# Ventricular septal defect with pulmonary arterial hypertension in an infant: Is there something more than what meets the eye?

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## 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

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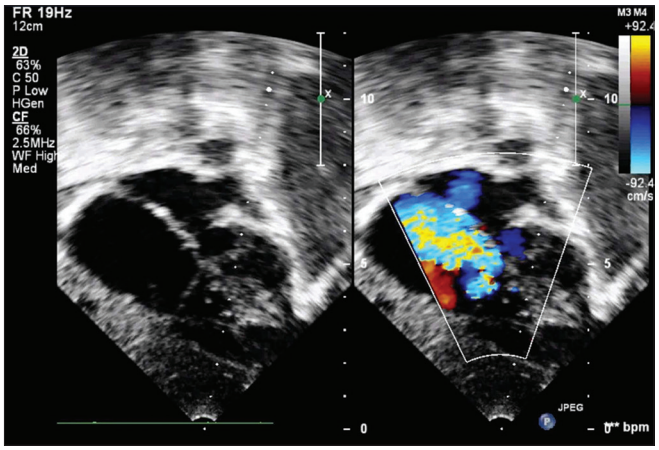


**Figure 1: Transthoracic echocardiogram (subcostal window) showing tiny residual VSD with large right to left shunt across VSD and PFO, with severe TR**

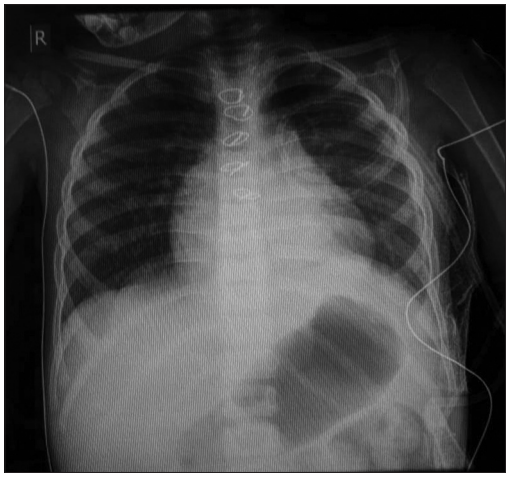

**Figure 2: Chest X ray showing dilated main pulmonary artery with pulmonary oligemia**

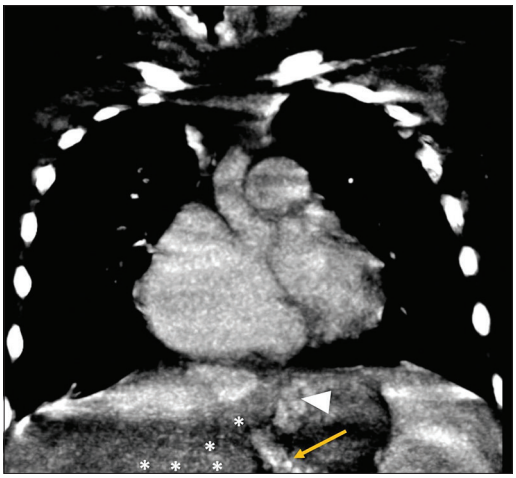

**Figure 3: Contrast enhanced computed tomography showing ill-defined portal vein (*) with peri-esophageal collaterals (arrow)**

sequelae of portal vein thrombosis [Figure 3]. No definitive connection between inferior vena cava and portal system suggestive of abernethy malformation could be delineated. Contrast-enhanced computed tomography of the chest also did not identify any pulmonary pathology or pulmonary arteriovenous malformation which could have caused PH. Liver function showed low albumin and mild elevation of bilirubin and transaminase (total bilirubin - 2.00 mg/dl, direct bilirubin - 0.90 mg/dl, total protein - 5.6 gm/dl, albumin - 3.0 gm/dl, aspartate aminotransferase - 84 IU/L, alanine aminotransferase - 71 IU/L, alkaline phosphatase - 354, and international normalized ratio – 1.07). Serum ammonia was mildly elevated (45 μmol/l), without any evidence of encephalopathy. Hence, a diagnostic possibility of portal vein agenesis, leading to porto-pulmonary hypertension was made. She was managed with intercostal drainage for the chylothorax which persisted for nearly a month. She gradually improved on octreotide infusion, diuretics, sildenafil, and bosentan and was discharged on pulmonary vasodilators and diuretic. The option of liver transplantation along with lung transplantation was discussed with the parents but was declined considering the prohibitively high risk.

## DISCUSSION

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 unrestrictive 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 unrestrictive 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, IIa – 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.

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### Conflicts of interest

There are no conflicts of interest.

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