Spastic paraplegia type 31

Description

Spastic paraplegia type 31 is one of a group of genetic disorders known as hereditary spastic paraplegias. These disorders are characterized by progressive muscle stiffness (spasticity) and the development of paralysis of the lower limbs (paraplegia) caused by degeneration of nerve cells that trigger muscle movement (motor neurons). Hereditary spastic paraplegias are divided into two types: pure and complicated. The pure types involve only the lower limbs, while the complicated types also involve the upper limbs and other areas of the body, including the brain. Spastic paraplegia type 31 is usually a pure hereditary spastic paraplegia, although a few complicated cases have been reported.

The first signs and symptoms of spastic paraplegia type 31 usually appear before age 20 or after age 30. An early feature is difficulty walking due to spasticity and weakness, which typically affect both legs equally. People with spastic paraplegia type 31 can also experience progressive muscle wasting (amyotrophy) in the lower limbs, exaggerated reflexes (hyperreflexia), a decreased ability to feel vibrations, reduced bladder control, and high-arched feet (pes cavus). As the condition progresses, some individuals require walking support.

Frequency

Spastic paraplegia type 31 is one of a subgroup of hereditary spastic paraplegias known as autosomal dominant hereditary spastic paraplegia, which has an estimated prevalence of one to 12 per 100,000 individuals. Spastic paraplegia type 31 accounts for 3 to 9 percent of all autosomal dominant hereditary spastic paraplegia cases.

Causes

Spastic paraplegia type 31 is caused by mutations in the *REEP1* gene. This gene provides instructions for making a protein called receptor expression-enhancing protein 1 (REEP1), which is found in neurons in the brain and spinal cord. The REEP1 protein is located within cell compartments called mitochondria, which are the energy-producing centers in cells, and the endoplasmic reticulum, which helps with protein processing and transport. The REEP1 protein plays a role in regulating the size of the endoplasmic reticulum and determining how many proteins it can process. The function of the REEP1 protein in the mitochondria is unknown.

REEP1 gene mutations that cause spastic paraplegia type 31 result in a short, nonfunctional protein that is usually broken down quickly. As a result, there is a reduction in functional REEP1 protein. It is unclear how *REEP1* gene mutations lead to the signs and symptoms of spastic paraplegia type 31. Researchers have shown that mitochondria in cells of affected individuals are less able to produce energy, which may contribute to the death of neurons and lead to the progressive movement problems of spastic paraplegia type 31; however, the exact mechanism that causes this condition is unknown.

Learn more about the gene associated with Spastic paraplegia type 31

- REEP1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Autosomal dominant spastic paraplegia 31
- Spastic paraplegia 31
- SPG31

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hereditary spastic paraplegia 31 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853247/)
- Genetic Testing Registry: Hereditary spastic paraplegia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0037773/)

Genetic and Rare Diseases Information Center

- Autosomal dominant spastic paraplegia type 31 (https://rarediseases.info.nih.gov/diseases/10817/index)
- Hereditary spastic paraplegia (https://rarediseases.info.nih.gov/diseases/6637/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- SPASTIC PARAPLEGIA 31, AUTOSOMAL DOMINANT; SPG31 (https://omim.org/entry/610250)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SPG31%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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