A Rare Case of Childhood Malformations, Abernethy Syndrome: Literature Review and Presentation of Two Cases

Özkan Akpınar,[1] Ömer Kubat,[1] Ramazan Kozan,[1] Koray Akkan,[2] Hakan Öztürk,[2] Sinan Sarı,[2] Ödül Eğritaş,[2] Buket Dalgıç,[2] Aydın Dalgıç[1]

Abstract

Abernethy syndrome is a rare congenital vascular anomaly. In this condition, blood from the portal system is diverted to the caval system through a portocaval shunt, entirely or partially bypassing the liver. Prevalence figures in the literature range from 1 per 30 000 population up to 1 per 50 000 population. Abernethy syndrome is classified into 2 types based on the absence (type 1) or presence (type 2) of an intrahepatic portal vein. Abernethy syndrome can manifest with hepatic encephalopathy, pulmonary hypertension, severe osteoporosis, gastrointestinal bleeding, cirrhosis, hepatopulmonary syndrome, and abdominal pain. Hepatocellular carcinoma is a less common presentation. Type 1 Abernethy syndrome is usually treated with a liver transplant, whereas type 2 is addressed by closing the shunt either endovascularly or surgically. Here, we present cases to highlight that surgical shunt ligation is an effective treatment for type 2 Abernethy malformation when the shunt cannot be closed endovascularly.

Key words: *Congenital extrahepatic portosystemic shunt, Hepatocellular carcinoma, Intrahepatic portal vein, Liver transplant, Surgical shunt ligation*

From the [1]Department of General Surgery and the [2]Department of Pediatric Gastroenterology, Gazi University, Ankara, Türkiye

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Corresponding author: *Özkan Akpinar, Department of General Surgery, Gazi University, Ankara, Türkiye*
E-mail: *ozkankpnrr@gmail.com*

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Introduction

Abernethy syndrome is a rare condition where portomesenteric blood bypasses the liver and drains into a systemic vein through a partial or complete shunt. This syndrome is divided into 2 types based on the shunt pattern between the portal vein (PV) and the systemic vein.[1] Abernethy malformation may be associated with congenital anomalies and acquired complications. Previous knowledge and experience with anatomy and embryology are crucial for proper interpretation of imaging findings. Computed tomography (CT), CT angiography, and magnetic resonance angiography can determine shunt anatomy and assess associated malformations. It remains important to distinguish Abernethy malformation from intrahepatic and acquired extrahepatic portosystemic shunts. Mild metabolic abnormalities should be treated with dietary changes and medical therapy, whereas symptomatic patients are eligible for definitive treatment.[2]

There are 2 types of shunts. Type 1 shunts fully divert portal blood into the vena cava and are often associated with a congenital absence of the PV. Conversely, type 2 shunts maintain an intact PV, with blood flow redirected through a side-to-side, extrahepatic connection to the vena cava. Type 1 shunts can be further classified based on the observed anatomy. In type 1A shunts, the splenic vein and superior mesenteric vein (SMV) drain separately into the inferior vena cava (IVC) or the left renal vein. In type 1B shunts, the SMV and splenic vein typically merge but do not supply the liver.[3] Typically, a patient with type 1 Abernethy malformation will require a liver transplant, and patients with type 2 are best treated with shunt occlusion through surgery or by endovascular procedure.[1,4]

