



PDCD10 gene

programmed cell death 10

Normal Function

The *PDCD10* gene (also known as *CCM3*) provides instructions for making a protein that appears to play a role in the structure of blood vessels. While the exact function of the PDCD10 protein is unclear, studies suggest that it works with other proteins to help strengthen the interactions between cells and limit leakage from blood vessels. This protein is also thought to be involved in pathways that signal cells to self-destruct (undergo apoptosis) when they have completed a certain number of cell divisions or accumulated errors in their DNA.

Health Conditions Related to Genetic Changes

Cerebral cavernous malformation

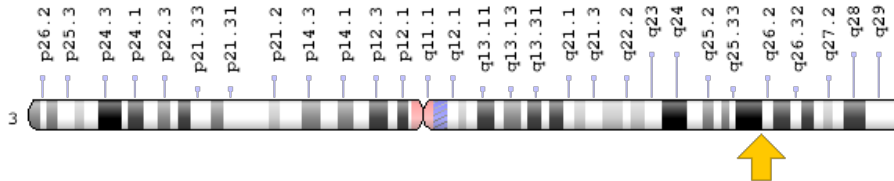
More than a dozen mutations in the *PDCD10* gene have been identified in families with cerebral cavernous malformations, which are collections of blood vessels in the brain that are weak and prone to leakage. These mutations include a deletion of the entire gene, deletion of small segments of DNA, and changes in single DNA building blocks (nucleotides). These mutations result in an abnormal or absent PDCD10 protein. It is unclear how mutations in the *PDCD10* gene lead to the formation of cerebral cavernous malformations.

Mutations in the *PDCD10* gene account for approximately 10 percent of familial cerebral cavernous malformation cases.

Chromosomal Location

Cytogenetic Location: 3q26.1, which is the long (q) arm of chromosome 3 at position 26.1

Molecular Location: base pairs 167,683,298 to 167,735,810 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- apoptosis-related protein 15
- CCM3
- cerebral cavernous malformation 3
- PDC10_HUMAN
- TFAR15

Additional Information & Resources

Clinical Information from GeneReviews

- Familial Cerebral Cavernous Malformation
<https://www.ncbi.nlm.nih.gov/books/NBK1293>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PDCD10%5BTIAB%5D%29%29+OR+%28programmed+cell+death+10%5BTIAB%5D%29%29+OR+%28TFAR15%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- PROGRAMMED CELL DEATH 10
<http://omim.org/entry/609118>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/PDCD10ID43399ch3q26.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PDCD10%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:8761
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:11235>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/11235>
- UniProt
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<https://ghr.nlm.nih.gov/gene/PDCD10>

Reviewed: November 2012

Published: November 12, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services