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Case Report

A rare Abernethy Ib malformation was initially misdiagnosed as chronic portal vein thrombosis in a 27-year-old female [☆]

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ABSTRACT

Abernethy malformation or congenital portosystemic shunt is a rare congenital vascular malformation and anomaly of the splanchnic venous system defined by diverting portal blood away from the liver. It is commonly associated with multiple congenital anomalies. Imaging modalities such as computed tomography or magnetic resonance have a crucial role in prompting diagnosis and determining the prognosis based on the type of malformation and associated anomalies. Misdiagnosis could be harmful and may lead to inappropriate treatment. We present a case of Abernethy malformation with a complete end-to-side shunt of portal venous flow into the systemic venous flow and complete bypass of the liver, which was initially misdiagnosed with portal venous thrombosis.

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Introduction

John Abernethy first reported the congenital absence of portal vein and congenital mesentericocaval shunt in 1793 [1]. Abernethy malformation, also known as Congenital extrahepatic portosystemic shunt (CEPS), is a rare clinical entity and is a condition in which portal blood is shunted partially or entirely into the systemic circulation via an abnormal communication of the portal system with the systemic circulation. Abernethy malformation can be classified into 2 types. In Type Ia, splenic

and superior mesenteric veins (SMV) drain separately into the inferior vena cava, and in Type Ib, draining occurs via a common trunk. In Type II, blood is shunted from the portal vein to the inferior vena cava [2]. The clinical presentation of these patients is varied, and these shunts are often unsuspected and picked up either on ultrasound or computed tomography/magnetic resonance imaging (CT/MRI) for evaluation of the varied symptomatology they cause, which includes jaundice, difficulty in breathing, cyanosis, clubbing, and abdominal mass. In ultrasound, failure to image the portal vein, which is not due to overlying bowel gas or decreased sonographic beam

Abbreviations: CEPS, Congenital extrahepatic portosystemic shunt; SMV, Superior mesenteric vein; IVC, Inferior vena cava; MRI, Magnetic resonance imaging.

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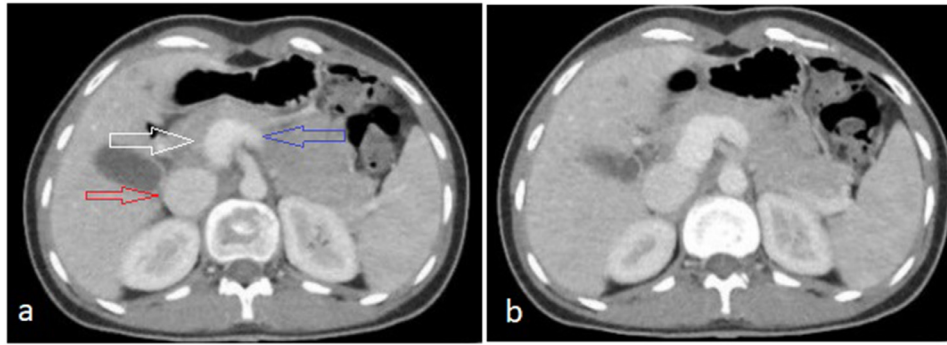


Figure 1 – Axial CT image of the upper abdomen in the portal venous phase shows splenic (blue arrow) and superior mesenteric (white arrow) veins joining to form a common trunk, which drains into IVC (red arrow).

penetration through severe fatty or cirrhotic liver, should raise the question of portal vein thrombosis [3]. We describe our experience with a 27-year-old woman with Abernethy malformation Ib, who was initially misdiagnosed by chronic portal vein thrombosis based on reported ultrasound, which was not confirmed by other imaging modalities.

Case presentation

A 27-year-old female presented to the emergency room with extrapyramidal symptoms and nonspecific vague abdominal discomfort. A physical exam reveals no abnormalities except jaundice. Blood work was notable for elevations in total bilirubin level (2.72 mg/dl, with a normal range of 0.3-1.2), direct and indirect bilirubin levels (0.86 and 1.86 mg/dl, with a normal range of <0.4 and 0.1-0.8 respectively), and decrease ceruloplasmin level (0.17 gr/l with normal range of 0.2-0.4), so for R/O of Wilson disease, the patient referred to GI clinic for complete work-up. Further evaluation was performed by abdominal sonography in the outpatient center, which revealed no abnormality except narrowed and echogenic portal vein compatible with chronic portal vein thrombosis.

