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# PEDIATRIC RARE CASE OF SUPRACARDIAC TOTAL ANOMALOUS PULMONARY VENOUS

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**ABSTRACT**

Total anomalous pulmonary venous return (TAPVR) is a rare congenital heart defect, and patients are usually symptomatic at a very young age. Survival to adulthood without surgical correction is extremely rare. Objective: presented pediatric rare case of supracardiac total anomalous pulmonary venous. Methods: This study was a case report that describing detailed account of a patient's diagnosis, treatment, and follow-up a rare case. This study presents a single case. Results*:* we present a rare case of a 2-month-old infant diagnosed with Supracardiac TAPVR accompanied by an Atrial Septal Defect (ASD). The condition was identified through a combination of echocardiography and cardiac CT imaging. In these conditions, a right-to-left shunt is necessary for survival, along with the need for prompt corrective surgery. TAPVR is often associated with heterotaxy syndrome and other congenital heart defects. Various cross-sectional imaging modalities are valuable in detecting and evaluating pulmonary venous development anomalies. Conclution: These modalities provide both anatomical and functional information. Early detection and diagnosis lead to the best management strategies for patients with TAPVR. Multidetector Computed Tomography (MDCT) is a non-invasive imaging technique that plays an increasingly important role in evaluating these anomalies.

Keywords: cardiovascular medicine; MDCT; radiology; supracardiac;TAPVR

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## INTRODUCTION

Total anomalous pulmonary venous return (TAPVR) is a rare and potentially life-threatening congenital heart disease characterized by abnormal drainage of the pulmonary venous system. In this condition, blood from the pulmonary veins does not flow into the left atrium but instead connects abnormally to the systemic venous circulation. A right-to-left shunt is typically present to sustain life. Depending on the type and degree of pulmonary venous obstruction, TAPVR can lead to pulmonary hypertension and congestive heart failure (Marini, et al., 2018) In severe cases, prompt diagnosis and surgical correction are crucial to reducing the morbidity and mortality associated with this condition (Maurya et al., 2021).

Total Anomalous Pulmonary Venous Return (TAPVR) is a rare congenital heart disease with an incidence of approximately 0.6 to 1.2 per 10,000 live births, in which all pulmonary veins connect to the systemic veins, the right atrium, or both (Maurya et al., 2021). TAPVR was first reported by Friedkowsky in 1868; however, it was only recognized as a distinct

congenital disease entity in 1942. This condition is uncommon, accounting for approximately 0.5–2.0% of all congenital heart diseases (CHD) or around 0.008% of all live births. Data from the National Cardiac Center Harapan Kita (PJN-HK) recorded eight TAPVR cases (five males and three females) in 2006, representing approximately 0.7% of total CHD cases (Raharjo et al., 2007).

TAPVR results from the failure of the pulmonary veins to establish proper drainage into the left atrium, leading to the development of abnormal connections that direct blood flow through systemic veins into the right atrium (Eray et al., 2013). This condition may occur in isolation or in association with other congenital heart diseases, particularly heterotaxy syndrome with atrioviscereal situs abnormalities and polysplenia/asplenia (Ivemark syndrome). Approximately one-third of TAPVR patients have additional cardiac lesions, including single ventricle, atrioventricular septal defect, transposition of the great arteries, hypoplastic left heart syndrome, or patent ductus arteriosus (PDA) (Goerne & Rajiah, 2023). Before the advent of cardiac surgery, nearly all children with TAPVR succumbed within the first few months of life. With advancements in treatment, the mortality and morbidity rates of isolated TAPVR have significantly decreased. A multicenter population-based study by Seale et al. found that 15% of patients had associated cardiac lesions (Mulia & Rahman, 2023).

