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.

Causes

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 Information

- What is genetic testing? [link]
- Genetic Testing Registry: Catecholaminergic polymorphic ventricular tachycardia [link]
- Genetic Testing Registry: Ventricular tachycardia, catecholaminergic polymorphic, 2 [link]

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov [link]

Other Diagnosis and Management Resources

- GeneReview: Catecholaminergic Polymorphic Ventricular Tachycardia [link]
- MedlinePlus Encyclopedia: Fainting [link]
- MedlinePlus Encyclopedia: Ventricular Tachycardia [link]
Additional Information & Resources

Health Information from 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

• KidsHealth from the Nemours Foundation: Arrhythmias

• MalaCards: catecholaminergic polymorphic ventricular tachycardia
  https://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

• 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/en/health-topics/arrhythmia/about-arrhythmia/tachycardia--fast-heart-rate

• Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/conghart.html

Clinical Information from GeneReviews

• Catecholaminergic Polymorphic Ventricular Tachycardia
  https://www.ncbi.nlm.nih.gov/books/NBK1289
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

Catalog of Genes and Diseases from 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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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1235548/
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