



CASE REPORT

A case of sinus venosus atrial septal defect misdiagnosed as primary pulmonary hypertension



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Abstract We present a case of sinus venosus atrial septal defect in a patient who was previously diagnosed as having primary pulmonary hypertension in a tertiary care center. Our findings are based on 2-dimensional trans-thoracic echocardiography, chest X-ray and surface electrocardiogram. A 26-year-old man, previously diagnosed as a case of primary pulmonary hypertension, presented to the emergency department (ED) with chest pain and breathlessness on exertion. Cardiac biomarkers were within their normal ranges. Surface electrocardiogram showed right atrial and ventricular overload with right axis deviation. Chest imaging noted enlarged central pulmonary vascularity with bilateral plethoric lung fields.

Trans-thoracic echocardiography showed a dilated right atria and ventricle with severe tricuspid regurgitation and severe pulmonary artery hypertension with an intact atrial septum. Surprisingly, the transoesophageal echocardiogram revealed the presence of a sinus venosus superior vena cava-type atrial septal defect with the right pulmonary vein draining into the right atria.

In this full-text version, we present a more detailed discussion of sinus-venous atrial septal defect associated with partial anomalous pulmonary venous return that was wrongly diagnosed as a case of primary pulmonary hypertension in a tertiary care center.

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1. Introduction

Sinus venous atrial septal defects (ASD) vary in that the atrial septum is intact except in the superior portion adjacent to the superior vena cava and can coexist with partial anomalous pulmonary venous connections. Diagnosis by trans-thoracic echocardiography (TTE) is difficult, although trans-oesophageal echocardiography (TEE) can contribute to the diagnosis of sinus venous defects and assessment of associated anomalies. However, even in tertiary care centers, facilities in developing countries may lack the ability to perform trans-oesophageal echocardiography (TEE) and cardiac catheterization, and sinus venous ASD is often misdiagnosed as primary pulmonary hypertension. We report a case of superior vena caval sinus venous ASD that was misdiagnosed and wrongly treated as primary pulmonary hypertension based on 2-dimensional trans-thoracic echocardiography (TTE), chest X-Ray and electrocardiographic (ECG) findings.

2. Case report

A 26-year-old male presented to the emergency department with chief complain of acute onset chest pain for 4 hours. The pain was retrosternal without any typical radiation and was associated with uneasiness and chest heaviness. On inquiring about past history, the patient noted that he had experienced breathlessness for the previous 3–4 months. This was gradual in onset and progressive in nature; initially he felt breathlessness on usual ordinal outdoor activities with slight limitations of his physical activities, and at present he became breathless on less than ordinal activities with marked limitation of physical activities. There is no history of seasonal or diurnal variation of

his breathlessness. That patient had no history of cough, hemoptysis, back pain, chest trauma, orthopnea or paroxysmal nocturnal dyspnea. There is no childhood history of acute rheumatic fever. The patient is non-hypertensive, non-diabetic, a non-smoker, a non-alcoholic and vegetarian in diet. There is no past history of pulmonary tuberculosis, systemic hypertension or diabetes mellitus. The patient belongs to a lower socioeconomic stratum. After receiving his medical records from a tertiary care centre in eastern Uttar Pradesh in north India, it was found that he was on oral diuretics, calcium channel blockers and sildenafil citrate for primary pulmonary hypertension for the last 2 years with some degree of symptomatic relief in between. However, he had left the treatment for the previous 6 months due to financial issues.

On examination, the patient was hemodynamically stable with a pulse rate of 110 per minute that was regular, normovolumic, and normal in character without any radio radial or radio femoral delay. All peripheral pulses are equally palpable with normal condition of the arterial wall. The patient's blood pressure was 128/78 mm Hg in the right arm while in a sitting position using an adult blood pressure measuring cuff. There was no significant difference between the upper limbs or between the upper and lower limbs. The patient was of adequate build and nutrition with a body mass index of 21. He was afebrile at the time of presentation, with an arterial oxygen saturation of 98% measured by a pulse oxymeter. Pallor, cyanosis, clubbing, icterus, edema and lymph node enlargement were absent. The jugular venous pressure was normal with prominent v wave and y descent. Hepatojugular reflux was absent. On examination of the cardiovascular system, we found that the S1 was normal and the S2 was normally split with a loud P2 and a grade III pansystolic murmur present at lower left parasternal area that increased in intensity on inspiration.

