Case Report

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Quadricuspid aortic valve in an asymptomatic young adult: a case report

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Introduction: Quadricuspid aortic valve (QAV) is a rare congenital heart disease, ranking behind bicuspid and unicuspid aortic valves in terms of the incidence of congenital aortic valve abnormalities. When symptoms are present, they are typically related to aortic regurgitation, manifesting as shortness of breath, nocturnal dyspnea, and palpitations, or aortic stenosis, which presents with exertional dyspnea, angina, or syncope.

Case presentation: We present the case of an asymptomatic male, diagnosed with a QAV during a routine examination. Despite the absence of clinical symptoms, a thorough general physical examination, including cardiac auscultation, revealed signs suggestive of valvular abnormality. The diagnosis was confirmed via transthoracic echocardiography.

Clinical discussion: For QAV, the diagnostic process typically includes transthoracic echocardiography, which allows for initial confirmation of the valve morphology. In addition, computed tomography (CT) coronary angiography provides detailed information on valvular morphology and helps identify any associated coronary stenosis. In cases of QAV, a CT aortogram is also crucial to assess potential aortic root dilation, a known complication of this congenital anomaly.

Conclusion: This case emphasizes the importance of routine physical examination in diagnosing rare congenital heart conditions like QAV, even in asymptomatic individuals. Early diagnosis and appropriate imaging are essential for timely management, particularly in preventing the progression of associated complications such as aortic regurgitation or root dilation.

Keywords: aortic regurgitation, computed tomography, congenital aortic valve abnormalities, echocardiography, quadricuspid aortic valve

Introduction

Quadricuspid aortic valve (QAV) is a rare congenital heart disease. The most frequently encountered aortic valve malformations include the bicuspid aortic valve and unicuspid aortic valve, followed by QAV. The overall incidence of QAV is 0.008% by autopsy; however, with the increasing use of two-dimensional transthoracic echocardiography (TTE), its incidence has risen to 0.043%[1]. The mean age at diagnosis is 43.5 ± 21.8 years[2]. The first case of QAV was described in 1862 when it was incidentally discovered during an autopsy by Balington[3].

Here we present a case of a 38-year-old male who was found to have QAV during a routine check-up, highlighting the importance of routine physical examination, especially cardiac auscultation – a clinical art that is increasingly being replaced by modern investigations, yet remains crucial for diagnosis. The case has been reported in line with the CARE 2017 guidelines[4].

HIGHLIGHTS

- Quadricuspid aortic valve (QAV) is a rare congenital anomaly of aortic valve.
- Thorough physical examination is crucial for diagnosis.
- Associated findings included aortic regurgitation, stenosis, and dilatation. Echocardiography remains a valuable initial diagnostic tool.
- The article also provides insights into the role of cardiac imaging modalities in QAV.

Case report

A 38-year-old regular military personnel member with no known comorbidities, a non-smoker, and a non-alcoholic, underwent a mandatory pre-deployment physical examination for his upcoming United Nations (UN) mission two years ago. He had no history of shortness of breath, chest pain, palpitations, or syncope. Additionally, there was no history of joint pain, back pain, redness of eyes, or grittiness. He also denied any unsafe sexual practices. Family history was unremarkable, and he had not undergone any previous interventions.

On general physical examination, he was of average build with no pallor, icterus, clubbing, cyanosis, or edema. On head-to-toe examination, there was no marfanoid habitus, and peripheral signs of aortic regurgitation (AR) were not appreciable. His pulse was regular at 64 beats per minute, with normal volume and character, and all peripheral pulses were palpable. His blood

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pressure (BP) was 150/70 mmHg in both arms, while BP in the bilateral thighs measured 170/80 mmHg. Chest examination was unremarkable. On cardiovascular examination, the apical impulse and beat were normal. On auscultation, an early diastolic murmur of grade 2/6 was heard over the neo-aortic area, conducted down to the left sternal border, and best heard in the sitting position, leaning forward with breath held in expiration. The murmur was augmented with squatting and isometric handgrip. Additionally, a pan-systolic murmur of grade 2/6 was heard over the left lower parasternal border, best heard on inspiration with no radiation. There was no palpable P2, left parasternal heave, or epigastric pulsations. Other systemic examinations were unremarkable. The patient was provisionally diagnosed with primary valvular heart disease with AR, tricuspid regurgitation (TR), and isolated systolic hypertension.

