Abernethy malformation: Congenital absence of the portal vein

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CASE PRESENTATION

A 20-year-old previously well man presented with unintentional 6.75 kg (15 lb) weight loss over a six-month period, vague abdominal discomfort and bilateral patchy rash. Blood work was notable for elevations in alanine aminotransferase (75 U/L [normal range <41 U/L]), aspartate aminotransferase (101 U/L [<40 U/L]), alkaline phosphatase (310 U/L [40 U/L to 129 U/L]) and total bilirubin (15.7 µmol/L [3.4 µmol/L to 17 µmol/L]) levels. An abdominal ultrasound revealed hepatic nodules.

On further characterization, magnetic resonance imaging revealed a portosystemic shunt (PSS) between the main portal vein and inferior vena cava (IVC) (Figure 1). Both vessels were dilated, with no definite intrahepatic portal venous branches identified (Figure 2). Multiple large regenerative nodules could also be identified. Hepatic portal venous Doppler confirmed an extrahepatic PSS, with a markedly distended infrahepatic IVC (Figure 3). The confluence of the splenic vein and superior mesenteric vein drained directly into the IVC. The morphology suggested a type Ib Abernethy malformation.

DISCUSSION

First reported in 1793 (1), Abernethy malformations are congenital extrahepatic PSS (CEPS). Only 80 cases have been described since the initial discovery, with most patients <18 years of age (2,3). Two types are classified based on the presence of the portal vein and its anastomosis with the IVC. Type I malformations have a congenital

absence of the portal vein and are predominantly found in females (74%) (4-7). Type II malformations have a hypoplastic portal vein leading to liver perfusion via a partial PSS.

CEPS have variable clinical presentations. Patients may be asymptomatic, or may present with nonspecific symptoms such as acute hepatic decompensation or cirrhosis. Importantly, CEPS are frequently associated with other congenital defects, especially type I, with cardiovascular abnormalities being highly associated (4-6,8,9). Atrial septal defect, patent foramen ovale, ventricular septal defect, patent ductus arteriosus, tetralogy of Fallot and dextrocardia can frequently be observed in CEPS (4-6,9). Associated gastrointestinal abnormalities include polysplenia, biliary and duodenal atresia, choledochal cyst and intrahepatic gallbladder. There are also associated anomalies in the genitourinary, skeletal and vascular systems (5,6,10). Except for malformations in the genitourinary system, which can be equally appreciated with type I and type II, most associated abnormalities are more common in type I CEPS (6,10).

Early recognition of PSS is important. PSS increases the risk of hepatic neoplasms including benign focal nodular hyperplasia, hepatocellular adenoma and regenerative nodules. Almost 50% of patients with CEPS have been found to have nodular liver lesions (7,11). Type I Abernethy malformations are associated with hepatocellular carcinoma and hepatoblastoma (3,5,8,11). Approximately 15% of CEPS cases result in hepatic encephalopathy (6). The significance of continued

Figure 1) Contrast-enhanced coronal magnetic resonance image of the abdomen revealing an abnormal shunt between the main portal vein and inferior vena cava (arrow)

Figure 2) Contrast-enhanced axial magnetic resonance image of the abdomen showing a dilated main portal vein and inferior vena cava (arrow), with no definite intrahepatic portal venous branches identified

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Figure 3) Doppler ultrasound demonstrated an extrahepatic portosystemic shunt, with a markedly distended infrahepatic inferior vena cava (arrow) and the confluence of the splenic vein and superior mesenteric vein draining directly into the inferior vena cava

monitoring and long-term follow-up is clear. There are no current published guidelines for follow-up of CEPS. Given the aforementioned risks, however, our recommendations are routine clinical assessments, biannual blood work (liver enzyme and liver function tests) and yearly imaging of the liver.

The type of PSS is particularly important in determining treatment. Type I shunts merit clinical, biochemical and imaging follow-up for an increased risk of malignancy and development of hepatic encephalopathy; their long-term treatment option may be limited to liver transplant. Type II malformation shunts can be occluded either surgically or via percutaneous transcatheter coil placement (12).

With modern advances in medical imaging, there has been an increase in the diagnosis of CEPS. Early noninvasive cross-sectional images via Doppler ultrasound, computed tomography or magnetic resonance imaging is key to providing an accurate diagnosis and classification, further directing the therapeutic course and clinical outcome (10).

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