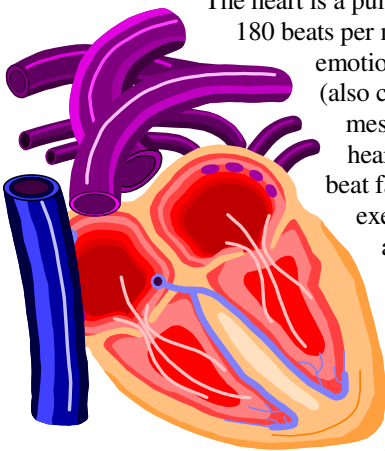


## Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Information sheet

### What is Catecholaminergic Polymorphic Ventricular Tachycardia?

CPVT is a rare condition that affects the heart of otherwise fit and healthy people. It causes the heart to beat abnormally quickly, usually at times of exercise (particularly swimming) or times of high emotion, and can result in dizziness, sudden loss of consciousness or even death. It most commonly occurs in children and young adults typically in the first or second decade of life. It was first recognised in 1975. In at least a third of cases, it is familial, (inherited) being passed down through the generations. It is a difficult condition to diagnose, because all of the tests taken at a time of rest are normal- including the electrocardiogram (ECG), and the echocardiogram (ultrasound of the heart). The diagnosis is usually made by detecting extra beats, or runs of fast rhythm arising from the bottom part of the heart, (ventricles), during an exercise test or on a 24 hour ECG.

### How does CPVT get its name?



The heart is a pump that initiates its own electrical signals, causing it to beat between at between 50 and 180 beats per minute depending on whether you are resting, exercising or under some form of emotional/physical stress. In response to stress the body releases adrenaline and noradrenaline (also called epinephrine and norepinephrine). Adrenaline and noradrenaline are chemical messengers that are collectively known as catecholamines that have a direct action on the heart. During exercise or emotional stimulation these catecholamines cause the heart to beat faster and the blood pressure to increase, thus enabling blood and oxygen to reach the exercising muscles and other vital organs. This response to stress is how the body copes and allows us to flee from danger or run to catch the bus, however it also works the same if the body is emotionally or physically challenged in some other way, such as severe illness.

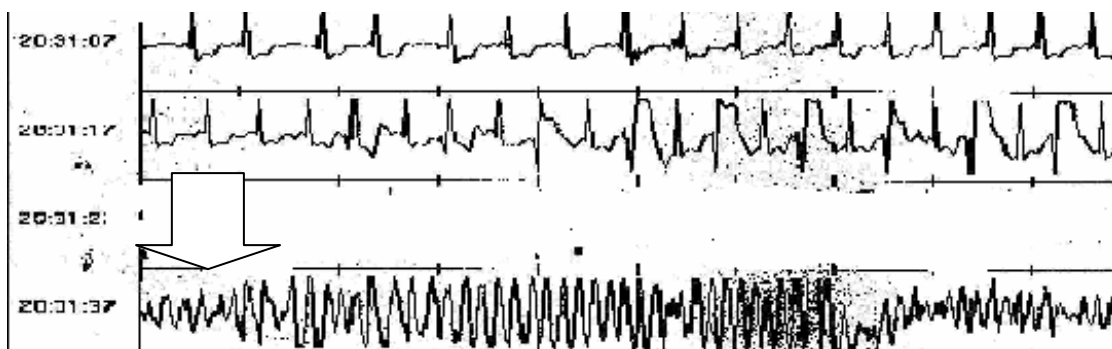
People with CPVT have hearts that respond abnormally to catecholamines, such that the heartbeat becomes rapid, chaotic and irregular at times of stress. A rapid heart rhythm is known as a “tachycardia”, and since this particular tachycardia arises from the main pumps or ventricles, it is known as a “ventricular tachycardia”. The word “polymorphic” literally means lots of different shapes- referring to the varying beats seen on the ECG. If such a rhythm sustains for more than a few seconds, insufficient blood reaches the brain, leading to a faint, collapse, or even death.

### How does it present?

CPVT typically presents with sudden collapse or severe dizziness, usually associated with exercise (or just after exercise stops) or intense emotional stress. First symptoms appear usually in the first or second decade of life, and may often be mistaken for epilepsy in children. It can also present quite out of the blue with sudden death.

## How can doctors make the diagnosis?

Symptoms may be reproduced by exercise test, and a 24-hour ECG (Holter) test can be useful. These tests are always performed in hospital under specialist supervision. Increasingly doctors use an implanted digital loop recorder, or “Reveal” device, which records the ECG continuously, and documents the heart rhythm during a collapse (see Fig 2 below).



