SGCD gene

sarcoglycan delta

Normal Function

The *SGCD* gene provides instructions for making the delta component (subunit) of a group of proteins called the sarcoglycan protein complex. The sarcoglycan protein complex is located in the membrane surrounding muscle cells. It helps maintain the structure of muscle tissue by attaching (binding) to and stabilizing the dystrophin complex, which is made up of proteins called dystrophins and dystroglycans. The large dystrophin complex strengthens muscle fibers and protects them from injury as muscles tense (contract) and relax. The dystrophin complex acts as an anchor, connecting each muscle cell's structural framework (cytoskeleton) with the lattice of proteins and other molecules outside the cell (extracellular matrix).

Health Conditions Related to Genetic Changes

Limb-girdle muscular dystrophy

At least 14 mutations in the *SGCD* gene have been identified in people with limb-girdle muscular dystrophy type 2F. Limb-girdle muscular dystrophy is a group of related disorders characterized by muscle weakness and wasting, particularly in the shoulders, hips, and limbs. People with limb-girdle muscle dystrophy type F have *SGCD* mutations in both copies of the gene in each cell (autosomal recessive inheritance pattern).

A small number of people with limb-girdle muscular dystrophy type 2F have developed a heart condition called dilated cardiomyopathy. Dilated cardiomyopathy is a form of heart disease that enlarges and weakens the heart (cardiac) muscle, preventing it from pumping blood efficiently. Dilated cardiomyopathy progresses rapidly and can be life-threatening.

Forms of limb-girdle muscular dystrophy caused by gene mutations that affect the sarcoglycan complex are called sarcoglycanopathies. *SGCD* gene mutations may prevent the sarcoglycan complex from forming or from binding to and stabilizing the dystrophin complex. Problems with these complexes reduce the strength and resilience of muscle fibers and result in the signs and symptoms of limb-girdle muscular dystrophy.

Familial dilated cardiomyopathy

MedlinePlus Genetics provides information about Familial dilated cardiomyopathy

Other disorders

A small number of people who develop dilated cardiomyopathy without skeletal muscle involvement have been found to have a mutation in one copy of the *SGCD* gene in each cell, an inheritance pattern called autosomal dominant.

Since other individuals with one *SGCD* gene mutation in each cell have normal heart muscle, some researchers question whether *SGCD* gene mutations are related to autosomal dominant dilated cardiomyopathy.

Other Names for This Gene

- 35 kDa dystrophin-associated glycoprotein
- 35DAG
- 35kD dystrophin-associated glycoprotein
- CMD1L
- DAGD
- delta-sarcoglycan
- delta-SG
- LGMD2F
- MGC22567
- placental delta sarcoglycan
- sarcoglycan, delta (35kDa dystrophin-associated glycoprotein)
- SG-delta
- SGCD_HUMAN
- SGCDP
- SGD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SGCD (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6444[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SGCD%5BTIAB%5D%29+OR+%28sarcoglycan,+delta%5BTIAB%5D%29%29+OR+%28%2835+kDa+dystrophin-associated+glycoprotein%5BTIAB%5D%29+OR+%2835DAG%5BTIAB%5D%29+OR+%28DAGD%5BTIAB%5D%29+OR+%28delta-sarcoglycan%5BTIAB%5D%

29+OR+%28delta-SG%5BTIAB%5D%29+OR+%28LGMD2F%5BTIAB%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

- SARCOGLYCAN, DELTA; SGCD (https://omim.org/entry/601411)
- CARDIOMYOPATHY, DILATED, 1L; CMD1L (https://omim.org/entry/606685)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6444)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SGCD[gene])

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Genomic Location

The *SGCD* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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