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Case Reports

Quadricuspid Aortic Valve: Imaging, Diagnosis, and Prognosis

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Abstract

Quadricuspid aortic valve is a rare congenital cardiac anomaly with an incidence of 0.008% to 0.043%. Its clinical course varies depending on cusp anatomy, function, and associated cardiac malformations. It frequently progresses to aortic valve regurgitation that may require surgical valve replacement. Detection has shifted from incidental discovery during autopsies or cardiac surgeries in the early 20th century to various cardiac imaging methods in recent decades. In addition to contributing to the literature, this report supports the use of transesophageal echocardiography more liberally to detect aortic valve abnormalities. The case presents a 48-year-old female patient with an incidentally discovered quadricuspid aortic valve.

Keywords: Quadricuspid aortic valve; echocardiography, transesophageal; aortic valve insufficiency

Case Report

Presentation and Physical Examination

A 48-year-old White woman was referred to the reporting institution for a cardiovascular consult because of a diagnosis of bicuspid aortic valve (BAV) made when she was 26 years old. The diagnosis of BAV was based on transthoracic echocardiography (TTE). She denied experiencing chest pain, syncope, and shortness of breath. The physical examination was unremarkable, except for a grade 1 systolic ejection murmur and a grade 2 diastolic murmur at the left sternal border.

Medical History

The patient has a history of gastroesophageal reflux disease, cholecystectomy, hysterectomy, and hernia repair.

Differential Diagnosis

Valve neoplastic involvement; valvular degeneration with or without calcifications, adherent thrombus, or vegetation; and aortic valve tumors (eg, papillary fibroelastoma and myxoma) were discussed.

Technique

Routine blood work, general chemistry, and the patient's lipid panel were well within the normal range. A transesophageal echocardiogram revealed a quadricuspid aortic valve (QAV) and a left ventricular ejection fraction of 63% (Fig. 1); a color Doppler echocardiogram revealed a QAV with moderate aortic valve regurgitation of grade 2 or

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higher (Fig. 2). Computed tomography (CT) angiography showed a QAV but was negative for aortic dilation, tortuosity, and dissection or coronary disease (Fig. 3).

Outcome

The patient was counseled regarding her condition, including the increased risk it presented for aortic valve regurgitation and thoracic aortic dilation. Because the patient was asymptomatic, with only an incidental finding of QAV, she was given instructions to schedule a regular follow-up in the cardiology clinic.

Key Points

- Quadricuspid aortic valve is a rare congenital cardiac anomaly.
- Aortic valve regurgitation or stenosis may be the initial presentation of such an anomaly.
- This report highlights the importance of TEE in detecting a case of this rare anomaly, which was diagnosed previously as BAV by TTE.

Abbreviations and Acronyms

BAV	bicuspid aortic valve
CT	computed tomography
QAV	quadricuspid aortic valve
TEE	transesophageal echocardiography
TTE	transthoracic echocardiography

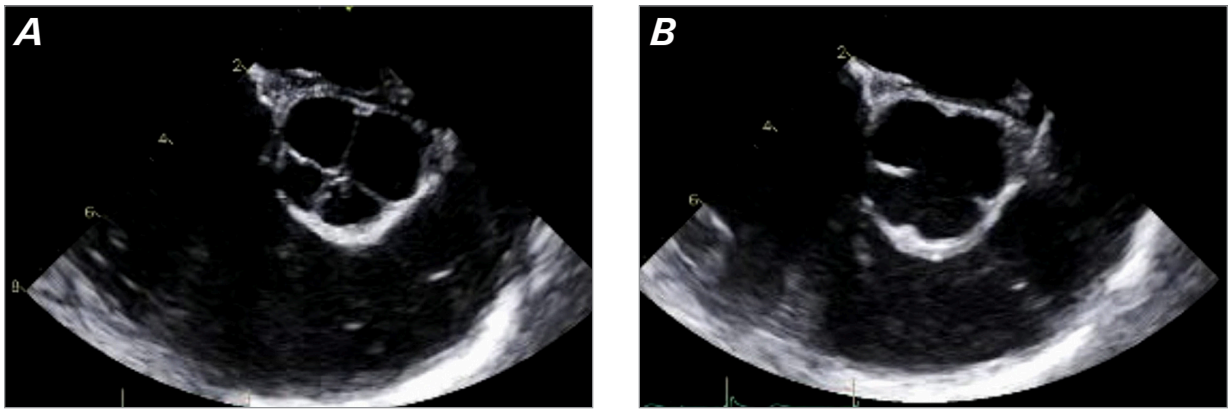

Fig. 1 A transesophageal echocardiogram's short-axis view shows quadricuspid aortic valve in **(A)** diastolic frame and **(B)** systolic frame.

