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## Case Report

# Abernethy malformation (Type 1B) presenting in a 6-year-old boy with hematochezia and hematuria: A case report <sup>☆</sup>

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### ABSTRACT

Abernethy malformation (Type 1B) presenting in a 6-year-old boy with hematochezia and hematuria: a case report Abernethy malformation is a rare congenital vascular abnormality defined by the diversion of portal blood flow to the inferior vena cava or its tributaries. Clinical presentations include neonatal cholestasis, liver tumors, and encephalopathy but variables in timing and symptomatology. Herein, we present a 6-year-old boy was referred to our hospital with complaints of hematochezia, hematuria, fecal, and urinary incontinence. A diagnosis of type 1b malformation was made depending on magnetic resonance angiography and cardiac catheterization findings, which demonstrated that the superior mesenteric vein and splenic vein joined to form a common trunk measuring 38 mm diameter and then drained into the dilated inferior vena cava with the absence of portal vein abnormalities in the liver. With further investigations, we indicated the presence of many arteriovenous malformations and urogenital abnormalities. The patient was managed conservatively.

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## Introduction

Congenital portosystemic shunts (Abernethy malformation) is a rare congenital malformation. In 1793, Abernethy reported

a case of this malformation for the first time. The patient was a 10-month-old female who died of an unknown cause. The postmortem examination showed that the portal vein joined the inferior vena cava (IVC) at the level of the renal vein with thickened hepatic artery. Many additional abnormalities were

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detected, such as dextrocardia, a right aortic arch, and multiple spleens [1]. The second case of Abernethy malformation was reported in 1883. The patient had a portosystemic end-to-side shunt, and the patient also had many abnormalities of the cardiac, gastrointestinal, urinary, bone, skin, and vascular systems [2]. Congenital portosystemic shunts are classified into intra- or extrahepatic and may accompany by cardiac abnormalities [3].

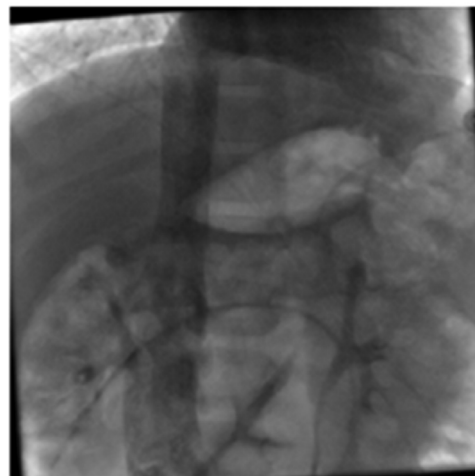
Morgan et al. classified the shunt into two types. Type I is defined by the absence of intrahepatic portal veins and is further sub classified into type IA and type IB. In IA, the superior mesenteric and splenic veins drain separately into the inferior vena cava. While in IB, the superior mesenteric vein and splenic vein form a common trunk that drains into the inferior vena cava. Type II shunt is defined by the presence of a significant collateral, patent intrahepatic vein [2].

Herein, we report – to the best of our knowledge – the first case of Abernethy malformation is Syria. This case demonstrates the importance of radiological imaging in the diagnosis of this rare malformation.

## Case presentation

A 6-year-old boy presented to our hospital with hematochezia and hematuria. His symptoms were associated with fecal and urinary incontinence. The child had a past surgical history of repaired inguinal hernia; otherwise, his past medical and familial history was unremarkable. His vaccination was appropriate for his age. On physical examination, the patient had gross developmental delay. The anthropometrics findings suggested retardation growth. His heart rate was 120 bpm and respiratory rate was 21 per minute. Blood pressure was 96/60 mmHg in the right upper limb. Four-limb blood pressure measurement was obtained. Examination of the cardiovascular and respiratory systems was normal. The abdomen was soft and slightly enlarged with no organomegaly or clinical ascites. Examination of the urogenital system revealed congenital angioma on the penis and enlarged scrotum. Abdominal and pelvic ultrasonography showed slightly hyperechoic, inhomogeneous liver, and thickened portal vein. No portal vein abnormalities in the liver parenchyma were observed.

