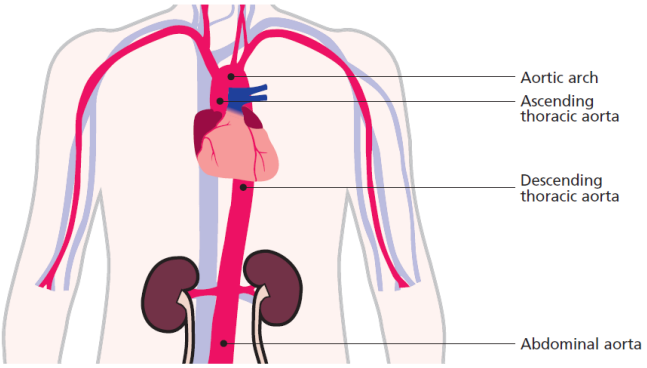
Familial thoracic aortic aneurysm and dissection: information for families

A thoracic aortic aneurysm is a bulge that occurs in the portion of the aorta in the chest. In time, particularly after a period of stress or exertion, the weakened area can split and tear – this is referred to as 'dissection'. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes and symptoms of familial thoracic aortic aneurysm and how it can be managed. An Easy Read information sheet is included for your child.

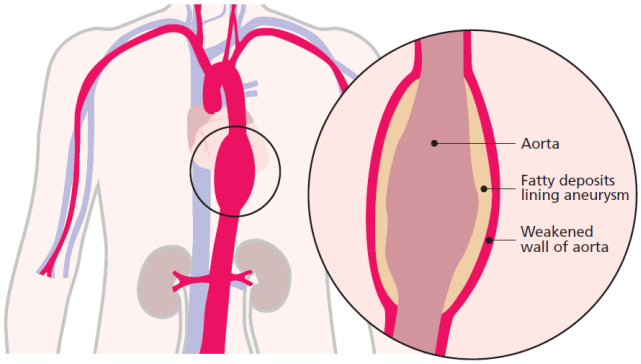
The aorta is a large blood vessel that carries blood from the heart to the rest of the body. There are two main sections of the aorta: the thoracic aorta is the portion that lies within the chest (thorax), and is further described as ascending – the first portion that leaves the heart – and descending – 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.



What is familial thoracic aortic aneurysm?

An aneurysm is the medical term used to describe 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.

A thoracic aortic aneurysm is a bulge that occurs in the portion of the aorta in the chest. The weakening of the blood vessel wall also makes it separate into thin layers along its length.



This means that the blood flow through the weakened section is abnormal, with blood collecting in between the separated layers, putting them under further pressure.

In time, particularly after a period of stress or exertion, the weakened area can split and tear – this is referred to as 'dissection'. The torn area

allows blood to escape from the aorta where it collects in the sac around the heart and lungs. This is a life threatening situation that needs emergency treatment.

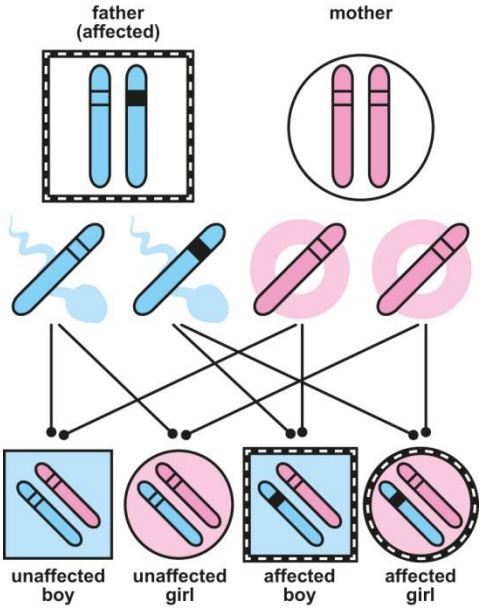
How common is familial thoracic aortic aneurysm?