Supplemental motion image is available for Figure 1.

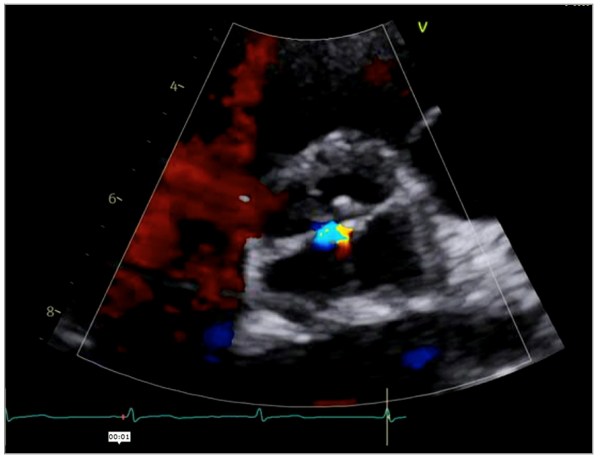

Fig. 2 A color Doppler echocardiogram's short-axis view shows a quadricuspid aortic valve.

Supplemental motion image is available for Figure 2.

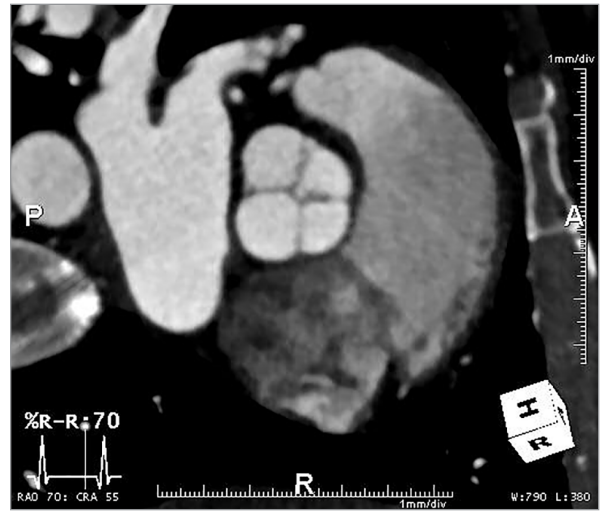

Fig. 3 A chest computed tomography angiogram's axial view shows a quadricuspid aortic valve.

