

Delayed diagnosis of an isolated partial anomalous pulmonary venous connection

Case Report

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Abstract: In this case report we describe the delayed diagnosis of a very rare congenital anomaly – isolated partial anomalous pulmonary venous connection. This congenital anomaly should be suspected at any age in the clinical setting of right heart volume overload, especially in the absence of a large atrial septal defect. Tomographic imaging modalities (computed tomography or cardiovascular magnetic resonance) not only allow the comprehensive structural and functional assessment of this anomaly, but also help assess the patient's suitability for surgical treatment. Surgery is the definitive treatment of a patient with a significant left-to-right shunt due to partial anomalous pulmonary venous connection.

Keywords: *Isolated partial anomalous pulmonary venous connection • Echocardiography • Computed tomography • , Cardiovascular magnetic resonance • Surgery*

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1. Introduction

Standard pulmonary venous anatomy consists of four pulmonary veins, two on each side, draining into the left atrium. Anomalous pulmonary venous connection results in an extracardiac left-to-right shunt, as pulmonary venous blood flows directly to the right atrium or indirectly through a variety of venous connections from the anomalous pulmonary vein [1]. Partial anomalous pulmonary venous connection (PAPVC) encompasses a specific group of congenital cardiovascular anomalies that are caused by the abnormal return of one or more, but not all, of the pulmonary veins [2]. The anomalous vein connects with a systemic vein resulting in the mixing of oxygenated blood from the pulmonary circulation

with the systemic venous blood before returning to the right-side of the heart.

The PAPVC is a rare congenital disease present in up to 0.7% in the general population [3-6]. The most common form of PAPVC is the left upper pulmonary vein (LUPV) that connects to the left innominate vein, which in turn drains into the superior vena cava (SVC) [6]. In other cases, small segments of the right upper pulmonary lobe connect directly to the SVC, usually above the level of the azygous vein. Other more uncommon PAPVCs include anomalous pulmonary vein connections to the coronary sinus, azygous vein, or the inferior vena cava [6]. PAPVC has traditionally been associated with ASD, found in 80% of patients in one paediatric series [7]. Thus, clinically significant isolated PAPVC is a very rare congenital disease because many individuals

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with a single anomalous pulmonary vein often are asymptomatic and have a normal life expectancy [8].

The primary aim of this educative case report was to present the delayed diagnosis of an isolated PAPVC and discuss the role of tomographic imaging modalities in the diagnosis of this very rare congenital anomaly. The secondary aim of this report was to emphasize the current knowledge about isolated PAPVC and discuss the indications for surgical repair relying on clinical case presented.

2. Case report

A 69-year-old male presented to our outpatient department because of palpitations, fatigue and dyspnoea during less than ordinary physical activity corresponding with the third New York Heart Association (NYHA) functional class. His medical history revealed a 3-year history of permanent atrial fibrillation (AF) and 8-year history of adequately treated arterial hypertension. Screening performed at local hospital was negative for pulmonary disease and chronic coronary heart disease. Due to above mentioned diseases for a couple of last years he was treated with beta-blocker, selective imidazoline I1-receptor agonist and warfarin maintaining international normalized ratio (INR) between 2.0 and 3.0.

On presentation, the patient was clinically stable with the blood pressure of 140/80 mm Hg, irregular heart rate of about 92 bpm. On heart auscultation, a holosystolic murmur was heard at the left mid-sternal border. The respiratory rate of the patient was about 20 breaths per minute. Lung auscultation was normal and abdominal palpation was unremarkable. Laboratory studies, including cardiac enzymes, thyroid function tests, liver function tests, electrolytes, and complete blood count, were all within normal limits. An electrocardiogram (ECG) showed AF, a heart rate of 96 beats per minute, a complete right bundle branch block and a left anterior hemiblock.

