

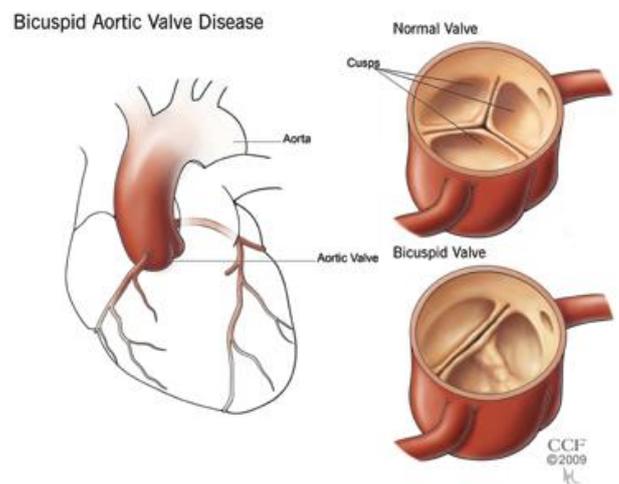
## Bicuspid aortic valve: information for families

The aortic valve sits at the base of the aorta – the blood vessel that takes blood from the heart to the rest of the body. The valve usually has three flaps or leaflets that open to let blood flow through to the aorta and close to keep it shut in between heartbeats. In bicuspid aortic valve, there are only two flaps inside the valve which affects how it functions. This information sheet from Great Ormond Street Hospital (GOSH) explains about the causes and symptoms of bicuspid aortic valve and how it can be treated.

Bicuspid aortic valve is a type of congenital (present at birth) heart defect.

It affects the aortic valve that sits between the aorta and the left ventricle (pumping chamber) of the heart. When the left ventricle squeezes to push oxygen-rich blood to the rest of the body, the aortic valve opens to let the blood through then closes to stop blood flowing backwards into the ventricle.

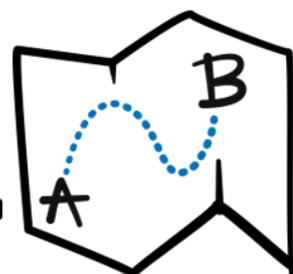
The aortic valve should have three flaps or leaflets but in bicuspid aortic valve, there are only two, which may also be malformed or not function as they should. This can cause narrowing of the valve if the leaflets cannot open fully (aortic stenosis). If the valve does not shut fully, oxygen-rich blood can flow back into the left ventricle (aortic regurgitation).



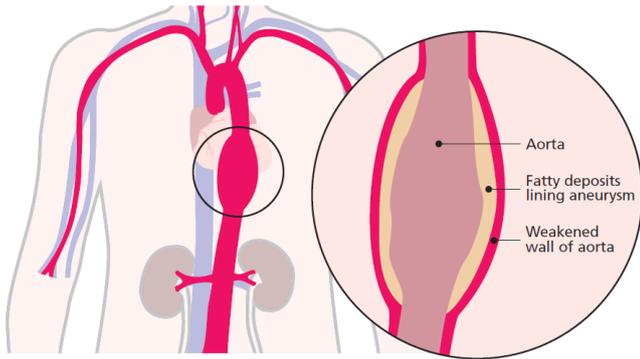
In addition to the problem valve, the walls of the aorta are weaker, which over time can lead to a bulge of aneurysm developing. The pressure of the blood travelling through the weakened section causes it to balloon outwards.

The severity of the aneurysm and how quickly it gets worse depends on two factors:

- The strength of the arterial wall – this in turn is influenced by the 'quality' of the connective tissue forming the arterial wall.



- The pressure being exerted on the arterial wall by the blood flowing through the artery – this is closely related to the heart rate and blood pressure.



## What causes bicuspid aortic valve?

Doctors do not fully understand what causes bicuspid aortic valve to develop during pregnancy. There may be a genetic component as it seems to run in some families. It can appear on its own or alongside other conditions such as Turner syndrome. It is more common in boys than girls but more research is needed to discover the reason for this. It affects around 1 in 100 people overall so is a relatively common type of congenital heart defect.

