Unicuspid Aortic Valve Presenting with Decompensated Critical Aortic Stenosis

Eunice Yang, MD, Madeline Schiminger, BS, RDCS (AE PE), and Jose A. Madrazo, MD,
Baltimore, Maryland

INTRODUCTION

Aortic stenosis is the most common valvular lesion in the United States, carrying an overall prevalence of 2.8% in adults >75 years of age. Congenital aortic valvular anomalies lead to accelerated progression toward aortic stenosis and therefore should be suspected when presenting in younger individuals. In this case we describe the first clinical presentation of decompensated critical aortic stenosis in a 46-year-old man with unicommisural unicuspid aortic valve (UAV). Echocardiography is indispensable in the evaluation of aortic stenosis, but echocardiographic diagnosis of UAVs is challenging, and these anomalies are often misdiagnosed as bicuspid aortic valves (BAVs). Patients with UAV require earlier and more extensive surgical intervention than their counterparts with BAVs. Thus, understanding clinical findings that prompt suspicion for UAV is necessary to trigger a more thorough evaluation of the aortic valvular complex and ensure timely clinical follow-up and intervention.

CASE PRESENTATION

A 46-year-old man with no known medical history reported a long-standing history of mild exertional dyspnea with concurrent chest pain that began when he was only 14 years old. Despite these long-standing symptoms, he reported no significant functional impairment until several months before his hospitalization, when he developed progressive fatigue and exercise intolerance. He found it increasingly difficult to keep up with his coworkers as a concrete mixer and stated that during the week before his admission, he was able to work for only 5 min before stopping to rest. The patient was able to climb one flight of stairs very slowly and could walk approximately two blocks on horizontal ground before stopping to rest. His New York Heart Association class III symptoms were accompanied by nonradiating dull chest pain. He had no known family history of premature coronary artery disease or sudden cardiac death and did not have routine medical care preceding his initial presentation to a local cardiology clinic.

Upon arrival to the clinic, the patient was hemodynamically stable, and his clinic notes described a grade 4/6 systolic ejection murmur heard at the right upper sternal border. Electrocardiography revealed normal sinus rhythm and severe left ventricular hypertrophy. He underwent cardiac positron emission tomography/computed tomography that showed nonobstructive coronary artery disease. The patient's symptoms continued to worsen, and his cardiologist sent him to a local emergency department before his outpatient echocardiographic examination could be completed. Upon arrival, the emergency department providers were concerned for an ST-segment elevation myocardial infarction given his J-point elevations from left ventricular hypertrophy and strain (Figure 1).

He was emergently taken to coronary angiography, which revealed nonobstructive coronary lesions, an anomalous circumflex artery originating from the right coronary, and a mean aortic transvalvular gradient >100 mm Hg. An intra-aortic balloon pump was placed, and the patient was transferred to the coronary care unit for further management.

On arrival at the coronary care unit, transthoracic echocardiography showed severe concentric left ventricular hypertrophy with hyperdynamic systolic function. The aortic valve appeared heavily sclerotic, with minimal leaflet excursion and a small opening on the anterior side of the aortic valve raising concerning for unicuspid unicommisural aortic valve (Figure 2A–2D, Videos 1-4). One of the most important defining features of UAV is the extension of the heavy calcification into the left ventricular outflow tract (Figure 2A and 2C, Videos 1 and 3).

There was minimal eccentric aortic regurgitation (Figure 2B and 2D, Videos 2 and 4). The visualized portions of the aorta appeared normal in caliber. The calculated aortic valve area by continuity equation was 0.3 cm^2. The maximal velocity through the aortic valve was 6.4 m/sec, with significantly elevated peak and mean pressure gradients of 164 and 127 mm Hg (Figure 3). There was no evidence of subvalvular narrowing, supravalvular narrowing, or coarctation, findings associated with congenitally stenotic aortic valves.

In the absence of significant comorbidities and because of clinical instability, the decision was made to proceed directly to surgical valve replacement to address clinical decompensation without additional imaging. Intraoperative transesophageal echocardiography confirmed unicommisural UAV (Figure 4, Videos 5-8).

The patient's family expressed strong concerns regarding long-term anticoagulation, as the patient had indicated a strong preference not to be on a medication that might place him at increased risk for bleeding. After weighing this strong patient preference as well as concerns for compliance with anticoagulation, the cardiac surgery team decided to remove the native valve and replace it with an On-X bioprosthetic valve (CryoLife, Kennesaw, GA), with the understanding that the patient would likely require a repeat valve procedure within his lifetime. Direct visualization of his explanted valve (Figure 5) confirmed a unicuspid unicommisural valve.

