

Abernethy malformation with portal vein aneurysm

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ABSTRACT

We present the case of a 24-year-old man who was incidentally diagnosed with congenital extrahepatic portosystemic shunt with portal vein aneurysm during an investigation for non-specific abdominal pain. These are rare anomalies, and to the best of our knowledge, this is the first case reported with both anomalies associated together. Ultrasound, including color Doppler, computed tomography, and magnetic resonance imaging were performed which revealed a side-to-side shunt between the extrahepatic portal vein and the inferior vena cava, with aneurysmal fusiform dilatation of the proximal intrahepatic portal vein which ended abruptly. Etiology, clinical significance, and management strategies with regard to these abnormalities are discussed.

Key words: • Abernethy malformation • portal vein aneurysm • computed tomography

The Abernethy malformation (congenital extrahepatic portosystemic shunt [CEPS]) is a rare condition. It 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. These types of Abernethy malformation are the following: type 1, the entire portal venous supply drains into the IVC with absence of the intrahepatic PV; and type 2, the portal venous blood partially drains into the IVC through side-to-side anastomosis (1). Patients with Abernethy malformation almost always have other associated anomalies such as liver and cardiac abnormalities. We present the case of a 24-year-old man who was incidentally diagnosed with congenital extrahepatic portosystemic shunt along with PV aneurysm. The latter association has not been previously described in the literature.

Case report

A 24-year-old man presented at our institution with complaints of abdominal pain associated with nausea and vomiting for 5 days. There was no history of fever or jaundice. His medical history was unremarkable. Physical examination revealed pallor, digital clubbing, and mild splenomegaly, but no icterus. Physical examination was unremarkable and there was no evidence of encephalopathy. Laboratory tests showed a hemoglobin level of 7.0 g/dL, erythrocyte sedimentation rate of 30 mm in the first hour, and normal liver function tests. Abdominal ultrasound revealed coarse heterogenous echotexture of the liver with an ill-defined large hypoechoic lesion in segments 7 and 8. The PV showed fusiform dilatation, measuring 25 mm at the porta, and it continued into a dilated intrahepatic portion which ended abruptly while coursing towards the left lobe. A small branch coursed towards the right lobe and probably represented the attenuated right branch of the PV. There was a side-to-side communication between the extrahepatic part of the PV and IVC. The splenic and superior mesenteric veins were prominent. Doppler examination (Fig. 1) showed hepatopedal flow in the PV, and flow towards the IVC in the portocaval shunt.

Dual-phase computed tomography (CT) (Fig. 2) confirmed the presence of a side-to-side shunt between the extrahepatic PV and the IVC, fusiform aneurysmal dilatation of the PV continuing into the left branch, which abruptly ended after a short distance, and an attenuated right branch of the PV. Distal branches of the PV were not seen. The mass lesion in the right lobe of the liver observed on ultrasound appeared as an ill-defined area of hypodensity on both unenhanced and enhanced images (arterial, portal venous, and delayed phases). Tributaries of the hepatic vein were seen to course through the lesion. The

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Received 20 January 2007; revision requested 6 March 2007; revision received 27 April 2007; accepted 7 May 2007.

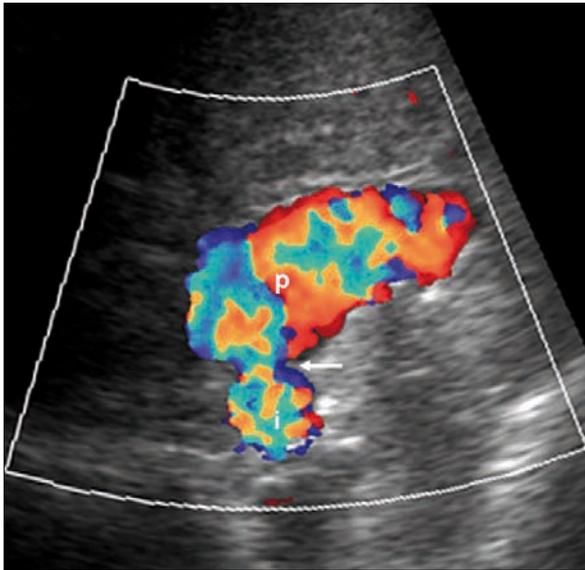


Figure 1. Doppler ultrasound image shows a communication (arrow) between the aneurysmal extrahepatic portion of the portal vein (p) and the inferior vena cava (i).

hepatic artery appeared normal. The splenic and superior mesenteric veins had normal orientation and these vessels joined to form the main PV.

Based on these findings, a diagnosis of congenital partial absence of PV with a portocaval shunt (Abernethy malformation type 2) associated with PV aneurysm and an area of intrahepatic perfusion alteration was made.