TAPVR accounts for about 1-2% of all congenital heart defects, with an estimated incidence of 5 to 7 cases per 100,000 live births. It affects both genders equally and can present in various anatomical forms, classified mainly into four types based on the location of the anomalous venous connection. The condition is often diagnosed in infancy due to severe clinical symptoms such as cyanosis, difficulty breathing, and poor feeding. If left untreated, TAPVR can lead to pulmonary hypertension, right-sided heart failure, and severe hypoxemia, contributing significantly to neonatal morbidity and mortality ((Bakker *et al*., 2019; Ghuge dan Rathi, 2020). The severity of TAPVR symptoms largely depends on the presence and degree of pulmonary venous obstruction. Infants with obstructed TAPVR usually present with respiratory distress and cyanosis soon after birth, requiring immediate medical intervention(Rahalkar dan Rahalkar, 2021). In non-obstructed cases, symptoms may be milder but can still include signs of heart failure and poor growth (Shi *et al*., 2017).

Echocardiography is the primary diagnostic tool for identifying TAPVR due to its accessibility and effectiveness in assessing cardiac anatomy and blood flown (Tongsong dan Nochizuki, 2023). However, the complexity of TAPVR may sometimes necessitate additional imaging techniques. Computed Tomography (CT) angiography and Magnetic Resonance Imaging (MRI) are advanced non-invasive options that offer detailed visualization of the pulmonary veins, helping to guide surgical planning without the need for invasive cardiac catheterization(Rocamora *et al*., 2022). Early and accurate diagnosis of TAPVR is critical for optimizing patient outcomes(Mulia dan Rahman, 2023). Timely surgical correction, which involves redirecting the pulmonary veins to the left atrium and closing any associated atrial septal defect, is the definitive treatment. The timing of surgery and the choice of the operative technique are influenced by factors such as the type of TAPVR and the presence of pulmonary venous obstruction(Qasim, 2015; Zhang *et al*., 2021).

Despite advancements in surgical techniques and perioperative care, the mortality rate in untreated cases remains high, with a significant number of infants succumbing to complications within the first year of life(Xiang *et al*., 2020). Early intervention can substantially improve survival rates, reduce the risk of long-term complications, and enhance the quality of life for affected individuals(Shentu *et al*., 2023). In this report, we present a rare case of a 2-month-old infant diagnosed with Supracardiac TAPVR accompanied by an Atrial Septal Defect (ASD). The condition was identified through a combination of

echocardiography and cardiac CT imaging. This case highlights the critical role of multi-modal imaging techniques in detecting complex congenital heart abnormalities and underscores the importance of early diagnosis and intervention in managing TAPVR. The purpose of this study is to report a rare pediatric case of Supracardiac Total Anomalous Pulmonary Venous Connection (TAPVC).

## METHOD

This study was a case report that describing detailed account of a patient's diagnosis, treatment, and follow-up. Case reports were often written as stories and can be a cornerstone of medical progress. Sample was a rare case and unique. The following criteria were used to define a rare case in this study: low prevalence, low incidence, high complexity, uniqueness of the case, and significant clinical implications. Case report describe and interpret an individual case, provide new ideas in medicine. Data was collected from document unusual or novel occurrences and document unexpected events that may yield new information. A critical review of the case report was conducted to assess the validity and reliability of the findings and to identify any limitations of the study.

## RESULT

Case description, diagnosis and management
Name: By. NMS
Gender: Male
Date of Birth: December 30, 2022
Age: 2 months
Address: Br. Pengembungan, Bongkasa Village
Medical Record Number: 22049607

### Anamnesis

*Chief Complaint*
Shortness of breath

*History of Present Illness*
The patient was referred from a regional hospital due to complaints of rapid breathing and worsening chest retractions that had persisted for one week prior to hospital admission. At 17 days old, the patient was previously hospitalized for five days due to jaundice. During that admission, the patient experienced respiratory distress and required assisted ventilation. The patient was reported to sweat on the head when active but was still able to breastfeed well. Cyanosis was denied.

*Past Medical History*
No history of previous illnesses or medical treatments.

