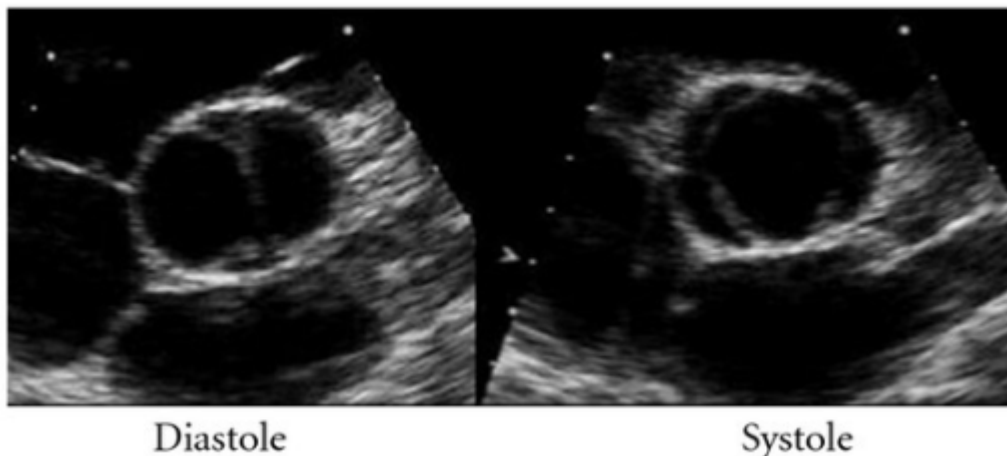


# Bicuspid Aortic Valve Disease — Symptoms and Treatment

[See online here](#)

**BAV is an aortic valve that only has two leaflets instead of three, and it's considered the most frequent cause of isolated aortic stenosis in adult patients. 1 - 2% of the population have BAV diseases and it is twice as common in males as in females. The majority will cause no problems, however, especially in later life, a bicuspid aortic valve may become calcified, leading to varying degrees of aortic stenosis and aortic regurgitation, which will manifest as murmurs. BAV might be associated with other cardiovascular anomalies, most commonly aortic coarctation.**

Bicuspid aortic valve-anterior-posterior commissure



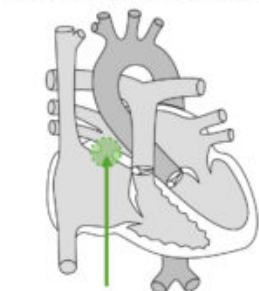
## Definition of Bicuspid Aortic Valve Disease

Bicuspid aortic valve (BAV) is an **inherited** form of heart disease where the aorta has two leaflets (bicuspid valve) instead of the normal three-leaflet valve. Most of the patients have one line of **coaptation** with a **low raphe**.

BAV may also occur with other cardiovascular anomalies, which include **coarctation of the aorta**, which is considered the most common congenital anomaly, **dilatation of aortic root** with a resultant **aortic aneurysm**, which may progress uncommonly into **aortic dissection**.

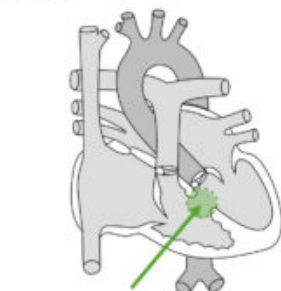
Less commonly, BAV may be associated with **other congenital heart diseases** like a **ventricular or atrial septal defect**, **hypoplastic left heart syndrome**, **patent ductus arteriosus**, and **Ebstein's anomaly**. The diagnosis is usually an accidental finding during an echocardiogram.

### Left to right shunting within the heart

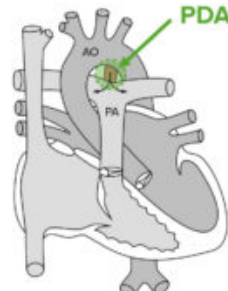


Atrial septal defect

Complete atrioventricular canal



Ventricular septal defect



Patent ductus arteriosus

Total anomalous pulmonary venous connection

Truncus arteriosus

## Epidemiology of Bicuspid Aortic Valve Disease

Bicuspid aortic valve disease is one of the prominent causes of death worldwide and considered one of the most common causes of inherited heart diseases.

### Prevalence in the United States

It has been estimated that 1 – 2% of the population in the United States of America have bicuspid aortic valves. Moderate to severe **aortic regurgitation** was the most common concomitant lesion in 12%, while mild **aortic stenosis** occurs in 5% only.

