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## Case Report

# Late Diagnostic of Bicuspid Aortic Valve with Severe Aortic Stenosis and Large Thoracic Aortic Aneurysm: Case Report and Review of the Literature.

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### Abstract:

Bicuspid aortic valve (BAV) is the most common congenital aortic valve anomaly and affects approximately 1%–2% of the population. Individuals with BAVs have higher risks of valvular dysfunction, endocarditis, and ascending aortic aneurysm and dissection than individuals with tricuspid aortic valves.

We report a fatal outcome of ascending aortic aneurysm in a women patient with bicuspid aortic valve complicated to aortic stenosis. The patient was qualified for elective surgery of replacement of the affected aorta with composite aortic valve-supracommissural ascending aorta. Unfortunately, patient ended up fatally during the open heart surgery.

Congenital BAV are common and in most cases remain undetected, BAVs may progress and become calcified, thus leading to varying degrees of severity of aortic stenosis. The thoracic aortic aneurysm in BAV frequently involves the proximal aorta, including the aortic root, ascending aorta, and aortic arch. While the ascending aortic aneurysm might be affected by both aortopathy and hemodynamics. Our patient, regrettably, was not diagnosed on time with BAV, thus preventive measures were not applied on him can avoid its serious consequences.

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### Introduction:

Bicuspid aortic valve (BAV) is the most common congenital aortic valve anomaly and affects approximately 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 [2]. However, BAV disease is not only a disorder of valvulogenesis; it probably represents a more global disorder of cardiac and vascular morphogenesis.

Diagnosis of many patients born with BAV does not happen until adulthood, however, up to 50–70% of patients with BAV experience some form of complication such as valvular dysfunction, endocarditis, and ascending aortic aneurysm and dissection [3]. The most common abnormality associated with BAV is dilatation of the thoracic aorta [4,5]. Svensson et al. showed that 20% of patients who underwent BAV surgery had a concomitant ascending aortic aneurysm that required surgery [6]. While the ascending aortic aneurysm in BAV might be affected by both aortopathy and hemodynamics and there is frequently involves the proximal aorta, but spares the aorta distal to the aortic arch [7].

### Case presentation:

We report the case of a 64-year-old women was referred to our cardiology department for the management of a progressively worsening dyspnea. She is at high cardiovascular risk (Age, high blood pressure and dyslipidemia), For the previous 2 years she had presented a Ischemic stroke that were never investigated. Echocardiography revealed severe aortic stenosis due to bicuspid aortic valve (BAV) with moderately severe concentric LV wall hypertrophy, an apical aneurysmal of the left ventricular, reduced ejection fraction and significant aortic arch dilatation. Holter monitor showed paroxysmal atrial fibrillation. Coronary angiography was normal. Patient was referred immediately for surgical intervention, replacement of the affected aorta with composite aortic valve-supracommissural ascending aorta. Unfortunately, patient ended up fatally during the open heart surgery.

