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Sinus venosus atrial septal defect and partial anomalous pulmonary venous connection in a patient with dextrocardia

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SUMMARY

An 85-year-old woman presenting with symptomatic pulmonary hypertension (PH) with a history of dextrocardia was referred to our facility for treatment of heart failure. Significant shunting was detected by measuring oxygen saturation during right heart catheterisation (RHC). CT with contrast revealed sinus venosus atrial septal defect (SVASD) and partial anomalous pulmonary venous connection (PAPVC), in which the left upper and middle lobar pulmonary veins returned to the superior vena cava and right atrium. Despite medical treatment, the patient died, and an autopsy was performed. SVASD and PAPVC are rare congenital anomalies. RHC with measurement of oxygen saturation and CT with contrast should be considered in patients with unexplained right atrial and ventricular enlargement or suspected PH.

BACKGROUND

Dextrocardia with situs inversus is a rare congenital abnormality in which the heart is a mirror-image reversal situated on right side of the thoracic cavity. The reported incidence ranges from 0.01% to 0.35% of all live births.¹ This condition is often accompanied by additional cardiac malformations.² Similarly, sinus venosus atrial septal defect (SVASD) is a rare congenital heart disease with an interatrial communication defect at the junction of the right atrium (RA) and vena cava, which accounts for 5%–10% of all types of atrial septal defects (ASD).³ SVASD is commonly associated with partial anomalous pulmonary venous connection (PAPVC), which is when blood from one or more pulmonary veins returns either directly to the RA or indirectly through a variety of systemic venous pathways that connect with the anomalous pulmonary vein. We describe a case of an older woman who presented with symptomatic pulmonary hypertension (PH) and was found to have SVASD and PAPVC with dextrocardia. To the best of our knowledge, this is the first reported case of a single patient who had SVASD and PAPVC with dextrocardia, and in which an autopsy was performed. In addition, we discuss the diagnostic and therapeutic challenges encountered.

CASE PRESENTATION

An 85-year-old woman with a history of dextrocardia with situs inversus was referred to our facility for treatment of decompensated heart failure. She had been experiencing a gradual increase in

exertional dyspnoea and bilateral lower extremity oedema for 2 months, although up to then she had been independent and able to walk. She had been diagnosed with heart failure 5 years prior to admission. At that time, transthoracic echocardiography (TTE) revealed right heart enlargement and pericardial effusion; however, the causes of heart failure were unclear. Her medical history also included atrial fibrillation, diabetes and interstitial pneumonia.

On physical examination, the patient had an irregular heart rhythm of 110 bpm. Her blood pressure was 127/91 mm Hg. SpO₂ was 90% under 5 L/min of oxygen using a nasal cannula. She had jugular vein distention and leg pitting oedema. Chest auscultation revealed crackles on both the lung bases.

INVESTIGATIONS

A 12-lead ECG showed atrial fibrillation, complete right bundle branch block and low voltage in precordial leads. Chest radiography (posterior-anterior view) showed cardiomegaly with the apex located to the right, bilateral pleural effusion and pulmonary congestion. Blood tests showed that the brain natriuretic peptide level was moderately elevated at 626 pg/mL, while troponin I was negative. The patient had mild renal dysfunction (creatinine, 0.80 mg/dL) and hyponatraemia (sodium, 126 mEq/L). She was intubated and admitted to the intensive care unit to maintain her oxygen level and manage her restlessness. TTE showed severe enlargement of the RA and right ventricle (RV), small left ventricular size with intraventricular septum displacement toward the left, and mild-moderate tricuspid regurgitation with PH (estimated RV pressure of 57 mm Hg). Transoesophageal echocardiography (TEE) showed a Patent Foramen Ovale (PFO). The initial clinical impression was PH, considering the medical history of interstitial pneumonia. However, KL-6 levels were not elevated. Right heart catheterisation (RHC) revealed an increase in oxygen saturation from 53.0% at the superior vena cava (SVC) to 87.5% in the high RA (table 1).