Transthoracic echocardiography (TTE) revealed no dilatation and hypertrophy of the left ventricle (LV) (LV end-diastolic diameter index 2.9 cm/m², normal value ≤ 3.1 cm/m²) with normal global and regional LV systolic function. However, the cavity of the right ventricle (RV) was moderately dilated (RV end-diastolic diameter 3.8 cm, normal value ≤ 2.7 cm) and diastolic septal displacement towards the LV was observed as a sign of RV volume overload. Additionally, the dilatation of both atria (left atrium (LA) 8.3 × 6.0 cm, normal values $\leq 5.6 \times 4.6$ cm; right atrium (RA) 7.8 × 5.6 cm, normal values $\leq 5.3 \times 4.5$ cm) and tricuspid valve annulus (diameter of tricuspid valve annulus 4.5 cm, normal value < 4.0 cm)

was observed (Figure 1A). Although tricuspid regurgitation (TR) was moderate-to-severe (regurgitation orifice according PISA (Proximal Isovelocity Surface Area) method was 0.51 cm², cut-off for severe TR is > 0.4 cm²; regurgitation volume 44 ml, regurgitation volume between 30 and 59 ml is considered as moderate TR) (Figure 1A) the pressure in the pulmonary artery was not elevated (pulmonary artery systolic pressure at rest was 28 mm Hg, normal value < 30 mmHg). There was no evidence of intracardiac shunts on TTE. In summary, the atriomegaly, RV volume overload, and moderate-to-severe TR without pulmonary artery hypertension (PAH) were diagnosed performing TTE. As the reason for RV dilatation and volume overload was not clear, the three-dimensional transesophageal echocardiography (TEE) was performed straight after TTE and excluded the atypical ASD (Figure 1B) and revealed only one right pulmonary venous entry (Figure 1C, arrow). A 1.5 T cardiovascular magnetic resonance (CMR) was performed in three months period after TEE for clarification of the diagnosis. CMR steady-state free precession (SSFP) sequence confirmed the findings of the TTE (Figure 1D). Additionally, on the CMR HASTE (Half Fourier Acquisition Single Shot Turbo Spin Echo) (Figure 1E) and VIBE (Volumetric Interpolated Breath-hold Examination) (Figure 1F) sequences, dilatation of the SVC was observed, and anomalous drainage of the right upper pulmonary vein (RUPV) into the SVC was suspected (arrows point to the anomalous connection). A chest computed tomography (CT) angiography performed in two months period after CMR confirmed the findings noted above of the anomalous RUPV drainage into the dilated SVC (Figure 1G and Figure 1H, RLPV, LUPV and LLPV – right lower, left upper and left lower pulmonary veins, respectively). The surgical repair of anomalous pulmonary vein together with tricuspid ring anuloplasty was recommended for the patient. While being on the waiting list for surgery patient changed his opinion and refused to undergo surgical treatment.

3. Discussion

The above described case illustrates an isolated congenital disease – anomalous RUPV without an associated ASD.

In case of isolated PAPVC the size of the left-to-right shunt is determined by the percentage of pulmonary venous return that drains anomalously. The most obvious determinant is the number of anomalously draining veins. In most cases significant shunting is associated with two or more anomalous draining veins. Thus, patients with single vein anomaly are generally asymptomatic and do