Case Report

Case 1

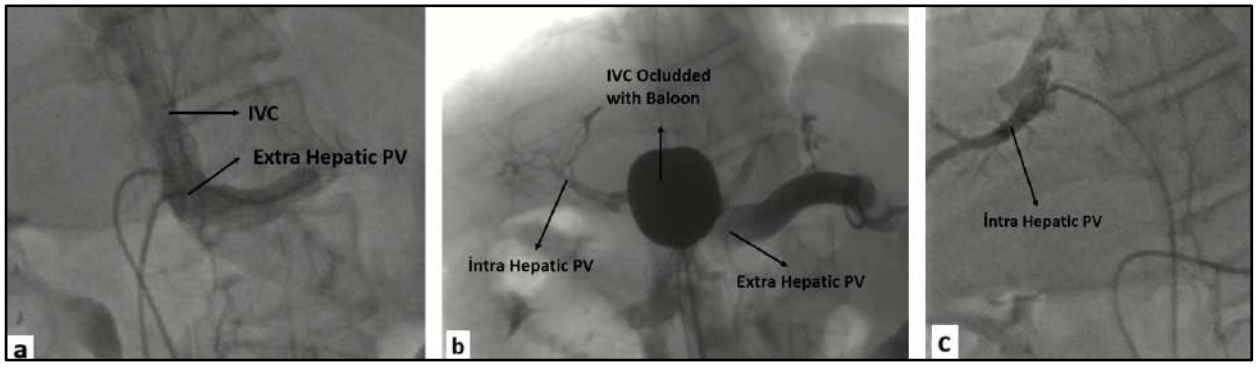
A 16-year-old male patient presented with abdominal pain, high liver function tests, mild consciousness disturbance, and frequent respiratory infections. Urine copper levels were also high; therefore, Wilson disease was initially suspected, and a liver biopsy was performed. However, the pathology was unremarkable, and the test results for the ATP7B gene mutation were negative. On abdominal Doppler ultrasonography, it was observed that the SMV and splenic vein opened into the IVC. In the initial evaluation, intrahepatic PV branches were not detected by Doppler ultrasonography and CT. Based on the noninvasive imaging results, the patient was diagnosed with type 1 Abernethy syndrome. Blood ammonia levels were measured at high values of 50 to 190 μmol/L (reference standard is 11-35 μmol/L).[5] No pathology was detected by echocardiography and gastroduodenoscopy. It was noted in the patient's history that he had 3 long bone fractures and that he was enrolled in a supplemental educational program to address low performance in the standard curriculum.

To evaluate the structure of the intrahepatic PV and shunt, the patient underwent digital subtraction angiography by interventional radiology (Figure 1). The femoral vein was accessed, and the extrahepatic PV was reached via the shunt for delivery of a radiopaque agent. We observed that all of the opaque material emptied into the IVC (Figure 1a). A balloon was then inflated above the shunt junction with the IVC, and opaque material was given again from the extrahepatic PV and splenic vein junction. This time, we observed that the opaque material went toward the liver, which indicated patency of the intrahepatic PV (Figure 1b). Direct intrahepatic PV catheterization showed intrahepatic tributary branches of PV, which confirmed that the malformation was type 2 Abernethy syndrome (Figure 1c).

We attempted to close the shunt between the PV and IVC by interventional radiology with an atrial septal defect closure device. However, we judged the shunt to be unsuitable for closure with angiographic intervention due to the large size of the shunt orifice and close proximity to the main PV trunk. Therefore, we used a surgical procedure to close the shunt. We considered the possibility of a complication with the intrahepatic PV flow (while monitored intraoperatively with Doppler ultrasonography), which could prevent closure of the shunt, and/or the possibility of a vascular injury. Hence, we contacted the patient's mother, who agreed to prepare and remain on-call as a donor, if an emergency living related LT were required.

The surgery was started with a right-sided hockey stick incision, and we proceeded posterior to the pancreas to reveal the hepatic artery, IVC, splenic vein, SMV, inferior mesenteric vein, and shunt (Figure 2). Before the shunt was surgically closed, we checked the patency of the intrahepatic PV branches; we closed the shunt with a Satinsky clamp and measured the intrahepatic PV flow with intraoperative Doppler ultrasonography (Figure 2,

Figure 1. Conventional Angiography for Differential Diagnosis of Abernethy Malformation



Abbreviations: IVC, inferior vena cava; PV, portal vein

a, Catheterization of the extrahepatic PV shows direct flow into the IVC. This image shows a malformation that appears to be typical of the type 1 condition. **b,** When the shunt was closed with a balloon through the IVC catheterization, the tributary branches of the PV into the liver were clearly visible, which indicated the malformation was type 2. **c,** Direct Intrahepatic PV catheterization shows intrahepatic tributary branches of PV, which confirmed the type 2 malformation.

