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CASE SERIES
Cardiac imaging

Association of ruptured sinus of Valsalva aneurysm and congenital ventricular septal defect: a case series

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Background Ruptured sinus of Valsalva aneurysm (RSOVA) is rare, and it is more common in Asians. Typically, the patient presents with acute/subacute shortness of breath (SOB) and chest pain. Echocardiography is the gold standard for diagnosis in most of these cases. Surgery has remained the first line of management.

Case summary We present two cases of RSOVA in which the patients presented to the emergency department with SOB. Their preoperative echocardiography results showed RSOVA into the right ventricle. During surgical repair, ventricular septal defect (VSD) was also found.

Discussion RSOVA is frequently associated with other congenital anomalies, and most often with VSD. In our cases, we believe that VSDs were missed preoperatively because either the large aneurysmal sacs covered the VSD or there was overlap between the two shunts. Additionally, in the first case, right ventricular pressure was high approaching systemic pressure, which probably reduced the shunt across the VSD. Early intervention is recommended to prevent endocarditis or enlargement of the ruptured aneurysm; long-term results were excellent after surgical repair. Most patients undergo surgery between 20 and 40 years of age, and the reported survival rate is 95% at 20 years. If left untreated, patients typically die of heart failure or endocarditis within 1 year after onset of symptoms.

Keywords Ruptured sinus of Valsalva • Echocardiography • Ventricular septal defect • Case series

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Learning points

- Ruptured sinus of Valsalva aneurysm (RSOVA) is more common in young Asian men. Patients presenting with RSOVA must have a vigilant preoperative assessment for coexisting cardiac defects that may have been missed, including transoesophageal echocardiography with a three-dimensional reconstruction, if it is available. Magnetic resonance imaging is an option if the patient is stable and can tolerate it.
- Surgical repair is still the modality of choice. In an era of less invasive procedures, percutaneous closure of RSOVA, particularly if it is not associated with infundibular ventricular septal defect (VSD), is feasible. Thus, diagnosing the existence of VSD becomes important when determining the most appropriate treatment pathway.

Timeline

Patient 1	
Day of admission	Admission due to shortness of breath (SOB). Transthoracic echocardiography (TTE) showed dilated sinus of Valsalva with a ruptured right coronary sinus into the right ventricle outflow tract (RVOT)
Day 6 of admission	Transoesophageal echocardiography (TOE) could not identify a ventricular septal defect (VSD)
Procedure day	Repair of the ruptured right coronary sinus of Valsalva, tricuspid valve, and VSD
Postoperative Day 1	Transfer from the intensive care unit to the high dependency unit
Postoperative Day 5	The patient was discharged home
7-Month follow-up	Follow-up with TTE showed no residual defects
Patient 2	
Day of admission	Admission due to SOB. TTE showed ruptured aneurysm of the right coronary sinus of Valsalva into the RVOT
Day 1 of admission	TOE could not identify VSD
Procedure day	Repair of ruptured right coronary sinus of Valsalva and VSD. Drainage of bilateral pleural effusion. Post-repair TOE showed a tiny flow over the VSD
Postoperative Day 2	Transfer from the intensive care unit to the high dependency unit
Postoperative Day 5	The patient was discharged home
5-Month follow-up	Follow-up with TTE showed the same tiny shunt through the VSD, with no significant changes

Case Presentations

Patient 1

A 23-year-old Pakistani man who had been diagnosed with a cardiac murmur as a child, presented to the emergency department (ED) after a few weeks of palpitations and progressive shortness of breath (SOB). On presentation, his vital signs were within normal limits, and he had normal oxygen saturation on room air. The neck veins were not congested. A continuous murmur was heard over the left sternal border, but the lungs were clear. Labs were normal except for the markedly elevated pro-B-type natriuretic peptide (pro-BNP) (4670 pg/mL). Electrocardiogram (ECG) showed sinus rhythm with a rightward axis deviation and incomplete right bundle branch block. Cardiomegaly and prominent bronchovascular markings were seen with a plain chest X-ray. Preoperative two-dimensional (2D) transthoracic echocardiography (TTE) showed a dilated sinus of Valsalva. It was 4.2 cm in diameter (normal < 4 cm) with a ruptured right coronary sinus into the right ventricle (RV) outflow tract, and there was continuous systolic and diastolic flow from the aorta to the RV. No ventricular septal defect (VSD) was detected (*Figure 1*, *Video 1*). The patient's left ventricular ejection fraction was 57% (normal > 50%). The right atrium and ventricle were severely dilated. There was severe tricuspid regurgitation and elevation of the RV systolic pressure at 104 mmHg. The tricuspid annulus was also severely dilated but the aortic valve showed normal anatomy and function. A computed tomography angiogram showed significant dilatation of the pulmonary artery, with enlargement of the right atrium and ventricle.