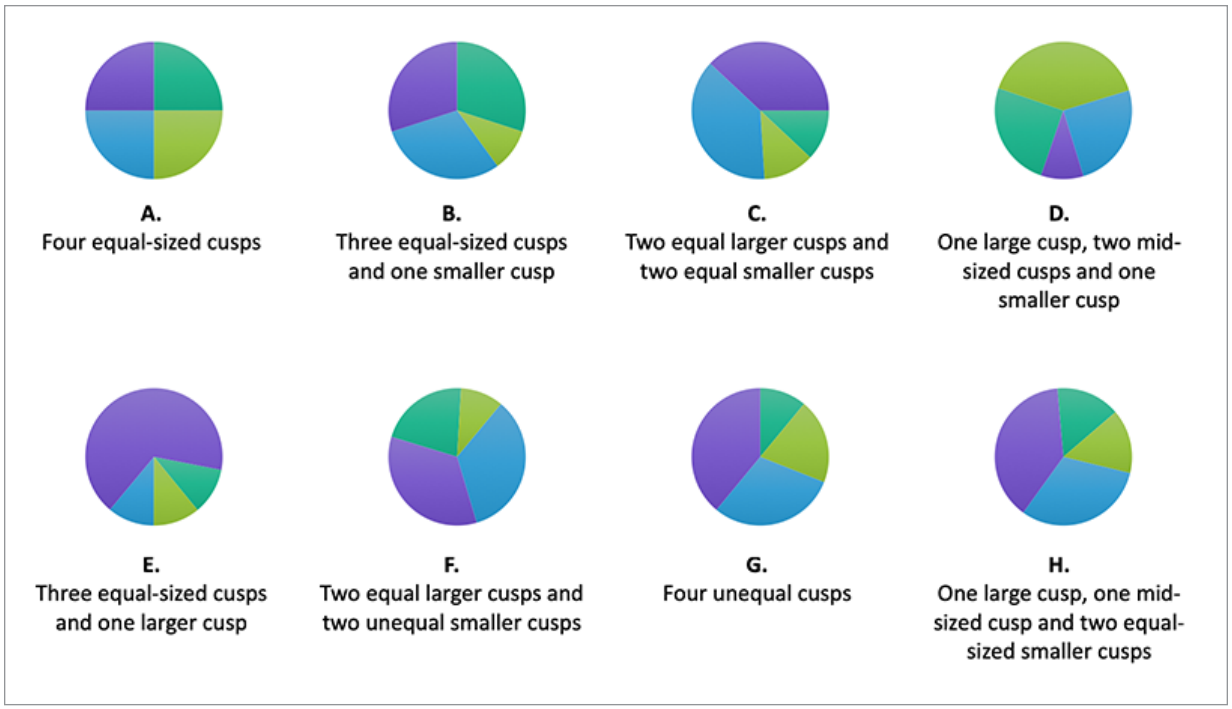

Fig. 4 Diagram of the common types of quadricuspid aortic valve cusp configuration according to the size of each leaflet.[7]

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Latest Follow-Up

Follow-up 37 months after the initial diagnosis of QAV showed a stable echocardiogram and no clinical symptoms. The patient was advised to repeat echocardiography in 2 years.

Discussion

Quadricuspid aortic valve is a rare congenital cardiac defect with approximately 200 reported cases worldwide in the literature.[1] With improving cardiac imaging techniques, including transesophageal echocardiography (TEE), cardiac CT, and cardiac magnetic resonance imaging, detection of QAV has increased.[2] The primary consequences of this anomaly are aortic valve regurgitation and thoracic aortopathy.[3] Quadricuspid aortic valve is a congenital cardiac anomaly where the normal tricuspid aortic valve has 4 equal or unequal cusps instead of 3 cusps. Its incidence is 0.008% on autopsy, 0.043% by 2-dimensional TTE, and 1% in patients scheduled for aortic valve surgery.[1] Though QAV is a congenital cardiac anomaly, patients are most commonly identified in their 50s, with a slight male predominance (1.6:1) in the reported cases.[4-6] Seven anatomic variations of QAVs (Fig. 4A through Fig. 4G) were described based on autopsy findings in 10 patients and previous reports of 97 cases.[7] Their classification was devised according to the relative size of each cusp in a QAV. Vali et al[6] reported an eighth variation characterized by 2 equal-sized smaller cusps and 2 unequal-sized larger cusps (Fig. 4H). The present patient has 2 equally larger cusps and 2 equally smaller cusps (Fig. 1A), similar to type C.

Although the mechanism of aberrant cusp formation has not been established, 1 widely accepted theory proposes that QAV arises from abnormal septation of the truncus arteriosus or endocardial cushions during embryogenesis.[3,8] Quadricuspid aortic valve usually occurs as an isolated cardiac anomaly (as seen in this patient) but has been reportedly associated with other structural cardiac defects, including hypertrophic obstructive cardiomyopathy, aberrant origin of coronary ostia, single coronary ostium, thoracic aortic dilation and aneurysm, ventricular septal defect, transposition of the great arteries, tetralogy of Fallot, and patent ductus arteriosus.[2,8,9]

Depending on the functional status of the valve and associated anomalies, the patient may be asymptomatic[10,11] or present with chest pain, syncope, shortness of breath, palpitations, fatigue, or peripheral edema.[2] Progressive aortic valve regurgitation is the most common clinical sequela for QAV.[3] Aortic valve stenosis, aortic valve infective endocarditis, thoracic aortic dilation, and aneurysm have also been reported as complications.[12,13] The unequal size of the cusps and abnormal leaflet coaptation may contribute to the progression of aortic valve regurgitation and gradual deterioration of the valves over time.[2,5,12] The risk of developing aortic valve infective endocarditis may be associated with unequal cusp size, leading to unequal distribution of stress. The risk of thoracic aortic dilation and aneurysm is similar to the aortopathy associated with BAV. Because this patient's aortic valve has 2 large and 2 small cusps, she will need regular follow-up and monitoring.

