

Information for patients, parents or carers

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)

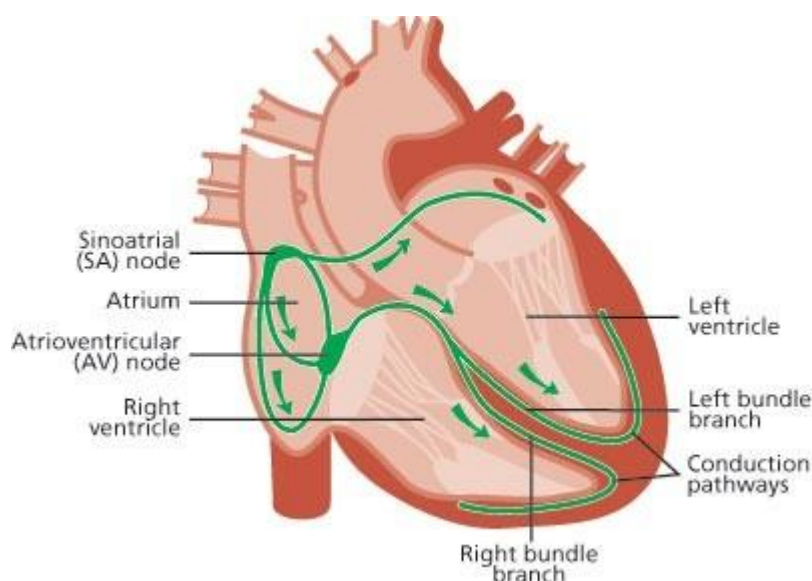
Introduction

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) is a rare heart condition which affects the electrical system of the heart. This information sheet explains the causes and symptoms of CPVT and how it can be managed.

The normal heart

The heart is a special kind of muscle which acts as a pump to keep blood moving. The pumping action of the heart muscle is triggered by electrical messages which travel through the walls of the heart, causing them to squeeze the blood out and around the body.

Each electrical message starts at the sinoatrial (SA) node in the heart. It travels through the walls of the top sections of the heart (the left and right atria), causing them to squeeze blood downwards into the bottom sections of the heart (the left and right ventricles). As the message travels through the bottom sections, it causes them to squeeze and pump blood out of the heart.



The electrical impulse through the heart is created by the movement of sodium, potassium and calcium ions across the cells. Ions are small particles which carry a tiny electrical charge. These ions move in and out of the cells via 'channels' which are found in the walls of the heart muscle cells.

What is Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)?

CPVT is a rare heart condition which affects the electrical system of the heart. In particular, the level of calcium inside the heart cells is not regulated properly, which makes people with CPVT especially vulnerable to abnormal heart rhythms. Because of the way that adrenaline (is a hormone that prepares the body for the ‘fight or flight’ response) interacts with calcium in the heart, people with CPVT are most at risk of developing an arrhythmia when they experience a sudden rush of adrenaline. This might be in response to exercise or because of an emotional stimulus (like a sudden shock or an upsetting event).

Although this is a very long and complicated sounding name, it is just a description of what happens in this condition:

- Catecholaminergic - catecholamine is a hormone that is released into your body to produce adrenaline. It is this “adrenaline rush” that can trigger an arrhythmia in people with CPVT.
- Polymorphic – this describes the type of arrhythmia that can occur in CPVT. It means that the arrhythmia is occurring in lots of different areas of the ventricles of the heart.
- Ventricular tachycardia (also called VT) – this is an abnormal heart rhythm originating in the bottom chambers of the heart. It causes the ventricles to beat extremely fast meaning they cannot pump blood out of the heart effectively. This results in a drop in blood pressure, leading to dizziness, fainting and, potentially, a cardiac arrest.

Another arrhythmia you may hear about is:

- Ventricular fibrillation (also called VF) – this is when all the muscle cells in the ventricle are contracting independently in a disorganised way, effectively stopping the heart muscle from making a co-ordinated movement to contract. Instead of pumping, the heart is tremoring (fibrillating). This means blood is not being effectively pumped out of the heart to the lungs and body. This is known as a cardiac arrest.

