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Case Report

Low Phospholipid Associated Cholelitiasis (LPAC) Syndrom Revealed by Acute Pancreatisis

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Abstract

Low Phospholipid-Associated Cholelithiasis (LPAC) is a genetic disorder characterized by intrahepatic lithiasis due to decreased secretion of phosphatidylcholine, leading to cholesterol stone formation in both the gallbladder and liver. LPAC is often linked to mutations in the MDR3/ABCB4 gene, which encodes the bile protein MDR3. This defective protein fails to transport phosphatidylcholine into the bile, resulting in cholesterol crystal accumulation in the intrahepatic bile ducts. A case study of a 27-year-old male, previously cholecystectomized, presented with epigastric pain, vomiting, and jaundice, revealing elevated lipase, liver enzymes, and bilirubin levels. Imaging showed pancreatic enlargement and the main bile duct and intrahepatic bile ducts dilation with lithiasis. Endoscopic retrograde cholangio-pancreatography confirmed the diagnosis and treatment with ursodeoxycholic acid was initiated.

Introduction

LPAC is a genetic disease that causes intrahepatic lithiasis. It is a very special form of gallstone disease, in which excess cholesterol is not eliminated in the bile, but the secretion of phosphatidylcholine decreases, which causes the formation of stones not only in the gallbladder, but also in the liver [1]. It is often caused by a mutation in the MDR3/ABCB4 (multidrug resistance/ATP-binding cassette, subfamily B, member 4) class III gene, which encodes the bile protein MDR3. MDR3 (now ABCB4) is a member of the ABC protein superfamily. It is a flippase that functions to transfer the phospholipid phosphatidylcholine from the inner membrane of the channel to the outer membrane. From there, phosphatidylcholine is washed into the bile with the help of bile acids. This genetic mutation results in a defective protein that partially or completely does not transport this essential phospholipid into the bile, leading to dissolution of cholesterol, which accumulates as crystals in the intrahepatic bile duct and duct [2]. LPAC syndrome usually presents as biliary symptoms or complications (eg, biliary pain, acute pancreatitis, cholecystitis, or cholangitis) in young patients who are not overweight [1].

Observation

A 27-year-old man, who has undergone a cholecystectomy 4 years ago, presented with epigastric pain radiating to the back and vomiting with no fever or other symptoms. At physical examination, there was an epigastric sensibility and a discrete jaundice. Biological tests revealed a high rate of lipase (≥ 3 ×upper limit of normal), a cytolysis associated with a cholestasis with aspartate aminotransferase (ASAT) 272 IU/L (normal: 0-55 IU/L), alanine aminotransferase (ALAT) 213 IU/L (normal: 5-34 IU/L), total bilirubin 18,5 mg/l (normal: 3-12 mg/l), direct bilirubin 14,2 mg/l (normal: 1-4 mg/dL), alkaline phosphatase (ALP) 80 IU/L (normal: 40-150 IU/L), and gamma-glutamyltransferase (GGT) 190 IU/L (normal: 9-36 IU/L). Contrast enhanced CT showed an enlarged pancreas, without loss of its physiological lobulations with infiltration of the peripancreatic fat and discrete dilatation of the intrahepatic bile

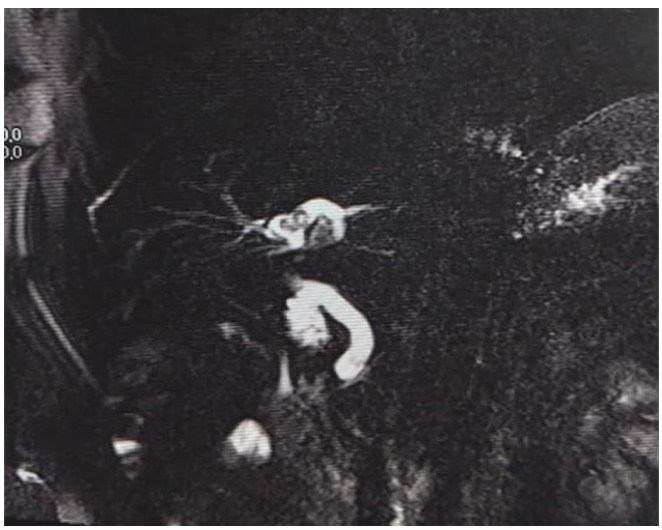

Figure 1: image of intrahepatic lithiasis on MRI.

