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.

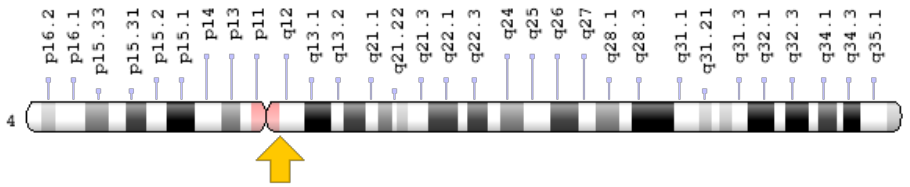
Some people with limb-girdle muscular dystrophy type 2E also develop a heart condition called dilated cardiomyopathy. Dilated cardiomyopathy is a form of heart disease that enlarges and weakens the heart (cardiac) muscle, preventing it from pumping blood efficiently. Dilated cardiomyopathy progresses 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+%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, BETA
  http://omim.org/entry/600900

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SGCB%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10806
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6443
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6443
- UniProt
  https://www.uniprot.org/uniprot/Q16585

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