The patient underwent a multiphasic abdominal CT scan with noncontrast, arterial, Porto venous, and delayed phases in our center. The liver shows normal parenchymal density and enhancement with an absent intrahepatic portal vein. Mesenteric and splenic veins form a short common trunk that directly drains into prominent IVC, with a complete end-to-side shunt of portal venous flow into the systemic venous flow, which causes complete bypass of liver and compatible with congenital extrahepatic portosystemic shunt, or Abernethy malformation type Ib (Figs. 1-3). Also, liver dynamic MRI was performed to evaluate liver parenchyma better and rule out possible liver nodules, which are more frequent in these patients. Liver MRI shows no abnormal signal intensity in liver parenchyma and confirmed the abnormal extrahepatic portosystemic shunt (Figs. 4 and 5).

The other work-up included urine copper (16ug/dl with normal range < 60), blood copper (74ug/dl with normal range 75-155), and repeated ceruloplasmin (0.37 gr/l with normal range 0.2-0.4) were in a normal range, and eye exam reveals

no Kayser-Fleischer rings. Also, brain MRI was performed, and some nonspecific high T2 signal foci were observed in both superficial and periventricular white matter. No specific findings were detected in basal ganglia, brain stem, and thalamus (Fig. 6), so the Wilson disease was excluded and diagnosed with Abernethy malformation type Ib, and the patient was referred to the Organ-Transplant committee for a liver transplant.

Discussion

Abernethy malformation is a congenital extrahepatic portosystemic shunt (CEPS) that develops between the Portomesenteric vasculature and the systemic veins, which was first reported in 1793 by John Abernethy [1]. The pathogenesis is linked to the complexity of the embryological development of the inferior vena cava and portal vein [4]. Risk factors have not been well-defined. Shunts may be associated with other congenital abnormalities but are also found incidentally as isolated aberrancies.

The portosystemic shunt disrupts the enterohepatic circulation with deranged metabolism of various substances, leading to adverse clinical manifestations. Presentation ranges from an incidental finding to hepatic encephalopathy and liver failure, depending on the type of abnormality. The portosystemic shunt ratio may predict progression to hepatic encephalopathy and cirrhosis. Decompensation may be precipitated by concurrent illness. Patients may be asymptomatic or present with nonspecific symptoms such as hypergalactosemia, hyperbilirubinemia, and hyperammonemia due to delayed hepatic metabolism of these metabolites as they bypass the liver in the first instance [5,6]. Pulmonary venous congestion results in hepatopulmonary syndrome, presenting clinically with dyspnea due to pulmonary hypertension and even syncope [7]. Hepatic encephalopathy may develop in longstanding cases, clinically manifesting with tremors, extrapyramidal symptoms, irritability, and altered sensorium. Regenerative nodular hyperplasia of the liver can also result from the liver's abnormal response to the absent portal flow and can progress to a hepatic tumor in the form of adenoma, hepatoblastoma, or hepatocellular carcinoma. Associated car-

