Congenital Aortic Valve Stenosis

35

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Keywords

Aortic valve stenosis • Echocardiography • Bicuspid aortic valve Unicuspid aortic valve • Aortic insufficiency (AI)

Definition and Morphology

Congenital aortic valve stenosis (AS) is a relatively common anomaly. Congenital AS occurs much more commonly in men, with a male to female ratio of 4:1. Related cardiac anomalies have been seen in up to 1/5 of patients. Patent ductus arteriosus (PDA) and coarctation of the aorta happen most often with AS [1].

Aortic valve (AV) may be unicuspid, bicuspid, or tricuspid or even a dome-shaped diaphragm. Unicuspid valves produce severe stenosis and are the most common anomaly found in valvular AS in children younger than 1 year. Congenitally bicuspid valves might be stenotic due to commissural fusion at birth, but more often they don't create severe stenosis in childhood and adolescence. A subgroup of cases with a bicuspid AV develops important aortic regurgitation (AR) demanding valve surgery. But most patients have normal function of valve till late in life, and superimposed calcific changes result in valve stenosis. When the AV obstruction is hemodynamically important, concentric hypertrophy of the left ventricular (LV) wall and also dilatation of the ascending aorta occur gradually.

Aortic dilation is present in more than 1/2 of young patients with normally functioning bicuspid AV. Aortic dilation must be monitored carefully with echocardiography. The section of maximal dilatation frequently involves the mid-part of ascending aorta but can also contain the aortic sinuses [1–3].

Clinical Findings and Physical Examination

In overall the children are asymptomatic; exercise fatigue and chest pain are unusual complaints and happen only when the stenosis is severe. The

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fundamental manifestations in adults are exertional dyspnea, angina, syncope, and finally heart failure. Most of the patients now are diagnosed before the onset of symptoms according to the finding of a systolic murmur on physical examination, with confirmation by echocardiography. In cases with bicuspid AV stenosis, symptoms typically occur at age 50–70 years though even in older age group, about 40 % of AS patients have a congenital bicuspid valve [1, 2].

The most common clinical presentation is a gradual decrease in exercise tolerance, fatigue, or dyspnea on exertion because of LV diastolic dysfunction, with too much rise in end-diastolic pressure leading to pulmonary congestion, and also due to restricted ability to increase cardiac output with exercise. More severe exertional dyspnea, orthopnea, paroxysmal nocturnal dyspnea, and finally pulmonary edema are late symptoms in cases with untreated AS [1–3].

Angina happens in approximately 2/3 of cases with severe AS, about 1/2 of them have associated significant coronary artery disease; angina usually results from the combination of the increased oxygen needs of hypertrophied myocardium and decrease of oxygen delivery secondary to the excessive compression of coronary vessels.

Syncope is most commonly produced by the reduced cerebral perfusion that happens during exertion when arterial pressure declines resulting in systemic vasodilation in the presence of a fixed cardiac output and also due to baroreceptor malfunction in severe AS. Syncope at rest may be caused by transient atrial fibrillation (AF) or due to transient atrioventricular block created by extension of the calcification of the AV into the conduction system [1–4].

Other late findings in patients with isolated AS include AF, pulmonary hypertension, and systemic venous hypertension. Of course, AS may be responsible for sudden cardiac death; this typically happens in cases who had formerly been symptomatic.

On physical examination, a sustained LV apical impulse with a fourth heart sound (S4) is a marker of severe stenosis. The slowly rising and low-volume carotid arterial pulse of severe AS can be noted in younger patients, but changes in arterial compliance frequently mask these findings in older patients. In younger patients with congenital AS, the flexible valve can result in an accentuated A2, and S2 may be normally split, even with severe valve obstruction. In addition, an aortic ejection sound can be audible due to halting upward motion of the AV and disappears when the leaflets become severely calcified. So, it is frequent in children and young adults with congenital AS but is unusual in adults with acquired calcific and rigid AS [1, 2].

