

Abernethy malformation with multiple aneurysms: incidentally found in an adult woman with Caroli's disease

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ABSTRACT

Abernethy malformation is a rare anomaly with partial or complete congenital absence of the portal vein and subsequent development of extrahepatic portocaval shunts. We present the case of a 28-year-old woman who was incidentally diagnosed with type II Abernethy malformation and multiple aneurysms during an investigation for nonspecific abdominal pain and fever. The patient had been diagnosed with Caroli's disease at the age of 10 and liver cirrhosis, portal hypertension a few years before. To the best of our knowledge, this is the first case reported with all such congenital anomalies associated together. Ultrasound, computed tomography, including three-dimensional reconstruction, and magnetic resonance imaging were performed which revealed a side-to-side shunt between the extrahepatic portal vein and the inferior vena cava, multiple aneurismal cystic dilation of the spleen artery and left renal artery, and extensive intrahepatic bile duct cystic dilation with calculus formation. Etiology, clinical significance and management strategies with regard to these abnormalities are discussed.

Key words: Abernethy malformation. Caroli's disease. Multiple aneurysm. Liver cirrhosis. Computed tomography.

INTRODUCTION

Abernethy malformation, also termed as congenital extrahepatic portosystemic shunt, is a rare condition. It was firstly described by John Abernethy in 1793 in a 10-month-old girl. The girl died of unknown cause, and the postmortem examination showed termination of the portal vein in the inferior vena cava at the level of the insertion of the renal veins and many other multiple congenital anomalies, such as dextrocardia, transposition of the great vessels, and polysplenia.¹ Abernethy malformation has been classified into two types based on the pattern of anastomosis between the portal vein (PV) and inferior vena cava (IVC), and the presence or absence of an intrahepatic portal venous supply. That is type I, the entire portal venous supply drains into the IVC with absence of the intrahepatic PV;

and type II, the portal venous blood partially drains into the IVC through side-to-side anastomosis.² Type I has a female prevalence and is always accompanied with anomalies such as liver and cardiac abnormalities, while type II has a male prevalence and is rarely accompanied with other anomalies.³ We present the case of a 28-year-old woman who was incidentally diagnosed with Abernethy malformation of type II and multiple aneurismal cystic dilation of the spleen artery and left renal artery, when she was in the treatment of biliary tract infection caused by Caroli's disease. The association of multiple aneurysms and Caroli's disease with Abernethy malformation of type II has not been previously described in the literature.

CASE REPORT

A 28-year-old woman presented at our institution on November 12, 2011 for complaints of right upper abdominal pain associated with intermittent fever for 7 days. The patient had a remarkable medical history. At the age of 10, she was diagnosed with Caroli's disease (congenital intrahepatic bile duct cystic dilation) and had been admitted to hospital many times for infection of biliary tract since 2003. In September 2004, she was diagnosed with liver cirrhosis, splenomegaly and portal hypertension

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during hospitalization for jaundice and biliary tract infection. In October 2011, an exploratory laparotomy for acute peritonitis was performed, and 2,000 mL purulent effusions were drained during the operation. Physical examination revealed mild tenderness in right upper quadrant and moderate splenomegaly, but no icterus. There was no evidence of peritonitis. Laboratory tests showed a hemoglobin level of 9.4 g/dL, white blood cell count of $4.47 \times 10^9/L$, platelet count of $94 \times 10^{12}/L$, and normal liver function tests. CA19-9 was 58.74 U/mL, while all other tumor markers were within a normal range. Abdominal ultrasound (Figure 1) showed a coarse heterogenous liver parenchyma with nodular changes, apparent intrahepatic bile duct dilation in the right liver lobe with sediment samples of cholestasis formation, and common bile duct dilation. Intrahepatic PV portion didn't show clearly, while the extrahepatic trunk of PV dilated significantly with portal embolization formation. Spleen enlarged remarkably. Doppler examination showed remarkable blood flow velocity reduction in the extrahepatic trunk of PV (9.44 cm/s), blood flow increase in the hepatic artery, and blood flow from the splenic vein towards the IVC in a splenic-caval shunt.

Dual-phase computed tomography (CT) (Figure 2) confirmed regional portal hypertension, liver cirrhosis, intrahepatic bile duct dilation of the right lobe with calculus formation, and common bile duct dilation. PV trunk dilated remarkably, and distal branches of the PV were not seen clearly. The hepatic artery appeared normal. The coronal CT image confirmed the presence of a side-to-side shunt between the dilated splenic vein and the left renal vein, which was observed in the Doppler examination as a splenic-caval shunt. 3D CT reconstruction showed multiple cystic aneurismal dilation of the splenic artery and left renal artery, and vascular disorders of the splenic hilum. The splenic and superior mesenteric veins had normal orientation and these vessels joined to form the normal main PV.

