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IMAGE | LIVER

Liver Involvement in Erythropetic Protoporphyria

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Case Report

A 19-year-old man presented with painless jaundice for 3 months. He had a 14-year history of recurrent, photosensitive, painful rashes without blistering on exposed areas of the face and hands. Examination revealed icterus, hyperpigmentation, hyperkeratosis, and leathery texture over affected hands with onycholysis, loss of lunulae, melanonychia, and firm hepatosplenomegaly (Figure 1). Laboratory tests revealed anemia with red blood cell hypochromasia and thrombocytopenia. There was evidence of advanced liver disease with marked hyperbilirubinemia, elevated INR, and hypoalbuminemia. Plasma and fecal protoporphyrin levels were elevated. Liver histopathology revealed portal tracts with mixed inflammatory infiltrates and brown-colored bile-laden macrophages with portal fibrosis and hepatocytes with aggregated cellular and canalicular brownish red-colored bile deposits (Figure 2). Red birefringence and clusters of brilliantly illuminated granules in a Maltese cross pattern on polarized light were also visible (Figure 3). Erythropoietic protoporphyria (EPP) diagnosis was made. The patient progressed liver failure at 8 weeks' follow-up, and is awaiting liver transplantation.

EPP manifests in childhood, rarely in early adulthood, with recurrent cutaneous photosensitivity without typical bullae or vesicles (characteristic of other acute porphyrias), leading to chronic skin changes. Hepatobiliary disease due to deficiency of ferrochelatase enzyme, the last enzyme step in heme biosynthesis, can also occur, with 5% developing advanced liver diease.[1] The spectrum of hepatobiliary disease associated with EPP includes cholelithiasis, mild parenchymal liver disease, progressive hepatocellular disease, and end-stage liver disease.[2] Liver disease develops due to precipitation of protoporphyrin in hepatocytes and biliary radicles. Diagnosis of EPP depends on demonstration of increased protoporphyrin levels in erythrocytes, plasma,

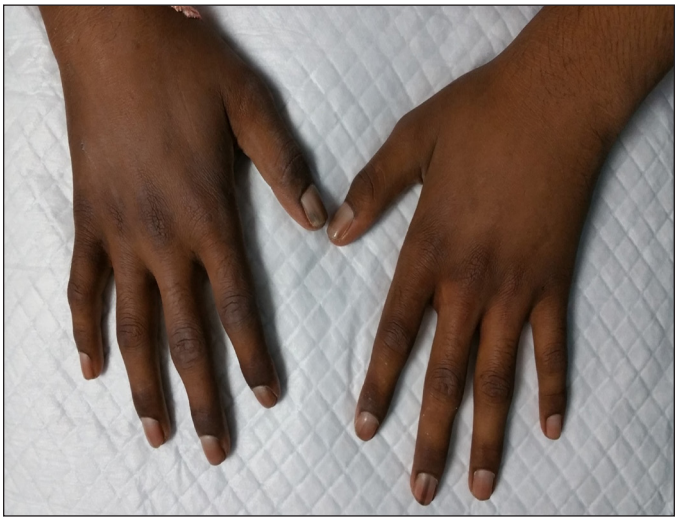

Figure 1. Hyperpigmentation, hyperkeratosis, and leathery texture of hands with onycholysis, loss of lunulae, and melanonychia.

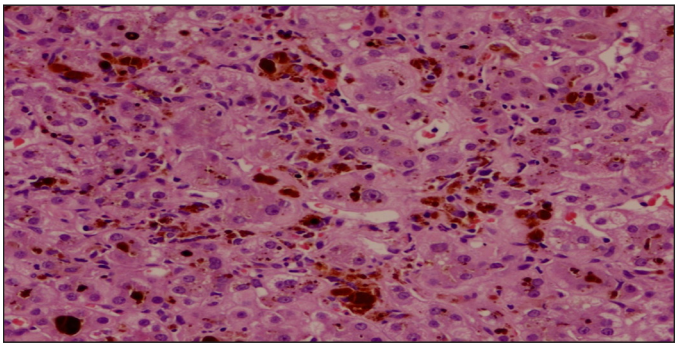

Figure 2. Liver histopathology showing portal tracts with mixed inflammatory infiltrates, brown-colored bile laden macrophages with portal fibrosis and hepatocytes, and aggregated cellular and canalicular brownish red-colored bile deposits (H&E staining, x20 magnification).

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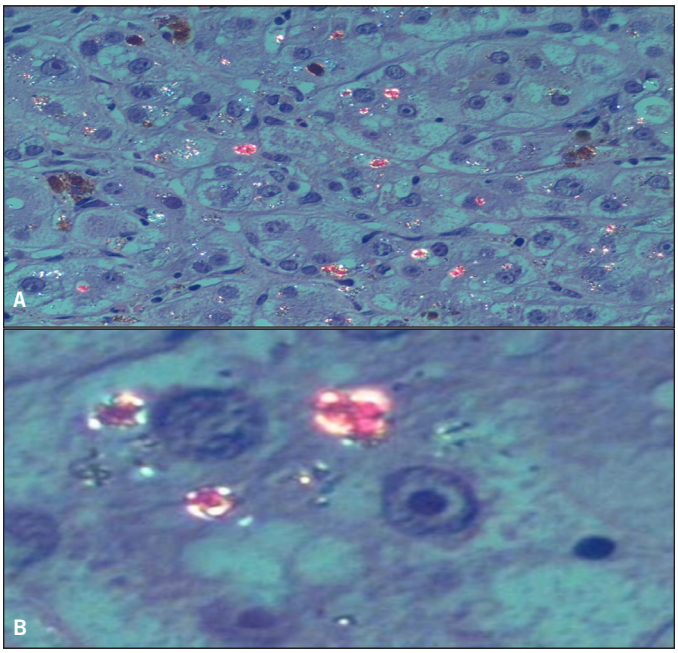

Figure 3. The tissue showed (A) red birefringence and clusters of brilliantly illuminated granules in polarized light at x20 magnification and (B) Maltese cross pattern (arrow) at x40 magnification.

and stool, with normal urine porphyrin levels. Very high plasma and erythrocyte protoporphyrin levels increase the risk of liver disease. Polarized microscopy of liver biopsy shows characteristic birefringent crystal deposits with Maltese cross patterns.[3]

Treatment includes blood transfusion to correct anemia, hemin to decrease protoporphyrin levels, ursodeoxycholic acid to increase excretion into bile, cholestyramine and activated charcoal to interrupt enterohepatic protoporphyrin circulation, haemodialysis or plasmapheresis to remove excess protoporphyrin, antioxidant and cytoprotective therapy with beta carotene and vitamin E, liver transplantation for advanced liver disease, and bone marrow transplant for correction of the defect.[4]

Disclosures

Author contributions: CA Philips wrote the manuscript and is the article guarantor. C. Bihari acquired the data and figures and wrote the manuscript.

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References

1. Sarkany RP, Alexander GJ, Cox TM. Recessive inheritance of erythropoietic protoporphyria with liver failure. *Lancet*. 1994;343(8910):1394–6.
2. Anstey AV, Hift RJ. Liver disease in erythropoietic protoporphyria: Insights and implications for management. *Gut*. 2007;56(7):1009–18.
3. Seth AK, Badminton MN, Mirza D, Russell S, Elias E. Liver transplantation for porphyria: Who, when, and how? *Liver Transpl*. 2007;13(9):1219–1227.
4. Thapar M, Bonkovsky HL. The diagnosis and management of erythropoietic protoporphyria. *Gastroenterol Hepatol*. 2008;4(8):561–566.