SPG7 gene

SPG7, paraplegin matrix AAA peptidase subunit

Normal Function

The *SPG7* gene provides instructions for producing a protein called paraplegin, which is a member of the AAA protein family. This protein family plays a role in many cellular activities, including regulation of cell components and proteins. Located within the inner membrane of the energy-producing centers of cells (mitochondria), paraplegin is one of the proteins that form a complex called the m-AAA protease. The m-AAA protease is responsible for assembling ribosomes (cellular structures that process the cell's genetic instructions to create proteins) and removing nonfunctional proteins in the mitochondria.

Health Conditions Related to Genetic Changes

Spastic paraplegia type 7

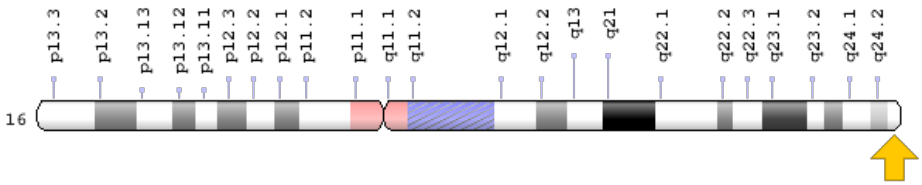
More than 10 mutations that cause spastic paraplegia type 7 have been found. Most of these mutations change single protein building blocks (amino acids) in the protein paraplegin. When paraplegin is mutated, it cannot organize with other proteins within the mitochondria to form the m-AAA protease. The buildup of unusable proteins in nerve cells, caused by the nonfunctional m-AAA protease, can impair mitochondrial functioning and diminish nerve cell signaling, leading to the major signs and symptoms of spastic paraplegia type 7.

Progressive external ophthalmoplegia

Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 89,508,379 to 89,557,768 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CAR
- cell adhesion regulator
- CMAR
- FLJ37308
- MGC126331
- MGC126332
- paraplegin, isoform 1
- PGN
- spastic paraplegia 7
- spastic paraplegia 7 (pure and complicated autosomal recessive)
- SPG5C
- SPG7_HUMAN

Additional Information & Resources

Clinical Information from GeneReviews

- Spastic Paraplegia 7
  https://www.ncbi.nlm.nih.gov/books/NBK1107

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG7%5BTIAB%5D%29+OR+%28paraplegin%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SPG7 GENE
  http://omim.org/entry/602783

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SPG7.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SPG7%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11237
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6687
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6687
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https://ghr.nlm.nih.gov/gene/SPG7

Reviewed: January 2008
Published: November 12, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services