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Journal

Texas Heart Institute Journal, 50(3)

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Publication Date

2023-05-01

DOI

10.14503/THIJ-22-8022

Peer reviewed

The Texas Heart Institute® Journal

Case Reports

Ruptured Sinus of Valsalva Aneurysm Diagnosed on Coronary Computed Tomography Angiography in a Patient With Trisomy 13 Syndrome

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Abstract

Trisomy 13 is a rare chromosomal disorder in which all or a percentage (mosaicism) of cells contain an extra 13th chromosome. Sinus of Valsalva aneurysms are rare, with an incidence of 0.1% to 3.5% of all congenital heart defects. This article reports the case of a patient with trisomy 13 with a new systolic murmur found to have a ruptured sinus of Valsalva aneurysm diagnosed on coronary computed tomography angiography. This is the first case to report sinus of Valsalva aneurysm rupture secondary to Streptococcus viridans endocarditis in a patient with trisomy 13 syndrome and highlights the importance of coronary computed tomography angiography in noninvasive imaging and surgical planning.

Keywords: Sinus of Valsalva; aneurysm; trisomy 13 syndrome; computed tomography angiography

Case Report

Presentation and Physical Examination

The patient was incidentally found during an outpatient clinic visit to have an asymptomatic new grade 2/6 systolic murmur, which was loudest at the lower left sternal border without radiation.

Medical History

The patient is a 30-year-old man with trisomy 13 with mosaicism, hypertension, type 2 diabetes, Wilms tumor status postnephrectomy, and no known congenital cardiac history.

Differential Diagnosis

Differential diagnosis was broad for valvular stenosis vs regurgitation. A septal defect was highest on the differential, because it is more common in patients with trisomy 13.

Management

The patient was referred for outpatient electrocardiogram, which revealed sinus tachycardia. Transthoracic echocardiography (TTE) showed a ruptured sinus of Valsalva aneurysm (SOVA) prolapsing into the right atrium, which was severely dilated, with mild to moderate tricuspid regurgitation. A restrictive left-to-right shunt (QpQs of 1.44-1), peak systolic gradient of 124 mm Hg (5.6 m/s), and diastolic gradient of 54 mm Hg (3.7 m/s) were noted.

Citation: Barforoshi S, Sheker C, Ullal AV, Manubolu V, Budoff MJ, Roy SK. Ruptured sinus of Valsalva aneurysm diagnosed on coronary computed tomography angiography in a patient with trisomy 13 syndrome. *Tex Heart Inst J*. 2023;50(3):e228022. doi:10.14503/THIJ-22-8022
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Shortly thereafter, he was admitted to inpatient care and underwent urgent coronary computed tomography angiography (CCTA) preoperatively to confirm the diagnosis and to make more precise preoperative measurements. Coronary computed tomography angiography showed a SOVA arising from the noncoronary cusp and measuring 2.4 cm in width, with a connection between the sinus and the right atrium, indicating a rupture (Fig. 1A, Fig. 1B). A patent foramen ovale was noted, and coronary arteries were unremarkable. Before cardiac surgery, poor oral hygiene prompted full mouth extraction to remove any potential odontogenic sources of infection. Under general anesthesia before surgery, transesophageal echocardiography (TEE) was performed and revealed a ruptured SOVA with a windsock deformity, consistent with the TTE and CCTA findings. The patient's ruptured SOVA was repaired using a XenoSure Biologic Patch (LeMaitre Vascular, Inc), and the patent foramen ovale was closed.

Outcome

Of note, although the blood cultures were negative throughout hospitalization, the aneurysmal aortic tissue culture was positive for *Streptococcus viridians*, and the patient was treated with a 4-week course of intravenous ceftriaxone. Postoperatively, he developed complete heart block that required a permanent dual-chamber pacemaker implant. Postsurgical CCTA (Fig. 2) and repeat TEE (Fig. 3) showed good repair with no evidence of a leak or aortic valve abscess.

Key Points

- Clinicians should recognize the increased risk for congenital heart defects in patients with trisomy 13 and perform early cardiac screening and lifelong follow-up.
- It is important to include sinus of Valsalva aneurysm on the clinical differential, as it can present with a wide variety of clinical manifestations from congestive heart failure and chest pain to, more rarely, a new heart murmur.
- Coronary computed tomography angiography should be considered for use as a first-line, noninvasive imaging modality for sinus of Valsalva aneurysm given its diagnostic accuracy and ability to guide treatment strategy.

Abbreviations and Acronyms

CCTA	coronary computed tomography angiography
SOVA	sinus of Valsalva aneurysm
TEE	transesophageal echocardiography
TTE	transthoracic echocardiography

Follow-up CCTA approximately 2 years later was negative for persistent communication between the sinus of Valsalva and the right atrium.

Discussion

Approximately half of SOVA cases are seen in patients with other congenital cardiac anomalies, most