The harsh systolic murmur of AS, loudest at the base of the heart and radiating to both carotids, is frequently but not always prominent and noticeable. Low-output states and also obesity can mask the findings. The murmur may radiate toward the apex, but the harsh component is vanished [1].

Electrocardiography

LV hypertrophy with or without strain is the hallmark point [1].

Chest Radiography

Cardiac ratio is overall normal or slightly increased in patients with congenital AS [1, 2].

Echocardiography

2D echocardiography delivers detailed information about the morphology of the AV, the LV function, and the presence or absence of related left-sided lesions. Doppler echocardiography is used to determine the severity of AS and the presence of associated AI.

Also comprehensive echocardiographic assessment should be done for patients with bicuspid AV; echocardiographic characteristic features include systolic doming with eccentric closing line of two visible cusps in the parasternal long-axis view and a single commissural line with two functional cusps in the parasternal

short-axis view. Specific care should be taken in studying the valve in both systole and diastole. Because in some patients with asymmetric leaflets and a noticeable and evident raphe, the valve can seem tricuspid in diastole; but the oval football shape of the systolic appearance of orifice indicates bicuspid AV [1, 4, 5] (Fig. 35.1).

The leaflets often are thickened and fibrotic and calcified with increasing age. With extensive calcification, it may be difficult to distinguish stenotic tricuspid from bicuspid AV [4, 5] (Fig. 35.2).

Also, Doppler provides peak instantaneous gradients which are higher than the peak-to-peak gradients derived from cardiac catheterization. Mean gradients as determined by Doppler and catheterization associate closely (Fig. 35.3).

Also, importantly, aortic dilation is present in more than 1/2 of young patients with normally functioning bicuspid AV in echocardiographic study. Aortic dilatation must be monitored and followed up carefully by echocardiography. The section of maximal dilatation frequently is seen in the mid-part of ascending aorta but can also contain the aortic sinuses. If the images of transthoracic echocardiography are limited, alternative imaging studies should include TEE, CT, or CMR. According to ACC/AHA guidelines, aortic root replacement is suggested for cases with a bicuspid AV whose aortic root size is 5 cm or larger [1, 5].

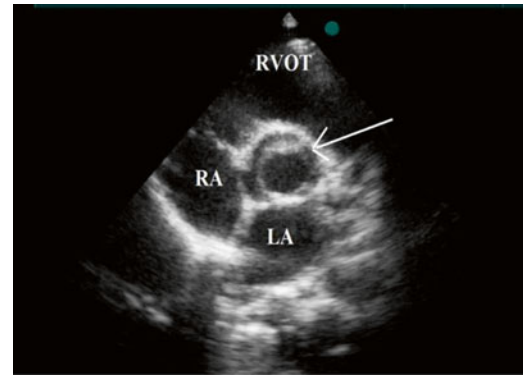

Fig. 35.1 Oval-shaped opening of the bicuspid aortic valve (BAV) in parasternal short-axis view; note the evaluation was done in systole (*arrow*). *LA* left atrium, *RA* right atrium, *RVOT* right ventricular outflow tract (Video 35.1)

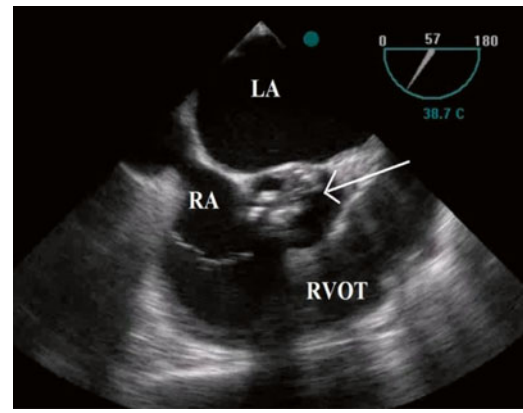

Fig. 35.2 Transesophageal echocardiography showing thickened and calcified BAV in short-axis view (*arrow*). *LA* left atrium, *RA* right atrium, *RVOT* right ventricular outflow tract (Video 35.2)

