Conditions + Treatments

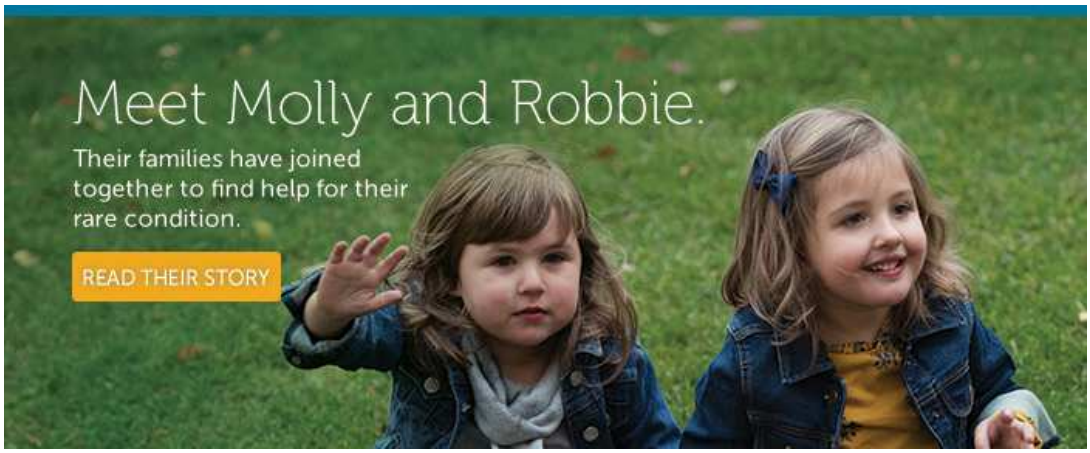
Spastic Parapalegia-47 (SPG47)

What is spastic parapalegia-47?

Spastic paraplegia-47 (SPG47) is an extremely rare, newly recognized genetic disorder resembling cerebral palsy. It is part of a group of disorders known as hereditary spastic paraplegia (HSP) and is sometimes called "HSP type 47."

SPG47 is present at birth and causes severe, progressive spasticity and muscle weakness, especially in the lower limbs. In infancy, children with SPG47 have very low muscle tone (**hypotonia**). As they grow, they develop spasticity (tight, stiff muscles that make movement difficult), together with cognitive deficits.

SPG47 is a recessive disorder caused by two copies of a mutation in a gene called AP4B1, one inherited from each parent. It is thought that these mutations affect the ability of cells in the nervous system to process proteins. Since very few cases have been reported to date, our understanding of the full effects of SPG47 is still emerging.



What are the symptoms of spastic parapalegia-47?

Virtually all children with SPG47 have:

- a "floppy" appearance in infancy due to low muscle tone
- increasing spasticity and paralysis in the lower limbs starting in early childhood
- intellectual disability
- **microcephaly** (a smaller than normal head)
- delayed motor development
- poor or absent speech development

Other known features of SPG47 can include the following (not every child will have these features):

- short stature
- late walking and later loss of the ability to walk independently
- dystonia (involuntary muscle contractions)
- seizures

Some children may also have facial differences that can include:

- high palate
- wide nasal bridge
- bulbous nose
- wide mouth
- protruding tongue
- short filtrum (the groove between the bottom of the nose and top of the lips)
- narrow forehead
- joint abnormalities, such as backwards bending of the knee or **hip dysplasia** (loose hip joint)
- flat feet or club feet

What is the long-term outlook for spastic parapalegia-47 (SPG47)?

Because of progressive leg spasticity, children with SPG47 often lose their ability to walk independently, requiring a walker or canes. Many become wheelchair-bound before their teens. Others can walk with a waddling gait. Some children known to have SPG47 have reached their early 20s, but long-term life expectancy for this condition is still unclear, as it was only first recognized in 2011. However, many children with hereditary spastic paraplegias in general have a normal life expectancy.

How is spastic parapalegia-47 treated?

There is unfortunately no specific treatment for SPG47. However, physical, occupational and speech therapies can help children develop and preserve motor and communication skills. Muscle relaxants and Botox injections can help relieve spasticity in the muscles, and physical therapy can increase strength and range of motion. At Boston Children's Hospital, we care for patients with spastic paraplegia in the **Department of Neurology**, the **Neuromuscular Center**, the **Cerebral Palsy Program** and the **Clinical Genetics** program.

Are my other children at risk?

Since SPG47 is a recessive disorder, the chances of you and your spouse having another affected child are 1 in 4. Genetic counseling is often helpful if you are planning to have more children.