DNAJC19 gene

DnaJ heat shock protein family (Hsp40) member C19

Normal Function

The *DNAJC19* gene provides instructions for producing a protein found in structures called mitochondria, which are the energy-producing centers of cells. While the exact function of the DNAJC19 protein is unclear, researchers believe that it helps transport other proteins into and out of mitochondria. The DNAJC19 protein may also assist in the proper assembly and disassembly of certain proteins.

Health Conditions Related to Genetic Changes

Dilated cardiomyopathy with ataxia syndrome

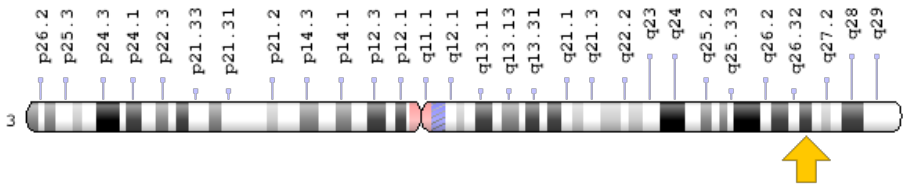
At least two mutations in the *DNAJC19* gene have been found to cause dilated cardiomyopathy with ataxia (DCMA) syndrome. This condition is characterized by heart problems, movement difficulties, slow growth, genital abnormalities in males, and other features affecting multiple body systems. *DNAJC19* gene mutations lead to the production of an abnormally shortened protein that likely has impaired function. In the Dariusleut Hutterite population of Canada, where DCMA syndrome is most frequently seen, the condition results from a mutation (written as IVS3-1G>C) that causes a disruption in the way the gene's instructions are used to make the DNAJC19 protein, resulting in deletion of part of the protein.

Researchers speculate that a lack of functional DNAJC19 protein alters the transport of proteins into and out of the mitochondria. When too many or too few proteins move in and out of the mitochondria, energy production and mitochondrial survival can be reduced. Tissues that have high energy demands, such as the heart and the brain, are especially susceptible to decreases in cellular energy production. This loss of cellular energy likely damages these and other tissues, leading to heart problems, movement difficulties, and other features of DCMA syndrome.

Chromosomal Location

Cytogenetic Location: 3q26.33, which is the long (q) arm of chromosome 3 at position 26.33

Molecular Location: base pairs 180,983,709 to 180,989,774 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DnaJ (Hsp40) homolog, subfamily C, member 19
- homolog of yeast TIM14
- mitochondrial import inner membrane translocase subunit TIM 14
- PAM18
- TIM14
- TIM14_HUMAN
- TIMM14
- translocase of the inner mitochondrial membrane 14

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Mitochondrion https://www.ncbi.nlm.nih.gov/books/NBK26894/
- The Cell: A Molecular Approach (second edition, 2000): Mitochondria https://www.ncbi.nlm.nih.gov/books/NBK9896/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DNAJC19%5BTIAB%5D%29+OR+%28TIM14%5BTIAB%5D%29+OR+%28PAM18%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- DNAJ/HSP40 HOMOLOG, SUBFAMILY C, MEMBER 19
  http://omim.org/entry/608977

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DNAJC19.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DNAJC19%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:30528
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:131118
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/131118
- UniProt
  https://www.uniprot.org/uniprot/Q96DA6

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