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UNMASKING THE HIDDEN DEFECT: A RARE CASE OF SINUS VENOSUS ATRIAL SEPTAL DEFECT COMPLICATED BY PULMONARY ARTERIAL HYPERTENSION

Cardiology

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ABSTRACT

Sinus venosus atrial septal defect (SVASD) is a rare congenital heart disease usually seen in adults and characterized by abnormal communication between the systemic and pulmonary circulations, often accompanied by abnormal coronary sinus and pulmonary venous connections. We report a case of a 52-year-old male who presented to the emergency department in an unconscious state following a syncopal episode while awaiting a cardiology consult. He regained consciousness after 20 minutes without post-syncopal confusion or focal neurological deficits. Clinical history revealed two years of exertional dyspnea, palpitations, and pedal edema, with recent worsening of symptoms. Clinical evaluation revealed hypotension, hypoxia, elevated jugular venous pressure (JVP), and signs of right heart strain. Investigations including echocardiography and contrast-enhanced CT of the chest showed cardiomegaly, dilated right atrium and ventricle, pulmonary artery dilation, and a sinus venosus atrial septal defect (ASD) of the superior vena cava (SVC) type. The patient was stabilized with diuretics and vasopressors and subsequently underwent successful surgical repair of the ASD. He demonstrated significant symptomatic improvement postoperatively and remains stable on follow-up. This case highlights the importance of considering congenital heart defects in adult patients presenting with unexplained syncope and signs of right heart overload.

KEYWORDS

INTRODUCTION

Atrial septal defects (ASDs) are among the most common forms of congenital heart disease diagnosed in adults. Sinus venosus ASD (SV-ASD), is a rarer subtype accounting for 5-10% of all ASD and presents diagnostic and management challenges.[1] Unlike the more prevalent ostium secundum ASD, SV-ASD is located at the junction of the superior vena cava (SVC) or, less commonly, the inferior vena cava (IVC) with the right atrium. It is frequently associated with partial anomalous pulmonary venous return (PAPVR), particularly of the right upper pulmonary vein draining into the SVC or right atrium, which contributes to significant left-to-right shunting [2,3]. The presence of PAH in SV-ASD patients not only complicates the clinical course but also significantly influences management decisions, especially regarding the timing and feasibility of surgical repair.

We report a case of a middle-aged male who presented with syncope and was later diagnosed with SV-ASD and significant PAH, highlighting the importance of timely recognition and multidisciplinary management of this rare but potentially life-threatening congenital cardiac anomaly.

CASE REPORT

A 52-year-old man, known case of Diabetes mellitus for 1 year was brought to the emergency department in an unconscious state. The patient was awaiting a cardiology consultation at GB Pant Hospital when he suddenly lost consciousness. As per the attendants who witnessed the event, there were no abnormal body movements, tongue biting, frothing from mouth or urinary incontinence during the event. He was brought immediately to the medicine emergency where he regained consciousness twenty minutes later. After the syncopal event, the patient's was not confused and had no motor or sensory deficits. He denied any symptoms such as nausea, vomiting, aura, chest pain, palpitations, sweating, dizziness, or blurred vision preceding the syncope. He had no history of similar event in the past. The patient never smoked or used drugs. He denied taking any medications.

Upon detailed history taking, he reported shortness of breath on exertion for the past two years, associated with self-limited exertional palpitations. There was also history of pedal edema for 2 years that appeared on prolonged standing and on exertion and used to get relieved as he lay down. There were no symptoms and rest and the patients was also able to do his routine daily activities without much difficulty. These symptoms were relatively stable for 1.5 years and the patients sought no treatment. However following an episode of fever with cough 6 months ago, these symptoms worsened and he was admitted in a private hospital for 5 days. During the course of hospital stay, a CECT chest was done that revealed Cardiomegaly with minimal pericardial effusion. The patient was referred to a higher centre for evaluation where he was planned for an Echocardiography but he did not follow up. Four months later, patient developed progressive pedal Edema and was taken to GBPH where an Echo showed Cardiomegaly with dilated RA RV and Low pressure TR, LV ejection fraction was 55-60%. Patient was on regular follow up from GBPH ever since and had presented for a repeat ECHO when he developed the episode of Loss of consciousness and was transferred to LNJP hospital. He had no family history of Sudden cardiac death, Rheumatic heart disease, coronary artery disease, heart failure, or congenital heart disease.