The patient also underwent primary repair of an anomalous left circumflex coronary artery and aortic root replacement after intraoperative transesophageal echocardiography revealed a mildly distorted aortic root with effacement of the sinotubular junction. His postoperative course was complicated by major bleeding in the setting of receiving a ticagrelor load during emergent angiography at the outside emergency department. He tolerated resuscitation and was ultimately discharged home with significant improvement in exercise tolerance. He recently followed up with his cardiac surgeon in an outpatient clinic, where he reported resolution of exertional dyspnea and chest pain.

From the Division of Cardiology, Johns Hopkins Hospital, Baltimore, Maryland.

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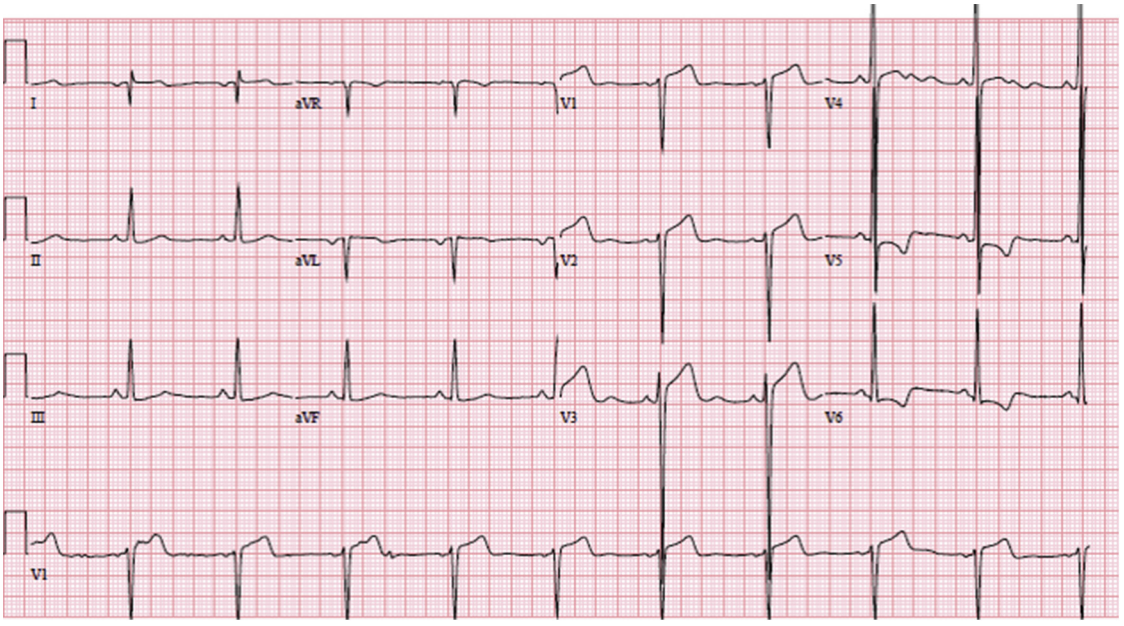

Figure 1 Electrocardiogram on admission to outside hospital showing normal sinus rhythm with J-point elevation and ST-segment changes concerning for left ventricular strain and left ventricular hypertrophy.