During the early 20th century, QAV was mostly found during autopsies or cardiac surgery. In 1983, Herman et al[14] noninvasively diagnosed a case of QAV using 2-dimensional TTE, and since then echocardiography has remained the primary imaging method of detection. Other diagnostic imaging modalities, such as TEE, cardiac CT, and cardiac magnetic resonance imaging, are superior to TTE for characterizing QAV anatomy and classification.[1,10] Transesophageal echocardiography, in particular, has better diagnostic accuracy than TTE and has emerged as the preferred diagnostic tool in recent years.[2,15] The visualization of 4 cusps makes the diagnosis of QAV straightforward, but many diseases and conditions can mimic QAV, leading to a lengthy list of differential diagnoses, including the cardiac benign neoplasms papillary fibroelastoma and myxoma.[16] In the past 2 decades, a study evaluated 126 patients who underwent aortic valve replacement and determined the sensitivity (61%), specificity (81%), and accuracy (77%) of the diagnosis of BAV with TTE as well as the sensitivity (86%), specificity (90%), and accuracy (89%) of diagnosis with TEE.[17] To the best of the authors' knowledge, no study has formally been conducted to determine the sensitivity, specificity, and accuracy of TEE over TTE for diagnosis of QAV. It is possibly the rare occurrence and limited number of known cases restrict such a study, and therefore patients may be incorrectly diagnosed with BAV by TTE. The present patient is a 48-year-old woman who presented with a murmur and a previous diagnosis of BAV by TTE. Repeated TTE failed to recognize her aortic valve as quadricuspid, which was identified after performing TEE. On a similar note, Garg et al[18] detected a rare type F QAV in a 52-year-old woman with a 7-year history of worsening dyspnea using 3-dimensional TEE, though the aortic valve initially appeared to be bicuspid on TTE. There have been reports of 3 other patients for whom TTE could not characterize aortic valve morphology but for whom TEE revealed the aortic valve to be quadricuspid.[11,19,20] Because of the limited number of QAV cases, it is important for the clinician to be aware that BAV diagnosed by TTE may represent QAV. Clinically, BAV is more likely to be associated with aortic valve stenosis, while QAV is associated with aortic valve regurgitation.

The course of management is guided by the severity of symptoms such as aortic valve regurgitation and thoracic aortic dilation and aneurysm. Aortic valve replacement or repair and thoracic aorta replacement or repair have been needed in a few instances.[21,22] Because there are no universal guidelines for QAV, regular follow-up with patients with QAV is imperative to monitor for early signs of valvular compromise, such as in this patient, whose condition was asymptomatic and monitored with echocardiography and CT angiography every 2 years.[23] Aortic valve regurgitation tends to occur around the sixth decade of life, often requiring aortic valve replacement.[1]

Again, QAV is a rare congenital cardiac anomaly, and its incidence may increase with the use of rapidly improving cardiac imaging techniques. Transesophageal echocardiography is considered the gold standard for diagnosis and follow-up of a patient with QAV.

