SPG7 gene

SPG7 matrix AAA peptidase subunit, paraplegin

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 100 mutations in the *SPG7* gene have been found to cause spastic paraplegia type 7. This condition is characterized by muscle weakness, progressive muscle stiffness (spasticity) in the legs, and difficulty walking. Most of the *SPG7* gene mutations change single protein building blocks (amino acids) in the paraplegin protein. When paraplegin is altered, 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

MedlinePlus Genetics provides information about Progressive external ophthalmoplegia

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

Tests Listed in the Genetic Testing Registry

- Tests of SPG7 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6687[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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)

Catalog of Genes and Diseases from OMIM

- SPG7 MATRIX AAA PEPTIDASE SUBUNIT, PARAPLEGIN; SPG7 (https://omim.org/entry/602783)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6687)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SPG7[gene])

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Genomic Location

The *SPG7* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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