The mean pulmonary artery (PA) pressure was 39 mm Hg, mean pulmonary capillary wedge pressure was 24 mm Hg and pulmonary vascular resistance (PVR) was 1.66. Chest CT with contrast revealed the presence of three anomalous pulmonary veins (figure 1). The first anomalous vein originated from the left upper lobe and drained into



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Table 1 Measurement of oxygen saturation

Position	Oxygen saturation (%)
SVC	53.0
IVC	38.5
High RA	87.5
Low RA	79.7
RV	81.7
PA	82.8
FA	91.8

The calculated pulmonary-to-systemic flow ratio (Qp/Qs) was 4.7 (table 2).

FA, femoral artery; IVC, inferior vena cava; PA, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

the SVC. The second anomalous vein also originated from the left upper lobe with drainage into the SVC-RA junction. The third anomalous vein originated from the left middle lobe with drainage into a large superior SVASD (figure 2). Right heart cardiomegaly, dilation of the PA, bilateral pleural effusions and pericardial effusion were also present. MRI was limited to atrial fibrillation. Pericardiocentesis was performed and approximately 1170 mL of pericardial fluid were evacuated.

DIFFERENTIAL DIAGNOSIS

The diagnoses of SVASD and PAPVC are challenging. Echocardiography is typically used to diagnose ASD. However, diagnosis of SVASD on TTE may sometimes be difficult because the atrial septum appears intact in most view.⁴ On the other hand, the validity of TEE in the diagnosis of SVASD and PAPVC has been reported.^{5 6} In our case, due to the unusual situation of dextrocardia with situs inversus, TEE was performed by an experienced operator; however, congenital anomalies could still not be detected. The clue to diagnosis was a step-up in oxygen saturation during RHC. A left-to-right shunt was detected and localised because of a significant step-up in blood oxygen saturation at elevated RA pressure. Careful interpretation of CT with contrast is helpful in providing accurate information on cardiac shunts and anomalous veins with right heart enlargement.

TREATMENT

The management of SVASD and PAPVC is considered individually based on the clinical presentation and degree of left-to-right shunt. Adults with SVASD show impaired functional capacity, right heart enlargement, and presence of left-to-right shunt ($Qp/Qs \geq 1.5$), which should be surgically repaired. There is one guideline that surgery is recommended when PA pressure is less than 50% of systemic pressure and PVR is less than one-third of systemic vascular resistance⁷; however, this should

Table 2 Measurement of right heart catheterisation

Parameter	Value
RAP (mm Hg)	26
RVP (mm Hg)	56/8
PAP (mm Hg)	59/25 (mean 39)
PCWP (mm Hg)	24
PVR (wood units)	1.66
CO Fick (L/min)	9.06
Qp/Qs	4.7

CO, cardiac output; PAP, pulmonary artery pressure; PCWP, pulmonary capillary wedge pressure; PVR, pulmonary vascular resistance; RAP, right atrium pressure; RVP, right ventricle pressure.

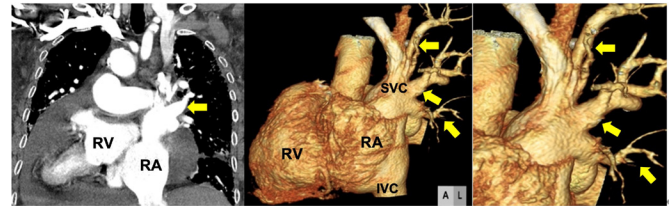


Figure 1 Chest CT with contrast and three-dimensional reconstruction shows partial anomalous pulmonary venous connection (yellow arrows) from both the left upper and middle lobe to SVC and RA. IVC, inferior vena cava; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

be individualised based on the anatomy and haemodynamics of each patient. Repair of PAPVC is recommended at the time of SVASD closure.⁷ Surgical treatment for SVASD is associated with low morbidity and mortality; however, older age at operation and New York Heart Association (NYHA) class III or IV symptoms are independent predictors of late mortality.⁸ Cardiac surgery in patients with dextrocardia has rarely been reported. In particular, dextrocardia with sinus inversus is a rare condition that may lead to surgical challenges due to the mirror-image anatomy of associated vasculatures.⁹ In our case, the patient was a poor surgical candidate because of her age. Extensive vascular remodelling was unlikely to be reversible by surgical intervention. In this situation, conservative management with diuretics is often performed. After discussion with the patient's family and cardiologist, the patient opted for conservative management. In addition, PA vasodilators can be considered in patients with Eisenmenger syndrome. However, there are few randomised controlled studies to guide the management of patients with PH associated with congenital heart disease.¹⁰ In our case, given the patient's high surgical risk, medical treatment without PA vasodilators was chosen. This is because RHC did not reveal Eisenmenger syndrome, and there was concern about decreased cardiac output by PA vasodilators, leading to reduced pressure in the right side of the heart and increased shunt flow. Diuretics and thoracentesis temporarily improved her respiratory status.

