ACG CASE REPORTS JOURNAL

CASE REPORT | PEDIATRICS

A Congenital Portosystemic Shunt in a Child With Heterotaxy, Situs Inversus, Polysplenia, and Interrupted Inferior Vena Cava With Azygous Continuation

Victoria Carvajal, DO[1], Saigopala Reddy, BSPH[2], Vani Gopalareddy, MD[3], Adrienne Bean, MD[4], and Gonzalo Wallis, MD[5]

[1]*Department of Pediatrics, Atrium Health, Levine Children's Hospital, Charlotte, NC*
[2]*University of North Carolina Chapel Hill Medical School, Raleigh, NC*
[3]*Department of Pediatric Gastroenterology, Hepatology, and Nutrition, Atrium Health, Levine Children's Hospital, Charlotte, NC*
[4]*Department of Pediatric Radiology, Atrium Health, Levine Children's Hospital, Charlotte, NC*
[5]*Department of Pediatric Cardiology, Atrium Health, Levine Children's Hospital, Congenital Heart Center, Charlotte, NC*

ABSTRACT

Congenital portosystemic shunts are rare vascular malformations in which portal venous blood from the intestines and spleen bypasses the liver and diverts directly into the systemic circulation through abnormal vessels. We report a case of a 4-year-old girl with heterotaxy syndrome, polysplenia, and situs inversus presenting with persistent hypoxemia who was found to have pulmonary arteriovenous malformations (PAVMs) and hypoxemia secondary to a congenital portosystemic shunt. Management of this patient's PAVMs involved endovascular occlusion of the portosystemic shunt with subsequent resolution of hypoxemia. PAVMs secondary to extrahepatic portosystemic shunt should be explored as a cause of progressive cyanosis in children with heterotaxy, polysplenia, and interrupted inferior vena cava with azygous continuation.

KEYWORDS: congenital portosystemic shunt; pulmonary arteriovenous malformation; hepatic factor; congenital heart disease

INTRODUCTION

Congenital portosystemic shunts (CPSSs) are rare vascular malformations in which portal venous blood from the intestines and spleen bypasses the liver and diverts directly into the systemic circulation through abnormal vessels. CPSS is estimated to affect 1 in 30,000 newborns.[1] CPSSs are frequently associated with other congenital anomalies, such as patent ductus arteriosus, tetralogy of Fallot, situs ambiguus, polysplenia, malrotation, biliary atresia, and genitourinary malformations.[2] Acquired pulmonary arteriovenous malformations (PAVMs) can also be associated with CPSS. These acquired PAVMs are low-resistance abnormal communications that develop between a pulmonary artery and a pulmonary vein, bypassing the pulmonary capillary bed, thereby creating a right to left shunt and subsequent hypoxemia. We report a case of acute hypoxemia and PAVMs in a patient with heterotaxy, polysplenia, and situs inversus found to have a large extrahepatic portosystemic shunt.

CASE REPORT

A 4-year-old girl with a history of heterotaxy with situs inversus, polysplenia, interrupted inferior vena cava (IVC), repaired atrioventricular canal defect, and resected subaortic membrane presented to the emergency department with hypoxemia at home noted to be 78%. Physical examination findings were notable for comfortable tachypnea and digital clubbing. The patient was placed on 4 L of high-flow nasal cannula, and chest radiograph obtained was unrevealing other than cardiomegaly. She was admitted for hypoxemia and further evaluation. Echocardiogram was obtained, which demonstrated normal biventricular function. The left heart had a multilevel left ventricular outflow tract obstruction with thickened left-sided atrioventricular valves with mild stenosis, mild regurgitation, and subsequent moderate left atrial dilation. There was also mild right-sided atrioventricular valve regurgitation with a gradient of 18 mm Hg, not suggestive of pulmonary hypertension. The

ACG Case Rep J 2023;10:e01201. doi:10.14309/crj.0000000000001201. Published online: December 4, 2023

Correspondence: Victoria Carvajal, DO (victoria.carvajal@atriumhealth.org).

