Pathological Case of the Month

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An otherwise healthy 13-month-old boy was taken to the pediatrician for evaluation of “crying when in the sun,” leathery appearance of the skin on the dorsum of the hands (Figure 1), and irritation and redness of the skin of the upper lip and bridge of nose. Five months later, when the child moved from a northern climate to Georgia, the mother noted marked worsening of the redness and sun-induced sensitivity of his skin. A clinical diagnosis of erythropoietic protoporphyria (EEP) was made. Laboratory studies showed a florescence (4+) of the red blood cells. Erythrocyte protoporphyrin level was 3.96 µmol/L (normal, <0.62 µmol/L). A biopsy specimen from an affected area of skin on the right dorsal hand is shown in Figure 2.

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Erythropoietic protoporphyria (EPP) is an autosomal dominant disorder of variable expressivity that usually presents between the ages of 2 and 5 years with complaints of burning, stinging, or itching of sun-exposed skin.1 There may be edematous, erythematous plaques or rare papulovesicular lesions. Chronic changes of the skin include dyspigmentation, papular thickening, or hyperkeratosis of the skin of the dorsal hands, shallow pitted or linear scars of the face (especially the nose), and linear furrows around the lips. Systemic symptoms are often absent but may include cholecystitis or cholelithiasis, hypersplenism, and rarely hepatic failure due to portal and periportal fibrosis.

The disease results from a congenital abnormality in the enzyme ferrochelatase, which is necessary for the biosynthesis of heme. The resultant accumulation of the heme precursor, protoporphyrin IX in the red blood cells, liver, bone marrow, and plasma leads to the photosensitivity and systemic symptoms described earlier. The protoporphyrin level in the serum and red blood cells is markedly elevated in patients with EPP, and diagnosis can be made by measuring the level of this protoporphyrin in the serum or erythrocytes.2 The route of elimination of protoporphyrin is via the feces; thus, one may also measure fecal protoporphyrin levels to confirm a diagnosis of EPP. The large protoporphyrin molecule is poorly soluble in water, therefore, unlike other types of porphyria, the urine porphyrin levels are normal in EPP.

Histological features of EPP are distinctive. In the sun-exposed areas of skin, there is marked thickening of the blood vessel walls in the papillary dermis. The blood vessels are surrounded by an eosinophilic, homogenized material that is periodic acid-Schiff-positive and diastase resistant (Figure 2 and Figure 3). Immunofluorescence studies have shown this material to be type IV collagen, consistent with reduplication of the perivascular basal lamina.3

Treatment consists of vigorous photoprotection, especially to UV-A radiation, as the protoporphyrin molecule photosensitizing affinity falls in the UV-A (400- to 410-nm) range. Patients should avoid sunlight, especially midday, whenever possible. Additional photoprotection should include use of UV-A-blocking sunscreens such as titanium oxide or zinc oxide.4 Protective clothing should also be worn. Since UV-A light is not filtered by ordinary window glass, patients are not fully protected even while indoors. Some florescent and incandescent indoor light sources also emit UV-A radiation and may cause problems for patients with EPP.

Beta-carotene (Solatene, Hoffman-LaRoche, Nutley, NJ), a natural constituent of several plant sources (ie, carrots, tomatoes, and oranges), has been used with benefit in patients with EPP. Cholestyramine may also encourage fecal protoporphyrin elimination.

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The opinions and assertions contained herein are the private ones of the authors and are not to be construed as official or reflecting the views of the Department of Defense or the US Army.

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