CLINICAL VIGNETTE

Aortic Dissection in a 40-Year-Old with Bicuspid Aortic Valve

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Case Report

A 40-year-old man with history of "congenital heart valve disease," hypospadias requiring surgical correction, and talipes equinovarus presented to an outside emergency room for anterior chest tightness ongoing for one day. His chest discomfort was acute in onset, occurred at rest, and worse with lying flat. There were no other associated symptoms. The patient denied any prior episodes of chest pain or tightness preceding this event. He was otherwise healthy with normal exercise tolerance. The patient denied fever, cough, nausea, diaphoresis, shortness of breath, lightheadedness, syncope, or congestive heart failure symptoms. In addition, he denied prior tobacco, methamphetamine, or cocaine use. Family history was notable for his mother who had cerebral aneurysms and his brother who had Chiari malformation.

In the emergency department, the patient was afebrile, pulse 96 BPM, blood pressure 125/74 mm Hg, respiratory rate of 15 breaths per minute, and oxygen saturation 98% on room air. He was well-appearing in no apparent distress with unremarkable cardiac and pulmonary exams. Electrocardiogram demonstrated diffuse ST elevation. Chest x-ray revealed an enlarged cardiac silhouette. Echocardiogram showed a severely dilated aortic root with Stanford type A aortic dissection, small pericardial effusion, bicuspid aortic valve, and moderate aortic regurgitation. CT angiogram confirmed a 7.6 cm in diameter ascending aortic aneurysm extending from distal edge of sinuses of Valsalva to just proximal to the left subclavian artery. In addition, there was a small pericardial effusion, concerning for hemopericardium. The descending thoracic aorta and abdominal aorta were unremarkable. He underwent emergent surgical repair which included graft repair of the ascending aorta, aortic valve replacement with a 25 mm bioprosthetic valve, and clipping of left atrial appendage. He was discharged on Metoprolol and Aspirin.

Background

Bicuspid aortic valve (BAV) is the most common congenital heart defect with an estimated prevalence of 0.5-2 percent and a roughly 3:1 male to female ratio.[1] BAV may exist in isolation or in association with other congenital and acquired cardiovascular disorders. The most common abnormality is thoracic aortic dilation, which is present in 50-70% of patients with BAV.[2] Other associated abnormalities include aortic coarctation, supravalvular stenosis, ventricular septal defect, atrial septal defect, patent ductus arteriosus, hypoplastic left heart, sinus of Valsalva aneurysm, coronary artery anomalies, aortic dissection, aortic stenosis, aortic regurgitation, and infectious endocarditis.[3] BAV is also associated with Turner Syndrome and intracranial aneurysms.[4] When there is the BAV-associated arthropathy, it mostly affects the proximal thoracic aorta. The most common site of aneurysm is the ascending aorta, followed by ascending aorta and aortic root, and then isolated aortic root. Aneurysms involving the innominate artery have also been described. Aortic dissection is one of the most serious complications of BAV.[5-7] It is estimated to be 5-10 times more common and occur on average 8 years earlier in patients with BAV compared to the general population.[5,6] Risk factors for aortic dissection associated with BAV include aortic size, male sex, family history of dissection, presence of other abnormalities such as aortic coarctation or aortic stenosis, and Turner syndrome.[3]

Etiology

BAV is considered a hereditary condition with incompletely understood inheritance, but can also occur sporadically.[7,8] There is a 9% prevalence of BAV in 1st degree relatives.[9] The developmental abnormalities leading to the formation of BAV are unknown, but may be due to abnormalities in neural crest cell migration and cell signaling.[3] The aortopathies associated with BAV are thought to occur primarily due to structural abnormalities such as decreased cystic medial degeneration, fibrillin, elastin fragmentation, and apoptosis that stem from the same genetic abnormalities leading to BAV.[10]

Diagnosis

Symptoms of BAV generally develop in adulthood and are usually due to the associated valvular and nonvalvular abnormalities. Physical exam findings of isolated, normal functioning BAV include an ejection click at the left lower sternal border or apex, often accompanied by a brief ejection murmur. The diagnosis of BAV is usually made by transthoracic echocardiography (TTE), which is 78-92% sensitive and 96% specific.[11] If TTE is indeterminate, transesophageal echocardiography or cardiac MR can be performed.

Management

The management of BAV involves surveillance, prevention, and early intervention for associated valvular and aortic patho-

logy. Patients with BAV should be regularly followed, with serial TTE imaging. The optimal imaging intervals are based on age as well as presence and progression of associated cardiovascular abnormalities.[12] First degree relatives should be screened for BAV with TTE. Exercise and activity recommendations range from no restrictions to complete avoidance of competitive sports, based on thoracic aortic diameter, presence of associated connective tissue disorder, and family history of thoracic aortic aneurysm and dissection.[12] Due to significant risk of aortic dissection and rupture, patients with BAV and dilated aorta should not drive if the ascending aorta measures greater than 6 cm. They should not drive commercially if the ascending thoracic aorta measures greater than 5.5 cm.

There is no specific medical therapy that improves the outcomes of BAV.[12] Theoretically, beta blockers may slow the progression of aortopathy by reducing aortic wall stress, but this has not been proven in this patient population. Angiotensin converting enzyme inhibitors or angiotensin receptor blockers have also been used without evidence of clinical benefit..

Surgery should be recommended as soon as the risk of watchful waiting exceeds the risk of surgical intervention.[13] Prophylactic aortic repair is recommended to prevent catastrophic complications such as aortic dissection and rupture. In the past decade, there has been an ongoing debate regarding the criteria for surgical intervention. The precise timing varies by individual patient, surgeon, and the treating institution. Asymptomatic BAL patients are recommended for surgical to the aortic root or ascending aorta if the aortic root or ascending aorta measures 5.5 cm or greater in size. In case the patient is at a low surgical risk, earlier surgical intervention may be considered at an experienced surgical center. Low surgical risk has been defined as a mortality risk of less than 4%, lack of frailty, absence of major organ system dysfunction which would not improve postoperatively (such as fixed pulmonary hypertension or dementia), and the lack of procedure-specific impediments, such as radiation damage or severe calcification of the ascending aorta. In such cases, surgical intervention is *reasonable* in asymptomatic patients if aortic root or ascending aorta measures between 5.0-5.5 cm in diameter and if there are additional risk factors for aortic dissection, such as aortic growth of at least 0.5 cm per year or family history of aortic dissection. In patients with BAV who plan to undergo surgical aortic valve replacement for severe aortic stenosis or regurgitation, it is *reasonable* to have surgical intervention to the ascending aorta if the ascending aorta measures greater than 4.5 cm in size.

Postoperatively, imaging of the aorta early after surgical correction should be considered to detect anastomotic leaks and pseudoaneurysms as well as establishing a baseline for future comparisons. In general, EKG-gated CT may be preferred as the imaging modality. In younger individuals less than 50 years age, MRI/MRA may be considered to limit radiation exposure secondary to repetitive CT scanning. Ongoing postoperative surveillance intervals should be individualized based on clinical, anatomic, and surgical features. The interval for repeat imaging after aortic surgery is determined by the extent of the initial operation and whether all areas of aortic dilatation were addressed during the initial surgery. When feasible, repeat studies should be performed at the same institution using similar imaging techniques and protocols to enhance comparison.

Conclusion

BAV is the most common congenital heart defect and is often associated with other cardiovascular disorders. The most common abnormality is thoracic aortic dilation, and one of the most feared complications is aortic dissection. Management includes surveillance, serial imaging, and prompt surgical intervention when the risk of watchful waiting exceeds the risk of surgical intervention.

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