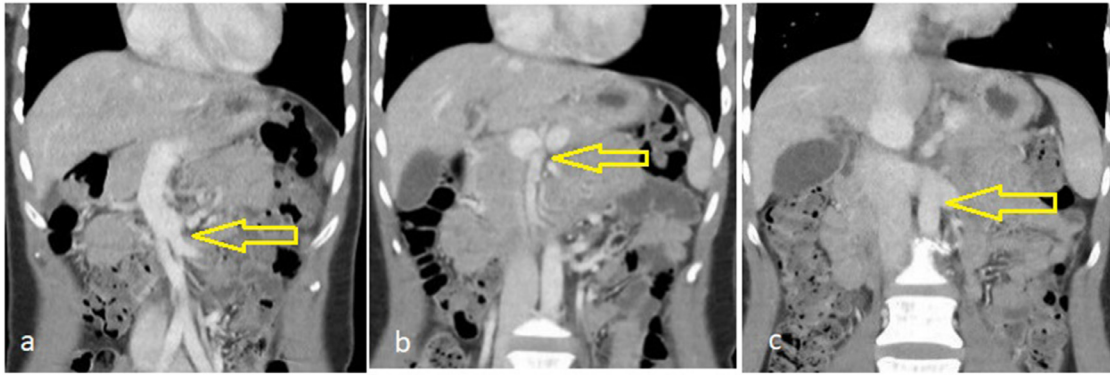


Figure 2 – Coronal reformatted CT image of the abdomen in the portal venous phase shows congenital extrahepatic shunt between the IVC and mesenteric and splenic veins common trunk (yellow arrows in a to c).



Figure 3 – Sagittal reformatted MDCT image demonstrates the extrahepatic portosystemic shunt between the IVC and extrahepatic portal vein (black arrow), bypassing the liver, and an absent portal vein is seen in the liver hilum.

diac and skeletal anomalies, biliary atresia, polysplenia, and hepatic tumors often coexist [8]. Prognosis depends on the presence and extent of associated cardiac or hepatic complications, so early diagnosis is essential.

Imaging plays an integral part in diagnosing these congenital extrahepatic portosystemic shunts. The first step in diagnosing the CEPS is to demonstrate communication between the portal and the systemic venous system. These may be end-to-side shunts where the PV terminates in the IVC or side-to-side shunts between the PV and IVC. The second step entails ruling out the acquired causes of nonvisualization of the PV, such as portal cavernoma or PV thrombus. An acute PV thrombus is seen on cross-sectional imaging as a hypodense, nonenhancing PV filled with thrombus. Long-standing portal thrombosis may result in empty porta hepatis. Webb et al. [9] first reported a sonographic demonstration of portal vein thrombosis. They described a characteristic empty porta hepatis in a complete portal vein block, appearing as a broad, diamond-shaped band of high-level echoes. As a rule, patients with congenital portosystemic shunts do not have features of portal hypertension, such as splenomegaly, varices, and collaterals [10].

Doppler ultrasonography is a safe and noninvasive modality for diagnosing the intrahepatic vasculature and flow direction and may demonstrate the shunt. It may pick up congenital shunts preoperatively; however, it may not detect associated anomalies and may also be unable to evaluate the retroperitoneum well, particularly in adult patients. Although color Doppler sonography first depicted the image of the absent portal vein in most cases, ultrasonography may fail to accurately detect the associated extrahepatic shunts because of

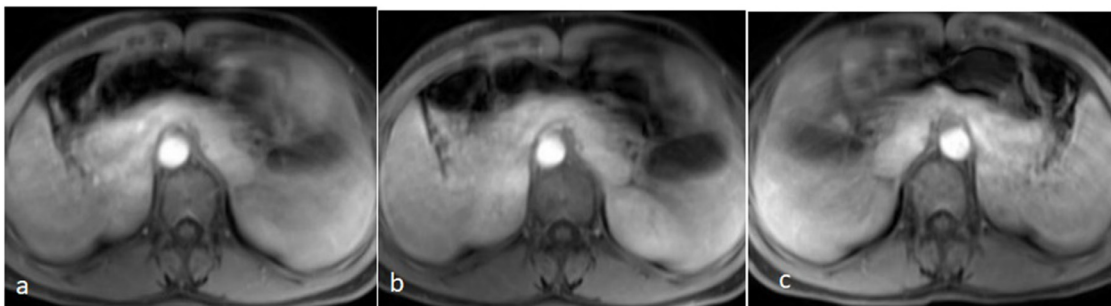


Figure 4 – Contrast-enhanced axial magnetic resonance image of the abdomen showing a dilated inferior vena cava and congenital extrahepatic shunt between the IVC and mesenteric and splenic veins common trunk.

