

# Pulmonary Arterial Hypertension in a Patient with a Portosystemic Shunt: Diagnostic Challenge



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## INTRODUCTION

Portosystemic shunt, especially congenital extrahepatic portosystemic shunt (CEPS), is a rare anomaly of the mesenteric vasculature in which the intestinal and splenic veins bypass the liver and drain directly into the inferior vena cava (IVC), the left hepatic vein, or the left renal vein.<sup>1,2</sup> This uncommon disease is frequently associated with other malformations that affect mainly women. CEPS, known as the “Abernethy malformation,” was first described by Abernethy in 1793.<sup>3,4</sup>

Pulmonary arterial hypertension (PAH) is a well-known complication of portal hypertension in chronic liver diseases, and when PAH coexists with congenital shunts, the condition is extremely rare.<sup>5,6</sup> There are no statistics in the medical literature; only a few isolated cases of PAH have been reported as a result of extrahepatic portosystemic shunts.<sup>7</sup> According to physiopathologic theory, when vasoactive substances present in the intestinal circulation (e.g., serotonin, histamine, estrogen, glucagon) bypass the liver without being metabolized and pass through a CEPS, the pulmonary parenchyma is directly affected.<sup>8</sup> The result is PAH caused by the induction of long-lasting pulmonary vasoconstriction and increased pulmonary vascular resistance due to vasoactive toxins that have not been metabolized correctly.<sup>9,10</sup>

In this report we describe a severe case of pediatric PAH with CEPS. This type of pathology is an uncommon cause of PAH in children, and we emphasize the need to arrive at an early diagnosis to permit earlier resolution, in order to avoid the consequences of a late-resolved shunt.

## CASE DESCRIPTION

We present the clinical case of a 15-year-old female patient who was referred to our pediatric cardiology service with clinical suspicion for an atrial septal defect for diagnostic evaluation and follow-up.

The patient was admitted in 2017. There were no perinatal records, and according to the patient’s history, she had irregular visits with health care practitioners, had no family doctor, and was attending school according to her age. She was receiving contracep-

tive treatment because of hormonal alteration during menarche. The patient was sedentary and did not report dyspnea at rest, but she had mild exertional dyspnea (New York Heart Association functional class II). The patient was 15 years of age, weighed 62 kg, was 163 cm tall, had an oxygen saturation level of 97%, and had blood pressure of 110/70 mm Hg. Cardiac examination revealed barrel chest, present and symmetric peripheral pulses, a normal first heart sound, a second heart sound with increased intensity with a diastolic murmur in the pulmonary area, and a systolic murmur at the apex. Pathologic electrocardiography and chest radiography were performed (Figure 1).

Color Doppler echocardiography was performed and demonstrated pulmonary hypertension with tricuspid regurgitation of 68 mm Hg, pulmonary regurgitation of 43 mm Hg, severe pulmonary artery (PA) dilatation of 4.3 cm, right atrial size of 17 cm<sup>2</sup>/m<sup>2</sup>, and IVC dilatation to 2.2 cm with adequate collapse. The estimated right ventricular systolic pressure was 82 mm Hg, which indicated severe PAH. The left ventricle was D shaped, with a normal left ventricular ejection fraction. There was no evidence of pericardial effusion. Because of the PAH diagnosis and because we could not rule out an atrial septal defect, it was decided to perform cardiac catheterization to initiate the PAH protocol (Figure 2).

On cardiac catheterization, mean pulmonary arterial pressure was 60 mm Hg, pulmonary vascular resistance was 14 Wood units/m<sup>2</sup>, and results of the pulmonary vasoreactivity test were negative. Consequently, PAH was diagnosed on right heart catheterization. On angiography, decreased arborization of the pulmonary arteries was seen in both lungs. Aortography showed the venous drainage of the mesenteric arteries, flowing into the portal vein (PV) and directly into the IVC. There was no evidence of congenital heart disease or pulmonary thromboembolism (Figure 3).