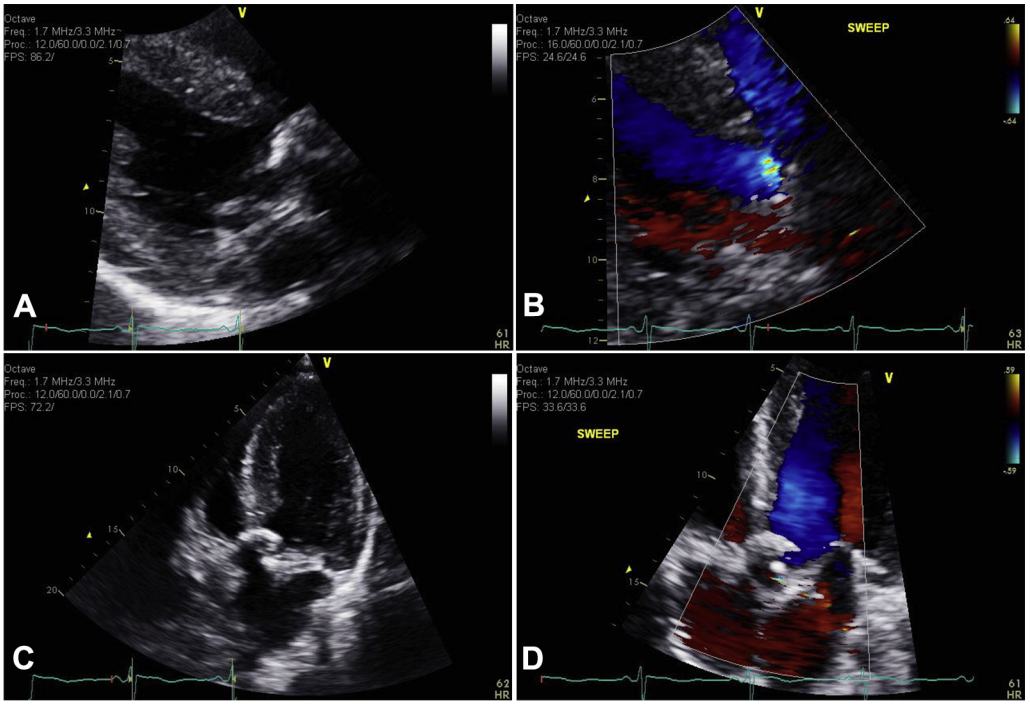

Figure 2 **(A)** Close-up image from transthoracic echocardiographic parasternal long-axis view showing heavily calcified, minimally mobile aortic valve. **(B)** Color flow Doppler parasternal long-axis view showing eccentric jet of aortic insufficiency (AI). **(C)** Transthoracic five-chamber apical view, demonstrating extension of aortic valve calcification from fused cusps into the left ventricular outflow tract. **(D)** Five-chamber apical view with color Doppler showing AI with main eccentric jet hugging the septum. See Videos 5-8.

DISCUSSION

Although BAV anomalies have been well described for hundreds of years, the first documentation of UAV was in the late 1950's.[1] UAVs are extremely rare, with an estimated prevalence of 0.02% in the general population.[2] Adults with UAVs experience accelerated aortic stenosis that outpaces their counterparts with BAVs, necessitating surgical intervention between their third and fifth decades of life. Although the overall prevalence of UAVs is low, patients with UAVs account for approximately 4% to 6% of individuals undergoing

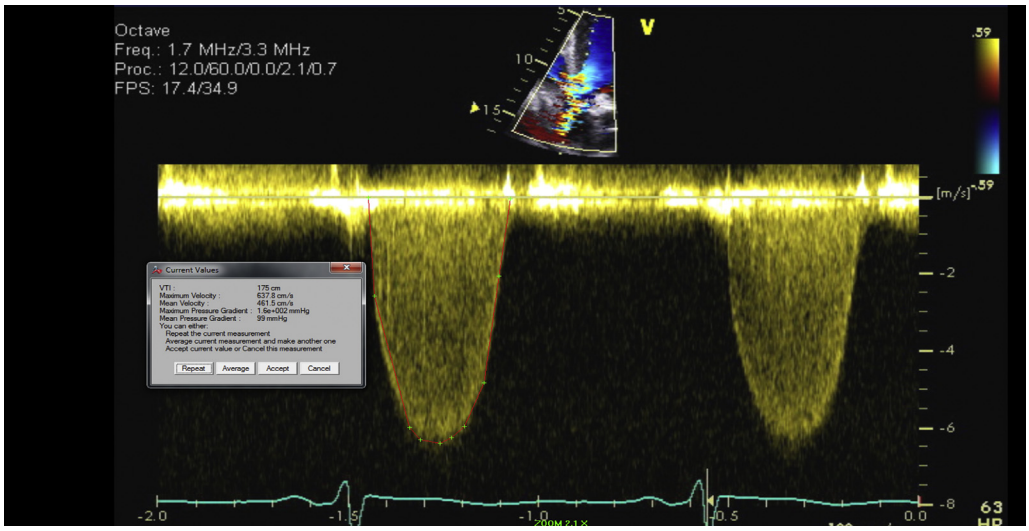

Figure 3 Gradient across aortic valve showing severely elevated maximal velocity of 6.4 m/sec.

