Diagnostic approach to this rare entity

Sinus venosus atrial septal defect with partial anomalous pulmonary vein return

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Case description

We report a 44-year-old patient, who was initially referred for routine cardiac evaluation, because of family history of sudden cardiac death and a probable new onset of a right bundle branch block (RBBB) on a regular electrocardiogram (ECG) (fig. 1). He stated that he had no cardiac or pulmonary symptoms. In particular, he denied previous unexpected syncope, palpitations or chest pain.

On transthoracic echocardiography, a normal left ventricular ejection fraction of 55% was found without any wall motion abnormalities. Furthermore, dilatation of the right ventricle with systolic dysfunction was found (fractional area change - FAC 33%) with preserved longitudinal function (tricuspid annular plane systolic excursion - TAPSE 24 mm, S' 14 cm/s) (fig. 2).

The patient had a family history of a sudden cardiac death and the possibility of arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) was initially raised. Advanced cardiac imaging was performed for further evaluation.

Cardiovascular magnetic resonance imaging (CMR) (figs 3 and 4) revealed a significant left to right shunt (Qp:Qs = 2:0) caused by a superior sinus venous defect with an anomalous right upper and right middle pulmonary vein connection, as shown in the special CT-based, 3D reconstructed pictures (figs 5 and 6). Furthermore, consistent with the initial echocardiography findings significant right ventricle dilatation and decreased systolic function were detected (fig. 7).

The transoesophageal echocardiography showed a partial anomalous connection of the right upper pulmonary vein into the superior vena cava and, in addition,

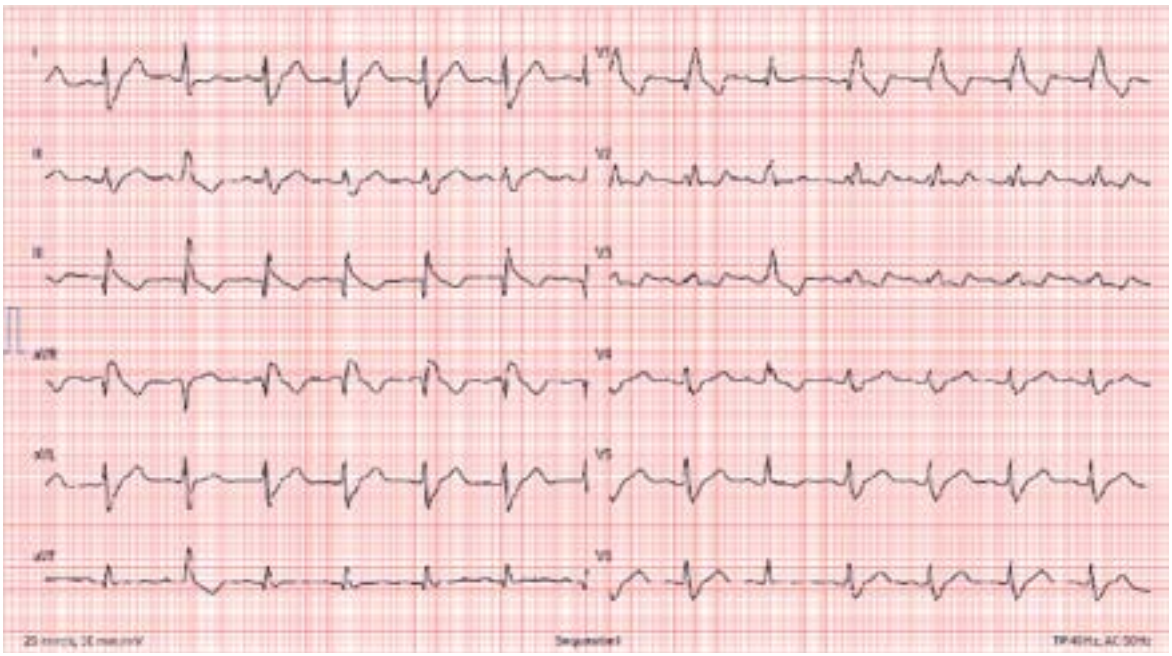

Figure 1: ECG with right bundle branch block.

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a superior sinus venous defect (13 × 17 mm) was confirmed, resulting in the significant left to right shunt. Apart from that, the accessory right middle pulmonary vein connected to superior vena cava and normal connection of the left pulmonary veins and right lower pulmonary vein was identified (fig. 8).