Thoracic computed tomography and abdominal computed tomography were performed. A hypodense parietal image was seen in the left branch bifurcation, a finding that could indicate a low probability of chronic pulmonary thromboembolism (Figure 4). The liver was slightly increased in size, with a heterogeneous segment intravenous structure with central enhancement after the administration of intravenous contrast, measuring 1.5 × 1.3 × 1.5 cm, associated with the absence of intrahepatic portal branches. Computed tomography demonstrated the presence of a type 1b extrahepatic portosystemic shunt (with the PV connected directly to the IVC; Figure 4).

Abdominal echocardiography showed the portosystemic shunt with reduction of the PV at the hepatic level.

We confirmed the diagnosis as PAH caused by CEPS (Abernethy type 1b). Hemodynamic intervention was planned but could not be performed because of the large size of the CEPS, and therefore surgical closure was performed. In January 2019 the patient was discharged after recovering from the surgical correction of her CEPS.

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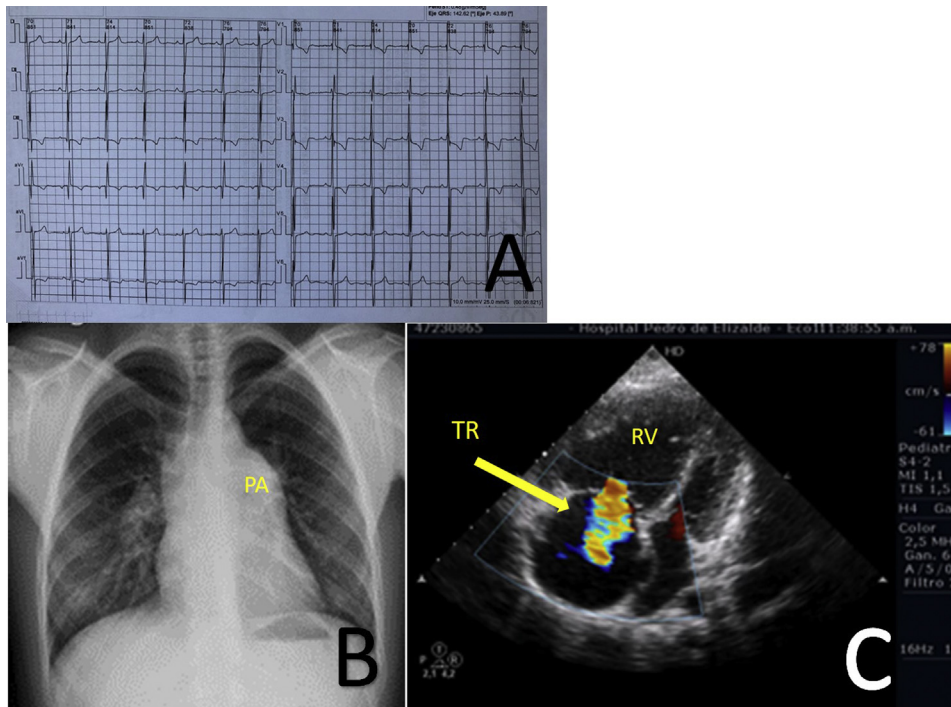
Keywords: Pediatric pulmonary hypertension, Pulmonary arterial hypertension, Congenital extrahepatic portosystemic shunt, CEPS, Abernethy malformation

