A note from the SADS Foundation.

We provide this information with the hope that informing physicians, other health care providers, and the public will encourage early and correct diagnosis and proper therapy, resulting in the reduction and ultimately elimination of cardiac arrest and sudden death from CPVT and other inherited arrhythmias.

Why do I need to know about CPVT?
CPVT (Catecholaminergic Polymorphic Ventricular Tachycardia) is a rare but potentially lethal yet treatable condition. It is far less common than LQTS but does affect otherwise healthy infants, children, adolescents, and adults at an unacceptably high rate. However, with increased awareness, genetic testing, and effective treatment options, CPVT can be diagnosed early and sudden death prevented. Still, this condition is often undetected prior to death and not recognized as the cause of death. Family members of individuals with unexplained death should be tested for CPVT and other genetic arrhythmias. CPVT is a treatable disorder and, with correct diagnosis and treatments, most deaths are preventable.

Physicians need to know:
• When to consider CPVT as a possible diagnosis.
• When to refer patients for diagnosis & treatment.
• About genetic testing for CPVT and other SADS conditions.
• How to develop a family pedigree and screen family members for CPVT.

Patients and Parents need to know:
• The warning signs and symptoms of CPVT.
• Who to see for proper testing.
• How to protect their children and themselves.
• How to expand their family pedigree and contact other family members who may be at risk.

What is CPVT?
CPVT is a condition that results in ventricular rhythm problems (bidirectional and polymorphic ventricular tachycardia and ventricular fibrillation) that can cause fainting or sudden death. Events usually occur with exercise or during stress. CPVT is caused by abnormalities in the way the heart muscle cells handle calcium.

What are the symptoms of CPVT?
Patients with CPVT experience fainting (syncope) usually with exercise or other activities that are associated with high adrenaline levels. Unfortunately, sudden death may be the first symptom in CPVT patients. Testing for CPVT includes a resting ECG that is usually normal and exercise testing to try to provoke the rhythm abnormalities. In children too young to perform exercise testing a 24-hour Holter monitor may help with the diagnosis. In children with CPVT the heart muscle function and the heart structure are normal.

Some CPVT patients experience syncope only. In these patients presumably the ventricular tachycardia returns to normal and the patient quickly regains consciousness, without disorientation or confusion. Some patients may experience slight fatigue afterwards; others feel fine and resume their regular activities. If the ventricular tachycardia persists longer, patients may then manifest a generalized seizure. In both the syncope and seizure presentations, the heart eventually caught itself, reverted back to normal sinus rhythm, and the “spell” is over. On the other hand, in some patients, the ventricular tachycardia degenerates into the heart rhythm known as ventricular fibrillation. This rarely reverts back to a normal rhythm without medical intervention. If the ventricular fibrillation is not converted, usually by electrical defibrillation, the outcome is sudden cardiac death or sudden cardiac arrest.

When should the diagnosis be suspected?
• In any young person with unexplained syncope (fainting), unexplained seizures, or unexplained cardiac arrest or sudden death especially during exercise or stress.

Usually, a careful history of the events surrounding the syncope differentiates CPVT induced syncope from the common faint, known as vasovagal or neurocardiogenic syncope. The CPVT syncope is usually precipitous and without warning. It often occurs during physical exertion or emotional stress. Conversely, in vasovagal syncope, most times there are warning symptoms, such as dizziness, blurring or blackening of vision, tingling or sweating, for seconds to even minutes prior to the syncope. Also, a precipitating event is usually present, commonly pain, injury, nausea, or an unpleasant or stressful experience.

• When there is a family history of unexplained syncope, unexplained seizures, or sudden death in young people especially in the setting of stress or exercise.
• When the autopsy is normal following the sudden and unexpected death of a young person.

How is the diagnosis made?
CPVT is diagnosed primarily by the identification of bidirectional ventricular tachycardia or polymorphic ventricular tachycardia. This is a wide QRS tachycardia that has a QRS appearance (morphology) that changes. This can be provoked with exercise. These patients have a normal resting ECG and normal QTc interval.

How is CPVT inherited, and who in a known or suspected family should be tested?
There are 2 known genetic variants of CPVT. Most commonly, CPVT is inherited by autosomal dominant transmission (RyR2). There is a less common autosomal recessive forms (CASQ2). This means that it generally affects boys and girls equally, and that each child of an affected parent has a 50% chance of inheriting the genetic abnormality. In a really large family, close to 50% of the children would inherit the gene. In average size families it can range from all to none as each child has an independent 50/50 chance of inheriting the gene. Once a family member is identified with CPVT, it is extremely important that other family members be tested for the syndrome. It is especially important to know which parent and grandparent has the abnormality, since brothers and sisters, aunts,
uncles, nephews, nieces, and cousins on the affected side are potentially at risk. There are forms of this disease that are inherited in an autosomal recessive manner. This means that both parents must have the gene defect to pass it on. This type is much less common. There are patients with CPVT that do not have either of the 2 identified mutations. Thus, all types of CPVT have not been characterized.

This prospective screening, by ECG and treadmill testing or Holter monitoring are extremely important so that all affected family members are identified and treated early in order to prevent the tragic and unnecessary sudden deaths that may occur.

What about genetic testing?
Since the original characterization of CPVT in 1995 it has been clear that the disease is familial. About 1/3 of patients have a positive family history. Genetic testing for the most common type of CPVT is commercially available and about 50% of CPVT patients will have a positive test.

What is the treatment and who should be treated?
All symptomatic patients should receive treatment. All children and young adults should be treated even if they do not have symptoms. This is because symptoms might occur and sudden death may be the first symptom. At present, it is not possible to tell which child or youth is destined to have symptoms. Thus, preventative treatment is required in all. The usual treatment involves taking beta-blocker medications daily. The dose of beta-blocker needs to be monitored closely carefully and exercise testing may help determine if the medication is effective.

Patients who continue to have symptoms in spite of appropriate doses of beta-blockers may also require additional medications or devices. There is conflicting data concerning the use of calcium channel blocking agents with the beta-blockers in CPVT. Patients who have experienced a cardiac arrest or continued syncope on beta-blockers usually receive an ICD (implantable defibrillator) in addition to the beta-blocker.

Medication compliance
It is very important that CPVT-directed medical therapies be taken every day and not missed or omitted. The medications are not curative; they only provide protection while being taken and the protective effect is gone within a day or two of stopping the medication. After that the risk of cardiac events is the same as if the patient had not taken the medication at all. Parents should teach their children about the importance of daily medication, and should make sure each daily dose is taken. Physicians need to discuss this directly with all patients, but particularly pre-teens and teenagers.

How can parents protect their kids?
- Make sure the children take their medication daily, no missing doses
- See the doctor regularly for follow-up. Growing children need medication dose changes regularly. Make sure you see the doctor at least once a year, more frequently during very rapid growth, and discuss the need for dose changes
- Be supportive if the doctor advises “no competitive sports for your child”. Support this advice, and help the child to understand that usual physical activities are suitable, but that competition may be dangerous. Channel their energies into sports without intense physical demands (golf, for example), or non-physical activities.
- Get additional medical advice if you are not comfortable with how things are going. Ideally, every patient/family with CPVT should be cared for by a heart rhythm specialist (cardiac electrophysiologist). Do not hesitate to obtain a second opinion if you have any questions about your child's treatment.
- Make sure your family has an AED (automatic external defibrillator) and/or your child’s school district has AED programs in their schools.

Supporting Families. Saving Lives
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