Abernethy malformation with unusual cardiac malformation: case report and literature review

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Abstract

Abernethy malformation, also known as congenital extrahepatic shunt, is a rare anomaly characterized by partial or complete diversion of the portal blood into the systemic venous circulation. The clinical manifestations of Abernethy malformation during childhood include neonatal cholestasis, failure to thrive, mental retardation, and other congenital defects. We report a case of Abernethy malformation Type II in a 9-year-old boy whose left ventricle was slightly enlarged because of several major aortopulmonary collateral arteries but normal laboratory examinations five years earlier. The characteristics of congenital heart disease in patients with Abernethy malformation are discussed. We propose that enlargement of the left ventricular with systemic-pulmonary collateral circulation should raise the suspicion of Abernethy malformation.

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Abstract: Abernethy malformation, also known as congenital extrahepatic shunt, is a rare anomaly characterized by partial or complete diversion of the portal blood into the systemic venous circulation. The clinical manifestations of Abernethy malformation during childhood include neonatal cholestasis, failure to thrive, mental retardation, and other congenital defects. We report a case of Abernethy malformation Type II in a 9-year-old boy whose left ventricle was slightly enlarged because of several major aortopulmonary collateral arteries but normal laboratory examinations five years earlier. The characteristics of congenital heart disease

in patients with Abernethy malformation are discussed. We propose that enlargement of the left ventricular with systemic-pulmonary collateral circulation should raise the suspicion of Abernethy malformation.

Keywords: Abernethy malformation; echocardiography; cardiac malformation; children; major aortopulmonary collateral arteries

Introduction

Abernethy malformation, also known as a congenital portal shunt or congenital portal vein loss, results from malformation of the visceral venous system[1]. The disease was identified in 1793 by John Abernethy in an autopsy of a female infant; it was named Abernethy malformation in 1997[2,3]. Two types of shunt have been defined: Type I is characterized by complete shunting of portal vein blood into the vena cava and is accompanied by congenital portal vein loss; Type II is characterized by an intact portal vein with an extrahepatic connection to the other side (the vena cava)[4]. Patients with Abernethy malformation have symptoms such as hematochezia, hematemesis, and abnormal liver function[5,6]. It may be associated with malformations such as hepatic nodules, musculoskeletal abnormalities, and congenital heart diseases[1,4,7-9].

Although other congenital heart deformities have been described in the literature, we believe the following is the first reported case of Abernethy malformations with major aortopulmonary collateral arteries (MAPCAs), an unusual cardiac malformation in which systemic blood drains into pulmonary circulation via several collateral vessels. This article also discussed the characteristics of congenital heart disease in patients with Abernethy malformation.

Case report

A 9-year-old boy with a history of cough and pneumonia and no previous history of abdominal pain was admitted to the cardiovascular center of Beijing Children's Hospital with a dilated left ventricle, even after taking oral coenzyme Q10 fructose sodium diphosphate and other cardiac nutritional drugs for five years. The patient's medical history included a normal electrocardiogram, myocardial enzyme tests, and normal liver enzymes. The patient was jaundiced at birth. Dandy-Walker syndrome with hydrocephalus was revealed by computed tomography (CT) following an episode of head trauma.

Several imaging modalities were used. Echocardiography revealed that the left ventricle was moderately enlarged, and the wall motion amplitude was enhanced. Color Doppler revealed an abundant blood flow signal from systemic circulation to pulmonary circulation on suprasternal long-axis imaging (**Figure 1A, 1B**). CT with contrast enhancement revealed MAPCAs; the size of the main portal artery was normal, but the right portal artery was significantly slimmer than the left portal artery. Most right portal artery flow into the inferior vena cava bypassed the liver, and the remaining blood drained into the liver **(Figure 1C)** . There were increased tortuous bronchial arteries and several vascular shadows in the mediastinum, most of which were in the mediastinum and hilum of the lung; some extended into the lung and returned to the pulmonary vein of the corresponding region. On this basis, a pulmonary arteriovenous malformation was diagnosed.

Selective angiography revealed that the thoracic aorta and the upper abdominal aorta-pulmonary artery collateral vessels (which were thickened and tortuous) supplied the left and right lungs, respectively. Laboratory tests showed serum glutamic pyruvic transaminase levels, glutamic oxalacetic transaminase, total bilirubin, indirect bilirubin, and direct bilirubin were elevated; levels of serum total protein and albumin were decreased **(Table 1)** . These findings suggested a diagnosis of Abernethy malformation Type II.