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ducts. MRI demonstrated a dilatation of the main bile duct and intrahepatic bile ducts upstream of lower bile duct lithiasis, associated with intrahepatic lithiasis.

An endoscopic retrograde cholangio-pancreatography was performed and showed lithiasis in the distal part of the main bile duct and dilatation of the intrahepatic bile ducts with visualization of lithiasis within it. A lithiasis of about 9 mm was extracted and a biliary stent has been placed after sphincterotomy. Post-operative course was regular with no complication occurred. Patient was discharged home with ursodeoxycholic acid 10 mg/kg daily treatment. Low phospholipid-associated cholelithiasis (LPAC) could be evocated in this case.

Discussion

LPAC syndrome is a specific form of symptomatic and recurrent cholelithiasis associated with ABCB4 dysfunction, which increases lithogenicity [3].

The prevalence of LPAC syndrome has not been precisely estimated, but it affects about 5% of patients with symptomatic cholelithiasis [1].

Cholelithiasis is symptomatic in LPAC syndrome and characteristic biliary pain leads to cholecystectomy in more than 90% of cases [4]. Recurrent symptoms after cholecystectomy are a source of worry because they are associated with intrahepatic lithiasis or lithiasis migration. Serious complications (pancreatitis, acute cholangitis, intrahepatic lithiasis) appear to be more common in men. Their recurrence after cholecystectomy indicates that they are more closely related to intrahepatic. It can be associated with biological cholestasis, particularly as regards GGT levels, and is likely to be connected with chronic cholangiocyte aggression [4].

Radiological examinations are essentiel to the positive diagnosis of LPAC syndrome. They must be performed and interpreted by a radiologist informed of the diagnostic suspicion. The rate of detection of signs of LPAC syndrome can vary from 5% (unexpected radiologist) to 90% (experienced radiologist) [4]. Ultrasound is mainly performed, and in 80-85% of patients "indicative" signs can be identified: intrahepatic hyperechoic foci with posterior acoustic shadow or "comet tail" images spreading along the portal axis, intrahepatic sludge, microlithiasis [5]. The diagnosis of LPAC syndrome uses two of three criteria: the appearance of biliary symptoms in patients younger than 40 years, recurrent biliary pain after cholecystectomy, and typical radiological features of intrahepatic micro- or microlithiasis [6]. Bile analysis is not recommended for routine clinical examination. Although a mutation in the ABCB4 gene occurs in only 56-65% of cases, its absence does not invalidate the diagnosis of LPAC syndrome [4].

Although the reason behind its efficacy is not completely understood, treatment with ursodeoxycholic acid (UDCA) seems to diminish symptoms of LPAC syndrome in the majority of cases [1,3,5] . Although the reason for the effectiveness of ursodeoxycholic acid (UDCA) is not fully understood, ursodeoxycholic acid (UDCA) appears to reduce the symptoms of LPAC syndrome in most cases. Such treatment is usually long-term, but it is not clear whether the treatment should be carried out throughout the patient's life. Improving understanding of this syndrome will facilitate patient screening and treatment [1].

A liver transplant may be indicated in those rare cases where end-stage liver disease develops [3].

Conclusion

LPAC syndrome is easy to diagnose and treat; therefore, it should no longer be overlooked. To increase its detection rate, all patients who experience recurrent biliary symptoms following an episode of acute pancreatitis should undergo an ultrasound examination performed by a radiologist with knowledge of the disease.

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