CASE REPORT

Abernethy Malformation Masquerading as Congenital Heart Disease: A Boy with Cyanosis, Clubbing and Hypoxia

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Competing interests

The authors declare that they have no competing interests.

Abbreviations

CPSS: Congenital portosystemic shunt; IVC: Inferior vena cava; HPS: Hepatopulmonary syndrome; PV:Portal vein.

Authors' contributions

DP wrote and designed the manuscript. RA and GC: provided procedural details and the radiological analyses. AV, AK and AK: provided intellectual input. All authors read and approved the final manuscript.

Consent for publication

Written informed consent was obtained from the patient's parents for publication of this case report including clinical data and the images.

INTRODUCTION

Abernethy malformation is an extrahepatic congenital portosystemic shunt (CPSS), an extremely rare condition characterized by the diversion of portal blood away from the liver. In this anomaly blood from splanchnic circulation drains into the inferior vena cava (IVC) through a shunt bypassing the liver, thereby causing an alteration in the metabolism of pulmonary vasoactive substances resulting in pulmonary vasodilatation, diffusion-perfusion mismatch, and eventually hypoxemia (1–3). The clinical manifestations of Abernethy malformation are highly variable and can be asymptomatic or symptomatic due to the shunting of blood such as hepatic encephalopathy or hepatopulmonary syndrome (HPS). Abernethy malformation can be classified into two types. Type I is defined by an absence of intrahepatic portal veins, and a lack of liver perfusion with portal blood. In type Ia, superior mesenteric and splenic veins drain separately into the IVC and in type Ib splanchnic blood drains via the common trunk into IVC (4). Type II is defined by side to side connection of the portal vein (PV) with IVC in which hypoplastic intrahepatic portal vein supplies some degree of portal flow to liver parenchyma. Prolonged untreated severe hypoxemia can result in irreversible changes in the pulmonary vasculature and its consequences become incapacitating for the patient. Early shunt closure resolves hypoxemia and clinical symptomatology.

Case

We present case of a 9-year-old boy from rural background with a height of 112 cm (<3rd centile) and weighing 19 kg (<3rd centile) who presented with cyanosis, clubbing, dyspnoea, and restricted growth since 4 years of age. Physical examination on admission revealed central cyanosis and digital clubbing with oxygen saturation on pulse oximetry (SpO_2) of 60% on room air. Blood investigations showed an elevated haemoglobin level of 15.5 g/L (normal range 11.5–15 g/L), a normal liver enzyme profile with aspartate aminotransferase (AST) 32 U/L (normal range 17–59 U/L), alanine aminotransferase (ALT) 19 U/L (normal range 21–72 U/L). He had an elevated serum ammonia level of 77 μmol/L (normal range 7–30 μmol/L) but no evidence of hepatic encephalopathy was found. His coagulation profile was normal with an international normalized ratio (INR) of 1.39; and activated partial thromboplastin time (APTT) of 30 s (normal range 24.6–29.9 s).

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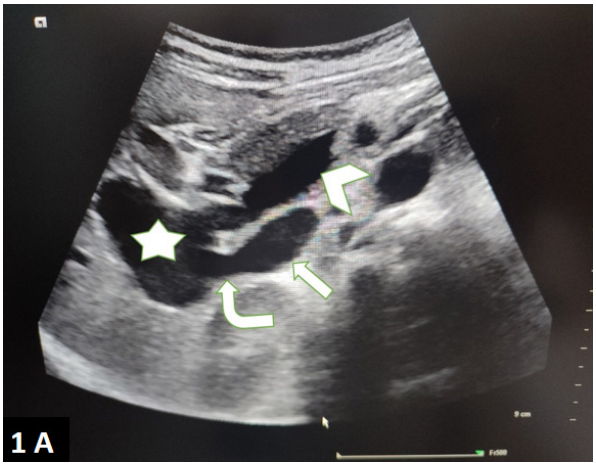
A chest X-ray showed mild cardiomegaly with increased pulmonary vascular markings. ECG was unremarkable. Echocardiography revealed features of mild left-sided volume overload and dilated left atrium, after that a contrast echocardiography was performed with agitated saline which showed the appearance of micro-bubbles in the left atrium after four cardiac cycles, suggesting pulmonary arteriovenous communication.

Doppler ultrasound showed dilated extrahepatic main PV with hypoplastic intrahepatic portal venous branches arising from the dilated segment and a large communicating shunt between IVC and main portal vein **(Figure 1A)**. Computed tomography (CT) angiography confirmed the doppler findings and diagnosis of type II Abernethy malformation was made **(Figure 1B)**. Hepatic veins were normal on Doppler and CT angiography.

