

Case Report

Bicuspid aortic valve with severe aortic stenosis: a case report

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Abstract

A bicuspid aortic valve (BAV) can be a serious disorder of heart valve in which the valve only has two leaflets or flaps that control blood flow through the heart. Between one and two percent of all people have this defect and it affects more men than women. This report presents a case of severe aortic stenosis with mild to moderate aortic regurgitation due to bicuspid aortic valve with hypertension. A 37 years old male presented with high record of blood pressure and occasional shortness of breath on exertion. Echocardiography (Color Doppler) revealed severe aortic stenosis with mild to moderate aortic regurgitation due to bicuspid aortic valve with moderately severe concentric LV wall hypertrophy. Surgical treatment (aortic valve replacement) was scheduled based on echocardiography findings. On surgical resection a well defined bicuspid aortic valve was found with calcification and friable valve leaflet. Histopathology of valve tissue shows large areas of calcification. Patient was discharged from hospital on 7th POD with an advice to attend cardiac surgery OPD after 1 month. Aortic valve replacement must be considered in this type of lesion.

Key words: Bicuspid aortic valve, aortic stenosis, aortic valve replacement, hypertension

Introduction

Bicuspid aortic valve (BAV) is a common congenital cardiac malformation affecting 1-2% of the population with strong male predominance,¹ in which the aortic valve only has two leaflets or flaps that control blood flow through the heart. (Figure-1) It may occur in isolation, or in association with other congenital heart diseases.² Individuals may have a normally functioning BAV and

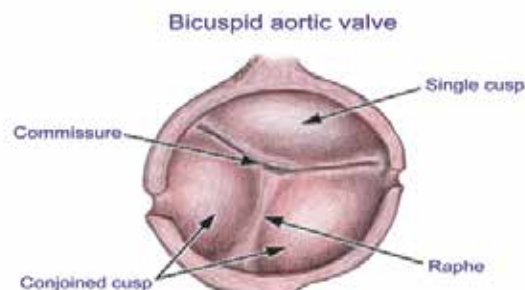


Figure-1: Bicuspid aortic valve

may be unaware of its presence and the potential risk of impending complications.³ They may typically remain asymptomatic until the third or fourth decade of life, when the valve becomes dysfunctional. They then require close follow-up and valve replacement may be warranted.⁴ BAV can be associated with abnormalities of the aortic wall such as coarctation of the aorta, aortic dissection, aortic aneurysm and turner's syndrome.⁴ It is now accepted that BAV is associated with both valve disease and aortic disease,⁵ thereby leading to increased morbidity and mortality, including aortic valve disorders, aortic wall abnormalities, endocarditis and other cardiovascular malformations.⁶ BAV has been identified at a prevalence of 4.6 cases per 1000 live births. The prevalence of BAV according to sex has been found to be 7.1 cases per 1000 among male neonates, and 1.9 per 1000 among female neonates. All newborns with BAV are asymptomatic. Mild aortic regurgitation has only been found in one neonate with BAV.⁶ Beta-blockers and statins are the possible medical treatment and aortic valve repair/replacement are indicated for patients with a severely diseased aortic valve. Rigorous follow-up throughout life is mandatory after BAV has been diagnosed. The aim of the present article was to describe the implications of BAV and to discuss diagnostic and treatment strategies.

Case report

A 37 years old male presented with high record of blood pressure and occasional shortness of breath on exertion since 11 months. One year later, patient was hospitalized with high record of blood pressure. On clinical examination it was diagnosed as a case of aortic stenosis. Then patient referred to our centre for further evaluation and management. Clinical examination showed normal body development. Bilateral radial pulses were palpable

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and regular at the rate of 72 beats/min, low volume and slow rising pulse. BP was 140/70 mm(Hg) in bilateral upper limbs in supine position. Cardiac examination revealed heaving apex in left fifth intercostal space, ejection systolic murmur of grade 4/6 in aortic area radiating to right carotid. General examination and other systems examination was unremarkable. On investigations, Blood investigations were essentially normal. Chest X-ray P/A view revealed cardiomegaly with left sided cervical rib. His ECG shows left ventricular hypertrophy (LVH). Echocardiography (Color Doppler) revealed severe aortic stenosis (peak pressure gradient 66 mm Hg, AV ring 23mm) with mild to moderate aortic regurgitation due to bicuspid aortic valve, severe concentric left ventricular hypertrophy & grade-I diastolic left ventricular dysfunction, LVEF-53%, PASP-27mmHg. Surgical treatment (aortic valve replacement) was scheduled based on echocardiography findings.

On surgical procedure, with all aseptic precaution, standard median sternotomy was done and cardiopulmonary bypass was established using single venous two stage canula. Cooling was done at 31.1°C. Then aortic cross clamp was applied, aortotomy done & blood cardioplegia was given. Then aortic valve dissection and replacement done with mechanical valve (21 St Jude mechanical valve) by interrupted pledged suture. Then aorta was closed and cross clamp off done sequentially followed by removal of SVC and aortic canula. Cross clamp time was 108 min and extracorporeal circulation time 180 min. On surgical resection, a well defined bicuspid aortic valve was found with calcification and friable valve leaflet.(Figure-2) Histopathology of valve tissue shows large areas of calcification. Patient was discharged on 7th POD with an advice to attend cardiac surgery OPD after 1(one) month. On follow up after one months, patient was doing well and echocardiogram (Doppler) findings were prosthetic aortic valve in situ with mild valvular leakage with grade -1 AR



Figure-2 : Resected specimen of bicuspid aortic valve

(peak pressure gradient 9.2 mm Hg), concentric LVH and LVEF-60%. Chest X-ray P/A view revealed cardiomegaly with prosthetic aortic valve in situ.

