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, \textit{RYR2} and \textit{CASQ2}. \textit{RYR2} gene mutations cause about half of all cases, while mutations in the \textit{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 \textit{RYR2} and \textit{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 \textit{RYR2} or \textit{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 \textit{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 \textit{RYR2} gene mutation from one affected parent. The remaining cases result from new mutations in the \textit{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
- Genetic Testing Registry: Ventricular tachycardia, catecholaminergic polymorphic, 2

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
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
- MalaCards: catecholaminergic polymorphic ventricular tachycardia
  http://www.malacards.org/card/catecholaminergic_polymorphic_ventricular_tachycardia
  https://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/abnormal-heart-rhythms/ventricular-tachycardia
- Orphanet: Catecholaminergic polymorphic ventricular tachycardia
  https://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+polymorphic+ventricular+tachycardia%5BTIAB%5D%29+AND+ventricular+tachycardia%5BTIAB%5D%29+OR+%28familial+polymorphic+ventricular+tachycardia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmhm%5D+AND+OR+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

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19879546
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19121813
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