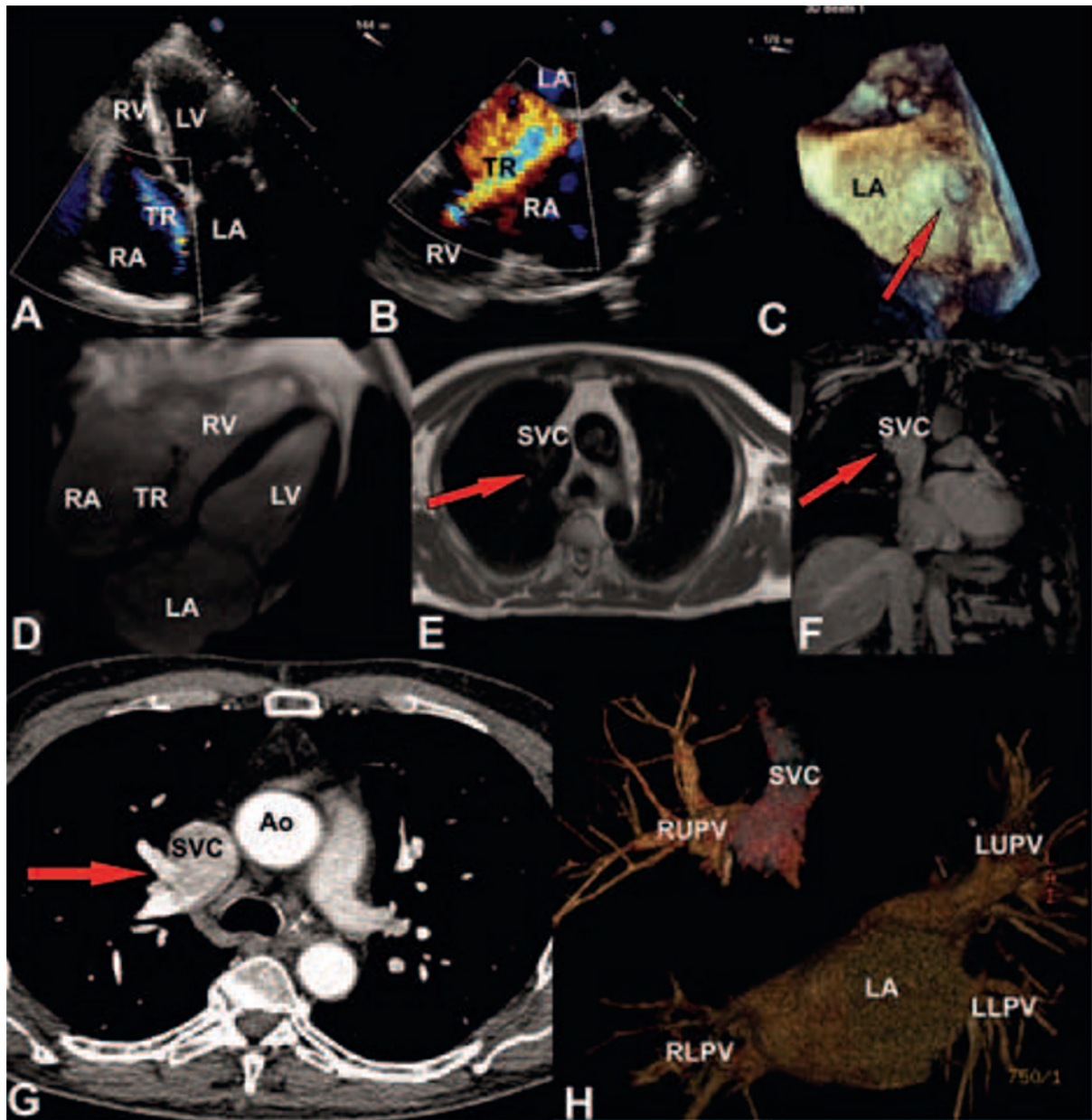


Figure 1. Ultrasound, CMR and CT images of 69-year-old male with isolated partial anomalous pulmonary venous connection. Figure 1A – apical four chamber heart view (TEE) showing atriomegaly and moderate-to-severe tricuspidal regurgitation (TR) (LA – left atrium, RA – right atrium, LV – left ventricle, RV – right ventricle). Figure 1B – the 2D TEE image showing intact atrial septum and moderate-to-severe TR. Figure 1C – 3D TEE image revealed only one right pulmonary venous entry (arrow). Figure 1D – four chamber CMR SSFP heart image showing atriomegaly, RV dilatation and moderate-to-severe TR. Figure 1E and 1F – CMR HASTE and VIBE axial images, respectively, showing dilatation of the superior vena cava (SVC) and anomalous drainage of the right upper pulmonary vein (RUPV) into the dilated SVC (arrows point to the anomalous connection). Figure 1G – contrast-enhanced axial chest CT depicts the anomalous RUPV drainage into the dilated SVC (arrow). Figure 1H – 3D volume-rendered CT image of the anomalous RUPV drainage into the dilated SVC and normal drainage of the right lower pulmonary vein (RLPV), left upper pulmonary vein (LUPV) and left lower pulmonary vein (LLPV) into LA.

not have significant hemodynamic and cardiac structural changes [6,8]. However, other factors are also important and may increase the significance of the shunt: the relative resistance to flow in the normally and anomalously draining lung segments, the relative capacitance of the RA and LA, and any stenosis in the arterial supply to

the anomalously draining lung segment [8]. Occasional patients (as the above described patient) with a single anomalous pulmonary vein do develop symptoms and signs of significant left-to-right shunting with advancing age, possible as the result of worsening LV diastolic function and LA capacitance [8]. Rarely, older patients

may present with pulmonary hypertension, but this is extremely unusual for patients with single pulmonary vein anomaly. We think that our patient have significant shunting despite of single pulmonary vein anomaly because he is symptomatic (fatigue and dyspnoea on physical exertion, palpitations) and has signs of right heart dilatation and volume overload with secondary moderate-to-severe TR. In clinical practise managing older patients with single-vein PAPVC is very important to rule out confounding diseases such as lung or coronary heart disease causing similar to significant shunting symptoms.

