SACS gene

sacsin molecular chaperone

Normal Function

The *SACS* gene provides instructions for producing a protein called sacsin. Sacsin is found in the brain, skin cells, muscles used for movement (skeletal muscles), and at low levels in the pancreas, but the specific function of the protein is unknown. Research suggests that sacsin may play a role in folding newly produced proteins into the proper 3-dimensional shape because it shares similar regions with other proteins that perform this function.

Health Conditions Related to Genetic Changes

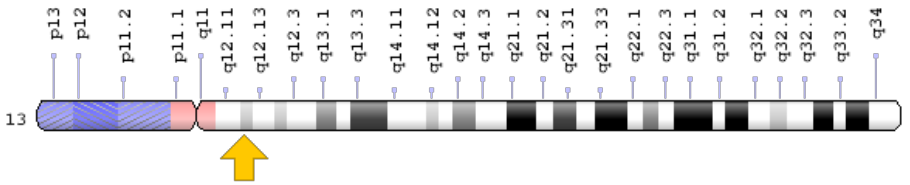
Autosomal recessive spastic ataxia of Charlevoix-Saguenay

Approximately 28 mutations in the *SACS* gene have been found to cause autosomal recessive spastic ataxia of Charlevoix-Saguenay, commonly called ARSACS. Two mutations have been found frequently in affected people from Quebec. One of these mutations deletes a DNA building block (nucleotide) called thymine at position 6594 in the *SACS* gene (written as 6594delT). This mutation is found in more than 90 percent of people with ARSACS in Quebec. The other mutation replaces the nucleotide cytosine with the nucleotide thymine at position 5254 in the *SACS* gene (written as C5254T). Both of these mutations lead to production of a sacsin protein that is abnormally short and nonfunctional. Mutations causing ARSACS in people outside of Quebec are varied and usually unique to that person or family. Most of these mutations either delete one or more nucleotides or replace one nucleotide with another nucleotide in the *SACS* gene. Mutations in the *SACS* gene cause the production of an unstable sacsin protein that does not function normally. It is unclear how the abnormal sacsin protein affects the brain and skeletal muscles and results in the signs and symptoms of ARSACS.

Chromosomal Location

Cytogenetic Location: 13q12.12, which is the long (q) arm of chromosome 13 at position 12.12

Molecular Location: base pairs 23,328,823 to 23,433,728 on chromosome 13 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARSACS
- DNAJC29
- KIAA0730
- PPP1R138
- SACS_HUMAN
- sacsin
- spastic ataxia of Charlevoix-Saguenay (sacsin)
- SPAX6

Additional Information & Resources

Educational Resources

- Clincal Methods (third edition, 1990): The Motor System and Gait https://www.ncbi.nlm.nih.gov/books/NBK391/

Clinical Information from GeneReviews

- ARSACS https://www.ncbi.nlm.nih.gov/books/NBK1255

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SACS%5BTIAB%5D%29+AND+%28spastic+ataxia%5BALL%5D%29+OR+%28sacsin%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

- SACSIN
  http://omim.org/entry/604490

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SACS.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SACS%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10519
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:26278
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/26278
- UniProt
  https://www.uniprot.org/uniprot/Q9NZJ4

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