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Case report

Type II Abernethy malformation in an adult: case report

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Abstract

Background: Abernethy malformations are vascular developmental anomalies characterized by hypoplasia or agenesis of the portal vein. These malformations have a variable clinical presentation due to associated complications and are often diagnosed incidentally through imaging studies, primarily in pediatric patients. **Case Summary:** We report the case of a 39-year-old female patient who presented with spider telangiectasias. Laboratory tests revealed pancytopenia and elevated liver transaminases. Given the complexity of the case, autoimmune diseases and portal vein thrombosis were ruled out through autoantibody testing and Doppler ultrasound. Incidentally, triphasic computed tomography of the abdomen revealed splenomegaly, an aneurysm of the inferior segmental artery of the left kidney, splenorenal collaterals, and portal vein hypoplasia consistent with Abernethy malformation type II. **Conclusions:** Abernethy malformation is rare in adults and was an incidental finding in this case. Although dermatological signs contributed to its detection, the relationship between the hemangioma and this condition, as well as its role as a potential physical sign, remains unclear.

Keywords

Vascular malformations, congenital anomalies, case study.

INTRODUCTION

Congenital extrahepatic portosystemic shunts, also known as Abernethy malformations, are vascular developmental anomalies characterized by hypoplasia or agenesis of the portal vein and diversion of portomesenteric blood flow to a systemic vein before reaching the portal bifurcation[1]. This condition was first described by John Abernethy in 1793[2]. According to Morgan and Superina[3], Abernethy malformations are classified into two main types: type I, characterized by complete absence of the portal vein with total diversion of portal blood to the inferior vena cava, and type II, involving portal hypoplasia with partial diversion of blood flow to the inferior vena cava through extrahepatic connections.

According to Franchi and colleagues[4], diagnosing this condition presents a significant challenge due to its varied clinical presentation, which may include hepatic tumors, hepatopulmonary syndrome, pulmonary arterial hypertension, portosystemic encephalopathy, heart failure, and glomerulonephritis. However, the diagnosis can also be an incidental finding. Baiges and colleagues[5] reported 66 cases of Abernethy malformation, 80% of which were diagnosed incidentally. Doppler ultrasound is the initial diagnostic tool for detection[6]. Other vascular malformations may coexist, although they are frequently underdiag-

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nosed[7]. Additionally, further laboratory testing is often required, depending on the patient's clinical presentation, including arterial ammonia levels, brain magnetic resonance imaging (MRI), electroencephalography, or liver biopsy[8].

The objective of this article is to report the case of a 39-year-old female patient who sought medical consultation for a dermatological concern and was found to have abnormal laboratory results. Follow-up assessments, including imaging studies, revealed incidental findings of multiple portosystemic shunts with portal hypoplasia consistent with a type II Abernethy malformation, accompanied by alterations in renal vasculature.

CASE DESCRIPTION

This case concerns a 39-year-old female patient from Azuay, Ecuador, who presented with telangiectasias as her primary reason for consultation. Her medical history included a pyogenic granuloma in the oral cavity and slow-healing lesions on her left foot. She had menarche at age 11, gravidity (G): 1, parity (P): 1, and one living child (LC): 1. She experienced pregnancy at age 15, with a vaginal delivery complicated by severe immediate postpartum hemorrhage. Prior to the pregnancy, her menstrual cycles were regular; however, she developed amenorrhea afterward.

On physical examination, erythematous skin lesions were observed on the palms, anterior chest, and face (**Figure 1**), which were confirmed as hemangiomas by biopsy. Additionally, there was a residual pyogenic granuloma lesion on the lower lip that disappeared upon digital pressure and was associated with mild tenderness on palpation. The patient displayed a complete absence of axillary and pubic hair.

Laboratory tests revealed pancytopenia and elevated liver transaminases: aspartate aminotransferase (AST) at 147 U/L (reference range: 0.0–32.0 U/L) and alanine aminotransferase (ALT) at 149 U/L (reference range: 0.0–35.0 U/L). Additionally, elevated alpha-fetoprotein levels were detected at 9.5 IU/ml (reference range: 0.0–5.8 IU/ml), prompting further imaging studies.

