

Quadricuspid Aortic Valve With Ostium Secundum Atrial Septal Defect



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INTRODUCTION

Quadricuspid aortic valve (QAV) is a rare congenital cardiac malformation with an estimated incidence of 0.003% to 0.043% of all congenital heart diseases.¹

Previously the diagnosis of QAV was made mostly during intraoperative or postmortem investigation, but now, with the advent of better imaging techniques such as transthoracic echocardiography, transesophageal echocardiography, and cardiac magnetic resonance imaging, more cases are being diagnosed in asymptomatic patients.

The importance of diagnosing this congenital anomaly in asymptomatic adults lies in the fact that >50% will require valve repair or replacement in the fifth or sixth decade of life because of worsening aortic regurgitation (AR) or stenosis.

CASE PRESENTATION

A 23-year-old man presented with a history of New York Heart Association functional class II dyspnea along with palpitations for 2 years. On physical examination, blood pressure was 120/80 mm Hg, pulse rate was 90 beats/min and regular, and a grade II/IV diastolic murmur was heard at the third left intercostal space. The murmur was increased by handgrip exercise and squatting position and decreased by standing from squatting and by Valsalva maneuver. Another grade III/VI ejection systolic murmur was present at the second left intercostal space. Electrocardiography showed normal sinus rhythm with right bundle branch block.

Two-dimensional transthoracic echocardiography was done using a 3.0-MHz transducer and a Vivid S5 cardiac ultrasound system (GE Healthcare, Milwaukee, WI), which showed a dilated right atrium and right ventricle, an ostium secundum atrial septal defect 34.6 mm in size (Figure 1, Video 1) with a left-to-right shunt, mild tricuspid regurgitation, and mild pulmonary arterial hypertension. The aortic valve was quadricuspid, with three equally sized cusps and one smaller cusp (type B QAV; Figures 2 and 3, Videos 2 and 3), with mild to moderate AR (Figure 4, Video 4). Left ventricular (LV) end-diastolic dimension, LV end-systolic dimension, and LV function were normal. Two-dimensional transesophageal echocardiography was done using the same ultrasound system for detailed

evaluation of the aortic valve, which confirmed the findings of two-dimensional transthoracic echocardiography.

Because AR was mild to moderate and LV dimensions were normal, the patient was advised to undergo surgical patch closure of the atrial septal defect with close follow-up for AR. Intraoperatively, findings of QAV were confirmed.

DISCUSSION

QAV is a rare congenital cardiac malformation with an estimated incidence of 0.003 to 0.043% of all congenital heart diseases.¹ QAV was first identified on autopsy by Balington in 1862; the first in vivo description appeared in 1968, by Robicsek *et al.* Systemic autopsy studies have estimated its incidence between 0.003% and 0.008%.

The embryology of QAV is unknown. Valve development starts during the fifth week, with two mesenchymal ridges that form the cephalic portion of the truncus arteriosus. These ridges fuse, descend into the ventricles, and form the aortopulmonary septum. Each semilunar valve consists of three areas of mesenchymal swelling that grow up, become excavated, and form three cusps. This process is well advanced by week 6 and virtually completed by week 9.

Various pathophysiologic mechanisms have been suggested that could alter the number of valve leaflets, including anomalous septation of the conotruncus, excavation of one of the valve cushions, and septation of a normal valve cushion as a result of an inflammatory episode.² Some studies have suggested that QAV may result from the division of one of the three mesenchymal ridges that normally give rise to three aortic valve cushions.^{3,4} The development of the aortic valve leaflets occurs temporally just after the formation process of the coronary arteries originating from the sinuses of Valsalva.⁵ For this reason, one could speculate that a single developmental abnormality might result in a variety of abnormalities in the aortic root. Therefore, abnormalities in septation of aortic cusps could be embryologically related to abnormal growth of coronary arteries.

True QAVs must be distinguished from pseudo-QAVs resulting from bacterial endocarditis or rheumatic fever. To be considered a truly congenital malformation caused by abnormal embryogenesis, each cusp should contain a corpus Arantii.

In 1973, Hurwitz and Roberts classified the quadricuspid semilunar valve according to their anatomic variations. They described seven variations, types A to G (Table 1). The most frequent anatomic variations are types A and B, and both are more likely to result in AR than other anatomic variants. According to Hurwitz and Roberts's classification system, our patient had three equal larger cusps and one smaller cusp (type B), leading to mild to moderate AR due to the unequal sizes of the cusps. Pure AR was found in almost 75% of cases, while 16% of the quadricuspid valve functions normally.^{6,7} Very rarely, QAV is associated with aortic stenosis.⁸

Congenital QAV is usually an isolated lesion, but several congenital cardiac abnormalities have been described with it. The most prevalent cardiac malformations associated with QAV are coronary artery anomalies, which have been reported in 10% of cases. Various forms

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VIDEO HIGHLIGHTS

Video 1: Parasternal short-axis view showing QAV in systole and diastole.

Video 2: Parasternal long-axis view showing mild AR.

Video 3: Apical four-chamber view showing ostium secundum atrial septal defect.

Video 4: Modified parasternal short-axis view showing color flow through atrial septal defect.

