VENTRICULAR SEPTAL DEFECT WITH PULMONARY ARTERIAL HYPERTENSION IN AN INFANT: IS THERE SOMETHING MORE THAN WHAT MEETS THE EYE?

Deepanjan Bhattacharya1, Deepa Sasikumar1, Arun Gopalakrishnan1, A Anoop2
1Department of Cardiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India, 2Department of Imaging Sciences and Intervention Radiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India

ABSTRACT
An 11-month-old girl was detected to have muscular ventricular septal defect with hyperkinetic pulmonary hypertension (PH) was urgently operated upon. On follow-up, her PH worsened, resulting in right ventricular dysfunction and was later detected to have absent portal vein.

Keywords: Absent portal vein, pulmonary hypertension, ventricular septal defect

INTRODUCTION
Large ventricular septal defects develop pulmonary arterial hypertension very early in life, and require early closure by infancy. However, sometimes pulmonary hypertension in an infant with large ventricular septal defect may have a totally different etiology as illustrated in our case.

CASE REPORT
An 11-month-old baby girl presented with incidentally detected congenital heart disease. She was born to a primigravida mother with uneventful antenatal period, had smooth perinatal transition, and did not have any history of recurrent respiratory tract infections, poor feeding, respiratory distress, or cyanosis.

On examination, she had a significant failure to thrive (weight - 6 kg). Cardiomegaly was noted with closely split second sound, with a loud pulmonary component, and no audible murmur or hepatomegaly. Electrocardiogram showed sinus rhythm with right bundle branch block. Echocardiogram revealed an 8 mm apical muscular ventricular septal defect (VSD) with severe hyperkinetic pulmonary hypertension (PH) (estimated mean pulmonary artery pressure 55 mmHg) and normal left ventricular function. She underwent surgical VSD closure following which there was reduction in PH (right ventricular systolic pressure [RVSP] 40 mmHg). She continued to be asymptomatic on follow-up.

However, at the age of 32 months, she was readmitted with large right chylothorax, congestive cardiac failure, and systemic desaturation to 85% and was found to have severe PH (RVSP = 100 mmHg), right ventricular dysfunction, 3 mm atrial septal defect, and a small residual apical VSD with bidirectional shunt [Figure 1]. Chest X-ray showed normal cardiothoracic ratio with dilated main pulmonary artery [Figure 2].

Ultrasound abdomen showed the absence of portal vein, with enlarged inferior vena cava, and hepatic veins. Computed tomography of the abdomen revealed ill-formed portal vein radicals with multiple peri-esophageal collaterals, suggestive of probable absent portal vein.

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

Access this article online
Quick Response Code:
Website:
www.annalspc.com
DOI:
10.4103/apc.apc_226_20

Access this article online
Quick Response Code:
Website:
www.annalspc.com
DOI:
10.4103/apc.apc_226_20

For reprints contact: WKHLRPMedknow_reprints@wolterskluwer.com

How to cite this article: Bhattacharya D, Sasikumar D, Gopalakrishnan A, Anoop A. Ventricular septal defect with pulmonary arterial hypertension in an infant: Is there something more than what meets the eye? Ann Pediatr Card 2021;14:554-6.

Address for correspondence: Dr. Deepa Sasikumar, Department of Cardiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India.
E-mail: deepaskumar@yahoo.com

Submitted: 30-Sep-2020 Revised: 26-Feb-2021 Accepted: 10-May-2021 Published: 25-Mar-2022

© 2022 Annals of Pediatric Cardiology | Published by Wolters Kluwer - Medknow
Figure 1: Transthoracic echocardiogram (subcostal window) showing tiny residual VSD with large right to left shunt across VSD and PFO, with severe TR

Figure 2: Chest X ray showing dilated main pulmonary artery with pulmonary oligemia

Figure 3: Contrast enhanced computed tomography showing ill-defined portal vein (*) with peri-esophageal collaterals (arrow)

Pulmonary hypertension in children is uncommon and is usually secondary to cardiac or pulmonary causes.\(^1\) Cardiac causes are predominant and include admixture lesions or acyanotic heart disease with unrestricted left-to-right shunting, which can lead to pulmonary vascular occlusive disease.\(^2\)

