Patient Information

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)

1. The Normal Heart

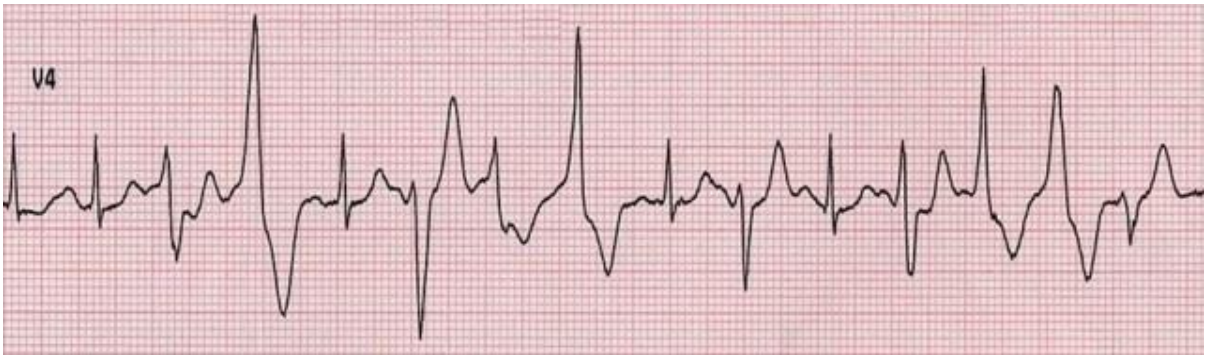
The heart is a special muscle that contracts regularly and continually, pumping blood to the body and the lungs. It has four chambers – two at the top (the atria) and two at the bottom (the ventricles). The pumping action of your heart is caused by a flow of electrical signals through the heart. These electrical signals repeat themselves in a cycle and each cycle causes one heartbeat. When the electrical activity of the heart is disturbed, known as an arrhythmia, it can affect your heart's ability to pump properly.

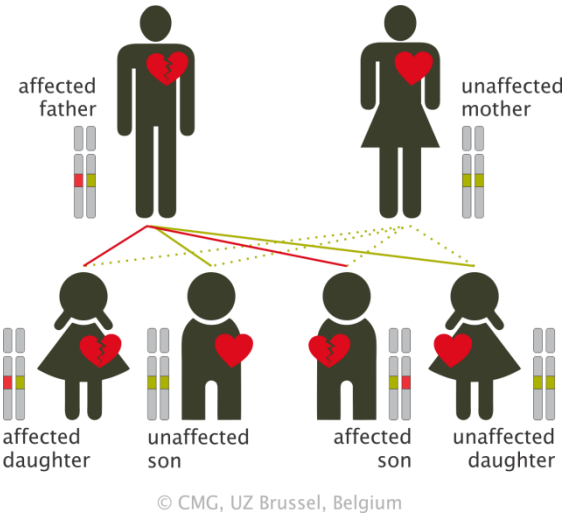
2. CPVT

CPVT is a rare heart disease which causes a type of fast arrhythmia known as ventricular tachycardia caused by physical exercise or emotional stress. CPVT is found mainly in children and young people, although it can be diagnosed at any age. The arrhythmias are caused by an abnormal control of the level of calcium inside the heart cells. If the level of calcium becomes too high, it can result in ventricular tachycardia. If this abnormally fast heart rate does not fix itself, blood cannot be pumped properly around the body and it can lead to dizziness, black-outs or even sudden death.

3. Prevalence & Inheritance

About 1 in every 10,000 persons has CPVT (the prevalence of the disease). CPVT is a genetic disease. This means that CPVT is caused by a defect (a mutation) in a gene that can be passed on through families. A gene is part of our DNA which contains a code for making a molecule (a protein). Every person has two copies of each gene that can be linked to CPVT. CPVT is caused by a mutation in the genes that contain codes for specific molecules (proteins) in the heart. A mutation in only one of the two copies of one of these genes (from the father or from the mother) is enough to develop CPVT. This is called an autosmal dominant disorder and a parent who carries it has a 50% chance (1 in 2) of passing the mutation to each child. The chance that a child will not inherit the mutation is also 50 percent. Sometimes, CPVT can also be an autosomal recessive disease. This means that you need mutations on both copies of a gene (from both father and mother) to develop CPVT. Whether CPVT is an autosomal dominant disease or an autosomal recessive disease depends on the gene and mutation. In some cases, a new (de novo) mutation can occur in the egg or sperm cells or in an embryo. In these cases, the child's parents do not have the mutation and CPVT, but the child does have CPVT and can pass the mutation to his or her own children.