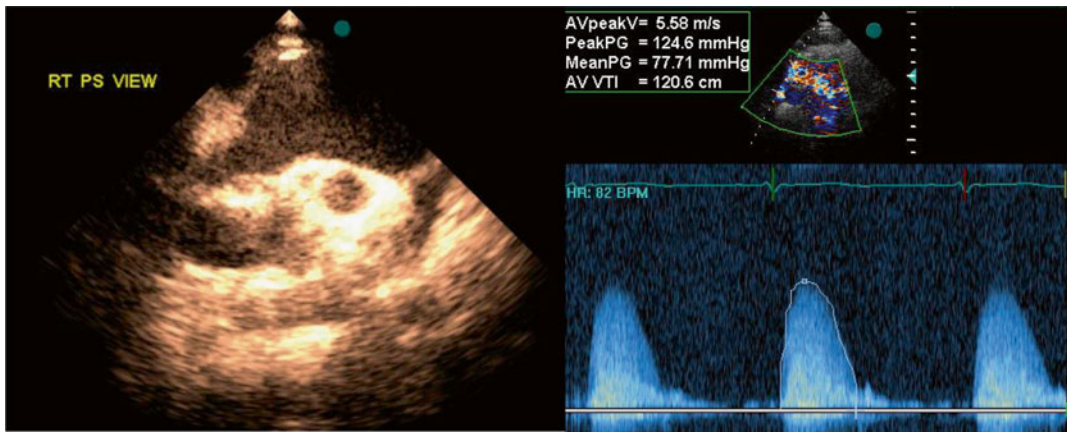

Fig. 35.3 (**a**, **b**) The max gradients should be evaluated by searching in all available windows. *RT PS* right parasternal always should be used

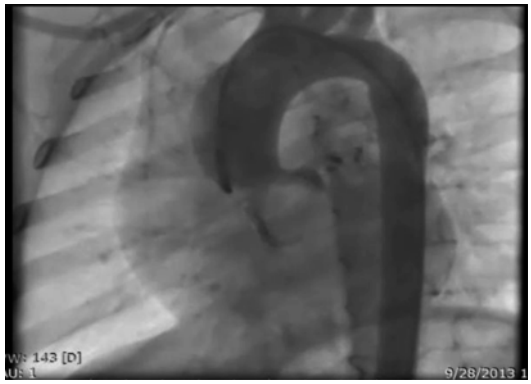

Fig. 35.4 The catheter course is arterial; injection was performed in aortic root and showed doming of AV cusps due to bicuspid aortic valve (Video 35.3)

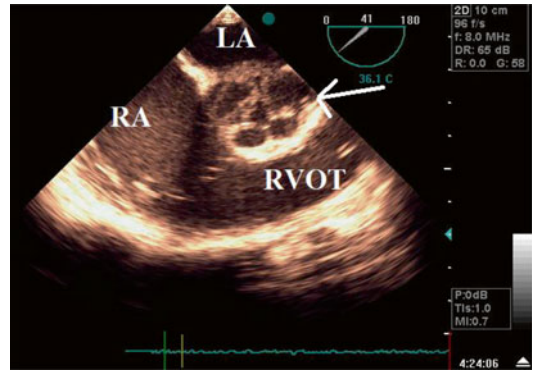

Fig. 35.5 Quadricuspid aortic valve in short-axis transesophageal echocardiography associated with significant aortic regurgitation

Catheterization

Nowadays, cardiac catheterization is rarely used to confirm the site and grade of severity of obstruction to LV outflow. Of course, catheterization is carried out when interventional therapeutic balloon aortic valvuloplasty is planned [5–7] (Fig. 35.4).

