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Clinical Image

Erythropoietic protoporphyria: Delayed presentation with decompensated liver disease

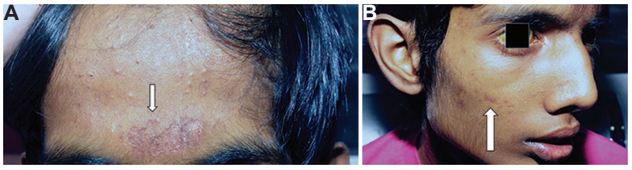

Fig. 1. (**A** and **B**) Repeated episodes of photosensitivity causing small erosions on exposed parts, especially the face, leading to scarring in the form of linear or circular pits (arrow).

Fig. 2. Nail changes in the form of photo-onycholysis due to photosensitivity (arrows).

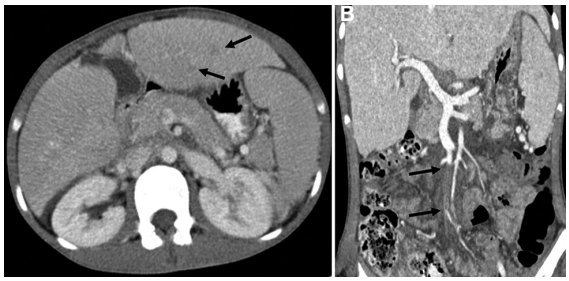

Fig. 3. (**A**) Axial contrast-enhanced computed tomographic (CT) image showing subtle lesions in the left lobe of liver (arrows). (**B**) Coronal CT portal venography image showing thrombosis of a segment of superior mesenteric vein (arrows).

A 17 yr old male child[†] presented to the department of Medicine, All India Institute of Medical Sciences, New Delhi, India, in April 2019, with complaints of jaundice and progressive abdominal distension for four months and recurrent non-specific abdominal pain for three months. He had received multiple antibiotics and antacids with no relief. Investigations revealed conjugated hyperbilirubinaemia with transaminitis and hepatosplenomegaly with Grade 3 oesophageal varices and superior mesenteric vein (SMV) thrombosis. Common causes of liver failure were ruled out. After probing multiple times, the patient gave non-specific history of painful non-blistering photosensitivity during summer in childhood and similar episodes before symptom onset at presentation. Repeated episodes of photosensitivity cause small erosions on exposed parts, especially face, leading to scarring in the form of linear or circular pits (Fig. 1). Nail changes in the form of photo-onycholysis were also present (Fig. 2). The skin lesions were initially not apparrently suggestive of erythropoietic protoporphyria (EPP) in any case. Screening for porphyria was negative in urine as protoporphyrin is not excreted in urine, however, plasma porphyrin screen showed peak at 634 nm,

[†]The child's assent and parents' consent obtained to publish clinical information and images.

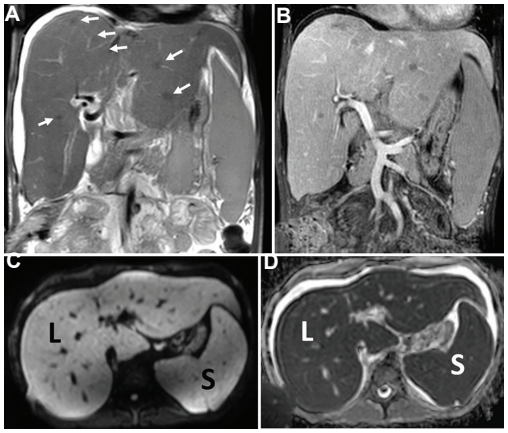

Fig. 4. Coronal T2-weighted (**A**) magnetic resonance images (MRI) showing multiple lesions in both lobes of the liver, appearing hypointense (arrows). **(B)** Coronal contrast-enhanced T1-weighted MRI showing multiple lesions in both lobes of the liver, appearing hypoenhancing (arrows). **(C)** Axial diffusion-weighted MRI showing diffuse restriction of diffusion of liver parenchyma. (**D**) Apparent diffusion magnetic resonance coefficient map showing diffuse restriction of liver parenchyma.

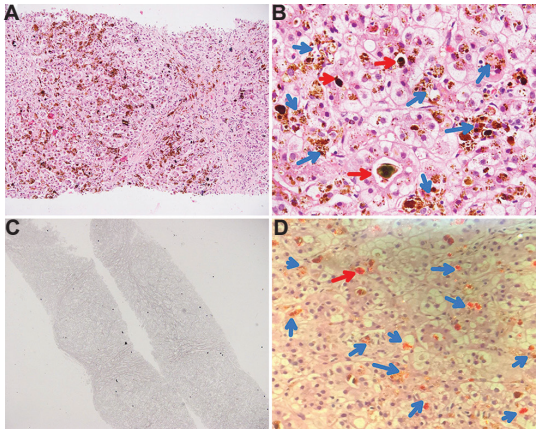

Fig. 5. (**A**) Sections examined from the liver biopsy showing distortion of lobular architecture, porto-portal bridging fibrosis and nodule formation (H and E, ×40). (**B**) There is canalicular cholestasis with cholestatic rosette formation (red arrows) and hepatocyte degeneration along with deposition of brown refractile pigment (blue arrows) in the hepatocytes (H and E, ×400). (**C**) Nodular architecture of the liver (×40) (reticulin stain). (**D**) The brown pigment displaying red fluorescence (blue arrows) and Maltese cross appearance (red arrow) on polarization.

suggestive of EPP. Initial contrast-enhanced computed tomographic abdomen had shown SMV thrombosis and subtle non-specific lesions in the left lobe of the liver (Fig. 3), so magnetic resonance imaging was planned which showed multiple hypointense lesions in the liver (Fig. 4). Liver biopsy of the lesions revealed deposition of brown refractile pigment, which displayed red fluorescence and Maltese cross on polarization which is a characteristic of EPP (Fig. 5). Treatment for EPP is liver transplantation followed by bone marrow transplantation. Unfortunately, the patient succumbed to liver failure within one month of diagnosis as there is no medical management after the liver gets involved.

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***Conflicts of Interest*:** None.

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