An abdominal ultrasound revealed hepatomegaly with a slightly heterogeneous size, increased echogenicity, and splenomegaly measuring 15 x 6.4 cm. A subsequent 3D reconstruction abdominal angiography (**Figure 2**) showed that the proximal third of the portal vein measured approximately 8.6 mm, with hypoplasia of the portal hilum and its branches. Multiple shunts were observed connecting the splenic and superior mesenteric veins to the inferior vena cava, along with a splenorenal shunt measuring approximately 10 mm. Additionally, tortuous collateral veins extended to the short gastric and splenorenal veins. The liver displayed parenchymal density changes and Glisson's capsule irregularities consistent with chronic liver disease, as well as dilated venous vessels at the lower pole of the left kidney. Based on these findings, Abernethy malformation was diagnosed as an incidental finding via computed tomography, and a lower segmental renal artery aneurysm was identified through Doppler color ultrasound (**Figure 3**).

Other conditions, such as systemic lupus erythematosus (negative antinuclear antibodies and anti-DNA antibodies), antiphospholipid syndrome (anticardiolipin IgG: 4.3 [reference value: <12 = negative], anticardiolipin IgM: 3.2 [<12 = negative], negative lupus anticoagulant, anti-β2 glycoprotein IgG: 3.1 U/mL [<12 U/mL = negative], anti-β2 glycoprotein IgM: 4.5 U/mL [reference value: <12 U/mL = negative]), and portal vein thrombosis (ruled out through hepatic Doppler ultrasound and thrombophilia screening) were excluded.

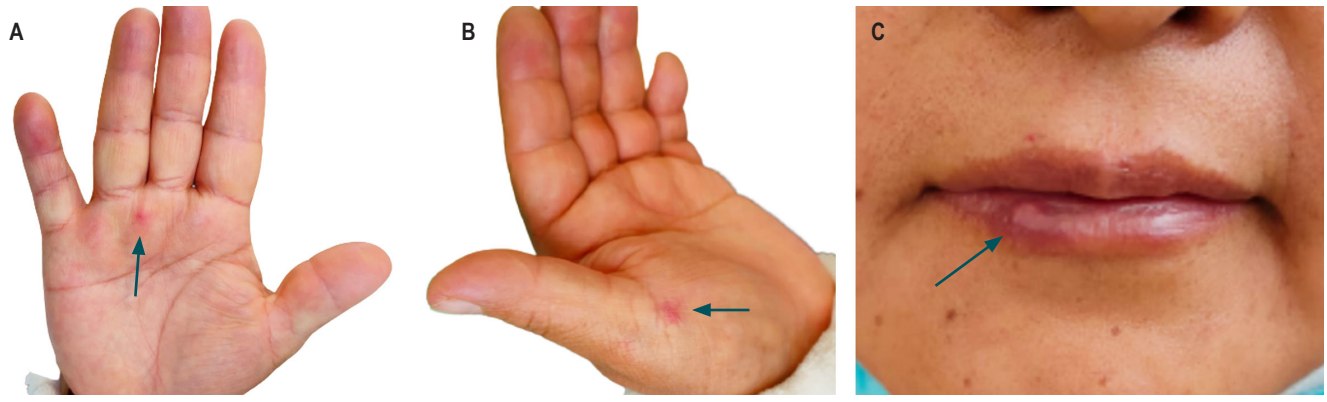


Figure 1. Cutaneous Lesions. **A.** Telangiectasia over erythematous skin in the middle of the Right palmar digital pad (arrow). **B.** Telangiectasia over erythematous skin in the left thenar eminence (arrow). **C.** Erythematous papule residual from the pyogenic granuloma on the lower lip (arrow). Author's File.

