INTRODUCTION

The most common congenital abnormality of the aortic valve is a bicuspid valve. Less common abnormalities include unicuspid and quadricuspid valves. The latter is a rare finding that is often associated with aortic regurgitation. Congenital aortic valve abnormalities such as this may be missed on standard transthoracic echocardiography (TTE). Advanced imaging modalities, including transesophageal echocardiography (TEE) and magnetic resonance imaging (MRI), are useful in providing detailed anatomical assessment of valve morphology.

Figure 1 Two-dimensional TTE images of the left ventricle from the parasternal long-axis view in diastole (A) and systole (B) showing severe left ventricular dilation. (C) Color Doppler imaging showing moderate-to-severe eccentric aortic regurgitation. (D, arrow) Pulsed-wave Doppler imaging of the descending thoracic aorta demonstrating diastolic flow reversal. (E) Continuous-wave Doppler of aortic regurgitation. The measured pressure half-time was 400 msec.

Figure 2 TEE image of the aortic valve in short-axis view demonstrating four cusps. LCC, left coronary cusp; NCC, noncoronary cusp; RCC, right coronary cusp.
Figure 3  TEE images of the QAV in short- (A-B) and long-axis views (C-D) with and without color Doppler demonstrating severe eccentric aortic regurgitation.

Figure 4  (A-B) TEE images of the QAV in short-axis view during systole demonstrating fusion of the right and noncoronary cusps and fusion of the left and accessory coronary cusps (arrows).
The following case illustrates the use of TEE in making this diagnosis in a patient with severe aortic regurgitation.

CASE PRESENTATION

A 52-year-old Hispanic male with a past medical history of uveitis and depression was seen in clinic for the evaluation of a cardiac murmur. He is generally active and exercises regularly including weight lifting, walking up to 5 miles/day, and climbing stairs without angina or dyspnea. Over the prior 6 months, however, he has noticed fatigue with these activities. He is a former smoker and has a family history of a sister who underwent cardiac surgery at age 52 for unclear indication. His current medications include artificial tears, prednisolone eye drops, and ibuprofen as needed for minor pain. He has no surgical history. On physical exam his blood pressure was 125/64, and his heart rate was 64 bpm and regular. He had no jugular venous distension and clear lungs. His point of maximal impulse was laterally displaced. Cardiac auscultation revealed a 3/6 early-mid diastolic murmur loudest at the apex. Routine labs including chemistries, complete blood count, coagulation panel, lipids, and liver function tests were unremarkable. A TTE was performed (Figure 1), which demonstrated a severely dilated left ventricle with preserved left ventricular ejection fraction of 55%-60%, mildly dilated aortic root measuring 3.9 cm in diameter, and a possible bicuspid aortic valve with moderate-severe eccentric aortic regurgitation. His left ventricular internal diameter in diastole and systole measured 7.0 cm and 5.3 cm, respectively, in the parasternal long axis view. He was subsequently referred for TEE, which showed a quadricuspid aortic valve (QAV; Figure 2) with severe aortic insufficiency by color Doppler (Figure 3, Video 1). There was also fusion of the right and noncoronary cusps as well as the left and accessory cusps creating a functionally bileaflet valve (Figure 4, arrows). His aortic root and ascending aorta were mildly dilated, measuring 4.1 cm and 3.9 cm in diameter, respectively (Figure 5). A cardiac MRI was also obtained that confirmed the QAV morphology (Figure 6) and severe aortic regurgitation with a calculated regurgitant volume of 64 mL.

Given his severely dilated left ventricle and recent exertional symptoms, he was referred for surgical aortic valve replacement. Preoperative coronary angiography was unremarkable. He underwent a minimally invasive aortic valve replacement with a 27-mm St. Jude mechanical valve and had an uneventful postoperative course. Follow-up TTEs at 6 months and 2 years postoperatively demonstrated a decrease in his left ventricular size back to normal (Figure 7).

DISCUSSION

QAV is an uncommon congenital cardiac malformation affecting approximately 0.01% of the population\(^1\) and is often discovered incidentally at the time of valve surgery or at autopsy. The true incidence is difficult to measure as this finding may be missed on standard TTE, although prior autopsy studies have confirmed the rare occurrence of this valvular deformity. It is frequently associated with progressive aortic regurgitation, with nearly 50% of affected patients going on to require aortic valve replacement surgery during their lifetime. Dilatation of the aortic root and ascending aorta has also been reported in a significant number of affected patients.\(^2\) In a recent series from the Mayo Clinic, 29% of QAV cases demonstrated aortic dilatation involving either the aortic root, ascending aorta, or both, and this finding was associated with a high incidence of moderate or greater aortic regurgitation.\(^3\) Aortic stenosis is a less common finding in patients with QAV, affecting 8% of cases in the

![Figure 5 TEE images demonstrating dilation of the aortic root (A) and ascending aorta (B).](image)

![Figure 6 Cardiac MRI demonstrated QAV morphology, leaflets numbered 1-4.](image)
aforementioned series. In addition, other structural congenital cardiovascular abnormalities including atrial and ventricular septal defects, nonaortic valvular abnormalities, coronary anomalies, and patent ductus arteriosus have been reported. In 1973, Hurwitz and Roberts described seven distinct subtypes of quadricuspid semilunar valve morphology based on the cusp size and distribution. Based on this classification scheme, our patient has type A with relatively equal size cusps. What is unique about this case, however, is the fusion of the right and noncoronary cusps as well as the left and accessory cusps, resulting in a functionally bicuspid aortic valve. Whether the aortic dilatation seen in QAV is due to a genetic aortopathy, as may occur in bicuspid aortic valve, or simply the result of hemodynamic consequences of progressive valve dysfunction is uncertain. Current guidelines do not include specific approaches to management of patients with QAV and associated valvular regurgitation and/or aortic dilatation. Clinical decision making with regards to surveillance imaging and indications for surgical intervention in QAV should therefore be made according to the standard guideline recommendations used in patients with tricuspid aortic valves.

**CONCLUSION**

QAV is a rare cardiac abnormality commonly associated with valve regurgitation and aortic dilatation. Although TTE remains the imaging modality of choice in evaluating valve anatomy and function, the reported incidence of QAV may be underestimated due to limitations in accurately characterizing aortic valve morphology with this modality (Figure 8). In patients with a suspected structurally abnormal aortic valve with significant regurgitation on TTE, additional imaging with TEE or cardiac MRI should be considered for further evaluation.

**Figure 7** Two-dimensional TTE images of the left ventricle from the parasternal long-axis view at end diastole at 6 months (A) and 2 years (B) postoperatively demonstrating decrease in LV internal diameter.

**Figure 8** Short-axis views of the aortic valve in diastole using TTE (A) and TEE (B) demonstrating the superiority of TEE in accurately evaluating aortic valve morphology.
SUPPLEMENTARY DATA

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.case.2017.01.013.

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