Zeynep Kamil Med J 2026;57(2):122–124
DOI: 10.14744/zkmj.2025.48254

CASE REPORT

Abernethy malformation as a rare cause of rectal bleeding and scrotal varicocele in childhood

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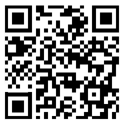

ABSTRACT

Abernethy malformation is a rare syndrome characterized by a congenital portosystemic shunt and the partial or total absence of the portal vein. This case report describes a 9-year-old male patient with type 1 Abernethy malformation presenting with scrotal varicocele and rectal bleeding. The patient presented with complaints of scrotal swelling and rectal bleeding. Physical examination revealed scrotal varicose veins, and laboratory tests showed no abnormalities except for a minimally prolonged INR. Radiological evaluation identified a portosystemic shunt between the splenic vein and the left iliac vein, leading to the diagnosis of type 1 Abernethy malformation. Further investigations for other congenital anomalies revealed no additional findings. Abernethy malformation is an extremely rare condition that may present with a variety of symptoms. In cases with findings such as rectal bleeding and scrotal varicocele, indicative of systemic collateral dilatation, Abernethy malformation should be considered among the rare differential diagnoses.

Keywords: Abernethy malformation, childhood varicosel, congenital extrahepatic portosystemic shunt, rectal bleeding, scrotal varicosel.

Cite this article as: Tetik Dinçer B, Urgancı N, Özel A. Abernethy malformation as a rare cause of rectal bleeding and scrotal varicocele in childhood. Zeynep Kamil Med J 2026;57(2):122–124.

Received: March 19, 2025 **Revised:** August 22, 2025 **Accepted:** August 27, 2025 **Online:** February 24, 2026

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Zeynep Kamil Medical Journal published by Kare Publishing. Zeynep Kamil Tıp Dergisi, Kare Yayıncılık tarafından basılmıştır.

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INTRODUCTION

Abernethy malformation, a rare vascular anomaly characterized by a congenital portocaval shunt and partial or complete absence of the portal vein, was first described by John Abernethy in 1793.[1] Although its incidence remains unclear, advances in imaging techniques have led to an increase in its diagnosis in recent years. While it may be incidentally detected during imaging in asymptomatic patients, it can also result in severe conditions such as hepatic encephalopathy or pulmonary hypertension.[2] Abernethy malformations are classified as type 1 when there is complete absence of the portal vein and a total portosystemic shunt (PSS), whereas patients with a hypoplastic portal vein and partial PSS are categorized as type 2.[3] This case report presents a type 1 Abernethy malformation in a patient who presented with scrotal varicosities and a history of rectal bleeding.

CASE REPORT

Written informed consent was obtained from the patient's legal guardians. A 9-year-old male patient presented with a 2-year history of scrotal swelling and intermittent rectal bleeding. His personal and family history was unremarkable. Vital signs were appropriate for his age. His height was 116 cm (3rd percentile), and his weight was 21 kg (3rd percentile). Physical examination revealed varicose structures in the scrotum and prepuce (Fig. 1). Other systemic examinations were normal. Laboratory tests showed no significant abnormalities except for a minimally prolonged INR (INR: 1.54 [normal range: 0.8–1.4]). Abdominal ultrasonography revealed the absence of the portal vein, with no pathological findings in the liver or evidence of splenomegaly. Contrast-enhanced abdominal MRI demonstrated portal vein aplasia and a tortuous venous structure extending from the splenic vein to the left iliac vein. Collaterals were observed in the scrotal and perianal regions (Fig. 2). The findings were consistent with type 1 Abernethy malformation, and the patient's symptoms were attributed to this condition. The patient was referred to pediatric gastroenterology for follow-up.

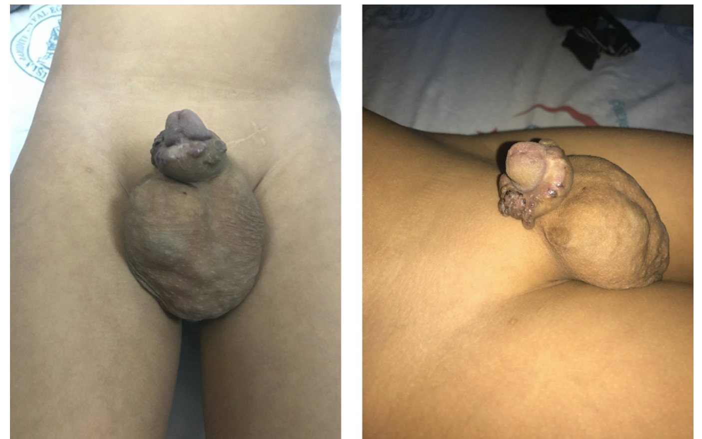

Figure 1: Varicose dilatation of testicles and papillomatous lesions on lymphangioma circumscriptum and angiokeratoma of the skin of prepisium.