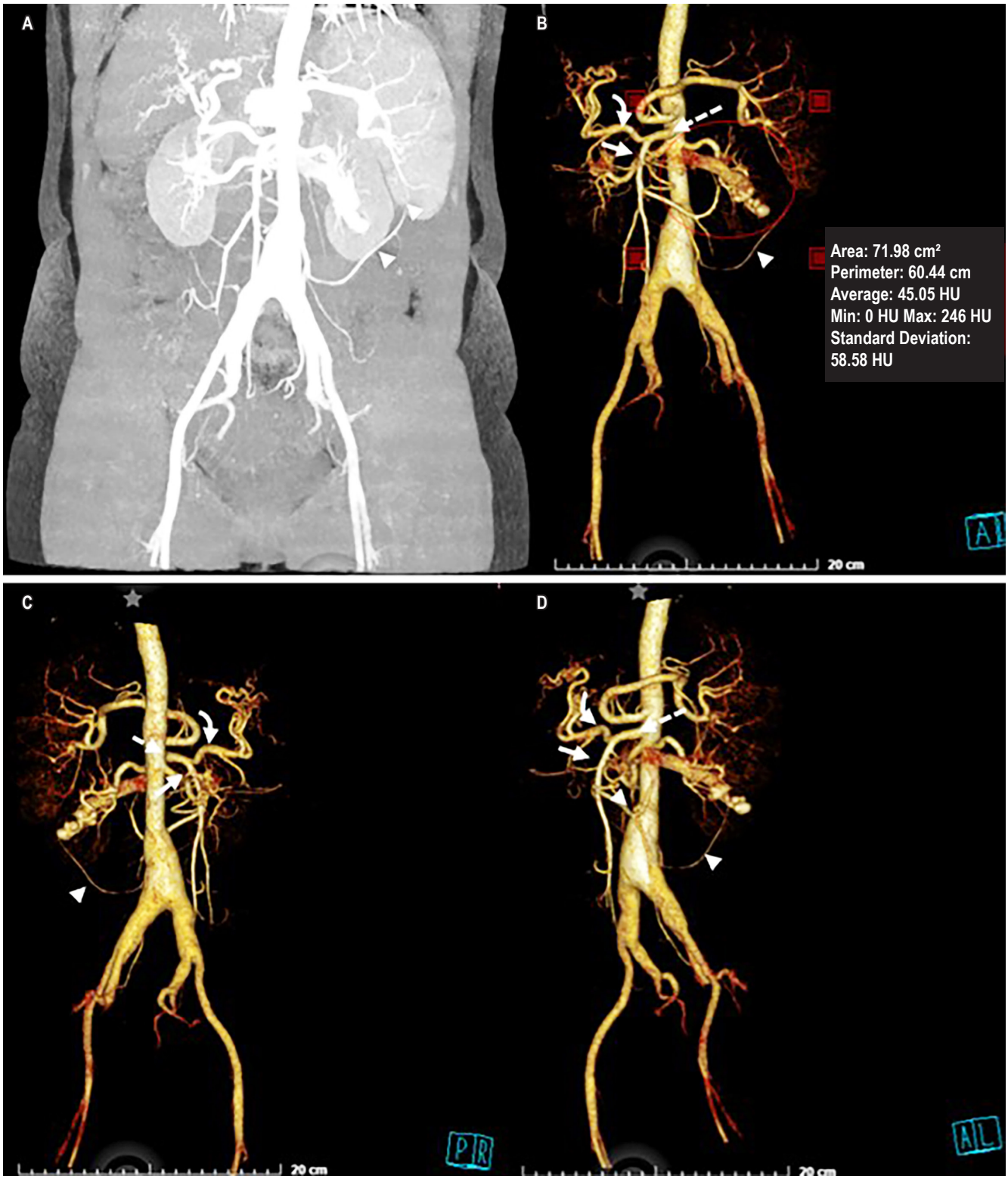


Figure 2. A. Abdominal angiography. **B.** 3D reconstruction, anterior view. **C.** Posterior view. **D.** Anterolateral view. The images show the junction of the superior mesenteric vein (straight arrow) and the splenic vein (curved arrow) draining directly into the superior vena cava (dashed arrow), along with a shunt connecting the splenic and superior mesenteric veins to the inferior vena cava. This shunt extends toward the left hypochondrium, descends near the lower renal pole, and reconnects with the inferior vena cava (arrowheads). Author's File.

Protein C and antithrombin II deficiencies were detected, prompting investigations for factor V Leiden mutation and G20210A prothrombin gene mutation, both of which yielded negative results, attributing the deficiencies to the patient's liver disease. Hormonal evaluation showed the following results: thyroid-stimulating hormone (TSH): 2.84 μIU/mL (reference range: 0.27–4.20 μIU/mL), free T_3: 1.8 pg/mL (reference range: 2.0–4.4 pg/mL), free T_4: 0.23 ng/dL (reference range: 0.93–1.70 ng/dL), prolactin: 3.4 ng/mL (reference range: 4.79–23.30 ng/mL), and morning cortisol: 3.2 μg/dL (reference range: 6.2–19.4 μg/dL), indicating hypopituitarism.

