The Lone Cusp: A Patient With a Regurgitant Unicuspid Valve With Aortic Aneurysm

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INTRODUCTION

Unicuspid aortic valve (UAV) is a rare congenital abnormality that affects only 0.02% of the adult population.[1] Its natural history is poorly understood, and it is commonly misdiagnosed as a bicuspid valve.[2] Aortic stenosis (AS) with or without concomitant aortic regurgitation (AR) is usually described in these cases.[3] Here we present a case of UAV complicated by an aneurysmally dilated aorta in a healthy 23-year-old patient with isolated AR.

CASE PRESENTATION

A 23-year-old patient with a known history of bicuspid aortic valve (BAV) presented to the clinic for a precollege wellness examination reporting no medical symptoms. The review of systems was negative. The patient had a history of BAV known since infancy, without any recent follow-up for years, and no family history of valvulopathy or aortopathy. The physical examination revealed a systolic and diastolic murmur loudest in the aortic area. The heart rate was normal, the rhythm was regular, and pulses were symmetrical. The rest of the physical examination, including vital signs, was otherwise unremarkable.

The patient was referred for transthoracic echocardiography (TTE) to characterize the murmur, and it was significant for a BAV with aortic root dilatation causing eccentric moderate AR (pressure half-time 386; Videos 1 and 2) and an ascending thoracic aortic aneurysm of 5.6 cm. The left ventricle had normal size and function (ejection fraction 61%, left ventricular internal diameter in diastole 6.0 cm; Video 3). Urgent computed tomographic angiography of the chest was ordered and confirmed the presence of a dilated aortic root measuring 4.5 cm and an aneurysm of the ascending thoracic aorta measuring 6.1 cm (Figure 1, Video 4).

Given the size of the aneurysm, the patient was referred to a cardiothoracic surgeon and scheduled for a valve repair. Preoperative cardiovascular magnetic resonance (CMR) was ordered to further characterize the valvular anatomy and revealed that the valve was in fact a unicommissural UAV (Figure 2, Videos 5 and 6).

The UAV was confirmed on intraoperative transesophageal echocardiography (TEE; Figure 3, Videos 7 and 8). The patient underwent a Ross procedure using a full root approach, using the patient's own pulmonic valve as an aortic autograft, and a decellularized pulmonary homograft. In addition, the patient underwent external annuloplasty of the valve root using a Dacron ring and replacement of the ascending aorta and proximal transverse arch using a Dacron graft. An image of the surgical specimen is provided, showing the patient's UAV with one functional commissure and two raphes (Figure 4).

Immediate postoperative TEE showed normal leaflet motion of the autograft with only trivial AR, a mean pressure gradient of 3 mm Hg, and normal leaflet motion of the pulmonic homograft (Video 9). The remaining distal arch and descending aorta were also inspected for dissection after decannulation, which was absent. Magnetic resonance angiography of the chest was obtained on postoperative day 4 and showed no evidence of pseudoaneurysm or leak. There were no postoperative complications, and the patient was discharged home on postoperative day 5.

The patient was seen 4 weeks after the procedure and was doing well. A genetics referral was made, and first-degree relative screening with echocardiography was initiated. The patient is currently doing well months after surgery, remains asymptomatic, and is returning to college.

DISCUSSION

The abnormality of the UAV was first described by Edwards[4] in 1958. Since then, many cases have been reported, but the natural history remains unclear. In their longitudinal study of almost 12 years, Novaro *et al.*[1] were the first to report that the incidence of UAV in the general adult population was 0.02%. It was then shown to be the cause of isolated AS in 4.9% of adults >20 years of age undergoing surgery.[5]

UAVs were also classified by Roberts and Ko[5] into unicommissural and acommissural UAVs, the first being more common in adults and the latter in children. The discrepancy is thought to be caused by the wider aperture of the unicommissural UAV, providing more latency before stenosis and calcification of the valve. The acommissural UAV, on the other hand, lends itself to early clinical presentation because of the severely reduced area of the aperture it causes at a young age, even at birth, presenting as signs of severe AS, left heart failure, and failure to thrive.[3]

Adult UAV usually presents as dyspnea, angina, or syncope around the third to fifth decades of life because of the degree of stenosis of the valve. These patients usually have severe AS with or without AR. Isolated AR is less common.[3] Our patient had isolated AR and was asymptomatic at the time of diagnosis. This sheds some light on the natural history of UAVs. It is entirely possible that our patient would have progressed to severe AS had the lesion not been treated.

