Ruptured Sinus of Valsalva Aneurysm in a Patient with DiGeorge Syndrome: Expanding the Cardiovascular Phenotype

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Sinus of Valsalva aneurysms (SVA) are uncommon cardiac anomalies. They represent only 0.1–3.5% of congenital heart defects. While rupture of an SVA can lead to acute left-to-right shunting and heart failure, its association with chromosome 22q11.2 deletion (DiGeorge syndrome) has rarely been documented.

Transthoracic echocardiography (TTE) revealed a continuous systolic-diastolic jet suggestive of aortic-to-right-atrial communication. TEE and contrast-enhanced computed tomography (CT) confirmed rupture of a right-coronary-cusp SVA into the right atrium. The patient underwent urgent surgical repair. Initial direct-suture closure was unsuccessful because of persistent flow and was converted to definitive pericardial-patch repair. Postoperative TTE demonstrated complete closure and preserved biventricular function. To the best of our knowledge, this case represents only the third known example of ruptured SVA in a patient with DiGeorge syndrome. It underscores the expanding cardiovascular phenotype of 22q11.2 deletion and highlights the role of multimodality imaging and timely surgical intervention.

SVA is an uncommon cardiovascular anomaly, accounting for approximately 0.09% of the general population. SVAs may present asymptomatically or rupture in up to 35% of cases, often resulting in life-threatening complications requiring urgent surgical intervention [1].

DiGeorge syndrome, velocardiofacial syndrome, or conotruncal anomaly face syndrome is a relatively common genetic disorder, resulting from a microdeletion on chromosome 22 [3]. This syndrome produces a spectrum of congenital anomalies involving the heart, palate, immune system, and facial features [2].

Although numerous structural cardiac abnormalities are well documented in association with 22q11.2 deletion, SVA has not previously been recognized and is only scarcely reported as part of this phenotype [3,4]. We report on a case of ruptured SVA of the right coronary cusp in a patient with DiGeorge syndrome, highlighting a previously under-reported cardiac manifestation of 22q11.2 deletion syndrome.

PATIENT DESCRIPTION

A 32-year-old male with genetically confirmed DiGeorge syndrome without known cardiovascular manifestations presented to the emergency department with palpitations. Given his hemodynamic stability and absence of concerning features, he was discharged for outpatient cardiac evaluation.

Ambulatory workup included a 24-hour electrocardiogram Holter demonstrating rare premature ventricular and atrial contractions and sinus tachycardia. TTE demonstrated a significant left-to-right shunt on color Doppler imaging, although the source remained unclear, potentially originating from either a SVA or ventricular septal defect.

At the Adult Congenital Heart Disease service, a repeat echocardiogram showed a continuous (systolic and diastolic) jet suspected to originate from the SVA into the right atrium [Figure 1]. The patient was admitted for further workup. During admission, he remained hemodynamically stable. Physical examination revealed a continuous systolic and diastolic murmur without stigmata of endocarditis. Workup for endocarditis, including three sets

of blood cultures and serologies for culture-negative endocarditis, returned negative. Rheumatoid factor, complements (C3, C4), antinuclear antibodies, and tests for human immunodeficiency virus and syphilis were all normal.

TEE showed a continuous flow from the right sinus of Valsalva into the right atrium confirming the diagnosis of ruptured right-coronary-cusp SVA [Figure 1]. Cardiac CT demonstrated a continuous jet from the right coronary cusp to the right atrium. Surgery was promptly performed. The initial defect closure via direct suture failed due to persistent flow through the fistula [Figure 1], and a definitive repair was achieved with placement of a surgical patch, yielding good surgical and hemodynamic outcomes.

COMMENT

This case represents a rare but clinically significant manifestation of DiGeorge syndrome. The initial non-specific presentation demanded careful echocardiographic assessment to identify a left-to-right shunt. Notably, our patient had no previously documented cardiac abnormalities, suggesting that SVA may develop later in life or remain undetected during routine pediatric screening, consistent with prior reports [3,4].

SVAs are rare defects stemming from congenital deficiency of elastin and abnormal bulbus cordis development leading to separation between the aortic media and annulus fibrosus [1]. The majority originate near the right (RCS) or non-coronary sinus (NCS) [1]. These lesions typically remain asymptomatic until rupture, which are more common in males (2:1) and can occur across a wide age range with a median age of approximately 45 years [1]. Congenital and genetic etiologies for SVA have been described in several congenital heart defects including tetralogy of Fallot, transposition of the great arteries, hypoplastic left heart syndrome, and bicuspid aortic valve [1], as well as in genetic syndromes with increased susceptibility to aneurysm formation (e.g., Marfan, Loeys-Dietz, Ehlers-Danlos, Turner, Noonan) [2].

Surgical repair remains the standard of care for ruptured SVA, with perioperative mortality < 4% and 10-year survival 87–99% [5]. Patch repair techniques are generally favored over direct suture due to reduced recurrence [5], while percutaneous closure may be considered in selected small, uncomplicated ruptures; long-term data remain limited and complex anatomy is a contraindication [5].

CONCLUSIONS

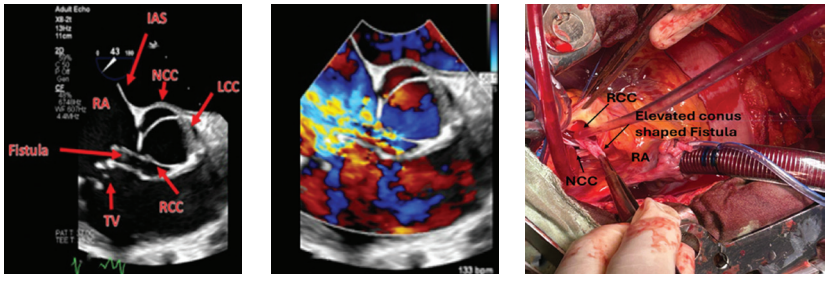
This case expands the cardiovascular phenotype of DiGeorge syndrome to include ruptured SVA. Clinicians should maintain vigilance for new murmurs and consider multimodality imaging. Lifelong surveillance of the aortic root may be warranted in 22q11.2 deletion syndrome.

Figure 1. Transthoracic echocardiography of parasternal short-axis at the aortic valve area

[A] Demonstrating apparent SVA on right coronary cusp level

[B] Color Doppler demonstrating regurgitant flow across SVA demonstrating ruptured SVA

[C] Intra-operative image of the conus shaped fistula



IAS = interatrial septum, LCC = left coronary cusp, NCC = non-coronary cusp, RA = right atrium, RCC = right coronary cusp, SVA = sinus of Valsalva aneurysm, TV = tricuspid valve

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