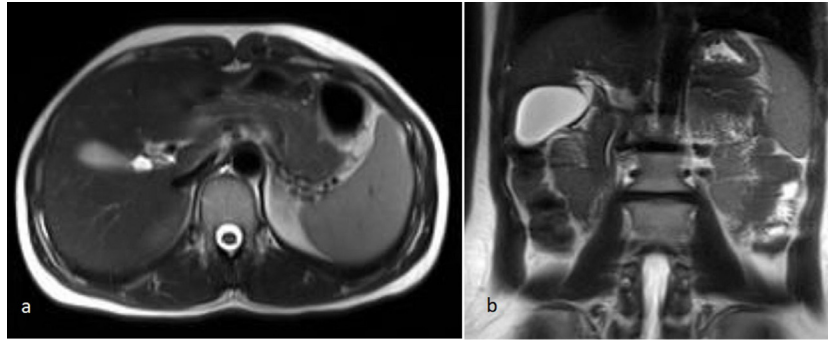


Figure 5 – T2 weighted hepatic MRI (axial in a, coronal in b) shows an absent portal vein in the liver hilum.

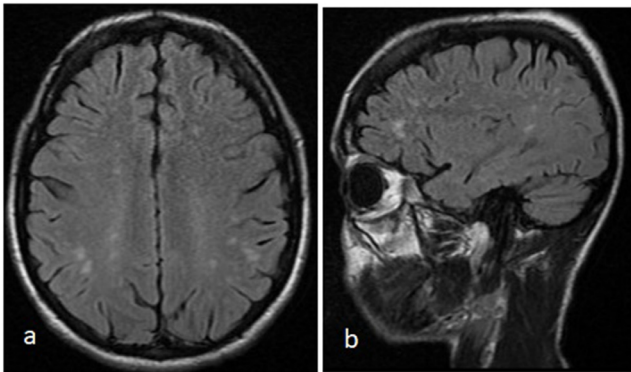


Figure 6 – Brain MRI shows nonspecific high signal intensity foci in both superficial and periventricular white matter at Flair and T2 weighted sequences.

its subtle US features. Experience and good ultrasound system, knowledge of the examination protocol, and familiarity with the ultrasound anatomy of abdominal vessels and portal vein abnormalities contribute to the accurate diagnosis. Ultrasound may not fully characterize liver lesions seen in these patients [11].

Multidetector CT (MDCT) is a fast and effective modality for the evaluation of patients with suspected or confirmed portocaval shunts; it displays all the information desired by the surgeon and the clinician, including the anatomy of the splenic and superior mesenteric veins (SMV), size and site of the shunt, presence or absence of the portal vein (PV) radicles. It helps to plan the therapy and even the follow-up of these patients. Contrast-enhanced magnetic resonance imaging (MRI) may provide similar information, and though CT has the advantage in speed and spatial resolution; however, MRI scores in the characterization of liver lesions and patients who need long-term follow-up [10].

Diagnosing CEPS requires exclusion of acquired shunts due to cirrhosis or portal-vein thrombosis. Acquired shunts manifest clinically with portal hypertension and imaging features of splenomegaly, cavernoma, or collaterals at the porta- or intraluminal filling defect in PV. Surgically created portosystemic shunts must also be excluded [12]. Knowledge of con-

genital extrahepatic mesenteric-systemic shunts is essential for accurate triage and selection of adequate management options. Determining the Type of shunt is crucial, as CEPS type I patients cannot undergo shunt closure; clinical, biochemical, and imaging follow-up with medical management is the only therapeutic option unless a liver transplant is possible. Type II shunts require early closure to prevent hepatic encephalopathy [13].

Conclusion

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 are essential to accurate diagnosis and classification, further directing the therapeutic course and clinical outcome. Not visualization of the portal vein is not always due to chronic thrombosis. Familiarity with congenital aberrant mesenteric drainage is important to accurately diagnose CEPS and Abernethy malformation, which is more critical for radiologists. The main limitations of the sonographic approach are its poor visualization of the splanchnic vessels and its highly operator-dependent. Despite its high sensitivity, in some patients, color-Doppler sonography is challenging to perform well, and the patency of the portal vein cannot be verified with certainty; however Current CT and MR technologies make the diagnosis of Abernethy malformation, and other CEPS shunts possible.

Patient consent

The authors confirmed that informed consent was obtained from the patient described in this report.

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