The results of upper gastrointestinal endoscopy were normal and serum alpha-fetoprotein and ceruloplasmin levels were normal. Echocardiography did not reveal any associated cardiac malformation. Dual-phase gadolinium-enhanced magnetic resonance imaging (MRI) was performed to further characterize the abnormalities. The findings confirmed the presence of the portocaval shunt (Fig. 3a) and PV aneurysm. The right lobe lesion was mildly hypointense compared with the liver on T1-weighted images and was poorly visualized on T2-weighted images being isointense compared with the liver. The scans showed no abnormal enhancement in arterial, venous, or delayed phases (Fig. 3b). T1-weighted MRI of the brain (Fig. 3c) revealed hyperintense signal in bilateral globus pallidus with generalized cerebral atrophy, suggesting portosystemic encephalopathy.

The patient was maintained on conservative management. At 6-month follow-up he was asymptomatic and

there was no change in the liver lesion or aneurysm on ultrasonography. In the event that hepatic encephalopathy develops in the future, surgical closure of the shunt will be considered at that time.

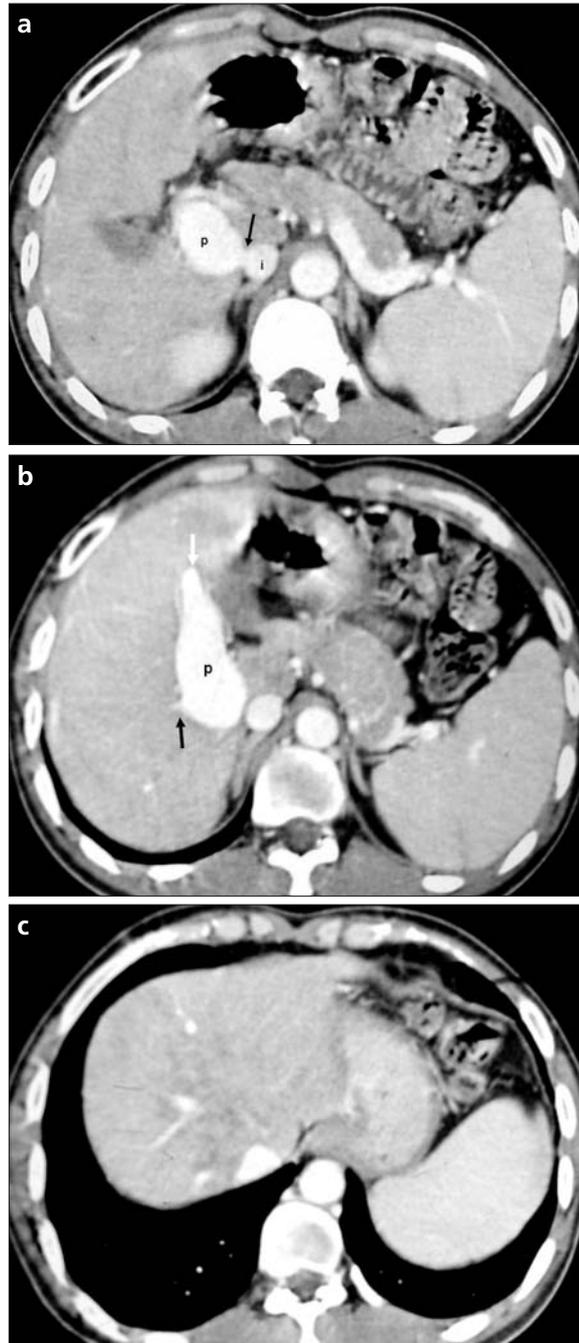


Figure 2. a–c. Axial contrast-enhanced CT image (a) shows the side-to-side shunt (arrow) between the aneurysmal extrahepatic portion of the portal vein (p) and the inferior vena cava (i). Axial contrast-enhanced CT image at a level slightly cranial to Fig. 2a (b) shows the aneurysmal portal vein (p) mainly continuing as the left branch which is getting tapered and ending abruptly (white arrow). Note the highly attenuated right portal vein (black arrow). Axial contrast-enhanced CT scan at a level cranial to Fig. 2b (c) shows ill-defined hypodense lesion in segments 7 and 8. The hepatic veins are seen to course through the lesion.

Discussion

Congenital extrahepatic portosystemic shunt is a rare congenital anomaly that was first described by Abernethy in 1793 at autopsy of a 10-month-old infant who died of unknown cause (2).