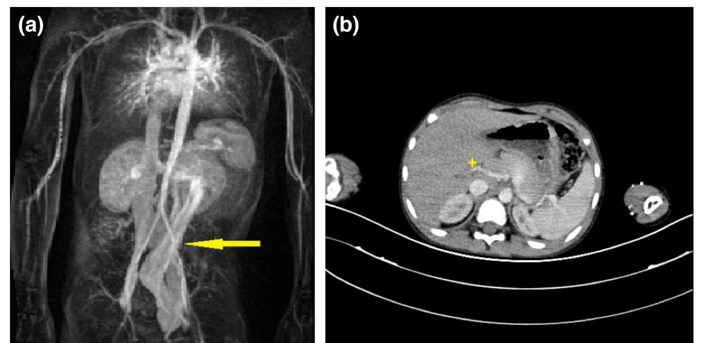

Figure 2: (a) Three-dimensional (3D) phase-contrast MR angiography. A venous, dilated and tortuous vein from the splenic vein was draining to the left iliac vein, extending toward the left iliac fossa. Inferior vena cava, left renal vein and SMA were dilated secondary to the increase in flow (yellow arrow). **(b)** Portal vein is invisible on triphasic CT of abdomen with iv contrast (yellow star).

DISCUSSION

Abernethy malformation is an extremely rare condition, with most information in the literature derived from case reports and small case series. Due to limited data on optimal treatment approaches, no consensus exists. However, life-threatening complications such as late-onset cirrhosis, hepatorenal syndrome, and pulmonary hypertension have been reported in the literature.[4] Additionally, patients diagnosed with Abernethy malformation have a high prevalence of liver lesions (e.g., focal nodular hyperplasia, regenerative nodules, adenomas) and an increased risk of developing liver tumors.[5] Therefore, early diagnosis is critical for enabling timely and appropriate treatment.

Patients with Abernethy malformation may also present with associated congenital anomalies.[1,2] Cardiovascular anomalies are particularly prominent, and patients diagnosed with Abernethy malformation should be screened for such anomalies to plan appropriate treatments. Conversely, Abernethy malformation should be considered in patients with cardiovascular anomalies as a potential associated condition.[6]

While asymptomatic patients with Abernethy malformation can be managed with regular follow-up, symptomatic patients require treatment. For type 1 cases, liver transplantation is typically required, whereas in type 2 cases, closure of the shunt is sufficient and can be achieved through either endovascular or surgical methods. [4] Although our patient did not present with cardiac anomalies or liver lesions, such conditions are frequently observed in Abernethy malformation and may require specific management.

CONCLUSION

Abernethy malformation is a rare condition that can present with diverse symptomatology depending on associated anomalies. This case report highlights a patient presenting with findings such as rectal bleeding and scrotal varicosel, attributed to portosystemic shunt-related collaterals. The patient was diagnosed with type 1 Abernethy malformation and placed under follow-up, with no additional anomalies detected in evaluations of other systems.

Statement

Ethics Committee Approval: This is a single case report, and therefore ethics committee approval was not required in accordance with institutional policies.

Informed Consent: Written informed consent was obtained from the patient's legal guardians

Conflict of Interest: The authors declare that there is no conflict of interest.

Financial Disclosure: The authors declare that they have not received any funding, grants, or other support during this study.

Use of AI for Writing Assistance: Not declared.

Author Contributions: Concept – Concept – BTD, NU, AÖ; Design – BTD, NU, AÖ; Supervision – NU, AÖ; Results – BTD, NU, AÖ; Materials – BTD, NU, AÖ; Data Collection and/or Processing – BTD, NU, AÖ; Analysis and/or Interpretation – BTD, NU, AÖ; Literature Search – BTD, NU, AÖ; Writing – BTD, NU, AÖ; Critical Reviews – BTD, NU, AÖ.

Peer-review: Externally peer-reviewed.

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