The patient underwent surgical repair. Intraoperative findings were similar to the findings preoperative TTE; additionally, there was a subaortic VSD. He underwent repair of the ruptured right coronary sinus of Valsalva and VSD using a tailored, synthetic Hemashield Dacron patch (HP) and repair of the tricuspid valve using a 30 mm annuloplasty ring. The intraoperative transoesophageal echocardiography (TOE) showed that there was a competent tricuspid valve with no residual shunt. The postoperative course was uneventful, and the patient was discharged on postoperative Day 5. He was followed up in our clinic on a regular basis for 6 months, complaining of occasional SOB. He underwent a 7-month follow-up TTE that showed no residual defects (*Figure 2*).

Patient 2

A 35-year-old Indian man with no medical history presented to the ED with SOB for 2 days, which had a gradual onset but a progressive course. On presentation, his vital signs were within normal limits and

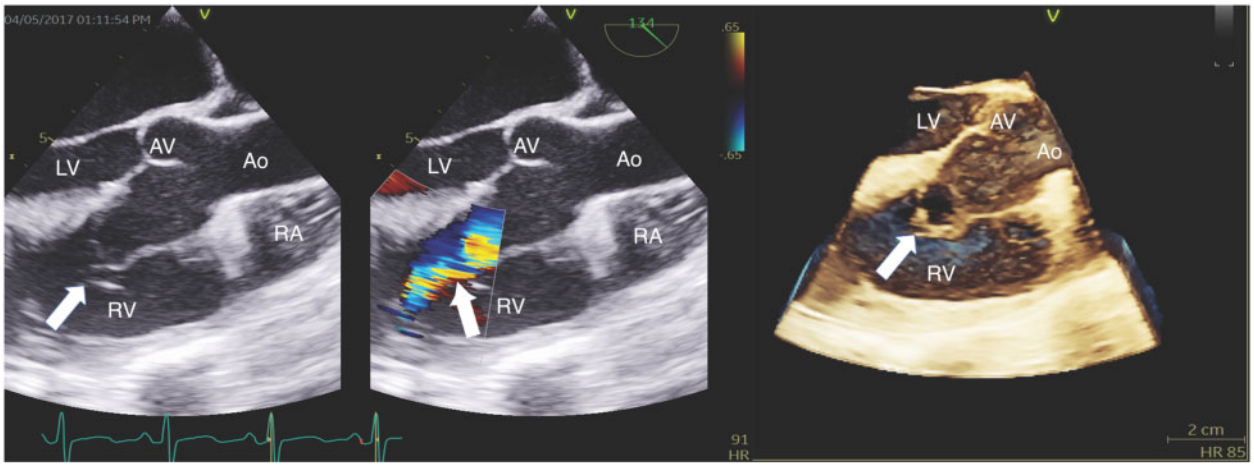


Figure 1 A transoesophageal echocardiography long-axis view (120°) with three-dimensional image showing the ruptured sinus of Valsalva aneurysm with a left-to-right shunt towards the right ventricle. Ao, aorta; AV, aortic valve; LV, left ventricle; RA, right atrium; RV, right ventricle.

he had normal oxygen saturation on room air. The neck veins were not congested. A continuous murmur was heard over the left sternal border. Chest examination showed decreased air entry into the lung bases with crepitation. Labs were normal except for the markedly elevated pro-BNP result (1325 pg/mL). ECG was normal. Chest X-ray showed increased bronchovascular marking with bilateral mild pleural effusion. Preoperative TTE and TOE showed a ruptured aneurysm of the right coronary sinus of Valsalva into the right ventricular outflow tract, with continuous systolic and diastolic flow from the aorta to the RV, but no VSD was detected (*Figure 3*, *Video 2*). The left ventricle ejection fraction was 55%. The aortic valve showed normal anatomy and function.