Routine investigations, including a full blood count, and renal/liver function tests were normal. The Venereal Disease Research Laboratory (VDRL) test was negative. A 12-lead electrocardiography (ECG; Fig. 1) showed sinus rhythm with left ventricular hypertrophy as per Sokolov-Lyon voltage criteria[5]. A chest radiograph was unremarkable. Transthoracic echocardiography (TTE; Fig. 2) revealed concentric left ventricular hypertrophy, a dilated ascending aorta, and a QAV. Color Doppler imaging showed moderate AR, mild aortic stenosis (AS), and mild TR. Continuous wave Doppler showed a normal left ventricular outflow gradient. No intracardiac shunts were detected. The ejection fraction was 65%. Computed tomography (CT) coronary angiography (Fig. 3) visualized the four cusps of the aortic valve, among which one was relatively larger than the other three, consistent with type E of the Hurwitz and Roberts classification. There was no evidence of coronary anomalies or stenosis. A CT aortogram (Fig. 4a and b) revealed a dilated tubular ascending aorta with a maximum transverse diameter of 5.1 cm. Considering the physical examination and investigation findings, a final diagnosis of QAV with moderate AR, mild AS, mild TR, ascending aortic fusiform aneurysm, and isolated systolic hypertension was made.

The patient was initiated on an angiotensin receptor blocker (ARB) in accordance with the 2020 American College of Cardiology/American Heart Association (ACC/AHA) guideline, which recommends ARBs or angiotensin-converting enzyme (ACE) inhibitors as the preferred treatment for hypertension, particularly when the systolic blood pressure exceeds 140 mmHg in the context of AR[6]. He was followed up every six months with TTE and annually with a CT aortogram. His two-year follow-up did not show any differences compared to his previous findings. Additionally, he had not developed any symptoms during his follow-up. Surgery was not recommended, as the patient was asymptomatic and did not meet the surgical criteria for either aortic regurgitation/stenosis or ascending aortic aneurysm.

Discussion

The precise mechanisms underlying the development of a QAV remain unclear. It has been hypothesized that the anomaly may result from an aberrant septation of the conotruncus or an abnormal division of one of the endocardial cushions, potentially triggered by an inflammatory episode. Alternatively, abnormal cusp formation could stem from a faulty fusion of the aorticopulmonary septum or excessive mesenchymal proliferation within the truncus arteriosus during embryogenesis[7]. QAV can be categorized into different types according to the size and shape of its cusps, as outlined by Hurwitz and Roberts. The primary types are: Type A, featuring four cusps of equal size; Type B, consisting of three cusps of equal size and one smaller cusp; Type C, with two larger cusps of equal size and two smaller cusps of equal size; and Type D, characterized by one larger cusp and three smaller cusps. Additionally, there are rarer types, such as Types E, F, and G. In Type E, the valve has three equal-sized cusps and one larger cusp. Type F consists of two equal-sized larger cusps and two unequal-sized smaller cusps. Type G is characterized by four cusps of varying, unequal sizes[8].

QAV typically presents as an isolated defect, although coronary artery anomalies are observed in approximately 10% of cases. This association may be explained by the embryological timing, as the development of the aortic valve leaflets occurs shortly after the formation of the coronary artery from the sinuses of Valsalva[9]. A case of sudden death in a 16-year-old teenager while walking to school has been reported, where the cause of death, discovered during necropsy, was the complete isolation of the orifice of the left coronary artery by the adherent aortic valve

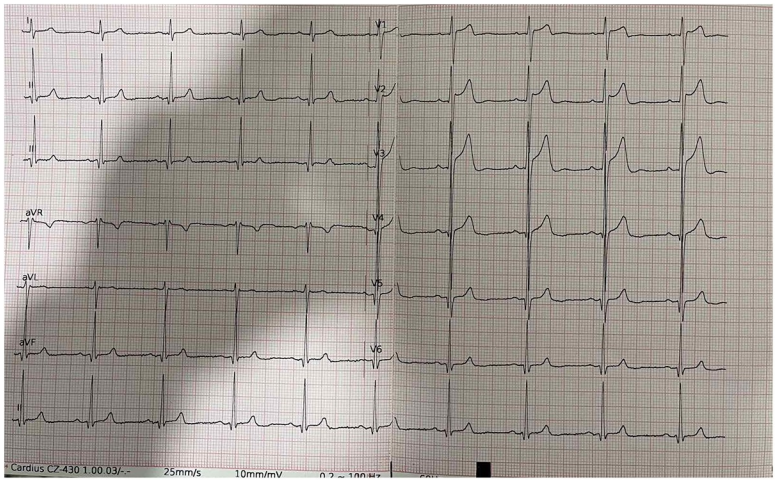

Figure 1. Electrocardiogram (ECG) showing left ventricular hypertrophy by voltage criteria.