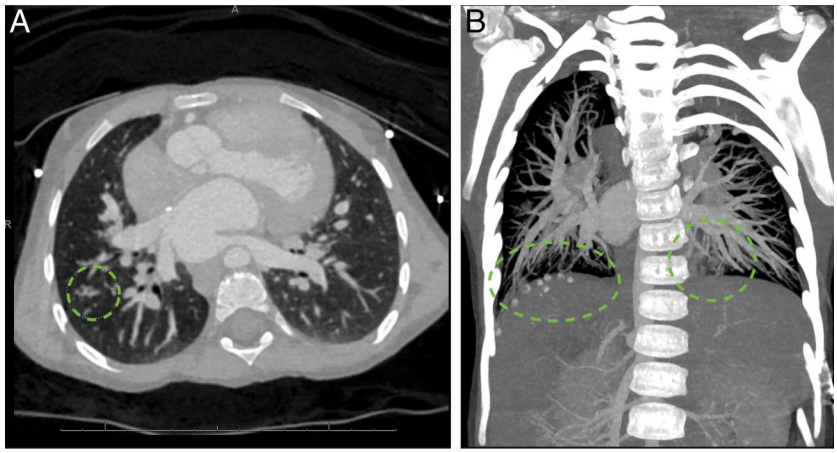

Figure 1. (A) Axial and (B) coronal maximal intensity projection computed tomography angiographic images demonstrating bilateral lower lobe supra-diaphragmatic peripheral arteriovenous malformations as outlined by dashed green circles.

echocardiogram did not reveal a significant cause of hypoxemia, and cardiac computed tomography (CT) yielded multiple PAVMs and dilated and tortuous pulmonary vessels were visualized (Figure 1). A massively dilated azygos vein was identified, possibly receiving drainage from the portal venous system. Subsequent abdominal ultrasound could not distinguish portosystemic shunt only, noting patent portal and hepatic vasculature. Abdominal CT with contrast detected the azygous vein in continuation with the IVC with anomalous drainage of the portal veins and splenic vasculature into the azygous vein described as a massive portosystemic shunt (Figure 2). Primary hepatic veins drained to the right atrium, and the liver surface was enlarged and nodular. The patient was diagnosed with a congenital extrahepatic portosystemic shunt as the portal and splenic vein emptied into the azygous vein. Laboratory test results were significant for hyperammonemia of 129, for which she was started on lactulose. The patient was discharged home on 2–3 L of supplemental oxygen with goal saturations above 80% and furosemide and chlorothiazide for underlying congenital heart disease.

A few weeks later, a liver biopsy was obtained and demonstrated scattered small portal veins and rare portal areas demonstrating enlarged hepatic arteries with multiple hepatic artery profiles and patchy mild perisinusoidal/perivenular fibrosis (Figure 3).

