World Journal of Pharmaceutical and Life Sciences

www.wjpls.org

Impact Factor: 8.21 Coden USA: WJPLA7

WE DESCRIBE THE CASE OF A 10-MONTH-OLD INFANT WITH CONGENITAL HEART DISEASE ASSOCIATED WITH A PULMONARY VENOUS RETURN ANOMALY

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DOI: https://doi.org/10.5281/zenodo.20444303

How to cite this Article: Mahdi Daher[*1], Nasma Morsad[2], Yacin Moumin[3], Najwa Benslima[4], Amal Rami[5]. (2026). We Describe The Case Of A 10-Month-Old Infant With Congenital Heart Disease Associated With A Pulmonary Venous Return Anomaly. World Journal of Pharmaceutical and Life Sciences, 12(6), 182–185.

Article Received on 05/05/2026 **Article Revised on 26/05/2026** **Article Published on 01/06/2026**

INTRODUCTION

Partial anomalous pulmonary venous return (PAPVR) accounts for approximately 3% of all congenital heart diseases. PAPVR is more common than total anomalous pulmonary venous return (TAPVR), with a prevalence of about 0.5% to 0.7% in the general population.[1] In most cases, PAPVR is asymptomatic and incidentally discovered during routine examinations.[2] The severity of symptoms correlates with the number of affected veins and the presence of an associated atrial septal defect (ASD). The clinical presentation of PAPVR is always similar to that of an ASD. Imaging plays a crucial role in diagnosing PAPVR. The prognosis is generally favorable in minor cases. Surgical management depends on the anatomical findings, clinical symptoms, and hemodynamic impact.

Here, we present a case of 10-month-old with trisomy 21 and PAPVR, followed for an ostium primum ASD, in whom a suspected PAPVR was further evaluated with a cardiac CT angiography.

CASE REPORT

A 10-month-old male infant with trisomy 21, followed for congenital heart disease. A fetal echocardiogram performed at 22 weeks of gestation confirmed an ostium primum ASD (Figure 1). At admission, the infant presented with tachycardia but no cardiac murmur or cyanosis. A chest X-ray was normal (Figure 2). The evaluation was further complemented by a cardiac CT angiography under sedation.

The cardiac CT angiography revealed that the right pulmonary veins drained into the right atrium, with an early left-to-right shunt (Figure 3), opacification of the suprahepatic veins, and the presence of a double superior vena cava (Figure 4). Additionally, the left superior pulmonary vein drained into the left atrium with an aberrant course (Figure 5). Right-sided cardiac chambers and suprahepatic veins were dilated, along with homogeneous hepatomegaly.

After sedation, the infant was transferred back to his hospital unit. Given that the right pulmonary veins drained entirely into the right atrium while the left pulmonary veins drained into the left atrium, the diagnosis of partial anomalous pulmonary venous return (PAPVR) associated with an ostium primum ASD was

confirmed. The next step in management remains under discussion, particularly regarding the indication for surgical intervention.

DISCUSSION

Pulmonary veins normally drain into the left atrium. Anomalous pulmonary venous return refers to abnormalities in the connection and/or drainage of pulmonary veins. A total defect is classified as total anomalous pulmonary venous return (TAPVR), while an incomplete defect (at least one anomalous pulmonary venous drainage) is classified as partial anomalous pulmonary venous return (PAPVR).[3] The underlying pathophysiology involves a failure in the confluence between the pulmonary venous sinus, the common pulmonary vein, and the left atrium.[4] PAPVR is more common on the right side (two-thirds of cases) than on the left (one-third of cases).

There are three types of right-sided PAPVR[1]

1. PAPVR to the superior vena cava (SVC) – the most common form, where the right upper and middle lobes drain into the segment of the SVC between the azygos vein entry and the right atrium.
2. PAPVR to the right atrium – a rarer presentation, exemplified by our case.
3. PAPVR to the inferior vena cava, known as the "scimitar syndrome."

Left-sided PAPVR typically drains into the left superior vena cava (LSVC) or the coronary sinus[2]

ASD is associated with PAPVR in 80% of cases.[5] ASD is the most common congenital heart defect after bicuspid aortic valve disease. ASDs are classified into four types based on the location of the defect: ostium secundum, ostium primum, sinus venosus, and coronary sinus.[6] The sinus venosus ASD is frequently observed in PAPVR cases, whereas ostium primum ASD, as seen in our patient, is part of atrioventricular septal defects (AVSD).[7]

AVSDs include several anatomical variants, typically classified as complete, intermediate, and partial forms. The partial form consists of an ostium primum ASD and a cleft mitral valve. AVSDs are associated with genetic abnormalities in 50% of cases, most commonly trisomy 21, which was present in our patient.[8]

Clinically, infants with PAPVR are often asymptomatic, and the condition is typically discovered incidentally. The extent of left-to-right shunting determines symptom severity. Small shunts are usually well tolerated and remain undiagnosed.[9] When significant, the shunt can cause right-sided heart dilation and failure.[10] Common clinical signs include dyspnea, tachycardia, cyanosis, and a heart murmur. Recurrent bronchitis and otorhinolaryngological infections have also been reported.[6,11]

PAPVR is rarely diagnosed prenatally. Fetal echocardiography between 12 and 16 weeks of gestation allows for an assessment of the four cardiac chambers and can detect a low atrial septal defect in the ostium primum position.[12] The larger the ostium primum defect, the easier the ultrasound diagnosis.