The patient experienced one episode of syncope and was later hospitalized due to a chronic subdural hematoma and subarachnoid hemorrhage, as evidenced by cranial computed tomography (**Figure 4**). She subsequently developed respiratory decompensation and was admitted to the intensive care unit due to bacterial hospital-acquired pneumonia. Later, a pleural effusion was detected, necessitating chest tube drainage, after which she remained hospitalized until stabilization.

Clinical follow-up of the non-traumatic chronic subdural hematoma was conducted due to the bleeding risk, and the patient showed favorable progress with reversal of the condition. At present, the patient has cirrhosis classified as Child-Pugh A and is being treated for hypopituitarism with hormone replacement therapy: 100 μg/day of levothyroxine and 5 mg/day of prednisone, with regular monitoring of blood hormone levels. A surgical resolution for the Abernethy malformation was offered, but the patient declined, as she remains stable at this time.

DISCUSSION

This report presents a case of type II Abernethy malformation diagnosed incidentally at the age of 39 during a dermatological consultation. Blood tests revealed pancytopenia, elevated liver transaminases, and elevated alpha-fetoprotein levels, prompting the need for imaging studies. These studies showed portal system hypoplasia with multiple portosystemic shunts and morphological alterations of the left kidney's inferior segmental artery. Despite the patient having received care within the national health system on multiple occasions, the malformations had not been previously detected due to a lack of clinical suspicion, as the patient did not present symptoms typically associated with this condition.

This malformation likely originates between the fourth and eighth weeks of gestation due to excessive regression

