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?
  /primer/testing/genetictesting
- 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/

Research Studies from ClinicalTrials.gov

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

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

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
  https://kidshealth.org/en/teens/arrhythmias.html
- MalaCards: catecholaminergic polymorphic ventricular tachycardia
  https://www.malacards.org/card/catecholam
  inergic_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

- Cerrone M, Napolitano C, Priori SG. Catecholaminergic polymorphic ventricular tachycardia: A paradigm to understand mechanisms of arrhythmias associated to impaired Ca(2+) regulation. Heart Rhythm. 2009 Nov;6(11):1652-9. doi: 10.1016/j.hrthm.2009.06.033. Epub 2009 Jun 30. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19879546
- Györke S. Molecular basis of catecholaminergic polymorphic ventricular tachycardia. Heart Rhythm. 2009 Jan;6(1):123-9. doi: 10.1016/j.hrthm.2008.09.013. Epub 2008 Sep 16. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19121813
- Hayashi M, Denjoy I, Extramiana F, Maltret A, Buisson NR, Lupoglazoff JM, Klug D, Hayashi M, Takatsuki S, Villain E, Kamblock J, Messali A, Guicheney P, Lunardi J, Leenhardt A. Incidence and risk factors of arrhythmic events in catecholaminergic polymorphic ventricular tachycardia. Circulation. 2009 May 12;119(18):2426-34. doi: 10.1161/CIRCULATIONAHA.108.829267. Epub 2009 Apr 27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19398665
- Katz G, Arad M, Eldar M. Catecholaminergic polymorphic ventricular tachycardia from bedside to bench and beyond. Curr Probl Cardiol. 2009 Jan;34(1):9-43. doi: 10.1016/j.cpcardiol.2008.09.002. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19068246
- Lahat H, Pras E, Olender T, Avidan N, Ben-Asher E, Man O, Levy-Nissenbaum E, Khoury A, Lorber A, Goldman B, Lancet D, Eldar M. A missense mutation in a highly conserved region of CASQ2 is associated with autosomal recessive catecholamine-induced polymorphic ventricular tachycardia in Bedouin families from Israel. Am J Hum Genet. 2001 Dec;69(6):1378-84. Epub 2001 Oct 25.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11704930
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1235548/
- Liu N, Ruan Y, Priori SG. Catecholaminergic polymorphic ventricular tachycardia. Prog Cardiovasc Dis. 2008 Jul-Aug;51(1):23-30. doi: 10.1016/j.pcad.2007.10.005. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18634915

- Mohamed U, Napolitano C, Priori SG. Molecular and electrophysiological bases of catecholaminergic polymorphic ventricular tachycardia. J Cardiovasc Electrophysiol. 2007 Jul;18(7): 791-7. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17578347
- Napolitano C, Priori SG, Bloise R. Catecholaminergic Polymorphic Ventricular Tachycardia. 2004 Oct 14 [updated 2016 Oct 13]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from http://www.ncbi.nlm.nih.gov/books/NBK1289/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301466
- Priori SG, Napolitano C, Memmi M, Colombi B, Drago F, Gasparini M, DeSimone L, Coltorti F, Bloise R, Keegan R, Cruz Filho FE, Vignati G, Benatar A, DeLogu A. Clinical and molecular characterization of patients with catecholaminergic polymorphic ventricular tachycardia. Circulation. 2002 Jul 2;106(1):69-74.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12093772
- Priori SG, Napolitano C, Tiso N, Memmi M, Vignati G, Bloise R, Sorrentino V, Danieli GA. Mutations in the cardiac ryanodine receptor gene (hRyR2) underlie catecholaminergic polymorphic ventricular tachycardia. Circulation. 2001 Jan 16;103(2):196-200.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11208676
- Scheinman MM, Lam J. Exercise-induced ventricular arrhythmias in patients with no structural cardiac disease. Annu Rev Med. 2006;57:473-84. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16409161

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/catecholaminergic-polymorphic-ventricular-tachycardia

Reviewed: December 2009
Published: November 13, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services