Functionally Bicuspid Quadricuspid Aortic Valve

Esad Vucic, MD, PhD, Krishna Prasad Kurpad, MD, Reenal Patel, MD, Yuliya Kats, MD, and Alfonso H. Waller, MD, *Newark and Livingston, New Jersey*

INTRODUCTION

A quadricuspid aortic valve (QAV) is a rare congenital defect, with an incidence in the general population of less than 0.05%.[1-3] Most frequently, QAV is an incidental finding found on imaging or autopsy. In a pooled series of autopsies conducted by Simonds *et al,* 2 cases of QAV were identified in 25,666 subjects (incidence, 0.008%).[3] The reported incidence of QAV based on transthoracic echocardiography (TTE) is 0.043% to 0.006%.[3]

The primary defect in a majority of QAV patients is isolated aortic regurgitation (AR), with a minority of patients developing aortic stenosis.[1] Symptomatic patients present in the fifth or sixth decade with heart failure resulting from AR, which is the predominant indication for aortic valve replacement.[1] Transesophageal echocardiography (TEE) plays a pivotal role in delineating the anatomy of QAV and for assessment of severity of AR.

We present a case of QAV mimicking a bicuspid aortic valve.

CASE PRESENTATION

A 60-year-old man with a medical history of hypertension, asthma, a heart murmur for 3 to 5 years, and polysubstance abuse presented to the emergency department with complaints of worsening shortness of breath on exertion over the past 2 to 3 weeks. A review of symptoms was negative for chest pain, palpitations, lightheadedness, or lower extremity edema. Significant physical examination findings included wide pulse pressure (blood pressure 157/56 mm Hg), tachypnea, jugular venous distension (3 cm above the clavicle), a decrescendo early diastolic blowing murmur, and a soft crescendo-decrescendo systolic murmur best heard in the aortic region; there was no peripheral edema. Laboratory studies were significant for elevations in B-type natriuretic peptide of 1,541 pg/mL and troponin of 0.06 ng/mL. An electrocardiogram demonstrated sinus rhythm with left ventricular hypertrophy and biatrial enlargement. The patient was started on medical therapy to optimize blood pressure.

From the Division of Cardiology (E.V., R.P., Y.K., A.H.W.) and Department of Medicine (E.V., R.P., Y.K., A.H.W.), Newark Beth Israel Medical Center, Newark, New Jersey; Department of Medicine, Rutgers-Saint Barnabas Medical Center (K.P.K.), Livingston, New Jersey; and Department of Medicine and Radiology, Rutgers New Jersey Medical School (A.H.W.), Newark, New Jersey.

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Correspondence: Esad Vucic, MD, PhD, 201 Lyons Avenue, Newark, New Jersey 07112. (E-mail: *esad.vucic@rwjbh.org*).

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VIDEO HIGHLIGHTS

Video 1: Three-dimensional TEE, midesophageal short-axis aortic view (50°) of the aortic valve demonstrates the quadricuspid valve, which is functionally bicuspid.

Video 2: Two-dimensional TEE, midesophageal short-axis view (50°) with color Doppler demonstrates the QAV with 4 almost equal-sized cusps. The valve is functionally bicuspid with a raphe between the NCC and RCC as well as between the LCC and the supernumerary cusp. There is moderate central AR.

Video 3: Two-dimensional TEE, midesophageal short-axis view (52°) of the aortic valve demonstrates the functionally bicuspid QAV with 4 almost equal-sized cusps and 2 distinct raphes.

Video 4: Two-dimensional TEE, midesophageal long-axis view (137°) with color Doppler demonstrates slightly eccentric AR. The vena contracta width was measured at 0.4 cm.

Video 5: Two-dimensional TEE, midesophageal long-axis view (119°) of the congenitally malformed aortic valve.

