DES gene
desmin

Normal Function

The DES gene provides instructions for making a protein called desmin. Desmin is found in heart (cardiac) muscle and muscles used for movement (skeletal muscle). Within muscle fibers, desmin proteins are important to help maintain the structure of sarcomeres, which are necessary for muscles to tense (contract). The desmin proteins surround rod-like structures called Z-discs that are located within the sarcomere. Desmin connects the Z-discs to one another, linking neighboring sarcomeres and forming myofibrils, the basic unit of muscle fibers. The connection of sarcomeres to each other to form myofibrils is essential for maintaining muscle fiber strength during repeated cycles of contraction and relaxation.

Health Conditions Related to Genetic Changes

Myofibrillar myopathy

More than 40 mutations in the DES gene have been found to cause myofibrillar myopathy. Most of these mutations change single protein building blocks (amino acids) in desmin. Mutated desmin proteins cluster together with other muscle proteins in the sarcomere to form clumps (aggregates). The aggregates prevent these proteins from functioning normally. A dysfunctional desmin protein cannot properly interact with Z-discs, leading to abnormalities of sarcomere structure and problems with the formation of myofibrils. DES gene mutations that cause myofibrillar myopathy impair the function of muscle fibers, causing weakness and the other features of this condition. People with DES gene mutations are more likely to have a weakened heart muscle (cardiomyopathy) than people with myofibrillar myopathy caused by mutations in other genes. In some cases, cardiomyopathy is the first symptom of this condition.

Arrhythmogenic right ventricular cardiomyopathy

Familial dilated cardiomyopathy

Other disorders

Mutations in the DES gene also cause a form of heart disease called dilated cardiomyopathy type 1I. This condition enlarges (dilates) and weakens the cardiac muscle, preventing it from pumping blood efficiently. DES gene mutations have also been shown to cause another form of cardiomyopathy called restrictive cardiomyopathy, in which the heart muscle is stiff and cannot fully relax after each
contraction. Although cardiomyopathy is a sign of myofibrillar myopathy, these forms of cardiomyopathy are not associated with weakness of the skeletal muscles.

Mutations in the DES gene can also cause an abnormal heartbeat (arrhythmia), which may lead to heart failure and sudden death.

Researchers are not certain why some mutations in the DES gene cause these heart problems instead of myofibrillar myopathy.

**Chromosomal Location**

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 219,418,377 to 219,426,739 on chromosome 2 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- DESM_HUMAN

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791
- Washington University, St. Louis: Neuromuscular Disease Center: Myofibrillar Myopathies
  https://neuromuscular.wustl.edu/musdist/lg.html#desmut

**Clinical Information from GeneReviews**

- Dilated Cardiomyopathy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1309
- Myofibrillar Myopathy
  https://www.ncbi.nlm.nih.gov/books/NBK1499
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DES%5BTI%5D%29+OR+%28desmin%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- CARDIOMYOPATHY, DILATED, 1I
  http://omim.org/entry/604765
- DESMIN
  http://omim.org/entry/125660

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DES.html
- ClinVar
- HGNC Gene Family: Intermediate filaments Type III
  https://www.genenames.org/cgi-bin/genefamilies/set/610
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1674
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P17661

Sources for This Summary

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- OMIM: DESMIN
  http://omim.org/entry/125660
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