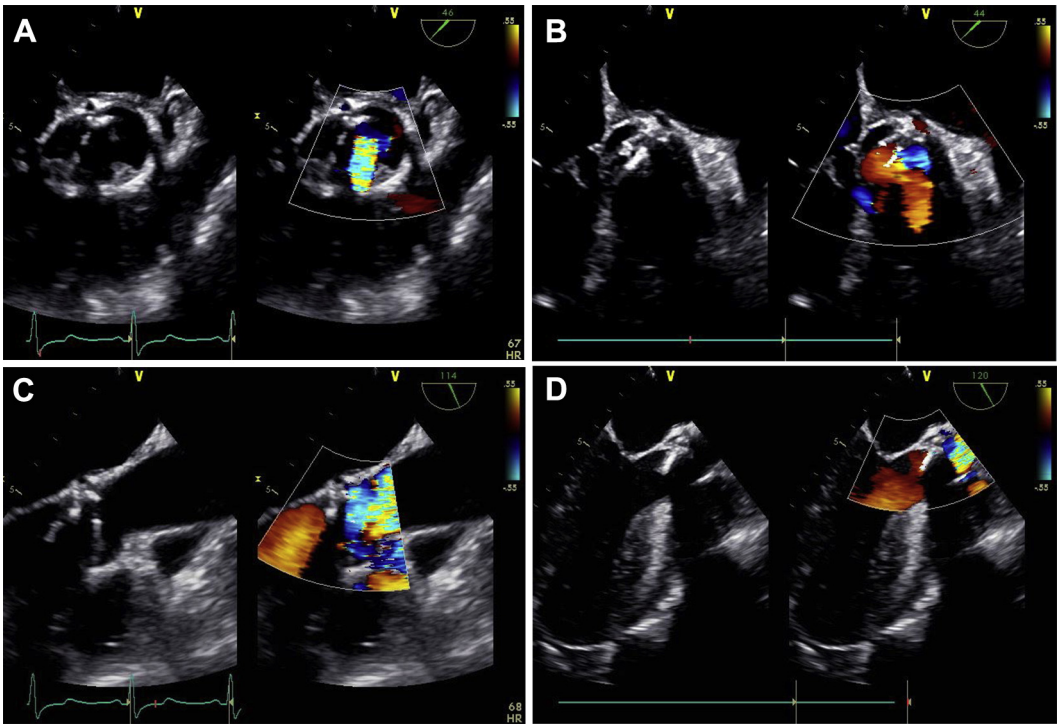

Figure 4 **(A)** Transesophageal echocardiographic (TEE) midesophageal aortic valve (AV) short-axis view showing en face view of heavily calcified, severely stenotic unicommisural UAV with and without color Doppler. **(B)** TEE midesophageal AV short-axis view showing a different angulation of the open commissure with a single attachment point. **(C)** TEE midesophageal long-axis view showing heavily calcified aortic root with a classic calcification pattern into the left ventricular outflow tract (LVOT), as well as an eccentric aortic regurgitation jet. **(D)** Long-axis midesophageal view taken at 120° also displaying unusual pattern of extended calcification into the LVOT. See Videos 5-8.

isolated aortic valve surgery, suggesting that most individuals with this condition will ultimately require surgical intervention.[3]

UAV and BAV anomalies arise through incomplete separation of valve tissue during the first trimester of fetal development, forming commissures at appropriately matured zones of apposition between valve leaflets and raphe at the fused zones of apposition.[4] UAVs most often present with a commissural opening extending from the peripheral zone of apposition to the centroid of the valve. The

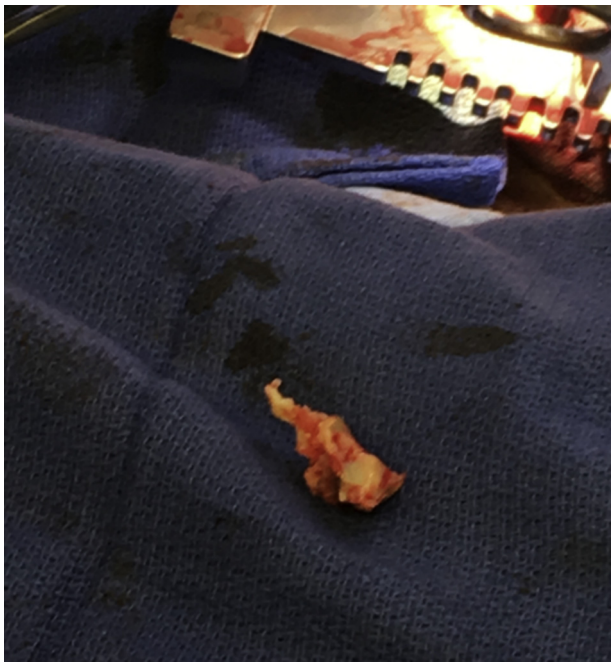

Figure 5 Direct visualization of explanted UAV.