## What are the symptoms of bicuspid aortic valve?

The severity of symptoms depends on how well the bicuspid aortic valve functions and whether there are any other associated problems such as an aneurysm or narrowed section of aorta.

Most children do not show any symptoms – if they only have an abnormal valve and no other problems, the only symptom may be a heart murmur that can be heard with a stethoscope. A mild narrowing can also cause no problems in childhood.

If the valve function is poor, children may become breathless on exercise and have chest pain. They may not be able to be as active as other children of a similar age. Rarely, children may faint or feel dizzy if blood flow is reduced.

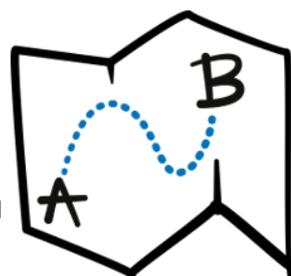
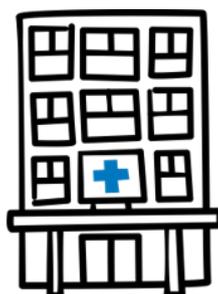
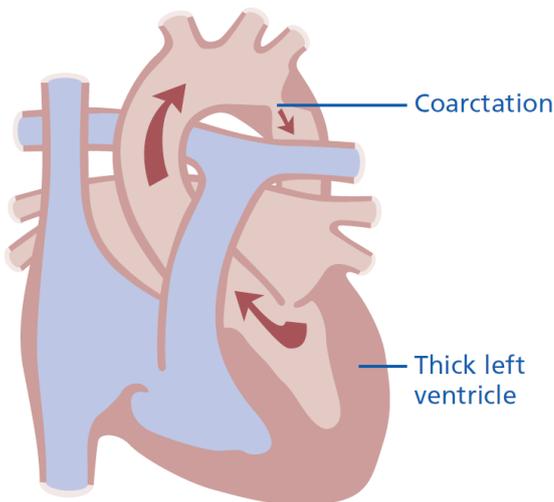
## How is bicuspid aortic valve diagnosed?

As well as a full physical examination, children will usually have an electrocardiogram (ECG) and echocardiogram (Echo) to look at their heart function. This may involve an exercise test to look at how the heart functions under stress. Imaging scans, such as a computed tomography (CT) or magnetic resonance imaging (MRI) scan may be used to look at and measure the diameter of all parts of the aorta. Cardiac catheterisation can also

In time, the weakened area can further dilate and eventually split and tear the layers of the arterial wall – this is referred to as ‘dissection’. The blood can then find its way within the wall layers from the torn area and extend the dissection causing severe pain and reducing the amount of oxygen reaching the body’s tissues.

The damaged wall is at high risk of rupture and consequent life threatening internal bleeding requiring emergency treatment, in the vast majority of cases involving open heart surgery.

The aorta may also have a narrowed section – called coarctation of the aorta – that means the left ventricle has to pump harder.



be used to look at the blood vessels and flow through them.

## How is bicuspid aortic valve treated?

If the valve is working relatively well and there is no narrowing or regurgitation, treatment may not be needed, although regular monitoring will be required to identify any problems as early as possible.

Medications to reduce blood pressure will reduce the stress on the bulging parts of the blood vessel(s) to reduce the risk of dilatation and eventually dissection. Specific types of medication include beta blockers, angiotensin converting enzyme (ACE) inhibitors and angiotensin receptor blockers (ARBs).

Narrowing of the valve or aorta can be managed using balloon dilatation. This is carried out during cardiac catheterisation. When the catheter is in place, the doctor blows up (inflates) the balloon carried inside it so that it stretches the narrowed section. Further X-rays are taken to check how much the balloon is inflated. At the end of the procedure, the balloon is deflated and with the catheter is brought back out through the blood vessel system.

Surgery to treat an aneurysm before dissection can be carried out through open chest surgery or endovascular surgery (repair from the inside of the blood vessel). Depending on the location of the bulge, surgery may involve a graft to replace the weakened portion.

If the aortic valve is damaged, the surgeon might decide to repair it or to replace it with an artificial valve. If a mechanical valve is used, the risk of blood clots developing, increases and anticoagulation (blood thinner) medication will be needed lifelong.