Autosomal dominant inheritance

4. Symptoms

CPVT most often affects children and young adults. The most common symptoms are palpitations or fainting and collapses, particularly during exercise. Diagnosing CPVT can be difficult, as the ECG is completely normal at rest but may be abnormal during an exercise test. However, once CPVT is diagnosed, treatments are available.

5. Diagnosis

The most common tools to make the diagnosis of CPVT are the medical and family history, physical examination, a heart electrical tracing (the electrocardiogram or ECG), and exercise testing. The exercise test and Holter monitoring also have an important role to control the success of the therapy and should therefore be periodically performed in all patients.

5.1. ECG (electrocardiogram)

This is the most basic test. Small sticky patches (electrodes) are put onto the chest and sometimes to arms and legs. These are connected by wires to an ECG recording machine, which picks up the electrical activity for a few seconds that makes the heartbeat. Sometimes additional or repeated ECG-tests are necessary.

5.2. Exercise test (stress test)

Exercise test is the same as the ECG described above, but is recorded before, during and exercising on a treadmill or an exercise bike. This records any changes in the electrical patterns that occur with exercise.

5.3. Holter monitoring

Holter monitoring involves a small digital machine, which can be worn on a belt round the waist. Four or six ECG electrodes from the machine are taped to the chest. It then records the electrical activity of the heart for 24-48 hours, or for up to seven days. During the monitoring all activities are listed in a 'diary'.

5.4. Cardiomemo and cardiac event recorders

These are more complicated versions of the Holter monitoring test described above. During any symptoms, the device can be triggered to record the heart's rhythm. The advantage of the cardiomemo is that it doesn't have any electrodes, so it just can be placed on the chest while having symptoms.

5.5. Echocardiogram (echo)

Echocardiogram uses ultrasound waves to look at the structures of the heart. An echocardiogram can detect different types of structural changes in the heart, for example heart muscle diseases and heart valve abnormalities. Areas of thinning of the heart muscle can also be identified. Patients with CPVT don't have structural problems, but often an echo is performed once to confirm this.

5.6. Genetic testing

In over half of CPVT families, a mutation can be identified in the RYR2-gene. In patients with autosomal recessive CPVT, two mutations in the CASQ2 gene can be found. The RYR2- and CASQ2-genes make two different proteins that are important for controlling the level of calcium inside heart cells.

6. Therapy

Often a medicine known as a beta blocker is prescribed to reduce arrhythmias. They are used in CPVT patients to slow the heart rate and reduce the effect of exertion or excitement on the heart. As beta-blockers make the heart beat slower, it allows blood to completely fill the chambers before it is pumped out. This leads to better heart function and to better circulation of blood around the body. Depending on the effect of the beta blockers, the therapy might be combined with other medicines such as flecainide. In patients where medications do not work or in patients after a cardiac arrest, an internal cardiac defibrillator

(ICD) or cervical sympathectomy may be considered. An ICD can correct most life-threatening arrhythmias. Cervical sympathectomy (also called cardiac denervation) is a surgical procedure to damage the nerves that release adrenaline and similar natural substances in the heart.

7. Lifestyle & Sports

There are key recommendations for patients (and families) who are diagnosed with CPVT, to prevent them from arrhythmias:

- in general avoid competitive and strenuous sports

- sport is permitted only after advise from an expert heart specialist.

- use of beta blockers (if prescribed)

- encourage relatives to be screened

The diagnosis of CPVT and the ability to pass on the condition can lead to anxiety and many other questions. Medical social workers or psychologists have experience with this and may be helpful for the patient and the family members.

8. Follow-up

The heart doctor (cardiologist) will advise on how often follow-up is needed depending on symptoms, age, and treatment.

9. Family Screening

If a mutation in a gene is found in a patient with CPVT, family members of this patient (to start with the first degree family members: mother, father, brothers, sisters and children) can have genetic testing at a genetic heart clinic. Family members in whom the same mutation is found, are called 'mutation carriers' and will be followed up by a cardiologist. Family members in whom the mutation is not found can be reassured. If there is not a mutation identified in a patient with CPVT, family members of this patient (to start with the first degree family members) are advised to see a cardiologist. CPVT patients can experience symptoms in childhood. Therefore genetic and heart testing, and timely treatment of the family members who are diagnosed with CPVT is important even in the first years of life (preferably before they start swimming lessons).

10. CPVT and Pregnancy

During pregnancy it is important to continue treatment with beta blockers. Sometimes it's necessary to switch the type of beta blocker, because not all types are suitable for use during pregnancy. When beta blockers are used during pregnancy, it is advised to plan for delivery in hospital, because of a possible low heart rate in the baby.

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