Diagnosing and Managing Bicuspid Aortic Valve

Azam Ansari, MD and Ryan Devine, BS

Echocardiography is the key to early diagnosis and effective management of this often silent cardiac abnormality.

Found in approximately 1% to 2% of the general U.S. population, bicuspid aortic valve (BAV) is the most common congenital heart abnormality.[1] Because the condition is often asymptomatic, its presence may go undetected for many years.[2] And though some people with BAV never experience complications, the condition frequently predisposes the aortic valve to degenerative calcification that can culminate in stenosis, incompetence, or both. In addition, the valve becomes increasingly susceptible to infective endocarditis. For these reasons, early recognition of BAV and periodic follow-up are of utmost importance.

Before the introduction of echocardiography into clinical practice, definitive BAV diagnosis was delayed until the patient underwent cardiac catheterization, angiography, or surgery—or until the excised valve was examined pathologically.[3] This contributed to the difficulty of detecting the condition (particularly in young, asymptomatic patients) and, thus, increased the vulnerability of many patients to infective endocarditis. Today, however, both transthoracic and transesophageal echocardiographic procedures can help providers diagnose the disease early in its course.

In this article, we describe two cases of congenital BAV: one in an adolescent and one in a middle-aged man. These cases illustrate the role of echocardiography in the timely diagnosis and appropriate management of BAV in patients of all ages. We also discuss the basic presentation, diagnosis, and management of the condition.

CASE 1: BAV IN ADOLESCENCE

A young man, aged 17 years, presented to a multispecialty practice for evaluation of a heart murmur that had been detected by his primary care provider. He was asymptomatic and had no history of rheumatic or scarlet fever. Physical examination revealed a grade I/VI early diastolic murmur following the aortic component of the second sound. The murmur radiated down the left parasternal area.

A transthoracic echocardiogram showed eccentric closure and two cusps of the aortic valve when opening (Figure 1 A and B). Application of color-flow Doppler revealed the presence of a mosaic color aortic regurgitation (AR) jet below the aortic orifice extending into the left ventricular outflow tract (Figure 1 C). The patient's left ventricular size was at the upper limit of normal, with normal left ventricular function.

In the absence of surgical indications, the patient was instructed in infective endocarditis prophylaxis before dental, genitourinary, or gastrointestinal surgery and counseled about periodic follow-up.

CASE 2: BAV IN MIDDLE AGE

A 50-year-old man was evaluated for a harsh basal systolic murmur, fatigue, and dyspnea on minimal exertion. He reported no chest pain, syncope, or history of rheumatic disease or scarlet fever.

After a transthoracic echocardiogram yielded suboptimal results, another echocardiogram was performed using the transesophageal route. A foreshortened four-chamber view in transverse axis revealed two aortic valve cusps of

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Dr. Ansari is a cardiologist in the department of cardiovascular medicine at Fairview Southdale Hospital, Edina, MN and Abbott Northwestern Hospital in Minneapolis, MN. He is also a fellow of the American College of Cardiology and the American Heart Association. **Mr. Devine** is a student of osteopathic medicine at Philadelphia College of Osteopathic Medicine, Philadelphia, PA.

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unequal length with doming of the left coronary cusp, an eccentric narrow orifice, and poststenotic dilation (50 mm) of the aortic root (Figure 2 A)—all of which indicated a BAV with aortic stenosis (AS). Color-flow Doppler imaging showed disturbed blood flow beginning below the aortic valve and extending into the aortic root through the stenotic orifice (Figure 2 B). Cardiac catheterization confirmed a peak mean gradient of 86 mm Hg across the aortic valve. Coronary angiogram was normal.

Because of a reduction in left ventricular systolic function (reflected by an ejection fraction of less than 50%), the evaluating physician recommended aortic valve replacement. During this procedure, the surgeon found the aortic valve to be thickened, partially calcified, severely distorted, and stenotic. Pathologic examination of the excised valve confirmed severe AS in a degenerated BAV.

After the prosthetic valve was placed, the patient's symptoms improved markedly, his left ventricular hypertrophy (LVH) regressed, and he was able to return to full-time employment. The patient also began oral anticoagulation therapy, with monitoring for an international normalized ratio of 2.5 to 3.

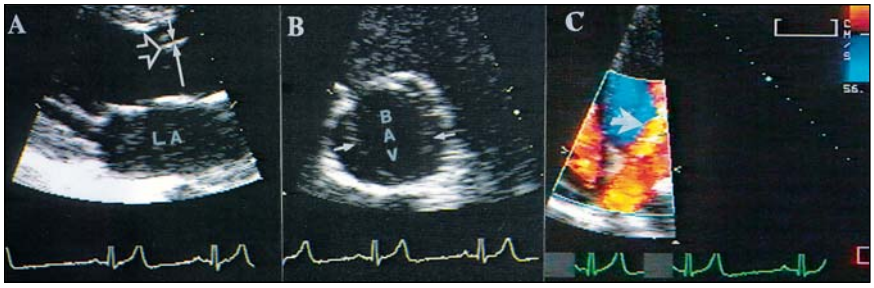

Figure 1. Transthoracic echocardiogram of a 17-year-old male patient with a congenital bicuspid aortic valve presenting as isolated aortic regurgitation. The parasternal long-axis view (A) shows eccentric closure (open, short arrow) of the aortic valve due to unequal length of the leaflets (thin arrows); the parasternal short-axis view (B) shows the aortic root, which contains an open bicuspid aortic valve; and the modified apical four-chamber view (C) focuses on the aortic regurgitation jet (short, closed arrow). Abbreviations: BAV = bicuspid aortic valve; LA = left atrium.