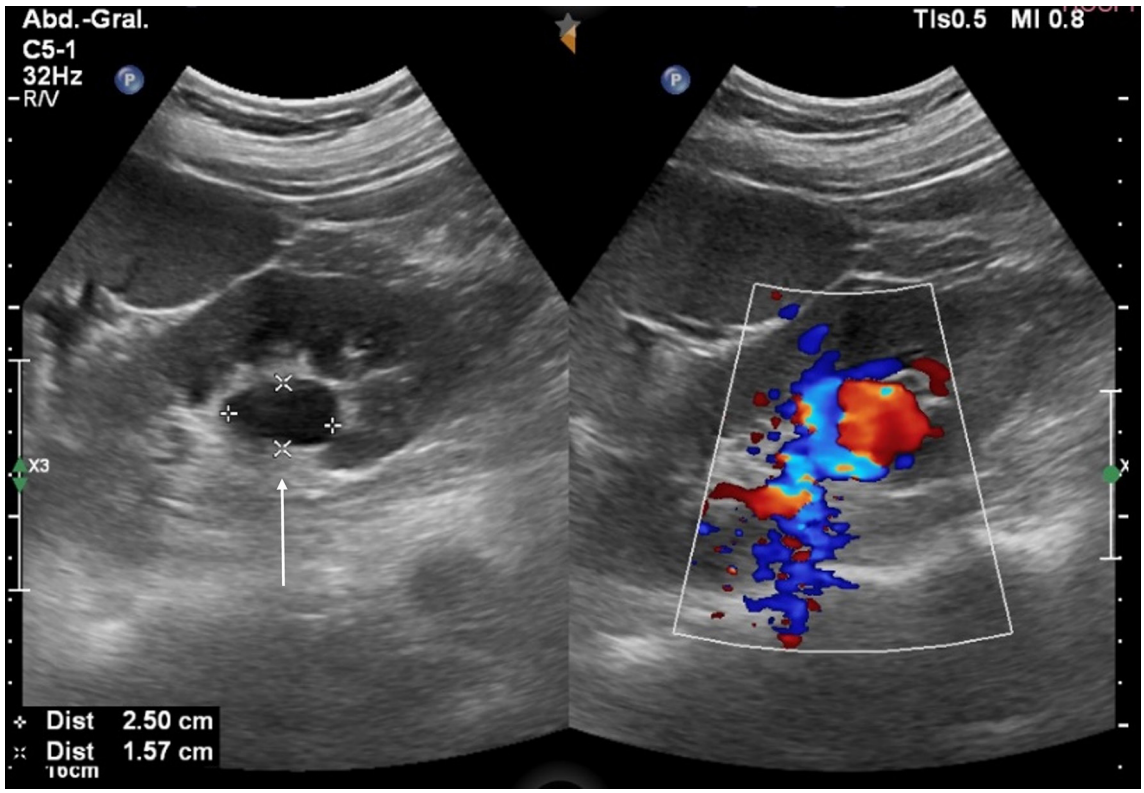


Figure 3. Color Doppler Ultrasound. Aneurysmal dilation of the inferior segmental artery with a yin-yang pattern measuring approximately 25 x 15 mm at the lower pole of the left kidney (arrow). Author's File.

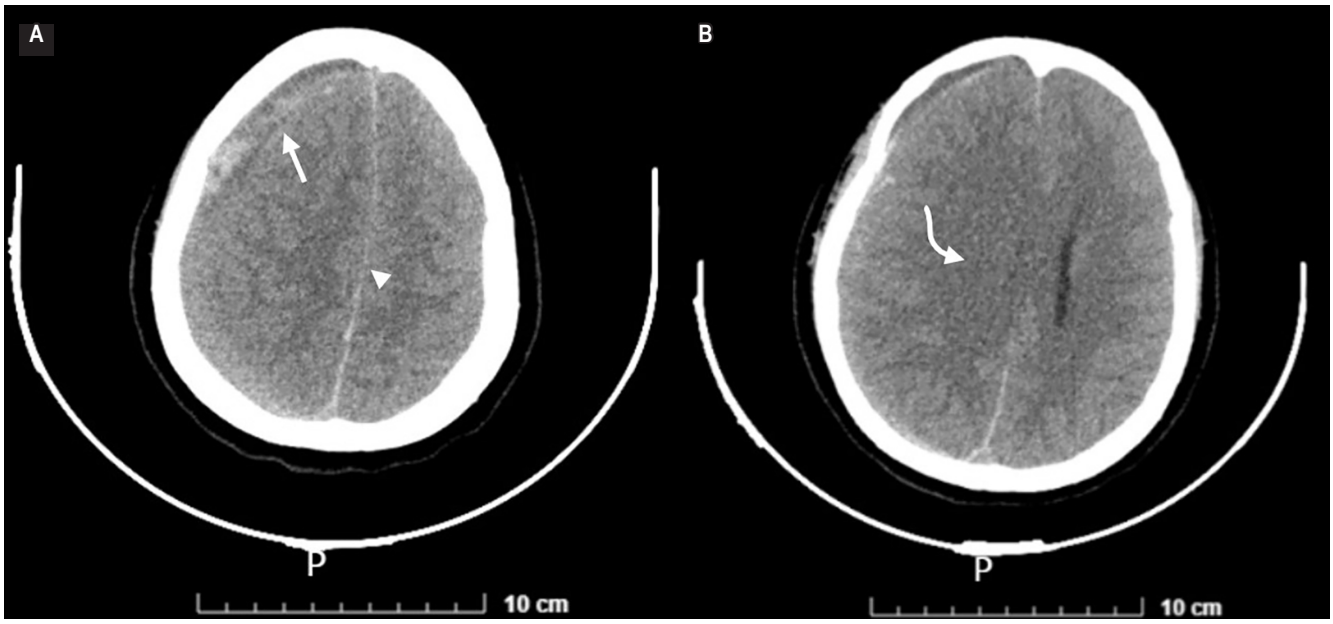


Figure 4. Cranial Computed Tomography. **A.** Chronic right subdural hematoma (arrow), with a 12 mm midline shift (arrowhead). **B.** Ventricular involvement (curved arrow). Author's File.

of the periduodenal vitelline veins, leading to the absence of the portal vein and diversion of blood flow into the systemic circulation[9]. It results from incomplete vascular remodeling between the symmetrical embryonic hepatoperihepatic circulations and the asymmetrical fetal system[10]. Another possible origin is agenesis of the ductus venosus, which predisposes the umbilical vein's oxygenated blood to bypass the liver and flow directly to the heart via persistent abnormal vessels, resulting in abnormal shunting and hypoplasia of the portal system[11].