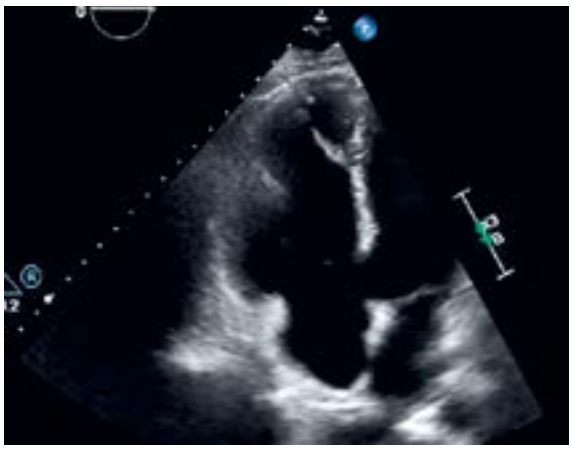

Figure 2: Transthoracic echocardiography showing right ventricular dilatation.

Volumetrie Linker Ventrikel (SSM nach Simpson):

Grösse [cm]	Gewicht [kg]	BSA [m²]	LV-Masse [g]	LV-Masse-Index [g/m²]	LV-EDV [ml]	LV-EDV-Index [ml/m²]	LV-SV [ml]	LV-EF [%]
175	102	2.23	112	51	167	75	98	59

Volumetrie Rechter Ventrikel (SSM nach Simpson, in transversal):

RV-EDV [ml]	RV-EDV-Index [ml/m²]	RV-SV [ml]	RV-EF [%]
353	159	168	48

Vorhöfe:

LA (4cv) [cm²]	RA (4cv) [cm²]
21	24

Norm LA und RA gemäss Andersen, JCMR 2005: <24cm²

MR-Flussmessung:

Lokalisation	Vorwärtsfluss [ml]	Rückwärtsfluss [ml]	Regurgitationsfraktion [%]
Aorta (Pars asc.)	93.7	1.4	1.5
Trunc. Pulm.	189.2	4.3	2.3

Qp/Qs Volume: 2.00 als Hinweis auf das Vorliegen eines Link-Rechs-Shunts.

Figure 3: Result of two dimensional phase contrast magnetic resonance imaging flow measurement, perpendicular to the ascending aorta and pulmonary artery.

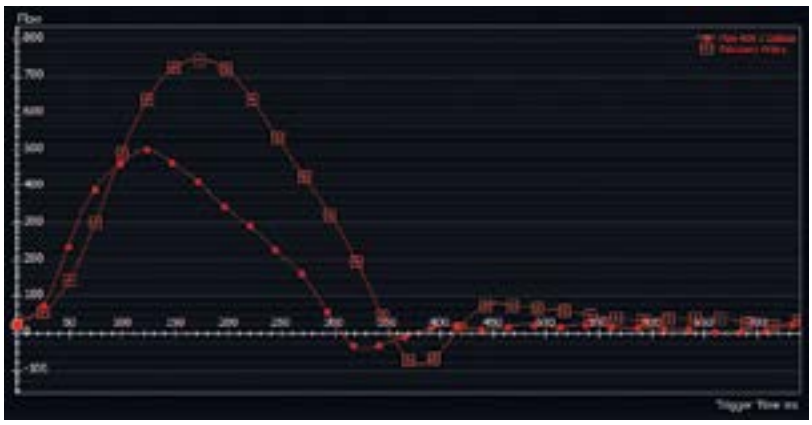

Figure 4: Graph with phase contrast magnetic resonance flow curves in the ascending and pulmonary artery.

To rule out pulmonary hypertension before planning surgery, right and left heart catheterisation was scheduled (fig. 9). Fortunately only mild post-capillary pulmonary hypertension was detected (PVR 0.3 Woods, Wedge 16 mm Hg and PA 22 mm Hg). As expected, measurement, elevated oxygen saturation in the right atrium and pulmonary artery was found.
Coronary artery disease and coronary anomaly were ruled out with coronary computed tomography angiography (fig. 10).

Discussion

Superior sinus venosus defect remains after secundum atrial septal defect (ASD) and primum atrial septal defect, the third most common congenital lesion and it is found in 5% of all cases [1]. It is associated with partial or complete anomalous drainage of the right pulmo-

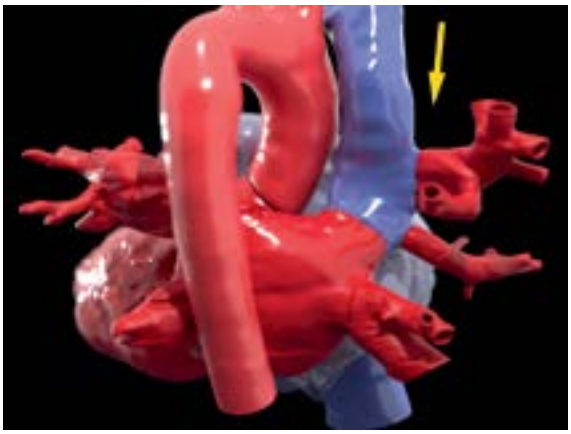

Figure 5: 3D reconstruction with volume rendering of computed tomography (CT) scan-posterior view- showing the partial anomalous drainage of the right and middle pulmonary vein into the superior vena cava (arrow).