The initial diagnosis of PAPVC may be made by echocardiography. The dilation of the RA and RV seen on echocardiography, especially in the absence of a large ASD, should prompt a careful search for PAPVC. The abnormal dilation of coronary sinus or other venous structures is a clue for the diagnosis of PAPVC [8]. Drainage of anomalous pulmonary vein into the SVC causes enlargement of this structure as in our case. In adults with limited acoustic windows, TTE may be inadequate. TEE may be helpful (especially in ruling out atypical ASD), but tomographic imaging modalities – CT and CMR – are superior. CT and CMR are ideally suited for diagnosis of PAPVC and have replaced cardiac catheterization as the primary imaging modalities when the venous anatomy is complex or when echocardiography is suboptimal [8]. Other tests, including chest radiography and ECG, may provide supportive evidence for the diagnosis of PAPVC but are not diagnostic [6].

Chest CT is the modality of choice for the diagnosis of PAPVC as in above described case. It noninvasively and accurately evaluates the presence, course, and number of anomalous veins, associated cardiovascular defects, and any other lung or vascular anomalies. Contrast-enhanced CT allows rapid acquisition of data with high spatial resolution and wide anatomic coverage. Axial images with multiplanar and three-dimensional reconstruction depict any anomalous vein with high sensitivity (100%) [10]. Owing to the increased use of CT to image coronary arteries in adults, more frequent diagnoses of single-vein PAPVC in the future is likely to be expected [8]. Thus, there is a challenge for clinicians to determine the true hemodynamic impact of PAPVC in order to prevent unnecessary surgery while not withholding therapy in patients who would benefit from it.

CMR is as good as CT for anatomic depiction of pulmonary venous structures. Advantages of CMR include lack of ionizing radiation, inherent soft tissue contrast, quantitative analysis of flow, and the ability to acquire multiple vascular phases using a single injection of contrast material (gadolinium) [11]. CMR can provide

additional information, including quantitation of heart chamber volumes, ventricular mass, and can quantify pulmonary blood flow when other modalities, such as echocardiography, yield equivocal findings. Despite the above mentioned advantages of CMR, in our case this imaging modality was not as informative as CT because of couple of reasons: good expertise of CMR specialist and clear clinical questions what to look for performing CMR are essential and, more important, quality of CMR images are very sensitive to inability to hold the breath and arrhythmia.

Surgery is the definitive treatment of PAPVC and is recommended with a significant left-to-right (at least 2:1) shunt. PAPVC of more than one vein usually demands repair [8]. Other indications for surgical treatment are: compression or obstruction of surrounding structures caused by the anomalous vein, recurrent pulmonary infections (especially with Scimitar syndrome) and concomitant surgical repair of other major cardiac lesions [6]. The benefit of surgical intervention must be weighed against the risk of the procedure. The surgical approach is patient specific given the wide variety of the involved anatomy and the presence of other associated cardiac anomalies. Significant left-to-right shunt and the presence of moderate-to-severe secondary TR with significant annular dilatation in our patient favoured surgical approach. The benefit and risk ratio for surgical approach in our case was in favour for surgery taking into account the absence of serious comorbidities and pulmonary hypertension. However, the patient refused to undergo suggested surgery. Thus, our patient was treated medically with bigger dose of beta-blocker to achieve optimal heart rate control, optimal dose of warfarin to maintain INR between 2.0 and 3.0 and angiotensin converting enzyme inhibitor to control elevated blood pressure. Initiation of diuretics was recommended as soon as first symptoms and signs of congestion occur.

4. Conclusions

Isolated PAPVC is a very rare congenital anomaly that should be suspected in any child or adult presenting with the dilatation of RV and RA seen on echocardiography, especially in the absence of a large ASD. Abnormal dilatation of coronary sinus or other venous structures is a clue. Tomographic imaging modalities (CT or CMR) not only allow the comprehensive structural and functional assessment of this anomaly but also help assess the patient's suitability for surgical treatment. Surgery is the definitive treatment of a patient with a significant left-to-right shunt due to PAPVC.

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None

Conflict of interest statement

Authors state no conflict of interest.

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