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Case Report

Abernethy syndrome presenting as an incidentally detected intrahepatic portosystemic shunt in an infant

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Abstract

Abernethy syndrome is a rare congenital anomaly in which blood from the splanchnic venous system partially or completely bypasses the liver through a portosystemic shunt and enters the systemic circulation. A subset of patients remains asymptomatic and are diagnosed incidentally, while others may present with complications such as portopulmonary hypertension, hepatopulmonary syndrome, portosystemic encephalopathy, or hepatic tumors. Early identification is important, as clinical outcomes depend on the degree of shunting and the presence of complications. In this report, we describe an asymptomatic infant diagnosed during Doppler ultrasonography, with documented spontaneous closure of the intrahepatic portosystemic shunt during follow-up and discuss the case in light of current literature to highlight the importance of careful monitoring.

Keywords: Abernethy syndrome; portosystemic shunt; intrahepatic shunt; spontaneous closure; portal venous anomaly; congenital shunt; vascular malformation

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Introduction

Abernethy syndrome was first described in the 18th century by the London surgeon John Abernethy (1). It is a rare vascular malformation in which venous blood from the intestine and spleen bypasses the liver and drains directly into the systemic circulation through an abnormal channel (2). Its estimated prevalence is approximately 1 in 30,000 live births. The incidence of persistent congenital portosystemic shunts is roughly 1 in 50,000 (3).

Shunts are classified as intrahepatic when the connection occurs between branches of the portal vein and the hepatic veins or the inferior vena cava, and as extrahepatic when the communication arises directly from the main portal vein before branching or when the portal vein is absent (4). Extrahepatic shunts are often associated with other congenital anomalies (2).

Clinical presentation varies mainly according to the proportion of blood flow diverted through the shunt (5). Findings range from asymptomatic patients diagnosed incidentally during evaluation for elevated transaminases or unrelated imaging, to severely symptomatic patients who may develop multi-organ dysfunction (2,6–8).

Diagnosis is usually made with Doppler ultrasonography of the liver and is commonly complemented with computed tomography (CT) or abdominal magnetic resonance imaging (MRI) (7,9–12).

Management depends on the shunt type and the presence of complications (13). Liver transplantation is the only definitive treatment for type 1 shunts, whereas type 2 and intrahepatic shunts may benefit from percutaneous or surgical closure (2,13–15). Intrahepatic shunts, unlike extrahepatic ones, may close spontaneously during the first year of life (2,6).

In this report, we present an asymptomatic infant diagnosed incidentally through imaging to raise awareness of this rare condition.

Case Presentation

A 46-day-old male infant was referred to our clinic after a portosystemic shunt was incidentally found on portal Doppler ultrasonography. The ultrasound was done because the patient had ongoing feeding difficulties and episodes of hypoglycemia. He was born at 40 weeks of gestation by spontaneous vaginal delivery. His early postnatal period included a 20-day NICU stay due to respiratory distress and hypoglycemia. At five weeks of age, he was hospitalized for five days with poor feeding and intermittent fever, and further evaluation was performed at that time.

During his assessment, the infant appeared well, active, and stable. Physical examination was normal, with no abdominal distention, hepatomegaly, or splenomegaly. Routine laboratory tests, including liver function tests and transaminases, were within normal limits. Ammonia and lactate levels were also normal.

Portal Doppler ultrasonography showed an intrahepatic portosystemic shunt measuring up to 2.5 mm, connecting the anterior segment branch of the right portal vein to the middle hepatic vein (Figure 1A–C). Hepatopedal flow was preserved in the main portal vein. Quantitative measurements such as flow velocity or shunt ratio were not assessed.

The patient was diagnosed with Abernethy syndrome and was followed regularly with clinical and radiological evaluations. Complete spontaneous closure of the shunt was confirmed at 8 months of age. The patient was followed for an additional 6 months after shunt closure and remained asymptomatic, with normal liver function tests and laboratory parameters during subsequent visits.

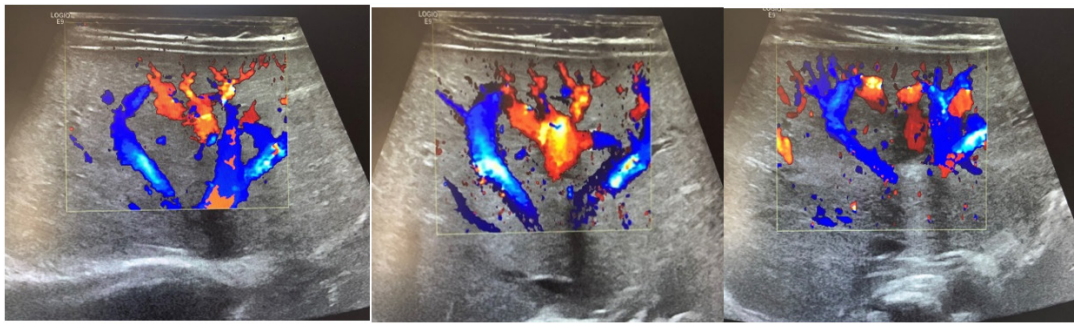

Figure 1. Color Doppler ultrasonography images (A–C) demonstrating a 2.5-mm intrahepatic portosystemic shunt connecting the anterior segment branch of the right portal vein to the middle hepatic vein.

