

SGCA gene

sarcoglycan alpha

Normal Function

The *SGCA* gene provides instructions for making the alpha 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. It 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

More than 70 mutations in the *SGCA* gene have been identified in people with limb-girdle muscular dystrophy type 2D. Limb-girdle muscular dystrophy is a group of related disorders characterized by muscle weakness and wasting, particularly in the shoulders, hips, and limbs. Forms of limb-girdle muscular dystrophy caused by gene mutations that affect the sarcoglycan complex are called sarcoglycanopathies.

The most common *SGCA* gene mutation occurs in about one-third of people with limb-girdle muscular dystrophy type 2D. This mutation replaces the protein building block (amino acid) arginine with the amino acid cysteine at position 77 in the alpha-sarcoglycan protein, written as Arg77Cys or R77C. The rest of the known *SGCA* gene mutations are specific to individual families or certain populations.

SGCA 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.

Other Names for This Gene

- 50 kDa dystrophin-associated glycoprotein

- 50-DAG
- 50DAG
- 50kD DAG
- adhalin
- ADL
- alpha-sarcoglycan
- alpha-SG
- DAG2
- DMDA2
- dystroglycan-2
- LGMD2D
- sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein)
- SCARMD1
- SG-alpha
- SGCA_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SGCA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6442\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6442[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SGCA%5BTIAB%5D%29+OR+%28sarcoglycan,+alpha%5BTIAB%5D%29%29+OR+%28%2850+kDa+dystrophin-associated+glycoprotein%5BTIAB%5D%29+OR+%2850-DAG%5BTIAB%5D%29+OR+%2850DAG%5BTIAB%5D%29+OR+%28adhalin%5BTIAB%5D%29+OR+%28alpha-sarcoglycan%5BTIAB%5D%29+OR+%28alpha-SG%5BTIAB%5D%29+OR+%28DAG2%5BTIAB%5D%29+OR+%28LGMD2D%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+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SARCOGLYCAN, ALPHA; SGCA (<https://omim.org/entry/600119>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6442>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SGCA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SGCA[gene]))

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Genomic Location

The *SGCA* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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