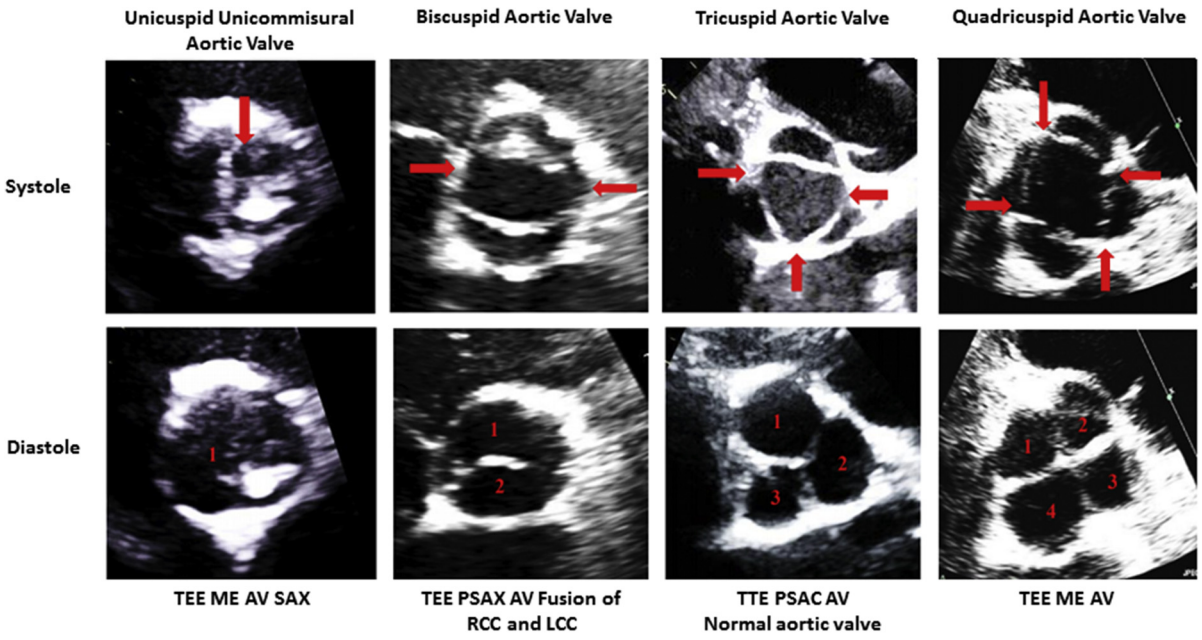

Figure 6 The number of points of commissural contact with the aortic root is a helpful way to identify the number of aortic valve cusps present. *Red arrows* indicate commissural attachment sites, with cusps numbered in the diastolic views. *LCC*, Left coronary cusp; *RCC*, right coronary cusp; *TTE PSAX AV*, transthoracic parasternal short-axis view focusing on the aortic valve.

preserved commissure typically lies between the noncoronary and left coronary cusps and arises from the persistence of the interleaflet triangle abutting the aortomitral curtain. This leads to a posteriorly directed slitlike opening pointing toward the aortic valve.[4,5] This formation of unicommisural UAV is described as the type 2 Sievers anomaly and is the most commonly encountered UAV in adults.[6] More rarely, UAVs can also be acommissural, unassociated with any preserved zones of apposition. This type of UAV has a pinhole-like orifice and is associated with neonatal hemodynamic compromise requiring urgent intervention.[7]

Unicommisural UAV cases are also notoriously difficult to diagnose using transthoracic echocardiography, with rates of determination of

Table 1 Summary of transthoracic echocardiographic findings that may help distinguish between UAV and BAV

UAV	BAV
• Heavily calcified aortic valve in a young patient • Eccentric coaptation closure line in long axis • Eccentric limited valve orifice systolic opening • Systolic cusp "doming"	
• One point of commissural contact with the aortic root • Low cusp height • Calcified posterior cusp prolapse into the LVOT • Two regurgitant jets: one eccentric through the middle of the valve, one at the level of the left and noncoronary cusps. Of note, visualization of two regurgitant jets is not always possible given limitations of echocardiography, calcification, and image quality.	• "Football" shape seen in systole in short axis • Two points of commissural contact with the aortic root

LVOT, Left ventricular outflow tract; *BAV*, bicuspid aortic valve; *UAV*, unicuspid aortic valve.

