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Quadricuspid Aortic Valve with Sinus of Valsalva Rupture

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A B S T R A C T

A 22-year-old female with no medical history presented to the emergency room with 2 weeks of rapidly worsening dyspnea on exertion, orthopnea, and cough. On cardiac auscultation, she was noted to have to-and-fro murmurs and a continuous murmur with signs of right heart failure. Echocardiographic images obtained showed moderate to severe aortic regurgitation, severe tricuspid regurgitation, and a "windsock" originating in the right coronary sinus of Valsalva and terminating in the right atrium. The aortic valve had four leaflets, with the right leaflet function compromised by the ruptured sinus, causing aortic regurgitation. The patient underwent resection of the sinus aneurysm and aortic valve replacement with a bioprosthetic valve. Quadricuspid aortic valves are uncommon and are rarely associated with sinus of Valsalva aneurysm. The prevalence in the general population, clinical progression, and prognosis of this unique congenital abnormality are reviewed.

Key Words. Quadricuspid Aortic Valve; Sinus of Valsalva; Congestive Heart Failure

Introduction

Congenital aortic valve abnormalities are most commonly of biscuspid morphology; quadricuspid aortic valves (QAV) are uncommon and are usually associated with other congenital cardiac pathology such as truncus arteriosus. Noninvasive evaluation of a young female with recent abrupt onset of biventricular failure revealed the concurrence of a quadricuspid aortic valve and ruptured sinus of Valsalva aneurysm.

Case Presentation

A 22-year-old Guatemalan female with no medical history presented to the emergency room with 2 weeks of gradual worsening dyspnea on exertion, orthopnea, and cough. She denied any fevers, nausea, weight loss, rashes, or recent illnesses. She denied a history of rheumatic fever, decreased exercise tolerance as a child compared with her peers, or any previous knowledge of a cardiac murmur.

On physical examination, the patient was afebrile, with a pulse of 110 beats per minute, a blood pressure of 108/47, respiratory rate of 20, and oxygen saturation of 100% on room air. She was alert and oriented and in no acute distress. Her jugular venous pressure was elevated at 20 cm H_2O with bounding carotid pulses and systolic bruits over both carotids. Her lungs were clear to auscultation with no rales, wheezes, or rhonchi. Cardiac examination revealed a right ventricular heave, with a single first heart sound followed by an ejection sound initiating a continuous murmur heard best at the lower left sternal edge (Movie S1). Mid systolic and early diastolic murmurs could be heard at the base (Movie S2). The extremities demonstrated no cyanosis, clubbing, or stigmata of endocarditis. There was +2 pitting pedal edema bilaterally.

An electrocardiogram showed sinus tachycardia, normal axis, and nonspecific T wave abnormalities in the anterior leads. The frontal plane axis was normal and there was no evidence of ventricular hypertrophy or atrial abnormalities. Frontal and lateral chest radiograph showed an enlarged cardiac silhouette with prominent pulmonary vasculature but no evidence of pulmonary edema or pleural effusion.

Serum electrolytes, complete blood count, and serial cardiac biomarkers were within normal limits. Her BNP level was elevated at 571 pg/mL. A urine pregnancy test was negative. Blood cultures that were drawn on admission grew no organisms.
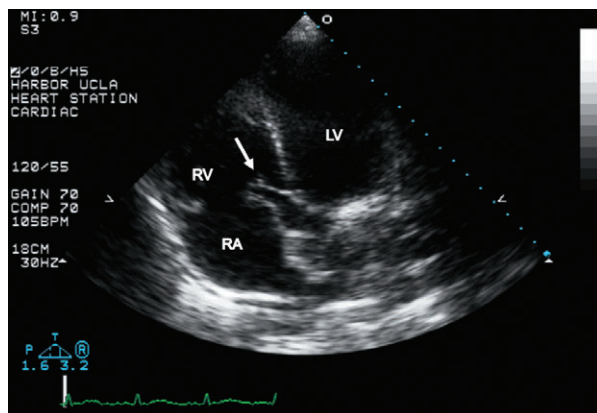
Figure 1. 2D transthoracic echocardiogram apical five-chamber view showing a tubular defect adjacent to the septal triscupid leaflet (arrow), representing the ruptured sinus of Valsalva. RA, right atrium; RV, right ventricle; LV, left ventricle.

