Quadricuspid aortic valve – A case report and literature review

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Abstract Quadricuspid aortic valve is a rare congenital cardiac entity. The recognition of quadricuspid aortic valve has clinical significance as it leads to aortic valve dysfunction in majority, commonly aortic regurgitation, severe enough for surgical correction. Preoperative diagnosis is essential as it is often associated with other congenital cardiac abnormalities including abnormally located coronary ostia. We report a case of quadricuspid aortic valve in a forty-five year old asymptomatic male and review the available literature.

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1. Introduction

Quadricuspid aortic valve is a rare variant of abnormal development of semilunar valves. The reported incidence is 0.008% in autopsy series, 0.043% by two dimensional transthoracic echocardiography and 1% of those posted for aortic valve surgery; however, being a rare anomaly the actual incidence is underreported. With advances in cardiac imaging, more cases are being reported. It frequently appears as an isolated congenital anomaly, but may also be associated with other malformations, especially with coronary anomalies.

Clear delineation of the anatomy of the quadricuspid aortic valve is difficult but of great clinical importance as these patients may present with progressive valvular aortic regurgitation (AR) and other cardiovascular abnormalities. We report a case of quadricuspid aortic valve in an asymptomatic patient who presented with a cardiac murmur on routine examination.

2. Case report

A forty-five year old asymptomatic male patient was referred from a military hospital for annual medical evaluation. Physical examination revealed regular heart beats (74 beats/min) and had a blood pressure of 110/80 mmHg. His carotid and peripheral pulses were normal. A grade 2/6 early diastolic murmur along the left sternal border without any radiation was heard. Physical examination of the other systems was unremarkable. The 12-lead electrocardiography was in sinus rhythm and normal. Chest radiograph was normal. Transthoracic echocardiography showed normal left ventricular dimensions, systolic functions and aortic valve opening. Color Doppler imaging demonstrated mild aortic regurgitation. On the parasternal long axis view no apparent aortic valve leaflet abnormality was seen; however, the short-axis view showed four aortic valve cusps of equal size in both systole and diastole. At end diastole, the commissural lines formed by the adjacent cusps resulted in an “X” configuration instead of the normal “Y” configuration (Fig. 1). In addition, color Doppler scans demonstrated incomplete coaptation of the aortic valve leaflets, resulting in a regurgitant orifice located in the center of the aortic valve. The patient underwent assessment by computed tomographic (CT) angiography and cardiac magnetic resonance imaging for possible aortic coarctation and coronary anomalies. No aortic coarctation or any other coronary abnormality was revealed; however, type A quadricuspid aortic valve was confirmed (Fig. 1).

At this stage no active treatment was indicated but regular follow-up was recommended.

2.1. Incidence

The bicuspid aortic valve is the most common aortic valve abnormality, occurring in approximately 2% of the population followed by unicuspid aortic valve. Quadricuspid aortic valve is a rare congenital cardiac disorder, with a reported incidence between 0.008% by autopsy to 0.043% by two dimensional echocardiography.
Quadricuspid aortic valve

2.2. Embryology

At the fifth week of normal embryogenesis, two mesenchymal ridges form in the cephalad portion of the truncus arteriosus. These truncoconal ridges fuse and descend in a spiral fashion into the ventricles to form the aorticopulmonary septum. At the junction of the conus and truncus, three mesenchymal swellings give rise to each semilunar valve. These swellings grow to form triangular-shaped valves which, with their covering endothelium become excavated on their distal aspect to form the cusps. The mechanism leading to the development of this congenital malformation is not well understood. Aberrant fusion of the aorticopulmonary septum or abnormal mesenchymal proliferation in the common trunk may lead to abnormal cusp formation. Quadricuspid aortic valve may represent a supernumerary variant of a primary aortic mesenchymal bud or an abnormal proliferation of the cusps. Other studies in Syrian hamsters have suggested that quadricuspid aortic valve may result from the division of one of the three mesenchymal ridges that normally give rise to three aortic valve cushions.