Abernethy malformation is a rare and underdiagnosed condition, with few reported cases. From its first description in 1793 until 2021, 323 cases have been documented, with type I malformation being more prevalent than type II[12]. Approximately 70% of diagnosed cases occur in patients under 18 years of age[13], with an estimated global incidence of 2.5 cases per year[14]. In Ecuador, a case involving a 6-year-old child was reported in 2017[15].

In this case, the dermatological concern was spider veins consistent with hemangioma, a physical sign previously reported by Kothari[16] in two pediatric cases. This finding led to an incidental imaging diagnosis via computed tomography (CT). Subsequently, the patient experienced an episode of syncope, a symptom also described by Lin and colleagues[17] in two reported cases. Other documented clinical presentations of Abernethy malformation include cholestasis, precocious puberty, and growth restriction[18].

In our patient, venous hypoplasia was detected via ultrasound and diagnosed incidentally through 3D reconstruction angiography. El-Medany and colleagues[19] reported cardiological presentations as the reason for consultation in their cases and used magnetic resonance imaging (MRI), computed tomography (CT), and ultrasound for diagnosis.

The complexity and rarity of this condition initially led to a clinical suspicion of portal vein thrombosis, as seen in the case of type I Abernethy malformation reported by Tamiru and colleagues[20], since the portal vein may appear poorly visualized in imaging studies, as occurred in both cases. Differential diagnoses, such as systemic lupus erythematosus and antiphospholipid syndrome, were also considered.

In this case, splenorenal collaterals, an aneurysm of the inferior segmental artery of the left kidney, and a shunt connecting the splenic and superior mesenteric veins to the inferior vena cava were identified. Therefore, other arteriovenous malformations must also be detected, similar to the case reported by Păcurar and colleagues[21], which involved type Ib Abernethy malformation with a hepatic artery originating from the superior mesenteric artery, hepatic veins forming a short common trunk before draining into the inferior vena cava, and a supernumerary right renal artery arising from the aorta.

The Morgan and Superina classification system[3] was used; however, other authors categorize Abernethy malformations into three types. Kobayashi and colleagues[22] des-

cribed types A, B, and C based on the location of portal blood drainage into the inferior vena cava, renal vein, or inferior mesenteric vein, respectively. Kanazawa and colleagues[23] classified hypoplasia according to its severity, as observed on angiography, into mild, moderate, or severe types.

Shunt closure can be performed via surgical ligation or endovascular balloon occlusion. However, conservative management may be appropriate for asymptomatic patients or those with incidental diagnoses[24]. Surgery is indicated in children with clinically significant disease, as spontaneous closure after the age of two years is unlikely, and complications can be severe[25]. The theoretical risk of portal vein thrombosis can be mitigated with anticoagulants such as heparin, although due to clinical heterogeneity, treatment must be individualized[26].

Early surgical intervention in pediatric cases leads to partial or complete regression of tumorous lesions, if present. Therefore, increased awareness of less common clinical presentations of portosystemic shunts is essential for early diagnosis[18], which can reduce complications and associated morbidity, improve the patient's quality of life, and optimize public healthcare system expenditures.

CONCLUSIONS

Abernethy malformation is rare in adult patients and, in this case, was an incidental diagnosis. The dermatological signs guided its discovery; however, although other cases of patients with Abernethy malformation and hemangioma have been reported, there is insufficient evidence to recommend this clinical feature as a reliable physical sign. This is further complicated by the existence of other conditions with similar clinical or imaging presentations. For these reasons, we encourage future researchers in this specific area to investigate whether a causal relationship exists and to provide a pathophysiological explanation, as well as to identify other vascular anomalies that may coexist in patients with extrahepatic portosystemic venous shunts.

Informed Consent and Patient Perspective

Written informed consent was obtained from the patient to publish her data anonymously. The patient reports an understanding of her condition and its severity and is currently willing to adhere to the planned clinical follow-up.

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Conflict of Interest

The authors declare no conflicts of interest regarding the research, authorship, or publication of this article.

Authorship Statement

All authors contributed to the collection, analysis, and interpretation of the data, as well as to the design and drafting of the content, and they approved the final version to be published.

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