Troyer Syndrome

Other names: Spastic paraplegia, 20 (SPG20) **Gene involved:** *SPAST*

What is Troyer Syndrome?

Troyer syndrome is an inherited autosomal recessive disorder that affects boys and girls equally. It is part of a group of disorders known as the hereditary spastic paraplegias. These disorders are characterized by progressive lower limb muscle stiffness and weakness that worsens as a person gets older. Sometimes families may have more than one child with Troyer syndrome.

What are the early signs and symptoms?

- Infants often have a lower birth weight than their healthy siblings, but their head size is usually normal size (appearing large for their body)
- Infants may struggle with their feeding and gain weight more slowly than usual
- Infants may drool excessively, and this can continue into adulthood
- Children are generally late in achieving the usual childhood developmental milestones particularly walking and talking

What Happens as Children Get Older?

- Children and adults with Troyer syndrome are shorter than their siblings. They may have a unique appearance with similar facial features to other people with Troyer syndrome.
- Walking is often clumsy from the start and parents often report their child trips more often
- Stiffness and weakness in the leg muscles, feet and hands slowly develops, with most children showing these signs before they are 10 years old
- Speech is generally slurred and unclear and worsens with age
- Children may find learning more difficult, but most can attend school like their siblings and do well with a little extra help and support
- Children and adults may struggle to control their emotions. They can have mood swings and become easily upset or laugh at inappropriate times.

Management

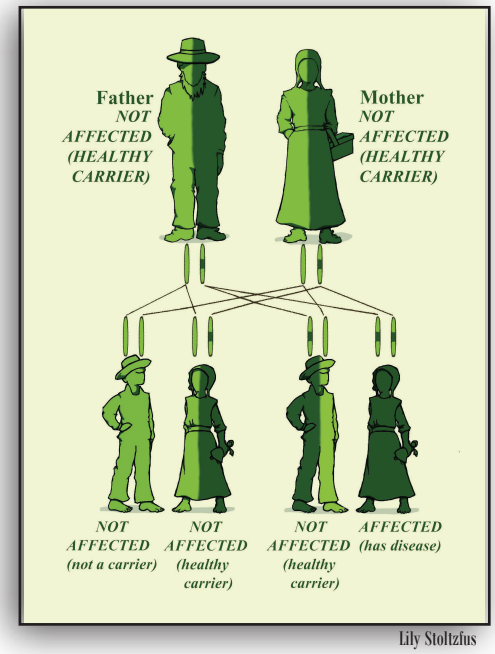
People with Troyer syndrome bring joy to their families. They will always require special support and therapy to reach their full potential. Although there is no cure for Troyer syndrome the following can be helpful

- Support from families and therapists improves mobility, communication skills and quality of life.
- Assistive walking devices and ankle and foot supports can help with walking
- Medications may help with excessive drooling and muscle stiffness

These helpful therapies should be coordinated through the doctor(s) assisting with the care of the affected child or adult.

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.



Lily Stoltzfus

Autosomal Recessive Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. Children with Troyer syndrome have a spelling change in BOTH copies of their *SPAST* genes. This means they have no working copy of the *SPAST* instruction.

People with one normal copy of the *SPAST* gene and one misspelled copy are called “healthy carriers”.

When two healthy carrier parents have a child, there is a:

- 1 in 4 chance the child will have Troyer syndrome, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *SPAST* gene and be neither affected nor be a carrier

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children’s Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**