OUTCOME AND FOLLOW-UP

The patient's advanced heart failure status progressed to persistent pleural effusion and respiratory failure type II. Despite receiving diuretic treatment, the patient died on the 67th hospital day.

An autopsy of the lungs and heart was performed. Gross findings were cardiomegaly (heart weight, 580g; figure 3), remarkable dilatation of the RA and RV, and compression of the left ventricle by a displaced interventricular septum (figure 4). A pericardial effusion was also observed. Two pulmonary veins originating from the left upper lobe communicated to the SVC

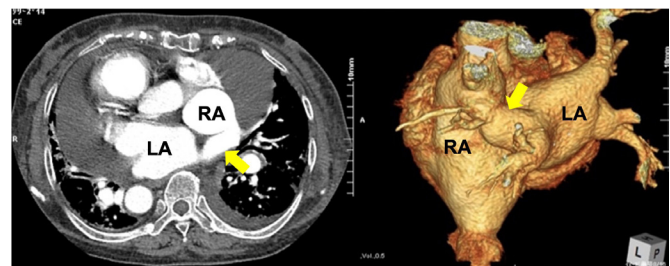


Figure 2 Chest CT with contrast and three-dimensional reconstruction shows sinus venosus atrial septal defect (yellow arrow). LA, left atrium; RA, right atrium.

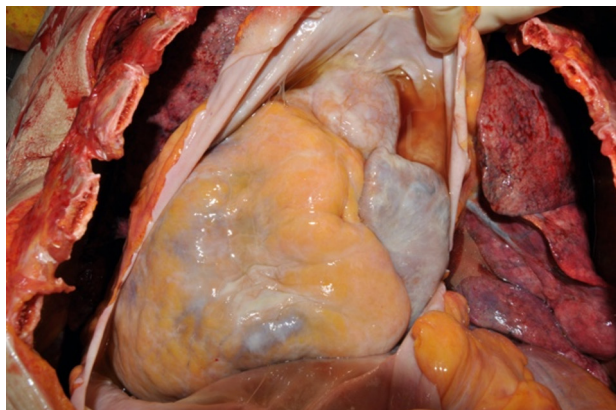


Figure 3 Gross finding of the heart showing a dextrocardia and cardiomegaly.

(figure 5). There was slit and tunnel-like communication, which was considered a PFO in the atrial septum. The lungs showed congestion and oedema. There were two lung lobes on the right side and three on the left side due to dextrocardia with situs inversus. Pathological finding showed PA Intimal proliferation and thickening, this suggested PH.

DISCUSSION

We describe a case of an older woman who presented with symptomatic PH and was found to have SVASD and PAPVC with dextrocardia. Our case highlights that measurement of oxygen saturation in RHC and CT with contrast was helpful in defining SVASD and PAPVC.

Dextrocardia, in which the heart is located on the right side of the thoracic cavity, is caused because the heart loops to the left instead of the right during the embryonic period. There are different types of dextrocardia; situs inversus with dextrocardia is the complete inversion of the position of the thoracic and abdominal visceral organs. On the other hand, SVASD result from the lack of septation between the pulmonary veins and the superior cava vein or RA.¹¹ SVASD is commonly associated with PAPVC¹²; more than 90% of patients with SVASD also have PAPVC.⁸ Dextrocardia is often accompanied by additional cardiac malformations¹; however, the incidence of congenital heart disease in dextrocardia with situs inversus is low, ranging from 2% to 5%.¹³ A case of congenital anomalies occurring in the same patient has never been reported before.