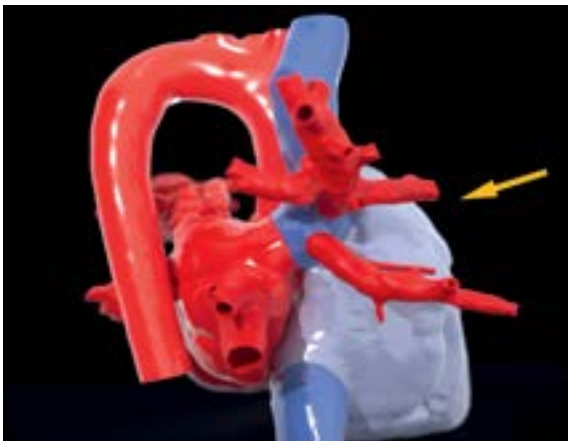

Figure 6: 3D reconstruction with volume rendering of computed tomography (CT) scan-lateral view- showing the partial anomalous drainage of the right and middle pulmonary vein into the superior vena cava (arrow).

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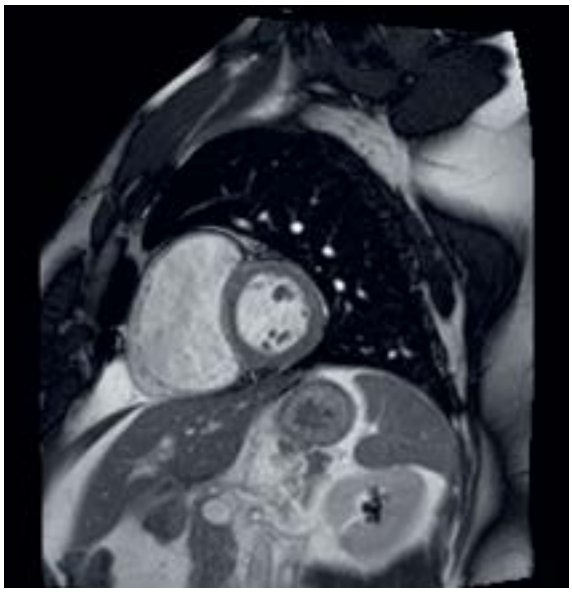

Figure 7: Short axis cine steady-state free precession (SSFP) CMR images, showing significant right heart dilatation.