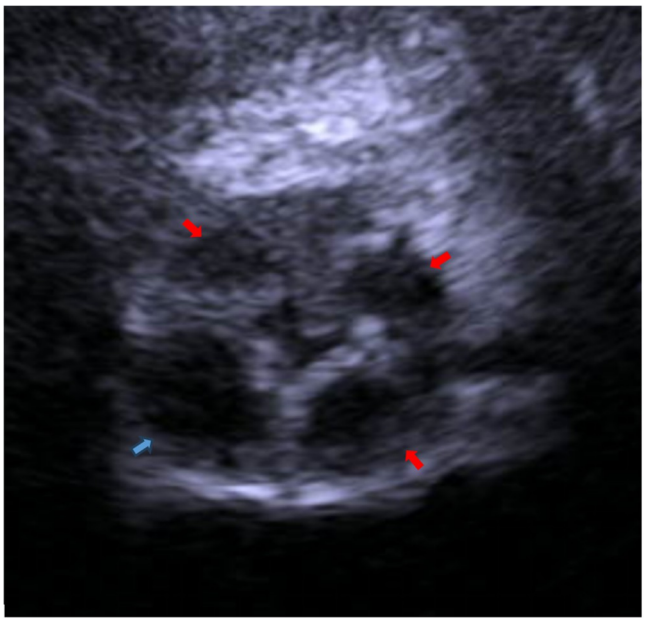

Figure 2. Transthoracic echocardiography (TTE) transverse scan image showing the four cusps of the aortic valve. Note that the aortic cusps are of different sizes, with one larger (blue arrow) than the other three cusps (red arrow), which are of similar size.

cusp[10]. Other associated anomalies include interatrial septal defects, interventricular septal defects, partial atrioventricular canal, subaortic stenosis, patent ductus arteriosus, Fallot's tetralogy, ascending aortic aneurysm, and non-obstructive hypertrophic cardiomyopathy[11]. Aortic dilatation and moderate to severe AR are present in 29% and 26% of cases, respectively. AS is less common than AR, accounting for up to 8% of cases, and is usually of mild severity[2]. Our patient presented with moderate AR and mild AS, though the most common clinical manifestation of a QAV is AR without AS. He also had an aneurysmal dilatation

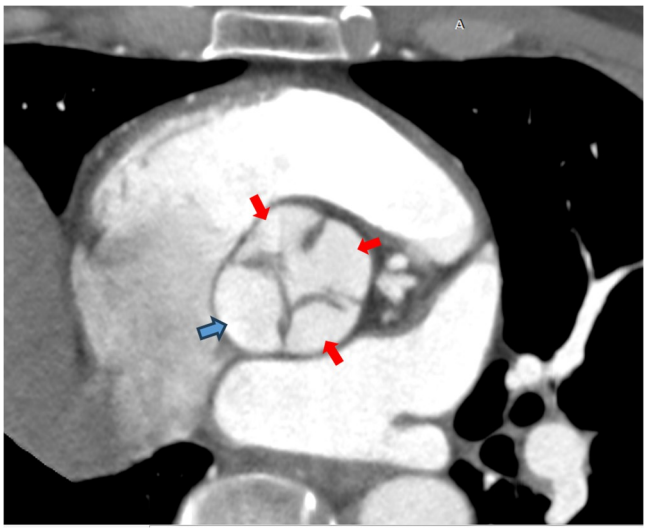

Figure 3. Computed tomography (CT) coronary angiography axial image showing a quadricuspid aortic valve. Note that the aortic cusps are of different sizes, with one larger (blue arrow) than the other three cusps (red arrow), which are of similar size.

