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Case Report

A case of the Abernethy malformation: An exceptionally rare clinical entity ☆

Jake Gibbons, MD

Baylor University Medical Center, Department of Radiology, 3500 Gaston Avenue, Dallas, Texas, 75246, USA

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ABSTRACT

I review a case of a 30-year-old woman who presented with diarrhea, nausea and vomiting for several days. She was ultimately diagnosed with acute gastroenteritis but was incidentally found to have a congenital extrahepatic portosystemic shunt, also known as the Abernethy malformation. The Abernethy malformation, first described by Dr John Abernethy in the year 1793, is an exceptionally rare clinical entity.

Introduction

The congenital extrahepatic portosystemic shunt (CEPS), termed the Abernethy Malformation, was first described by Dr. John Abernethy in the year 1793 as he performed a post-mortem examination on a 10-monthold girl who died of an unknown cause [1]. In patients with the Abernethy malformation, the splanchnic veins drain directly into the systemic vasculature, often via the inferior vena cava. The underlying cause is thought to be secondary to aberrant embryological development of the peri-intestine venous loop, ultimately resulting in an absent or hypoplastic portal vein with abnormal mesenteric-caval communication [2]. The clinical presentation is variable. Although some patients present in the neonatal period with manifestations of hepatic dysfunction, many remain asymptomatic or minimally symptomatic into adulthood.

Case summary

A 30-year-old woman presented to the emergency department with diarrhea, nausea and vomiting for several days. She also described multiple prior episodes of mental fogging or confusion. She reported no chronic medical conditions, however, as a child required median sternotomy due to congenital heart disease. The specific cardiac anomaly requiring repair was unknown by the patient. She was on no medications. She reported a family history of hypertension and diabetes.

She had a blood pressure of 103/65 mmHg. Her heart rate was mildly elevated at 112 beats per minute. She had a normal respiratory rate and oxygen saturation. On physical examination, she had dry mucous membranes. She had a normal cardiopulmonary exam. Her bowel sounds were hyperactive, but the abdomen was nontender. Her laboratory studies, including complete blood count, comprehensive metabolic panel and

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E-mail address: jakegibbons07@gmail.com

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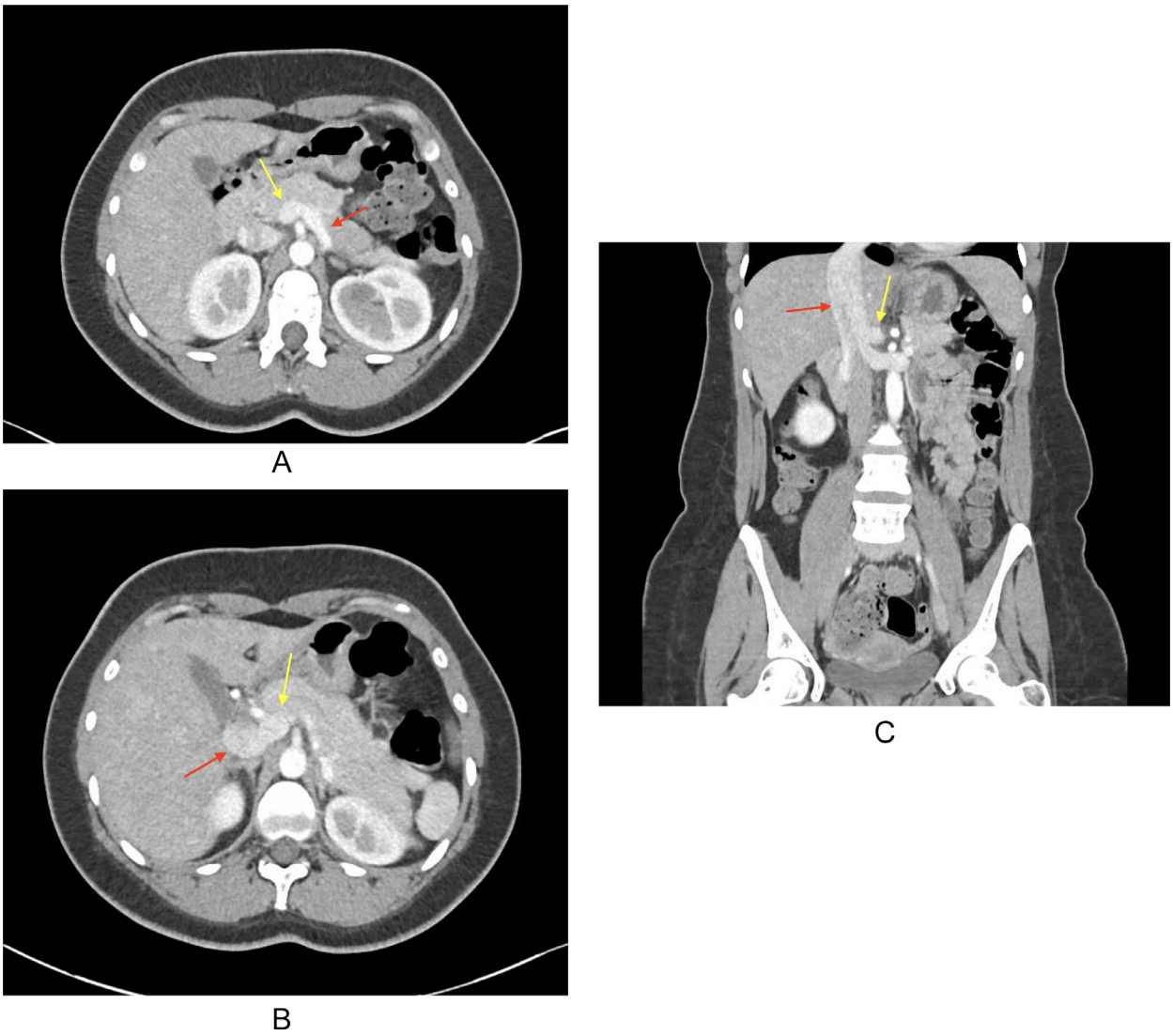

