



SPG11 gene

SPG11, spatacsin vesicle trafficking associated

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.

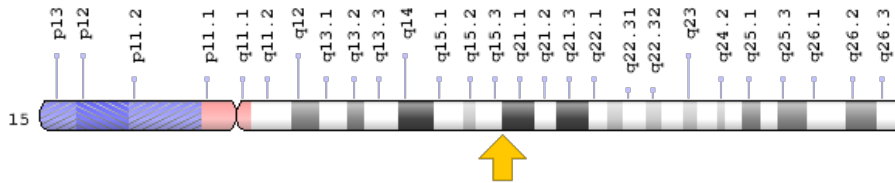
Amyotrophic lateral sclerosis

Charcot-Marie-Tooth disease

Chromosomal Location

Cytogenetic Location: 15q21.1, which is the long (q) arm of chromosome 15 at position 21.1

Molecular Location: base pairs 44,562,696 to 44,663,678 on chromosome 15 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Clinical Information from GeneReviews

- Spastic Paraplegia 11
<https://www.ncbi.nlm.nih.gov/books/NBK1210>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG11%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SPG11 GENE
<http://omim.org/entry/610844>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SPG11.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SPG11%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:11226
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:80208>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/80208>
- UniProt
<https://www.uniprot.org/uniprot/Q96J17>

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