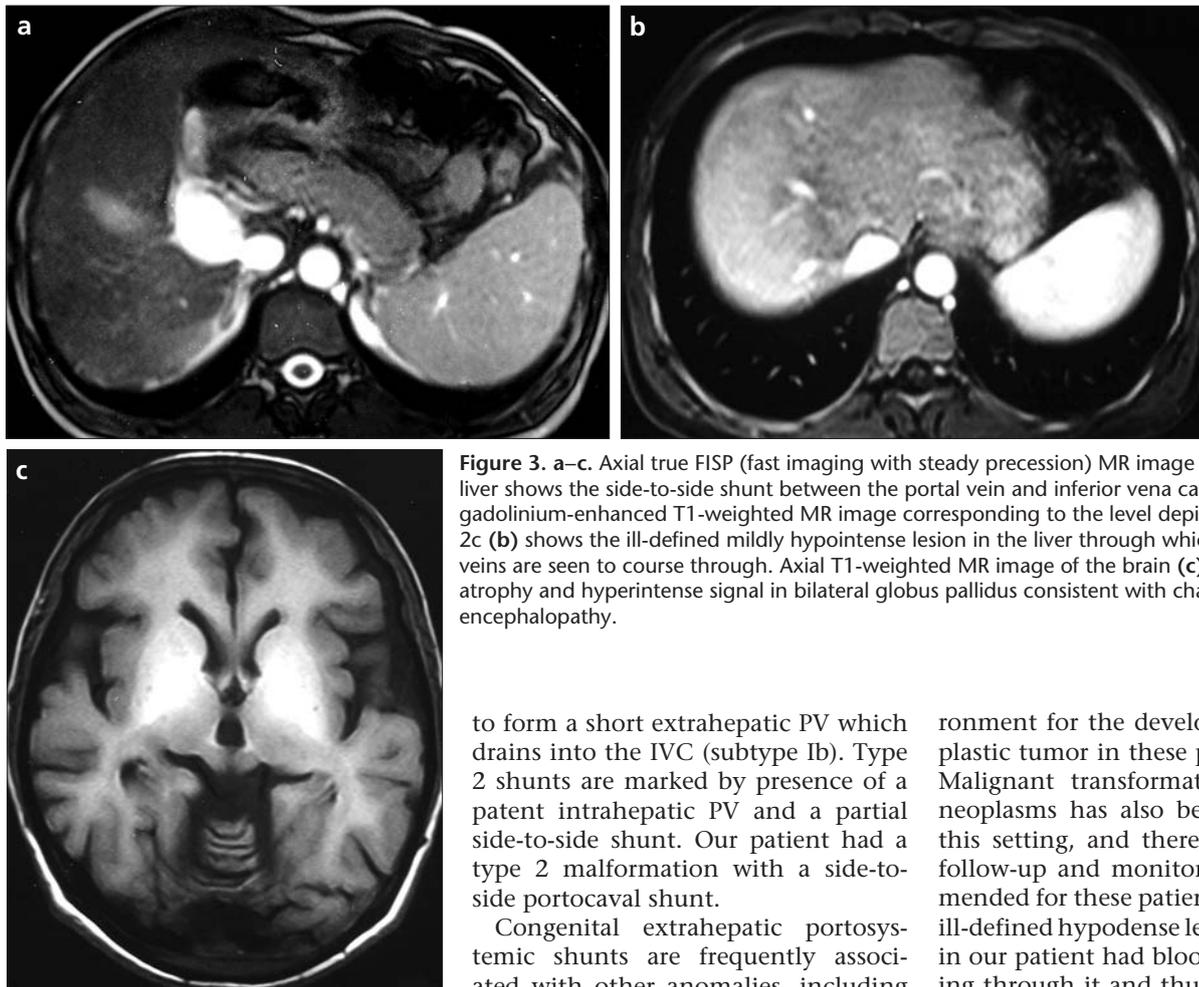


Figure 3. a–c. Axial true FISP (fast imaging with steady precession) MR image (a) of the liver shows the side-to-side shunt between the portal vein and inferior vena cava. Axial gadolinium-enhanced T1-weighted MR image corresponding to the level depicted in Fig. 2c (b) shows the ill-defined mildly hypointense lesion in the liver through which the hepatic veins are seen to course through. Axial T1-weighted MR image of the brain (c) shows brain atrophy and hyperintense signal in bilateral globus pallidus consistent with changes of hepatic encephalopathy.

With advances in imaging techniques, cases with this malformation are being identified more frequently. To the best of our knowledge, only 22 cases of type 2 Abernethy malformation have been reported to date (3). None of the cases of Abernethy malformation reported in the literature had an associated PV aneurysm.

The portal venous system develops between the fourth and tenth weeks of embryonic life by selective apoptosis of some portions of the vitelline veins. The IVC also develops at this time, which leads to the potential for congenital portosystemic shunts (3).