Management and Follow-up

Nowadays, therapeutic balloon aortic valvuloplasty has virtually replaced primary surgical valvotomy in children. Of course, aortic valvotomy is a so safe and effective palliative surgery with excellent results in relief of symptoms. AI can infrequently be progressive and need valve replacement. Furthermore, after valve commissurotomy, the valve leaflets remain deformed, and progressive degenerative changes containing calcification will cause important stenosis in future years. So prosthetic AV replacement is needed in about 35 % of cases within 15–20 years of the first operation. For those adolescents requiring AV replacement, the surgical options consist of replacement with a mechanical AV, an aortic homograft, or a pulmonary autograft in the aortic position (Ross procedure). Collecting evidence shows that the pulmonary autograft may be better than the aortic homograft. In the Ross technique, the patient's pulmonary valve is removed and used to replace the diseased AV, and the right ventricular outflow tract is recreated with a pulmonary valve homograft [6–8].

Quadricuspid Aortic Valve

Definition and Diagnosis

Quadricuspid aortic valves (QAV) are infrequent but well-recognized reason of important aortic valve insufficiency (AI). The first instance was reported in 1862. Generally QAV cases traditionally have been exposed incidentally in surgery or postmortem study; however, with advances in echocardiographic studies, many cases are now being diagnosed antemortem. The preoperative identification of QAVs is important as they can be related with abnormal coronary arteries' ostium. Using the transesophageal echocardiography (TEE) is so helpful in the diagnosis, because a higher-frequency transducer is used, and also it is at closer proximity to the heart. In diastole, in the short-axis view of the aortic valve (AV), the commissural lines of adjacent cusps result in an X conformation rather than the Y shape of the normal tricuspid AV [9–11] (Fig. 35.5; Videos 35.4 and 35.5).

Though the echocardiographic studies may propose the size of the cusps of AV, they do not correlate with surgical findings, always. TEE has also

revealed displacement of coronary ostium. Newly, cine magnetic resonance imaging (MRI) has also been used for the diagnosis of QAV and the associated lesions. Hurwitz and Roberts defined the seven common anatomic variants of QAV depending on the size of the leaflets. Of course, more than 85 % of the reports are of type A, B, and C [12].

Presentation and Outcome

AI usually develops due to fibrotic thickening with incomplete coaptation of leaflets. Indeed, with the unequal distribution of stress on cusps and also abnormal leaflet coaptation, AI may occur. AI is not frequently seen in young cases with QAVs. Aortic stenosis (AS) may be present but is uncommon and rare. In one of the previously reported studies, of 108 patients with QVA, 73 had pure AI and 28 cases were normal, and in 7 cases there was evidence of mixed AI and AS; in no case there was pure AS (Video 35.6). Of course, it appears that valvular dysfunction has a tendency to deteriorate in adult life and often needs surgery around the sixth decade. The definite risk of infective endocarditis (IE) is not clear, but there have been documented cases of IE affecting a QAV [13, 14].

Associated Abnormalities

Anomalies of coronary artery ostium and course represent in less than 1 % of all congenital heart disease and can associate with QAV. So for surgeon, it is important to be aware and conscious of any displacement of the coronary artery origin to prevent ostial obstruction during fixing of the prosthetic AV ring. Also, QVAs have been reported in association with other abnormalities, including nonobstructive hypertrophic cardiomyopathy, ventricular septal defect, pulmonary valve stenosis, fibro-muscular subaortic stenosis, supravalvular stenosis, and left coronary artery atresia. Fenestrations of the aortic cusps in QAV are seen with advancing age. In addition, QVA has also been reported in association with Ehlers-Danlos syndrome [14].

Management

When a QAV is found on echocardiography, follow-up study is needed as progress to severe AI is to be expected. Valves with four equal cusps are not less likely to develop significant AI. There are a few confirmed cases of IE affecting a QAV, so IE prophylaxis is recommended [12–14].

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