On physical examination after regaining consciousness, the patient was alert and well-oriented to time, place, and person. He had a Blood pressure of 84/50 mm Hg, pulse was 96 beats/minute, SpO2 was 85% on room air and respiratory rate was 20/min. He was started on Nor-Adrenaline infusion and low flow oxygen and responded promptly. There was pitting pedal edema up to the shins, and the jugular venous pressure was elevated to 8 cm of H_2O. On cardiovascular examination, the apex was localised to 5th ICS 7cm from sternal border, normal in character. There was right ventricular parasternal heave grade 3 and a palpable sound was heard in the left parasternal 2nd ICS. On auscultation a grade 3 pansystolic murmur was appreciated in left 5th ICS parasternally that increased in intensity on inspiration. S3 was also appreciated in left 5th parasternal ICS and a loud P2 was heard in left parasternal 2nd ICS. There were no other sounds or murmurs. Abdomen was soft, non-tender and not distended. Bowel sounds were normal. Respiratory auscultation revealed normal Vescicular breath sounds with no added sounds.

Routine blood investigations, including cardiac enzymes, are provided in Table 1. ProBNP levels were 5148pg/ml, Trop T and I levels were within normal range, and CPK T and MB levels were borderline elevated. His ABG was suggestive of type 1 respiratory failure while Chest Xray showed Cardiomegaly with Clear lung fields and minimal pleural effusion. ECG suggestive of bifascicular block (RBBB with

LPFB- Right axis deviation) with ST-T changes in chest leads and inferior leads likely suggestive of strain. CECT of the chest revealed dilated RA and RV measuring 60mm and 58mm respectively. The main pulmonary trunk, right and left pulmonary arteries were also dilated and measured 43mm, 33mm and 25mm in transverse diameter respectively, suggestive Pulmonary Hypertension. A detailed ECHO was sought given these findings and it revealed a Sinus venosus ASD of SVC type with dilated RA and RV and mild low pressure TR. LA was normal and Left ventricular ejection fraction was 55-60%.

The patient was started on diuretics after the BP had stabilised and showed improvement. Three days later, he had minimal pedal Edema and was able to maintain saturation of 95% on Room air with a BP of 118/74mmHg. The patient was then referred to cardiothoracic surgery where he underwent a successful ASD repair. The patient was discharged from the hospital five days postoperatively on oral aspirin 75 mg daily, metoprolol succinate 50 mg twice daily, and Lasilactone 10/50 once daily. He is currently on regular follow up from Cardiology department and has good good cardiac rehabilitation.

Table 1: Routine Blood Investigations Of Patient Including Cardiac Biomarkers

PARAMETER	Patient's value	NORMAL RANGE
Hemoglobin	13.2g/dl	13-16g/dl
TLC	8500	4000-11000/cc
DLC	64/33/2/1	
Platelet Count	1.52lac	1.5-4lac
ESR	09	
Total Bilirubin	0.5mg/dl	0.2-1.3mg/dl
Alanine Transaminase	52 U/L	<50 U/L
Alkaline Phosphatase	134 U/L	38-126 U/L
CPK Total	124	10-120 ug/L
CPK MB	28	5-25 IU/L
Aspartate Transaminase	72 U/L	17-59 U/L
Blood Urea	33 mg/dl	15-39 mg/dl
Serum Creatinine	0.8mg/dl	0.66-1.25 mg/dl
Sodium	128 mmol/L	135-145 mmol/L
Potassium	4.3 mmol/L	3.5-5 mmol/L
Ddimer	394	
proBNP	5148pg/ml	<300pg/ml
Trop T	<0.01	
Trop I	<0.01	

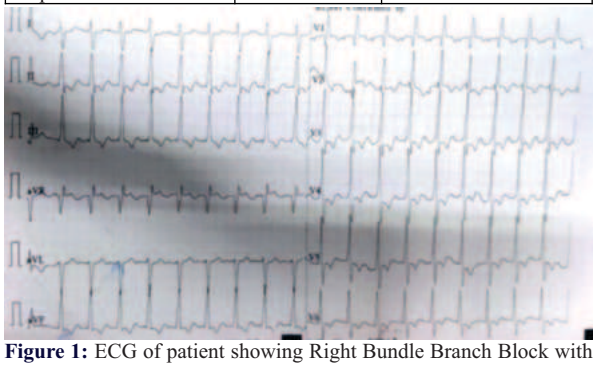

Figure 1: ECG of patient showing Right Bundle Branch Block with Right axis deviation with ST-T changes in chest leads and inferior leads

