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Authors:
Luísa Martins Figueiredo, Luis Lourenço, David Horta, Alexandra Martins

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A complex case of low-phospholipid-associated cholelithiasis syndrome

Luísa Martins Figueiredo, Luís Lourenço, David Horta and Alexandra Martins

Gastroenterology Department. Hospital Professor Doutor Fernando Fonseca. Amadora, Portugal

Correspondence: Maria Luísa Marques Martins Figueiredo
e-mail: luisa_mmfigueiredo@hotmail.com

We present the case of a 40-year-old female patient with a family history among first-degree relatives of gallstone disease and a medical history of choledocholithiasis, cholestatic parameters and intrahepatic biliary ducts (IHBD) dilatation. She underwent magnetic resonance cholangiopancreatography (MRCP), which showed diffuse dilation of IHBD and intrahepatic stones, without extrahepatic biliary tract alterations. The liver biopsy revealed ductopenia and fibrosis around small ducts. She was treated with ursodeoxycholic acid (UDCA) at 750 mg/d and remained asymptomatic. Two years later, she complained about pruritus, right hypochondrium pain and dark urine, accompanied by elevated liver tests. Abdominal ultrasound showed dilated IHBD and multiple bile duct stones, without gallstones. Endoscopic retrograde cholangiopancreatography (ERCP) confirmed multiple stones of the common hepatic duct and lithiasis in the left hepatic duct (Fig. 1). A plastic biliary stent was placed after sphincterectomy.

A genetic mutation screening of the ABCB4 and ABCB11 genes was performed, which was positive for low-phospholipid-associated cholelithiasis syndrome (LPAC) spectrum mutations. Following the first episode, four ERCPs were performed and stones were extracted after fragmentation with an intraductal laser, placement of plastic biliary stents and a nasobiliar probe (Fig. 2). An MRCP was repeated, with persistence of lithiasis of the left and right IHBD (Fig. 3). One year later, a portal vein thrombosis was diagnosed and she was referred to the Hepatic Pre-Transplant Clinic.

DISCUSSION

LPAC syndrome is a specific form of symptomatic and recurring cholelithiasis, associated with a defect in ABCB4 function that increases lithogenicity (1-3). The recommended medical therapy is UDCA (1,3). A liver transplant may be indicated in the rare cases that progress to end-stage liver disease (2,3).

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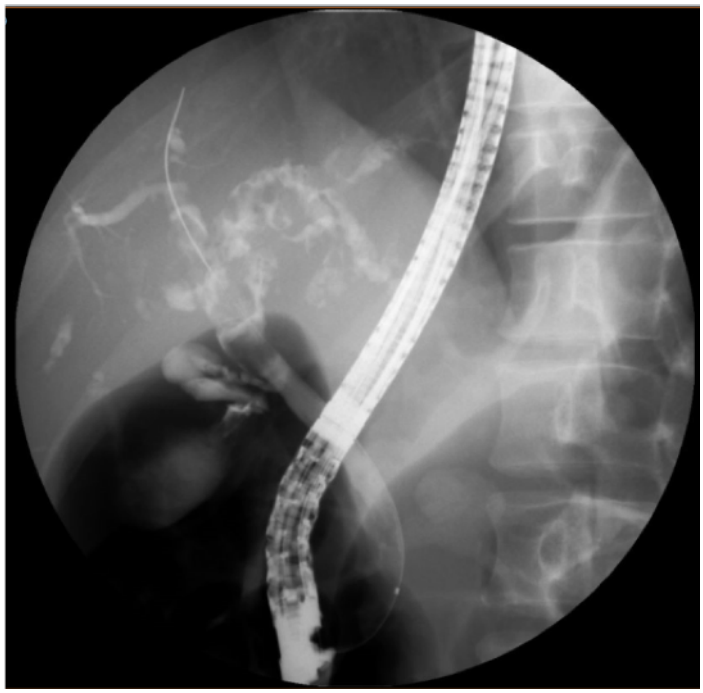

Fig. 1. Proximal CBD with large multiple stones (15-20 mm), causing a marked dilatation at this level and upstream. Bile duct stones of more than 10 mm in the left hepatic duct and irregularity of the right hepatic duct in relation to lithiasis.

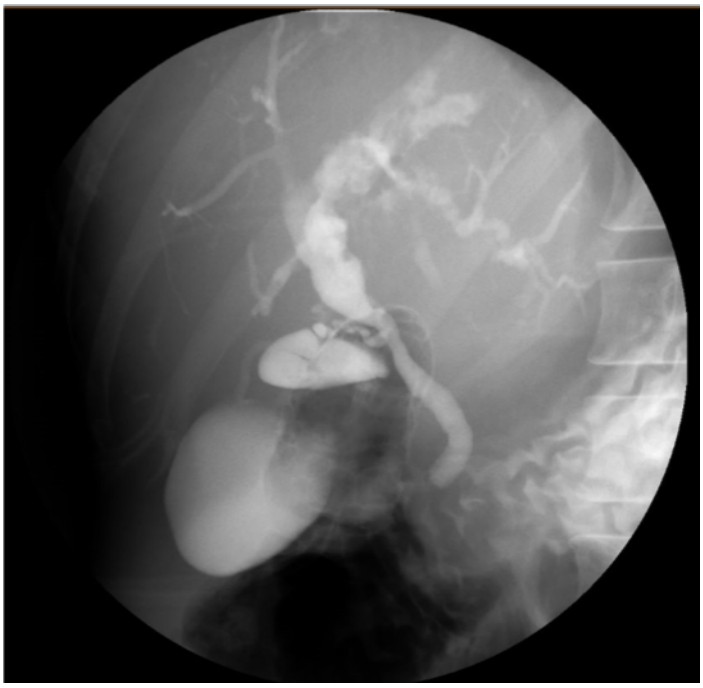

Fig. 2. The last ERCP with rarefaction of segmental branches, without choledocholithiasis.

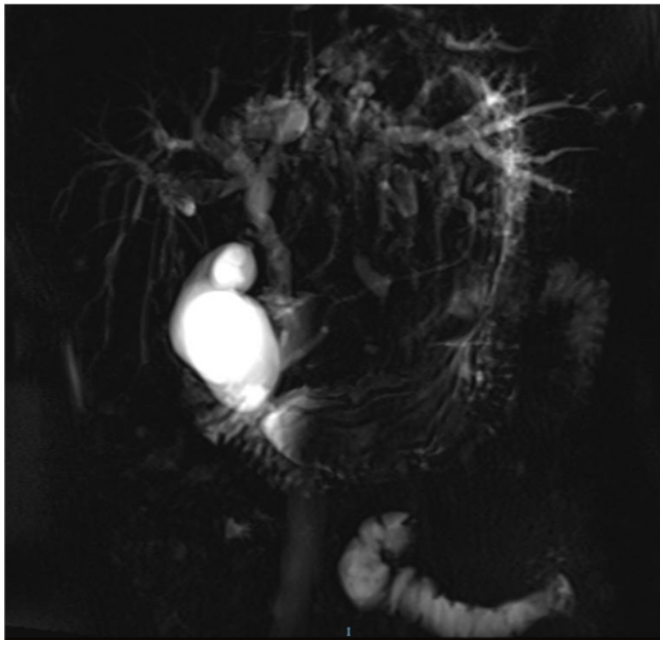

Fig. 3. Marked dilation of the left IHBD with several points suggestive of stenosis and several defects of repletion, suggestive of lithiasis. In addition, the right IHBD also has several defects suggestive of bile duct stones with no gallstones, despite being less dilated and regular.