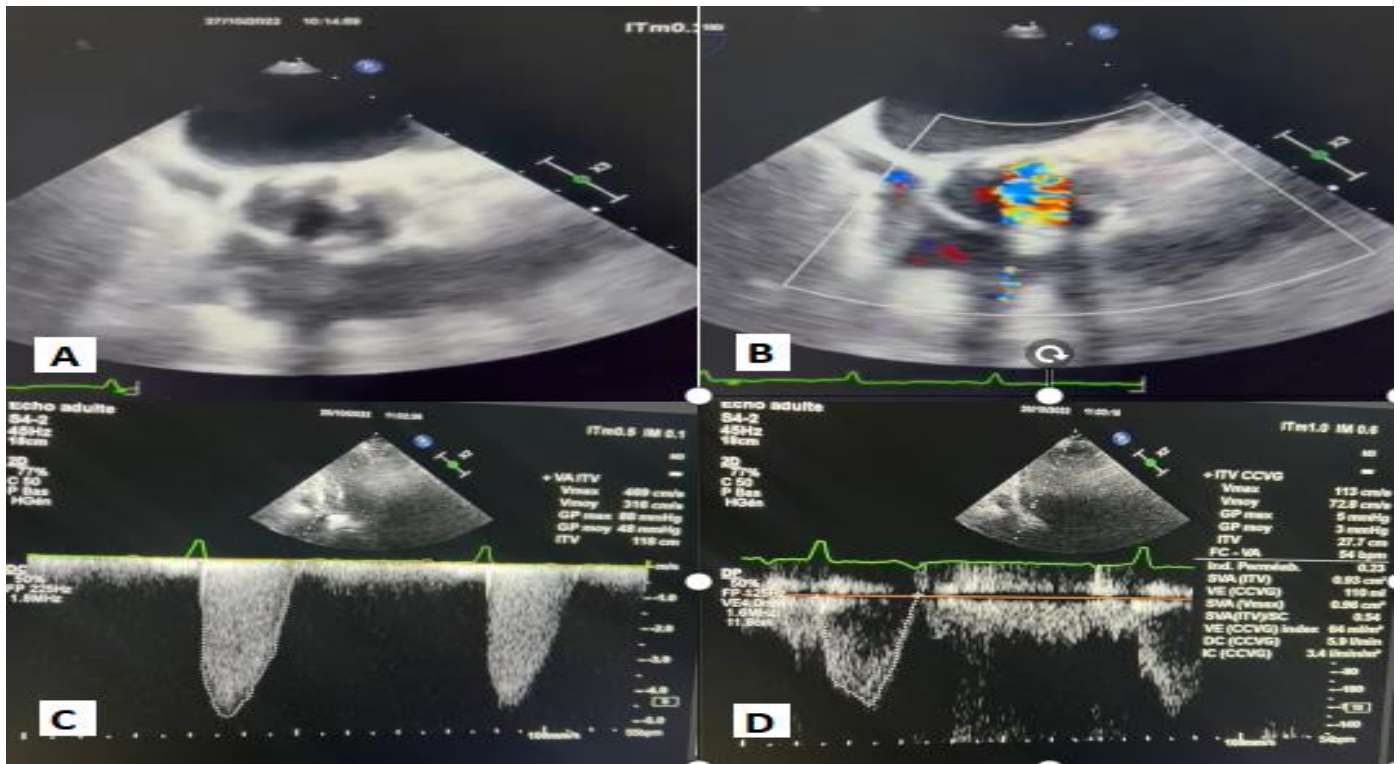


Figure 1: Transthoracic and transesophageal echocardiography images shows a BAV type 0 (A,B), and sévère aortic sténosis (C,D).

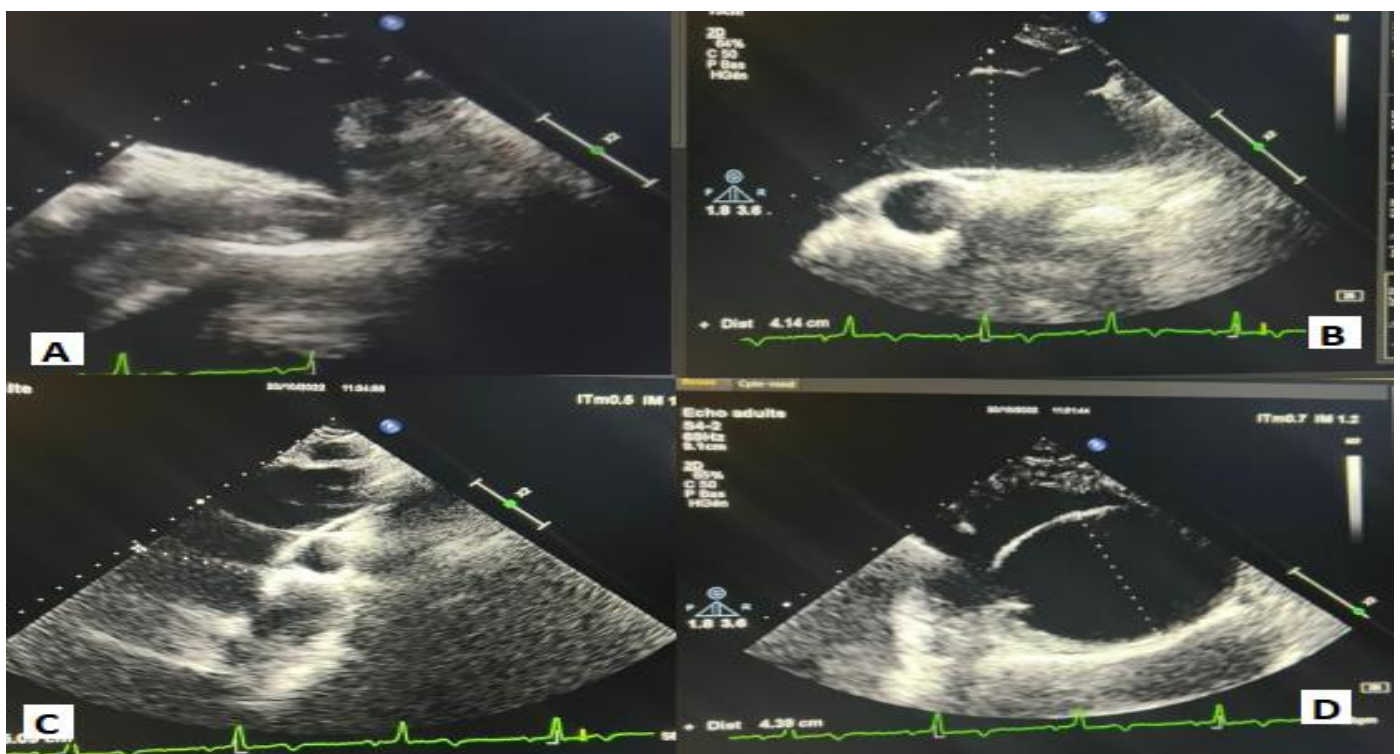


Figure 2: Echocardiography image of aorta: A, B and C Transthoracic echocardiography image on suprasternal view shows Aneurysm of Aortic Arch confirmed by transesophageal echocardiography.