Fig. 1 – (A) Axial contrast-enhanced CT through the abdomen showing the superior mesenteric vein (yellow arrow) joining the splenic vein (red arrow) to create a common trunk. (B) Axial contrast-enhanced CT through the abdomen showing the common trunk (yellow arrow) formed from the confluence of the superior mesenteric vein and splenic veins joining into the inferior vena cava (red arrow). (C) Coronal contrast-enhanced CT through the abdomen showing the common trunk (yellow arrow) formed from the confluence of the superior mesenteric vein and splenic veins joining into the inferior vena cava (red arrow).

urinalysis, were normal. A serum ammonia level was not checked by the clinical team.

A computed tomography (CT) scan of the abdomen and pelvis was obtained. This showed an absence of the portal vein with the splenic vein and superior mesenteric veins combining into a singular trunk which joined the inferior vena cava, therefore bypassing the liver entirely (Fig. 1). The entirety of the hepatic vascular supply arose from the hepatic artery. The hepatic parenchyma was heterogeneous, which was thought to be secondary to the above-described vascular anomalies. The patient was diagnosed with viral gastroenteritis and discharged from the emergency department. Prior to discharge, she was instructed to follow up with hepatology as an outpatient.

She was seen by hepatology, at which time contrast-enhanced magnetic resonance imaging (MRI) of the abdomen was also completed. In addition to the congenital portosystemic shunt, the MRI showed a 2.5 cm lesion within the left hepatic lobe most compatible with a hepatic adenoma (Fig. 2). There were numerous additional lesions that were subsequently characterized as focal nodular hyperplasia on a subsequent MRI with Eovist contrast (Fig. 3). The patient's described episodes of confusion were thought to represent hepatic encephalopathy. Therefore, she was started on lactulose. Given the high coexistence of congenital heart disease in patients with the Abernethy malformation, she was referred to cardiology. Transthoracic echocardiography showed a normal heart in both structure and function.

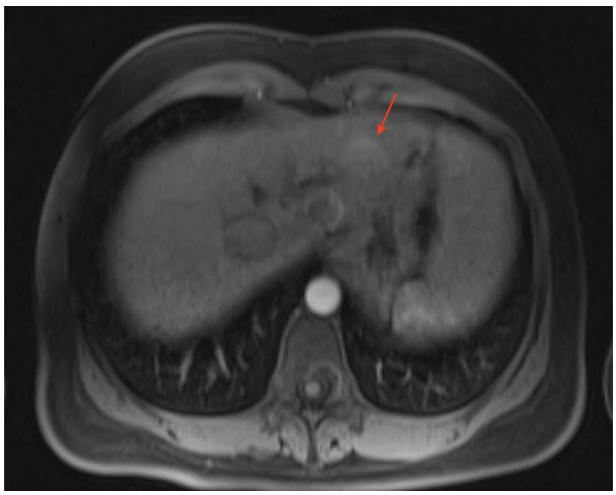

Fig. 2 – Axial contrast-enhanced MRI of the abdomen in the late arterial phase demonstrates a 2.4 cm arterially enhancing lesion compatible with an adenoma (red arrow) in segment 2 of the left hepatic lobe.