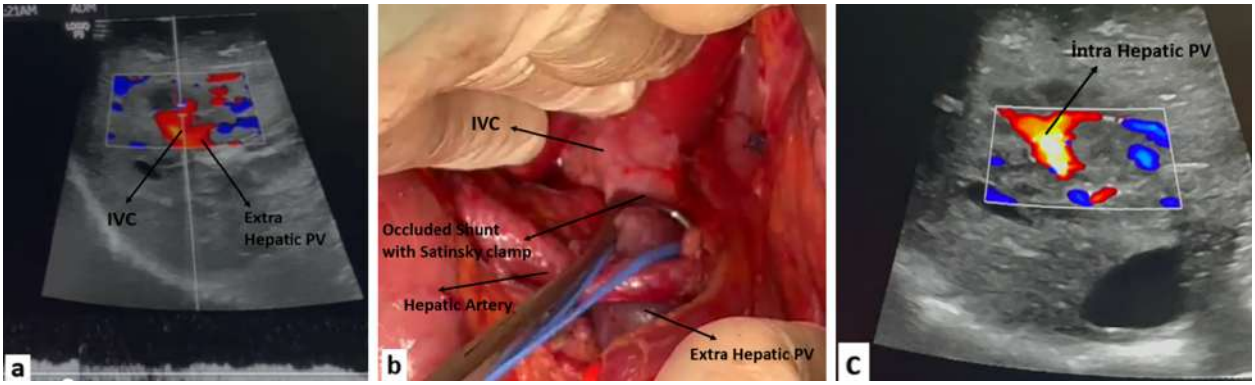
b and c). We confirmed a flow rate of 13 cm/s (reference standard is 20-30 cm/s), which was deemed sufficient at this stage; hence, we applied vascular clamps on both ends of the shunt, and both ends were sewn using 5.0 polypropylene sutures (Prolene) (Figure 3).[6] The patient had no postoperative complications.

Case 2

A 6-year-old female patient was discovered to have an extrahepatic portocaval shunt during a routine pediatric checkup, as revealed by abdominal Doppler ultrasonography. Subsequent CT angiography of the abdomen confirmed the diagnosis of type 2 Abernethy malformation. The patient had a history of frequent respiratory tract infections and central cyanosis during infancy. She remained under observation without medication by pediatric cardiology for an asymptomatic patent foramen ovale. No other anomalies or symptoms were observed.

The patient first underwent evaluation of the size of shunt and composition of the intrahepatic PV by digital subtraction angiography. After confirmation that the flow in the intrahepatic PV had been transferred to the

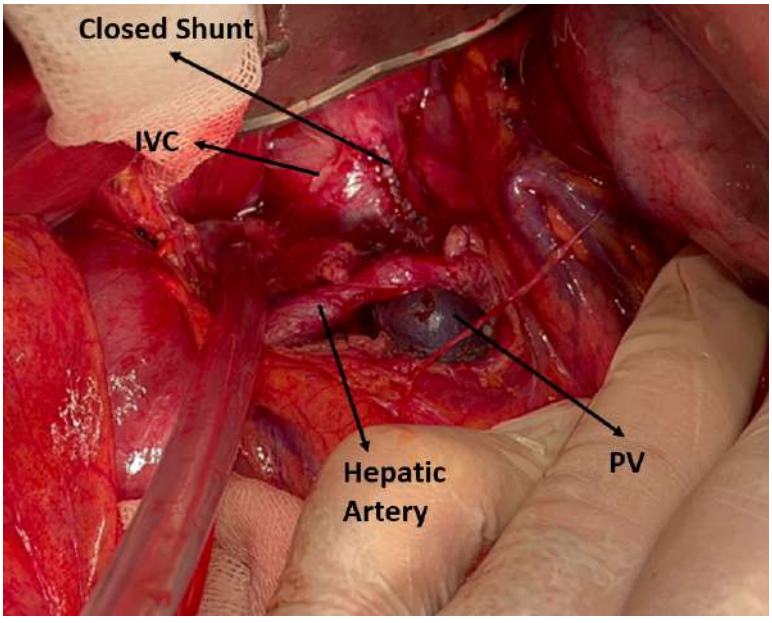
Figure 2. Intraoperative Examination of Intrahepatic Portal Vein Branches After Shunt Occlusion



Abbreviations: IVC, inferior vena cava; PV, portal vein

a, Intraoperative Doppler ultrasonography shows absence of PV circulation in intrahepatic area, and extrahepatic PV flows direct into the IVC. **b,** Abernethy shunt was closed with the Satinsky clamp. **c,** Intrahepatic branches of PV.