Ventricular tachycardia and ventricular fibrillation can be very dangerous and if left untreated can result in sudden death.

It is important to note that not all people with CPVT will have these arrhythmias; however, people with this condition do have an increased risk compared to other people in the population. Fortunately, there are treatments available to help manage the heart rhythm in people with CPVT – these will be explained later.

What causes CPVT?

Genes are like instructions which tell our bodies how to grow and develop. They work by building proteins, which have a huge range of functions in different parts of the body. Some genes carry instructions for building the proteins involved in calcium transport and storage in the heart.

In people with CPVT, these genes have a change that prevents the proteins from developing or working properly. This results in the heart muscle cells of these people being unable to regulate calcium levels properly. Genetic changes like this are called “variants” or “mutations”. This means that they are changes from the normal genetic code that we would expect for that gene.

In some people, the genetic change that causes CPVT occurs in them for the first time. This is called a de novo change. These de novo genetic changes happen around the time of conception, and they are random—there is nothing we can do to cause or prevent them. In people with a de novo gene change, there is no family history of CPVT because they are the first person in the family to be affected.

In other people, the genetic change that causes CPVT has been inherited from one of their parents, which means that the condition first arose in an earlier generation of the family and has been passed down to them.

It is important that when a person is diagnosed with CPVT, their close family members are also screened and tested as some people may not be aware that they have the condition.

How is CPVT inherited?

Everyone inherits two copies of each of their genes, one copy from their mother and one from their father. This means that we all carry two copies of each gene. In some health conditions, only one copy of a gene has to be abnormal to cause signs of the disease to arise. This is called 'autosomal dominant' inheritance. In other conditions, both copies of a gene need to have changes in order for signs of the disease to occur. This is called "autosomal recessive" inheritance.

In the case of CPVT, it is possible for the condition to be inherited in either way; it just depends on which gene is affected.

What are the symptoms of CPVT?

Many people with CPVT may never experience any symptoms during their lifetime. As they don't show any symptoms the condition can remain undiagnosed for a long time. Other people with CPVT may experience symptoms such as palpitations, chest pain, dizziness, feeling lightheaded (pre syncope) or a sudden faint or blackout (syncope). The symptoms of CPVT can be well managed using medications. This is discussed in more detail further on in this leaflet.

As mentioned above, people with CPVT are at an increased risk of developing abnormal heart rhythms, known as arrhythmias. These arrhythmias can be life threatening if left untreated. People with a diagnosis of CPVT should be seen regularly in a specialist inherited cardiovascular disease clinic, where the risk of abnormal arrhythmias can be properly assessed and managed.

How is CPVT syndrome diagnosed?

CPVT may be suspected if a person has an unexpected faint or blackout which is due to an abnormal heart rhythm. Unfortunately, for some people, the first time CPVT may be suspected is after a life-threatening event or sudden death in a family member, especially if there have been no symptoms beforehand. CPVT can also be diagnosed incidentally following a routine ECG for something else entirely.

If CPVT is suspected in your family, or if your child has had symptoms which could have been caused by CPVT, your GP or paediatrician (if your child has one) will refer your child to see a cardiologist with specialist knowledge of inherited cardiac conditions. The cardiologist will order a number of tests to clarify whether or not your child has CPVT.

CPVT can be diagnosed by seeing a particular type of arrhythmia on the patient's ECG. Because this arrhythmia is often triggered by an adrenaline rush in people with CPVT, the most reliable diagnostic test for this condition is an exercise test, which stimulates the release of adrenaline in the patient. This involves having an ECG undertaken during physical activity. Usually this is running on a treadmill, but sometimes if children are too young for a treadmill, we will exercise them by running on the Children's Cardiac Ward whilst attached to ECG monitoring.