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Keywords: Bicuspid aortic valve, Unicuspid aortic valve, Aortic stenosis, Aortic regurgitation, Thoracic aortic aneurysm

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VIDEO HIGHLIGHTS

Video 1: Two-dimensional TTE, parasternal short-axis view, demonstrates the aortic valve, which appears to be constructed of only two cusps united by two commissures at each side.
Video 2: Two-dimensional TTE, parasternal long-axis view without (*left*) and with (*right*) color flow Doppler, demonstrates the turbulent eccentric AR flow (in a "mosaic" pattern, which appears as a mix of bright blue and yellow in the picture), highlighted with a *red circle* in the first 2 sec of the video.
Video 3: Two-dimensional TTE, parasternal long-axis view, demonstrates a normal-size left ventricle with normal systolic function and a dilated ascending aorta.
Video 4: Three-dimensional cardiac computed tomography, volume-rendered reconstruction of the heart and vessels, cine rotational display, demonstrates the ascending aortic aneurysm.
Video 5: CMR, phase-contrast through-plane sequence at the aortic valve, demonstrating the unicommissural unicuspid morphology.
Video 6: CMR, coronal display, balanced steady-state free precession sequence at the aortic valve, demonstrates the unicommissural unicuspid morphology and the ascending aortic aneurysm.
Video 7: Two-dimensional TEE, midesophageal aortic valve short-axis (50°) view, without (*left*) and with (*right*) color flow Doppler, demonstrates the unicommissural UAV, origin of AR, and dilated aortic root.
Video 8: Two-dimensional TEE, midesophageal aortic valve long-axis (122°) view without (*left*) and with (*right*) color flow Doppler, demonstrates eccentric, moderate AR (*circle*).
Video 9: Postoperative two-dimensional TEE, midesophageal aortic valve long-axis (140°) view with color flow Doppler, demonstrates mild AR through the pulmonary autograft.

View the video content online at www.cvcasejournal.com.

Aortopathy is present in almost half of patients with UAV, with the prevalence ranging between 42% and 62% in most large studies.[2,3] UAV is associated with aortic dissection, coarctation, and aneurysmal dilation of the ascending aorta. Compared with patients with tricuspid aortic valve, those with UAV have an 18-fold increase in dissection risk, while those with BAV have a 9-fold increase.[3]

Often, UAV is misdiagnosed as BAV on initial imaging; TTE has been shown to have sensitivity of only 27% and specificity of 50%. Although TEE increases the sensitivity to 75% and specificity to 86%,[6] it suffers from the same limitations as TTE because of the high degree of calcification present on the valves at the time of diagnosis. Newer diagnostic modalities and even multimodality imaging incorporating cardiac multidetector computed tomography or CMR promise superior performance in detecting UAV.[3]

Although UAV and BAV have been at times described as different phenotypes of the same disease,[7] making the right diagnosis is important because of the twice increased risk for dissection and because of its relevance to treatment. The treatment for UAV has not been standardized, and BAV guidelines have sometimes been applied, albeit with some slight variations. This increased risk for dissection could prove to be the information needed to proceed with surgery in borderline cases, such as when the aortic root or ascending aorta is between 5.0 and 5.5 cm.[8] Additionally, transcatheter aortic valve implantation has been widely used for the correction of severe AS in recent years and has been used in certain cases of BAV with a Class 2b weak recommendation in the 2020 American College of Cardiology and American Heart Association valvular disease guidelines. However, it is not currently recommended to use this treatment in patients with UAV, due to insufficient data.[3,8] Furthermore, in general, UAV requires surgical treatment at least one to two decades earlier than BAV. This is a consequence of the earlier presentation and increased likelihood of more severe AS.[7]

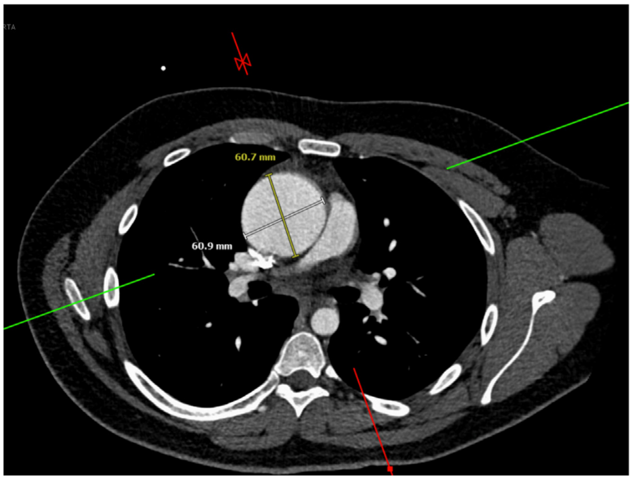

Figure 1 Computed tomographic angiography of the chest, transverse section, demonstrates the aneurysm of the ascending aorta.