Interventions

The patient was taken under general anaesthesia for endovascular management of the CPSS closure. Both jugular and femoral access were taken. The shunt was cannulated from both sides (jugular and femoral). Balloon occlusion of the shunt was performed and multiple catheter angiograms were taken to establish the patency of the intrahepatic portal venous branches **(Figure 2A)**. After documenting the patency of intrahepatic portal venous branches and good intrahepatic circulation, the decision of shunt closure was taken. Angiogram showed a shunt with diameter of 12 mm and length of 7 mm. The shunt closure was done with a 16 mm diameter and 8 mm long vascular plug device (Cera, Lifetech Scientific Corporation, China). The device was deployed across the shunt via the femoral route and angiograms were taken from the jugular route, which showed a visualization of the intrahepatic portal branches with good hepatic portal circulation and closure of the shunt **(Figure 2B)**.



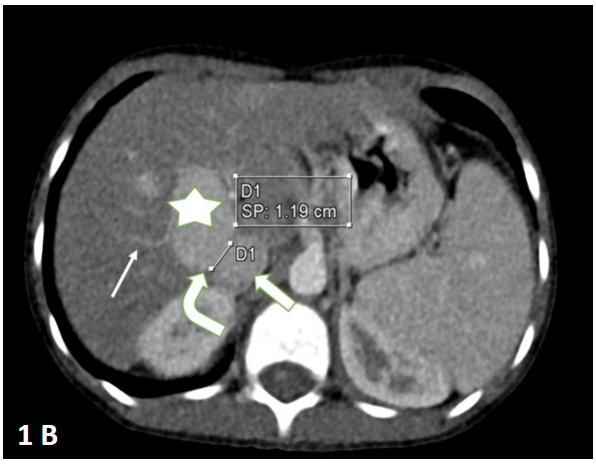
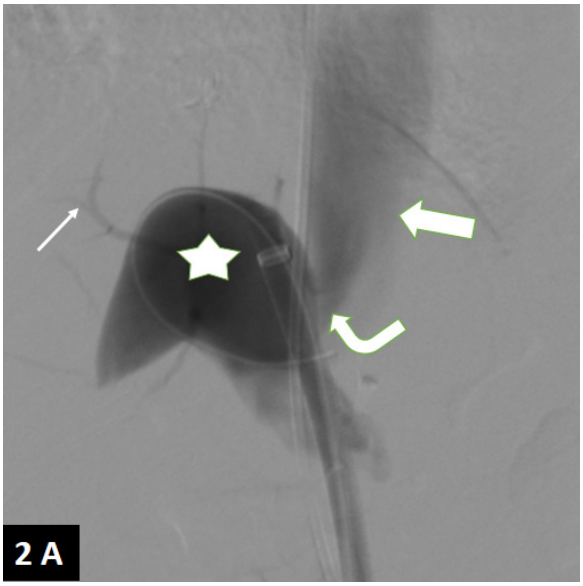


Figure 1:

(A) Ultrasonography image showing side to side communication (curved arrow) between main portal vein (star) & IVC (solid arrow) and also the patent superior mesenteric vein (arrow head).

(B) Corresponding CT scan image at the similar level showing side to side communication (curved solid arrow) between main portal vein (star) & IVC (solid arrow), shunt size (line) and also the patent hypoplastic intrahepatic portal venous branches (simple arrow).



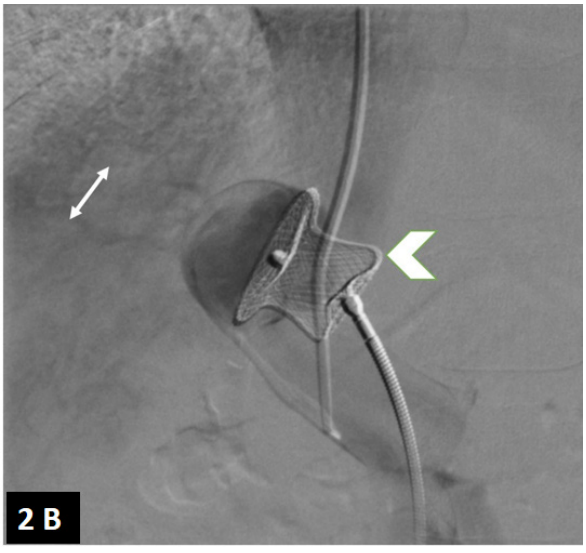


Figure 2:

(A) Digital substraction angiogram showing side to side communication (curved arrow) between main portal vein (star) & IVC (solid arrow) along with patent hypoplastic intrahepatic portal venous branches (simple arrow).