**Race:** It has been suggested in a report that BAV is less prevalent in African-Americans.

**Sex:** BAV is more common in males with the male-to-female ratio being 3:1.

**Age:** Bicuspid aortic valve may be present in patients of any aged person, ranging from birth to 11th decade.

## Classification of Bicuspid Aortic Valve Disease

BAV is composed of two unequal leaflets, where the larger leaflet is called a **conjoined leaflet**, and two commissures are most commonly present in the anteroposterior direction giving **Lt. & Rt. cusps**. The commissures can present less commonly on the Rt. & Lt. side of the annulus resulting in anterior and posterior cusps. The presence of partially fused commissure is called a **“high raphe,”** which usually predisposes the valve for stenosis.

BAV has **heterogeneous morphologic phenotypes and function**, so it results in **different treatment strategies**.

Three characteristics of a systematic classification are found:

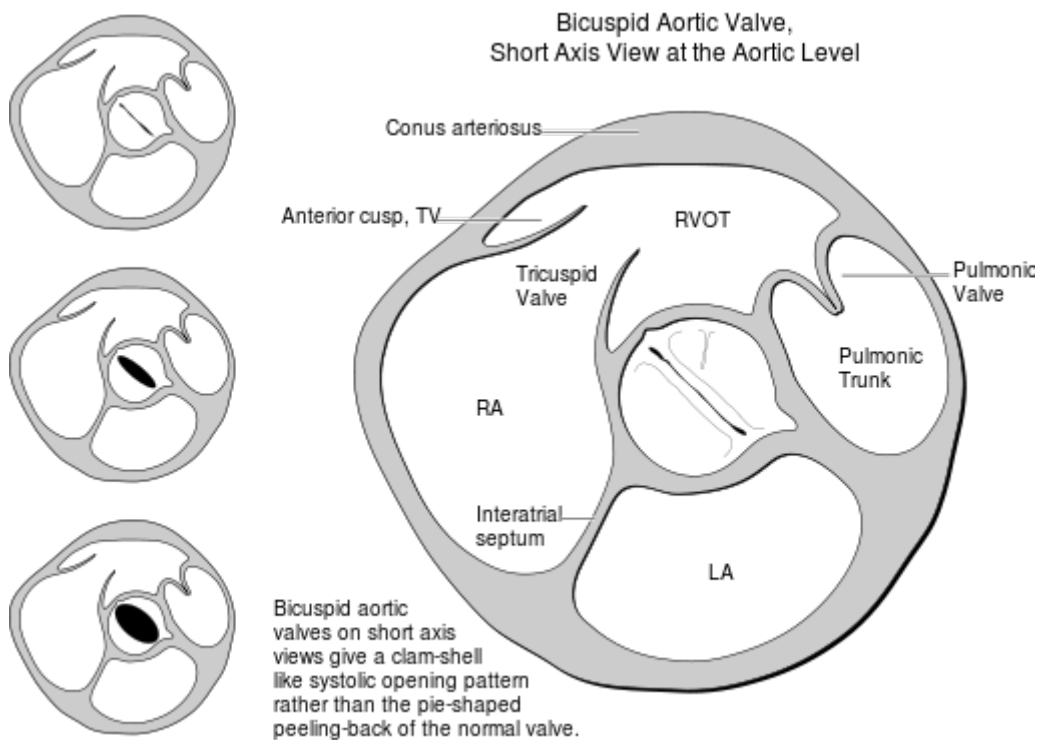
- Number of raphes
- The spatial position of cusps or raphes
- Functional status of the valve

The first characteristic is the most significant one and therefore it is termed as “the type.” It is further divided into 3 major types:

- Type 0 (no raphe)

- Type 1 (one raphe)
- Type 2 (two raphes)

The other two are supplementary characteristics.



*Image:* "Heart Bicuspid Aortic Valve Anatomy." by Patrick J. Lynch, the medical illustrator. License: [CC BY 2.5](https://creativecommons.org/licenses/by/2.5/)

## Pathophysiology of Bicuspid Aortic Valve Disease

The bicuspid aortic valve may result from **the inadequate production of fibrillin-1** during the process of the **valvulogenesis**.

The BAV can be **competent** with no regurgitation, and can also become **incompetent** as a result of **aortic dilatation**, prolapsed of cusp or underlying **infective endocarditis**.

**The fusion of aortic valve leaflets** commonly (up to 80%) occurs between RT. coronary and LT. coronary leaflets, where this type of fusion is less commonly associated with stenosis or regurgitation in children. The **fusion between the RT. coronary and non-coronary leaflets** is more commonly associated with future **aortic valve regurgitation and stenosis** in children. Fusion occurs least commonly between the non-coronary and LT. coronary leaflets ( $\approx 2\%$ ).

