SGCE gene
sarcoglycan epsilon

Normal Function

The SGCE gene provides instructions for making a protein called epsilon (ε)-sarcoglycan, whose function is unknown. The ε-sarcoglycan protein is found within the outer membrane of cells in tissues throughout the body, but it is most abundant in nerve cells (neurons) in the brain and in muscle cells. Researchers suspect that in the brain the ε-sarcoglycan protein plays a role in the functioning of synapses, which are the connections between neurons where cell-to-cell communication occurs.

People inherit one copy of most genes from their mother (the maternal copy) and one copy from their father (the paternal copy). Both copies are typically active, or "turned on," in cells. However, only the paternal copy of the SGCE gene is active. This sort of parent-specific difference in gene activation is caused by a phenomenon called genomic imprinting.

Health Conditions Related to Genetic Changes

Myoclonus-dystonia

More than 110 mutations in the SGCE gene have been found to cause myoclonus-dystonia, which is a movement disorder characterized by involuntary muscle twitches in the neck, torso, and arms (myoclonus). Most of these mutations lead to an abnormally short, nonfunctional ε-sarcoglycan protein that is quickly broken down. Other mutations prevent the protein from reaching the cell membrane where it is needed. This lack of functional protein seems to affect the regions of the brain involved in coordinating and controlling movements (the cerebellum and basal ganglia, respectively) and leads to the involuntary movements characteristic of myoclonus-dystonia. It is unknown why SGCE gene mutations seem to affect only these areas of the brain.

Myoclonus-dystonia occurs when mutations affect the paternal copy of the SGCE gene. More than 95 percent of individuals who inherit an SGCE gene mutation from their mothers do not show signs or symptoms of the condition. Rarely, individuals who inherit an SGCE gene mutation from their mothers will develop features of myoclonus-dystonia. It is unclear why a gene that is supposed to be turned off is active in these rare cases.
Chromosomal Location

Cytogenetic Location: 7q21.3, which is the long (q) arm of chromosome 7 at position 21.3

Molecular Location: base pairs 94,584,980 to 94,656,205 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DYT11
- ESG
- sarcoglycan, epsilon
- SGCE_HUMAN

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: ε-sarcoglycan
  https://www.ncbi.nlm.nih.gov/books/NBK6317/#A37969

Clinical Information from GeneReviews

- SGCE Myoclonus-Dystonia
  https://www.ncbi.nlm.nih.gov/books/NBK1414

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SGCE%5BTIAB%5D%29+OR+%28epsilon+sarcoglycan%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+last+1800+days+AND+5Bdp%5D
Catalog of Genes and Diseases from OMIM

- SARCGLYCAN, EPSILON
  http://omim.org/entry/604149

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SGCE.html

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

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:8910

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/O43556

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17200151

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12634861

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17853490

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25209853
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4495322/

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  http://omim.org/entry/604149