

## SPART gene

spartin

### Normal Function

The *SPART* gene provides instructions for producing a protein called spartin, whose function is not entirely understood. Researchers believe that spartin may be involved in a variety of cell functions, from breaking down proteins to transporting materials from the cell surface into the cell (endocytosis). Spartin is found in a wide range of body tissues, including the nervous system.

### Health Conditions Related to Genetic Changes

#### Troyer syndrome

One mutation has been identified in people with Troyer syndrome. This mutation deletes a DNA building block (nucleotide) in the *SPART* gene. Specifically, this mutation deletes the nucleotide adenosine at position 1110 in the gene, written as 1110delA. This mutation results in an abnormally shortened, nonfunctioning spartin protein. It is unclear how this mutation causes the signs and symptoms of Troyer syndrome.

### Other Names for This Gene

- KIAA0610
- SPARTIN
- spastic paraplegia 20
- spastic paraplegia 20 (Troyer syndrome)
- spastic paraplegia 20, spartin (Troyer syndrome)
- SPG20
- SPG20\_HUMAN
- TAHCCP1
- trans-activated by hepatitis C virus core protein 1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SPART ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23111\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23111[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SPG20%29+OR+%28SPART%29%29+OR+%28spartin%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- SPARTIN; SPART (<https://omim.org/entry/607111>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23111>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SPART\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SPART[gene]))

## References

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## **Genomic Location**

The *SPART* gene is found on chromosome 13 (<https://medlineplus.gov/genetics/chromosome/13/>).

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