Portosystemic shunt anomalies have been classified into two types (4). Type 1 shunts are characterized by the absence of the intrahepatic PV and complete end-to-side shunt, and have two subtypes, (i) separate drainage of the superior mesenteric and splenic veins into the IVC, iliac veins, or renal veins (subtype 1a) and (ii) superior mesenteric and splenic veins joining

to form a short extrahepatic PV which drains into the IVC (subtype 1b). Type 2 shunts are marked by presence of a patent intrahepatic PV and a partial side-to-side shunt. Our patient had a type 2 malformation with a side-to-side portocaval shunt.

Congenital extrahepatic portosystemic shunts are frequently associated with other anomalies, including congenital heart disease, polysplenia, biliary atresia, malrotation, duodenal atresia, annular pancreas, situs inversus, anomalies of the renal tract, and skeletal anomalies. These shunts are more common with type 1 than with type 2 malformations.

Congenital extrahepatic portosystemic shunts are also associated with an increased frequency of hepatic neoplasms, which may be benign (focal nodular hyperplasia, hepatocellular adenoma, or nodular regenerative hyperplasia) or malignant (hepatocellular carcinoma or hepatoblastoma) (3, 5–8). It has been proposed that the diversion of hepatotrophic substances in the splanchnic venous blood, such as insulin and glucagon, away from the liver results in alterations of development, function, and regenerative capacity of the liver. This diversion, along with increased arterial hepatic flow, may also contribute to the development of hepatic neoplasms (5). It has been suggested that the disequilibrium of hepatic circulation between the hepatic artery and PV may provide an envi-

ronment for the development of neoplastic tumor in these patients (9, 10). Malignant transformation of benign neoplasms has also been reported in this setting, and therefore, long-term follow-up and monitoring are recommended for these patients (11, 12). The ill-defined hypodense lesion in the liver in our patient had blood vessels coursing through it and thus it represented a perfusion anomaly rather than a neoplasm. Also, the lesion remained unchanged on 6-month follow-up with ultrasound. However, we have advised our patient to undergo regular follow-up at 6-month intervals.

Patients with Abernethy malformation may have some other manifestations, such as diversion of gut-derived toxins to the systemic circulation leading to hepatic encephalopathy or diversion of vasoactive mediators into the systemic circulation leading to dilatation of intrapulmonary vessels and hepatopulmonary syndrome (6). Our patient had MRI features consistent with portosystemic encephalopathy although there was no clinical evidence of this complication. Digital clubbing was also seen in our patient. Digital clubbing has been described in various hepatic diseases including biliary atresia, sclerosing cholangitis, cirrhosis, Wilson's disease, and tumors such as epithelioid hemangioendothelioma. Because vasoactive and angiogenic factors originating from the liver normally control the pulmonary circula-

tion, liver disease and portosystemic shunts result in hypoxemia due to ventilation perfusion mismatch (13). This hypoxemia leads to neoangiogenesis and vascular hyperplasia which in turns leads to digital clubbing. It has also been postulated that damaged liver produces or fails to metabolize substances responsible for the local changes in bone metabolism in these patients (14).

Currently, a diagnosis of Abernethy malformation is usually made by non-invasive cross-sectional imaging techniques such as ultrasound, CT, or MRI, which show the shunt and any intrahepatic PV branches. However, liver biopsy may be necessary in patients with suspected type 1 malformation since an occasional patient may have small PV radicles which cannot be seen on ultrasound but can be observed on liver biopsy (15, 16).

PV aneurysm is a focal fusiform or saccular dilatation of the portal venous system. A segmental dilatation of more than 20 mm is considered to be an aneurysm (17). It can be congenital or acquired. The acquired causes of PV aneurysm include chronic liver disease, portal hypertension, trauma, liver biopsy, pancreatitis, and tumor invasion of the PV. If none of these acquired causes are identified the aneurysm can be considered as congenital (17, 18). To date, only 25 cases of congenital PV aneurysm have been described (17). There has been no report in the literature of a PV aneurysm associated with Abernethy malformation.

Determining the type of shunt is particularly important in the planning of treatment. In patients with type I malformation, occlusion of the shunt is not an option since it represents the only drainage route for the mesenteric

venous blood. Hence, these patients merit clinical, biochemical, and imaging follow-up; for those who develop severe hepatic encephalopathy or malignant liver nodules, liver transplantation is the only treatment option (3, 15). For patients with type 2 malformations and serious symptoms such as hepatic encephalopathy, shunt occlusion can be performed, either surgically or by percutaneous transcatheter coil placement (15).

PV aneurysm management depends on the size of the aneurysm, symptoms, complications, and the clinical condition of the patient. Conservative management with regular follow-up of aneurysm size and symptoms is usually required in most cases. Surgical treatment should be considered if the aneurysm increases in size or there is thrombus formation (17).

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