Clinical guidelines recommend that ASD should be diagnosed by imaging techniques with demonstration of shunting across the defect and evidence of RV volume overload and any associated anomalies. Patients with unexplained RV volume overload should be referred to an ACHD centre for further diagnostic studies to rule out obscure ASD or PAPVC.¹⁴ In our case, the clue to diagnosis was a step-up in oxygen saturation

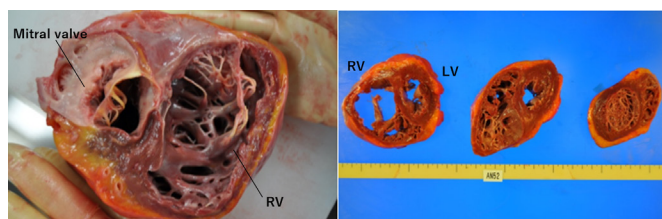


Figure 4 Cross-section of the heart showing remarkable dilatation of the right ventricle (RV). LV, right ventricle.

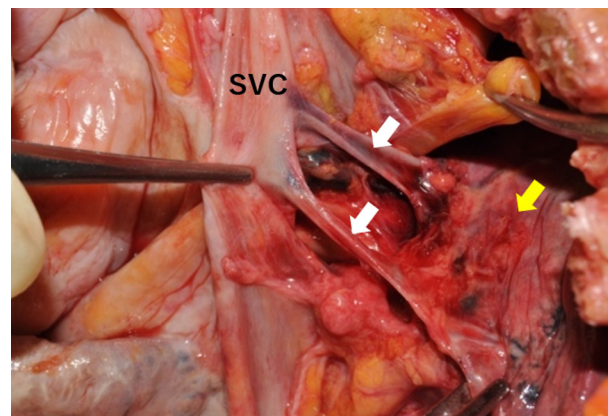


Figure 5 Gross finding of connection SVC and anomalous veins (white arrows) from left upper lobe (yellow arrow). SVC, superior vena cava.

during RHC. Suspecting shunt disease, we carefully checked the CT with contrast and were able to diagnose these congenital heart diseases. Moreover, three-dimensional (3D) reconstruction of CT images leads to a better understanding of complex abnormalities.

Several patients who had large unrepaired ASD survived for 80 years or even longer like this case. However, untreated large defects are associated with a reduced life expectancy. The mortality rates of unrepaired ASD are low for the first two decades (0.6% and 0.7% per year, respectively), increasing to 5.4% per year in the fifth decade and 7.5% per year in the sixth decade and over with 90% of patients die by age 60 years.¹⁵ There are case reports that show diagnosis and management in patients with PAPVC and SVASD during late adulthood, these patients were not able to undergo surgery due to age or severe PH.^{16 17} Surgical treatment for SVASD is associated with low morbidity and mortality; however, older age at operation and NYHA class III or IV symptoms are independent predictors of late mortality.⁸ Early detection of the condition is important to lead to successful surgical repair.

In this case, there are several haemodynamic questions. The patient presented with normal PVR and elevated PCWP, suggestive of underlying left heart failure as the aetiology for PH, in general. As for the elevated PCWP, it may be due to the left ventricular-right ventricular interdependence; dilatation of the RV shifts the interventricular septum toward the left and these changes may contribute to low cardiac output state by decreasing

Learning points

- ▶ We should understand the anatomy of patients with dextrocardia by at least transthoracic echocardiogram and CT scan prior to consider them not having a congenital heart disease.
- ▶ The CT with contrast should be performed, especially if echocardiogram does not explain the aetiology of right heart enlargement.
- ▶ Right heart catheterisation with measurement of oxygen saturation could be clue for diagnosis in patients with unexplained right atrial and ventricular enlargement or suspected pulmonary hypertension.
- ▶ Congenital heart diseases require early recognition that might lead to successful surgical intervention.

LV distensibility, preload and ventricular elastance.¹⁸ In calculation, PVR is underestimated due to elevated PCWP. Based on the pathological findings, PVR may be increased.

We report that SVASD and PAPVC with dextrocardia remained asymptomatic until the development of significant PH in an 85-year-old woman. Both PAPVC and SVASD have the potential to remain undiagnosed for many years. These congenital anomalies should be systematically evaluated in patients with unexplained right atrial and ventricular enlargement or suspected PH. RHC with measurement of oxygen saturation and CT with contrast, especially 3D reconstruction, are useful for diagnosis. Recognition of the condition is significant as early detection makes surgical repair a feasible option.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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