The most common treatment is the aortic valve replacement, which has well-documented long-term results but suffers from prosthesis- and anticoagulation-related complications. Aortic valve repair techniques including bicuspidization and tricuspidization have been used successfully, but the long-term data are sparse. Last, the Ross technique, which consists of replacing the affected valve with the patient's own pulmonic valve, provides good long-term viability without the need for anticoagulation. However, it does require technical expertise in the procedure and should be done at high-volume centers only.[2] Another consequential limitation of the Ross procedure is the likely need for another procedure 15 to 20 years later due to the development of regurgitation or stenosis in the pulmonic homograft.[9]

Our case highlights the importance of follow-up of congenital aortic disease. Although there are no specific guidelines for UAV, the 2022 American College of Cardiology and American Heart Association guidelines for the diagnosis and management of aortic disease currently recommend lifelong surveillance of patients with BAV, without specifying the frequency of the imaging.[10] Furthermore, the prevalence of BAV in first-degree relatives of BAV patients is 9% to 20%, and screening first-degree relatives of patients with BAV is indicated when aortic root or ascending aortic dilation is present (Class 1, strong recommendation) and should be considered in all patients with BAV regardless of aortopathy (Class 2a, moderate recommendation).[10] Had our patient not presented for a wellness examination and the aortic aneurysm not been diagnosed, they would have remained at a significant risk for dissection.

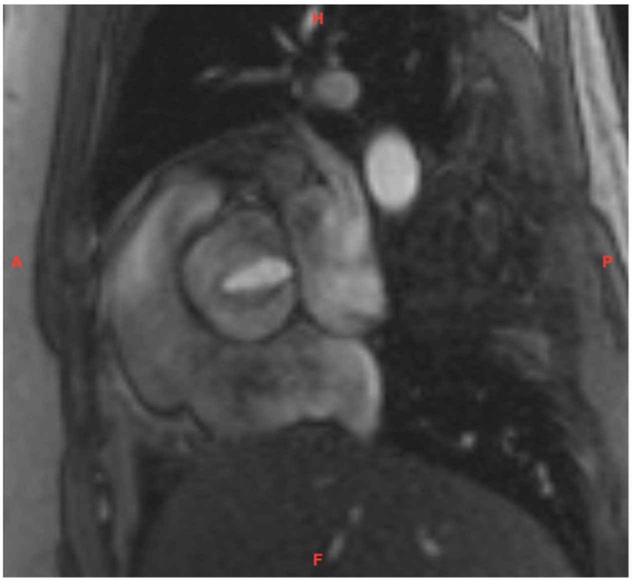

Figure 2 CMR, through-plane phase contrast sequence, sagittal display at the aortic valve in systole, demonstrates the patient's unicommissural UAV.

CONCLUSION

UAV is a rare abnormality that can be initially misdiagnosed as BAV. It is worth considering more accurate imaging, such as TEE and CMR, in younger patients with BAV, as the exact valve morphology may alter management. In such patients, it is important to bear in mind the importance of surveillance and follow-up, early after diagnosis, especially after repairs such as the Ross procedure with known and predictable sequelae.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with the following guidelines: CARE guidelines for case reports.