The reported incidence is around 10 people in every 100,000, although this may be an underestimate. Unfortunately, many people may be unaware that they have familial thoracic aortic aneurysm until a serious event happens. Research has shown that males and females are affected in equal numbers and all races and ethnic backgrounds can be affected.

What causes it?

In around 20 per cent of cases, there is a genetic component to the condition – that is, a faulty gene is passed on from parent to child. A number of faulty genes have been identified, most of which are involved in the development of the smooth muscles cells that make up the blood vessel walls and holds them in shape.

The faulty gene is passed on in an autosomal dominant manner, so a child only has to inherit the faulty gene from one parent to develop the condition. However, we know that having the faulty gene does not automatically mean that a child will develop an aneurysm – for some reason, some do not develop one which implies that other factors may be present as well as genetics.



Familial thoracic aortic aneurysm is also associated with other conditions affecting the development of connective tissue – such as skin, muscles and ligaments – such as Ehlers-Danlos syndrome, Marfan syndrome and Loeys-Dietz syndrome.

The risk of having familial thoracic aortic aneurysm also increases if aneurysms are present elsewhere in the body, for instance in the blood vessel system within the brain or the abdominal aorta. It is also more common in people with certain congenital (present at birth) heart conditions and autoimmune disorders (where the body mistakenly attacks itself rather than foreign invaders such as germs). Part of the assessment of these associated conditions will usually include screening for familial thoracic aortic aneurysm.

What are the symptoms of familial thoracic aortic aneurysm?

Only five per cent of people with familial thoracic aortic aneurysm show symptoms – unfortunately the remainder only become aware following dissection. Where symptoms of the aneurysm are present, they can include chest pain, swelling, swallowing difficulties, wheezing and coughing up blood. If dissection occurs, the chest pain

becomes acute (sharp and sudden) with a weakening pulse and pallor. Symptoms can occur at any age from childhood to old age. Usually the artery develops the bulge first but occasionally, the artery wall can tear without a bulge developing.

How is it diagnosed?

Familial thoracic aortic aneurysm is often 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) scan that produces very detailed pictures of internal structures. Sometimes, a magnetic resonance imaging (MRI) scan shows the aneurysm as can a transoesophageal echocardiogram, which is similar to a regular echo except that the ultrasound probe is inserted down the throat into the oesophagus or foodpipe rather than pressed against the skin over the heart.

How is it treated?

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

Medications to reduce blood pressure will reduce the stress on the bulging part of the aneurysm which may be enough to reduce the risk of dissection. Regular monitoring of the aneurysm will be needed – this is often carried out using echo and CT scans as used for diagnosis. The doctors will measure the diameter (width) of the bulge and the rate at which it is getting bigger. They may set a maximum diameter after which surgery will be needed.

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 an aortic graft – taking a healthy section of aorta from lower down and using it to replace the weakened portion.

If the aortic valve joining the aorta to the heart is damaged, this may be replaced with a mechanical valve. As the artificial valve increases the risk of blood clots developing, anticoagulation (blood thinner) medication will be needed lifelong.

An alternative to grafting and/or valve replacement is to insert a ‘stent’ – an expandable metal or plastic cage – inside the damaged portion of the blood vessel so that cells can grow into and over the cage strengthening the blood vessel wall.

If dissection occurs, an aortic graft and/or valve replacement will be needed – however, it is always preferable to carry out surgery before dissection occurs as the outcomes of surgery are significantly higher.

What is the outlook for children and young people with familial thoracic aortic aneurysm?

When familial thoracic aortic aneurysm has been diagnosed, life-long monitoring will be required and surgery planned when needed.

As there is a genetic component to this condition, screening of close family members (brothers, sisters, parents) might be suggested. If symptoms are not present, monitoring may occur on an annual basis, depending on the size of the bulge.

People with familial thoracic aortic aneurysm 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.

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.

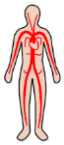
Further information and support

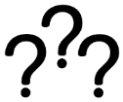
The British Heart Foundation offers support and advice to anyone with heart problems – call their helpline on 0300 330 3311 or visit their website at www.bhf.org.uk

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 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.