2.3. Anatomic variations

On the basis of the variation in size of the aortic cusps, quadricuspid aortic valve has been classified into seven types by Hurwitz and Roberts (Fig. 2):

- a. All four cusps of equal size;
- b. Three cusps with equal size, the fourth cusp smaller in size (most common type);
- c. Two larger cusps and two smaller cusps;
- d. One larger cusp, two cusps of intermediate size and one smaller cusp;
- e. Three cusps with equal size and one of bigger size;
- f. Two cusps of equal size and two cusps with unequal, smaller size; and
- g. All four unequal sizes (the less frequent variant).

Of the ninety-seven cases in which comment on the size of the various leaflets has been made by Hurwitz and Roberts, the distribution of types is as in Table 1.

A possible new type of quadricuspid aortic valve, type H, which comprises 2 equally sized smaller cusps and 2 larger cusps has been described lately. Recently simplified classification of quadricuspid aortic valve (type I to type IV) was published. Type I and type II are the same as the previously described types A and B. In our patient, the quadricuspid aortic valve appeared to be of type A with 4 equal-size cusps.

2.4. Functional expression

The exact natural history is not well established. In a literature review by Tutarel, aortic regurgitation was the most common pathology, where it was found in approximately 75% of the cases, and about 9% of the cases had both aortic stenosis and regurgitation, with only 16% having a normal functioning aortic valve. About 45% of the cases in the same series required surgery for significant symptoms and progressive valvular dysfunction. Another review of seventy cases by Janssens et al. detected thirty-nine valves (56%) with significant insufficiency, twenty-six of which required surgical replacement. The lowest incidence of abnormal valvular function is found in those with type B (67.5%). Aortic stenosis is very rare (0.7%), although in the series case described by Yotsumoto et al. the reported prevalence is between 7% and 12%. The quadricuspid aortic valve dysfunction is minimal or absent in children and adolescents.

2.5. Associated abnormalities

Quadricuspid aortic valve is associated with other congenital anomalies in tune of 18.3%. These congenital anomalies include coronary artery abnormalities, ventricular septal defect, patent ductus arteriosus, pulmonary stenosis, ruptured sinus of valsalva, complete heart block, and hypertrophic cardiomyopathy. Quadricuspid aortic valve has also been reported in association with Ehlers–Danlos syndrome. The association with anomalous coronary arteries (10% of cases), in particular single coronary artery, is especially important as there has been a reported sudden cardiac death caused by complete isolation of the left coronary artery by an adherent cusp of a quadricuspid aortic valve.

2.6. Diagnosis

Transthoracic two dimensional echocardiography provides information about the morphology of the aortic valve (number of cusps, degree of thickening, vegetations); aortic root size, left ventricular size, and color Doppler diagnose aortic regurgitation.

Three-dimensional transthoracic echocardiography is frequently used nowadays.

Sometimes, the quadricuspid aortic valve may not be discovered by transthoracic echocardiography and a transesophageal examination is necessary. It provides additional data regarding the morphology of the aortic valve (presence of prolapse) and measurement of several other parameters useful for valve surgery.

Another diagnostic method is aortography. The first case diagnosed at aortography in the left anterior oblique view was reported by Peretz et al. in 1969. It can describe the aortic cusps in terms of mobility, number, and calcification and assess the severity of the aortic regurgitation. CT angiography (CTA) is known to be highly accurate compared to other imaging techniques.