Laparoscopy and portal shunt ligation, and intraoperative portal vein angiography were performed. The left branch of the portal vein merged into the inferior vena cava through the venous catheter, and the development of the intrahepatic portal vein was poor. After venous catheter ligation, the development of the intrahepatic portal vein was more significant. The body of the pulmonary artery collateral circulation was closed with wave divisions of 6-mm and 3-mm diamond spring coils, respectively, to block the main body pulmonary collateral circulation blood vessels (**Figures 1D-F)** .

The patient remained well one month after surgery. Echocardiography revealed no apparent abnormal blood flow sign in section of the superior sternal fossa. The inner diameters of the cardiac chambers were smaller than before. The liver function also improved (**Table 1**). Postoperative CT revealed that the left hepatic portal vein and inferior vena cava were truncated and not filled. The patient remains in follow-up.

Discussion

Abernethy malformation is a congenital portal vein malformation caused by abnormal selective preservation and degeneration of the peri-intestinal yolk vein plexus during the formation of the portal vein system during the embryonic period[1]. The disease is rare, affecting an estimated 1/30000 live births[7,9]. Various degrees of preservation and degeneration lead to diverse vascular variations[2]. In 1994, Morgan et al. divided Abernethy malformation into two types: Type I, characterized by intrahepatic portal vein complete loss and complete side-to-side shunt; and Type II, characterized by the presence of intrahepatic portal vein and partial side-to-side shunt[10]. Depending on the confluence of the superior mesenteric vein and the splenic vein, Type 1 Abernethy malformation can be subsided into two types. Type 1 is more common than Type 2[8]. In our case, the intrahepatic vein was present, and there was an extensive shunt between the hepatic portal vein and the inferior vena cava. Type II Abernethy malformation was identified.

The clinical manifestations of Abernethy deformity differ depending on the location of the abnormal shunt vessels and combined malformations, which are divided into three classes[11]: (1) abnormal liver function, liver fatty change, and severe cases such as a regenerative nodule or tumor formation caused by insufficient liver bloody supply[11]; (2) symptoms related to shunt, including hyperammonemia, hypergalactosemia, hepatic encephalopathy, liver lung syndrome, pulmonary hypertension, heart failure, and gastrointestinal bleeding[12,13]; (3) symptoms associated with coexisting congenital abnormalities such as congenital heart disease[7]. Zhang et al. reported 12 children with Abernethy malformation, nine of whom presented with hematochezia, and all had abnormally thick inferior mesenteric vein entering the inferior vena cava through the iliac vein in the pelvic cavity[5]. Other less common symptoms include abnormal liver transaminase, jaundice, and hypoxemia[5]. Abnormal laboratory tests are usually caused by untreated portal vein blood entering the vena cava system and include elevated serum ammonia and bile acids. Patients with Abernethy malformation and congenital heart diseases may initially present with symptoms related to cardiac dysfunction[7]. In the present case, total bile acids and total bilirubin decreased significantly after portal shunt ligation.

Abernethy's deformity is rare but often associated with other malformations. Congenital heart disease is one of the most common associated malformations. Mistinova et al. reviewed 84 cases with congenital absence of the portal vein and found that 65% were female, and 22% had heart disease[1]. Christiane et al. reviewed the characteristics of 316 patients with a congenital portosystemic shunt in the literature and found that the percentage of patients associated with heart disease was also 22%[4]. Patients with cardiac disease are more common in Type 1 than Type 2 (68% vs. 32%)[4]. The most common type is a ventricular septal defect, followed by atrial septal defect, patent ductus arteriosus, atrioventricular septal defect, and coarctation[4]. In the present case, we reported a patient with Type 2 Abernethy malformation and MAPCAs, which is not found in the literature. Congenital MAPCAs can be associated with or without heart defects. Those with heart defects tend to have cyanotic defects. MAPCAS without cyanotic congenital heart defects are rare and are associated with premature birth, lung tissue infection, and pulmonary dysplasia[14]. Our patient was born at term and had no other heart defects. In another study, the authors found that 27.4% of patients with congenital portosystemic shunts had cardiac disease[7].