Discussion

Congenital portosystemic shunts are rare abnormalities, and their association with other syndromes and malformations has become clearer as more cases have been reported (7). In addition to the intrahepatic and extrahepatic classification, Morgan and Superina proposed an additional system for extrahepatic shunts. They defined type 1 shunts as *end-to-side* connections in which all portal venous blood is diverted, and the intrahepatic portal branches are severely hypoplastic or completely absent. Type 2 shunts are *side-to-side* connections, where only part of the portal flow is diverted, and the liver still receives some degree of portal perfusion (16). Type 1 shunts are further classified into type 1A and type 1B. Type 1A involves complete congenital absence of the portal vein, whereas in type 1B, the superior mesenteric vein and splenic vein join to form a portal-like trunk that drains directly into the inferior vena cava (4). Other congenital anomalies are reported more frequently in type 2 extrahepatic shunts (2,4,15). The Morgan and Superina classification is summarized in Table 1.

Table 1. Classification of Congenital Extrahepatic Portosystemic Shunts (Morgan & Superina Classification)

Type	Description	Portal Venous Anatomy	Hepatic Perfusion
Type 1A	Complete diversion through an end-to-side shunt	Congenital absence of portal vein; SMV and splenic vein drain separately	No hepatic portal flow
Type 1B	Complete diversion through an end-to-side shunt	SMV + splenic vein form a portal-like trunk draining into IVC	No hepatic portal flow
Type 2	Partial diversion through a side-to-side shunt	Intrahepatic portal branches intact	Partial hepatic portal flow

Summary of the Morgan and Superina classification of congenital extrahepatic portosystemic shunts (16).

Clinical features vary depending on the underlying physiology and the amount of blood bypassing the liver. Reduced hepatic perfusion may lead to intrauterine growth restriction (17), which is seen in about 50% of children with congenital portosystemic shunts. Neonatal cholestasis is another possible early presentation (2).

Because metabolites and vasoactive substances from the splanchnic circulation bypass the liver, serum galactose and ammonia levels may increase. These changes can lead to portosystemic encephalopathy, hepatopulmonary syndrome, or pulmonary arterial hypertension. According to the literature, hypergalactosemia occurs in nearly 70% of newborns with congenital portosystemic shunts (2). Conversely, about 60% of newborns with persistent hypergalactosemia but no enzyme deficiency are found to have a congenital portosystemic shunt (18).

Pulmonary arterial hypertension is another important complication and may occur at any age and

in all shunt types. It can even be the first presenting symptom. Because pulmonary hypertension may progress to right heart failure and death, patients with a known shunt should be monitored with a careful history, physical examination, and echocardiography (2,15,19). Another complication that may occur especially in the neonatal period is severe and persistent hypoglycemia, likely due to reduced hepatic insulin clearance (20). This patient had a history of hypoglycemia in the neonatal period, which aligns with this mechanism.

For type 1 shunts, liver transplantation is the only definitive treatment (2). For shunts in which the intrahepatic portal system is intact, surgical ligation or percutaneous closure performed by an experienced interventional radiologist are possible management options (15,21). Intrahepatic shunts may close spontaneously during the first year of life (2,6). In this patient, follow-up Doppler ultrasonography showed spontaneous resolution of the shunt.

Conclusion

Abernethy syndrome is a rare congenital condition that may affect several organ systems and present with a wide range of findings, including hypertransaminasemia, neonatal cholestasis, hypoglycemia, hyperammonemia, and hypergalactosemia. In addition to these, serious complications such as pulmonary arterial hypertension, portopulmonary syndrome, and hepatic encephalopathy may also develop. Even when patients are asymptomatic, regular clinical assessment and imaging are important to identify complications early and guide appropriate follow-up. Awareness of this condition can help clinicians recognize subtle presentations and prevent delayed diagnosis.

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Conflict of Interest

The authors declare that they have no conflict of interest regarding content of this article.

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Ethical Declaration

Informed consent was obtained from the participant and Helsinki Declaration rules were followed to conduct this study.

Authorship Contributions

Concept: MET, Design: MET, YSO, Supervising: MET, YSO, GT, Financing and equipment: MET, GT, Data collection and entry: MET, YSO, Aİ, GT, Analysis and interpretation: MET, YSO, Aİ, GT, Literature search: MET, YSO, Writing: MET, YSO, Aİ, GT, Critical review: MET, YSO, Aİ, GT

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