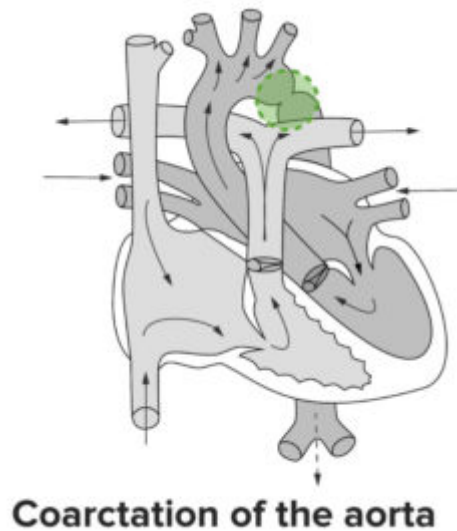
Recent studies pointed towards an association between **NOTCH1** mutations and BAV.

## Clinical Features of Bicuspid Aortic Valve

### Symptoms

Patients with a normal functioning bicuspid aortic valve can be completely **asymptomatic**. Symptoms usually occur as a result of the development of **aortic stenosis**, **aortic regurgitation** or both. If BAV is associated with **coarctation** or

**interrupted aortic arch**, symptoms will usually develop as a result of these underlying associated conditions, rather than due to the BAV itself.



Those who do have complications may experience:

- Dyspnea
- Fatigue
- Tachycardia
- Light-headedness
- Fainting

Note:

**Uncommissural valve** is usually associated with a critical form of aortic stenosis and may manifest with **severe congestive heart failure** in early infancy.

## Signs

Clinical examination is usually normal in patients with BAV, and the diagnosis is usually discovered accidentally by the echocardiography.

### General examination

Generally, look at features of inherited disorders such as: **Williams syndrome** (e.g., elfin facies with mental retardation) or **Turner syndrome** (female with short stature, webbed neck, and broad chest).

### Cardiac examination

- **Normal precordium** examination
- 1st heart sound usually unaffected
- 2nd heart sound usually has normal splitting with inspiration. Splitting of the 2nd heart sound starts to disappear with more aortic stenosis up to **paradoxical splitting** in very severe stenosis.
- **Medium-pitched systolic ejection clicks** during all phases of respiration, just after the 1st heart sound can be heard in the apex.
- Aortic stenosis may develop a **soft, harsh ejection systolic murmur** at the first aortic area with **possible radiation into the carotids**. With more stenosis, **thrills** can be palpable.

# Differential Diagnosis

Following are the related general differential diagnoses:

- **Coarctation of the aorta:** A section of the aorta is narrowed, especially the descending part of the aorta.
- **Interrupted aortic arch:** The ascending and descending aorta is completely separated and there is a gap between them. The aortic valve is normal, unlike in a bicuspid aortic valve.
- **Pediatric aortic valve insufficiency:** The blood comes back to the left ventricle after ejection into the ascending aorta, but, in bicuspid aortic valves, blood does not flow back into the left ventricle unless a complication has developed.
- **Pediatric rheumatic heart disease:** can result in rheumatic aortic stenosis (AS) or aortic regurgitation (AR) and presents with similar clinical findings to the bicuspid aortic valve.

## Investigation for Bicuspid Aortic Valve

### Chest X-ray

- May show prominent ascending aorta
- Cardiomegaly due to LV enlargement can be explained by the occurrence of aortic regurgitation.
- Not helpful in the screening of BAV



[Image:](#) "Left ventricular hypertrophy in short axis view." by Patrick J. Lynch, medical illustrator. License: [CC BY 2.5](#)

### ECG

Usually shows non-specific changes, such as:

- Left ventricular hypertrophy
- Atrial enlargement
- [Arrhythmias](#)

## Echocardiography

Echocardiography was considered the primary tool in the diagnosing of BAV.

- The reliability of diagnosis for BAV has been improved significantly by the presence of **cross-sectional and Doppler echocardiography** which has a specificity of 96%, a sensitivity of 78%.
- Diagnosis of BAV can be confirmed by the visualization of **two cusps and two commissures** using short-axis view.
- Other supportive features include:
  - Cusp redundancy
  - Eccentric valve closure
  - Single coaption line between cusps during diastole.

## MRI and CT

In 2015, an article was published that discussed the optimal diagnostic utility in confirming the diagnosis of the bicuspid aortic valve. The following observations were noted in this review article:

- Cardiovascular magnetic resonance (CMR) imaging is superior to echocardiography in assessing the morphology of BAV (96 versus 73%).
- Echocardiography is unable to confirm the diagnosis of BAV in many cases because of the severity of the condition and extensive calcification.
- The current standard of care for a patient with BAV is to perform serial CMR to evaluate the progression of the disease.
- Multi-slice computed tomography is helpful in evaluating the size of the BAV.
- The gold standard for assessing the severity of stenosis in BAV is the clinical judgment of the surgeon.