### Table 1 Quadricuspid valve types and function.

<table>
<thead>
<tr>
<th>Valve type</th>
<th>% of total</th>
<th>AR (% of valve type)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>32</td>
<td>77</td>
</tr>
<tr>
<td>B</td>
<td>41</td>
<td>60</td>
</tr>
<tr>
<td>C</td>
<td>15</td>
<td>71</td>
</tr>
<tr>
<td>D</td>
<td>03</td>
<td>100</td>
</tr>
<tr>
<td>E</td>
<td>02</td>
<td>0</td>
</tr>
<tr>
<td>F</td>
<td>02</td>
<td>100</td>
</tr>
<tr>
<td>G</td>
<td>05</td>
<td>80</td>
</tr>
</tbody>
</table>
to conventional coronary angiography and has been validated in the preoperative setting for chronic aortic regurgitation.\textsuperscript{20}

Cardiac magnetic resonance imaging is especially helpful in the evaluation and monitoring of this condition. It provides accurate and reproducible assessment of regurgitation severity and left ventricular size, the identification of aberrant coronary ostia, and the screening for associated anomalies, all in a single comprehensive investigation.\textsuperscript{20} It also helps in prognostication of patients with regurgitant aortic valve lesions: patients with a regurgitant fraction <26\% have a good prognosis and patients with a regurgitant fraction >33\% require valve replacement within a few years.\textsuperscript{27}

Often, the quadricuspid aortic valve is an intraoperative accidental discovery, in cardiac surgery, usually for severe aortic regurgitation. Yotsumoto et al.\textsuperscript{33} reported four cases diagnosed intraoperatively, from the series of nine cases with quadricuspid aortic valves.

More than two hundred cases of quadricuspid aortic valves have been published, most of them have been diagnosed by echocardiography (51\%), followed by surgery (22.6\%), autopsy (15.6\%), and aortography (6.5\%).\textsuperscript{9,28}

2.7. Management

Management of the quadricuspid aortic valve patients depends on the extent of aortic regurgitation and its associated lesions. For aortic regurgitation, the indication is same as regurgitation caused by other reasons, such as degenerative disease. When there is an indication for surgical intervention, the treatment of choice is aortic valve replacement. However, successful surgical repair has also been reported in several cases.\textsuperscript{29}

Iglesias et al.\textsuperscript{30} reported a case in which quadricuspid aortic valve was converted into a tricuspid valve by suturing the common commissure between the right coronary cusp and the supranumerary cusp. With the advances in percutaneous interventions, there has been a case report of stenotic quadricuspid aortic valve being successfully repaired with transcatheter aortic valve implantation.\textsuperscript{31} The average age reported for valve replacement is fifty-four years in the forty-six cases of quadricuspid aortic valves operated on. However, the youngest patient undergoing valvular repair for a quadricuspid valve with significant regurgitation was only five years old.\textsuperscript{32} It is very important for the surgeons to know the presence of quadricuspid aortic valve since there is a possibility of abnormally placed coronary ostia (specially the left) which may be damaged during aortic valve replacement.

2.8. The risk of infective endocarditis

Few cases of infective endocarditis associated with a quadricuspid aortic valve have been reported, but it is so far unclear whether quadricuspid aortic valve is more prone to infectious risk. In patients with four equally sized cusps the risk of bacterial endocarditis is lower because of the lack of asymmetry or flow disturbance. However, Matsuoka et al.\textsuperscript{33} presented a case of bacterial endocarditis in a 75 years old patient, diagnosed with quadricuspid aortic valve with equally sized cusps. In valves with unequal cusps, uneven distribution of stress and incomplete juxtaposition during diastole may lead to progressive aortic insufficiency and gradual deterioration over the years, and thus increasing the risk for endocarditis. Endocarditis prophylaxis is no longer recommended in the management of patients with quadricuspid aortic valve.\textsuperscript{28}

2.9. Conclusion

In conclusion, the incidence of quadricuspid aortic valve, a rare congenital cardiac disorder, is reported to be on an increase with the increasing use of echocardiography, computed tomography angiogram and cardiac magnetic resonance imaging. The great majority of patients have abnormal function and often require surgery for aortic valve dysfunction, most commonly for aortic regurgitation, usually in the fifth and sixth decades. As such, patients with quadricuspid aortic valve would require careful clinical evaluation and close follow-up to manage them appropriately.

Conflict of interests

None.

References


