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Porphyria Cutanea Tarda Masquerading as Systemic Sclerosis: Two Cases Demonstrating an Important Clinical Observation

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Porphyria cutanea tarda (PCT) is a heme biosynthesis disorder characterized by photosensitive bullae and skin fragility[1]. Sclerodermoid changes and dystrophic calcification are uncommon, occurring in 18% and 8% of patients, respectively[1]. Scarring alopecia in PCT, or alopecia porphyrinica, is rarely described, with 5 reports to date[2,3,4,5,6,7].

A 75-year-old man with longstanding facial and scalp tightening and scalp biopsy demonstrating cutaneous sclerosis overlying dystrophic calcification was previously diagnosed with systemic sclerosis (SSc). Antinuclear antibody was positive (1:1280), while anticentromere, Scl-70, and RNA-polymerase-III were negative. Examination revealed facial and scalp sclerosis with ectropion (Figure 1) and 2 scalp ulcers with rock-hard calcinotic nodules. There was no edema or sclerosis of the hands/feet, or nailfold changes. He denied Raynaud phenomenon (RP). PCT was suspected and 24-h urine porphyrins revealed uroporphyrin 427 nmol/24 h (normal < 30), confirming this diagnosis.

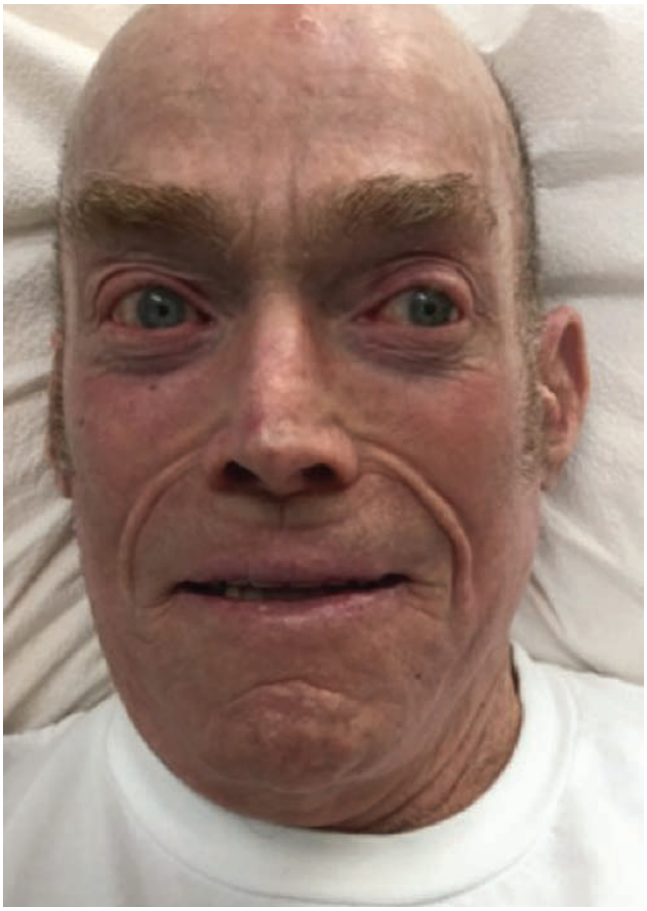

Figure 1. A man in his 70s with facial and scalp sclerosis as well as prominent ectropion as presenting signs of porphyria cutanea tarda.

An 80-year-old female with a history of SSc was referred for alopecia, with scalp biopsy resembling morphea or SSc.

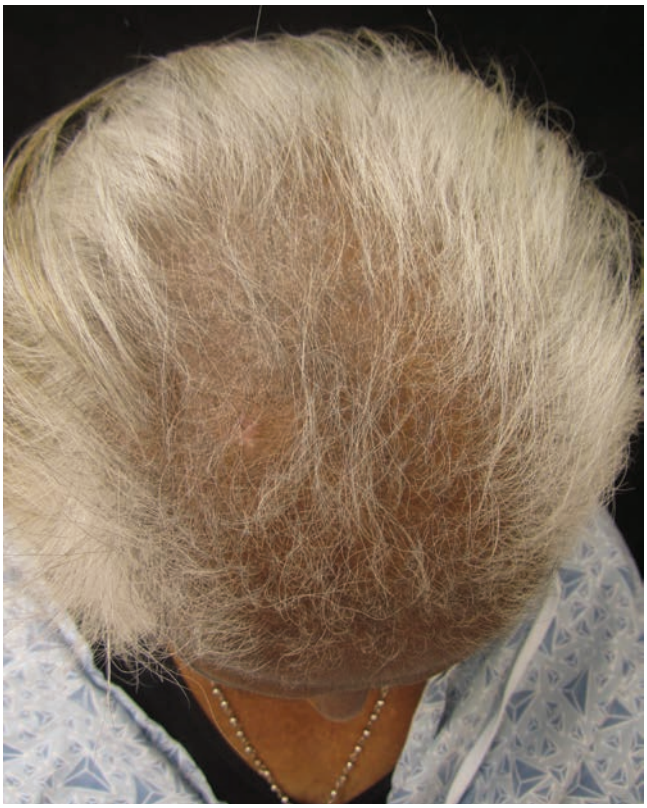

Figure 2. A woman in her 80s with scarring alopecia of the vertex scalp as a presenting sign of porphyria cutanea tarda.

She reported progressive hyperpigmentation, denying photosensitivity, blistering, extremity swelling/tightening, and RP. Examination revealed scarring alopecia (Figure 2), malar hypertrichosis, and hyperpigmentation of the face, trunk, and extremities. Elevated urine (879.5 mcg/24 h; normal < 22) and plasma (24.9 mcg/dl; normal < 1) uroporphyrins confirmed PCT.

Atypical sclerodermatous features, including alopecia porphyrinica, may suggest PCT, even in the absence of photosensitivity or blistering. To our knowledge, our cases are the first ones reported masquerading as SSc; PCT must be considered in the differential diagnosis of sclerotic skin disease. Because biopsy does not distinguish PCT from SSc and morphea, investigation with porphyrin levels is imperative for diagnosis.

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