Postnatally, chest X-rays is typically the first imaging test but is often normal, as in our case. However, it can reveal cardiomegaly and bilateral pulmonary vascular overload in cases of significant left-to-right shunting.[1] While chest X-rays helps assess pulmonary and cardiac repercussions, it does not exclude the diagnosis of PAPVR.[13]

Echocardiography with color Doppler is the key diagnostic and follow-up tool in neonates and infants.[6] A segmental and functional sequential analysis provides detailed anatomical information.[3] The presence of an ASD with left-to-right shunting can be identified. In the absence of an ASD, unexplained right-heart dilation should prompt a systematic search for pulmonary venous or systemic venous return anomalies.[7] However, echocardiography has a limited field of view, making cross-sectional imaging techniques (CT angiography or cardiac MRI) necessary for further evaluation.[10]

Cardiac CT angiography is the modality of choice for diagnosing PAPVR and is typically performed after echocardiographic suspicion.[13] It provides detailed anatomical and functional assessment. In neonates and infants, sedation is required as breath-holding is not feasible. However, motion artifacts from a high heart rate (>100 bpm) can affect image quality. Proper contrast administration is critical for a successful scan.[14] Contrast volume is adjusted based on weight (2 mL/kg), with an injection rate varying by age and venous access quality (0.5 mL/s in neonates).[13,4] Images are acquired in axial planes with coronal and sagittal reconstructions for a comprehensive evaluation of thoracic vascular structures. Right-sided PAPVR can be visualized as drainage into the right SVC, inferior vena cava, or directly into the right atrium, as seen in our case.

The overall prognosis is favorable and depends on associated congenital heart defects or malformative syndromes.[6] Management is multidisciplinary, involving pediatric cardiologists, specialized radiologists, and congenital cardiac surgeons. Isolated PAPVR with insignificant shunting does not require surgical correction.[2] However, in symptomatic cases with a shunt fraction exceeding 50% and pulmonary hypertension, surgery is indicated. The procedure involves tunneling the anomalous pulmonary veins to the left atrium and closing the ASD.[9]

In our patient, the clinical course remained stable, so surgery was deferred in favor of continued monitoring. This case highlights the rarity of this cardiovascular anomaly in routine clinical practice.

CONCLUSION

Partial anomalous pulmonary venous return is a rare, often asymptomatic congenital anomaly frequently associated with ASD. Most cases are diagnosed incidentally.

Echocardiography is the primary diagnostic tool, with confirmation via CT or MRI. Early diagnosis and surgical management, particularly in cases associated with congenital heart defects or malformations, are critical for prognosis.

This report highlights a case of PAPVR associated with an ostium primum ASD, in which cardiac CT angiography was instrumental in confirming the diagnosis.

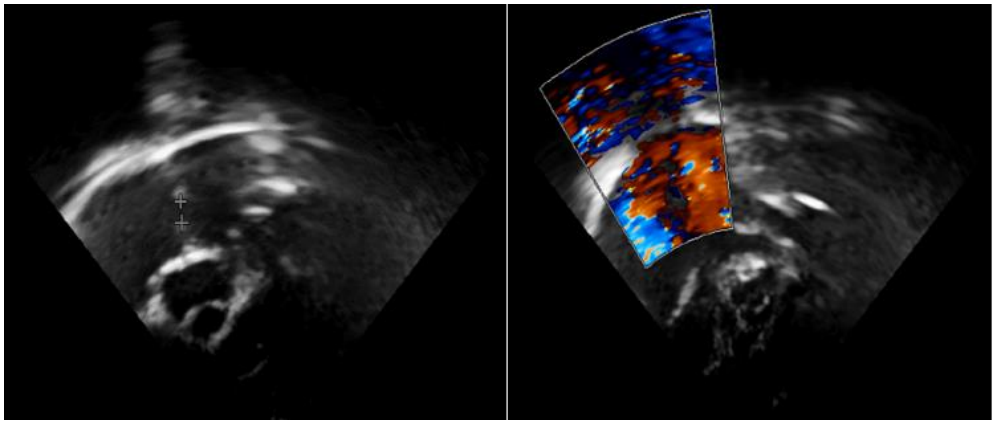

Figure 1: Fetal echocardiography demonstrates a primum-type atrial septal defect (ASD).