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A TTE demonstrated a moderately calcified aortic valve with moderate AR (vena contracta width, 0.4 cm). Left ventricular internal diameter during diastole and systole was noted to be 5.2 and 4.0 cm, respectively. Left ventricular ejection fraction was 47%. A TEE was then performed to further evaluate the mechanism and severity of AR. The TEE demonstrated an underdeveloped QAV that was functionally bicuspid with a raphe between the right and noncoronary cusps and a raphe between the left and supernumerary coronary cusps (Figures 1 and 2, Videos 1-3). Moderate AR (Figure 3, Videos 2, 4, and 5) was noted with a vena contracta width of 0.4 cm (Figure 3). The aortic valve area by two-dimensional planimetry was 1.9 cm^2. The aortic root and ascending aorta were normal in size, measuring 3.6 and 3.4 cm, respectively. A cardiothoracic surgeon was consulted for aortic valve surgery, and the patient was referred to addiction medicine for rehabilitation before surgery. Surgical results were not available at the time of this case submission.

DISCUSSION

A QAV is a rare congenital defect. Based on the anatomical variations of the cusp size, Hurwitz *et al*[4] identified 7 subtypes among QAV with 4 equal cusps (type A) and 3 equal cusps and 1 smaller cusp (type B), accounting for 76% of cases. Nakamura *et al*[5] described a simplified

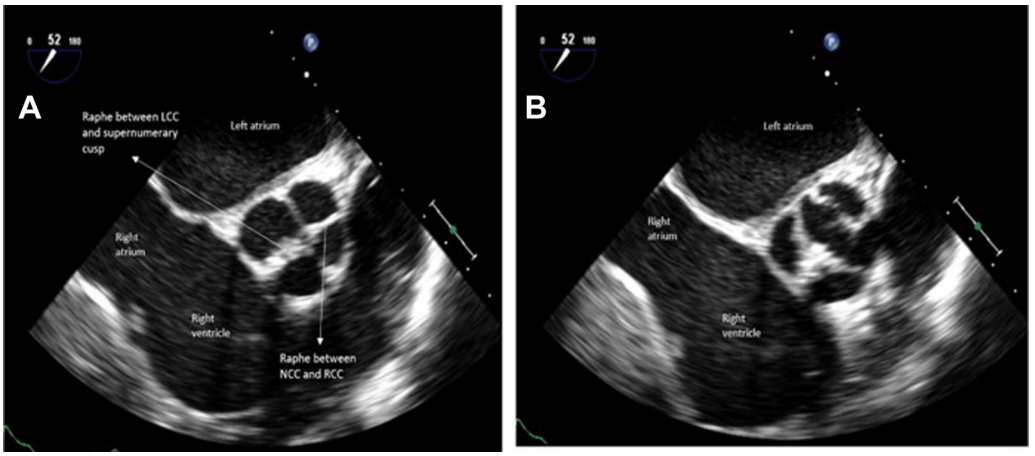

Figure 1 Two-dimensional TEE, midesophageal short-axis view (52°) of the aortic valve. **(A)** Quadricuspid aortic valve in diastole demonstrating raphe between noncoronary cusp (NCC) and right coronary cusp (RCC) and a raphe between left coronary cusp (LCC) and supernumerary cusp. **(B)** Quadricuspid aortic valve that is functionally bicuspid in systole.

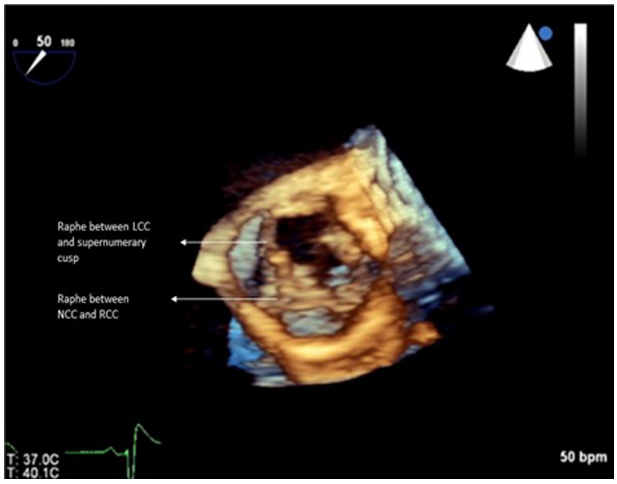

Figure 2 Three-dimensional TEE, midesophageal short-axis view (50°) of the aortic valve, systolic phase, demonstrates the QAV with raphe between noncoronary cusp (NCC) and right coronary cusp (RCC) and a raphe between left coronary cusp (LCC) and supernumerary cusp.