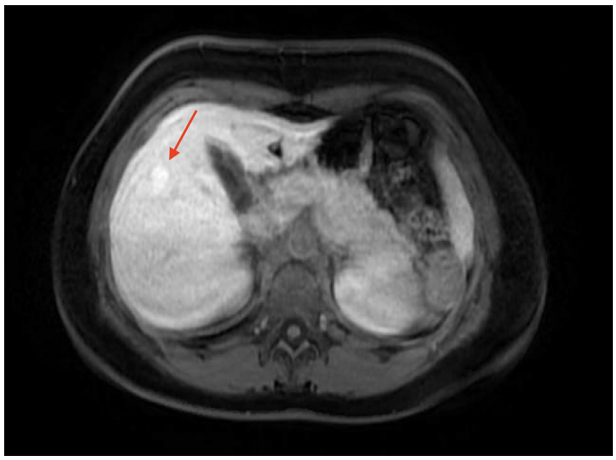

Fig. 3 – Axial MRI of the abdomen with Eovist contrast in the delayed phase (20 minutes following injection) showing a region of focal enhancement (red arrow) within the right hepatic lobe compatible with focal nodular hyperplasia.

Discussion

The Abernathy malformation can be divided into two subtypes. In Type 1, there is complete diversion of blood from the splanchnic system into the systemic venous system with absence of the portal vein. The Type 1 cases are further subclassified as either Type 1a or Type 1b. In patients characterized as Type 1a, the splenic and superior mesenteric veins drain separately into the inferior vena cava, renal vein or iliac vein. On the contrary, in Type 1b the superior mesenteric vein and splenic vein join together to form a common trunk that drains into a systemic vein, typically the inferior vena cava or the right atrium. Type 2 shunts are characterized by an intact, though hypoplastic, intrahepatic portal venous system with partial systemic shunting of portal blood through a side-to-side extrahepatic communication [3].

The clinical presentation of CEPS is variable. Patients may present in the neonatal period with growth restriction, cholestasis and hepatic encephalopathy; others may remain asymptomatic or minimally symptomatic into adulthood [4–6]. Many patients are diagnosed after being evaluated for other developmental abnormalities, since there is a high coincidence of comorbid processes such as congenital heart disease [1]. Biliary atresia, situs inversus, polysplenia and malrotation are other frequently reported associations [7].

With portosystemic shunting of blood, there is deranged metabolism of many substances, such as ammonia, leading to various clinical manifestations including hepatic encephalopathy [2]. The liver in patients with CEPS undergoes regenerative nodular hyperplasia as a response to absent or altered portal venous flow, which ultimately may progress to the development of hepatic neoplasms such as hepatocellular carcinoma, hepatoblastoma, and hepatic adenoma [8]. The development of hepatopulmonary syndrome has been reported in some patients [2].

Patients with CEPS can be diagnosed noninvasively though either ultrasound, CT or MRI. Treatment of such patients depends on the underlying type of shunting, either complete (Type 1) or partial (Type 2). In patients with Type 1 shunting, intervention with occlusion or embolization of the shunt is not performed, as the shunt is the only pathway of blood flow for the splanchnic vessels. Clinical, biochemical and imaging follow-up with medical management is the only treatment option unless liver transplantation is possible [2]. In patients with Type 2 shunting, however, there is a role for either surgical or transcatheter shunt occlusion with or without surgical reconstruction of the portal vein [9,10].

Conclusion

The Abernethy malformation is an exceptionally rare clinical entity with fewer than 200 cases reported in the literature to date [11]. Patients may present in the neonatal period or remain asymptomatic to minimally symptomatic into adulthood. Treatment is variable and depends on the degree of portosystemic shunting, with medical management and eventual liver transplantation serving as the only options for patients with Type 1 disease. A coordinated approach with multiple subspecialists is necessary in the management of such patients given the high incidence of comorbid congenital anomalies affecting other organ systems.

Patient consent

Written consent for publication was obtained from the patient. Approval to publish this manuscript has been given by my institution, its legal representative and an ethics committee. Publication of this document is in accordance with the local legislation.

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