

SPG11 gene

SPG11 vesicle trafficking associated, spatacsin

Normal Function

The *SPG11* gene provides instructions for making the protein spatacsin. Spatacsin is active (expressed) throughout the nervous system, although its exact function is unknown. Researchers speculate that it may help control the activity of particular genes (gene expression) or play a role in the transport (trafficking) of proteins. Spatacsin may also be involved in the maintenance of axons, which are specialized extensions of nerve cells (neurons) that transmit impulses throughout the nervous system.

Health Conditions Related to Genetic Changes

Spastic paraplegia type 11

More than 65 mutations in the *SPG11* gene have been found to cause spastic paraplegia type 11. Most of these mutations change the structure of the spatacsin protein. The effect that the altered spatacsin protein has on the nervous system is not known. Researchers suggest that mutations in spatacsin may cause the signs and symptoms of spastic paraplegia type 11 by interfering with the protein's proposed role in the maintenance of axons.

Amyotrophic lateral sclerosis

MedlinePlus Genetics provides information about Amyotrophic lateral sclerosis

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Other Names for This Gene

- FLJ21439
- KIAA1840
- spastic paraplegia 11 (autosomal recessive)
- SPATACSIN
- SPTCS_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SPG11 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80208\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80208[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SPG11%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+2160+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SPG11 VESICLE TRAFFICKING ASSOCIATED, SPATACSIN; SPG11 (<https://omim.org/entry/610844>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/80208>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SPG11\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SPG11[gene]))

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Genomic Location

The *SPG11* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

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