

Quadricuspid Aortic Valve May Be Misdiagnosed as Bicuspid Aortic Valve

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Abstract

Quadricuspid aortic valve (QAV) is a rare congenital cardiac anomaly with incidence of 0.008-0.033%. The most common complication of QAV is aortic regurgitation (75%). Other complications include aortic stenosis, left ventricular hypertrophy, bundle branch blocks, and abnormal displacement of the ostium in the right coronary artery. It is vital that patients with QAV who present with progressive aortic regurgitation undergo valve replacement or repair within an appropriate amount of time. This case report focuses on the presentation of a 20-year-old man who was easily overlooked with transthoracic echocardiography (TTE) during childhood when he was diagnosed with aortic stenosis secondary to bicuspid aortic valve. The patient presented to our cardiology clinic for evaluation of a new diastolic murmur. TTE revealed QAV, moderate aortic insufficiency, and mild aortic stenosis.

Keywords: Quadricuspid aortic valve; Aortic regurgitation; Aortic stenosis; Transthoracic echocardiogram

Introduction

A quadricuspid aortic valve (QAV) is an uncommon congenital heart defect characterized by the presence of four cusps, instead of the usual three found normally in the aortic valve [1]. It is a defect that occurs during embryological development of the aortic trunk during gestation [2]. The incidence of 0.008-0.033% is nearly equal in men and women [1]. Previously, this congenital defect was diagnosed at the time of surgery or at postmortem examination. With advances in echocardiography, more cases are being diagnosed before surgery. However,

few cases were misdiagnosed as bicuspid aortic valve despite the use of echocardiography [3]. Echocardiography series incidence rates range from 0.013% to 0.043% [4]. There is an increased risk of developing post-natal aortic regurgitation and other heart-related diseases; therefore, patients with this condition should be carefully monitored [5]. More than half of these patients need valve surgery in adulthood mainly due to progressive aortic regurgitation. Here, we present a 20-year-old man with systolic murmur since childhood who was told by his pediatrician that he has a bicuspid aortic valve.

Case Report

A 20-year-old man who presented with a prior diagnosis of bicuspid aortic valve, which was diagnosed at another medical center during childhood, presented to our cardiology clinic for evaluation of a new diastolic murmur. He had no family history of coronary artery disease, sudden death, or known valvular abnormalities. The patient was asymptomatic. On physical examination, his systemic blood pressure was 142/68 mm Hg, and his pulse was 90 beats/min. His carotid and peripheral pulses were found to be normal. A 2/6 diastolic decrescendo murmur was audible over the left sternal border, also a grade-2 systolic ejection murmur was audible in the second right sternal border. Chest X-ray was normal and the electrocardiogram displayed normal sinus rhythm.

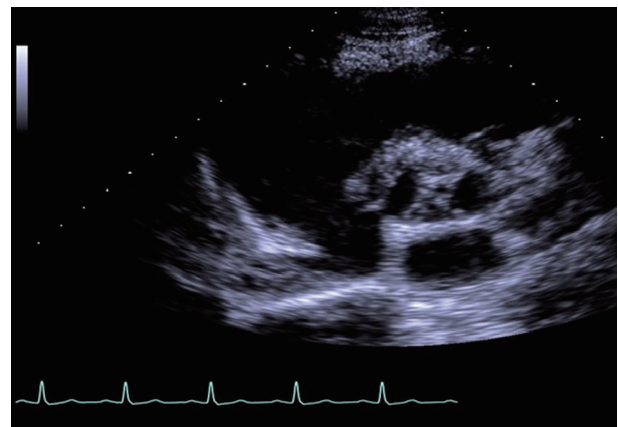


Figure 1. Short axis view of aorta and aortic valve with four leaflets.

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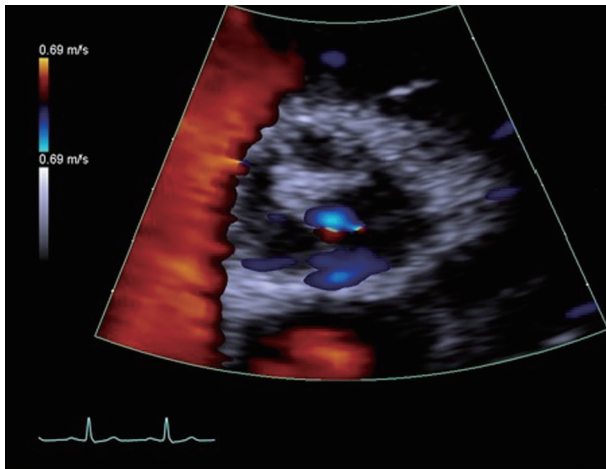


Figure 2. Short axis view of aortic valve with color Doppler - aortic regurgitation.

Electrocardiogram showed normal sinus rhythm with sinus arrhythmia, rightward axis, and minimal voltage criteria for left ventricle hypertrophy that may be normal variant. Transthoracic echocardiography (TTE) revealed left ventricle ejection fraction 55-60% with normal diastolic function and cavity size. On the short axis, a QAV was found with four unequal cusps (Fig. 1, 2). There was moderate aortic insufficiency (vena contracta 0.5 cm) (Fig. 3) with mild aortic stenosis (maximum velocity 2.6 m/s, mean gradient 14 mm Hg). The diagnosis was explained in detail to the patient, and he was asked to follow up with cardiology for a clinical re-evaluation and TTE every 12 months.

