Case Report

A Case Report of Undiagnosed Bicuspid Aortic Valve: A Global Health Challenge

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A Case Report of Undiagnosed Bicuspid Aortic Valve: A Global Health Challenge

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Abstract: Introduction and importance: Congenital bicuspid aortic valve (BAV) is a common cardiac malformation predisposing individuals to aortic stenosis, regurgitation, and aortopathy. This risk persists even after aortic valve replacement (AVR). **Case presentation:** We report a case of a male in his 60s with a history of BAV who underwent bioprosthetic aortic valve replacement (BVR) and ascending aorta/root repair in 2014. The patient was noncompliant with follow-up and presented nine years later with severe bioprosthetic aortic stenosis and an 8.0 cm ascending aortic aneurysm with no symptoms. Emergent redo BVR and aneurysm repair were successful. **Clinical discussion:** It should be emphasized that successful outcomes for BAV patients require a lifelong commitment to surveillance, even after surgical intervention. Noncompliance can lead to life-threatening consequences like bioprosthetic valve deterioration or progressive aortic dilation. **Conclusion:** This case highlights the importance of early BAV diagnosis for ongoing monitoring, accessible follow-up care after complex cardiac procedures, and addressing patient barriers to treatment adherence.

Keywords: aortic aneurysm; aortic valve replacement; aortopathy; bicuspid aortic valve; bioprosthetic valve; surveillance

Introduction

Bicuspid aortic valve (BAV) is a common congenital heart defect that affects approximately 0.5-2% of individuals in the United States (up to 6.5 million people, with an estimated annual incidence of 0.7%) [1]. The lack of obvious symptoms in early life often leads to BAV going undiagnosed for years, as was the case with a male in his 60s in our report. This is concerning because even when detected later, BAV carries a high risk of serious cardiovascular complications, such as aortic aneurysm, aortic valve pathologies (stenosis, insufficiency), and associated aortic complications (root dilatation, rupture, dissection) [2], ultimately leading to the highest associated mortality rate among congenital heart diseases (CHD) [3]. The frequent asymptomatic presentation of BAV patients, especially during childhood [4], highlights the body's ability to compensate for underlying cardiac abnormalities. The prevalence of undiagnosed BAV remains a concern, as reliable epidemiological data indicate that many cases may go unnoticed [5]. This case report is presented in accordance with the CARE 2020 Guidelines.

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Case Presentation

A 67-year-old Hispanic male with a history of hypertension, dyslipidemia, hypothyroidism, and bicuspid aortic valve (BAV) presented for an initial evaluation in 2014. His past medical history also included kidney stones and left ureteral lithotripsy. A diagnostic workup revealed severe aortic regurgitation (AR), a dilated aortic root (4.5 cm), and a dilated ascending aorta (4.7 cm). Transesophageal echocardiogram (TEE) and cardiac catheterization confirmed these findings, with minimal non-obstructive coronary artery disease (CAD) and a left ventricular ejection fraction (LVEF) of 55%. An aortic root angiogram confirmed severe aortic regurgitation (4+ AR), a dilated LV cavity, a dilated aortic root, and a dilated ascending aorta, with no aneurysm or dissection observed. Cardiovascular surgery was consulted. The patient underwent successful repair of the ascending aorta and aortic root, as well as a bioprosthetic aortic valve replacement with a Carpentier-Edwards Magna valve. The postoperative course was complicated by transient atrial fibrillation, which resolved, and he was discharged on carvedilol, rosuvastatin, aspirin, and amiodarone (which was discontinued shortly afterward).

Despite the successful intervention, the patient was completely lost to medical care for over nine years, remaining noncompliant with preventive medications and follow-up appointments. This lapse in care presented a significant concern for potential complications.

After 9 years, in 2023, the patient re-emerged in the cardiology clinic for cardiac clearance before prostate surgery. Remarkably, he was completely asymptomatic at this time. However, an echocardiogram revealed an LVEF at the lower limits of normal (50-55 percent), severe bioprosthetic aortic stenosis (AVA 0.6 cm^2, peak gradient 60 mmHg), mild aortic regurgitation (AR), and a massive 8.0 cm ascending aortic aneurysm. No dissection was observed.

Recognizing the urgency of the situation, the patient was immediately referred to a tertiary care center for emergency surgery, including repair of the large ascending aortic aneurysm and a second bioprosthetic aortic valve replacement (BVR). The surgery was successful, and the patient tolerated it well, with no significant postoperative complications.

Discussion

The case report demonstrates the challenges in diagnosing BAV, especially in asymptomatic individuals, which can potentially result in delayed detection and subsequent complications. Factors such as limited awareness among healthcare providers, lack of access to screening, and the asymptomatic nature of BAV contribute to underdiagnosis, particularly in low-resource settings [6]. This case report also highlights BAV as a significant global health concern, especially in regions with limited access to cardiac screening and care, necessitating increased awareness, screening efforts, and access to cardiac care, particularly in resource-limited regions. Personalized medical management is essential for BAV-related conditions, which may involve close monitoring, medication, aortic valve interventions, or surgery to address related aortopathy [7].