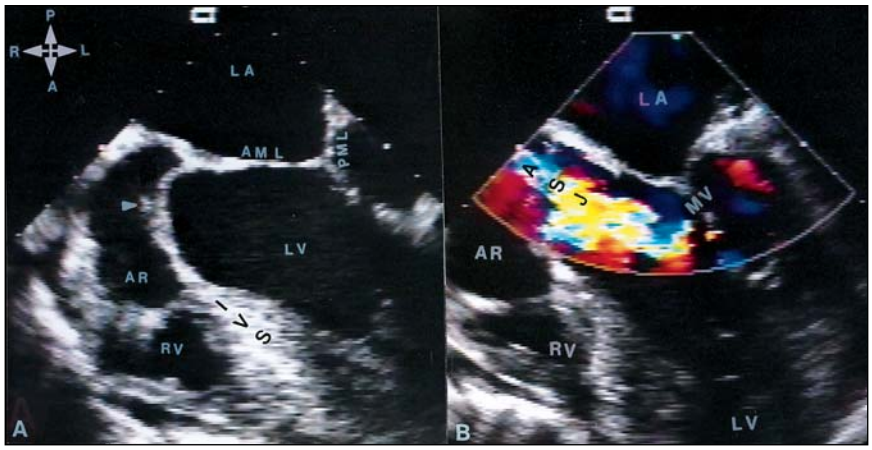

Figure 2. Transesophageal echocardiogram of a 50-year-old male patient with severe isolated aortic stenosis originating in a bicuspid aortic valve. The left ventricular outflow tract view (A) shows two unequal aortic valve leaflets with an eccentric, slit-like, narrow opening (small arrowhead). The same view with color flow Doppler application (B) shows a mosaic color jet originating below the valve and traversing through the narrow orifice. Abbreviations: AML = anterior mitral leaflet; AR = aortic root; ASJ = aortic stenosis jet; IVS = interventricular septum; LA = left atrium; LV = left ventricle; MV = mitral valve; PML = posterior mitral leaflet; RV = right ventricle. Compass: A = anterior; L = left; P = posterior; R = right.

RECOGNIZING AND EVALUATING BAV

Clinicians should consider the possibility of BAV when AS occurs in isolation, in the presence of an ejection systolic click in the second or third right or left intercostal space, or in the presence of other congenital heart diseases (such as coarctation of the aorta, high membranous ventricular septal defect, patent ducts, or aneurysm of the sinus of Valsalva). The incidence of BAV is higher in patients with these defects than in the general population. As the first case illustrates, it's also important to suspect congenital BAV in any patient whose physical examination suggests isolated AR and whose history includes neither rheumatic nor scarlet fever.

Transthoracic echocardiography is the gold standard for confirming a BAV diagnosis. Echocardiographic features of BAV and its sequelae have been described in detail elsewhere.[4,5] It's important to note that echocardiography plays a key role not only in diagnosing patients with BAV but also in obtaining important data about the natural history of the disease. Changes in the structure and function of the valve can be

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noted as early as one year or as late as 10 to 20 years after initial diagnosis. About 30% of patients may remain asymptomatic until they are about 70 years old.[4]

NONSURGICAL MANAGEMENT

All patients with confirmed BAV, asymptomatic or symptomatic, should receive lifelong infective endocarditis prophylaxis before dental, genitourinary, or gastrointestinal surgery as recommended by the American Heart Association.[6] They also need periodic surveillance to detect any transition from normal to abnormal hemodynamic status for timely and appropriate intervention. Echocardiography and exercise testing often helps in such routine evaluations. In general, if the mean aortic valve gradient exceeds 40 to 50 mm Hg, or if the aortic valve surface area is 1 to 1.5 cm^2 in the presence of LVH, surgical replacement of the BAV should be considered, even in an asymptomatic patient. The idea is to minimize the possibility of sudden death, which occurs in 1% to 2% of patients with these cardiac parameters.

Percutaneous retrograde catheter balloon dilation of stenotic BAV (valvuloplasty) can relieve symptoms temporarily but does not prolong survival.[7] For this reason, balloon valvuloplasty of stenotic BAV must be regarded as a palliative measure for select patients with contraindications to surgery or as a preliminary procedure in patients who eventually will receive definitive surgery.

VALVE REPLACEMENT

Degenerative BAV that results in AS or AR has no effective medical therapy. These sequelae are mechanical in nature and require surgical valve replacement. Even in the presence of such clear surgical indications, however, the risk of valve replacement (1% mortality and 1% morbidity) must be weighed against the risks of delaying the procedure, particularly in patients with early symptoms. Asymptomatic patients generally have a good prognosis without valve replacement, and should be offered nonsurgical management as described in the previous section.[8]

In evaluating the surgical candidacy of a patient with BAV and AR, clinicians should consider the patient's symptoms, exercise capacity, left ventricular dimensions, and ejection fraction. Decreasing exercise capacity, failure of ejection fraction to rise during exercise, and increasing dimensions of the left ventricle on serial echocardiography are important indications for surgical intervention.

The long-term relative survival rate after aortic valve replacement is excellent. At 10 years, the age-corrected survival rates among patients who undergo this surgery approach normal.[9]

After valve replacement, it's important that patients with BAV continue to practice infective endocarditis prophylaxis. If the replacement valve is mechanical, the patient also should receive anticoagulation therapy, with a target international normalized ratio of 2.5 to 3. Bioprosthetic valve implantation obviates the need for anticoagulation unless the patient has atrial fibrillation.

SUMMING UP

BAV is common in patients with isolated AR or AS and should be considered in any patient presenting with either symptom. Through the use of echocardiography, the cornerstone for BAV diagnosis and evaluation, clinicians can help reduce the morbidity and mortality associated with the condition by optimizing management. ●

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