A rare pediatric cardiac anomaly: Quadricuspid aortic valve with aortic regurgitation

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ABSTRACT

A quadricuspid aortic valve is rarely diagnosed in children, but it can be associated with significant aortic regurgitation. It is important for pediatric cardiologists to be aware of this pathologic entity. We present a nine-year-old male, diagnosed with a quadricuspid aortic valve and mild aortic regurgitation.

Keywords: Aortic valve, aortic regurgitation, quadricuspid

CASE REPORT

A 9-year-old male was referred to the cardiology clinic for evaluation of an asymptomatic diastolic heart murmur. On physical examination, a 2/4 early, medium-frequency, diastolic murmur was audible at the right mid-sternal border. The remainder of his physical examination was unremarkable and an electrocardiogram was normal. Transthoracic echocardiography showed a type B quadricuspid aortic valve (QAV) (three cusps of equal size and one smaller cusp) associated with mild aortic regurgitation by color Doppler imaging, likely from a coaptation defect [Figure 1 and Video 1].[1] The left ventricular end-diastolic dimension measured by M-mode was normal (4.58 cm, z-score + 0.3) with a shortening fraction of 31%. The remainder of the cardiac anatomy was normal, including the origins of the coronary arteries.

Two years later, the patient remained asymptomatic and the echocardiography showed stable, mild aortic regurgitation [Figure 2], now associated with mild left ventricular dilation [Figure 3]. By M-mode the left ventricular end-diastolic dimension measured 5.82 cm (z-score + 2.8), the end-systolic dimension was 3.7 cm (z-score + 1.9), and the shortening fraction was 35%.

DISCUSSION

A QAV is an extremely rare congenital heart malformation. Often found incidentally during an autopsy, the incidence is between 0.008 and 0.043%.[2,3] Diagnosis of a QAV during childhood is uncommon, as most are diagnosed in adulthood. The mean age of diagnosis is 50.7 years and there is a slight male predominance.[4]

Aortic regurgitation is the most prevalent hemodynamic abnormality associated with a QAV and appears in up to 75% of the patients.[4] This rarely develops in childhood and is thought to be a consequence of mechanical stress leading

Figure 1: Parasternal short axis view of the transthoracic echocardiogram showing two-dimensional and color Doppler diastolic images of a quadricuspid aortic valve with mild insufficiency, due to abnormal leaflet coaptation, in a nine-year-old male
to leaflet fibrosis and malcoaptation over time.\cite{1} Many require aortic valve replacement later in adulthood.\cite{4} Our patient exhibited mild aortic insufficiency due to abnormal leaflet coaptation at only nine years of age, which caused a diastolic murmur and led to his early diagnosis.

Unlike bicuspid aortic valve, aortic stenosis is rare among those with a QAV.\cite{4} Up to 18% of the patients will have other cardiac malformations, most common being a coronary artery anomaly.\cite{4} Atrial septal defect, pulmonary valve stenosis, hypertrophic cardiomyopathy, ventricular septal defect, and subaortic stenosis have also been described.\cite{4,5}

Hurwitz and Roberts classified QAV into seven types, A through G [Figure 4].\cite{1} Our patient had a type B valve with three cusps of equal size and one smaller cusp. Types A and B are the most common forms.\cite{3,5} Some have argued that type B valves are more likely to develop regurgitation because the single smaller cusp leads to unequal stress distribution, progressive trauma, and malcoaptation, with age.\cite{5} However, a recent review suggests that both forms are often regurgitant.\cite{5}

Although QAV is rare and uncommonly diagnosed in children, pediatric cardiologists should be familiar with this congenital malformation and its associated cardiac anomalies. Currently, our patient has mild aortic regurgitation with mild left ventricular dilation and preserved shortening. He will require a lifelong cardiology follow-up, with a possible need for intervention later in life.

REFERENCES

1. Hurwitz LE, Roberts WC. Quadricuspid semilunar valve. Am J Cardiol 1973;31:623-6.
2. Simonds JP. Congenital malformation of the aortic and pulmonary valves. Am J Med Sci 1923;166:584-95.
3. Feldman BJ, Khanderia BK, Warnes CA, Seward JB, Taylor CL, Tajik AJ. Incidence, description and functional assessment of isolated quadricuspid aortic valves. Am J Cardiol 1990;65:937-8.
4. Tutarel O. The quadricuspid aortic valve: A comprehensive review. J Heart Valve Dis 2004;13:534-7.
5. Timperley J, Milner R, Marshall AJ, Gilbert TJ. Quadricuspid aortic valves. Clin Cardiol 2002;25:548-52.

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An 18-month-old male child was referred to our institute with complaints of fast breathing and repeated episodes of lower respiratory tract infections requiring hospitalizations in the past. On examination, he was underweight with no evidence of cyanosis or clubbing. There was a pansystolic murmur of grade III/VI along the lower left sternal border. The arterial oxygen saturation was 99%. His chest X-ray showed levocardia, cardiothoracic ratio of 0.6 with a left ventricular type apex and plethoric pulmonary vasculature. The echocardiogram was suggestive of levocardia, situs solitus, and congenitally corrected transposition of the great arteries (CCTGA) with SLL loop. There was large subpulmonic ventricular septal defect with inlet extension and left to right shunting. There is no left ventricular outflow tract obstruction [Figure 1a]. In addition, an echogenic membrane was noted just above the left-sided atrioventricular valve (morphological tricuspid valve) and below the left atrial appendage in the modified apical four-chamber view. The valve and subvalvular apparatus were structurally normal with a valvular annulus of 15 mm (Z value = −1.12). The membrane was causing severe obstruction of right ventricular inflow with pressure gradients of 26/18 mm of Hg [Figure 1b and c]. Surgical correction with resection of membrane and closure of ventricular septal defect was advised but declined by the family.

**DISCUSSION**

CCTGA accounts for less than 1% of the cases of congenital heart diseases. CCTGA is known to be associated with ventricular septal defect, atrial septal defect, and obstruction in right ventricular outflow tract, tricuspid valve anomalies, and coarctation of the aorta. Abnormalities of the morphological tricuspid valve (left atrioventricular valve in SLL loop) are frequently found during autopsy in CCTGA,[1] These abnormalities commonly resemble the Ebstein anomaly of a right-sided tricuspid valve in hearts without ventricular inversion.[2] Occurrence of obstructive lesions of right ventricular inflow is rare. A review of the literature revealed only a few individual case reports and a single autopsy series. Allwork et al.[3] in their autopsy study of 32 cases of CCTGA, noted abnormalities of the morphological tricuspid valve in 91% of the cases; most of them were Ebstein malformations and only five had supravalvular stenosing ring. Marino et al.,[1] in their echocardiographic study of 42 cases of CCTGA, found only two cases with obstruction in right ventricular inflow. Both the cases also had subpulmonary obstruction. Supratricuspid membrane often originates from the left atrial wall below the left auricle, rarely from the tricuspid annulus. The pathophysiological consequences of such a membrane are similar to the supramitral ring in hearts with concordant atrioventricular and ventriculoarterial connections. Both supratricuspid and supramitral are believed to develop after incomplete division of endocardial cushion tissue.[3] Toscano et al., classified the supramitral ring into two categories based on the involvement of the valve: Supramitral type and intramitral