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References

1. Khan SKA, Tamin SS, Araoz PA. Quadricuspid aortic valve by cardiac magnetic resonance imaging: a case report and review of the literature. *J Comput Assist Tomogr.* 2011;35(5):637-641. doi:10.1097/RCT.0b013e318224a129
2. Yuan SM. Quadricuspid aortic valve: a comprehensive review. *Braz J Cardiovasc Surg.* 2016;31(6):454-460. doi:10.5935/1678-9741.20160090
3. Hayakawa M, Asai T, Kinoshita T, Suzuki T. Quadricuspid aortic valve: a report on a 10-year case series and literature review. *Ann Thorac Cardiovasc Surg.* 2014;20 Suppl:941-944. doi:10.5761/atcs.cr.13-00125
4. Tutarel O. The quadricuspid aortic valve: a comprehensive review. *J Heart Valve Dis.* 2004;13(4):534-537.
5. Suraci N, Horvath SA, Urina D, Rosen G, Santana O. Quadricuspid aortic valve: case series and review of literature. *Echocardiography.* 2019;36(2):406-410. doi:10.1111/echo.14240
6. Vali Y, Rajendra R, Nishtar S. A previously undescribed type of quadricuspid aortic valve: type H. *J Heart Valve Dis.* 2010;19(6):792-793.
7. Hurwitz LE, Roberts WC. Quadricuspid semilunar valve. *Am J Cardiol.* 1973;31(5):623-626. doi:10.1016/0002-9149(73)90332-9
8. Attaran RR, Habibzadeh MR, Baweja G, Slepian MJ. Quadricuspid aortic valve with ascending aortic aneurysm: report of a case and discussion of embryological mechanisms. *Cardiovasc Pathol.* 2009;18(1):49-52. doi:10.1016/j.carpath.2007.07.012
9. Tsang MYC, Abudiab MM, Ammash NM, et al. Quadricuspid aortic valve: characteristics, associated structural cardiovascular abnormalities, and clinical outcomes. *Circulation.* 2016;133(3):312-319. doi:10.1161/CIRCULATIONAHA.115.017743
10. Okmen AS, Okmen E. Quadricuspid aortic valve without severe dysfunction despite advanced age. *Tex Heart Inst J.* 2009;36(5):486-488.
11. Pasanisi G, Mazzanti G, Sassone B. A quadricuspid aortic valve in an asymptomatic 40-year-old man: a case report. *J Med Case Rep.* 2018;12(1):234. doi:10.1186/s13256-018-1755-3
12. Takeda N, Ohtaki E, Kasegawa H, Tobaru T, Sumiyoshi T. Infective endocarditis associated with quadricuspid aortic valve. *Jpn Heart J.* 2003;44(3):441-445. doi:10.1536/jhj.44.441
13. Watanabe Y, Taketani Y, Takei Y, Tanaka K, Watanabe Y. Complete heart block resulting from quadricuspid aortic valve penicillin-resistant pneumococcal endocarditis: a case report. *Circ J.* 2003;67(3):275-276. doi:10.1253/circj.67.275
14. Herman RL, Cohen IS, Glaser K, Newcomb EW III. Diagnosis of incompetent quadricuspid aortic valve by two-dimensional echocardiography. *Am J Cardiol.* 1984;53(7):972. doi:10.1016/0002-9149(84)90546-0
15. Alegret JM, Palazón O, Duran I, Vernis JM. Aortic valve morphology definition with transthoracic combined with transesophageal echocardiography in a population with high prevalence of bicuspid aortic valve. *Int J Cardiovasc Imaging.* 2005;21(2-3):213-217. doi:10.1007/s10554-004-3901-9
16. Saith S, Saith S, Murthy A. Quadricuspid aortic valve: an introduction for clinicians. *Cardiol Res.* 2022;13(1):2-10. doi:10.14740/cr1308
17. Takeda H, Muro T, Saito T, et al. Diagnostic accuracy of transthoracic and transesophageal echocardiography for the diagnosis of bicuspid aortic valve: comparison with operative findings. *Osaka City Med J.* 2013;59(2):69-78.
18. Garg P, Kamaruddin H, Orme R, Watt V. Type F congenital quadricuspid aortic valve: a very rare case diagnosed by 3-dimenional transoesophageal echocardiography. *Open Cardiovasc Med J.* 2014;8:23-25. doi:10.2174/1874192401408010023
19. Armen TA, Vandse R, Bickle K, Nathan N. Three-dimensional echocardiographic evaluation of an incidental quadricuspid aortic valve. *Eur J Echocardiogr.* 2008;9(2):318-320. doi:10.1016/j.euje.2007.03.041
20. Dencker M, Stagmo M. Quadricuspid aortic valve not discovered by transthoracic echocardiography. *Cardiovasc Ultrasound.* 2006;4:41. doi:10.1186/1476-7120-4-41
21. Jagannath AD, Johri AM, Liberthson R, et al. Quadricuspid aortic valve: a report of 12 cases and a review of the literature. *Echocardiography.* 2011;28(9):1035-1040. doi:10.1111/j.1540-8175.2011.01477.x
22. Plaunova A, Gulkarov I, Tortolani AJ, Worku B. Surgery for a quadricuspid aortic valve: case report and comprehensive review of the literature. *J Heart Valve Dis.* 2015;24(2):260-262.
23. Khatun N, Kaliounji A, Alkoutami SS, Francois J, John S. Quadricuspid aortic valve: an incidental finding in an elderly man. *Cureus.* 2023;15(5):e39536.