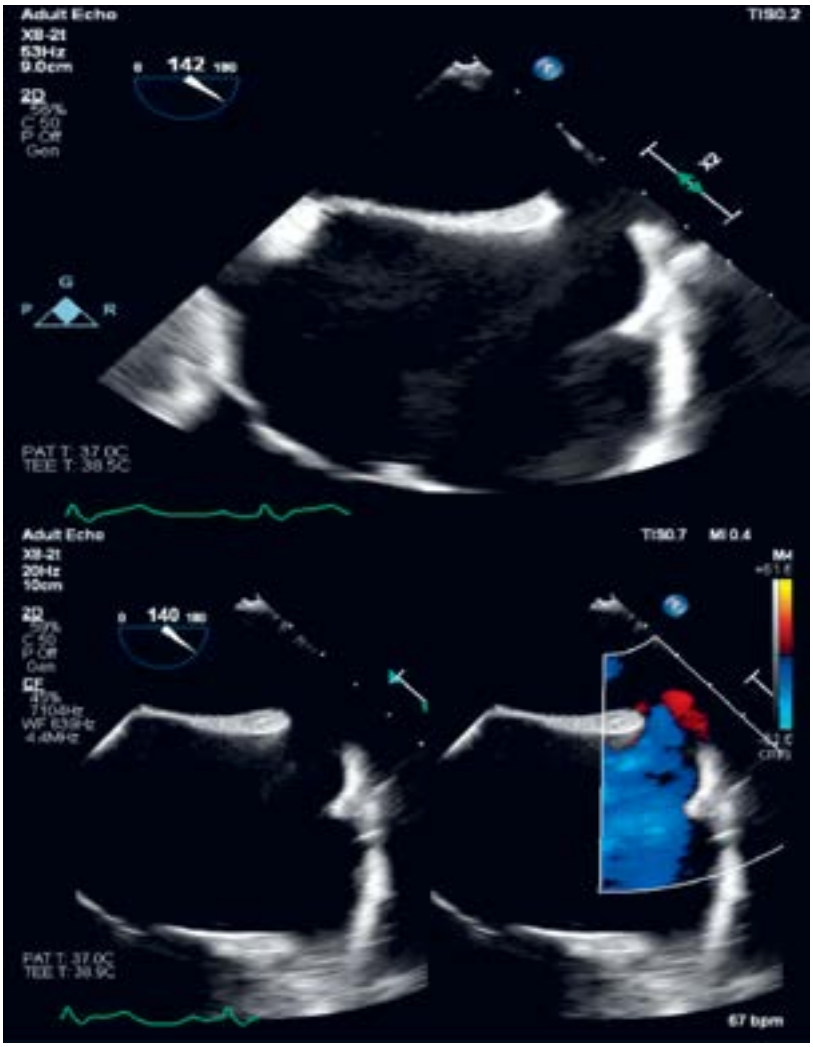

Figure 8: Transoesophageal echocardiography showing sinus venosus atrial septal defect.

nary veins into the superior vena cava or right atrium in about 80–90% [2].

Isolated partial anomalous pulmonary venous connection is a rare congenital anomaly. The first report of anomalous vein drainage was by Winslow in 1739 [3]. It is found in <1% of the population [4]. Right-sided partial anomalous pulmonary venous connections into the superior vena cava are the most common (90% right vs. 17% left) [5]. An ASD should be evaluated in every patient with suspected pulmonary hypertension and right ventricle dilatation.

In most cases the partial pulmonary venous connection is found incidentally in computed tomography (CT) or magnetic resonance imaging (MRI) due to lack of clinical symptoms. The total anomalous pulmonary venous connection is seen in about 1–5% of congenital heart diseases and can in young age cause early cyanosis and death [6]. Pulmonary veins are also an origin of supraventricular arrhythmias and the knowledge of anomalous pulmonary venous drainage is essential for planning of interventions including catheter ablation [7].

Unrepaired anomalous pulmonary venous connection with a significant shunt leads slowly to increase in the pulmonary vascular resistance and development of pulmonary arterial hypertension. If an atrial septal defect is also present, finally it comes to the shunt reversal and formation of an Eisenmenger physiology, which is connected with high surgical risk and is one of the contraindications of further surgical repair [1].

Echocardiography is the initial diagnostic tool in the evaluation of any congenital heart disease. Because of limited field of views and picture quality, this technique is in most of cases insufficient to detect the anomalous pulmonary venous connection. The right heart catheterization represents the gold standard for assessing the pulmonary hemodynamics and detection of the possible shunt location by exposure of step up in oxygen saturation.

The CT scan is providing an excellent additional diagnostic benefit including the 3 dimensional reconstructed images (videos 1 und 2). Cardiovascular magnetic resonance (CMR) helps to detect a complex anatomy and more importantly to quantify the shunt size, especially when invasive tests are not conclusive.

Patients newly diagnosed with partial anomalous pulmonary venous connections should be referred to a special grown up cardiac heart disease center (GUCH) for evaluation of corrective surgery. According to the 2020 guidelines of the European Society of Cardiology for the management of GUCH patients, the septal defect closure should be evaluated in case of significant shunt (Qp:Qs >1,5) and pulmonary vascular resistance between 3–5 Wood units (class IIA indication) [1], and is

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Hämodynamik	Ort	[mmHg] a/s	[mmHg] v/d	[mmHg] m/ed	HR
Ruhe	PCW	20	20	16	
Ruhe	PA	32	16	22	
Ruhe	RV	41	7	10	
Ruhe	RA	13	12	10	
Sauerstoffsättigung					
Ruhe	AO	93	%		
Ruhe	PA	85	%		
Ruhe	RA	83	%		

Figure 9: Right heart catheterisation showing an elevated saturation in the pulmonary artery.

Figure 10: 3D reconstruction of a cardiac CT scan showing the anatomy of the coronary arteries.

recommended (class IB indication) in evidence of RV volume overload and no signs of pulmonary arterial hypertension or left ventricle disease [1].

In our case after careful consideration and interdisciplinary evaluation with colleagues from the grown up cardiac heart disease centre (GUCH) the patient was sent for further evaluation of an invasive management.

The patient underwent a repair operation including xeno-pericardial patch closure of the sinus venosus defect and reestablishment of the pulmonary circulation.

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Disclosure statement
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