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Porphyria Cutanea Tarda Due to Primary Hemochromatosis

To the Editor:

A 56-year-old woman with no history of comorbidities or medication intake presented with a 1-year history of blisters and erosions affecting the dorsum of the hands, forearms, cheeks, and forehead. She complained of pain and burning at those sites, and lesions were refractory to potent topical corticosteroids and antibiotics. On physical examination, she had a localized eruption consisting of erosions, blisters, and milia on the dorsum of hands (Figure 1A). On the face, she presented with erosions and mild hypertrichosis. Porphyria cutanea tarda was suspected and laboratory work including urine porphyrins, complete blood count, iron kinetics, liver function test, HIV, and hepatitis B, C serology were assessed. Results from urine porphyrins showed elevated concentration of uroporphyrin, coproporphyrin I, coproporphyrin III, heptacarboxylic, pentacarboxylic, and hexacarboxylic porphyrins. Serum ferritin and liver function test were abnormal. Genetic test for the C282Y mutation for the hemochromatosis gene was positive. Primary hemochromatosis presented by porphyria cutanea tarda diagnosis was made, and the patient was successfully treated using phlebotomy (Figure 1B).

Hemochromatosis is one of the most frequent autosomal recessive hereditary diseases in European Caucasians with a prevalence of 1:200 to 1:500.[1] Porphyrias are clinically and genetically heterogeneous metabolic diseases arising from predominantly inherited catalytic deficiencies of specific enzymes involved in heme biosynthesis.

Elevated serum iron markers point toward an association with hemochromatosis, which can be found in 2%-27% of patients with porphyria cutanea tarda.[2] In the present case, blisters and milia related to porphyria cutanea tarda are the clinical manifestations of underlying primary hemochromatosis. There is a clear-cut association between porphyria cutanea tarda and hemochromatosis; the latter interferes with the transferrin receptor, causing a decrease in the affinity with which the receptor binds transferrin. This interaction may modulate cellular iron uptake and decrease ferritin levels. When a mutant or nonfunctional variant of the hemochromatosis gene is present, it leads to enhanced accumulation of iron in peripheral tissues.[3] Two separate gene mutations in the hemochromatosis gene (C282Y and H63D) have been identified as responsible for the disease. Treatment consists of removing probable secondary disease triggers, photoprotection, phlebotomy as an efficient treatment of

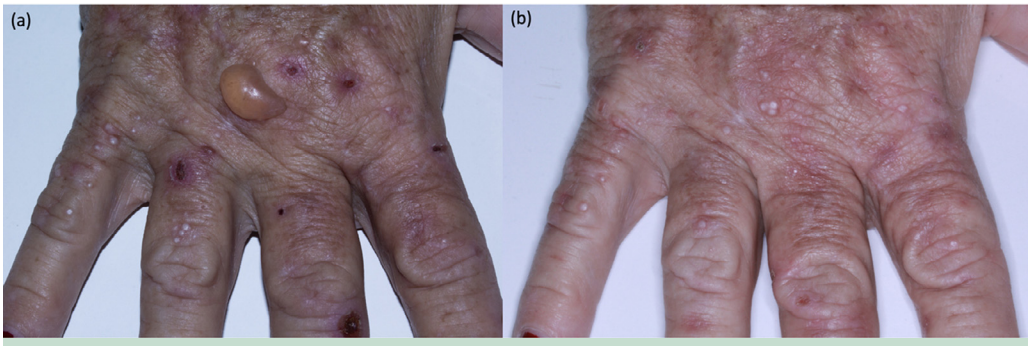

Figure 1 **(A)** Clinical image shows blisters, erosions, and milia. **(B)** Clinical image after 2 phlebotomy sessions.

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iron removal, resulting in improvement of hepatic URO-D activities, and the use of hydroxychloroquine.[4]

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