The co-existence of Abernethy malformation and cardiac raises the suspicion that these two diseases might be related. Some authors suggested that abnormal hemodynamics might cause the abnormal persistence of an embryonic vitelline vein[1], while other authors held that portosystemic shunts might cause a "congestive effect" and the heart defects would result from adaptive change[15]. In some cases with multiple cardiovascular diseases, angiogenesis might play a more critical role[16]. Nevertheless, there is no definitive explanation for the relationship between these diseases; further research is needed.

In addition to cardiovascular anomalies, other deformities may be associated with Abernethy malformation.

A small number of patients are affected by Turner syndrome, Caroli syndrome, Goldenhar syndrome, and Down syndrome[8]. The child in our case was incidentally found to have Dandy-Walker syndrome (revealed by a head CT performed after a traumatic brain injury). Before that time, no neurological abnormalities were found. Other deformities may be found in the urinary and male genital tract, the spleen, brain, and skeletal system[1]. More than 40% of patients with Abernethy malformation may have hepatic lesions such as focal nodular hyperplasia, adenoma, hepatocellular carcinoma, and cirrhosis[1,8]. The relationship between these two diseases might involve absent or decreased portal vein flow and the contemporaneous increased supply of the hepatic artery[8]. There was no hepatic lesion in our case before surgery and during follow-up. However, the follow-up was relatively short, and additional attention should be paid to hepatic lesions in the future.

Conclusion

The diagnosis of Abernethy malformation complicated with a cardiac malformation is challenging. Due to technical limitations and pulmonary air interference, MAPCAs are often undiagnosed. Suprasternal long-axis ultrasound is suggested, especially when the heart has no clear shunt and a dilated left ventricle. In these cases, MAPCAs should be excluded first.

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	TBIL (μmol /L)	TBA (μmol /L)	ALT (U/L)	AST (U/L)	PT (s)	LVEDD (mm)
Before surgery	37.6	114.6	16.2	34.6	13.6	49
After surgery	17.90	6.73	13.0	29.3	12.9	40
Normal	3.4-20.5	0-10	5-40	5-40	9.4-12.5	38.8-42.19

TBIL, total bilirubin; TBA, total bile acid; BA, blood ammonia; ALT, alanine aminotransferase; AST, aspartate aminotransferase; PT, prothrombin time; LVEDD, left ventricular end diastolic diameter

Table 1 Changes in laboratory examination and heart size before and after surgery

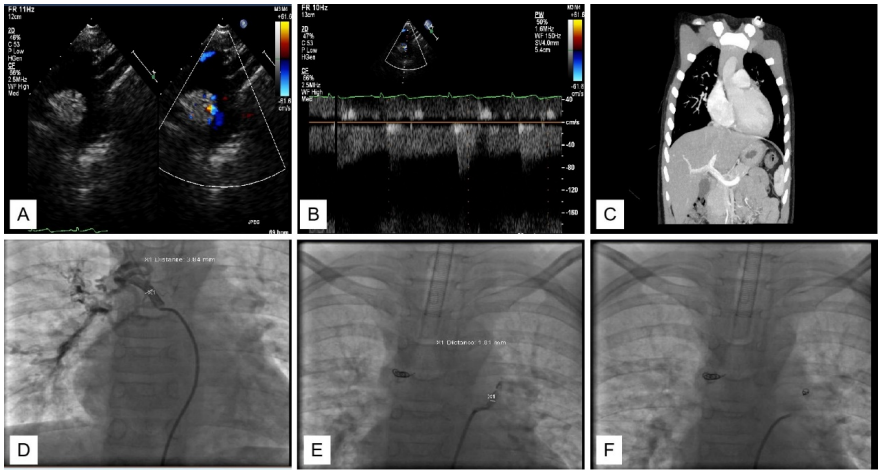

Figure 1. (A) Echocardiography revealing several small tortuous blood flow signals between the aorta and the pulmonary artery. (B) Doppler ultrasonography revealing continuous left-to-right shunt signals at the arterial level with a step-like change. (C) Enhanced CT revealing the left portal vein draining into the

inferior vena cava. (D-F) Selective angiography revealing the thoracic aorta's body and the upper abdominal aorta-pulmonary artery collateral vessels.