The patient underwent surgical repair. A small subaortic VSD was discovered intraoperatively along with the ruptured sinus of Valsalva. The VSD was repaired using a pledgeted prolene stitch after repairing the Valsalva defect using a double-tailored HP. Postoperative TOE showed a tiny shunt through the VSD. The patient had an uneventful postoperative course and was discharged on postoperative Day 5.

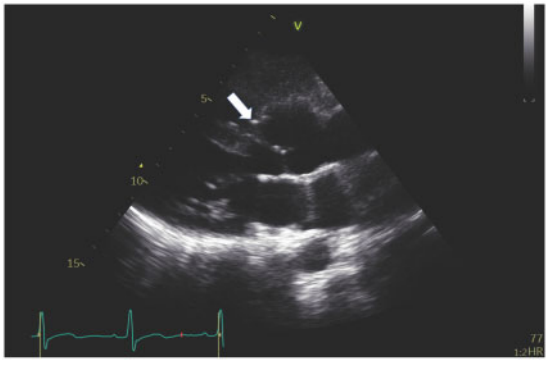


Figure 2 Postoperative transthoracic echocardiography long-axis parasternal view showing the repair site.

He was followed up by our clinic on a regular basis for 7 months with no complaints, and a 5-month follow-up TTE showed the same tiny shunt through the VSD with no significant change (*Figure 4*).

Discussion

Sinus of Valsalva aneurysms is relatively uncommon congenital heart anomaly, with an overall incidence ranging from 0.1% to 3.5% reported in literatures.[1] This type of aneurysm is predominant in men (4:1), and its incidence is highest in the Asian population.[2] The aneurysm is commonly found in the right coronary sinus (77%), less commonly in the non-coronary sinus (23%), and rarely in the left coronary sinus.[3]

Aneurysms of the sinus of Valsalva are thought to result from the absence of normal elastic and muscular tissue, which creates thinning of the aortic sinus walls.[4] These aneurysms are frequently associated with other congenital anomalies, most often VSD (30–60%), bicuspid aortic valve (15–20%), and aortic regurgitation (44–50%).[5,6] In the infundibular variant of VSD, the lack of an anatomical muscular conal septal support in the right ventricular outflow tract creates an unsupported aortic sinus and aortic valve cusp to prolapse into the probable VSD orifice, which is dragged into the right ventricular outflow tract. The whole sinus usually becomes aneurysmal, and its final appearance must be distinguished from the classic windsock shape of the ruptured sinus of Valsalva aneurysm (RSOVA).[7]

The combined association between a membranous VSD and a sinus of Valsalva aneurysm could be a consequence of defectively fused both sides of the distal bulbous septa in the foetal life, where the base composes the right and non-coronary sinuses of Valsalva. The resulting congenital deficiency along the aortic annulus may be exceedingly correlated to other defects in the ventricular membranous septum.[8,9] A sinus of Valsalva aneurysm can be asymptomatic for long periods of time, but occasionally it causes symptoms that are secondary to the right-sided ventricular outflow obstruction or compression of the coronary arteries.

