



Troyer syndrome

Troyer syndrome 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). Troyer syndrome is a complex hereditary spastic paraplegia.

People with Troyer syndrome can experience a variety of signs and symptoms. The most common characteristics of Troyer syndrome are spasticity of the leg muscles, progressive muscle weakness, paraplegia, muscle wasting in the hands and feet (distal amyotrophy), small stature, developmental delay, learning disorders, speech difficulties (dysarthria), and mood swings. Other characteristics can include exaggerated reflexes (hyperreflexia) in the lower limbs, uncontrollable movements of the limbs (choreoathetosis), skeletal abnormalities, and a bending outward (valgus) of the knees.

Troyer syndrome causes the degeneration and death of muscle cells and motor neurons (specialized nerve cells that control muscle movement) throughout a person's lifetime, leading to a slow progressive decline in muscle and nerve function. The severity of impairment related to Troyer syndrome increases as a person ages. Most affected individuals require a wheelchair by the time they are in their fifties or sixties.

Frequency

About 20 cases of Troyer syndrome have been reported in the Old Order Amish population of Ohio. It has not been found outside this population.

Causes

Troyer syndrome is caused by a mutation in the *SPART* gene. 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.

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
- Cross-McKusick syndrome
- Hereditary Spastic Paraplegia
- spastic paraparesis, childhood-onset, with distal muscle wasting
- spastic paraplegia 20, autosomal recessive
- spastic paraplegia, autosomal recessive, Troyer type
- SPG20

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Troyer syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0393559/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Troyer+syndrome%22+OR+%22Paraplegia%2C+Spastic%22+OR+%22Spastic+Paraplegia%22>

Other Diagnosis and Management Resources

- GeneReview: Hereditary Spastic Paraplegia Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1509>
- GeneReview: Troyer Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1382>
- 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

- Troyer syndrome
<https://rarediseases.info.nih.gov/diseases/5372/troyer-syndrome>

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>
- National Institute of Neurological Disorders and Stroke: Troyer syndrome
<https://www.ninds.nih.gov/Disorders/All-Disorders/Troyer-syndrome-Information-Page>

Educational Resources

- MalaCards: spastic paraplegia 20, autosomal recessive
https://www.malacards.org/card/spastic_paraplegia_20_autosomal_recessive
- Merck Manual Consumer Version
<https://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/spinal-cord-disorders/hereditary-spastic-paraparesis>
- Orphanet: Hereditary spastic paraplegia
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=685

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>
- Troyer Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1382>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28troyer+syndrome%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SPASTIC PARAPLEGIA 20, AUTOSOMAL RECESSIVE
<http://omim.org/entry/275900>

Medical Genetics Database from MedGen

- Troyer syndrome
<https://www.ncbi.nlm.nih.gov/medgen/97950>

Sources for This Summary

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Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735361/>

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