Discussion

QAV is a rare congenital malformation and the first case of the QAV was reported by Balington in 1862 [2]. Embryologically, the aortic valve is formed when the truncus arteriosus separates into aortic and pulmonary trunks. Within the walls of the aortic and pulmonic trunks, three pads of mesenchymal tissue develop inward to form the primordia of the semilunar cusps. Abnormal cusp formation results from either aberrant fusion of the aorticopulmonary septum or from abnormal mesenchymal proliferation in the common trunk resulting in abnormal aortic valve cusps [6].

On the basis of the variation in size of the aortic cusps, QAV has been classified into seven types (lettered A through G), which help in predicting the likely outcome, treatment planning, and are important in identifying the different types of juxtaposition of the valves. The majority of patients with QAV are classified as one of the first three types: type A shows four equal cusps; type B shows three equal cusps and one smaller cusp; type C shows two equal larger cusps and two equal smaller cusps. The remaining classes are significantly less common: type D shows one large, two intermediate, and one small cusp; type E shows three equal cusps and one larger cusp; type F shows two equal larger cusps and two unequal smaller cusps; type G shows four unequal cusps [7]. Based on

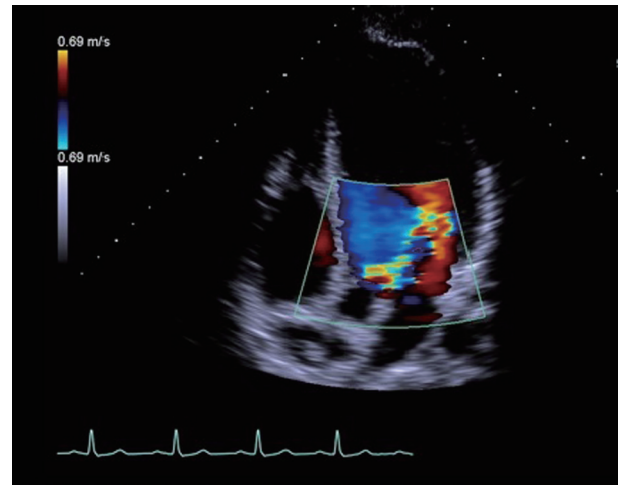


Figure 3. Five chambers view showing aortic regurgitation.

this classification system, our patient would be considered as having QAV type G.

QAV usually appears as an isolated congenital anomaly, but may also be associated with other malformations, including aortic regurgitation (75%), aortic stenosis (18%), coronary artery disease anomaly, atrial septal defect, pulmonary valve stenosis, hypertrophic cardiomyopathy, ventricular septal defect, and subaortic stenosis [2]. Infective endocarditis in patients with QAV has been reported in several cases [8-10].

With ongoing development and reduced costs in echocardiography and imaging, including better training of technicians, TTE is increasingly used in common practice. This has resulted in easier and more frequent diagnosis of QAV. It is to be noted that transesophageal echocardiography (TEE) is more sensitive and specific than TTE [11]. On echocardiography, QAV is identified by its characteristic "X" configuration during diastole, and appears rectangular during systole [12]. Recent case reports have been published of cardiac magnetic resonance imaging and multidetector CT imaging, both with excellent image quality, to diagnose QAV [13]. However, these modalities are undoubtedly more expensive and relatively more cumbersome than TTE or TEE, and focus should be placed on improving proper diagnosis via echocardiography to avoid those potential hurdles. Our patient was misdiagnosed as having a bicuspid aortic valve by TTE during childhood, similar to other cases of QAV reported previously [3, 14]. Given this, we would infer that a QAV could potentially be misdiagnosed as a bicuspid aortic valve if diagnosis is given based on a single view or snapshot of the valve. Obtaining multiple images from varying viewpoints might serve to decrease the likelihood of misdiagnosing quadricuspid valves as bicuspid.

The surgical indication of QAV patients depends on the severity of aortic regurgitation and its associated complications. A majority of patients with QAV potentially require surgery due to severe aortic regurgitation. This is especially true in the geriatric population secondary to progressive valvular deterioration [2]. The indication for surgery remains the same regardless of the etiology of the aortic regurgitation [13]. However, surgeons should pay attention to the origin of the coronary ar-

tery, and avoid injury when operating. As aortic valve repair is still a big challenge for cardiac surgeons, valve replacement was the most widely performed operation in such patients [15]. Some surgeons have tried tri-cuspidized or bi-cuspidized repair techniques and obtained good short-term results. Long-term follow-up has been shown to help avoid complications associated with valve replacement [16]. In fact, close follow-up is also beneficial in patients who are not surgical candidates (e.g. QAV with mild aortic regurgitation) [17].

Conclusion

QAV is an unusual congenital anomaly which was unintentionally found in surgery or autopsy in previous years. Aortic insufficiency is the most common abnormality associated with QAV. Nowadays, in tandem with advances in echocardiographic imaging and technique may reveal more cases of QAV and alter the incidence rate. Physicians should be aware of this rare congenital anomaly since more than half these patients will need either valve repair or replacement due to progressive aortic regurgitation.

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