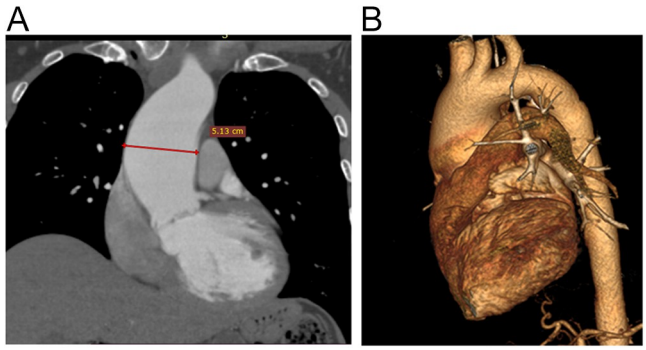

Figure 4. (A) Computed tomography (CT) aortogram coronal image showing a dilated tubular ascending aorta. (B) CT reformatted coronal image showing the dilated tubular ascending aorta.

of the ascending aorta. An ascending aorta diameter of ≥4.0 cm is considered dilated (ectasia), while a diameter of ≥4.5 cm is classified as an aneurysm[12]. Mild TR was also found in our case. Although QAV primarily affects the aortic valve, severe left heart dysfunction can indirectly cause secondary TR, while direct structural abnormalities of the tricuspid valve are not typically associated with QAV[13]. Given the patient's normal left ventricular systolic function, the TR is unlikely to be related to the QAV. A potential explanation could be valvular regurgitation induced by physical exertion, considering the patient's role as an active-duty army member[14]. No coronary artery anomalies were observed. The observed systolic hypertension, in this case, can be attributed to the increased workload on the heart as it compensates for the regurgitant volume. While chronic hypertension is a recognized cause of aortic root dilation and AR, it is relatively uncommon in hypertensive individuals under the age of 60[15].

In early life, particularly in individuals younger than 18 years, cases of QAV are typically asymptomatic and often identified incidentally through echocardiography or during autopsy. Symptoms such as dyspnea, fatigue, and palpitations generally manifest later in life, typically after the fourth decade, as valvular dysfunction progresses[7]. Our patient was asymptomatic, likely due to being in his late thirties. Progressive development of aortic regurgitation often necessitates surgical intervention by the age of 50 years[16]. However, patients can remain asymptomatic until the sixth decade of life, particularly if the valvular dysfunction is mild or progresses slowly[3]. Conversely, there is also evidence of significant aortic regurgitation requiring valvular repair as early as 5 years of age, highlighting the variability in the clinical presentation and progression of QAV[17]. Advanced cases, particularly those left untreated, can progress to heart failure, significantly impacting the patient's quality of life. The chronic volume overload from severe AR and left ventricular dysfunction contribute to this deterioration.

Asymptomatic cases of QAV with mild dysfunction should be kept under regular follow-up with echocardiography to assess valve and left ventricular function. Surgical indications for QAV include severe AR, severe AS, or a dysfunctional QAV in combination with other conditions, such as the occlusion of the left coronary ostium[7]. For patients with AR and relatively intact cusps, repair may be considered, involving either tricuspidalization or bicuspidalization, depending on the condition of the leaflets and the surgeon's preference. However, the majority will require valve replacement[18]. Regarding the ascending

aortic dilatation, surgery is recommended if the patient develops symptoms related to the condition, or for asymptomatic individuals if the aortic dilation reaches 5.5 cm or more. For individuals with a family history of dissection, surgery is also advised if the aneurysm is 5.0 cm or larger. Additionally, surgery may be considered if the aneurysm shows rapid growth, defined as an increase of more than 0.3 cm per year over two consecutive years or more than 0.5 cm in a single year[19].

Detailed history and physical examination are essential for identifying cases like these. Additionally, a focused laboratory examination, including regular follow-up, holds clinical significance for further management and interventions. Moreover, clinicians should always consider the possibility of QAV when AR or AS is revealed on TTE, as bicuspid and unicuspid aortic valves are not the only congenital aortic valve abnormalities and there are instances when the QAV is missed even in cases of documented AR[20].

Conclusion

This case highlights the physical examination findings and possible associations of QAV. The article also provides insights into the role of cardiac imaging modalities in the diagnosis and follow-up of QAV.

Ethical approval

Not applicable.

Consent

Not applicable.

Sources of funding

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author's contribution

P.K.: Contributed in conceptualization, manuscript writing, mentor, and editing; K.C.S.: Conceptualization, manuscript writing, literature review and editing; A.B.: Contributed in literature review; A.K.: Contributed in literature review and editing; all authors have read and approved the manuscript.

Conflicts of interest disclosure

All the authors declare to have no conflicts of interest relevant to this study.

Research registration unique identifying number (UIN)

Not applicable.

Guarantor

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Provenance and peer review

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Data availability statement

It will be open access and publicly available as per the journal guideline.

Patient perspective

I am grateful to my physician for identifying my cardiac condition. I will be under regular follow-up, as I may require surgical intervention soon. However, I am saddened that I won't be able to join the UN mission.

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