A transthoracic echocardiogram (TTE) revealed a mildly dilated left ventricle, with an estimated ejection fraction of 45–50% and no regional wall motion abnormalities. The right ventricular systolic function was moderately reduced, and the right atrium was dilated. The severity of the triscupid regurgitant jet was difficult to assess because of its overlap with the windsock Doppler jet. The right ventricular systolic pressure (RVSP) was estimated at 46 mm Hg. A mobile tubular "windsock" structure emanating from the aortic root could be seen adjacent to the septal leaflet of the tricuspid valve (Figure 1, Movie S3) through which continuous wave Doppler demonstrated uninterrupted flow throughout systole (increasing to a peak gradient 114 mm Hg) and diastole (declining to a gradient of 37 mm Hg) (Figure 2). Although it could not determined with certainty from the transthoracic echocardiographic images whether the windsock entered the right atrium or ventricle, the peak systolic gradient through the fistula was comparable with the magnitude of systemic arterial pressure indicating a low pressure recipient chamber, namely the right atrium (Figure 3).

Transesophageal echocardiography (TEE) confirmed that the windsock was 18 mm in length and 7 mm in diameter, originated in the right sinus of Valsalva of a quadricuspid aortic valve and terminated in the right atrium (Figures 3–5, Movie S4). Severe aortic regurgitation was noted (Movie S5), and no ventricular septal defect (VSD) was seen.

At operation, the aortic valve was confirmed to have four leaflets with associated sinuses,
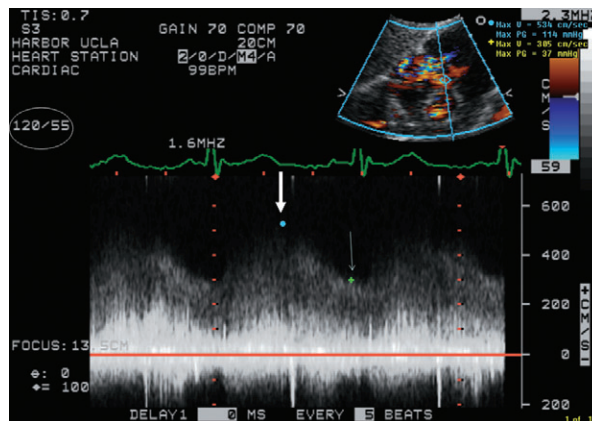

Figure 2. Continuous wave Doppler across the defect, showing uninterrupted flow through systole and diastole, with a peak systolic gradient of 114 mm Hg (thick arrow), and a diastolic gradient of 37 mm Hg (thin arrow), similar to systemic blood pressures (circle), indicating the origin of the defect was the aortic root.
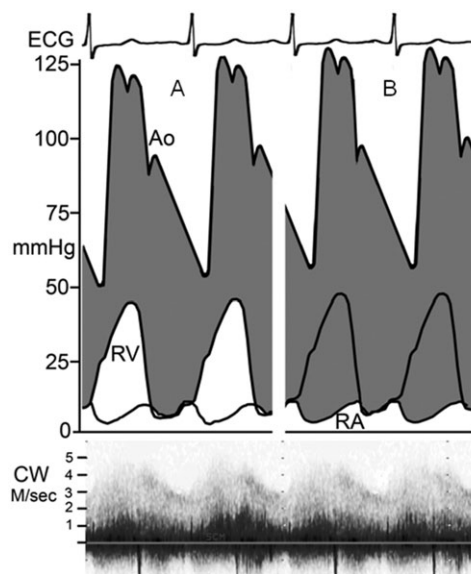
Figure 3. Diagrammatic depiction of estimated intracardiac pressures (top) are aligned with continuous wave Doppler velocity through windsock deformity (bottom). Because the peak systolic gradient was similar to the magnitude of systolic blood pressure, the velocities confirmed that the terminus of the windsock was located in a low pressure chamber such as the right atrium (B). Given the right ventricular systolic pressure was 46 mm Hg, if the windsock entered the right ventricle, the peak systolic gradient would be lower given the smaller pressure difference between the aorta and right ventricle (A). Ao, aortic pressure; RV, right ventricular pressure; RA, right atrial pressure.
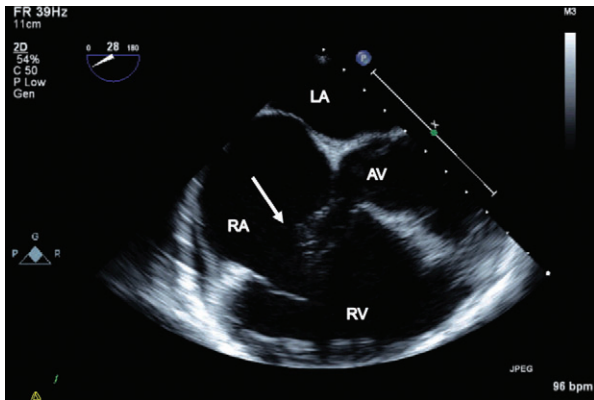
Figure 4. Transesophageal echocardiogram, 28 degree view of the short axis through the aortic valve showing the windsock deformity emanating from the sinus of Valsalva (arrow). RA, right atrium; RV, right ventricle; LA, left atrium; AV, aortic valve.
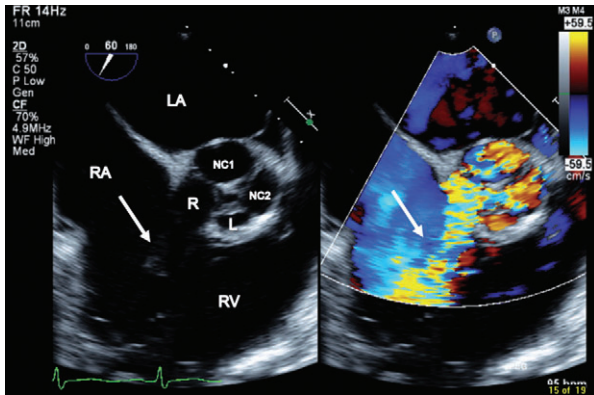
Figure 5. Transesophageal echocardiogram, 60 degree view of the short axis aortic valve showing simultaneous color Doppler flow during diastole and the windsock (arrow). The closed aortic valve has a "X" appearance, and the Doppler flow shows diastolic flow both through the ruptured sinus of Valsalva and through the aortic valve indicating aortic regurgitation. R, right coronary cusp; L, left coronary cusp; NC1, noncoronary cusp #1; NC2, noncoronary cusp #2; RA, right atrium; LA, left atrium; RV, right ventricle.