Given these radiological findings, a diagnosis of bile duct infection on the background of Caroli's disease with calculus formation, liver cirrhosis, congenital partial absence of PV with a portocaval shunt (Abernethy malformation of type II), regional portal hypertension, and multiple aneurismal cystic dilation of the splenic artery and left renal artery was made.

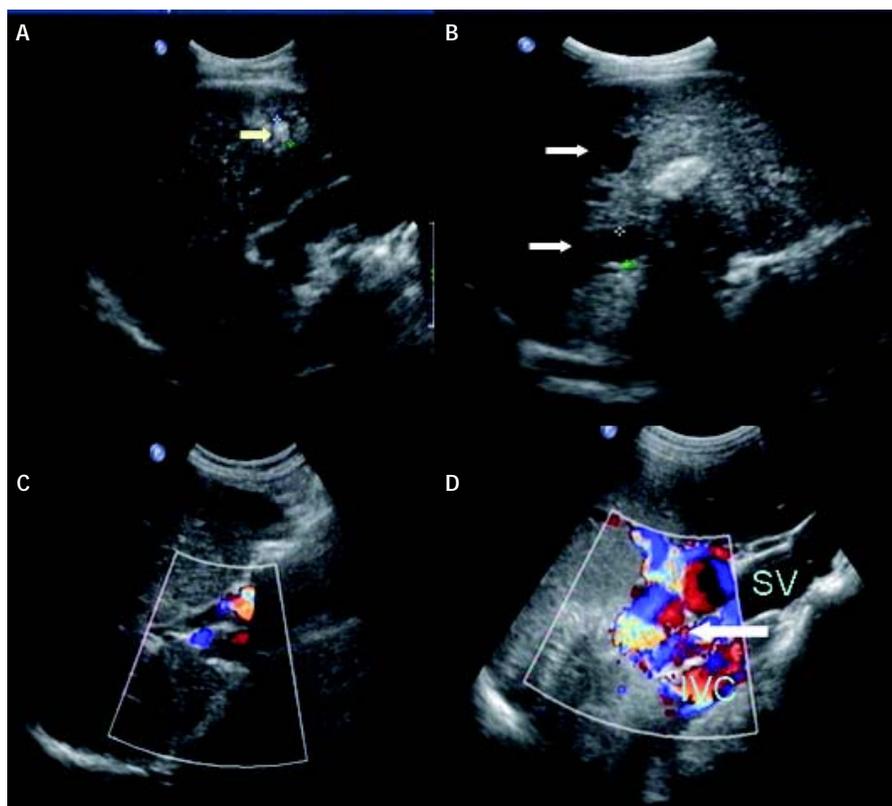


Figure 1. Abdominal two-dimensional ultrasound and colored Doppler ultrasound image. **A.** Shows the heterogenous change of the liver and a nodule (arrow) in the segment 5. **B.** Shows dilated intrahepatic bile duct (arrow) of the right liver lobe. **C.** Shows sparse intrahepatic PV blood flow (red point) and increased hepatic arterial blood flow (yellow point). **D.** Shows a communication (arrow) between the dilated splenic vein (SV) and the inferior vena cava (IVC).