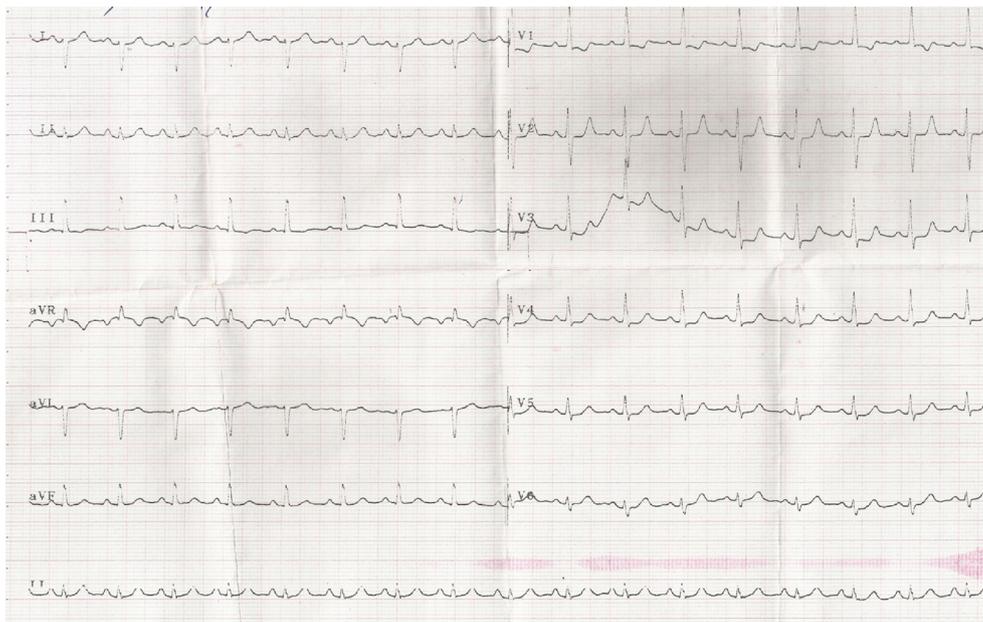


Figure 1 Electrocardiogram of patient showing right axis deviation with RAA and RVH. RAA-Right atrial abnormality, RVH-Right ventricular hypertrophy.

Other systemic examinations were within the normal limits. A provisional diagnosis of primary pulmonary hypertension with right ventricular ischemia was made, and IV treatment with diuretics, a calcium channel blocker and opioid analgesics for pain relief was initiated. Upon further investigations, we found that the patient's complete blood count and kidney and liver function tests were normal. Quantitative assessment of cardiac biomarkers found that they were within the normal range. A surface electrocardiogram suggested right axis deviation with right atrial abnormality and right ventricular hypertrophy (Fig. 1). A chest X-ray PA view was suggestive of right atrial, right ventricular enlargement with a prominent pulmonary artery and plethoric lung fields (Fig. 2). The 2D echocardiography was suggestive of a dilated right atrium and ventricle with severe TR and severe PAH with an intact atrial and ventricular septum (Figs. 3 and 4). All of the



Figure 2 Chest X-ray showing RAA, RVH, dilated pulmonary artery with plethoric lung fields.

investigative findings were in favour of his previous diagnosis of primary pulmonary hypertension (PPH) except for the chest X-ray findings. Thus, we performed transoesophageal echocardiography (TEE) to rule out any possibility of sinus venosus ASD. Surprisingly, the TEE examination revealed a superior vena cava-type sinus venosus atrial septal defect with a prominent left to right shunt with dilated right atrium and right ventricle and an anomalous right upper pulmonary venous drainage into the right atria (Fig. 5). The patient's discharge was postponed in view of the sinus venosus ASD, and a catheterization study was scheduled for the following day. The catheterization confirmed the TEE findings with increased pulmonary blood flow (increased Qp) and reversible pulmonary arterial hypertension. The patient was referred to the Cardiothoracic Department for surgical closure of the defect. The elective surgical correction of this abnormality was performed under extracorporeal circulation without any complications. After right atriotomy, the interatrial defect was closed with the use of a Dacron device. Enlargement of the superior vena cava was performed with a pericardiectomy that allowed closure of the right atriotomy. At a post-surgical 3-month follow-up, the patient showed marked improvement in his symptoms of breathlessness and chest pain.