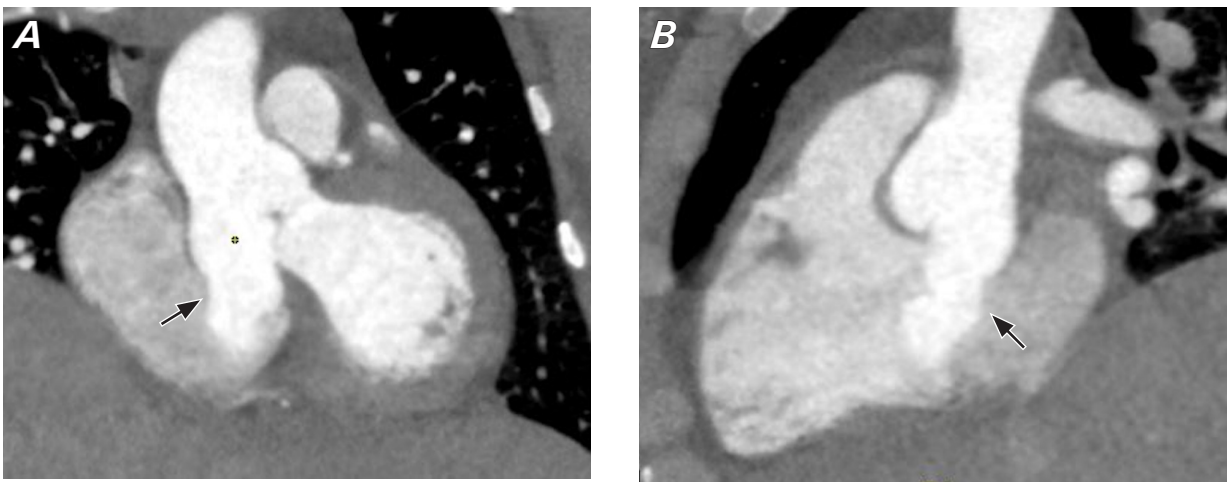

Fig. 1 Coronary computed tomography angiography images before surgical repair, with **A**) coronal and **B**) sagittal views indicating a ruptured sinus of Valsalva aneurysm (arrows) connecting to the right atrium.

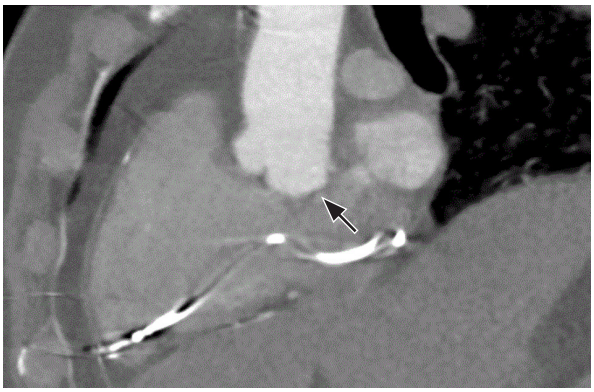

Fig. 2 Coronary computed tomography angiography, sagittal view, postsurgical repair. The prior site of ruptured sinus of Valsalva aneurysm (arrow) after successful repair is noted with no leak.

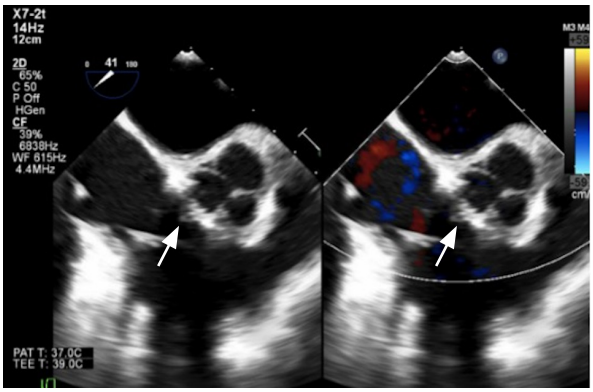

Fig. 3 Transesophageal echocardiogram with Doppler on the right, midesophageal at the level of the aortic valve, shows postsurgical repair of the ruptured noncoronary cusp (arrow) with no leak.

commonly ventricular septal defect (31%) and aortic regurgitation (44%).[1] It has also been reported in genetic syndromes including 22q11.2 deletion (DiGeorge syndrome), Down syndrome, Wildervanck syndrome, and Noonan syndrome. There is 1 known case report on SOVA in a patient with trisomy 13.[2] To the best of the authors' knowledge, this case report is the first of a SOVA rupture secondary to *Streptococcus viridians* endocarditis in a patient with trisomy 13 syndrome.

Ruptured SOVAs vary widely in clinical manifestations, including congestive heart failure, acute chest pain with dyspnea, and cardiac arrest. Here, the patient had a rare presentation of a ruptured SOVA that only presented with a murmur, which has been described in very few cases.[3] Because prognosis after treatment is excellent, with a 15-year survival rate of 90%, prompt and accurate diagnosis is crucial. Treatment for SOVA repair is surgical, involving cardiopulmonary bypass; techniques include aortotomy through the aortic root, through the cardiac chamber the aneurysm ruptured into, or through a dual approach.[1] Our patient developed complete heart block postrepair that necessitated pacemaker placement, which is a very rare complication and only reported about once in a review of 53 cases of operative outcomes.[4]

Although imaging is essential in the diagnosis, surgical planning, and postsurgical follow-up of SOVA, there is no established consensus on selection of imaging modality for workup.[5] Transthoracic echocardiography followed by TEE is traditionally the first line; however, CCTA is being increasingly used.[6] In this case presentation, SOVA was first suspected on TTE, but the diagnosis was confirmed preoperatively with noninvasive CCTA, which importantly spared the patient multiple invasive imaging modalities before surgical management. In addition, CCTA offers high-quality anatomic imaging with multiformat reconstructions and volume-rendered techniques that allow it to serve as a comprehensive approach for preoperative planning. This case highlights the utility of CCTA, a relatively noninvasive imaging modality for initial imaging compared with TEE, in the diagnosis and treatment strategy of SOVA rupture.

Published: 24 May 2023

Author Contributions: Shiva Barforoshi, Chandana Sheker, Ajayram V. Ullal, and Venkat Manubolu contributed to study concept and design, acquisition of data, data analysis and interpretation, and drafting and revision of the manuscript for intellectual content. Matthew J. Budoff and Sion K. Roy contributed to revision of the manuscript for intellectual content.

Conflict of Interest Disclosure: None

Funding/Support: None

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