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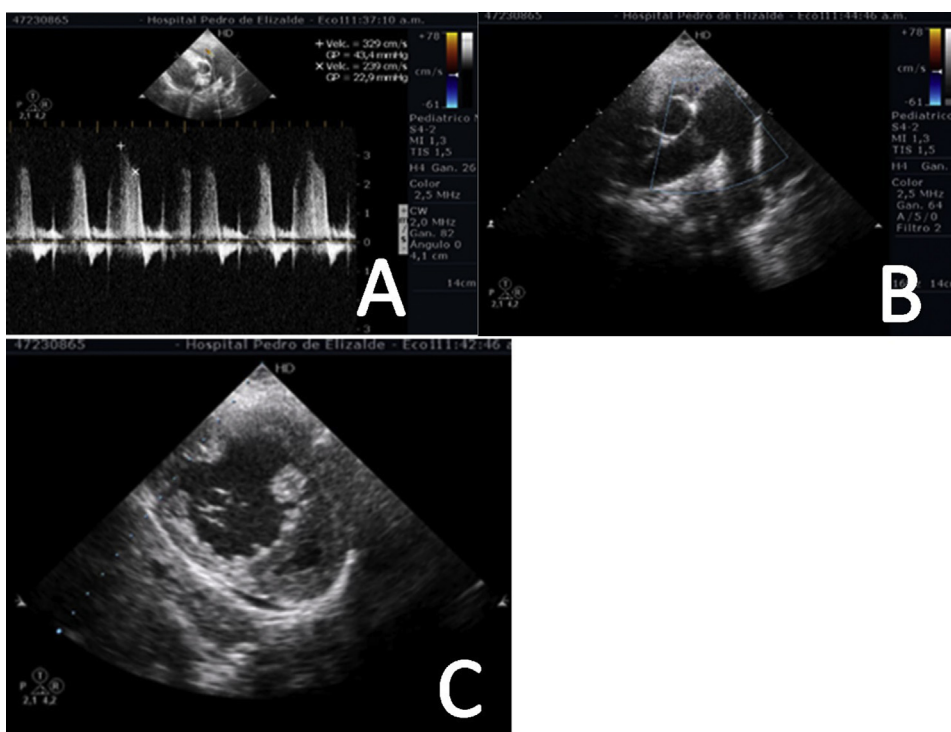
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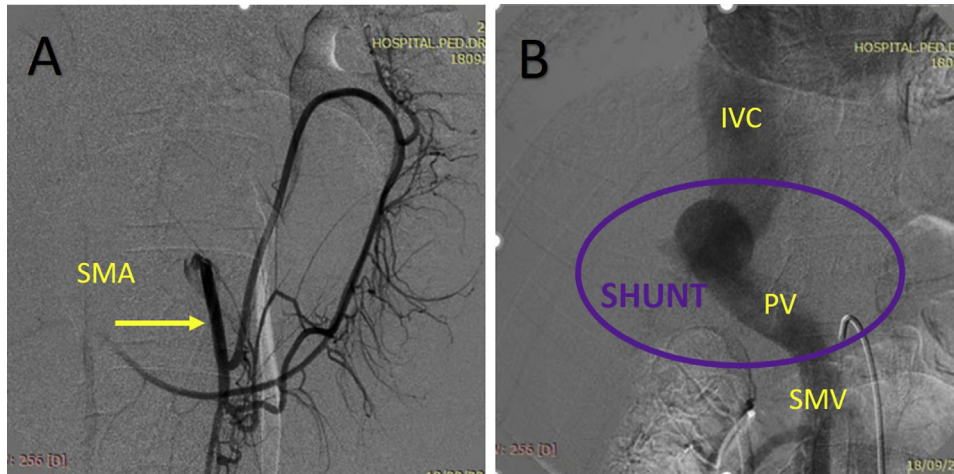
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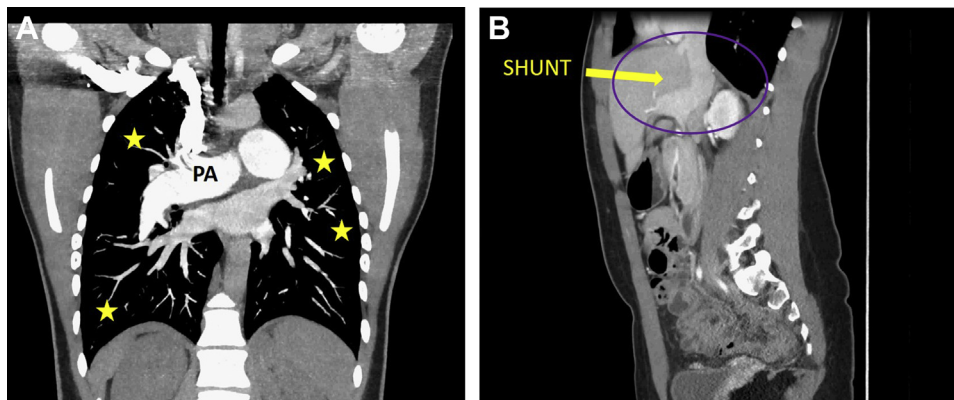
**Figure 1** Initial studies. **(A)** Electrocardiography demonstrated regular sinus rhythm with right-axis QRS  $120^\circ$  and right ventricular overload. **(B)** Chest radiography showed decreased vascularity of the distal portion of both pulmonary arteries and PA truncus dilatation. **(C)** Echocardiography (apical four-chamber view, color Doppler) depicting significant tricuspid regurgitation (TR). RV, Right ventricle.