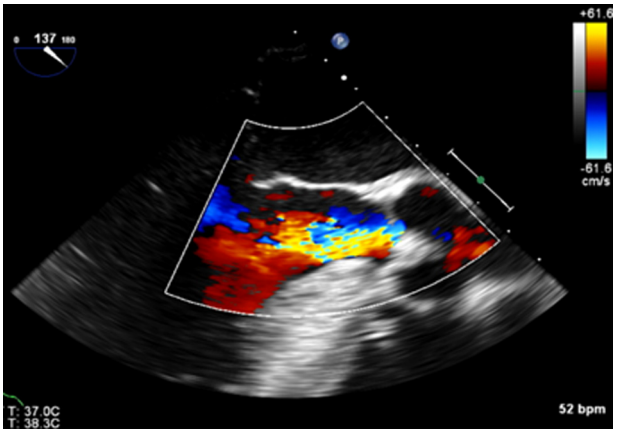

Figure 3 Two-dimensional TEE, midesophageal long-axis view (137°) with color Doppler, diastolic phase, demonstrating moderate AR. The vena contracta width was measured at 0.4 cm.

classification of QAV into 4 anatomical types based on the location of the supernumerary cusp.

To understand the QAV, it is necessary to understand the embryological development of aortic valves. Embryologically, semilunar valves typically form from the conotruncal and intercalated endocardial cushions of the outflow tract, and formation is completed by 9 weeks.[6] Left and right coronary cusps of the aortic valve are formed from the conotruncal cushion. In contrast, right-posterior and left-anterior intercalated cushion gives rise to the noncoronary cusp of the aortic valves.[6] Any abnormality in the development and fusion of the cushion leads to congenital anomalies such as a QAV. However, the exact mechanism leading to the formation of a QAV is unknown. A QAV is likely due to either aberrant fusion of the aorticopulmonary septum or abnormal proliferations in the common trunk.[4,7,8] Furthermore, embryologic development of the aortic valve is very closely related to the development of the coronaries as the coronary artery develops before the formation of the valve. Hence, although most frequently QAV is an isolated congenital anomaly, the most common associated disorders include coronary artery and coronary ostium anomalies.[4,7] This includes saccular aneurysm of the noncoronary sinus and a coronary ostium, abnormal takeoff of the right coronary artery with an anomalous coronary artery near the left ostium, and displaced right coronary orifice.[7] These associated anomalies are significantly important when these patients undergo aortic valve replacement to prevent ostial obstruction of the coronary arteries.

Our patient under discussion has a type A QAV based on the Hurwitz and Roberts classification (4 equal-sized cusps) and type 1 QAV based on the Nakamura classification (supernumerary valve between the left coronary and right coronary cusps). The aortic valve has 2 raphes, which are fused tangentially between the right and noncoronary cusps and between the left and supernumerary cusps,

leading to the valve being functionally bicuspid. Transthoracic echocardiography is useful in identifying a QAV. Unlike the diagnosis of a bicuspid aortic valve, a QAV is identified during diastole rather than systole. From the parasternal short-axis view, during diastole the aortic commissures of a QAV form a classic *X* shape as opposed to the *Y* shape seen with the trileaflet valves.[9] Transesophageal echocardiography is also an accurate imaging tool to confirm the diagnosis of a QAV especially if the TTE is inconclusive. Transesophageal echocardiography displays the 4 cusps, identifies the type of QAV, and may be able to visualize possible displacement of the coronary ostia.[8] In a database search of 186 cases of QAV, isolated AR was found in almost 75% of cases, AR with stenosis in just over 8%, isolated stenosis is less than 1%, and normal function in about 16% of cases.[10,11] Most commonly, patients present in the fifth to sixth decade of life with heart failure. The most common indication for surgical intervention is severe AR.[7] The finding of an anatomic QAV functioning as a bicuspid aortic valve is exceptionally rare, and the implications of this unusual anatomy are unknown. This case report also highlights the utility of TEE in delineating the underlying anatomy and mechanism of the AR.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for experiments involving humans.

CONSENT STATEMENT

The authors declare that since this was a non-interventional, retrospective, observational study utilizing de-identified data, informed consent was not required from the patient under an IRB exemption status.

FUNDING STATEMENT

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DISCLOSURE STATEMENT

The authors report no conflict of interest.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2022.09.007.

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