with a small right coronary leaflet, fusion of the right and left coronary leaflets, and two distinct noncoronary sinuses. The proximal windsock orifice was immediately below the right coronary artery ostium, and the fistulous structure could be completely everted and pulled into the aortic root.

The windsock was excised (Figure 6) and the proximal orifice was closed with a pericardial patch with 4–0 Prolene sutures. The aortic valve was replaced with a 23 mm Epic bioprosthetic valve (St. Jude Medical, St. Paul, MN, USA). The

Figure 6. Surgically excised "windsock."

patient did well postoperatively and was discharged in good condition.

Discussion

Quadricuspid aortic valve is a rare congenital abnormality, whose incidence has varied in different studies. Simonds[1] in a series of 6252 autopsies did not identify any cases of QAV, but in a pooled series of 25 666 autopsies, he found two cases for an incidence of 0.008%. Feldman,[2] in a more recent survey with the advent of 2D echocardiography found an echocardiographic incidence of 0.043% in 13 805 echocardiograms.

The hollow region enclosed by the three aortic cusps between the aortic valve annulus and the sinotubular ridge is known as the sinus of Valsalva. Multiple etiologies for aneurysmal dilatation and rupture of the sinus of Valsalva have been cited, including congenital causes, endocarditis, cystic medial necrosis, atherosclerosis, trauma, and syphilis. A congenital sinus of Valsalva aneurysm, which is usually associated with dilation of a single sinus, has been classified into four types by Sakakibara and Konno.[3] A Type I aneurysm develops in the left part of the right coronary sinus and projects into the outflow tract of the right ventricle. Type II develops in the central part of the right coronary sinus and protrudes into the right ventricle, penetrating the crista supraventricularis. Type III originates in the posterior part of the right coronary sinus, and can project into either the right ventricle or atrium. Type IV develops from the right part of the noncoronary sinus and projects into the right atrium near the septal leaflet of the tricuspid valve. Congenital sinus of Valsalva aneurysms most commonly originate from the right sinus

(65–85%), followed by the noncoronary sinus (10–30%), and the left sinus (<5%).[4]

The embryology of quadricuspid aortic valves remains unknown. Proposed mechanisms include anomalous septation of the conotruncus, excavation of one of the valve cushions, aberrant fusion of the aorticopulmonary septum, or septation of a normal valve cushion as a result of inflammation.[5–7] Animal studies have suggested that QAVs may result from the division of one of the three mesenchymal ridges that give rise to three aortic valve cushions.[8,9] In relation to congenital defects of the sinus of Valsalva, there is also speculation that a single developmental abnormality may result in various congenital defects in the aortic root, given that development of aortic valve leaflets occurs after development from coronary artery origins of the sinus of Valsalva.