**Figure 2** Echocardiography. **(A)** Short-axis view at large vessel level with truncus and PA dilatation. **(B)** Doppler of pulmonary regurgitation. **(C)** Short-axis view of the left ventricle, with deviation of the interventricular septum and compression of the left ventricle by the right ventricle.



**Figure 3** Cardiac catheterization. **(A)** Arteriography of the superior mesenteric artery (SMA). **(B)** Levophase of the same image displays the Abernethy shunt, where the superior mesenteric vein (SMV) drains into the PV, and the PV drains directly into the IVC.



**Figure 4** Angiotomography. **(A)** Thoracoabdominal front view: pulmonary arborization pathology and severe PA dilatation. **(B)** Side abdominal view: the abdominal venous return drains directly into the IVC through the PV, without intrahepatic circulation, showing the Abernethy shunt. *Star* indicates severe decrease in pulmonary vasculature.

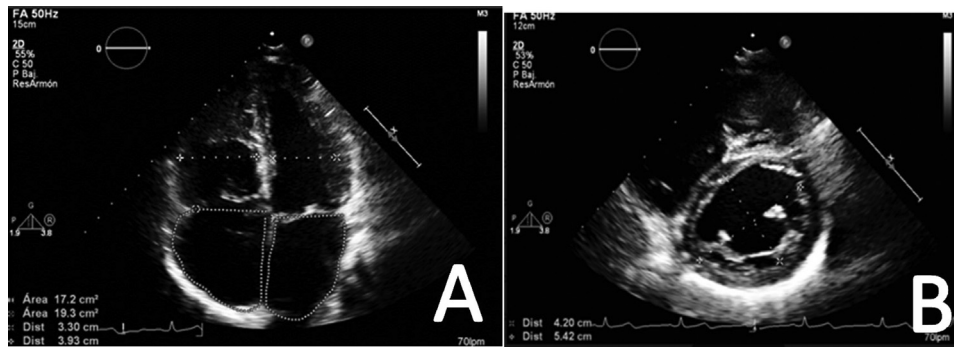
The patient is asymptomatic (New York Heart Association functional class I) 2 years after her diagnosis and 6 months since surgery. Follow-up cardiac examination revealed barrel chest, a calm precordium, and present and symmetric peripheral pulses. The first heart sound was normal, the second heart sound had increased intensity, and no murmurs were appreciated. Color Doppler echocardiography revealed mild PAH with tricuspid regurgitation of 30 mm Hg, pulmonary regurgitation of 19 mm Hg, right atrial dimension of 12 cm<sup>2</sup>/m<sup>2</sup>, no ventricular dilatation (left ventricle 39 mm, right ventricle 31 mm), PA ring size of 30 mm, and truncus size of 50 mm. Severe dilation of the PA and branches remains (Figure 5).

