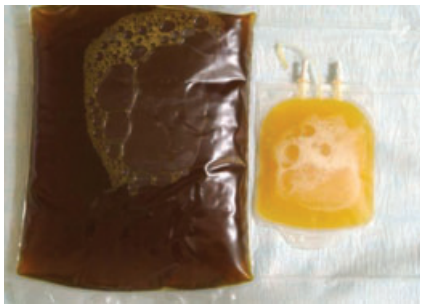
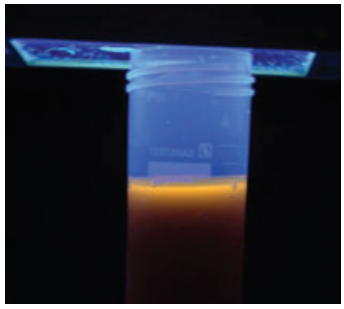
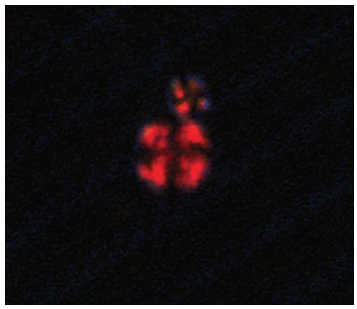


A



B



C

Erythropoietic protoporphyria

Robertson D. Davenport

This 56-year-old woman had experienced solar sensitivity since age 15, resulting in pain and blistering. Her father, two of her seven siblings, and one of her two children had similar solar sensitivity. Three years ago she was diagnosed with erythropoietic protoporphyria. Liver biopsy at that time showed cirrhosis with prominent periportal cholestatic changes. Now she presented with encephalopathy and ascites. The patient was listed for liver transplantation because of worsening hepatic function. Laboratory examination showed a normocytic, normochromic anemia with burr cells and target cells. Free erythrocyte protoporphyrin (FEPP) was markedly elevated at 2037 μg per dL (normal range, 1-10) and zinc-complexed erythrocyte protoporphyrin (ZnEPP) was elevated at 663 μg per dL (normal range, 10-38). Plasmapheresis followed by hematin infusion three times per week was initiated in an attempt to lower the protoporphyrin levels to protect the transplanted liver. The plasma from the first procedure was a deep red-brown color (A; normal plasma is shown on the right for comparison). Under ultraviolet light, the plasma showed an orange fluorescence (B). After 10 apheresis and infusion treatments, FEPP was 1827 μg per dL and ZnEPP was 845 μg per dL. The patient was successfully transplanted, during which she received 22 units of red blood cells and 22 units of fresh-frozen plasma. The liver was cirrhotic and contained crystals visible under polarized light having a Maltese cross-like pattern characteristic of protoporphyrin (C). Four days after transplantation, FEPP was 164 and ZnEPP was 37. Presently, the management plan is to follow the FEPP level and reinstitute hematin infusions if it is significantly elevated. Ultimately, hematopoietic progenitor cell transplantation from an unaffected sibling is contemplated.

Erythropoietic protoporphyria is an autosomal dominantly inherited deficiency of ferrochelatase, the final step in heme synthesis. Protoporphyrin concentrations are elevated in red blood cells, plasma, bile, and feces. Photosensitivity usually begins in childhood. Gallstones are common. Liver disease is unusual but can be severe. Liver transplantation is at present the only adequate intervention in cases of hepatic failure but will not prevent on-going protoporphyrin accumulation. In this case, plasmapheresis and hematin infusion had little effect in the short term. However, effective exchange transfusion strikingly reduced the protoporphyrin level.

ABBREVIATIONS: FEPP = free erythrocyte protoporphyrin; ZnEPP = zinc-complexed erythrocyte protoporphyrin.

From the University of Michigan Medical School, Ann Arbor, Michigan.

Address reprint requests to: Robertson D. Davenport, MD, Department of Pathology, 1500 E. Medical Center Dr., Ann Arbor, MI 48109-0054; e-mail: rddvnprt@umich.edu.

Received for publication February 18, 2004; revision received March 12, 2004, and accepted March 12, 2004.

TRANSFUSION 2004;44:1401.