

Quadricuspid aortic valve diagnosed by transthoracic echocardiography in childhood

Figen Akalın, Banu Demirel, Pınar Ürenden, Neşe Bıyıklı

Department of Pediatrics, Marmara University Faculty of Medicine, İstanbul, Turkey

SUMMARY: Akalın F, Demirel B, Ürenden P, Bıyıklı N. Quadricuspid aortic valve diagnosed by transthoracic echocardiography in childhood. *Turk J Pediatr* 2010; 52: 339-341.

Quadricuspid aortic valve is a rare congenital malformation of the heart leading to significant aortic regurgitation or stenosis. Its diagnosis by transthoracic echocardiography is difficult. Most of the cases are diagnosed during surgery or autopsy. Associated abnormalities of the coronary arteries should also be searched, since surgical injury may have devastating results. We herein present an eight-year-old girl found to have a quadricuspid aortic valve during evaluation of chronic renal disease and systemic hypertension.

Key words: aortic valve disease, quadricuspid, aortic regurgitation, pediatric echocardiography.

The quadricuspid aortic valve is a rare form of congenital valvular malformation. Its incidence ranges between 0.008-0.033% in autopsy series¹. It is often detected incidentally by transesophageal echocardiography (TEE), during surgery or by postmortem examination, but is one of the well-recognized causes of aortic incompetence requiring surgical intervention². Its importance lies in its association with coronary abnormalities, which may lead to surgical catastrophe, if not diagnosed preoperatively. Hence, early recognition and follow-up are critical in these patients. Diagnosis of quadricuspid aortic valve is often missed during transthoracic echocardiogram (TTE). This report describes a case of quadricuspid aortic valve detected incidentally during routine TTE in a child with chronic renal disease.

Case Report

An eight-year-old girl diagnosed with chronic renal failure was referred to the pediatric cardiology clinic of Marmara University Hospital for evaluation of cardiac function and end-organ injury. Her initial cause of referral was failure to thrive and high levels of creatinine; unilateral renal agenesis and pyelonephritis sequela were considered the cause of renal failure. She had been followed in the pediatric nephrology

clinic for two years with medical treatment and was not on dialysis. Enalapril 5 mg/day was given as antihypertensive treatment, which was not sufficient for blood pressure control. Prenatal, natal and family histories were uneventful. Physical examination revealed her weight and height to be under the 3rd percentile. Blood pressure was 120/90 mmHg (systolic pressure 90 to 95th percentiles, diastolic pressure >99th percentile). Cardiac auscultation revealed a diastolic decrescendo murmur heard throughout the entire diastole in the tricuspid region. Telecardiogram and electrocardiogram were normal. TTE disclosed a hypertrophic left ventricle; systolic and diastolic functions were within the normal range (ejection fraction=62%). The aortic valve was found to have four equal-sized cusps; the right and left coronary arteries originated from aortic sinuses opposite to each other, and moderate aortic regurgitation was present (Figs. 1-3). Since the aortic regurgitation was not severe enough for surgical intervention, it was decided to follow her clinically with regulation of systemic hypertension and prophylaxis for endocarditis during surgical procedures.

Discussion

Quadricuspid aortic valve is a rare congenital abnormality. The first case was reported

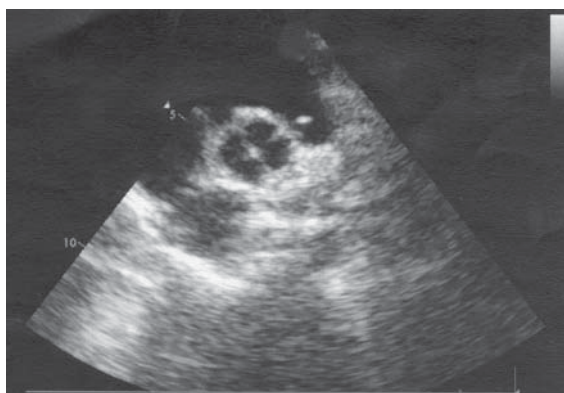


Fig. 1. Transthoracic parasternal short-axis view demonstrating the quadricuspid aortic valve with four equal cusps.

in 1862 from necropsy data, and the first echocardiographic report was from 1984³. Since then, the frequency of this diagnosis has been increasing. However, less than 200 cases have been reported to date. Most of the cases were recognized incidentally. Its recognition during childhood is even rarer. Aortic regurgitation is the most common hemodynamic abnormality, and a few cases of aortic stenosis were reported⁴.