Discussion

Congenital bicuspid aortic valves are common and in most cases remain undetected until infection or calcification supervenes. These valves may run in families in a multifactorial inheritance pattern or occasionally in an autosomal dominant one.⁹ In our case, chromosomal analysis could not be done due to technical limitation. It has been noted that BAV is inheritable. The pathogenesis of BAV is unknown. Experiments on syrian hamsters have revealed that BAV does not occur consequent to improper development of the conotruncal ridges, conotruncal malseptation, valve cushion agenesis or lesions acquired after normal valvulogenesis. Fusion of the right and left valve cushions at the beginning of valvulogenesis appears to be a key factor in BAV formation.¹⁰ A recent study has demonstrated that BAVs with fused right and noncoronary leaflets and those with fused right and left leaflets are different etiological entities. BAVs with fused right and noncoronary leaflets result from a morphogenetic defect that occurs before cardiac outflow tract septation on the basis of an exacerbated nitric oxide-dependent epithelial-to-mesenchymal transformation. On the other hand, BAVs with fused right and left leaflets result from anomalous septation of the proximal portion of the cardiac outflow tract, caused by dysfunctional neural crest cells.¹¹ Deficient fibrillin-1 content in the vasculature of BAV patients may trigger matrix metalloproteinase production, thereby leading to matrix disruption and dilation.¹² It has been noted that the fibrillin-1 content was remarkably reduced in the aorta of BAV patients, compared with that of patients with a tricuspid aortic valve.¹³⁻¹⁵ We could not add any information about this case in the process of development of bicuspid aortic valve.

BAVs may progress and become calcified, thus leading to varying degrees of severity of aortic stenosis or aortic regurgitation, or both, which may eventually necessitate surgical intervention.¹⁶ In our reported case, there was no clinical feature or any limitation of patient's physical activity before one year of admission in the hospital. BAV is recognized as a frequent cause of aortic stenosis in adults. Aortic stenosis has been found in 72% of adults with BAV. The stenotic valves were obstructed by nodular, calcareous masses, but commissural fusion was present in only eight cases. In this case, there was commissural fusion with diffuse calcification of valve leaflet which is supported by adhesion.¹⁶ Primary aortic regurgitation without infective endocarditis was uncommon, and 32%

had an apparently normally functioning aortic valve.¹⁷ Among the 600 patients analyzed, 213 (36%) had pure aortic stenosis, 265 (44%) had pure aortic regurgitation and 122 (20%) had combined stenosis and regurgitation. BAVs represented 18%, as the third most important cause of aortic disorder following degenerative and rheumatic changes, followed by infective endocarditis (5%).¹⁸ In 388 patients with severe aortic valve disease alone, BAVs were found in 45% of the patients with aortic stenosis and 24% of the patients with aortic regurgitation. In 110 patients with severe combined aortic and mitral valve disease, BAVs were found in only 12%.¹⁹ A double blind placebo controlled study illustrated that the patients recruited into the astronaut study were younger, with less severe aortic stenosis. The population of BAV patients was large and accounted for 48.9%.²⁰ From echocardiography, the patients with a stenotic BAV had significantly larger anatomical aortic valve areas than effective aortic valve areas. The discrepancy relating to jet eccentricity was much bigger than that of the patients with a stenotic tricuspid aortic valve, thus indicating greater severity of valve dysfunctional hemodynamics. In other words, the jet eccentricity correlated with BAV.²¹

A bicuspid aortic valve was detected before cardiac surgery in patients in whom echocardiograms were performed by TEE and by TTE. Serial assessment of the aortic valve by echocardiography is a valuable tool to evaluate the functional status of the valve as well as to measure the aortic diameter, chamber dimensions, and ventricular function.²² However, echocardiographic identification of a BAV can be obscured in severe stenosis and after cuspal fusion secondary to inflammation.

In general, patients with mild-to-moderate valvular dysfunction and normal left ventricular (LV) dimensions and function should be monitored by echocardiography at regular intervals. Aortic valve replacement is indicated for severe valvular dysfunction, symptomatic patients, and/or those patients with evidence of abnormal LV dimensions and function.²³ Because many of these patients will require cardiac surgery during their lifetime, early referral to a surgeon with experience in aortic valve surgery is recommended. Our patient reported to us a bit lately due to economical support and lack of proper awareness. Use of the pulmonary autograft (Ross procedure) for aortic valve replacement has been advocated as an important alternative to prosthetic valve implantation, particularly in younger patients.^{24,25} In this case, we could not perform Ross procedure due to lack of our team orientation.

In summary, bicuspid aortic valves are common congenital defects and with the virtual disappearance of

rheumatic fever in the developed world are likely to become the most important intrinsic cardiac predisposition for infective endocarditis (IE). Among those with bicuspid valves, IE is severe and tends to occur in the fourth and fifth decades of life requiring major surgery in most cases with significant mortality. Echocardiography should be undertaken in all young adults in whom murmurs are detected. Emanuel et al⁸ suggested echocardiography for first and second degree relatives of patients also undergo. The value of trans oesophageal echocardiography remains to be established. Recommendations such as avoiding exercises involving weight lifting, maintaining a normal blood pressure and relatively slow heart rate, control of cholesterol levels, and avoiding infection can also be made to these patients.²⁴ The detection of a bicuspid aortic valve will not only make it possible to offer antibiotic prophylaxis for IE but should also increase the index of clinical suspicion of endocarditis if such patients also have fever and malaise.

This report presented a rare case of severe aortic stenosis with mild to moderate aortic regurgitation due to bicuspid aortic valve. Aortic valve replacement must be considered in this type of lesion.

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