A 5-year-old boy, born in the United States to Pakistani parents, had a history of skin problems and the passage of pink-red urine since birth. He developed severe phototoxic reactions, including vesicles and bullae with resultant scarring, in areas of the skin exposed to the sun. His face was most severely affected (Figure 1). The dorsum of both hands demonstrated hyperpigmentation with irregular areas of hypopigmentation and skin atrophy (Figure 2). In addition to the cutaneous changes, on physical examination he was found to have hepatosplenomegaly and red-stained teeth.

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The differential diagnosis of CEP includes other types of porphyria that are characterized by photosensitivity, such as hepatoerythropoietic porphyria and porphyria cutanea tarda. Although hepatoerythropoietic porphyria and porphyria cutanea tarda may present within the first few years of life, the photosensitivity seems to diminish with age and is followed by hypertrichosis, hyperpigmentation, and sclerodermalike scarring. Erythropoietic protoporphyria usually presents in childhood with burning or stinging sensations of the skin followed later by photocutaneous lesions. Vesicles and bullae are rare in this form of porphyria. Children with xerodermia pigmentosum have severe photosensitivity but normal porphyrin metabolism.

The prognosis of CEP is poor, frequently culminating in death in early adulthood and occasionally in the neonatal period. The only preventive measure is the absolute avoidance of sunlight exposure. Protection from trauma to the skin and aggressive treatment of cutaneous infections may help delay scarring and mutilation. Other therapies that have shown some benefit are oral beta carotene to reduce sun sensitivity, erythrocyte transfusions and splenectomy for the hemolytic anemia, and the oral administration of adsorbents such as charcoal and cholestyramine. Allogenic bone marrow transplantation may be curative in patients with severe phenotypes.

Some students of the porphyrias believe that the clinical picture of individuals with CEP, particularly their avoidance of sunlight, the mutilated skin, red teeth, and hypertrichosis, led to the development of the folklore of the werewolf in medieval Europe.

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