## Complications of Bicuspid Aortic Valve

Normal functioning BAV can undergo **gradual damage** due to continuous abnormal stresses on the valve; therefore, the complications which may occur with BAV are aortic stenosis, aortic regurgitation, and infective endocarditis.

### Aortic stenosis

AS is considered the **most common surgical complication** in patients with BAV and most patients will require surgical intervention in the middle age. Stenosis of a bicuspid aortic valve has a higher chance to develop in individuals older than 20 years.

It is caused by progressive **sclerosis**, **calcification** and eventually **stenosis**. A **high level of serum cholesterol** is associated with more risk to develop progressive sclerosis of the bicuspid aortic valve.

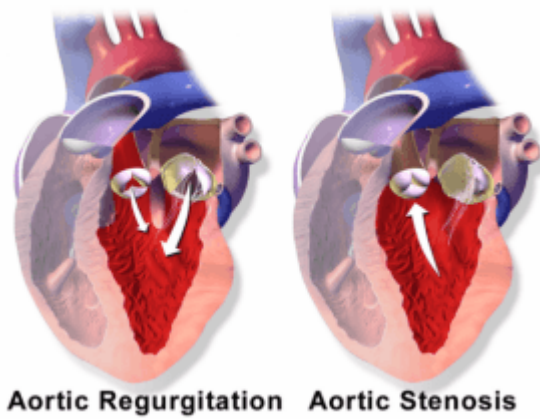


Image: „Aortic Valve Regurgitation vs. Aortic Valve Stenosis“ by BruceBlaus. License: [CC BY 3.0](https://creativecommons.org/licenses/by/3.0/)

## Aortic regurgitation

AR is another common complication of BAV that may present alone or concomitantly with AS. AR usually develops as a result of **prolapse of aortic cusps, dilated root of the aorta** or underlying **infective endocarditis**.

## Infective endocarditis

Infective endocarditis incidence is more in bicuspid aortic valve than the normal tri-leaflets valve (Incidence is about 2%). The outcome result of infective endocarditis in BAV is generally worse than the normal aortic valve.

## Management of Bicuspid Aortic Valve

The bicuspid aortic valve has primarily a **surgical treatment**. Many other aspects have to be considered during the evaluation of the most appropriate surgical interventions in patients with BAV. Most of the patients having this disease are young when the surgery is considered; therefore, they will live a longer life which can lead them to an increased risk of valve-related morbidities, such as:

- Valve thrombosis
- Thromboembolism
- Hemorrhage after mechanical valve implantation.

Accurate assessment of BAV morphology should reveal the size of ascending aorta; the extent and rate of aortic dilatation progression to determine the appropriate surgical approach for each individual; therefore, an **effective risk assessment** of the difficulty of the surgical procedure and associated comorbidities is required and it should be considered during the decision-making process.

**A regular follow-up assessment** is important for the recognition of possible complications and the prevention of progressive deterioration of the aortic valve with age.

### General measures

**High cholesterol level** is considered a risk factor that may accelerate the sclerosis of BAV and deterioration of the valve condition predisposing to undesirable complications;

therefore, a strict healthy diet is important for limiting fat calories up to 30% of total calories.

Patients with BAV don't need to restrict their activities as long as the valve is working properly. Those who develop AS or AR may need to avoid strenuous exercise.

### Medical treatment

- No specific medical treatment can be useful for BAV.
- Medical therapy can only be used for the treatment of existent cardiac problems such as **heart failure**.
- The American Heart Association (AHA) is no more recommending prophylactic treatment against IE unless the patient has a previous episode of IE.

### Surgical management

The definitive treatment for the bicuspid aortic valve (BAV) with severe AS or AR is **aortic valve replacement**. The guidelines for indications of surgery of BAV are the same as of aortic stenosis and regurgitation. **Transcatheter aortic valve replacement** is the standard procedure for the treatment of severe BAV stenosis.

**Regular follow-up echocardiography** to assess aortic root is important to guide for the possibility of elective aortic root surgery.

Based on ESC guidelines for the treatment of valvular heart diseases, surgery is indicated in any patient with aortic root diameter  $\geq 55$  mm.

Surgery with aortic root diameter  $\geq 50$  -54 mm in patients with BAV should be considered if associated with any:

- **Family history of aortic dissection** or unexplained sudden cardiac death
- Rapid growth progression of **aortic dilatation**
- Others (**hypertension**, associated **aortic coarctation**, a female patient requiring **pregnancy**)
- Increase in aortic diameter  $\geq 2$  mm/year in follow-up examinations.

Surgery with aortic root diameter  $\geq 45$  mm in patients with BAV should be considered if another concomitant valvular surgery is indicated to be done.

## References

[Aortic Dilatation in Patients with Bicuspid Aortic Valve](#) via nejm.org

[A classification system for the bicuspid aortic valve from 304 surgical specimens](#) via nih.gov

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