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.

Other Names for This Gene

- DYT11
- ESG
- sarcoglycan, epsilon
- SGCE_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SGCE (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8910[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SGCE%5BTIAB%5D%29+OR+%28epsilon+sarcoglycan%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SARCOGLYCAN, EPSILON; SGCE (https://omim.org/entry/604149)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/8910)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SGCE[gene])

References

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Genomic Location

The *SGCE* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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