*Family Medical History*
No family history of congenital heart disease, hypertension, or diabetes mellitus.
The patient is the second child of two siblings, and the older sibling is reported to be healthy.

*Maternal Pregnancy History*
The mother reported no complaints during pregnancy.
No history of hypertension, hyperglycemia, fever, or abnormal vaginal discharge during pregnancy.
Routine antenatal care (ANC) visits were conducted with a midwife.

*Birth History*
The patient was born via normal spontaneous vaginal delivery assisted by a doctor.
The newborn cried immediately after birth.
Birth weight: 2700 grams.
Birth length and head circumference were not recalled.

*Immunization History*
The mother reported that the patient had received complete basic immunizations.

*Nutritional History*
Breastfeeding: Since birth, duration of 18 months, on demand.
Formula milk: Not given.
Rice porridge: Not given.
Soft rice (nasi tim): Not given.
Solid adult food: Not given.

*Developmental Milestones*
Ability to hold head upright: Not yet recorded.

**Physical Examination**
*Anthropometric Status*
Birth weight (BW): 2700 g
Birth length (BL): Not recalled
Head circumference: Not recalled
Chest circumference: Not recalled
Current weight: 3.3 kg
Current height: 52 cm
Weight-for-length (PB/BB): -2 to -1 SD
Weight-for-age (BB/U): -2 to -1 SD
Height-for-age (TB/U): -2 to -1 SD

*Vital Signs*
General condition: Moderately ill appearance
Heart rate: 141 bpm, adequate volume
Respiratory rate: 45 breaths per minute, regular
Axillary temperature: 36.6°C
$SpO_2$: 94% on room air

*General Status*
Head: Normocephalic
Eyes: No pallor, no jaundice, pupils isocoric and reactive to light (+/+)
ENT: Appears normal, no secretions
Mouth: Lips and tongue not cyanotic
Neck: No lymph node enlargement
Thorax: Symmetrical (+), retractions present
Heart: Normal S1 and S2, regular rhythm
Systolic murmur at ICS 5 midclavicular line, grade III/VI
Lungs: Vesicular breath sounds (+/+), no rhonchi or wheezing
Abdomen: No distension, normal bowel sounds
Liver and spleen not palpable
Genitalia: Within normal limits
Extremities: Warm, capillary refill time (CRT) <2 seconds, no edema, Cyanosis (+)

***Echocardiography Examination***

The results of the Multislice Computed Tomography (MSCT) cardiac scan in axial, sagittal, and coronal views, both with and without contrast, performed on February 3, 2023, showed the following findings (Figure 1):