## DISCUSSION

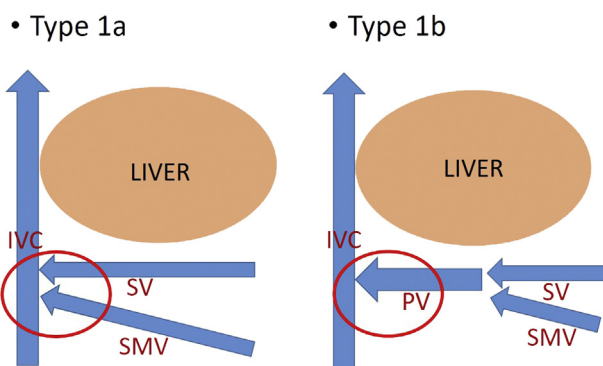
Congenital portosystemic venous shunts are best classified as intrahepatic or extrahepatic. Intrahepatic connections are created between branches of the portal vein, after its division, and the hepatic veins of the IVC. In CEPS the anastomoses are established outside the liver between the portomesenteric vasculature, before the division of the PV and systemic vein.<sup>4</sup>

CEPS, also known as Abernethy malformation, is classified into two types according to the presence or absence of portal blood flow within the liver parenchyma (Figure 6).<sup>3,4</sup> In type 1 (Abernethy type 1 or end-to-side shunt), there is total absence of intrahepatic portal flow. The entire portal venous supply drains into the IVC, with absence of the intrahepatic PV. This represents a congenital atresia of the PV, in which the superior mesenteric and splenic veins can be attached to the IVC separately (type 1a) or by confluence (type 1b). Type 1 has a female prevalence and is often complicated by congenital heart defects, liver masses, and gastrointestinal and vascular abnormalities.<sup>2</sup> In type 2 (Abernethy type 2 or side-to-side shunt), portal flow is partially preserved. There is PV hypoplasia with a partial derivation of the portosystemic shunt. Type 2, which is even rarer, has a male prevalence and usually does not involve associated malformations.<sup>2</sup>

CEPS can cause a broad spectrum of clinical manifestations. PAH is a major complication of CEPS. In patients with CEPS, a long-standing pulmonary vasoconstriction may occur because of increased levels of humoral substances in the lung tissue. When CEPS is detected, cardiac catheterization should be considered to determine whether the shunt can be corrected by shunt closure. This pathologic state of portosystemic venous shunt may be cured if the shunt vessel can be closed.<sup>7</sup>



**Figure 5** Late postoperative echocardiography. **(A)** Apical four-chamber view showing improved right ventricular cavity size. **(B)** Left ventricular short-axis view with adequate morphology.



**Figure 6** Different types of CEPS. Complete portosystemic shunts that do not perfuse the liver via the PV are defined as type 1. They are subclassified into type 1a (the splenic vein [SV] and superior mesenteric vein [SMV] drain separately) and type 1b (both veins drain together after joining to form a common trunk). Partial shunts with some remaining degree of portal perfusion to the liver are defined as type 2. Modified with permission from Yi *et al.*<sup>1</sup> and Alonso-Gamarra *et al.*<sup>4</sup>

## CONCLUSION

In pediatric patients, if the cause of PAH is unknown, all possible causes of secondary PAH should be considered, including the most uncommon ones. CEPS is rare, especially in pediatric patients, and should be ruled out when facing this type of patient. Resolution can be improved, and the damage caused by PAH may even be reversed.

Improvement of parameters and the consequent reduction of pulmonary vascular resistance after the correction was applied indicate that vasoconstriction by vasoactive substances played an important role in our patient.

In the case presented here, the large PA dilatation and the noninvolution of the diameter should be followed up, with

further discussion of the need for anticoagulation and surgical approach.

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