



Catecholaminergic polymorphic ventricular tachycardia

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a condition characterized by an abnormal heart rhythm (arrhythmia). As the heart rate increases in response to physical activity or emotional stress, it can trigger an abnormally fast and irregular heartbeat called ventricular tachycardia. Episodes of ventricular tachycardia can cause light-headedness, dizziness, and fainting (syncope). In people with CPVT, these episodes typically begin in childhood.

If CPVT is not recognized and treated, an episode of ventricular tachycardia may cause the heart to stop beating (cardiac arrest), leading to sudden death. Researchers suspect that CPVT may be a significant cause of sudden death in children and young adults without recognized heart abnormalities.

Frequency

The prevalence of CPVT is estimated to be about 1 in 10,000 people. However, the true prevalence of this condition is unknown.

Genetic Changes

CPVT can result from mutations in two genes, *RYR2* and *CASQ2*. *RYR2* gene mutations cause about half of all cases, while mutations in the *CASQ2* gene account for 1 percent to 2 percent of cases. In people without an identified mutation in one of these genes, the genetic cause of the disorder is unknown.

The *RYR2* and *CASQ2* genes provide instructions for making proteins that help maintain a regular heartbeat. For the heart to beat normally, heart muscle cells called myocytes must tense (contract) and relax in a coordinated way. Both the *RYR2* and *CASQ2* proteins are involved in handling calcium within myocytes, which is critical for the regular contraction of these cells.

Mutations in either the *RYR2* or *CASQ2* gene disrupt the handling of calcium within myocytes. During exercise or emotional stress, impaired calcium regulation in the heart can lead to ventricular tachycardia in people with CPVT.

Inheritance Pattern

When CPVT results from mutations in the *RYR2* gene, it has an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder. In about half of cases, an affected person inherits an *RYR2* gene mutation from one affected parent. The remaining cases result from new mutations in the *RYR2* gene and occur in people with no history of the disorder in their family.

When CPVT is caused by mutations in the *CASQ2* gene, the condition has an autosomal recessive pattern of inheritance. Autosomal recessive inheritance means that both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- bidirectional tachycardia induced by catecholamines
- Catecholamine-induced polymorphic ventricular tachycardia
- CPVT
- familial polymorphic ventricular tachycardia
- FPVT

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Catecholaminergic polymorphic ventricular tachycardia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1631597/>
- Genetic Testing Registry: Ventricular tachycardia, catecholaminergic polymorphic, 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2677794/>

Other Diagnosis and Management Resources

- GeneReview: Catecholaminergic Polymorphic Ventricular Tachycardia
<https://www.ncbi.nlm.nih.gov/books/NBK1289>
- MedlinePlus Encyclopedia: Fainting
<https://medlineplus.gov/ency/article/003092.htm>
- MedlinePlus Encyclopedia: Ventricular Tachycardia
<https://medlineplus.gov/ency/article/000187.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Fainting
<https://medlineplus.gov/ency/article/003092.htm>
- Encyclopedia: Ventricular Tachycardia
<https://medlineplus.gov/ency/article/000187.htm>
- Health Topic: Arrhythmia
<https://medlineplus.gov/arrhythmia.html>

Genetic and Rare Diseases Information Center

- Catecholaminergic polymorphic ventricular tachycardia
<https://rarediseases.info.nih.gov/diseases/4421/catecholaminergic-polymorphic-ventricular-tachycardia>

Educational Resources

- Cincinnati Children's Hospital Medical Center: Fast Arrhythmias
<https://www.cincinnatichildrens.org/patients/child/encyclopedia/diseases/arrhythmia>
- Disease InfoSearch: Familial Ventricular Tachycardia
<http://www.diseaseinfosearch.org/Familial+Ventricular+Tachycardia/2777>
- KidsHealth from the Nemours Foundation: Arrhythmias
<http://kidshealth.org/en/teens/arrhythmias.html>
- MalaCards: catecholaminergic polymorphic ventricular tachycardia
http://www.malacards.org/card/catecholaminergic_polymorphic_ventricular_tachycardia
- Merck Manual Consumer Version: Ventricular Tachycardia
<https://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/abnormal-heart-rhythms/ventricular-tachycardia>
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=Catecholaminergic%20Polymorphic%20Ventricular%20Tachycardia&type=profile>
- Orphanet: Catecholaminergic polymorphic ventricular tachycardia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3286

Patient Support and Advocacy Resources

- American Heart Association
http://www.heart.org/HEARTORG/Conditions/Arrhythmia/TypesofArrhythmias/Tachycardia_UCM_302018_Article.jsp
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/conghart.html>

GeneReviews

- Catecholaminergic Polymorphic Ventricular Tachycardia
<https://www.ncbi.nlm.nih.gov/books/NBK1289>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Tachycardia%2C+Ventricular%22+OR+%22catecholaminergic+polymorphic+ventricular+tachycardia%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Tachycardia,+Ventricular%5BMAJR%5D%29+AND+%28%28catecholaminergic+polymorphic+ventricular+tachycardia%5BTIAB%5D%29+OR+%28catecholaminergic+%5BTIAB%5D+AND+ventricular+tachycardia+%5BTIAB%5D%29+OR+%28familial+polymorphic+ventricular+tachycardia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC, 1, WITH OR WITHOUT ATRIAL DYSFUNCTION AND/OR DILATED CARDIOMYOPATHY
<http://omim.org/entry/604772>
- VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC, 2
<http://omim.org/entry/611938>

Sources for This Summary

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