Some other tests that the cardiologist may order include:

- Echocardiogram – this is an ultrasound scan of the heart, and it is carried out to make sure that the structure of the heart is normal

- Electrocardiogram (ECG) – this test is used to measure the electrical activity of the heart, using sensors (stickers) that are stuck to the chest
- 24-hour ambulatory ECG monitor (sometimes called a Holter monitor) - this is similar to an ECG, except that the sensors are worn for 24 hours to monitor the electrical activity of the heart for a longer period of time.

In some cases, where there is a family history of CPVT, genetic testing may be used to see if members of the family carry the gene change that can cause CPVT. To carry out genetic testing, a blood sample is collected and sent to a specialist laboratory for testing. It is important to be aware that this type of testing may not be available to all families. The testing process and the implications of the potential test results will always be discussed in detail with yourself before any genetic testing is undertaken.

How is CPVT treated?

There are treatments available to help manage the heart rhythm in people with CPVT – your consultant and specialist nurse will tell you more about these.

- Medication

CPVT cannot be cured, but there are several options to manage the symptoms and protect against irregular heart rhythms. Most people with CPVT take a medication called a beta-blocker, which helps to regulate the heart rate and reduces the risk of arrhythmias. For some people with CPVT, beta-blockers alone are an effective treatment option. However, many people with this condition also need to take an additional medication called flecainide, which works alongside their beta-blocker to help manage symptoms and reduce the risk of arrhythmias developing. Taken together, these medications can be very effective in managing CPVT for most people.

- Left Cardiac Sympathetic Denervation

In some instances, medication alone may not be enough to reduce the risk of arrhythmias developing. As mentioned earlier, often the cause of arrhythmias in patients with CPVT is due to the adrenaline rush that a person can experience. One way to reduce the effect of an adrenaline rush is to carry out a procedure called a left cardiac sympathetic denervation (LCSD). This is also sometimes called a sympathectomy.

The heart rate is controlled by the central nervous system. When there is an adrenaline rush, this can trigger the nervous system to tell the heart to beat faster. This can sometimes cause arrhythmias to occur in people with CPVT. The left cardiac sympathetic denervation (LCSD) procedure involves cutting one of the nerves connected to the heart. This reduces the amount of arrhythmias that could occur. This procedure is only needed in a small number of patients with CPVT. If your cardiologist thinks this type of procedure is needed, more details can be given.

- Implantable Cardioverter Defibrillator (ICD)

In a small number of cases, an individual with CPVT may continue to develop arrhythmias, despite taking medications and having a sympathectomy. Other people with CPVT may have suffered a cardiac arrest previously. In both cases, these individuals are at a greater risk of developing a dangerous arrhythmia (and therefore have an increased risk of sudden death). For these people, an implantable cardioverter defibrillator (ICD) may be considered. An ICD is a small, surgically implanted device that is able to monitor the heart rhythm continuously. If the ICD detects an arrhythmia, it is able to treat this by delivering a burst of electrical energy to the heart, sometimes referred to as a “shock”. This therapy returns the heart back into a normal rhythm.

Life with CPVT

As well as clinical treatment, people with CPVT and their family also benefit from support such as psychology.

Receiving a diagnosis of CPVT can be worrying for everyone involved, so we usually suggest that patients and their families receive some psychological support. This allows them to talk through their feelings and have some help coming to terms with their new diagnosis. Our clinical nurse specialists are available to help answer any questions you may have about CPVT, and they can help explain the condition to children and young people.

Some of the symptoms that are described in CPVT, such as palpitations and chest pain, are very similar to the symptoms of anxiety. If a person with CPVT feels anxious about their condition, then they may experience symptoms caused by this anxiety. These can easily be misinterpreted as cardiac symptoms caused by the condition itself, and this can lead to further worry and further worsening of anxiety symptoms. We have psychologist working within our team to help our patients develop coping strategies to overcome these difficulties.

