

BICUSPID AORTIC VALVE

Bicuspid aortic valve (BAV) is a defect of the aortic valve that is present at birth. The aortic valve allows oxygen-rich blood to flow from the heart to the aorta. The aorta is the major blood vessel that brings blood to the body. Normally, there are three valve flaps (leaflets) in the aortic valve. In a person who has a bicuspid aortic valve, there are only two valve flaps (leaflets) in the aortic valve. Valves are passages for the blood to pass through before leaving each chamber of the heart. The valves prevent blood from flowing backwards into the heart.

Bicuspid aortic valve is one of the most common congenital heart defects, appearing in 4.6 of every 1,000 live births.



In bicuspid aortic valve, the aortic valve may not be able to completely stop blood from leaking back into the heart (aortic regurgitation). The aortic valve may also become stiff and not open as well, causing the heart to have to pump harder than usual to get blood past the valve (aortic stenosis). The ascending aorta also may become enlarged (dilated or aneurysmal) with this condition.

What other names do people use for bicuspid aortic valve?

Bicuspid aortic valve (BAV) is also sometimes called bicommissural aortic valve.

How prevalent is bicuspid aortic valve?

Bicuspid aortic valve is one of the most common congenital (at birth) heart defects, appearing in 4.6 of every 1,000 live births. Bicuspid aortic valve is more common in males than females. The prevalence of BAV is 7.1 in 1,000 male babies, and 1.9 in 1,000 female babies. It is estimated that 1-2 percent of the population is affected by BAV.

What are the characteristics of bicuspid aortic valve?

People with bicuspid aortic valve can have abnormal coronary arteries, aortic aneurysm, or an abnormal thoracic aorta (the portion of the aorta that passes through the upper chest), as well as unstable high blood pressure.

Some of the signs of bicuspid aortic valve include:

- Enlarged heart
- Heart murmur (systolic click, aortic stenosis, aortic regurgitation)
- Weak pulse in the feet and ankles if associated with a coarctation of the aorta

While there are often no symptoms in childhood, there are some children with severe bicuspid aortic valve who experience symptoms of the disease. If the aortic valve becomes narrowed, a child may tire easily, have chest pain, difficulty breathing, rapid and irregular heartbeat (palpitations), loss of consciousness (fainting), or pale skin. A severely leaking bicuspid aortic valve can lead to symptoms of heart failure, including difficulty breathing.

What causes bicuspid aortic valve?

The exact cause of bicuspid aortic valve is not yet known. Bicuspid aortic valve is often seen in babies with coarctation (narrowing) of the aorta and other diseases in which there is a blockage of blood flow on the left side of the heart. Bicuspid aortic valve disease may have an underlying genetic defect, but no one specific gene defect is related to bicuspid aortic valve disease.

Bicuspid aortic valve may exist alone or it may be associated with other congenital heart defects. Importantly, bicuspid aortic valve can also be present in aortic aneurysm syndromes, including Loeys-Dietz syndrome and familial thoracic aortic aneurysm syndromes related to other gene mutations.

Bicuspid aortic valve is highly heritable, and may be entirely determined by genetics. In large family studies, approximately 9 percent of first degree relatives of the person with a bicuspid aortic valve are found to also have a bicuspid aortic valve. This suggests an autosomal dominant inheritance (only need to get the abnormal gene from one parent) with a variable expression (range of features).

How is bicuspid aortic valve diagnosed?

Bicuspid aortic valve can be diagnosed through medical testing, most often with an echocardiogram. Often it is not diagnosed in children because the bicuspid aortic valve can work properly for many years before any symptoms occur. It is possible that bicuspid aortic valve can be diagnosed in children with more severe cases.

A medical professional may perform a magnetic resonance imaging test (MRI) of the heart or an ultrasound of the heart (echocardiogram) to detect a bicuspid aortic valve. A transesophageal echocardiogram may also diagnose a bicuspid aortic valve.

How is bicuspid aortic valve managed?

People who have been diagnosed with bicuspid aortic valve should be under the ongoing care of a specialist in heart valve disease who can monitor changes in the heart, valves, and aorta over time.

Complications can include:

- Aortic regurgitation, the backward flow or leaking of blood back into the heart
- Stenosis, a stiffening of the aortic valve that makes the heart work harder to pump the blood through the valve
- Aortic aneurysm, a bulging or enlargement of the ascending aorta
- Aortic dissection, a ripping or tearing of the aorta
- Congestive heart failure, due to severe valve dysfunction
- Aortic coarctation, narrowing of the aorta

Treatments include:

For people with symptoms related to a severely malfunctioning bicuspid aortic valve, the treatment is to surgically replace (or in some instances repair) the valve. The majority of people with bicuspid aortic valve will require surgery to repair or replace the valve and, in many, the dilated part of the aorta.

