



## 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.

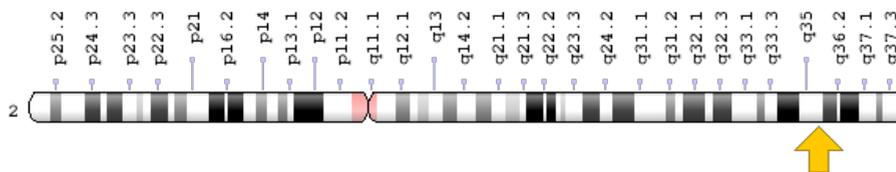
Mutations in the *DES* gene can also cause an abnormal heartbeat (arrhythmia), which may lead to heart failure and sudden death.

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 219,426,734 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DESM\_HUMAN

## Additional Information & Resources

### Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Muscle Contraction  
<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>

### 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](http://atlasgeneticsoncology.org/Genes/GC_DES.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=DES%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:2770](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2770)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:1674>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1674>
- UniProt  
<https://www.uniprot.org/uniprot/P17661>

### **Sources for This Summary**

- Claeys KG, van der Ven PF, Behin A, Stojkovic T, Eymard B, Dubourg O, Laforêt P, Faulkner G, Richard P, Vicart P, Romero NB, Stoltenburg G, Udd B, Fardeau M, Voit T, Fürst DO. Differential involvement of sarcomeric proteins in myofibrillar myopathies: a morphological and immunohistochemical study. *Acta Neuropathol.* 2009 Mar;117(3):293-307. doi: 10.1007/s00401-008-0479-7. Epub 2009 Jan 17.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19151983>
- OMIM: DESMIN  
<http://omim.org/entry/125660>
- Ferrer I, Olivé M. Molecular pathology of myofibrillar myopathies. *Expert Rev Mol Med.* 2008 Sep 3; 10:e25. doi: 10.1017/S1462399408000793. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18764962>

- Goldfarb LG, Dalakas MC. Tragedy in a heartbeat: malfunctioning desmin causes skeletal and cardiac muscle disease. *J Clin Invest*. 2009 Jul;119(7):1806-13. doi: 10.1172/JCI38027. Epub 2009 Jul 1. Review. Erratum in: *J Clin Invest*. 2011 Jan 4;121(1):455.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19587455>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2701871/>
  - Schröder R, Schoaser B. Myofibrillar myopathies: a clinical and myopathological guide. *Brain Pathol*. 2009 Jul;19(3):483-92. doi: 10.1111/j.1750-3639.2009.00289.x. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19563540>
  - van Spaendonck-Zwarts KY, van Hessem L, Jongbloed JD, de Walle HE, Capetanaki Y, van der Kooi AJ, van Langen IM, van den Berg MP, van Tintelen JP. Desmin-related myopathy. *Clin Genet*. 2011 Oct;80(4):354-66. doi: 10.1111/j.1399-0004.2010.01512.x. Epub 2010 Jul 21. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20718792>
- 

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/DES>

Reviewed: January 2011  
Published: August 17, 2020

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