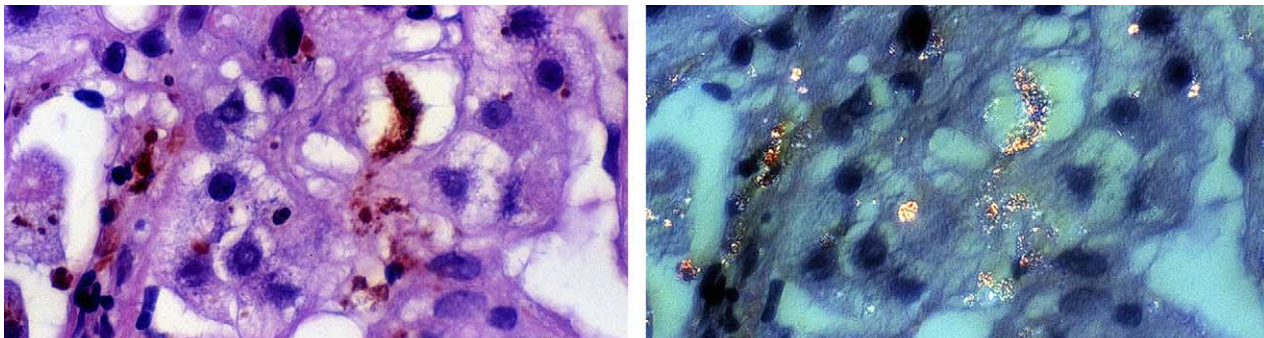


Images in Hepatology

Associate Editor: Dominique Charles Valla

Erythropoietic protoporphyria in an adolescent female



A 16-year-old black female presented with a history of chronic hepatitis of unknown etiology and intermittent episodes of right upper quadrant and back pain; she reported no photosensitivity. Previous evaluation included negative serology for HBV, HCV, EBV, and autoimmune hepatitis; normal serum levels of ceruloplasmin and α -1-antitrypsin, and 24-h urine for copper. Admission laboratory values showed mild elevations of bilirubin (total 3.56 mg/dl, conjugated 1.46 mg/dl), transaminases (ALT 224 U/l), GGT (341 U/l), and lipase (1838 U/l), as well as anemia (hematocrit 29.5%). Ultrasound demonstrated only gallbladder sludge with small stones. At cholecystectomy the liver appeared cirrhotic; an intraoperative liver biopsy was performed.

Histological examination showed well-developed cirrhosis, slightly distorted lobular architecture, hepatocyte ballooning degeneration, and a large degree of cholestasis (left panel). Under polarized light birefringent material was seen within the accumulated bile pigment, some in the form of Maltese crosses (right panel). Erythropoietic protoporphyria (EPP) was diagnosed based on histological features and elevated levels of erythrocyte protoporphyrin (7624 mcg/dl) and serum protoporphyrin (7.5 mcg/dl).

In EPP there is an abnormality in heme biosynthesis, specifically a defect of ferrochelatase, resulting in the accumulation of excess protoporphyrins and porphyrin precursors in the liver and skin [1]. Photosensitivity is the characteristic clinical finding in EPP, but progressive liver disease represents a more serious and life-threatening complication. Protoporphyrin-induced liver damage is mediated by the progressive accumulation of protoporphyrin in hepatocytes which interrupt bile flow and cause damage [1]. Grossly, the liver is enlarged, firm, and black in color. Histologically, there is nodular cirrhosis, hepatocellular necrosis, portal inflammation, and cholestasis; pigment is birefringent when examined by polarization microscopy [2]. Liver transplantation has been successfully done for end-stage liver disease due to EPP. Measures should be taken to avoid prolonged or excessive exposure to operating lights during surgery, which can lead to phototoxic effects to the skin and intra-abdominal organs [3].

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