There are two types of aortic valve surgery: aortic valve replacement and aortic valve repair. The surgery that is used to repair a bicuspid aortic valve is called bicuspid aortic valve repair; this is an operation that requires a surgeon with special expertise. When a bicuspid aortic valve is replaced, the types of valves that are most commonly used are a biological tissue valve and a mechanical aortic valve. Less commonly, human homograft valve replacement or the Ross procedure are performed for bicuspid aortic valve disease. Biological valve replacement involves replacing the aortic valve with a new valve made of tissue. A human homograft aortic valve replacement involves replacing the aortic valve with a new valve that has been removed from a donated human heart. Mechanical valve replacement involves replacing the aortic valve with a new valve which is made completely of mechanical parts.

Some people with leaking bicuspid aortic valves which do not require surgery are treated with medications including ACE inhibitors or ARB drugs in an attempt to lessen the leaking.

Some children are treated with catheter-based ballooning of a severely narrowed bicuspid aortic valve.

Association with aortic aneurysm:

Bicuspid aortic valve is one of the most common disorders associated with an enlargement or aneurysm of the ascending aorta. The enlarged aorta is believed to be caused by an underlying defect in the connective tissue of the aorta wall, and is not only related to a narrowed or leaking valve. In fact, the aortic enlargement or aneurysm may occur years after the bicuspid aortic valve is replaced. It is very important to carefully image the ascending aorta in people with a bicuspid aortic valve to evaluate for the presence of an aneurysm and to continue to image (with echocardiogram, CT scan, or MRI) over time. People with a bicuspid aortic valve and aneurysm are at risk for a tear in the aorta (aortic dissection) and this risk increases as the aorta enlarges further.

What is the life expectancy of someone with a bicuspid aortic valve?

There is no evidence that the life expectancy for a person with bicuspid aortic valve is shorter than the life expectancy for the general population.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.

Bicuspid Aortic Valve Disease

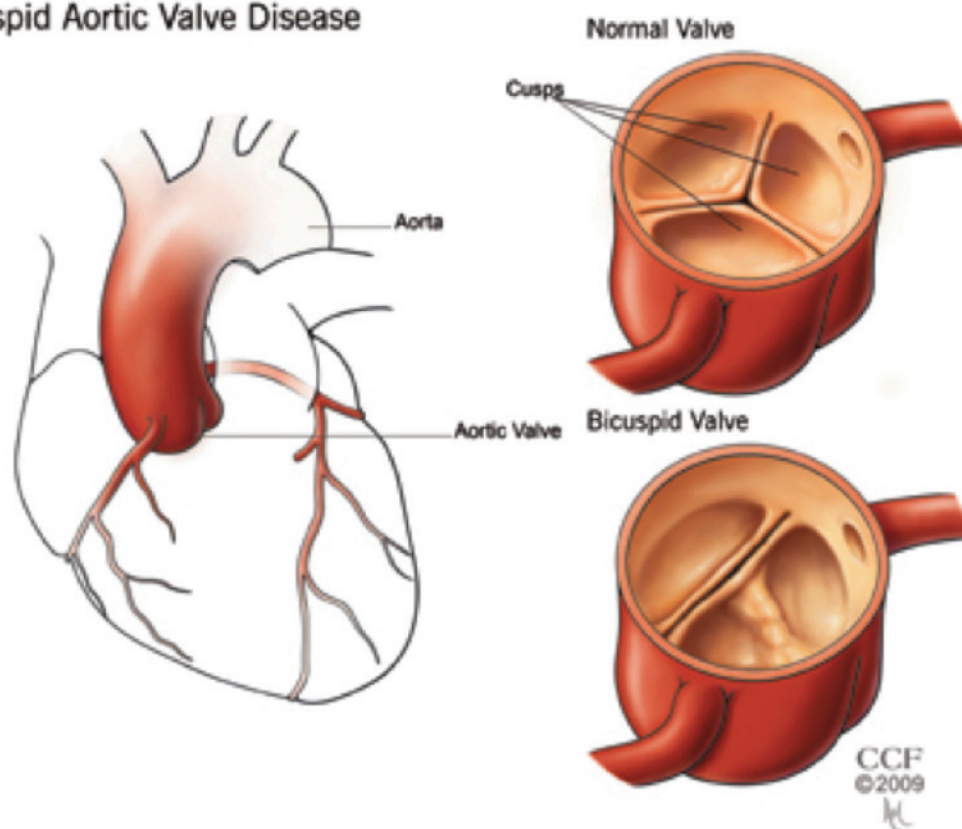


Figure courtesy of the Cleveland Clinic