3. Discussion

Sinus venosus defect (SVD) is a rare cardiac abnormality consisting of a disturbed connection of the vena cava to the right atrium and an abnormal pulmonary vein connection to the vena cava. Thus, the malformation provokes an interatrial shunting outside the interatrial septum.¹ SVD is found in 2% to 10% of patients with cardiac atrial septal defects. Clinical manifestations range from benign to

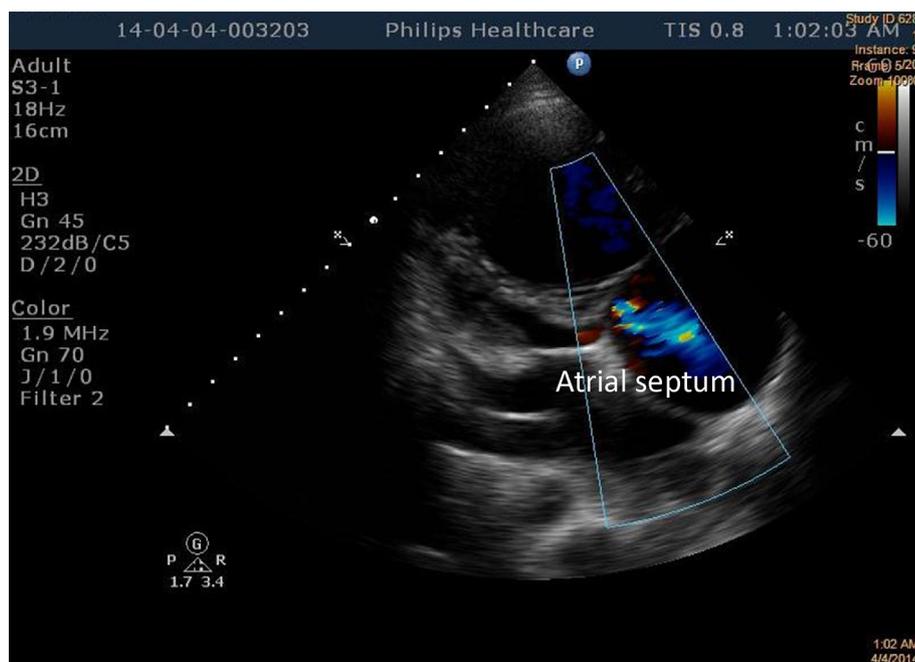


Figure 3 Two dimensional ECHO (PLAX view) showing intact atrial septum.

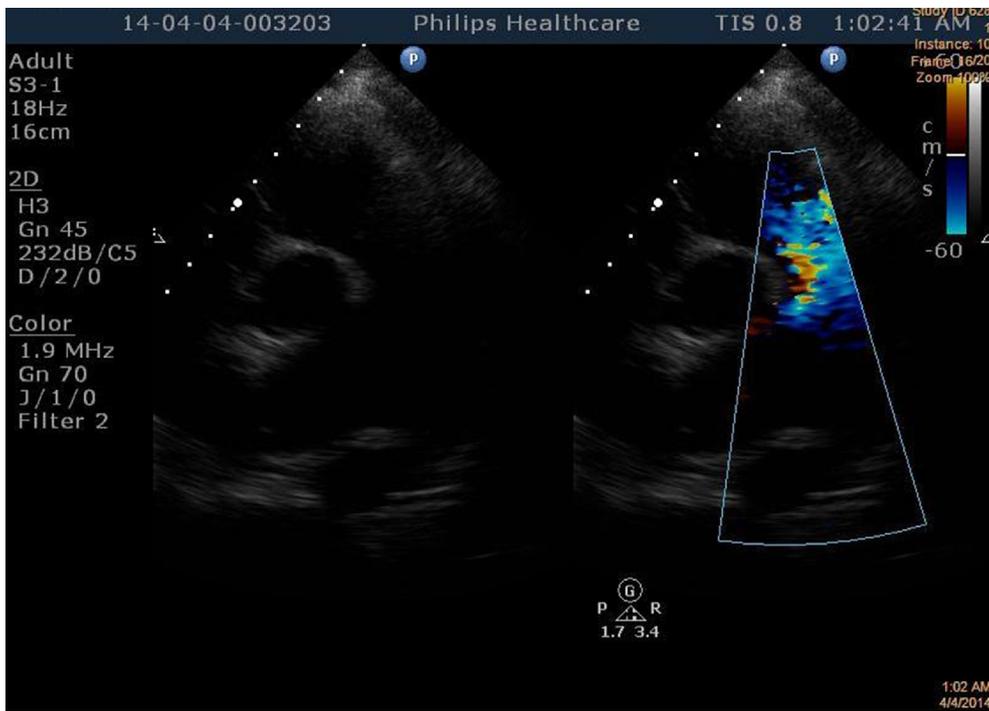


Figure 4 Two dimensional ECHO showing dilated pulmonary arteries.