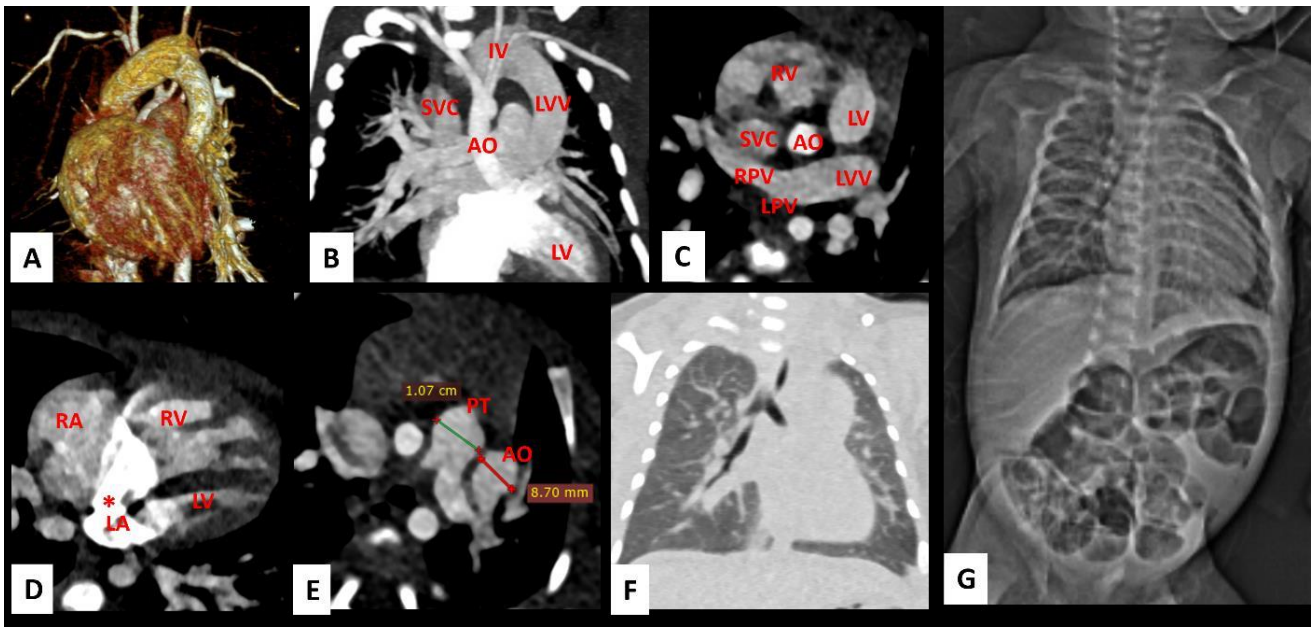


Figure 1. Results *MSCT in infant* NMS, Age 2 month with *Supracardiac* (*TAPVR)*

- The right and left pulmonary veins were observed to converge, forming a left vertical vein that drained into the left innominate vein.
- There was a defect in the septum connecting the right and left atrium measuring approximately 8.3 mm.
- The measured diameters of the aortic structures were as follows: aortic root approximately 8.7 mm, ascending aorta approximately 6.4 mm, aortic arch approximately 4.7 mm, and descending aorta approximately 5.3 mm, with no signs of stenosis, intraluminal thrombus, or intimal flap.
- The pulmonary trunk was measured at approximately 10.7 mm, with no signs of stenosis or intraluminal filling defects.
- The ratio of the pulmonary trunk to the ascending aorta was greater than 1.
- The main right and left pulmonary arteries were both measured at approximately 6.1 mm, with no signs of stenosis or intraluminal filling defects.
- Venous system: no signs of stenosis or intraluminal filling defects were noted in the branches of the right and left pulmonary veins.
- Confirmation of vascular diameters according to age was requested. The normal diameters for ages 0-12 months (mean age of 8 months) are: ascending aorta 10-14.8 mm, descending aorta 8.3-11.3 mm, main pulmonary artery 9.9-13.3 mm, right pulmonary artery 6.5-8.7 mm, left pulmonary artery 5.8-10.4 mm (Akay et al., 2009. Diameters of normal thoracic vascular structures in pediatric patients. Surgical and Radiologic Anatomy, 31(10), pp. 801–807).
- The heart size appeared to be within normal limits, with a cardiothoracic ratio (CTR) of 55%. Right atrial enlargement was observed.
- Heart valves: no calcification was observed on the aortic or mitral valves.
- Pericardium: no effusion, thickening, or calcification was noted.
- There were no mural thrombi or intracardiac masses in the left atrium or ventricle.
- No pulmonary embolism or dissection was observed.
- Extra-cardiac structures: no abnormalities were detected.
- Ground-glass opacities were observed in the superior and inferior lobes of both lungs.
- The trachea and main bronchi on both sides were patent.

- Skeletal findings: a complete fracture of the right clavicle (1/3 medial) with displacement and callus formation was noted.

Based on the contrast-enhanced MSCT cardiac findings, the patient was diagnosed with Total Anomalous Pulmonary Venous Return (TAPVR) type I (Supracardiac) accompanied by signs of pulmonary hypertension, an atrial septal defect (ASD) with right atrial enlargement (RAE), pulmonary edema, and a displaced complete fracture of the right clavicle (1/3 medial) with callus formation, indicating incomplete union.