The exact cause of BAV disease remains unclear, and it has been associated with various genetic syndromes (Shone complex, Kabuki syndrome, Marfan syndrome) as well as genetic variations and mutations [8,9]. Genes essential for heart development, such as GATA4, GATA5, GATA6, and NKX2-5, play key roles, and mutations in these genes can disrupt normal heart formation [10]. Genes involved in neural crest cell migration, such as ROBO4, are crucial for proper valve development [11]. Disruptions in genes that regulate the extracellular matrix (ECM), including NOTCH1, TGFBR1/2, and SMAD6, can also contribute to BAV by affecting the developing valve's structural integrity and signaling environment [10]. Furthermore, BAV can also be associated with connective tissue disorders caused by mutations in genes like FBN1, leading to weakened aortic tissue [12]. Understanding the genetic basis of BAV is important for identifying individuals at risk globally, implementing early screening, and guiding management strategies. Addressing genetic factors associated with BAV requires a collaborative global effort, with increased focus on research, awareness campaigns, and broader access to clinical interventions to improve outcomes and reduce the burden of BAV-related complications.

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Bicuspid aortic valve (BAV) often leads to complications like aortic stenosis, aortic regurgitation, endocarditis, bicuspid aortopathy (aortic dilatation or aneurysms), aortic coarctation, patent ductus arteriosus, and coronary artery problems [13]. This case emphasizes the significant link between bicuspid aortic valve (BAV) and aortopathy, particularly ascending aortic (AA) aneurysms. It shows that aortopathy in BAV patients can progress independently of the severity of aortic valve dysfunction, as the patient needed urgent aneurysm repair despite the potentially protective effect of bioprosthetic aortic stenosis.

Due to BAV's genetic predisposition and associated hemodynamic changes, early identification and regular monitoring of first-degree relatives (parents, siblings, children) are essential. The prevalence of BAV among first-degree relatives of individuals with BAV stands at approximately 10–15% [14]. Guidelines recommend regular monitoring of BAV patients and their first-degree relatives for aortopathy, underscoring the importance of early detection and intervention to mitigate complications. Deciding when to perform surgery on BAV patients is complex and individualized, considering multiple factors [15-17]. The 2022 ACC/AHA guidelines strongly recommend surgery if the aortic diameter reaches 5.5 cm or more. In exceptional cases (aorta between 5.0-5.4 cm), earlier surgery might be considered depending on additional risk factors such as a family history of dissection, rapid growth, aortic coarctation, or "root phenotype" aortopathy. Surgery with an aortic diameter of ≥4.5 cm is also recommended for aortic valve replacement or repair [18]. While existing guidelines provide a framework for care, recent studies offer conflicting views on whether early surgery might benefit BAV patients without apparent symptoms [19]. While bioprosthetic valve implantation can be effective, patients should be aware that structural valve deterioration (SVD) can begin as early as eight years post-procedure, significantly accelerating around the 10-year mark [20-22].

To address the global challenge of BAV disease, more specialized BAV clinics are needed within existing multidisciplinary aortic teams (MATs). Recently, some centers, like the Melman Comprehensive Bicuspid Aortic Valve Program at the Bluhm Cardiovascular Institute and the University of Calgary BAV Clinic, have established standalone clinics specifically for BAV [23]. In these clinics, a 'heart team' must make decisions based on agreed-upon guidelines, the clinic's protocols, and each patient's unique situation. Training and resources should also be provided to help regular clinics become experts in BAV, fostering international collaboration for knowledge exchange. Increased research funding should prioritize better risk prediction, such as blood biomarkers [24], for predicting rapid BAV disease progression. To find ways to slow aortic dilation in BAV patients, clinical trials of potential drug therapies and global BAV databases to facilitate large-scale studies are needed. Finally, to promote equity, telemedicine can connect underserved areas with specialized care, while knowledge-sharing and training programs should focus on building capacity in lower-resource regions.

This case illustrates the challenges faced by BAV patients, where even successful treatment doebarrrsn't eliminate the need for lifelong vigilance. Therefore, long-term aortic surveillance and medication adherence are essential in BAV patients, even after valve replacement, regardless of symptoms. Several potential barriers to treatment adherence warrant careful consideration, including economic factors (cost of regular follow-up and imaging), logistical challenges (access to specialized care), and patient understanding of their condition's severity and the importance of surveillance. Additionally, the psychological impact of living with BAV, potentially including anxiety or fear related to future complications, should be addressed. As the patient ages, caregivers or a support network may become essential in ensuring appointment attendance, medication adherence, and emotional support, all of which are crucial for successful long-term BAV management. Open communication between the healthcare team, the patient, and their caregivers is essential to identify individual barriers and collaboratively develop strategies to promote optimal treatment adherence. By addressing these barriers comprehensively, healthcare providers can enhance patient engagement and improve long-term outcomes in BAV management.

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Conclusion

While BAV presents ongoing challenges, this case highlights the importance of timely diagnosis, personalized care, surveillance, and addressing barriers to adherence for optimal long-term outcomes. Timely detection and management of bioprosthetic valve deterioration and aortopathy can help prevent life-threatening complications and the need for high-risk emergency surgery.

Patient's Perspective

I was shocked and saddened when the doctor told me that I had a bicuspid aortic valve, as I had never experienced any symptoms beforehand. The possibility of undergoing surgery scared me a lot. But with the help of my family and the medical team, I decided to face it bravely. Recovery from surgery was difficult; adhering to new medicines caused exhaustion. However, this experience instilled a sense of daily gratitude and appreciation.

Managing BAV requires constant care and adhering to treatments, which can be demanding sometimes. Despite this, I'm committed to not letting it define me. I proactively manage my condition, seeking support and resources whenever I need them. The future holds uncertainty, but I approach it with resilience and hope. This experience taught me the importance of staying strong and relying on the community for support. If you are newly diagnosed, know you are not alone. I'm grateful for the care received and the lessons learned along the way. Every day is precious; I cherish each one.

Consent: Written consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-chief of this journal on request.

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