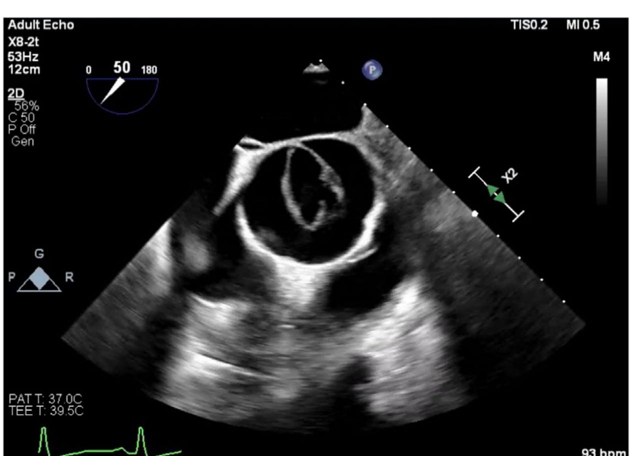

Figure 3 Two-dimensional TEE, midesophageal aortic valve short-axis view in systole, demonstrates the unicommissural UAV with mildly restricted opening.

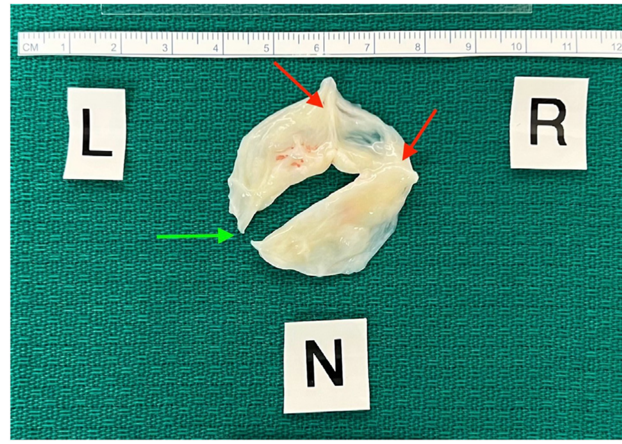

Figure 4 Surgical specimen, patient's UAV, demonstrates one functional commissure (*green arrow*) and two raphes (*red arrows*).

CONSENT STATEMENT

Complete written informed consent was obtained from the patient (or appropriate parent, guardian, or power of attorney) for the publication of this study and accompanying images.

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DISCLOSURE STATEMENT

The authors report no conflict of interest.

SUPPLEMENTARY DATA

Supplementary data to this article can be found online at https://doi.org/10.1016/j.case.2023.12.020.

REFERENCES

1. Novaro GM, Mishra M, Griffin BP. Incidence and echocardiographic features of congenital unicuspid aortic valve in an adult population. J Heart Valve Dis 2003;12:674-8.
2. Naito S, Sequeira-Gross T, Petersen J, Holst T, Reichenspurner H, Girdauskas E. Focus on a rare clinical entity: unicuspid aortic valve disease. Expert Rev Cardiovasc Therapy 2020;18:625-33.
3. Pan J. Unicuspid aortic valve: a rare congenital anomaly. Cardiology 2021;147:207.
4. Edwards JE. Pathologic aspects of cardiac valvular insufficiencies. AMA Arch Surg 1958;77:634-49.
5. Roberts WC, Ko JM. Frequency by decades of unicuspid, bicuspid, and tricuspid aortic valves in adults having isolated aortic valve replacement for aortic stenosis, with or without associated aortic regurgitation. Circulation 2005;111:920-5.
6. Chu JW, Picard MH, Agnihotri AK, Fitzsimons MG. Diagnosis of congenital unicuspid aortic valve in adult population: the value and

limitation of transesophageal echocardiography. Echocardiography 2010;27:1107-12.
7. Krepp JM, Roman MJ, Devereux RB, Bruce A, Prakash SK, Morris SA, et al. Bicuspid and unicuspid aortic valves: different phenotypes of the same disease? Insight from the GenTAC Registry. Congenit Heart Dis 2017;12: 740-5.
8. Otto C, Nishimura R, Bonow RO, Carabello BA, Erwin JP 3rd, Gentile F, Jneid H, et al. 2020 ACC/AHA guideline for the management of patients with valvular heart disease: a report of the American college of Cardiology/American heart association Joint Committee on clinical Practice guidelines. J Am Coll Cardiol 2021;77:e25-197. Section 5.1.1.2.
9. Brown KN, Kanmanthareddy A. Ross procedure for aortic valve replacement. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023.
10. Isselbacher E, Preventza O, Hamilton Black J 3rd, Augoustides JG, Beck AW, Bolen MA, et al. 2022 ACC/AHA guideline for the diagnosis and management of aortic disease: a report of the American Heart Association/American College of Cardiology Joint Committee on clinical Practice guidelines. J Am Coll Cardiol 2022;146:e334-482.