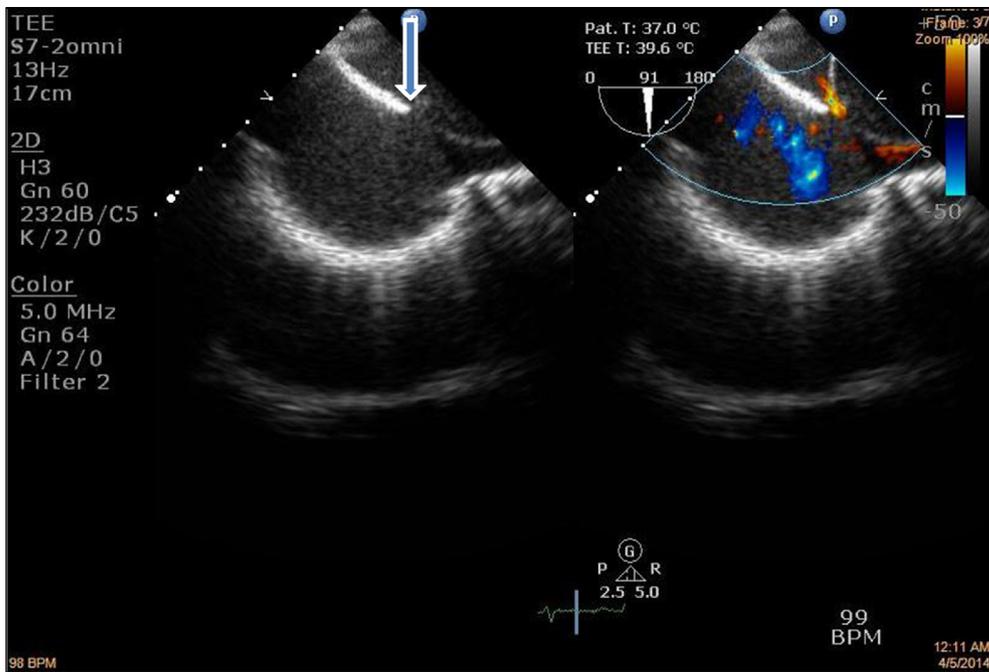


Figure 5 TEE showing superior vena cava type sinus venosus atrial septal defect (marked by arrow).

severe, with most patients having minimal, if any, functional limitation to exercise. The primary cardiac disturbances include pulmonary hypertension, arrhythmias,² and extrinsic compression of the pulmonary artery when the giant remnant valve of sinus venosus is present.³ Patients with SVD have a shorter life expectancy. Diagnosis of SVD is often more difficult than for other forms of ASD and may require special imaging, such as transoesophageal

echocardiography, magnetic resonance imaging (MRI), or computed tomographic scanning, and the possibility of a sinus venosus ASD should be considered for any patient with unexplained right atrial and right ventricular dilation. Catheter closure is not possible, and the treatment is surgical.

This case emphasizes the usefulness of TEE in the setting of dysnea and chest pain with a pre-hospital

diagnosis of primary pulmonary hypertension, especially in young patients. Discovery of SVD in this setting is rare, but in this case, it led to decisive surgical management. This type of malformation can be overlooked at conventional transthoracic echocardiography because of its posterior (far field) location. TEE is ideally suited to diagnose SVD, given the proximity of the transducer to the defect. TEE is accurate for the diagnosis of SVD should be undertaken in any patient with unexplained dilation of the right side of the heart. The best view on TEE for recognition of ASD sinus venosus (SVC type) is a bicaval view.⁴ ECG-gated multislice CT appears to be a promising tool in exploring cardiac morphological abnormalities and has been described recently in a case of SVD as a supplement to echocardiography.⁵ The goal of diagnostic testing is to confirm that PAH exists and to identify its underlying cause. In the case reported here, the echocardiogram was suggestive of pulmonary hypertension. In patients in whom there are sufficient cardiac anomalies on TTE, explaining PAH does not require further diagnostic testing, but if there is no evidence of cardiac anomaly on TTE to clarify the PAH, TEE should be performed. Sinus venosus defect may carry a worse prognosis than other forms of ASD and may need to be treated at a younger age either via medical therapy and/or surgical closure. Surgical repair of ASD, including sinus venosus defect, in patients over 40 years of age, increases long-term survival and decreases the incidence of heart failure.^{6,7}

4. Summary

Atrial septal defects can be misdiagnosed as primary pulmonary arterial hypertension in adults. With the increased use of trans-oesophageal echocardiography (TEE), careful assessment of these defects and their

associated pulmonary venous anomalies in patients may be beneficial as surgical repair of these defects can improve outcomes.

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