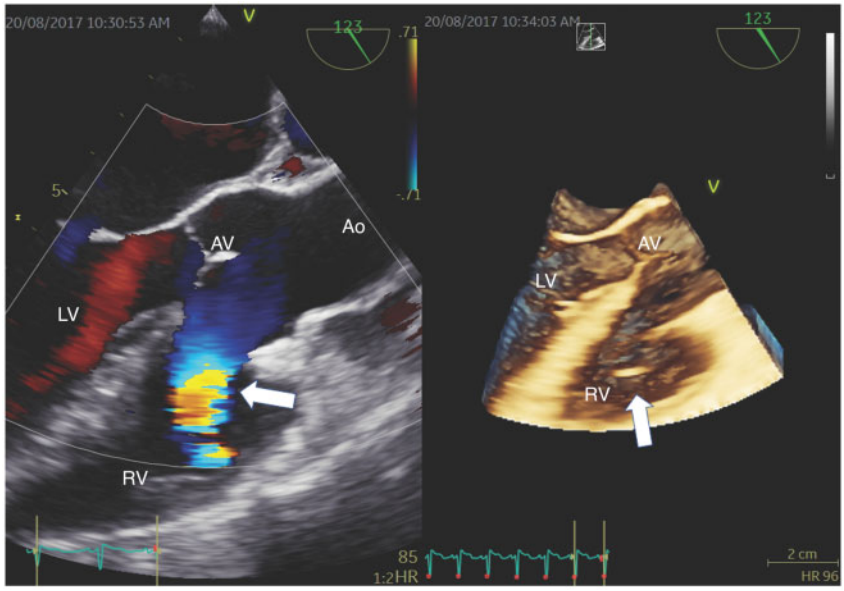


Figure 3 Left image showed a transoesophageal echocardiography long-axis view (120°) showing the ruptured sinus of Valsalva aneurysm with a left-to-right shunt towards the right ventricle. The right image shows a three-dimensional view of the ruptured sinus. Ao, •••; AV, •••; LV, left ventricle; RV, right ventricle.

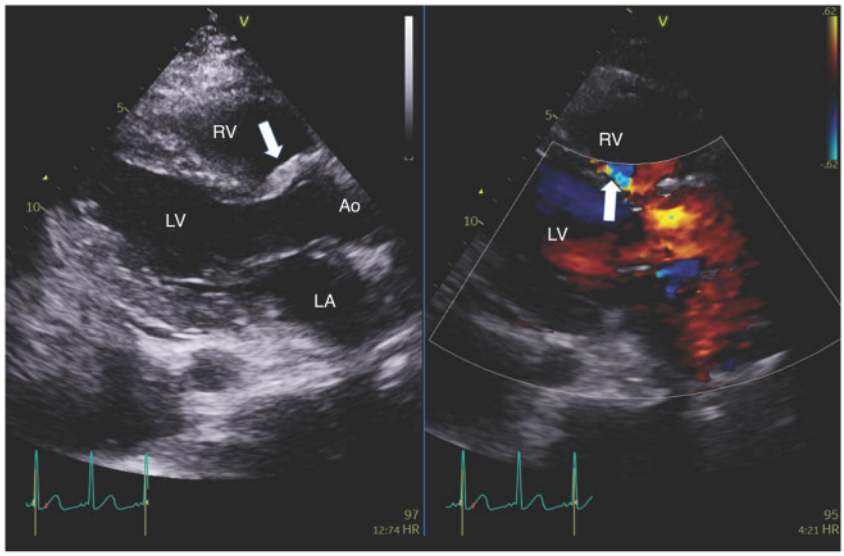
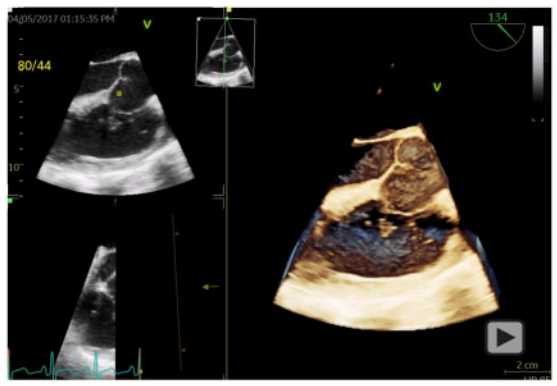
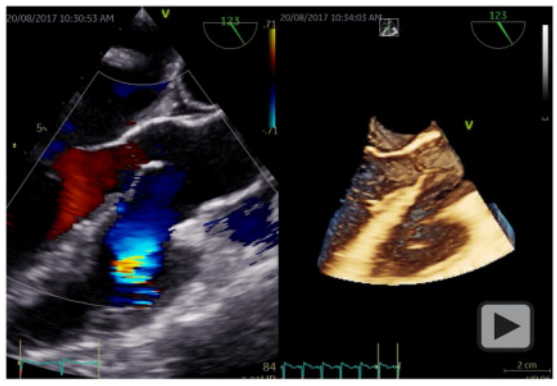


Figure 4 A postoperative transthoracic echocardiography long-axis parasternal view showing the repair site with a tiny residual flow through the ventricular septal defect. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.

