Aortopathy: information for families

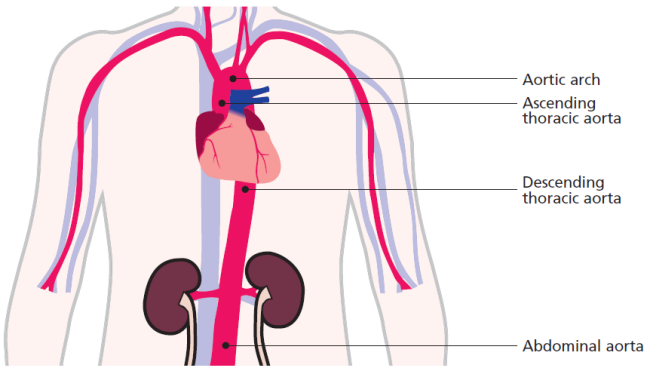
This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of the group of conditions affecting the aorta – the large artery that carries blood from the heart to the rest of the body. These conditions are usually referred to as aortopathies. An Easy Read information sheet is included for your child.

The aorta is a large blood vessel that carries blood from the heart to the rest of the body.

There are two main sections of the aorta: the thoracic aorta is the portion that lies within the chest (thorax), and is further described as ascending – the first portion that leaves the heart – and descending – the rest of the thoracic aorta after the aortic arch where blood travels downwards.

The other portion is the abdominal aorta, which lies within the abdomen.

Many other arteries branch off of the aorta to take blood to specific areas of the body – for instance, the carotid arteries carry blood to the brain and the femoral arteries carry blood to the legs and feet.



What is an aortopathy?

Aortopathy is the term used for a condition that affects the aorta. There are three main groups of aortopathy:

Genetic conditions

- Marfan syndrome
- Familial thoracic aortic aneurysm and dissection
- Loeys-Dietz syndrome
- Vascular Ehlers-Danlos syndrome

Aortopathy associated with congenital (present at birth) heart conditions or following certain heart operations

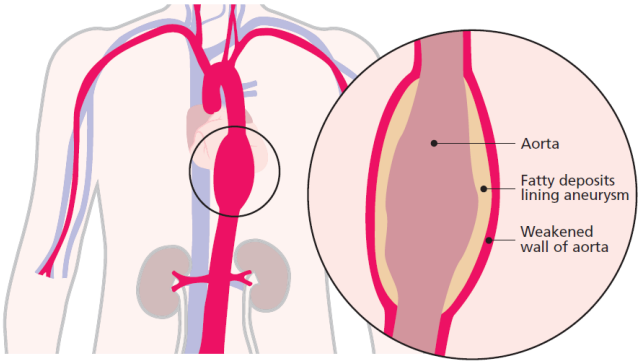
- Coarctation of the aorta
- Bicuspid aortic valve
- Tetralogy of Fallot, transposition of the great arteries and similar conditions
- Ross, Fontan, Norwood and arterial switch procedures

Acquired aortopathy

- Atherosclerosis (hardening of the arteries)
- Systemic arterial hypertension (high blood pressure)

In every patient the genetic component together with conventional risk factors contribute to the onset and progression of aortic disease. In some syndromes, the genetic background play an extremely important role and specific tests and management are needed.

Aorthopathy increases the risk of developing an aneurysm. An aneurysm is the medical term used to describe dilatation in the form of a bulge that develops in a blood vessel wall. It happens when an area of the blood vessel wall is weaker so the pressure of the blood travelling through it causes it to balloon outwards.



The severity of the condition 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.

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.

How common are aortopathies?

In many cases, aortopathies are quite rare so the actual incidence is not known – in addition, some people may not be diagnosed with an aortopathy until they have a catastrophic event. Where incidence figures are known, they vary depending on the type of aortopathy, for instance:

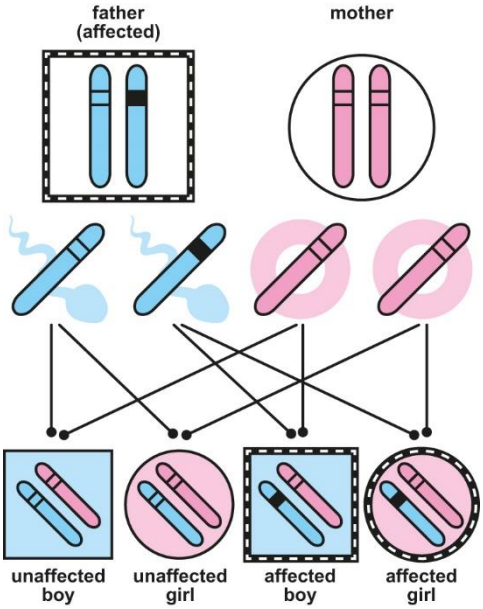
- Marfan syndrome affects 1 in every 3000 to 5000 people
- Bicuspid aortic valve affects 0.5 to 2 per cent of the population and is more common in males than females
- Vascular Ehlers Danlos is estimated to affect 1 in 90,000 to 250,000 people
- Familial thoracic aortic aneurysm and dissection affects 1 in 10,000 people
- Turner syndrome affects 1 in 2500 girls

As research is carried out into aortopathies, more reliable incidence figures may become available.

What causes aortopathies?

Again, this varies depending on the type of aortopathy. Many have a genetic component – that is, they are caused by a faulty gene. This faulty gene can develop out of the blue (sporadically, so there is no one else in the family with the condition) or it can be passed on from parent to child.