Figure 3. Case 1: End of Surgery, With Abernethy Shunt Closed and Sutured



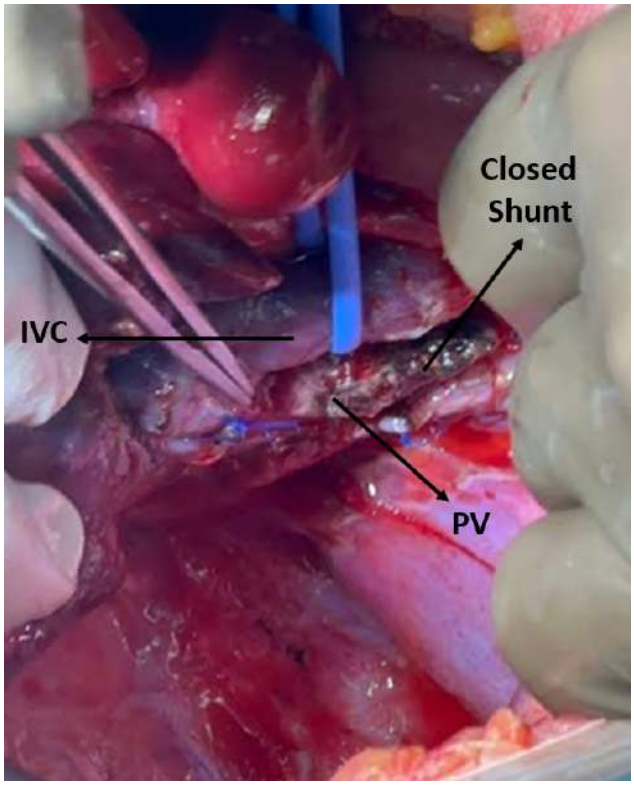
Abbreviations: IVC, inferior vena cava; PV, portal vein

hepatic veins without complications, we decided to close the shunt. Initial attempts by interventional radiology to close the shunt were unsuccessful due to its diameter. Consequently, we promptly proceeded with surgical closure. During the surgery, it was necessary to dissect the liver slightly to expose the shunt (Figure 4). The shunt was then sewn and closed with 5.0 Prolene sutures. There were no postoperative complications.

Discussion

Abernethy syndrome, also known as congenital extrahepatic portosystemic shunt, is a rare congenital anomaly. It is characterized by the absence or malformation of the PV that causes portal blood to flow directly into the systemic circulation. This condition presents a significant challenge in pediatric medicine due to its various clinical manifestations and potential complications. Here, we outlined the diagnosis and management of Abernethy syndrome in 2 pediatric patients, emphasizing the importance of timely intervention and multidisciplinary collaboration.[6]

Figure 4. Case 2: End of Surgery, With Abernethy Shunt Closed and Sutured



Abbreviations: IVC, inferior vena cava; PV, portal vein

Treatment for Abernethy syndrome often involves a combination of medical management and surgical intervention, depending on symptom severity and associated complications.[7] The decision for surgical shunt ligation in the 2 cases we have presented in this report was based on the clinical presentation of each patient, including aspects of hepatopulmonary syndrome, consciousness disturbance, and recurrent gastrointestinal bleeding. This approach aimed to redirect portal blood flow back into the liver, to restore hepatic function and minimize further complications.

Surgical shunt ligation in pediatric patients with Abernethy syndrome requires careful consideration of factors such as patient age, underlying liver function, anatomic variations, shunt diameter, and potential long-term outcomes. Preoperative imaging studies, including Doppler ultrasonography, CT angiography, and conventional angiography, are crucial for delineation of the vascular anatomy, detection of patency of PV branches in the liver, and guidance of surgical planning. Intraoperative monitoring techniques, such as transesophageal echocardiography and PV pressure measurements, play a critical role to ensure procedural success and safety.[8]

Postoperative monitoring and multidisciplinary follow-up are essential after surgical shunt ligation to assess the effectiveness of the intervention, monitor potential complications, and optimize long-term outcomes.[9] In the 2 cases we have presented here, significant improvement in the clinical status of the patients was observed postoperatively, with resolution of both hepatopulmonary syndrome and consciousness disturbance. These outcomes underscore the importance of early diagnosis and timely intervention for mitigation of the adverse effects of Abernethy syndrome on the health and quality of life for pediatric patients.[10]

Conclusions

Abernethy syndrome presents a rare but substantial clinical challenge in pediatric medicine and requires a comprehensive and multidisciplinary approach to diagnosis and management. The cases we have presented in this report underscore surgical shunt ligation as a viable treatment option for select patients with Abernethy syndrome and offers to provide improved clinical outcomes and quality of life. Further research and collaboration are warranted to enhance our knowledge of this complex

congenital anomaly and to optimize therapeutic strategies for affected individuals.

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