Sudden rupture is often the first manifestation of the lesion.[10] Rupture of the aneurysmal sac may occur spontaneously, or it may be precipitated by exertion, blunt trauma, or cardiac catheterization. A substantial number of ruptures develop long after puberty but before age 40 years, with a mean age of 35.8 years and a range of 13–65 years.[11] Rupture into the RV is most common (60–90%), followed by rupture into the right atrium (10%) and the left atrium (2–3%). VSDs, as well as aortic valve regurgitation and bicuspid aortic valve, are frequently associated with RSOVA, particularly when aneurysms rupture into the RV.[12,13] The principal VSD associated with RSOVA is the supracristal plane (or infundibular).[14]



Video 1 A TOE long axis view (120°) with three-dimensional image showing the RSOVA into the RV.



Video 2 A TOE long-axis view (120°) showing the RSOVA with a left-to-right shunt towards the RV with a three-dimensional view of the ruptured sinus.

The characteristics of a sinus of Valsalva aneurysm share some anatomical dissimilarities between Asian patients and their western counterparts. For example, an aneurysm of the sinus of Valsalva in Asian patients is distinguished by a higher incidence, more aneurysms arising from the right coronary sinus (85.8% vs. 67.9%), more tendency to aneurysmal rupture into the RV (72.5% vs. 60%), a higher incidence of combination with VSDs (52.4% vs. 37.5%), and a lower incidence of bicuspid aortic valve associated defect (0.6% vs. 7.8%). However, Asian and Western patient series reported a similar incidence of association with aortic regurgitation (33.6% vs. 32.7%).[15]

Transthoracic echocardiography successfully identifies sinus of Valsalva aneurysm in most cases. However, TOE may be necessary in as many as 25% of cases. A ruptured aneurysm that is identified using echocardiography frequently has a 'windsock' appearance; it has an elongated tubular structure that expands and collapses with the cardiac cycle. It arises from the body of the aortic sinus and extends into an adjacent cardiac cavity. An associated VSD may occasionally remain undetected if it is occluded by the wall of the aneurysm. Some case reports of patients with RSOVA reported that VSD was missed, and therefore, TOE is recommended for complete evaluation.[16]

In addition, three-dimensional (3D) echocardiography is primarily useful in the precise anatomical delineation of RSOVA, particularly if it is associated with VSD.[17–20] In the presence of a coexisting VSD, the large RSOVA shunt overlaps VSD flow, which may be difficult to recognise on 2D echocardiography. Thus, 3D echocardiography identifies the two abnormal flows in most cases. Magnetic resonance imaging (MRI) also allows for accurate evaluation of the anatomy, including areas that are difficult to assess so it may be considered in stable patients.

In our cases, we believe that VSDs were missed preoperatively because either the large aneurysmal sacs covered the VSD or there was overlap between the two shunts. Additionally, in the first case, right ventricular pressure was high, approaching systemic pressure, which probably reduced the shunt across the VSD.

Early repair of RSOVA is recommended to prevent endocarditis or enlargement of the ruptured aneurysm, and long-term results are excellent after surgical repair.[1,11] Most patients undergo surgery between 20 and 40 years of age, and the reported survival rate is 95% at 20 years. If left untreated, patients typically die of heart failure or endocarditis within 1 year after symptom onset.

In an era of less invasive procedures, transcatheter closure is a feasible alternative to surgical repair of RSOVA, either primary or with a residual shunt postsurgical repair.[21,22] However, surgical repair is the preferred method to treat RSOVA with infundibular VSD. Transcatheter closure may be attempted in RSOVA combined with perimembranous VSD.[21]

In conclusion, patients presenting with RSOVA must have a vigilant preoperative assessment for coexisting cardiac defects that may have been missed by TTE. We recommend TOE with a 3D reconstruction if available, to identify coexisting cardiac defects. MRI is an option if the patient is stable and can tolerate it.

Lead author biography

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Supplementary material

Supplementary material is available at *European Heart Journal - Case Reports* online.

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Slide sets: A fully edited slide set detailing this case and suitable for local presentation is available online as Supplementary data.

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

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