Tabel 1.

E*chocardiography* Februari 1st 2023

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| **LA/Ao ratio** | 1.1 | | **IVSd** | 0.3 | Cm | **AV MaxPG** | 5 | mmHg |
| **Ao** | 0.6 | cm | **LVIDd** | 1.1 | Cm | **Dao MaxPG** | 4 | mmHg |
| | | | **LVPWd** | 0.3 | Cm | **PV MaxPG** | 18 | mmHg |
| **E/A** | - | | **EF Teich** | 66.1 | % | | | |
| **E/e' average** | - | | **RWT** | 0.5 | | | | |
| | | | **TAPSE** | 1.2 | Cm | | | |
| | | | | | | | | |

| |
|---|
| 2nd echo with acyanotic CHD ec susp PS, mild TR (ref dr. Putu Jerry Eka Rahayu Pande, Sp.A, Mangusada Hospital) SpO2 98% : |
| |
| Atrial situs solitus, susp TAPVR supracardiac (RPV and LPV drain through innominate vein to SVC) dilated SVC, AV-VA concordant, RAE, RVE, moderate ASD II bidirectional shunt dominan R to L shunt (diam 7.3 mm, posterior rim minimal, anterior 5.4 mm, LASD 1.3 mm), no PDA, no VSD, no CoA, left aortic arch, mild TR, dilated MPA (diam 11.5 mm, normal 5 – 10 mm), D shaped LV at systolic and diastolic phase,no pericardial effusion,normal LV and RV systolic function. |
| |

| |
|---|
| Result(s): Susp supracardiac TAVPR, moderate ASD II bidirectional shunt dominan R to L shunt, PAH, mild TR<br>Comment(s): Initiation heart failure therapy furosemide/ captopril/ spironolactone/digoxin : 3 mg/ 0.75mg/ 3.125mg/ 15mcg BID IO<br>Planning : CT-Cardiac and CT-angio Pulmonal with sedation<br>Monitor ARI, CHF, FTT, PHT, IE<br>Re-echo accordng to clinical condition |

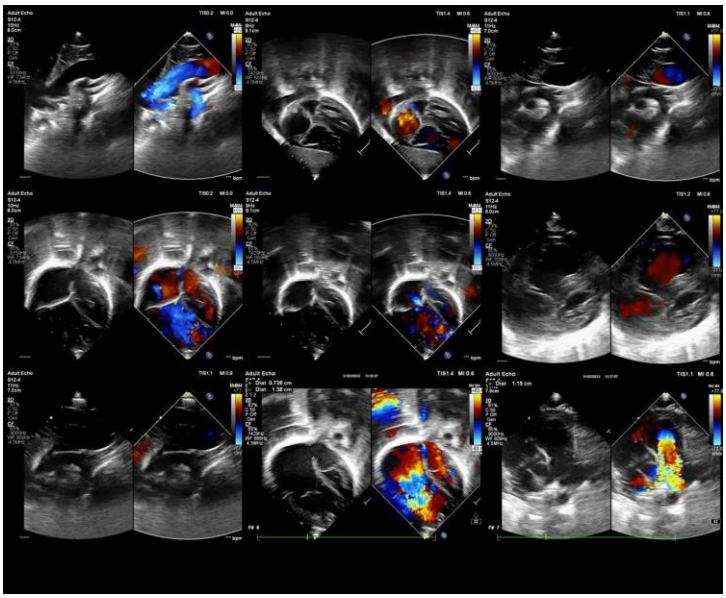

Figure 2. Results *echocardiography in infant* NMS, Age 2 month with *Supracardiac* (*TAPVR)*

After undergoing various follow-up examinations, the patient was diagnosed with moderate heart failure, suspected supracardiac Total Anomalous Pulmonary Venous Return (TAPVR), and moderate atrial septal defect (ASD) II with a bidirectional shunt, predominantly right-to-left shunt, with good nutritional status. The patient received the following supportive therapy:

- Fluid requirement: 330 mL/day
- Breast milk: 42 mL every 3 hours
- Furosemide: 3 mg orally every 12 hours
- Captopril: 0.75 mg orally every 12 hours
- Spironolactone: 3.125 mg orally every 12 hours
- Digoxin: 15 mcg orally every 12 hours

A multidisciplinary team meeting (BTKV Conference) involving pediatric cardiology, radiology, and other relevant departments was held to discuss the patient's condition and future management. The conclusion from this meeting was that the patient had supracardiac TAPVR, and a re-routing procedure for TAPVR was planned. While awaiting the procedure, the patient continued to receive supportive medical therapy on an outpatient basis. Unfortunately, the patient experienced recurrent respiratory tract infections and passed away in April 2023 after suffering from repeated respiratory infections.

The transthoracic echocardiography findings in this patient were consistent with supracardiac TAPVR, showing a connection to the left innominate vein (LIV), with right and left pulmonary veins draining into the left vertical vein, the left innominate vein, and then into the superior vena cava (as illustrated in Figure 3)

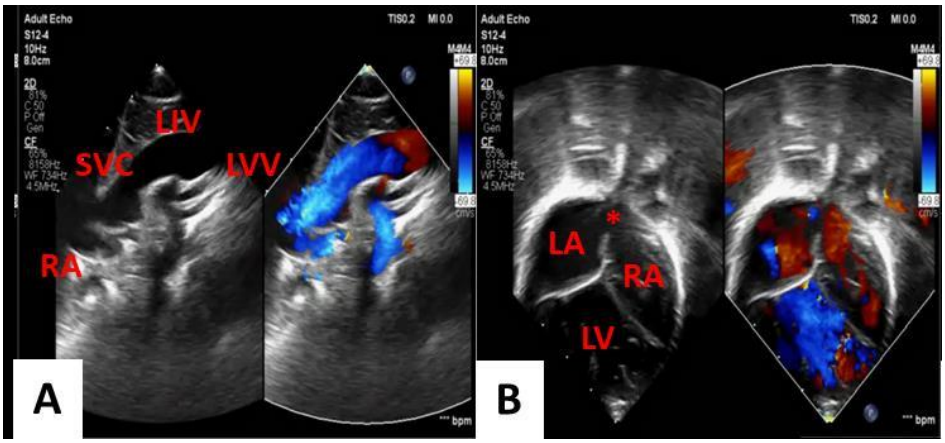


Figure 3. T*ransthoracic echocardiography results in patient that showed supracradiac TAPVR* with conection in left innominate vein (*LIV*) (A). There was also *right to left shunt* as *ASD* in Picture B*

## DISCUSSION

The patient in this case was a two-month-old male infant presenting with rapid breathing accompanied by chest wall retractions and sweating while feeding, along with mild cyanosis but no signs of heart failure or a family history of heart disease. Physical examination revealed tachypnea and a pansystolic murmur on auscultation at the left fifth intercostal space. According to existing literature, infants or children with Total Anomalous Pulmonary Venous Return (TAPVR) commonly present with symptoms of heart failure (such as difficulty breathing, trouble feeding, or tachypnea) and/or cyanosis. Therefore, TAPVR should be considered as a differential diagnosis in infants or children with these clinical features. However, the diagnosis of TAPVR cannot be based solely on symptoms and clinical signs, as its presentation is similar to other congenital heart diseases (CHD), necessitating further diagnostic investigations (Raharjo et al., 2007; Lyen et al., 2017).