Porto-pulmonary hypertension in children can result from shunting of portal venous blood into the systemic circulation either due to portal vein agenesis or due to congenital (abernethy) or acquired (primary liver disease) portosystemic shunts.\(^3\) In these situations, there is an imbalance between vasoconstrictors and vasodilators due to reduced hepatic metabolism, and this promotes pulmonary vasoconstriction.\(^4,5\)

In our case, the presence of pulmonary hypertension at an early age with significant shunting through unrestricted ventricular septal defect led us to consider the posttricuspid left-to-right shunt as the cause for PH. However, the worsening PH and right ventricular dysfunction with chylothorax on follow-up made us reconsider the diagnosis. The deficiency of portal venous radicles helped to identify portal vein agenesis and subsequent porto-pulmonary hypertension as the etiology.\(^6\)

Agenesis of the main portal venous branches is a rare cause of pulmonary arterial hypertension in the absence of liver dysfunction or portal hypertension, with <20 cases published in literature. The portal vein originates from the vitelline venous system at
4–10-week gestation, and its absence results in a congenital portosystemic shunt with absent or reduced perfusion of the liver. Since development of inferior vena cava is closely associated, venous malformations are not uncommon.[7] Morgan and Superina proposed a classification system in 1994 (Type I – absent portal perfusion, Ia – no confluence between splenic vein and superior mesenteric vein, Ib – splenic and superior mesenteric veins form a confluence; Type II – portalhepatic venous anastomosis with partial shunt, Ila – congenital, IIb – acquired).[8] Type I is usually congenital with hyperammonemia without any encephalopathy and present in children with female preponderance and associated cardiac malformations, while Type II is usually acquired and present in middle age with encephalopathy and elevated ammonia levels.[9]

Liver transplantation is indicated only when medical management fails with the occurrence of diffuse hepatoblastoma or severe portosystemic encephalopathy. Liver transplantation has to be considered before severe pulmonary arterial hypertension or pulmonary arteriovenous malformations develop.[10] Our child had severe pulmonary hypertension and right ventricular dysfunction and is not a candidate for liver transplantation. Concomitant liver and lung transplantation carries high perioperative mortality risk and was not a viable option considering the dearth of suitable organ donors. Optimal medical management with pulmonary vasodilator therapy remains the only option for this child.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Fasnacht MS, Tolsa JF, Beghetti M, Swiss Society for Pulmonary Arterial Hypertension. The Swiss registry for pulmonary arterial hypertension: The paediatric experience. Swiss Med Wkly 2007;137:510-3.
2. Jin H, Yang J, Zhang Q, Du J. Epidemiology and clinical management of pulmonary hypertension in children. Korean Circ J 2012;42:513-8.
3. Ohno T, Muneuchi J, Ihara K, Yuge T, Kanaya Y, Yamaki S, et al. Pulmonary hypertension in patients with congenital portosystemic venous shunt: A previously unrecognized association. Pediatrics 2008;121:e892-9.
4. Hervé P, Lebrec D, Brenot F, Simonneau G, Humbert M, Sitbon O, et al. Pulmonary vascular disorders in portal hypertension. Eur Respir J 1998;11:1153-66.
5. Raevens S, Geerts A, Van Steenkiste C, Verhelst X, Van Vlierberghe H, Colle I. Hepatopulmonary syndrome and portopulmonary hypertension: Recent knowledge in pathogenesis and overview of clinical assessment. Liver Int 2015;35:1646-60.
6. Sharma M, Rameshbabu CS. Collateral pathways in portal hypertension. J Clin Exp Hepatol 2012;2:338-52.
7. Joyce AD, Howard ER. Rare congenital anomaly of the portal vein. Br J Surg 1988;75:1038-9.
8. Morgan G, Superina R. Congenital absence of the portal vein: Two cases and a proposed classification system for portosystemic vascular anomalies. J Pediatr Surg 1994;29:1239-41.
9. Bellah RD, Hayek J, Teele RL. Anomalous portal venous connection to the suprahepatic vena cava: Sonographic demonstration. Pediatr Radiol 1989;20:115-7.
10. Hu GH, Shen LG, Yang J, Mei JH, Zhu YF. Insight into congenital absence of the portal vein: Is it rare? World J Gastroenterol 2008;14:5969-79.