Embryologically, the aortic valve is formed when the truncus arteriosus partitions into aortic and pulmonary valves. The semilunar cusps develop from three small pads of connective tissue in the wall of the aortic trunk. Deviation from these leads to an aortic valve with one, two, four, or even five cusps. On the basis of the morphology, seven types of quadricuspid aortic

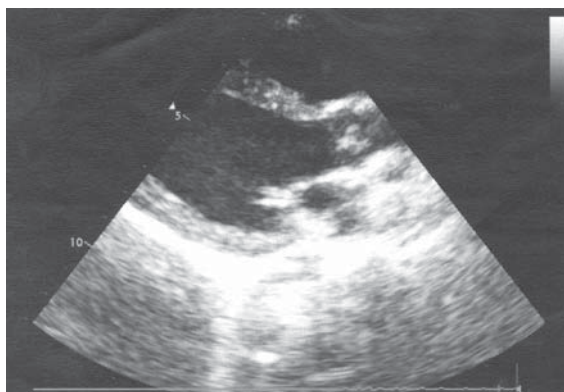


Fig. 2. Transthoracic parasternal long-axis view of the quadricuspid valve forming the typical "W" shape.

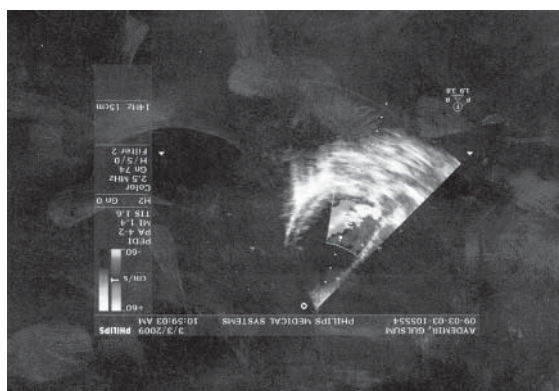


Fig. 3. Apical five-chamber view demonstrating the aortic regurgitation.

valve are recognized: 1) three equally large cusps, one small cusp; 2) four equal cusps; 3) two equally large, two equally small cusps; 4) one large, two intermediate, one small cusps; 5) three equally small, one large cusps; 6) two equally large, two unequally small cusps; and 7) four unequal cusps³. Our case falls in the second group.

Quadricuspid aortic valve may be associated with coronary abnormalities (including single coronary artery, displaced coronary ostium, etc.). Patent ductus arteriosus, subaortic stenosis, ventricular septal defect, deficient mitral valve, and aneurysmatic dilatation of the ascending aorta have also been reported as accompanying heart disease³. However, the lesion is most commonly found as an isolated defect.

Systemic disorders such as renal failure associated with this anomaly have not been reported to date, which is another unusual aspect of our case. Renal agenesis was found in our patient, which was another developmental congenital abnormality. We do not know whether this is a coincidental association or whether the same genetic or intrauterine exposure caused these two abnormalities.

The physiopathology of the valve dysfunction is poorly understood. Anatomical abnormalities of the cusps could induce unequal shear stress leading to fibrosis and incomplete coaptation³. However, insufficiency is also observed in cases of quadricuspid valve with four equal cusps. In our patient, the aortic valve had four equal-sized cusps, the coronary anatomy

was normal, and moderate aortic regurgitation was present.

Due to asymmetric mechanical stress around the four cusps, there is progressive deterioration and increased risk of endocarditis in these patients⁴. Hence, the patients should be on regular follow-up, and infective endocarditis prophylaxis must be advised whenever indicated³. The functional aspect of the quadricuspid valve is mainly represented by pure insufficiency in adulthood.

Surgical intervention is needed in patients with severe aortic regurgitation; however, aortoplasty is usually not successful, and artificial valve replacement is required. Preoperative diagnosis is essential for prevention of possible injuries to the abnormally located coronary arteries.

In conclusion, quadricuspid aortic valve is a rare congenital malformation, which can be diagnosed by two-dimensional transthoracic echocardiography. The anatomy of the aortic valve, number of cusps and pattern of coronary arteries should be determined before surgery in patients with aortic regurgitation.

REFERENCES

1. Attaran RR, Habibradah MR, Baweja G, Slepian MJ. Quadricuspid aortic valve with ascending aortic aneurysm: report of a case and discussion of embryologic mechanisms. *Cardiovasc Pathol* 2009; 18: 49-52.
2. Formica F, Sangalli F, Ferro O, Paolini G. A rare cause of severe aortic regurgitation: quadricuspid aortic valve. *Interac Cardiovasc Thorac Surg* 2004; 3: 672-674.
3. Kasliwal RR, Chuq̄h S, Manoca S. Quadricuspid aortic valve with severe aortic regurgitation. *Indian Heart J* 2006; 58: 158-159.
4. Yıldırım SV, Gümüř A, Cořkun I, Türköz R. Quadricuspid aortic valve: a rare cause of aortic regurgitation and stenosis. *Turk J Pediatr* 2008; 50: 500-502.
5. Saxena P, Konstantinov IE, Downie S, Newman MA. Quadricuspid aortic valve: a rare clinical entity. *Heart Lung Circ* 2009; 18: 50-51.
6. Godefroid O, Colles P, Vercauteren S, Louagie S, Marchandise B. Quadricuspid aortic valve: a rare etiology of aortic regurgitation. *Eur J Echocardiogr* 2006; 7: 168-170.