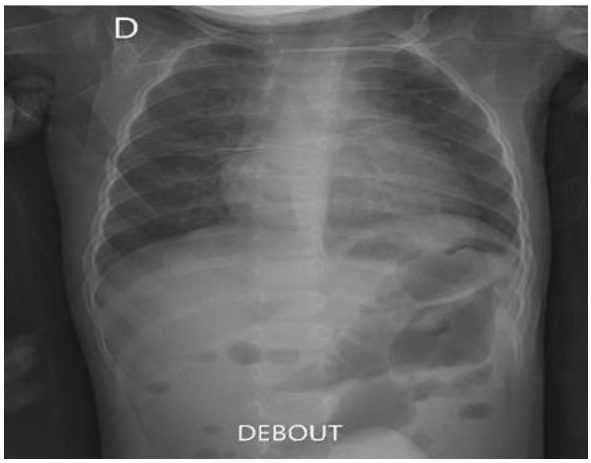

Figure 2: Anteroposterior (AP) supine chest radiograph shows no evidence of cardiomegaly (cardiac index < 0.55). No focal lesion is detected in either lung field, and the thoracic osseous structures appear unremarkable.

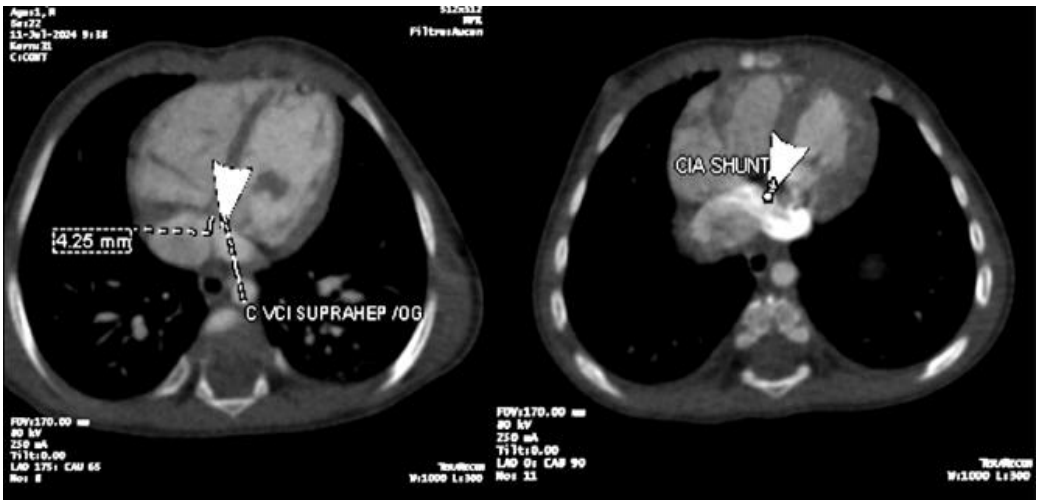

Figure 3: Cardiac CT angiography (axial view) demonstrating a left-to-right shunt.

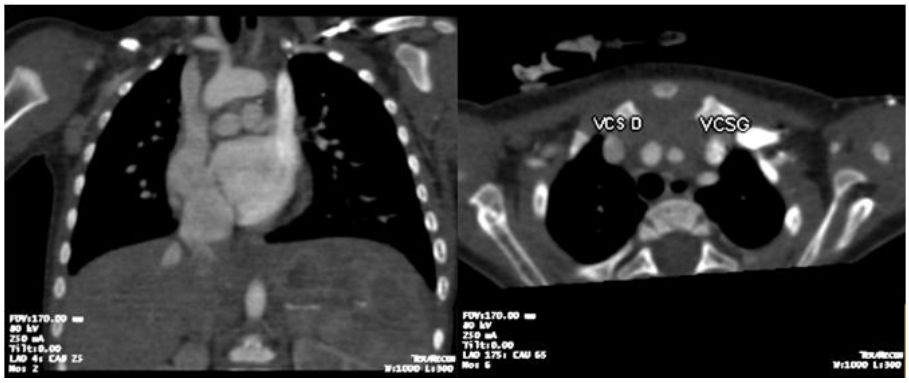

Figure 4: Cardiac CT angiography (coronal and axial view) that demonstrates duplication of the superior vena cava.

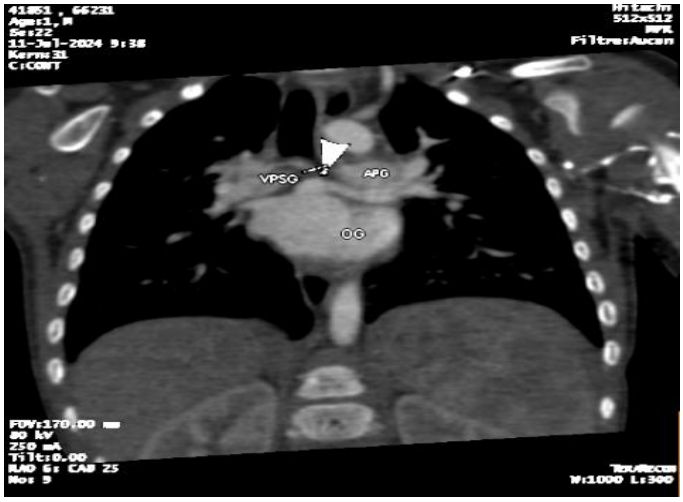

Figure 5: Cardiac CT angiography showing abnormal connection of the left superior pulmonary vein to the anterosuperior aspect of the left atrium.

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