

Title:

**A complex case of low-phospholipid-associated
cholelithiasis syndrome**

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DOI: 10.17235/reed.2020.6625/2019

Link: [PubMed \(Epub ahead of print\)](#)

Please cite this article as:

Figueiredo Luísa Martins, Lourenço Luis, Horta David,
Martins Alexandra. A complex case of low-phospholipid-
associated cholelithiasis syndrome . Rev Esp Enferm Dig
2020. doi: 10.17235/reed.2020.6625/2019.



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ACCEPTED MANUSCRIPT

1 **IPD 6625**

2 **A complex case of low-phospholipid-associated cholelithiasis syndrome**

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

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

31

32 **DISCUSSION**

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

37

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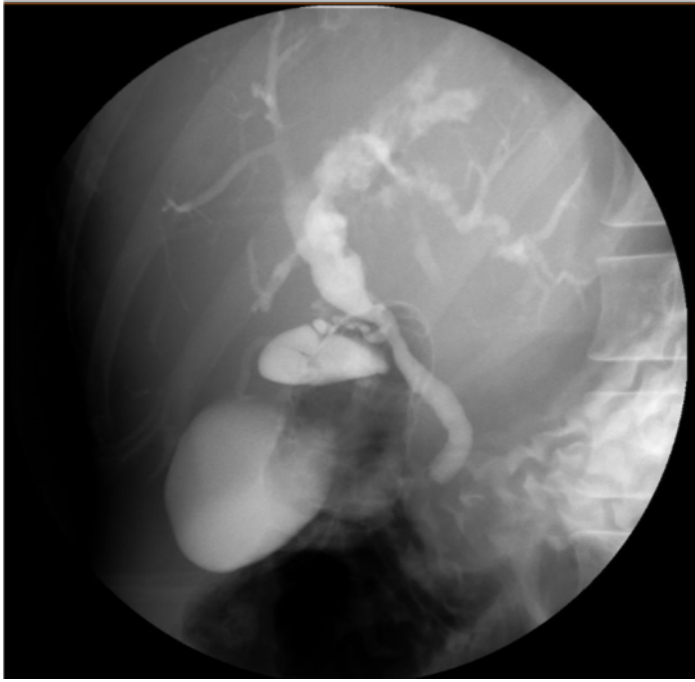


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48 Fig. 1. Proximal CBD with large multiple stones (15-20 mm), causing a dilatation at
49 this level and upstream. Bile duct stones of more than 10 mm in the left hepatic duct and
50 irregularity of the right hepatic duct in relation to lithiasis.

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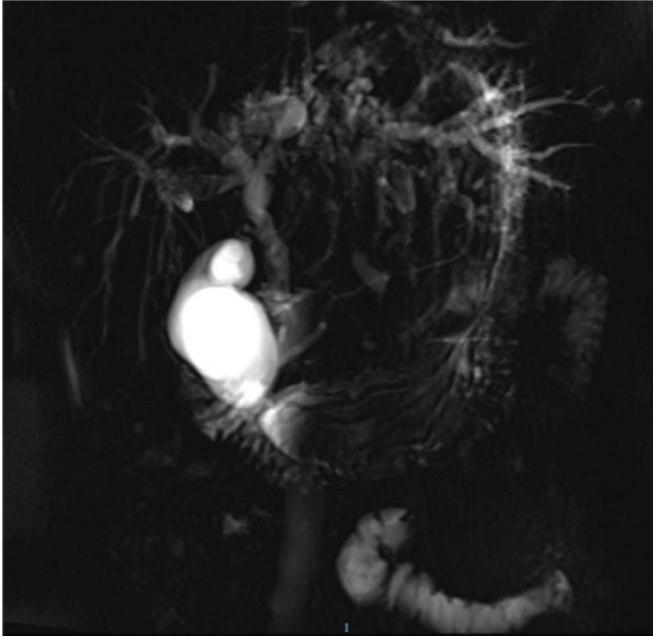


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53 Fig. 2. The last ERCP with rarefaction of segmental branches, without choledocholithiasis.

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

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