SGCB gene
sarcoglycan beta

Normal Function

The *SGCB* gene provides instructions for making the beta 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**

Approximately 50 mutations in the *SGCB* gene have been identified in people with limb-girdle muscular dystrophy type 2E. Limb-girdle muscular dystrophy is a group of related disorders characterized by muscle weakness and wasting, particularly in the shoulders, hips, and limbs.

Some people with limb-girdle muscular dystrophy type 2E also develop 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 is life-threatening in many cases.

Forms of limb-girdle muscular dystrophy caused by gene mutations that affect the sarcoglycan complex are called sarcoglycanopathies. *SGCB* 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.
**Chromosomal Location**

Cytogenetic Location: 4q12, which is the long (q) arm of chromosome 4 at position 12

Molecular Location: base pairs 52,020,695 to 52,038,319 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- 43 kDa dystrophin-associated glycoprotein
- 43DAG
- A3b
- beta-sarcoglycan
- beta-SG
- LGMD2E
- sarcoglycan, beta (43kDa dystrophin-associated glycoprotein)
- SG-beta
- SGC
- SGCB_HUMAN

**Additional Information & Resources**

**Educational Resources**

- University of Washington Neuromuscular Disease Center
  https://neuromuscular.wustl.edu/musdist/lg.html#2e
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SGCB%5BTIAB%5D%29+OR+%28%28beta-sarcoglycan%5BTIAB%5D%29+OR+%28LGMD2E%5BTIAB%5D%29+OR+%2843DAG%5BTIAB%5D%29+OR+%28beta-SG%5BTIAB%5D%29+OR+%2843+kDa+dystrophin-associated+glycoprotein%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D+AND+Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SARCOGLYCAN, BETA
  http://omim.org/entry/600900

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SGCB%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6443

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q16585

Sources for This Summary


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10679964

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17558114

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15937871

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3279956/

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