UAV of about 14% to 25%, increasing to 69% to 75% (with specificity of 86%) only when using intraoperative transesophageal echocardiography.[8,9] The valve is most commonly mistaken for a BAV, as heavy calcification and two-dimensional imaging of the three-dimensional aortic root limit the ability to distinguish between raphe and true zones of apposition in the short-axis view during diastole.[8] Visualization of the points of contact of the commissures with the aortic root on short-axis views in systole is a useful way to identify the number of aortic valve cusps present (Figure 6).

These heavy calcifications pose similar limitations in diagnosis of UAV using alternative imaging modalities, such as computed tomography, which is further hindered by relatively low temporal resolution.[10] Interestingly, heavy calcifications may fool the eye even on direct visualization of the valve; in a small study involving surgically excised stenotic aortic valves, valve structure analysis by surgeons during surgery was congruous with cardiac pathologist analysis in only 59% of cases.[11] Three-dimensional transesophageal echocardiography enhances accuracy of UAV diagnosis by providing real-time volume-rendering images and should be performed when UAV is suspected.[12] Several clinical and imaging findings suggestive of UAV diagnosis should prompt focused evaluation of the aortic valve complex (Table 1).

Because most adults with UAVs will require surgery by middle age, the accurate diagnosis of this valvular anomaly is crucial in planning the timing of intervention. Several clinical and imaging findings should raise suspicion for UAV and prompt a more thorough evaluation of the aortic valve. A high index of suspicion should be maintained for young patients presenting with severe mixed aortic valve dysfunction, aortic annular dilation with normal sinus of Valsalva, eccentric valvular orifice during systole (systolic doming), eccentric coaptation line of the cusp in the long-axis view, low cusp height, and calcified posterior cusp prolapse into the left ventricular outflow tract. On Doppler imaging sequences, the presence of two regurgitant jets, one eccentric through the middle of the valve and another at the level of the left and noncoronary cusps, should also trigger further evaluation.

Management of UAVs hinges on the assumption that the natural disease progression, once severe, is similar to BAVs. BAV anomalies and associated aortopathy are well recognized, with evidence of its natural history, associated risk for dissection, and outcomes of various surgical techniques. Similar to their bicuspid counterparts, UAVs are also associated with aortopathies and typically present with aortic root or ascending aortic dilatation, thought to arise from genetic contribution as well as abnormal hemodynamic shear stress throughout the cardiac cycle from mixed valvular dysfunction.[2,13] However, the data on this association are limited by small cross-sectional studies in a disease with a clearly mutable phenotype over time. To this point, although certain studies report ascending aortic dilation >4.5 cm in more than half of the study cohort,[8] other studies report no pattern of aortic dilation associated with UAVs, despite noting the presence of more extensive aortopathy.[6] To address this discrepancy, a prospective research trial was designed to identify associated comorbidities in patients with UAVs. The most common associations were dilation of the aortic annulus (71% of total cases) and less dilatation in the ascending aorta than their counterparts with BAVs.[9] In addition to aortopathy, other associated disorders include aortic coarctation, an aberrant right subclavian artery, a single coronary artery or anomalous coronary anatomy, and ventricular septal defects.[10]

Preoperative determination of UAV phenotype is important for appropriate surgical planning of planned repairs. UAV repair typically requires reconstruction plasty, whereas BAV repair is most often corrected with a resuspension plasty. Survival appears significantly improved when valve operations are performed with concurrent aortic root repair.[14] Indeed, this small single-center prospective study suggests that annular reduction and stabilization with extra-aortic ring annuloplasty should be considered at the time of UAV repair to avoid late dilation and recurrent valvular failure. Finally, small studies have suggested that intraoperative transesophageal echocardiographic measurement of the percentage difference between the longest and shortest coaptation lengths may be predictive of risk for early redo surgery.[15]

CONCLUSION

Unicommisural UAV is a challenging echocardiographic diagnosis. Nevertheless, echocardiography plays a critical role in the identification, characterization, and management of the aortic valve and remains a cornerstone in the diagnosis of this rare anomaly.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at 10.1016/j.case.2018.04.002.

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