes alcohol abuse, smoking, estrogen, hepatitis C, and hemochromatosis gene *(HFE)* mutations.[1]

Diagnosis: Plasma porphyrins are increased in patients with porphyrias causing blistering skin lesions. The fluorescence spectrum of plasma and urine can distinguish, erythropoietic protoporphyria, and PCT.[1] A predominance of uroporphyrin and heptacarboxyl porphyrin in urine, producing coral pink fluorescence is diagnostic of PCT.

Treatment: Phlebotomies to reduce ferritin levels is the treatment of choice.[4] Deferoxamine, an iron chelator,[5] or low-dose hydroxychloroquine (100 mg) or chloroquine (125 mg) twice weekly is usually effective when repeated phlebotomies are contraindicated.[1]

PCT should be considered as a diagnostic possibility in patients undergoing evaluation for excessive hair growth. The typical location of hypertrichosis is the sun-exposed areas, sparing other androgen-dependent areas, other features, such as photosensitivity, skin blisters, cola-colored urine, and examination of urine under ultra violet light helps to differentiate this condition from other endocrine causes of excessive hair growth.

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