CASE REPORT

TAPVR in a kindergartner presenting with fatigue

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SUMMARY

We present a case of late diagnosis of cyanotic heart disease in a kindergartner, as a contemporary reminder of the importance of the history and clinical examination in the assessment of paediatric patients. In addition, this case illustrates the complementary diagnostic value of various cardiac imaging modalities in understanding pulmonary venous drainage.

BACKGROUND

While most cyanotic heart disease is discovered in the newborn period, this is not universally true. Also, while approximately 50% of school age children may have an innocent murmur, the quality of the murmur and associated clinical features should be considered carefully.¹ We present a case of late diagnosis of cyanotic heart disease as a contemporary reminder of the importance of careful clinical examination for children.

CASE PRESENTATION

A previously healthy 4.5-year-old girl was seen by her paediatrician several times for persistent decreased exercise tolerance and fatigue. A heart murmur and her presentation eventually prompted a chest X ray (CXR). The CXR showed cardiomegaly and central vascular congestion without pulmonary infiltrate and led to paediatric cardiology referral. Weighing 14.8 kg (15th centile) and standing 104.8 cm tall (50th centile), she had dusky lips,

with normal vital signs except room air oxygen saturation was 93%. She had an increased right ventricular (RV) impulse, normal S1 and widely split S2 with 2/6 systolic ejection murmur at upper sternal borders bilaterally, but no diastolic murmur or gallops.

INVESTIGATIONS

Echocardiogram (ECHO) showed RV dilation with large atrial septal defect (ASD) with superior extension and possible partial anomalous pulmonary venous return (PAPVR); yet, full definition of pulmonary veins eluded ECHO. Cardiac magnetic resonance (CMR) was, therefore, performed with Siemens 1.5T Symphony magnet (Siemens Healthcare, Malvern Pennsylvania, USA). CMR revealed total anomalous pulmonary venous return (TAPVR) with a large ostium secundum ASD. The pulmonary venous confluence (PVC), situated superior to the left atrium (LA), received venous drainage from all segments of right and left lungs. This PVC drained via a dilated vertical vein into the innominate vein, and subsequently to the right superior vena cava and right atrium (RA; [figure 1A](#)). Blood traversed right to left across a large, superiorly positioned ASD into the LA, leading to cyanosis. Using CMR velocity encoded cine imaging of the main pulmonary artery and aorta, the estimated pulmonary (Qp) to systemic (Qs) flow was 4:1, where normally Qp equals Qs. The RV appeared severely dilated, with end-diastolic

Figure 1 (A) Gradient echo (FLASH) cardiac MR coronal image (Siemens 1.5T Symphony magnet with matrix 192×192, TR 54.8, TE 3.67, slice thickness 4 mm) showing pulmonary venous confluence superior to left atrium LA (top panel), vertical vein (middle panel) and dilated right superior vena cava (bottom panel). (B) Gradient ECHO (FLASH) short axis CMR image ((Siemens 1.5T Symphony magnet) matrix 192×156, TR 54.0, TE 3.5, slice thickness 6 mm) showing right ventricular dilation due to increased blood flow from total anomalous pulmonary venous return.

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Figure 2 (A) Intraoperative photo showing the dilated vertical vein (VV). (B) Intraoperative photo showing pulmonary venous confluence (PVC) and left atrium (LA).

volume index equal to 166 mL/m^2 (normal $\sim 73 \pm 9 \text{ mL/m}^2$) and end-systolic volume index 54 mL/m^2 (normal $\sim 25 \pm 5 \text{ mL/m}^2$; ² Figure 1B). Biventricular systolic function was preserved.

TREATMENT

Using aortic bicaval cannulation for bypass, antegrade cardioplegia and cooling, the repair was able to be successfully completed without circulatory arrest. After ligation and division of the vertical vein (figure 2A), a wide non-obstructive direct anastomosis was created between the PVC and the back of the LA (figure 2B). The patient was discharged from the hospital on post operative day 4.

OUTCOME AND FOLLOW-UP

At 3-year follow-up, she is an acyanotic, asymptomatic, active child with a normal examination and normal ECHO.

DISCUSSION

TAPVR is an uncommon cyanotic heart disease typically diagnosed in the newborn period. TAPVR may be one of several varieties: supra cardiac, where pulmonary veins drain to a vertical vein, innominate vein, superior vena cava and then RA ($\sim 50\%$ of cases); infra cardiac to hepatic system to RA (20%); cardiac to the coronary sinus or RA directly (20%); or mixed (10%). Pulmonary venous obstruction is possible with any variety, but more common with infra diaphragmatic TAPVR. An atrial shunt is essential for survival and leads to variable degrees of cyanosis. Clinical presentation depends on the presence or absence of obstruction to pulmonary venous return. Patients with obstructed TAPVR are extremely ill soon after birth, with severe cyanosis, and may have low cardiac output and respiratory distress. Patients with TAPVR without pulmonary venous obstruction (our case) are often asymptomatic at birth, with difficult to detect cyanosis, but will usually present within the first year of life with tachypnoea, feeding difficulties, failure to thrive or repeated respiratory infections.

While our patient was well until age 3 years, her mother reported persistence in seeking care during the year prior to diagnosis, because her daughter fatigued more easily than other children. The mother's persistence, and then eventual recognition of the pathological character of this child's systolic ejection murmur, prompted the CXR, which led to cardiology referral.

The child's subtle cyanosis escaped detection by the paediatrician, likely due to the torrential pulmonary blood flow (ie, high Qp:Qs), but was confirmed by pulse oximetry during cardiology appointment. With the advent of neonatal pulse oximetry testing prior to hospital discharge, rare cyanotic heart diseases like this may be discovered earlier. Her dynamic chest wall points to the importance of palpation during physical examination. This and her widely split second heart sound reflect the increased right heart preload and increased pulmonary blood flow with delayed closure of pulmonary valve. Although Qp:Qs was 4:1, a diastolic inflow murmur (from increased inflow across the tricuspid valve) was not heard. Typically, TAPVR requires urgent surgical correction within the first few months of life due to a high mortality rate of 80% by 1 year, if left unrepaired. TAPVR can, as in our case, present in child or adulthood, in the setting of a widely patent atrial septum and no obstruction to pulmonary venous return; here, prognosis is better despite late repair.^{3 4}

Learning points

- ▶ This case illustrates the importance of careful clinical examination for any symptomatic child. The practitioner should pay close attention to qualities that differentiate an innocent murmur from a pathological murmur, including murmur quality, grade, location of optimal auscultation, and ancillary clinical findings like precordial activity, quality of second heart sound and presence/absence of cyanosis. (Innocent murmurs are typically vibratory and soft, increasing with supine positioning and not associated with other clinical abnormalities.) This attention to detail will lead to appropriate and timely paediatric cardiology referral.
- ▶ In addition, this case illustrates a rare case of cyanotic heart disease diagnosed outside the newborn period.
- ▶ The case shows the complementary nature of ECHO and CMR for diagnosis of congenital heart disease.

Contributors AJC is responsible for recognition of this child's disease and referral for cardiac MRI. He has reviewed this manuscript for accuracy. JMP assisted with

manuscript writing and with performing of CMR scan. MMS recognised this as a unique case worthy of case report. She supervised CMR scanning, making the diagnosis of TAPVR. MMS initiated this case report, obtained permission from the family for publication of medical images, and wrote the manuscript with the assistance of the others, especially JMP. JST performed the surgery, provided intra operative images and reviewed this manuscript for accuracy.

Competing interests None.

Patient consent Obtained.

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