In some cases, families are referred to our service for cardiac screening tests because they have lost a close family member due to CPVT. Our psychologists are also available to offer bereavement counselling for those who have lost someone close to them.

Lifestyle advice

Having CPVT will mean lifelong follow-up in the cardiology clinic to monitor the condition, to manage medication doses and to continually assess risk.

Having CPVT may mean that some careers are unsuitable for your child – for instance, the armed forces or professional sports. You can talk to the team about this at any time.

Day-to-day management of CPVT involves identifying the triggers for symptoms and avoiding them wherever possible. It is also important to treat any prolonged period of diarrhoea and vomiting as this can cause dehydration and the loss of potassium and sodium, which can increase the risk of arrhythmias. Re-hydration sachets contain a good balance of minerals as well as fluid to replace any losses, you can purchase these from your local pharmacy. If this period of sickness continues, seek immediate medical advice. You may need to visit your local accident and emergency centre for rehydration, blood tests and an ECG.

When your child is diagnosed with CPVT you will receive a documentation called an Emergency Health Care Plan (EHCP), this is an official document that gives information about their condition, events that might occur and how healthcare professionals should act to ensure the best possible outcomes for your child. You will be given a written copy and sent an electronic version to handover/show to professionals in an event. For further information please contact your child's specialist cardiac rhythm and inherited cardiac conditions nurse.

Exercise

As high intensity exercise and strenuous activity can trigger arrhythmias in CPVT, we advise that highly competitive or prolonged vigorous exercise should be avoided. Maintaining a healthy and balanced lifestyle is equally important for good general health, once your child has had exercise restrictions relaxed. We would encourage children and young people with CPVT to participate in PE, swimming, recreation and games but they must be always supervised by an adult. It is important that they continue to take the medication that has been prescribed and that they attend the cardiology clinic regularly for close monitoring. Each time they attend clinic; the cardiologist will ensure that their medication doses are correct and working well. If your child has any symptoms during exercise i.e. palpitations, dizziness they must stop exercising immediately, rest until symptoms stop or if immediately concerned, seek immediate medical attention.

If you have specific questions relating to exercise and CPVT for your child, you can speak with your cardiologist or one of the clinical nurse specialists for further advice.

Further information and support

Contact details

Paediatric rhythm and ICC nurse specialist: 0191 213 9633

Children's cardiac nurse specialist: 0191 213 7344 Email address: nuth.ccns@nhs.net

The cardiac nurse specialists are around 09.00am to 5.00pm Monday to Friday. If there is no answer please leave a voicemail, and they will endeavour to return your call within 24 hours. If urgent, please contact ward 23, Freeman Hospital for advice.

Ward 23 Freeman Hospital: 0191 2137023 (24 hours)

The Patient Advice and Liaison Service (PALS) can offer on-the-spot advice and information about the NHS. You can contact them on freephone 0800 032 02 02 or email pals@nhct.nhs.uk.

<https://www.northumbria.nhs.uk/patients-and-visitors/share-your-feedback/patient-and-advice-liaison-service-pals#9882d87e>

Useful websites

The Arrhythmia Alliance supports anyone affected by a heart rhythm problem. Visit their website at www.heartrhythmcharity.org.uk

CRY (Cardiac Risk in the Young) is another organisation offering advice and support to families of children with heart problems. Visit their website at www.c-r-y.org.uk

SADS UK offers support and advice about heart conditions that can lead to sudden unexpected death. Visit their website at www.sadsuk.org

The British Heart Foundation is the main organisation in the UK offering advice and support to anyone affected by heart disease. Visit their website at www.bhf.org.uk

If you would like further information about health conditions and treatment options, you may wish to have a look at the NHS website at www.nhs.uk

If you would like to find accessibility information for our hospitals, please visit www.accessable.co.uk

Information adapted from Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT): Information from families. Great Ormond Street Hospital for Children.

Information produced by (F Pugh Cardiac Rhythm and ICC Nurse Specialist)

Date: November 2025

Review date: November 2028