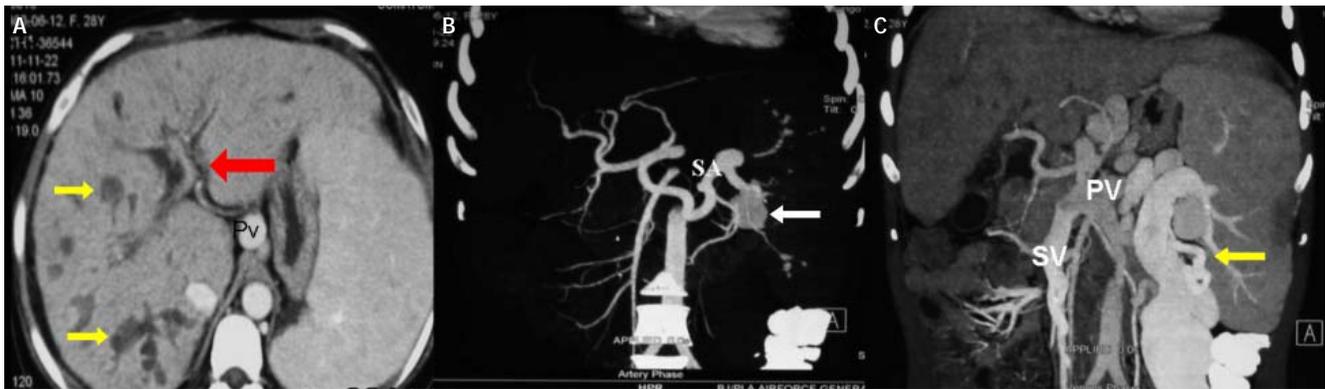


Figure 2. Axial, coronal and three-dimensional CT image. **A.** Shows that the liver and spleen increase obviously, intrahepatic bile ducts of the right lobe dilate with calculus formation (white arrow), and PV trunk (PV) dilates remarkably. The hepatic artery branches normally (red arrow). **B.** Shows the cystic aneurysmal dilation (arrow) of the splenic artery (SA), which exists to the left renal hilum. **C.** Shows the collateral vessel of side-to-side shunt (arrow) between the splenic vein (PV) and the left renal vein, which connects to the inferior vena cava. The vascular disorders of the splenic hilum and the normal form of the superior mesenteric vein (SV) can be seen.

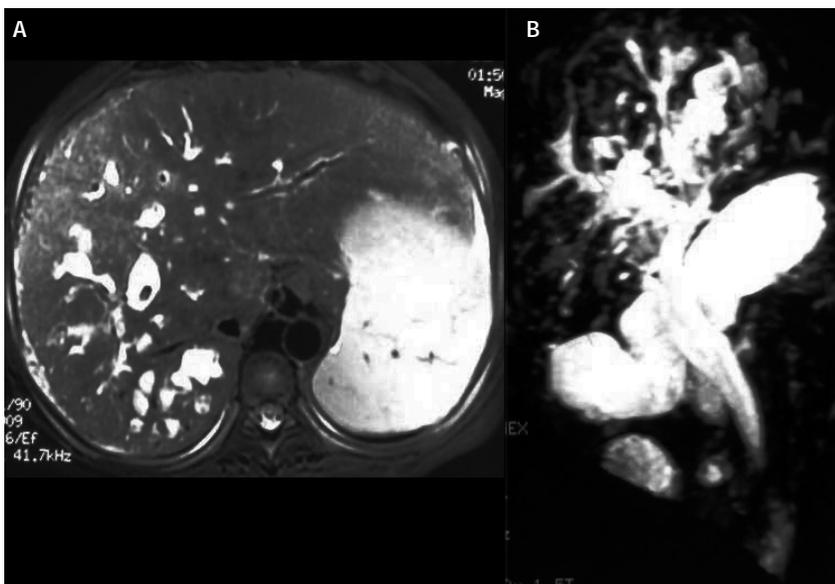


Figure 3. Axial (A) and MRCP (B) image of the liver MRI. **A.** Shows the extensive intrahepatic bile duct dilation of the right lobe and extensive hepatic fibrosis. **B.** Shows the intrahepatic and extrahepatic bile duct system dilation.

The results of upper gastrointestinal endoscopy were normal and esophageal varices were not found. Echocardiography didn't reveal any associated cardiac malformation. Radiographic inspection and renal function test showed no evidence of an associated autosomal recessive polycystic kidney disease. Because of the abnormal level of CA19-9 and the nodular changes of liver found in ultrasound, magnetic resonance imaging (MRI) was performed to determine whether malignant tumors were present on the liver. The findings confirmed the presence of extensive intrahepatic cystic dilatation of bile duct (mainly in the right hepatic lobe), extensive hepatic fibrosis and common bile duct dilation, and eli-

minate the presence of hepatic malignant tumors (Figure 3).

In consideration of the existence of liver cirrhosis and portal hypertension, and her medical history of abdominal operation, surgical therapy wasn't performed. The patient was maintained on conservative therapy, and the symptoms of abdominal pain and fever relieved after 7 days. At 1-month follow-up, she was asymptomatic and there was no change on ultrasonography. The possibilities of hepatic encephalopathy and liver function failure to develop in the future have been told to the patient. The patient will consider the surgical mode to choose including liver transplantation at that time.