**Discussion:**

Congenital BAV are common and in most cases remain undetected, until infection or calcification supervenes and 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 [8]. In our reported case, there was presented with progressively worsening dyspnea related to AS due and BAV that was diagnosed late at the age of 64 years.

BAV is recognized as a frequent cause of aortic stenosis in adults. Aortic stenosis has been found in 72% of adults with BAV. the

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stenotic valves were obstructed by nodular, calcareous masses, but commissural fusion was present in only eight cases [8]. In this case, there was a diffuse calcification of valve leaflet without commissural fusion.

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%) [9]. 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 [10].

In patients that present with valvular dysfunction earlier in life (<50 years old), Aortic Inuffisance is more common. however, later in life (>50 years old) aortic stenosis (AS) is more prevalent [11].

The prevalence of aortic dilation in BAV is reported from 20% to 84%, depending on the population studied and the definition of aortic dilation [12,13]. The mechanism responsible for the occurrence and progression of aortic dilatation has also not yet been elucidated in detail. The researchers suggest two theories for the cause of aneurysms in patients with BAV: the hemodynamic theory and the aortopathy theory. The exact molecular and cellular pathways involved in BAV aortopathy remain unknown. However, MMP-2 (matrix metalloproteinase-2) has been identified as a key molecular modulator and a circulation biomarker of aortic dilation in patients with BAV. An increase in MMPs, enzymes that process or degrade the extracellular matrix, is associated with the development of aortic aneurysms. A study of patients with TAA comparing patients with BAVs and patients with TAVs, found that MMP-2 was increased by 34% in patients with BAVs [14]. Therefore, an increase in collagen turnover and a decrease in collagen crosslinking may be a factor in the formation of aneurysms in patients with BAVs [15].

While aortopathy is important in TAA formation in patients with BAVs, the hemodynamic theory cannot be ignored. BAVs compared to TAVs alter the hemodynamic blood flow through the valve, diversely impacting the aorta, leading to different locations of dilation. While the fusion subtypes contribute to flow patterns, so do valvular dysfunctions, such as AI and AS. Most frequently, the hemodynamic change in patients with BAV affects the ascending aorta, but not the aortic root. AS creates a high-velocity jet that increases shear stress on the ascending aorta [16]. Those with BAV and a dilated aorta are at risk for type A aortic dissection [17-19], and there require lifelong surveillance of the aortic root and ascending aorta. In a prospective study of 90 adults with BAV, the mean increase in ascending aortic diameter was 0.47 mm/y (range, 0.2–2.3 mm/y) over a 4.8-year follow-up [17]. Surveillance imaging such as TTE, a CT or MR can document current aortic diameters and permit calculation of aortic growth rates [20,21]. Among a cohort of adult patients with BAV (mean age, 55-17 years) without a TAA at baseline (ie, the baseline aortic diameter was <4.5 cm), 13% went on to develop a TAA at 14,6 years after diagnosis, and the 25-year risk of TAA was 26% [13]. For many adults, an aortic root, ascending aortic, or both diameter >4.0 cm is considered dilated and should therefore be monitored overtime with surveillance imaging to detect progressive dilation. The timing of surgery to replace the aorta in BAV disease depends on the morphology and diameter of the aorta, aortic valve function, rate of aortic growth, family history, patient characteristics, patient wishes, and the expertise of the surgeon and institution [22,23]. Aortic valve replacement is indicated for severe valvular dysfunction, symptomatic patients, and/or those patients with evidence of abnormal LV dimensions and function [24]. 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, and is recommended instead of treating BAV as a homogenous disease, it should be treated based on the different subtypes and associated valvulopathy/aortopathy. Cluster A BAV (malignant form) root phenotype with aortic root aneurysm and AI should be treated more aggressively with surgical resection at 5 cm for asymptomatic patients, as for patients with MFS, while Cluster B BAV (benign form) without aortic root aneurysm could be treated less aggressively as for patients with a trileaflet aortic valve with surgical resection at 5.5 cm for asymptomatic patients. Concomitant elective surgery of the aorta should be

considered in both Cluster A and B when undergoing clinically indicated AVR and the aorta measures  $\geq 4.5$  cm [25].

Our patient, regrettably, was not diagnosed on time with BAV, thus preventive measures were not applied on him can avoid its serious consequences.

**Conclusion :** The bicuspid aortic valve is the most common congenital cardiac anomaly but in most cases remain undetected. The case report shows a course of a BAV and its most serious complications, aortic stenosis and aneurysm aortic. Optimal management of patients with BAV disease and associated ascending aortic aneurysms often requires a thoughtful approach, carefully assessing various risk factors of the aortic valve and the aorta and discerning individual indications for ongoing surveillance, medical management, and operative intervention.

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