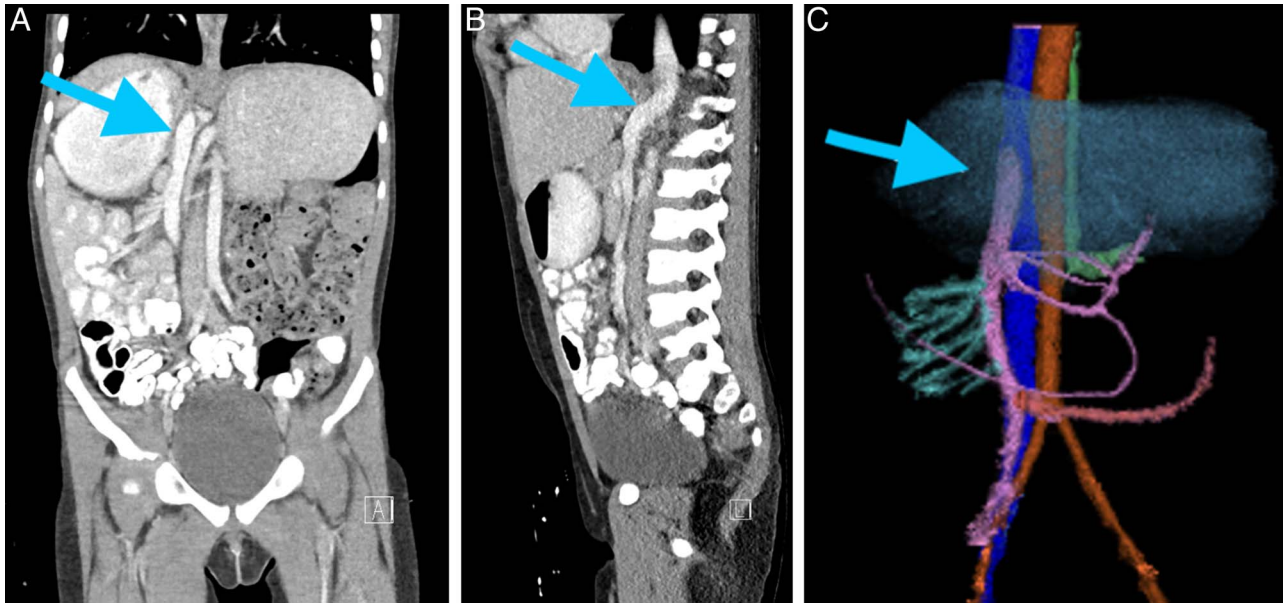

Figure 2. Coronal (A) and sagittal (B) computed tomography angiographic images with 3D volume rendered reconstruction (C) demonstrating the anomalous drainage of the splanchnic veins to the systemic venous system bypassing the liver with azygous continuation of the inferior vena cava. No portal vein is present. The draining mesenteric veins form an anomalous vein that inserts into the retro-hepatic azygous continuation of the inferior vena cava creating a portosystemic as indicated by blue arrows.

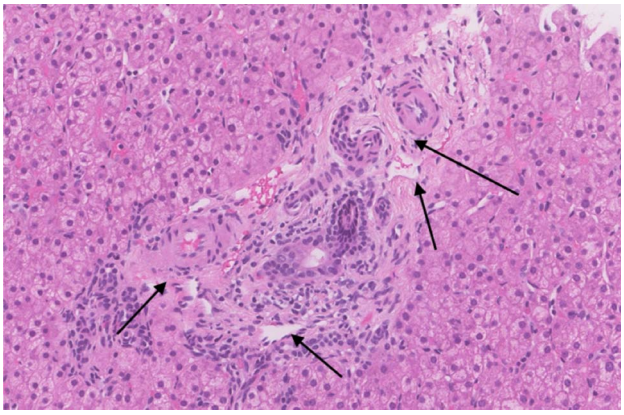

Figure 3. Liver biopsy demonstrating small patent hepatic portal veins as indicated by arrows.

There was no evidence of definitive loss of portal veins or areas suggestive of fibrous obliteration of portal veins, classifying the shunt as an extrahepatic portosystemic shunt type II. Azygous and portal venogram was performed. Portal manometry demonstrated a 12 mm Hg pressure gradient of the portal vein with shunt test occlusion. A 14 mm × 10 mm Amplatzer plug was placed by interventional radiology, with intravascular ultrasound confirming good placement of the occlusion device. Interval abdominal magnetic resonance angiography 1 month later continued to demonstrate shunt closure with an increase of the portal venous diameter from 3–4 mm to 8–10 mm. At the 2-month follow-up, hyperammonemia resolved. Hypoxemia and activity tolerance improved, and daytime home oxygen was discontinued 10 days after CPSS occlusion.

DISCUSSION

This case highlights the complex relationship between the vascular system of the gastrointestinal tract, liver, and pulmonary vasculature. The history of heterotaxy, situs inversus, and interrupted IVC with PAVMs should clue in providers to add CPSS to their differential diagnosis because these are associations that have been previously recognized in patients with CPSS.[1] In the literature, there are small studies noting an increased prevalence in patients with congenital heart disease.[3] It is important to detect CPSS early because cardiopulmonary sequelae associated with CPSS, such as high-output heart failure, are among the most severe and life-threatening for these patients.[4]