**Fig 2: Reveal™ Event recorder ECG showing onset of Ventricular tachycardia. The arrow shows the fast abnormal heart rhythm starting, not how much faster each beat is in comparison with the two lines above which show a normal rhythm (line 1, and the heart beat becoming more unstable with some different looking beats occurring during line two before the fast rhythm begins in line 3. Patient symptoms included dizziness and collapse.**

Sudden collapse or severe dizziness may be associated with heart rhythm disturbance such as CPVT. Typically these symptoms may occur with little or no warning, and are usually associated with exercise (or just after exercise stops) or intense emotional stress. Typically cases present in the first or second decade of life, and may often be mistaken for epilepsy in children. Symptoms of the disease may be reproduced by exercise test or by using other forms of adrenergic stimulation such as drugs that increase the heartbeat. These tests are always performed in hospital under Specialist supervision.

The first case of CPVT was reported in 1975, with further cases published in 1995. Familial incidence of the condition has been manifest in 30% of cases to date.

### Treatment:

CPVT usually responds well to beta-blockers, although beta-blockers are usually used in combination with implantable defibrillators (AICDs; automatic implantable cardioverter defibrillators) that stop the fast chaotic heart rhythm by treating it with a small electric shock. Beta-blocker medications work to stop adrenaline and other catecholamines from speeding the heart up and therefore keep the heart rate under control. It is important that if you are taking beta-blockers for this condition that you continue to take them and do not miss any doses. It is good to establish a usual routine to taking this medication so that it is difficult to ‘forget’, i.e. before brushing your teeth at night, or in the morning.

### Genetic Diagnosis of CPVT:

At this time 3 genes are thought to be responsible for CPVT. Ryanodine (RYR2) mutations (abnormalities in the gene sequence), are thought to be responsible for the autosomal dominant (this means that each child born has a 50% chance of inheriting the mutation). In the heart ryanodine is located in the working part of the heart muscle cells that are responsible for making the heart beat.

The role of ryanodine is to release calcium inside the cell in order for each heartbeat to occur, however this does not work properly as in the case of the abnormal ryanodine, then calcium may build up and not be realised as it should. When adrenaline is present and the heart rate speeds up the working cells are likely not able to cope with the increased workload and this throw the heart into the chaotic heart rhythm depicted in figure 1.0.

Calsequestrin CASQ2 binds to calcium inside the working areas of the cardiac muscle cells (sarcoplasmic reticulum), it is thought that mutations in the calsequestrin prevent adequate binding occurring particularly when the cell is having to work hard in the presence of catecholamines such as adrenaline. The build up of calcium because it cannot adequately attach itself to calsequestrin for transport is thought to cause the chaotic heart rhythm also seen in figure 1.0.

Ankyrin B mutations have also been implicated in CPVT, and these may also be linked to LQT (Long QT type 4 ). Work is still being done to establish this link.

### **Other sources of information:**

There are a number of physicians in New Zealand who have a special interest in the management of families with heart rhythm disorders and some of these are listed below. There are many web sites to visit, of varying quality. A good start is [www.SADS.org](http://www.SADS.org).

### **Physicians with skills in the management of heart rhythm problems:**

Dr Ian Crozier, Cardiologist, Christchurch Hospital, Christchurch

Dr Joanne Dixon, Clinical Geneticist, Wellington Hospital, Wellington

Dr Spencer Heald, Cardiologist, Waikato Hospital, Hamilton

Dr David Heaven, Cardiologist, Middlemore Hospital, Auckland

Dr Margaret Hood, Cardiologist, Green Lane Hospital, Auckland

Dr Nigel Lever, Cardiologist, Wellington Hospital, Wellington

Dr Hugh McAlister, Cardiologist, Waikato Hospital, Hamilton

Dr Jon Skinner, Cardiologist, Green Lane Hospital, Auckland

Dr Warren Smith, Cardiologist, Green Lane Hospital, Auckland

Many of these Doctors travel to smaller hospitals during the course of their work if you are unsure please ask your Doctor or Specialist.

### **Registry of Cardiac Inherited Disease in New Zealand:**

We are in the process of developing this registry and it is not yet truly national. A significant registry exists in Wellington under the care of Joanne Dixon and a new registry, based in Auckland, is being developed as part of the inherited cardiac disease service. This is co-ordinated by Jackie Crawford (Senior Cardiac Technologist) and Drs Jon Skinner and Dr Jim Stewart. Ultimately we hope to amalgamate these registries to form a national service. The Cardiac inherited disease group is a group of Specialists who meet once a month to discuss heart rhythm problems (many of the Doctors listed above are members of this group, but it also includes, Genetic counsellors, pathologists, Laboratory Technologists and others dedicated to diagnosis of heart rhythm disorders.

If you have any other questions you would like to ask, please do not hesitate to contact the registry co-ordinator and/or discuss your queries with your Specialist.

**If you have any queries regarding your rights as a participant in the Cardiac Inherited Disease Registry you may phone the Health Advocate Trust: Phone (09) 623 5799 Auckland**

**Or contact Mata Forbes RGON; Co-ordinator/Advisor, Maori Health Services, Auckland Healthcare Services Ltd, Auckland Hospital Grafton; Mobile 021 348 432  
Work Phone (09) 307 4949 Ext. 7972.**

**This study has received Ethical Approval from the Auckland Ethics Committee: Study AKX/02/00/107**