SPART gene

spartin

Normal Function

The *SPART* gene provides instructions for producing a protein called spartin. Spartin is found in a number of body tissues, including the brain. Research shows that spartin likely plays a role in regulating the activity of endosomes, which are structures inside the cell that are involved in sorting, transporting, and recycling proteins and other materials. Spartin may also play a role in mitochondrial function. Mitochondria are the energy-producing centers inside the cell.

Spartin is also believed to regulate the size and number of lipid droplets inside the cell. Specifically, spartin may be involved in delivering lipid droplets to the cell's recycling center. Lipid droplets help cells use and store fats, which are an important energy source. Spartin may also be involved in transporting materials from the cell surface into the cell (endocytosis).

Health Conditions Related to Genetic Changes

Troyer syndrome

Some variants (also called mutations) in the *SPART* gene cause Troyer syndrome. Troyer syndrome is a type of hereditary spastic paraplegia (also called hereditary spastic paraparesis), a group of conditions that cause progressive stiffness (spasticity) and weakness of the leg muscles. The first known variant associated with Troyer syndrome was found in the Old Order Amish population in Ohio. This variant deletes a single DNA building block (nucleotide) in the *SPART* gene. Specifically, this variant deletes the nucleotide adenosine at position 1110 in the gene. This change results in an abnormally short, nonfunctioning spartin protein. Troyer syndrome has since been diagnosed in other populations and additional variants have been found.

The variants that cause Troyer syndrome reduce the amount of functioning protein inside the cell. Cells that do not have enough normal spartin cannot break down lipid droplets properly, which allows fat molecules to build up inside the cell. This buildup of fat molecules in the cells of the brain is believed to contribute to the signs and symptoms of Troyer syndrome.

Other Names for This Gene

- KIAA0610
- SPARTIN
- SPG20
- TAHCCP1

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])

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)

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])

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Genomic Location

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

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