Our patient's presentation and physical examination was notable for dyspnea at rest and was found to have digital clubbing, which can also be seen with telangiectasias, palmar erythema, and hyperemic lips in cases of CPSS. Our patient's symptomatology was found to be related to the right to left shunting that occurs at the level of the PAVMs with subsequent hypoxemia. Pulmonary hypertension is a rare complication of PAVMs and is more often seen in hepatic AVMs due to excessive preload or in cases associated with hereditary hemorrhagic telangectasias. Other signs of cardiopulmonary presentation of a patient with a CPSS include portopulmonary hypertension and syncope related to high-output failure or hoarseness. CPSS can also present with neonatal cholestasis and hepatic complications such as focal nodular hyperplasia, hepatocellular adenoma, hepatoblastoma, or hepatocellular carcinoma. Once CPSS is suspected, the current recommendation for screening is an abdominal ultrasound with Doppler.[4] It is standard at many institutions to obtain an abdominal ultrasound to characterize splenic anatomy of patients with heterotaxy [5]; however, abdominal Doppler is not routinely included in abdominal ultrasounds. Detection of CPSS has more frequently been identified on prenatal ultrasounds, and this is perhaps an area to focus on in the future, particularly in patients with congenital heart disease. Interestingly, it was the CT angiography of the chest that first detected the CPSS and abdominal ultrasound thereafter was unable to characterize the shunt. Magnetic resonance angiography of the abdomen was then used to characterize the patient's anatomy. Although abdominal ultrasound with Doppler is the gold standard, more specific imaging should be obtained if suspicion for CPSS is high.

It was decided to pursue intervention with occlusion of the portosystemic shunt because of systemic complications of hypoxemia and hyperammonemia. Before shunt occlusion, a liver biopsy was obtained showing portal vein patency with an acceptable pressure gradient of the portal system for closure. Shunts can be closed by interventional radiology endovascular methods or surgically in 1–2 steps depending on the level of portal pressure during occlusion test.

There are proposed hypotheses of the role of hepatic factor, portal venous factor, and substance P and their role in formation of PAVMs.[6] By occluding the shunt, the patient had restored portal blood flow to the liver through her interrupted IVC, thereby re-establishing the portohepatic relationship. Resolution of patient's hypoxemia suggests that PAVMs functionally resolved likely related to improved regulation of factors of the liver and portal vein.

DISCLOSURES

Author contributions: V. Carvajal and S. Reddy made substantial contributions to the conception or design of the work and interpretation of data for the work. V. Carvajal drafted the work, revising it critically for important intellectual content along with the help of S. Palareddy. V. Gopalareddy reviewed content as well as G. Wallis. A. Bean helped with image acquisition of images and interpretation. She also assisted with legends for the figures in the manuscript. Final approval of the version to be published was reviewed by V. Gopalareddy, G. Wallis, and A. Bean. V. Gopalareddy is the article guarantor.

Financial disclosure: None to report.

Informed consent was obtained for this case report.

Received April 19, 2023; Accepted October 5, 2023

REFERENCES

1. Bernard O, Franchi-Abella S, Branchereau S, Pariente D, Gauthier F, Jacquemin E. Congenital portosystemic shunts in children: Recognition, evaluation, and management. *Semin Liver Dis*. 2012;32(4):273–87.
2. Newman B, Feinstein JA, Cohen RA, et al. Congenital extrahepatic portosystemic shunt associated with heterotaxy and polysplenia. *Pediatr Radiol*. 2010;40(7):1222–30.
3. Sokollik C, Bandsma RH, Gana JC, van den Heuvel M, Ling SC. Congenital portosystemic shunt: Characterization of a multisystem disease. *J Pediatr Gastroenterol Nutr*. 2013;56(6):675–81.
4. Mclin V, Franchi-Abella S, Debray D, et al. Congenital portosystemic shunts: Current diagnosis and management. *J Pediatr Gastroenterol Nutr*. 2019;68(5):615–22.
5. Oleszczuk-Raschke K, Set PA, von Lengerke HJ, Tröger J. Abdominal sonography in the evaluation of heterotaxy in children. *Pediatr Radiol*. 1995;25(Suppl 1):S150–6.
6. Vettukattil JJ. Pathogenesis of pulmonary arteriovenous malformations: Role of hepatopulmonary interactions. *Heart*. 2002;88(6):561–3.