Abdominal magnetic resonance angiography showed that the superior mesenteric vein joins the splenic vein and forms a common trunk measuring 38 mm in diameter (Fig. 2). This trunk descended in front of the left kidney, lateral to the aorta, then crossed the midline to form a loop around itself and finally drained into the inferior vena cava, which was dilated through the left common iliac vein. Several arteriovenous malformations emanating from superior mesenteric, left internal iliac, and left renal arteries were detected. The child was finally diagnosed with Abernethy malformation type Ib. Echocardiography, cardiac catheterization, and brain magnetic resonance angiography were obtained and revealed no abnormalities (Fig. 1). Scrotal ultrasonography demonstrated normal testes



**Fig. 1 – The dilated inferior vena appears during performing the cardiac Catheterization.**



**Fig. 2 – Abdominal MRA shows that superior mesenteric vein and splenic vein join to form a common trunk which form a loop around itself and drains into inferior vena cava.**

and edematous changes in the scrotum with some vessels inside.

## Discussion

In this paper, we will discuss a case of a 6-year-old boy with type 1b of Abernethy malformation, the diagnostic means, the different clinical manifestations, and treatment choices. Diagnosis of Abernethy malformation can be made depending on imaging techniques, ultrasonography (US), computed tomog-

raphy (CT), and magnetic resonance imaging (MRI). Compared to CT scans, MRI is preferred for children because of the lack of radiation and safety. It was reported that CT angiography and MR angiography were sufficient to detect portosystemic shunts [4]. We were able to identify the portosystemic shunt using MRA and cardiac catheterization.

The clinical presentations of the congenital portosystemic shunt are highly variable. Hyperammonemia, neurological abnormalities, cardiac anomalies, liver tumors, and hepatopulmonary syndrome may present. Other clinical manifestations include hematuria and hematochezia. In our patient, the common trunk drained into the inferior vena cava through the left common iliac vein. Developmental delay was noticed, which can be explained by hepatic encephalopathy and increased blood ammonia levels. Hepatic encephalopathy is a common clinical manifestation of Abernethy malformation and was reported in many cases [5,6]. Blood from mesenteric circulation bypasses the liver and goes directly into the systemic circulation. Toxic substances, which are removed in healthy people by the liver, pass directly into the systemic circulation and reach the central nervous system causing different neurological clinical manifestations. Cases of congenital biliary atresia, cystic dysplasia of the kidneys, ectopic kidneys, and hypospadias were reported in patients with Abernethy syndrome. In our patient, enlarged scrotum and penis angioma were present.

Treatment of Abernethy malformations depends on the type of malformation, the presenting symptoms, complications, and comorbidity. Treatment of Abernethy malformation depends on the type of portocaval fistulas, the symptoms, and the associated complication. Two main treatment options aim to relieve hepatic encephalopathy and maintain normal liver function. In type I, closure of the shunt is usually not performed since is the only outflow tract for the mesenteric venous blood. Hence, liver transplantation is indicated for patients with symptomatic type I malformation who are unresponsive to medical management. While in type II, the treatment includes closure of the shunts, which can be performed laparoscopically or with open surgical ligation [7,8]. Type I patients need clinical, biochemical, and imaging follow-up [8].

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## Conclusion

Although Abernethy malformation is a rare condition, it can seriously affect the patients' quality of life. Therefore, it should be kept in mind. This malformation causes a wide variety of symptoms depending on the types and locations of the portosystemic shunt. The radiograph study here represented the most critical element in diagnosis and exploring for potential abnormalities that accompany Abernethy malformation. Management of this malformation should be considered in a case-by-case model.

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## Consent

Written informed consent was obtained from the patient's father for publication of this case report and accompanying images.

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## Patient consent

Written, informed consent for publication of this case was obtained from the patient.

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## Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:[10.1016/j.radcr.2022.06.045](https://doi.org/10.1016/j.radcr.2022.06.045).

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