CLINICAL VIGNETTE

An Asymptomatic Young Male with Bicuspid Aortic Valve with Severe Aortic Insufficiency

Sandra Vizireanu, M.D., and Gopi Manthripragada, M.D., FACC

Introduction

As the most prevalent congenital cardiac anomaly, bicuspid aortic valve is found in approximately one percent of the population with a 2-3 to 1, male to female ratio.[1] It is a heritable disorder with clinical studies reporting 9% prevalence in first degree relatives.[2]

It is associated with other congenital cardiac lesions, and when diagnosed in adults possible complications include aortic aneurysm and dissection, endocarditis and stenotic, or regurgitant aortic valves.

Case Report

A 28-year-old healthy male with no known medical history presents to his primary care physician for an annual physical examination. He is active without any limiting symptoms and has not seen a physician for a decade. On physical exam, he was noted to have a 3/6 holo-diastolic murmur best heard at the left upper sternal border and audible throughout the precordium. A subsequent echocardiogram showed moderately increased left ventricular size (left ventricular end diastolic and systolic dimensions measured 6.2cm and 4.4cm, respectively), preserved left ventricular systolic function with an ejection fraction of 60-65%, bicuspid aortic valve, and severe aortic regurgitation. Laboratory data were unremarkable with the exception of subclinical hypothyroidism. A CT angiogram showed mild ectasia of the ascending aorta with a maximal transverse diameter of 42mm. There was no aortic coarctation, and the thoracic and abdominal aorta were of normal caliber.

Discussion

Despite being asymptomatic, the patient's severe aortic regurgitation, increased ventricular and aortic size prompted a discussion on surgical management for his bicuspid aortic valve. Recent studies suggest that those with at least one of three risk factors–age >30, moderate or severe aortic regurgitation, and moderate or severe aortic stenosis–have higher cardiac event rates.[3,4] Baseline ascending aorta >40mm was an independent predictor for eventual surgery (risk ratio, 10.8; 95% confidence interval, 1.8 to 77.3; $P<0.01$).[3]

Medical therapy includes beta-blockade (ACC/AHA IIA recommendation), and angiotensin receptor blockers based on shared pathophysiology with Marfan's syndrome.[5,6] Surgical aortic valve replacement is a Society of Thoracic Surgeons (STS) and ACC/AHA Class I indication for symptomatic patients with severe aortic regurgitation regardless of left ventricular systolic function or asymptomatic patients with ejection fraction ≤ 50%. In asymptomatic patients with normal left ventricular function and left ventricular dilation, surgery may be considered (left ventricular end systolic dimension >5cm is a IIA recommendation, left ventricular end diastolic dimension >6.5cm IIB based on 2014 ACC/AHA Guidelines).[7, 8]

The incidence of aortic dissection in those with bicuspid aortic valves is lower than previously thought with recommendations for aortic root repair if the aorta measures 5.5cm in greatest dimension (Class I indication). In those undergoing surgery for severe aortic stenosis or regurgitation, aortic surgery is reasonable if the aorta measures >4.5cm.[8]

Diagnosis

A thorough personal and family history is important, as well as a thorough physical exam. Exam findings are dependent on the degree of valvular dysfunction and presence of associated abnormalities. It should include careful exam of arterial pulses for aortic coarctation. Clinical presentation is variable and can be detected by asymptomatic auscultatory findings, valve dysfunction, bacterial endocarditis, or incidentally noted thoracic aortic aneurysm on radiographic imaging. Diagnosis is confirmed by transthoracic echocardiography with findings of systolic doming, eccentric valve closure, leaflet redundancy, leaflet prolapse, presence of a raphe, elliptical orifice of valve during systole, distinct opening pattern, and a dilated aortic root or ascending aorta.[9] Transthoracic echocardiography has sensitivity of 92% and specificity of 96% for detecting bicuspid aortic valves,[9] with valvular calcifications lowering accuracy. Transesophageal echocardiography is not necessary for diagnosis. CT angiogram may be warranted to evaluate the size and anatomy of the aortic root and ascending aorta. Thoracic aortic aneurysm, when present, must be quantified by aortic segment, diameter, and adjusted for body surface area. Symptoms may not always be elicited from the patient, and in those who are asymptomatic, an exercise stress test can help assess functional limitations.

Conclusion

After reviewing his test results, our patient elected to obtain a second opinion. He underwent successful bio-prosthetic aortic valve replacement and has had an uneventful recovery thus far. The diagnosis of bicuspid aortic valve, particularly when associated with significant valvular heart disease, can be challenging for both patients and their providers. Young, otherwise healthy individuals find it understandably difficult to realize they will need operative repair for a previously undiagnosed condition. Some elect to postpone surgery as long as possible, while others proceed more urgently. Further, they are faced with a choice of lifelong anticoagulation with a mechanical valve or the relatively less durable bio-prosthetic valve. For the provider, it is imperative to have a low threshold to obtain cardiac imaging in patients with unexplained cardiac murmurs, determine the ideal timing of surgery, and provide adequate screening to first degree relatives.

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