Porphyria Cutanea Tarda

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A 44-year-old man with human immunodeficiency virus infection, untreated hepatitis C virus infection, and active alcohol abuse presented with bilateral blistering on the dorsal hands of 2 months' duration (Figure 1). The blisters were pruritic and photoexacerbated and resolved spontaneously with scarring. Examination revealed tense bullae limited to the dorsal hands in various stages of healing, including milia and scarring. Biopsy specimens for histopathologic evaluation and direct immunofluorescence revealed pauci-inflammatory subepidermal clefting and homogeneous deposition along the basement membrane and within walls of superficial dermal vessels with multiple conjugates, consistent with porphyria cutanea tarda (PCT; Figure 2). The patient's baseline ferritin concentration was found to be higher than 800 ng/dL (to convert to pmol/L, multiply by 0.02247). Serum porphyrins demonstrated increased total porphyrins; fractionation showed increased uroporphyrin, confirming PCT.

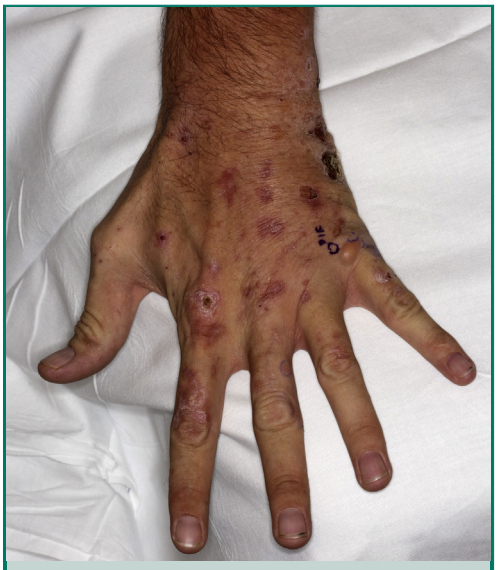

FIGURE 1. Intact and ruptured blisters with overlying hemorrhagic crust on the left hand.

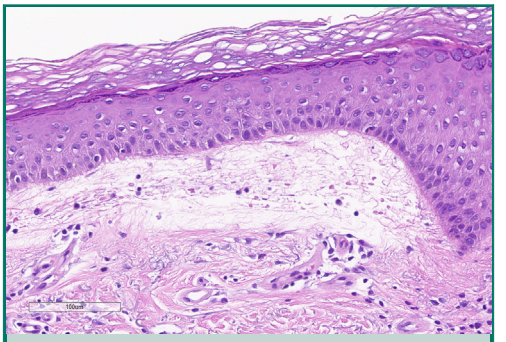

FIGURE 2. A pauci-inflammatory subepidermal blister on histopathologic examination (hematoxylin and eosin, magnification ×200).

PCT, the most common type of porphyria, is caused by inhibition of hepatic uroporphyrinogen decarboxylase (UROD). Pathogenesis involves cutaneous phototoxicity due to deposition of alternative metabolic products from excess hepatic porphyrinogen accumulation. Approximately 20% of cases are associated with familial UROD mutations,[1,2] whereas 80% of patients present with acquired PCT with absence of UROD mutations. These patients often have liver damage, frequently from alcohol, hepatitis C, human immunodeficiency virus infection, or hemochromatosis.[2-5] Typical presenting features include bullae and dyspigmentation of sun-exposed areas. First-line treatment is therapeutic phlebotomy of 450 mL every 2 weeks until ferritin concentration is less than 20 to 25 ng/dL.[2] Additional management recommendations include broad-spectrum sun protection, alcohol cessation, and hepatitis C treatment. Hydroxychloroquine can be used when phlebotomy is contraindicated.[2]

Although PCT is readily treated and nonfatal, relapses may follow successful treatment.[2]

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