DISCUSSION

Congenital extrahepatic portosystemic shunts are a rare congenital anomaly. Since Abernethy described this anomaly in 1793, only 134 cases have been reported in medical literature to January 2012, and most of them were reported in these 10 years because of advances in imaging techniques.⁴ To the best of our knowledge, only 37 cases of type II Abernethy malformation have been reported.⁵ None of the cases of Abernethy malformation reported in the literature had associated multiple aneurisms and Caroli's disease.

The portal vein is normally formed in 4-10-week-old embryos by the selective involution of the per-intestinal vitelline venous loop. The left umbilical vein and the right and left vitelline veins flow into the liver bud, and within the liver bud these veins form the hepatic sinusoid and the ductus venosus, respectively. Blood flow through these veins then drains into the suprahepatic portion of the IVC. Next, the proximal and distal ends of the left vitelline vein disappear, and the remaining right vitelline vein mainly forms the portal vein. During this step, the left umbilical vein and the portal vein supply blood to the liver bud. The abnormal development of these vessels during this stage causes Abernethy malformation.⁶

Abernethy malformation can be anatomically classified into two groups. Type I is characterized by the absence of an intrahepatic portal venous supply and complete end-to-side portocaval shunt, while type II is characterized by the presence of a patent intrahepatic portal venous supply and a partial side-to-side shunt.⁷ Type I can be further subclassified into type Ia and type Ib, based on whether or not the splenic vein (SV) and the superior mesenteric vein (SMV) drain together to form a common trunk.⁸ This patient had a type II Abernethy malformation with a side-to-side portocaval shunt between the splenic vein and left renal vein.

Congenital extrahepatic portosystemic shunts are frequently associated with other congenital anomalies in cardiovascular system, brain, skeleton and genito-urinary tract,⁹⁻¹¹ but these congenital anomalies are much more common with type I than with type II, and multiple cystic aneurismal dilation of the splenic artery and left renal artery have never been reported with Abernethy malformation. Although left sided IVC, polysplenia, two splenic veins, congenital choledochal cyst and biliary atresia have been reported in patients of type I Abernethy malformation,¹² congenital hepatobiliary

anomaly of Caroli's disease has never been reported, especially in patients of type II.

Because there was an increased frequency of malignant hepatic neoplasms with patients of cirrhosis caused by extrahepatic portosystemic shunts,¹³ we choose MRI to further make sure the hepatic nodules' nature of the patient. It has also been suggested that the disequilibrium of hepatic artery and PV may provide an environment for the development for neoplastic tumor in Abernethy malformation patients,⁶ so regular follow-up is necessary for this patient.

A diagnosis of Abernethy malformation can now be made by noninvasive cross-sectional imaging technologies, such as ultrasound, CT, and MRI. For patients with suspected extrahepatic portosystemic shunts, three-dimensional reconstruction of the vessels can be very helpful.¹⁴ In this case, we used 3D CT reconstruction of the vessels to show the side-to-side portosystemic shunt, as well as multiple cystic aneurismal dilation of the splenic artery and left renal artery. Although congenital factors can be the primary reason of the aneurysms, the probabilities of blood vessel remodeling connected with liver cirrhosis can't be excluded. MRI showed extensive hepatic fibrosis of the patient, which maybe caused by recurrent intrahepatic infection and chronic liver injury and promoted the formation of liver cirrhosis. Evidence of congenital hepatic fibrosis wasn't clear according to the patient's medical history.

The treatment planning for Abernethy malformation depends on the type of shunts and the associated conditions. In patients with type I, liver transplantation is the only effective method for those who develop severe hepatic encephalopathy or malignant tumors, because occlusion of the shunt will obstruct the only drainage route of the mesenteric venous blood.¹⁵ In patients with type II, shunt occlusion can be performed in case of serious symptoms such as hepatic encephalopathy, and intrahepatic PV blood flow should be evaluated by angiography before the operation.¹⁶ For this patient, Abernethy malformation of type II was incidentally diagnosed and bile tract infection symptoms were the main clinical manifestation. Conservative management was chosen, due to:

1. Congenital intrahepatic bile duct dilation couldn't be treated by partial hepatectomy because of the existence of serious liver cirrhosis.
2. Bile duct exploration and drainage couldn't be performed because regional portal hypertension may lead to massive hemorrhage.

3. There was no clear surgical indication for portal hypertension because the patient's had no medical history of gastrointestinal bleeding.
4. It isn't necessary to deal with aneurysms and extrahepatic portosystemic shunts because there were no serious symptoms associated with them. In fact, liver transplantation would be the only effective treatment option. The patient was informed about the future possibility of a liver transplantation and the need for regular follow-up to monitor the hepatic masses.

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