[View the video content online at www.cvcasejournal.com.](http://www.cvcasejournal.com)

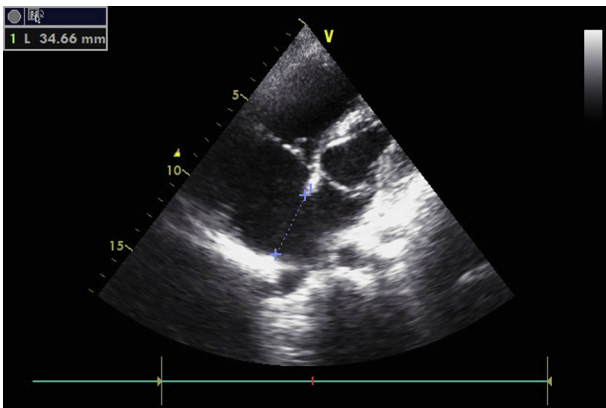


Figure 1 Transthoracic echocardiography, modified apical four-chamber view, showing ostium secundum atrial septal defect.

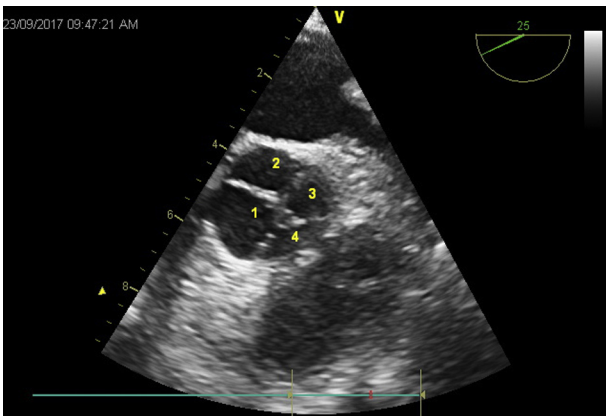


Figure 2 Transesophageal echocardiography showing QAV in diastole.

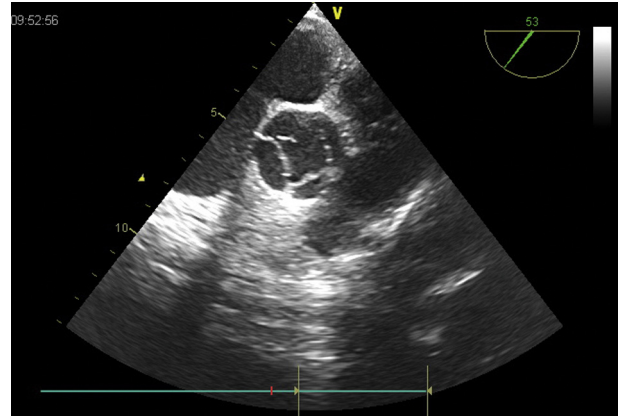


Figure 3 Transesophageal echocardiography showing QAV in systole.

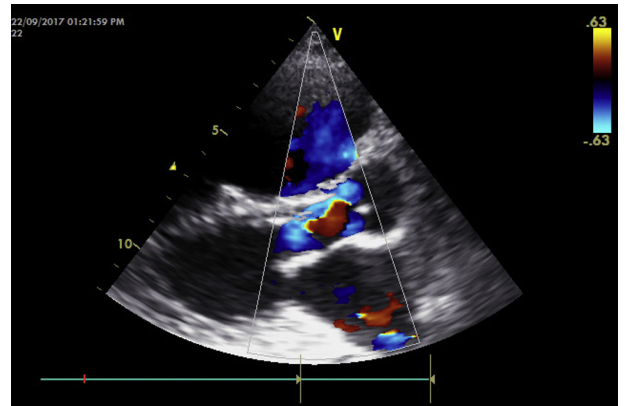


Figure 4 Transthoracic echocardiography, parasternal long-axis view, showing mild AR.

Table 1 Hurwitz and Roberts's anatomic classification of QAV²

Type	Description
A	Four equal cusps
B	Three equal cusps and one smaller cusp
C	Two equal larger cusps and two equal smaller cusps
D	One large, two intermediate, and one small cusp
E	Three equal cusps and one larger cusp
F	Two equal larger cusps and two unequal smaller cusps
G	Four unequal cusps

Types A, B, and C are represented in >85%, while type D is very rare.

of presentation of such anomalies have been reported, including anomalous position of one of the coronary ostia, single coronary ostium, coronary-pulmonary artery fistula, and occlusion of a coronary artery ostium by valve tissue, which can be fatal. It is very important to diagnose these associated anomalies before repair or replacement of aortic valve to avoid ostial obstruction.

Atrial septal defect, ventricular septal defect, patent ductus arteriosus, pulmonary stenosis, subaortic stenosis, hypertrophic cardiomyopathy, congenital complete heart block, and also cases of bacterial endocarditis

affecting a QAV have been reported. Very few cases of QAV in association with atrial septal defect are described in the literature.⁹⁻¹²

Although QAV is a congenital malformation, diagnosis is usually late. Wider use of echocardiography has made detection of QAV easier and hence more frequent.¹³ The characteristic echocardiographic finding in the short-axis view is an X-shaped commissure pattern during diastole and a rectangular appearance during systole.¹⁴ Transthoracic echocardiography has an important role in detecting congenital anomalies preoperatively. However, transthoracic echocardiography may be

suboptimal for recognizing QAV, and with more liberal use of transeosophageal echocardiography, computed tomography, and magnetic resonance imaging, the prevalence of QAV is increasing.

CONCLUSION

The importance of diagnosing QAV in asymptomatic adults lies in the fact that >50% will require valve repair or replacement in the fifth or sixth decade of life because of worsening AR or stenosis. Repair may be feasible in some patients with regurgitation, but most require replacement. Hence, patients with QAV are not simply of academic interest; rather they should undergo regular and close follow-up and have aortic valve repair or replacement at an appropriate time before LV decompensation to avoid morbidity, mortality, and complications.

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SUPPLEMENTARY DATA

Supplementary data related to this article can be found at <https://doi.org/10.1016/j.case.2019.03.004>.

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