(B) Digital substraction angiogram showing post deployment of the vascular plug (arrow head) showing stasis and hepatic parenchymal blush (double arrow).

Discussion

In the presence of cyanosis and clubbing clinicians usually think about congenital heart disease until proven otherwise; however, there are uncommon conditions like hemoglobinopathy, Abernethy malformation and diffuse pulmonary arteriovenous fistula that can also result in cyanosis. Most of these patients end up in cardiothoracic surgery or cardiology clinics. Hence, special attention and detailed workup is required in children with to children with unexplained cyanosis where cardiac causes have been ruled out. Similarly, our case was misdiagnosed, mistaken as well as managed managed as cyanotic heart disease for many years before coming to our hospital. In resource poor settings, the diagnosis of CPSS can be missed or delayed due to lack of awareness, resources and technical expertise. However, now a days it can be picked up easily due to advancements in imaging techniques. Diagnosis of CPSS is often delayed and difficult due to varied clinical presentations. These patients reach the health care facility when complications have set in. Abernethy malformation may lead to severe complications such as liver tumours, hepatic encephalopathy, HPS, or pulmonary hypertension. The HPS is one of the common complications and has been reported in at least 20 children with Abernethy malformation (5). HPS is a triad of liver diseases, arterial hypoxemia and pulmonary vascular dilatation. Although HPS typically develops in the setting of cirrhosis and portal hypertension, it may also occur in the absence of parenchymal liver disease in association with portosystemic shunting. It is believed to be attributed to the exposure of the pulmonary vascular bed to vasoactive mediators, which are derived from the intestinal tract entering the systemic circulation without being metabolized in the liver (3). Prolonged untreated cases result in pulmonary vasodilatation, pulmonary arteriovenous fistula, diffusion-perfusion mismatch, and eventually hypoxemia.

Treatment of CPSS depends on the type, the presenting symptoms, complications and comorbidity. Treatment may vary from endovascular occlusion of the shunt, surgical correction to even liver transplantation (5, 6). The optimal timing of the shunt closure is not defined in pediatric patients with CPSS (7). It is proposed that even in the absence of overt symptoms, early intervention prevents pulmonary and other complications (8).

Liver transplantation is the treatment of choice for Abernethy malformation type I due to the lack of an apparent intrahepatic portal venous channel. But recently published experiences by several authors (9) point out that many patients with CPSS type I malformations might have small portal vein radicals which cannot be seen on ultrasonography but could be visualized on shunt angiography. The balloon occlusion test of the fistula can also be done to decide on a single stage or a two-staged shunt closure procedure (9).

Transcatheter occlusion/surgical ligation of the shunt is the therapeutic option for type II Abernethy malformation (9). The successful transcatheter closure of CPSS has been conducted in children with type II Abernethy malformation with good outcomes (9). In general, exercise tolerance and resting oxygen saturation of these patients are increased after the procedure. However, symptoms like exertional dyspnea, cyanosis, and pulmonary hypertension are only partially improved in some patients when treatment is delayed. Shunt closure results in restoration of intrahepatic portal blood flow in most patients (9). Symptomatic improvement and stabilization of pulmonary, cardiac and neurological functions are seen in patients post shunt closure (9).

In our case, device closure of the shunt was successful, as his serum ammonia levels normalised over 1-month and hypoxemia got corrected over 6 months. A limitation of our case was that, we did not measure the portal venous pressure before shunt closure. Ideally, a balloon occlusion of shunt followed by measurement of portal venous pressure should be done before shunt closure as it can help in deciding whether a one- or two-stage procedure is needed to decrease post-procedure liver related morbidity. At a follow-up of 1 year, his growth and exercise tolerance improved, and resting oxygen saturation increased from 60 to 98% on room air. No complication was noted during a 1-year follow-up after the shunt occlusion. Early diagnosis and appropriate management of Abernethy malformation may lead to an improved prognosis.

In conclusion, Abernethy malformation is a rare cause of cyanosis and clubbing in children. Clinicians must suspect Abernethy malformation complicated with HPS in children in the setting of unexplained cyanosis, clubbing, dyspnea, abnormally high serum ammonia levels and dilated portal veins with abnormal drainage after demonstration of portosystemic fistulas on imaging.

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