Spastic paraplegia type 7

Spastic paraplegia type 7 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 the lower limbs. The complex types involve the lower limbs and can also affect the upper limbs to a lesser degree; the structure or functioning of the brain; and the nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat, and sound (the peripheral nervous system). Spastic paraplegia type 7 can occur in either the pure or complex form.

Like all hereditary spastic paraplegias, spastic paraplegia type 7 involves spasticity of the leg muscles and increased muscle weakness. People with this form of spastic paraplegia can also experience exaggerated reflexes (hyperreflexia) in the arms; speech difficulties (dysarthria); difficulty swallowing (dysphagia); involuntary movements of the eyes (nystagmus); mild hearing loss; abnormal curvature of the spine (scoliosis); high-arched feet (pes cavus); numbness, tingling, or pain in the arms and legs (sensory neuropathy); disturbance in the nerves used for muscle movement (motor neuropathy); and muscle wasting (amyotrophy). The onset of symptoms varies greatly among those with spastic paraplegia type 7; however, abnormalities in muscle tone and other features are usually noticeable in adulthood.

Frequency

The prevalence of all hereditary spastic paraplegias combined is estimated to be 2 to 6 in 100,000 people worldwide. Spastic paraplegia type 7 likely accounts for only a small percentage of all spastic paraplegia cases.

Causes

Mutations in the SPG7 gene cause spastic paraplegia type 7. The SPG7 gene provides instructions for producing a protein called paraplegin. Located within the inner membrane of the energy-producing centers of cells (mitochondria), paraplegin is one of the proteins that form a complex called the m-AAA protease. The m-AAA protease is responsible for assembling ribosomes (cellular structures that process the cell's genetic instructions to create proteins) and removing nonfunctional proteins in the mitochondria. When there is a mutation in paraplegin, the m-AAA protease cannot function correctly. Nonfunctional m-AAA proteases cause a build up of unusable proteins in the mitochondria of nerve cells, which can result in swelling of the cell, reduced cell signaling, and impaired cell movement, leading to the major signs and symptoms of spastic paraplegia type 7.
Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Autosomal Recessive Hereditary Spastic Paraplegia
- Hereditary Spastic Paraplegia
- hereditary spastic paraplegia, paraplegin type
- spastic paraplegia 7

Diagnosis & Management

Genetic Testing Information

- What is genetic testing? 
/primer/testing/genetictesting
- Genetic Testing Registry: Spastic paraplegia 7

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22spastic+paraplegia+type+7%22+OR+%22Spastic+Paraplegia%2C+Hereditary%22+OR+%22Spastic+Paraplegia%22

Other Diagnosis and Management Resources

- GeneReview: Hereditary Spastic Paraplegia Overview 
  https://www.ncbi.nlm.nih.gov/books/NBK1509
- GeneReview: Spastic Paraplegia 7 
  https://www.ncbi.nlm.nih.gov/books/NBK1107
- Spastic Paraplegia Foundation, Inc.: Treatments and Therapies 
  https://sp-foundation.org/understanding-pls-hsp/treatments.html

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Neurologic Diseases 
  https://medlineplus.gov/neurologicdiseases.html
- Health Topic: Neuromuscular Disorders 
  https://medlineplus.gov/neuromusculardisorders.html
Health Topic: Paralysis
https://medlineplus.gov/paralysis.html

Health Topic: Peripheral Nerve Disorders
https://medlineplus.gov/peripheralnervedisorders.html

Genetic and Rare Diseases Information Center

Hereditary spastic paraplegia
https://rarediseases.info.nih.gov/diseases/6637/hereditary-spastic-paraplegia

Additional NIH Resources

National Institute of Neurological Disorders and Stroke: Hereditary Spastic Paraplegia
https://www.ninds.nih.gov/Disorders/All-Disorders/Hereditary-spastic-paraplegia-Information-Page

Educational Resources

MalaCards: spastic paraplegia 7, autosomal recessive
https://www.malacards.org/card/spastic_paraplegia_7_autosomal_recessive

Merck Manual Consumer Version

Orphanet: Hereditary spastic paraplegia
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=685

Orphanet: Spastic paraplegia type 7
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=99013

Patient Support and Advocacy Resources

National Ataxia Foundation
https://ataxia.org/

National Organization for Rare Disorders (NORD): Hereditary Spastic Paraplegia
https://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/

RareConnect
https://www.rareconnect.org/en/community/hereditary-spastic-paraplegia

Clinical Information from GeneReviews

Hereditary Spastic Paraplegia Overview
https://www.ncbi.nlm.nih.gov/books/NBK1509

Spastic Paraplegia 7
https://www.ncbi.nlm.nih.gov/books/NBK1107
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG7%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days+%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SPASTIC PARAPLEGIA 7, AUTOSOMAL RECESSIVE
  http://omim.org/entry/607259

Medical Genetics Database from MedGen

- Spastic paraplegia 7

Sources for This Summary