We each carry two copies of the vast majority of our genes, with one copy inherited from each parent. Most types of aortopathy are passed on in an autosomal dominant manner, which means a child only has to inherit one faulty copy of the relevant gene from one parent to develop the condition. Every child of an affected person has a 50 per cent chance of inheriting the faulty gene.



How the aortopathy appears and the symptoms it causes can be very different within the same family. Different members of the family may show different symptoms in varying degrees of severity, despite carrying the same genetic mutation. This variability is called ‘incomplete penetrance’.

Over 30 individual genes have been identified as playing a part in aortopathy. These play an important role in the development and growth of the cells that make up the blood vessel walls (and other organs) and hold them in shape.

As a result of this gene mapping, if a family member has been diagnosed with an aortopathy and the specific faulty gene has been identified, other members of the family can be screened to see if they have the condition too, even if they are not showing any symptoms.

What are the symptoms of aortopathy?

Most likely there will be no symptoms to suggest aortopathy. Large size aneurysms can sometimes cause pain depending on their location or if they are pressing on nearby structures.

In the vast majority of cases symptoms may only become apparent when an aneurysm dissects or ruptures. The symptoms of a dissected aneurysm include sharp and severe chest pain, often moving to the back, which can be associated with other symptoms like dizziness or fainting, numbness of one limb, stroke-like symptoms and many others depending on the location of the arterial dissection. Dissection is a life threatening condition that needs emergency treatment.

How are aortopathies diagnosed?

In the majority of cases, aortopathies are diagnosed unexpectedly as a result of screening for another reason entirely.

The usual method of diagnosis is through imaging scans, such as an echocardiogram (echo) which uses sound waves to show blood flow through the heart and surrounding blood vessels or a computed tomography (CT) or magnetic resonance imaging (MRI) scan that produce very detailed pictures of internal structures.

Unfortunately however, some people only become aware that they have aortic disease after dissection of an artery. When a young person presents with an acute vascular event such as dissection or rupture of an artery when they do not have the usual risk factors, such as uncontrolled high blood pressure, the doctors may suspect a genetic aortopathy.

The first step is to investigate and monitor the heart and blood vessel system, possibly with blood tests to look for faulty genes. First-degree family members (parents, brothers and sisters) might also be advised to have similar tests. If the specific faulty gene has been identified in a family member, predictive genetic testing might be available for relatives.

How are aortopathies monitored?

Aortopathy can be a progressive condition, that is, it may worsen as a child grows older and bigger, so regular monitoring is needed in almost all cases. The aim of monitoring is to identify any

aneurysms, measure their size accurately and monitor their growth as the child grows.

Often doctors will set a ‘maximum’ diameter of an aneurysm after which they will plan surgical treatment – this will vary depending on the type of aortopathy. The same imaging scans used to diagnose an aortopathy are also used to monitor it (echocardiogram, MRI and CT scans).

Usually monitoring is carried out every six months to a year in children and teenagers, but may be needed more often if the size of the aneurysm changes or it is getting close to the maximum diameter.

How are aortopathies treated?

Options for treatment vary depending on the location of any aneurysm, its severity and whether it has already dissected.

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).

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 (which joins the aorta to the heart) 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 happens next?

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

People with an aortopathy 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.

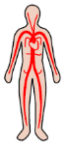
Specific lifestyle, exercise, career and pregnancy advice might differ based on the underlying genetic condition as some syndromes involve other systems and organs and carry specific risks under particular circumstances (such as the management of pregnancy in Loeys-Dietz syndrome and vascular Ehlers-Danlos syndrome). The team will offer tailored counselling and advice based on the diagnosis.

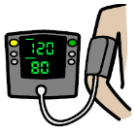
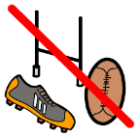
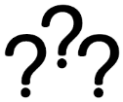
Further information and support

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 . 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
- The Marfan Association offers support and advice to anyone with Marfan syndrome or similar conditions. Call them on 01252 810 472 or visit their website at www.marfan-association.org.uk
- The Marfan Foundation (U.S. based) offers support and advice to anyone with Marfan syndrome and related and rare conditions. A vast selection of comprehensive and up to date resources for parents, teenagers and children (and health providers) can be found on their website at www.marfan.org
- The Loeys-Dietz Syndrome Foundation (LDSF) Provides a support network for anyone affected by Loeys-Dietz syndrome. Visit their website at www.loeysdietz.org/en/
- Annabelle's Challenge is the leading charity in the UK for Vascular Ehlers-Danlos Syndrome, with their website at www.annabelleschallenge.org
- Ehlers Danlos Support UK offers support and advice to anyone with vascular EDS. Call them on 0800 907 8518 or visit their website at www.ehlers-danlos.org/about-eds/typesof-eds/vascular-ehlers-danlos-syndrome
- The Circulation Foundation offers support and advice to anyone with vascular (blood vessel) disease or associated conditions. Call them on 020 7205 7151 or visit their website at www.circulationfoundation.org.uk

All about aorta problems

	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.
	Some people have a bulge in the aorta. This is called an aneurysm (said: ann-you-is-em).
	In a few people, this can make their chest sore. They may find it hard to breathe.
	The bulge might show on an echo.

	You might need a CT or MRI scan as well.
	The bulge makes the walls of the aorta weak. This means they could tear, so blood can leak into the chest. This need to be fixed quickly if it happens.
	If the doctors find the bulge before it tears, they might want you to have a heart operation to fix it.
	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 the bulge is getting bigger or not.
	Please ask us if you have any questions.