TAPVR arises due to an embryological development defect in the cardiac venous system, where the pulmonary veins fail to connect to the left atrium, instead draining into the systemic circulation, either directly or indirectly into the right atrium and then to the left atrium through what is known as a right-to-left shunt. The survival rate of patients with TAPVR heavily depends on the presence of an intracardiac right-to-left shunt, which can be in the form of a patent foramen ovale (PFO) or an atrial septal defect (ASD) (Lyen et al., 2017). Echocardiography using color Doppler ultrasound is a non-invasive diagnostic tool that is fairly accurate in detecting TAPVR. Reports indicate its sensitivity ranges from 85-100%, with a specificity of 99-100% (Bakker et al., 2019). Key echocardiographic indicators include right atrial enlargement, elevated pulmonary pressures, and a right-to-left shunt through a PFO or ASD, which can strongly suggest the diagnosis of TAPVR. During echocardiography, it is crucial to determine whether the pulmonary veins are draining into the left atrium or another location, as well as to identify any potential obstructions. Different echocardiographic views are used to locate the types of TAPVR: the suprasternal view for supracardiac TAPVR, the four-chamber view for cardiac TAPVR, and the subcostal view for infracardiac TAPVR (Kumar et al., 2016).

The findings were confirmed by a contrast-enhanced Multislice Computed Tomography (MSCT) cardiac scan (Figure 21), which revealed a snowman appearance of the cardiac silhouette, along with abnormal drainage of both right and left pulmonary veins through the left vertical vein and left innominate vein into the superior vena cava. There was also evidence of superior vena cava dilation and an ostium secundum atrial septal defect (ASD) measuring approximately 8.3 mm, accompanied by right atrial enlargement. Based on these imaging results, the patient was radiologically diagnosed with supracardiac TAPVR, characterized by a right-to-left shunt through an atrial septal defect in the ostium secundum. Additionally, there was an increased pulmonary trunk-to-ascending aorta ratio, indicating pulmonary hypertension and ground-glass opacity in both lungs, suggestive of pulmonary edema.

The patient was diagnosed with supracardiac TAPVR, the most common form of TAPVR, accounting for approximately 50% of all cases. Other types include cardiac, infracardiac, and mixed-type TAPVR (Katre et al., 2012; Munsi et al., 2015). The patient was managed with routine follow-ups and supportive medical therapy. Although a re-routing procedure for TAPVR was planned, the patient unfortunately passed away several months later due to recurrent respiratory infections. Medical therapy is typically administered in cases with heart failure symptoms resulting from increased pulmonary blood flow. However, definitive treatment for TAPVR is surgical correction (Raharjo et al., 2007; Bae et al., 2022). Surgical intervention involves redirecting the pulmonary venous drainage to the left atrium (LA). For supracardiac and infracardiac TAPVR, the procedure includes anastomosing the pulmonary venous confluence to the LA, whereas for intracardiac TAPVR, a patch is used to channel the pulmonary venous flow to the LA (Shentu et al., 2023). In non-obstructive cases, elective surgery should ideally be performed within the first six months of life to prevent irreversible pulmonary artery remodeling. Associated defects, such as patent foramen ovale (PFO) and atrial septal defect (ASD), are closed during surgery. In obstructive TAPVR, urgent surgery is required at an earlier age, with a higher post-operative mortality rate compared to non-obstructive cases (Shen and Ungerleider, 2001; Konduri and Sanjeev, 2023).

## CONCLUSION

Congenital pulmonary venous drainage anomalies refer to a condition in which the pulmonary veins do not return to the left atrium but instead drain into the systemic circulation. Total Anomalous Pulmonary Venous Return (TAPVR) is classified into supracardiac, cardiac,

infracardiac, or mixed types, depending on the drainage location. In these conditions, a right-to-left shunt is necessary for survival, along with the need for prompt corrective surgery. TAPVR is often associated with heterotaxy syndrome and other congenital heart defects. Various cross-sectional imaging modalities are valuable in detecting and evaluating pulmonary venous development anomalies, which represent one of the more complex types of congenital heart diseases, such as in the case of TAPVR. These modalities provide both anatomical and functional information. Early detection and diagnosis lead to the best management strategies for patients with TAPVR. Multidetector Computed Tomography (MDCT) is a non-invasive imaging technique that plays an increasingly important role in evaluating these anomalies.

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