If dissection occurs, an aortic graft and/or valve replacement will be needed as an emergency operation – however, it is always preferable to carry out surgery before dissection occurs as the outcomes of surgery are significantly better when surgery is planned ahead and not carried out as an emergency.

## What is the outlook for children and young people with bicuspid aortic valve?

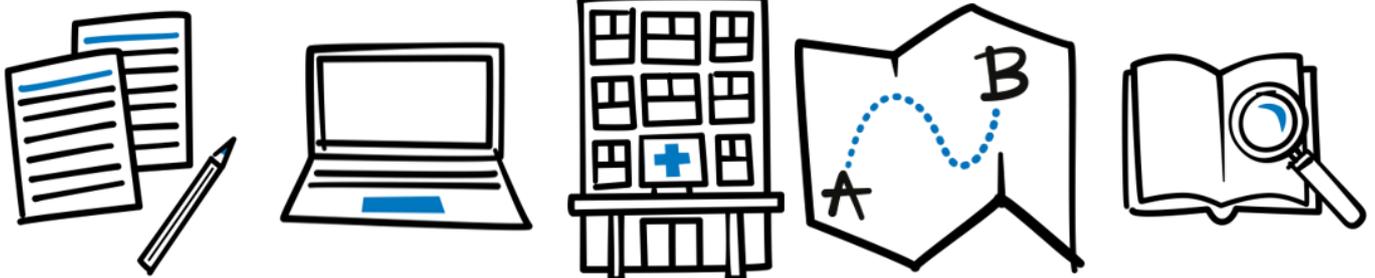
When bicuspid aortic valve has been diagnosed, life-long monitoring will be required and surgery planned when needed. As there is a genetic component to some cases of bicuspid aortic valve, screening of close family members (brothers, sisters, parents) might be suggested.

People with bicuspid aortic valve will have to make some lifestyle adaptations – contact sports and energetic sports will not be advisable due to the risk of dissection through direct trauma to the chest or sudden increases in heart rate and blood pressure. However, gentle exercise is possible and encouraged – the team will explore suitable options. For example, longer distance running at a steady pace is preferable to short sprint distances.

Pregnancy should be planned wherever possible so that the extra strain put on the blood vessel system by the developing fetus is monitored appropriately. Doctors may suggest surgical treatment of any aneurysms before pregnancy to reduce the risk of dissection.

A healthy lifestyle, eating a balanced diet, avoiding smoking and drinking alcohol in moderation should be followed and with regular monitoring as described above, children and young people can expect a normal lifespan.

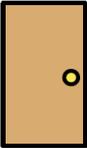
## Further information and support

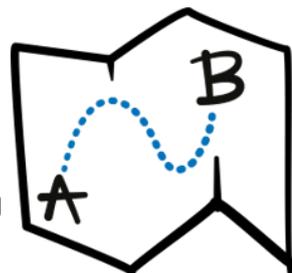


Contact the Inherited Cardiovascular Diseases Unit at GOSH. Call them on 020 7829 8839 or 020 7405 9200 ext 5124 or 5305 or 5139 or email: [icvd@gosh.nhs.uk](mailto:icvd@gosh.nhs.uk). You can also contact the team through MyGOSH once you have registered.

- The British Heart Foundation offers support and advice to anyone with heart problems – call their helpline on 0300 330 3311 or visit their website at [www.bhf.org.uk](http://www.bhf.org.uk)

## All about bicuspid aortic valve

	Your heart is in your chest. It squeezes and relaxes to pump blood around the body.
	The biggest blood vessel in the body is called the aorta (said: ay-or-ta). It carries blood from the heart to the rest of the body.
	There is a valve or door where the aorta joins the heart. Sometimes, this door is not formed properly.
	This can cause problems with blood flowing backwards or the heart having to pump harder to get blood through any narrow bits of the aorta.
	In a few people, this can make their chest sore. They may find it hard to breathe.
	You will usually need to take medicines every day. These will lower your blood pressure.
	Blood pressure shows how hard the heart is working to pump blood around the body.
	You may have to give up some sports, especially ones where you could get knocked in your chest.





You will need to have regular check-ups with the doctor to see if you need more treatment.



Please ask us if you have any questions.