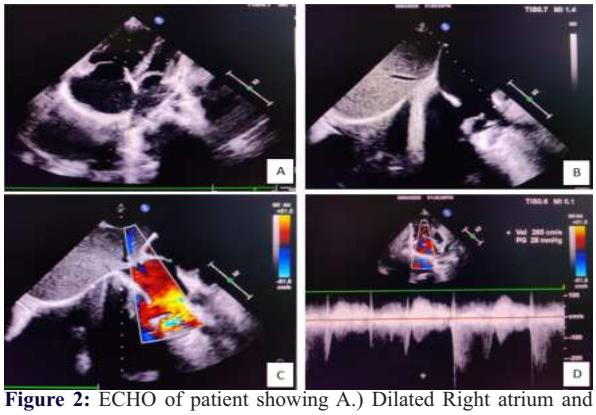

Figure 2: ECHO of patient showing A.) Dilated Right atrium and Right Ventricle. B.) Sinus venosus ASD of SVC type measuring 1.9cm. C.) SVC type ASD showing left to right shunt D.) Mild gradient across RA and RV

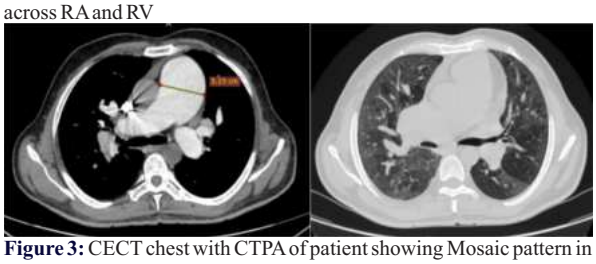

Figure 3: CECT chest with CTPA of patient showing Mosaic pattern in bilateral lung parenchyma, cardiomegaly with Dilated Right heart chambers with dilated Main, right and left pulmonary arteries, suggestive of Pulmonary hypertension with right sided volume overload

DISCUSSION

Unrepaired ASDs account for 7–10% of all congenital heart defects and are the second most common congenital heart defect in adults [4]. The overall incidence of ASDs is approximately 1.6–2.4 per 1,000 live births [5]. SV-ASD typically occurs near the junction of the superior vena cava (SVC) and the right atrium, although it may also occur near the inferior vena cava (IVC). It allows left-to-right shunting of blood, leading to chronic volume overload of the right atrium (RA) and right ventricle (RV), and eventually pulmonary overcirculation.

In many cases, especially in the SVC type, the defect is associated with PAPVR, particularly involving the right upper pulmonary vein draining into the SVC or RA. Interestingly, not all SV-ASDs have PAPVR. According to a retrospective echocardiographic and surgical review, 10–15% of SV-ASDs occur without demonstrable anomalous pulmonary venous drainage [2]. This subset presents diagnostic challenges and may delay appropriate surgical correction.

Patients typically remain asymptomatic during early life and may present in adulthood with exertional dyspnea, fatigue, palpitations (often due to atrial arrhythmias), right heart failure, syncope (in cases of advanced PAH or arrhythmias). Over time, the left-to-right shunt can lead to pulmonary vascular remodeling and increased pulmonary artery pressure, progressing to Eisenmenger's physiology in extreme cases. The progression from precapillary PAH to Eisenmenger syndrome (reversal of shunt direction) marks a critical threshold where surgical closure is contraindicated due to the risk of right ventricular failure. [6]

Diagnosis of SV-ASD can be challenging with transthoracic echocardiography (TTE) alone due to its posterior and superior location. Transesophageal echocardiography (TEE), cardiac MRI, or cardiac CT angiography are often required to confirm the diagnosis and evaluate associated PAPVR. [7]

Surgical repair remains the mainstay of treatment for SV-ASD, especially when diagnosed in adulthood before irreversible pulmonary vascular disease ensues. Closure of the defect, often accompanied by redirection of anomalous pulmonary veins if present, results in symptom relief and improved right heart function.[8][9] However, in patients with severe pulmonary arterial hypertension, surgical intervention requires cautious evaluation, as abrupt closure of the shunt can lead to right ventricular failure from an acute rise in afterload. Medical management prior to surgery focused on hemodynamic stabilization, while postoperative management including β-blockers for arrhythmia prevention and anticoagulants reduce thromboembolic risk.

Early diagnosis and surgical repair of SV-ASD confer excellent long-term outcomes. Postoperative follow-up focuses on monitoring for arrhythmias, right heart function, and residual shunting. The presence of longstanding pulmonary hypertension, as in this case, may necessitate long-term pulmonary vasodilator therapy or more frequent surveillance.

CONCLUSION

This case emphasizes the importance of considering congenital heart disease in patients presenting with right heart dilatation and severe PAH. Sinus venosus ASD, while rare, should be a differential in adult patients with unexplained right heart enlargement or pulmonary

hypertension. The association with PAPVR is strong, but not universal. Up to 30% of uncorrected ASD cases develop PAH, which dramatically worsens prognosis if left untreated. Timely diagnosis via advanced imaging and prompt surgical correction are key to preventing irreversible cardiopulmonary complications.

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