Spastic paraplegia type 8

Description

Spastic paraplegia type 8 is part 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). Hereditary spastic paraplegias are divided into two types: pure and complex. The pure types involve only the nerves and muscles controlling the lower limbs and bladder, whereas the complex types also have significant involvement of the nervous system in other parts of the body. Spastic paraplegia type 8 is a pure hereditary spastic paraplegia.

Like all hereditary spastic paraplegias, spastic paraplegia type 8 involves spasticity of the leg muscles and muscle weakness. People with this condition can also experience exaggerated reflexes (hyperreflexia), a decreased ability to feel vibrations, muscle wasting (amyotrophy), and reduced bladder control. The signs and symptoms of spastic paraplegia type 8 usually appear in early to mid-adulthood. As the muscle weakness and spasticity get worse, some people may need the aid of a cane, walker, or wheelchair.

Frequency

The prevalence of all hereditary spastic paraplegias combined is estimated to be 1 to 18 in 100,000 people worldwide. Spastic paraplegia type 8 likely accounts for only a small percentage of all spastic paraplegia cases.

Causes

Mutations in the *WASHC5* gene cause spastic paraplegia type 8. The *WASHC5* gene provides instructions for making a protein called strumpellin. Strumpellin is active (expressed) throughout the body, although its exact function is unknown. The protein's structure suggests that strumpellin may interact with the structural framework inside cells (the cytoskeleton) and may attach (bind) to other proteins.

WASHC5 gene mutations are thought to change the structure of the strumpellin protein. It is unknown how the altered strumpellin protein causes the signs and symptoms of spastic paraplegia type 8.

Learn more about the gene associated with Spastic paraplegia type 8

- WASHC5

Inheritance

Spastic paraplegia type 8 is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- Autosomal dominant spastic paraplegia 8
- Hereditary spastic paraplegia 8
- Spastic paraplegia 8
- SPG 8

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hereditary spastic paraplegia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0037773/)
- Genetic Testing Registry: Hereditary spastic paraplegia 8 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1863704/)

Genetic and Rare Diseases Information Center

- Autosomal dominant spastic paraplegia type 8 (https://rarediseases.info.nih.gov/diseases/9591/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 8, AUTOSOMAL DOMINANT; SPG8 (https://omim.org/entry/603563)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SPG8%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Hedera P, Rainier S, Alvarado D, Zhao X, Williamson J, Otterud B, Leppert M,Fink JK. Novel locus for autosomal dominant hereditary spastic paraplegia, onchromosome 8q. Am J Hum Genet. 1999 Feb;64(2):563-9. doi: 10.1086/302258. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9973294) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1377766/)
- Meijer IA, Valdmanis PN, Rouleau GA. Spastic Paraplegia 8. 2008 Aug 13[updated 2020 May 21]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE,Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet].Seattle (WA): University of Washington, Seattle; 1993-2024. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1827/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301727)
- Rocco P, Vainzof M, Froehner SC, Peters MF, Marie SK, Passos-Bueno MR, Zatz M.Brazilian family with pure autosomal dominant spastic paraplegia maps to 8q: analysis of muscle beta 1 syntrophin. Am J Med Genet. 2000 May 15;92(2):122-7. doi: 10.1002/(sici)1096-8628(20000515)92:23.0.co;2-b. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10797436)
- Valdmanis PN, Meijer IA, Reynolds A, Lei A, MacLeod P, Schlesinger D, Zatz M, Reid E, Dion PA, Drapeau P, Rouleau GA. Mutations in the KIAA0196 gene at theSPG8 locus cause hereditary spastic paraplegia. Am J Hum Genet. 2007Jan;80(1):152-61. doi: 10.1086/510782. Epub 2006 Dec 1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17160902) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1785307/)

Last updated March 1, 2009