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

MedlinePlus Genetics provides information about Arrhythmogenic right ventricular cardiomyopathy

Familial dilated cardiomyopathy

MedlinePlus Genetics provides information about 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.

Other Names for This Gene

- DESM_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DES (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1674[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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

- DESMIN; DES (https://omim.org/entry/125660)
- CARDIOMYOPATHY, DILATED, 1I; CMD1I (https://omim.org/entry/604765)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1674)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DES[gene])

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Genomic Location

The *DES* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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