Before the widespread use of 2D echocardiography, most cases of QAV were discovered on autopsy or intraoperatively at aortic valve replacement. On the short axis view of the aortic valve in diastole, the commissural lines formed by the adjacent cusps result in a "X" configuration rather than the "Y" formation seen in a normal tricuspid aortic valve. However, echocardiographic findings may not always correlate with surgical findings, and it may not be possible to visualize the aortic leaflets adequately with TTE. In a study by Timperly and colleagues, 50 of 114 cases previously reported in the literature were diagnosed by TTE or TEE[6], implying that the incidence is likely underestimated in the general population and will likely increase as more studies are performed and image quality continues to improve.

Hurwitz,[5] in 1973, described seven common anatomic variants of quadricuspid semilunar valves in a review of 197 previously reported cases. Timperley[6] did a review of 114 QAV cases previously reported in the literature up until 2002. In 97 cases in which the size of the leaflets were described, over 85% of the reports were type A (valves with four equal leaflets), type B (three equal cusps with one smaller cusp), or type C (two equal larger and two equal smaller cusps) based on the Hurwitz classification.

Previously, it was believed that valves with four equal leaflets, or type A, were more likely to function normally. Given that valvular regurgitation usually develops because of fibrous thickening with incomplete coaptation, it was though that unevenly sized leaflets would be more predisposed to develop regurgitation. However, in Timperley's review of the 30 type A QAVs reported, 80% had

significant regurgitation and 40% required valve replacement. In the type B group, there was a higher proportion of normally functioning valves (32%). Out of the 114 cases reported, 46 required aortic valve replacement at the average age of 54 years.

QAVs have been reported in association with other cardiac abnormalities. Egred[10] reported the discovery of a sinus of Valsalva fistula and QAV on magnetic resonance imaging, with the aortic regurgitant jet entering the right ventricle. Pitta[11] reported a QAV with severe aortic regurgitation with unruptured sinus of Valsalva aneurysm. Unger[12] reported echocardiographic findings of a QAV with a ruptured sinus of Valsalva aneurysm, and a VSD.

QAVs are often functionally normal, and management depends on the development of regurgitation, which usually does not occur until adulthood. If a QAV is found on echocardiography, follow-up assessment is needed to monitor for progression to severe regurgitation, regardless of morphology as seen in the number of Type A QAVs that required aortic valve replacement. Endocarditis prophylaxis is recommended as there are three confirmed cases of endocarditis affecting a QAV[6].

Besides aortic valve replacement, other surgical options may include suture valvuloplasty or annulovalvuloplasty, both of which have been used in valve repair for truncus arteriosus patients with truncal quadricuspid valves.[13] The suture valvuloplasty technique consists of suturing the edges of a prolapsing leaflet to either one or both adjacent leaflets, creating a functional tricuspid or bicuspid semilunar valve. The annulovalvuloplasty method involves excising the prolapsed leaflet, and then using pledgetted sutures to tighten the annulus, therefore remodeling the truncal valve into a smaller and more competent valve. If the prolapsed leaflet involves the coronary sinus of Valsalva, the involved coronary artery can be reimplanted into the neighboring sinus of Valsalva prior to leaflet resection and tightening of the annulus.

In conclusion, we present a patient who presented with a ruptured sinus of Valsalva in association with a quadricuspid aortic valve with aortic regurgitation. Using bedside examination and noninvasive imaging, an accurate diagnosis of a complex lesion was established and confirmed in the operating room without the need for invasive diagnostic testing. The very recent onset of symptoms and the near-normal electrocardiogram

suggested that the lesions were functionally well tolerated until the sinus of Valsalva aneurysm ruptured causing an abrupt onset of symptoms of increasing severity.

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Conflict of interest: None.

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Supporting Information

Additional Supporting Information may be found in the online version of this article:

Movie S1. Cardiac auscultation of the patient along the left third intercostal space sternal border revealing an ejection sound which initiates a continuous murmur caused by flow through the ruptured sinus of Valsalva aneurysm. Both aortic systolic and diastolic pressures are higher than right pressures; thus, the murmur persists through both phases. In addition, a right ventricular heave, bounding carotid pulsations and elevated jugular venous pressure are seen.

Movie S2. Cardiac auscultation of the patient along the right third intercostal space sternal border revealing both a systolic and diastolic murmur, consistent with a to-and-fro murmur from aortic regurgitation, with a superimposed continuous murmur. Bounding carotid pulsations are also present.

Movie S3. Cinematic view of Figure 1.

Movie S4. Cinematic view of Figure 5 showing continuous flow in both systole and diastole through the ruptured sinus of Valsalva with the windsock deformity ending in the right atrium.

Movie S5. Transesophageal echocardiogram, 129 degree view long-axis view showing the left ventricular outflow tract showing moderate to severe aortic regurgitation.

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