Gillette Children's Specialty Healthcare

Conditions

Spinal Muscular Atrophy

What Is Spinal Muscular Atrophy (SMA)?

Spinal muscular atrophy (SMA) is a condition affecting the motor nerves that control muscular function. SMA makes activities such as crawling, walking, breathing and eating difficult for children.

The nerves affected by SMA don't have enough of an important protein, so they can't carry signals from the brain to the muscles. Without those signals, muscles weaken and decrease in mass (also known as atrophy).

SMA is caused by a change (mutation) in the survival motor neuron 1 gene (SMN1 gene). The SMN1 gene makes a protein known as survival motor neuron protein, which helps motor neurons grow and function properly.

Motor neurons control muscle movement. When a gene mutation decreases the amount of protein, a child loses motor neurons over time. That means muscle control gets worse over time as well.

The most common SMN1 change is a deletion. That means a piece of the gene is missing. Most people who have SMA have two deletions—one in each copy of the SMN1 gene. Roughly 5 percent of people who have SMA have one deleted copy, as well as a different change in the second copy of the SMN1 gene.

What Causes Spinal Muscular Atrophy (SMA)?

SMA is a hereditary condition that affects both males and females. Children who have SMA have a mutation in both copies of the SMN1 gene.

Parents of children who have SMA are usually carriers. That means one copy of their SMN1 gene has a mutation, while the other copy is normal. If both parents are carriers, there is a one in four chance that a child will have SMA. Carriers don't show symptoms of SMA, and usually have no history of the condition in their family.

In most cases, both parents of a child who has SMA are carriers. In a small number of cases, SMA develops from a new change (mutation), in the formation of either the egg or the sperm. Roughly one in 50 people are SMA carriers.

Types of Spinal Muscular Atrophy

The type of SMA is determined by the age of onset and the severity of symptoms.

SMA I

Also called infantile onset or Werdnig-Hoffmann disease, SMA I is the most severe form of the condition. Symptoms of SMA I usually develop by the time an infant is 6 months old. Most infants diagnosed with SMA I show severe muscle weakness and floppiness (also known as hypotonia). They have difficulty gaining strength and can't sit on their own. The muscle weakness also causes difficulty with swallowing and breathing.

SMA II

Sometimes called intermediate SMA, SMA II causes muscle weakness and hypotonia, but the problems are less severe than with SMA I. Babies and toddlers with type 2 spinal muscular atrophy learn to sit on their own, but have trouble standing or walking. As they grow, some children lose the ability to stand and move freely. At that point, they need to use mobility devices, such as powered wheelchairs

SMA III

Also called late-onset SMA (or Kugelberg-Welander disease), SMA III develops in children 18 months or older. Children who have SMA III walk on their own, but might have trouble jumping or using stairs. The severity of this form varies widely. Some people aren't diagnosed until adulthood. In those cases, the condition is sometimes called SMA IV, or adult-onset SMA.

Spinal Muscular Atrophy Symptoms and Effects

The symptoms of SMA syndrome differ depending on type. The most severe form of SMA usually affects essential functions important during your infant's first six months of life.

Less severe forms of SMA often have milder symptoms that don't develop until later in life. Even in milder cases, the disease gets worse over time (it's progressive)—but not to the same degree as with severe cases.

If your infant has SMA, you might look for symptoms such as:

- Delays in development of larger movements (gross motor skills)—or loss of skills already mastered.
- Breathing problems.
- Trouble feeding.
- Limp or floppy muscles.
- Poor head control.

- Lack of movement.
- Muscle weakness that grows worse.
- Failure to thrive.

If your child has SMA, you might look for symptoms such as:

- Delays in gross motor development, such as walking—or the loss of skills already mastered.
- Frequent respiratory infections that get worse with each infection.
- Nasal-sounding speech.
- Posture that gets worse over time.
- **Scoliosis** that gets worse over time.

Spinal Muscular Atrophy Diagnosis and Treatment

If your child has SMA, a health care professional might use the following tests and examinations to make a diagnosis:

- **Physical exam:** The physical exam tests gross motor abilities.
- **Electromyography Testing (EMG) :** An EMG involves inserting a very thin needle (electrode) into the muscle. The electrode sends a mild electrical current through the nerves to measure the muscle's ability to respond to stimulation.
- **Genetic testing:** This blood test shows any changes (mutations) in the SMN1 gene responsible for SMA.
- **Muscle biopsy test:** Doctors remove a small sample of tissue and examine it under a microscope, looking for signs of muscle degeneration.

Although there is no cure for SMA, supportive care can make its complications manageable. Once Gillette Children's Specialty Healthcare specialists make a diagnosis, your family will get a customized treatment plan.

Regular checkups are important for slowing the progression of symptoms and avoiding secondary complications. As children reach adulthood, we continue to provide age-appropriate SMA care through **Gillette Lifetime Specialty Healthcare** for teens (16 and older) and adults.

Patients who have SMA often see experts in these specialties and services at Gillette:

Medical and Specialties

- Nursing.

- **Neurology.**
- **Pulmonology and respiratory care.**
- **Rehabilitation medicine.**
- **Orthopedics.**
- **Neuropalliative care.**
- **Sleep medicine.**
- **Medical genetics and genetic counseling.**
- **Assistive Technology.**

Testing and Diagnostic Services

- Molecular genetic tests.
- Functional ability tests.
- Manual muscle tests.
- **Pulmonary function tests.**
- **Sleep studies (polysomnography).**
- **Swallowing study (fluoroscopy and videofluoroscopy).**
- **Seating evaluations.**
- **X-rays.**

Rehabilitation Therapies

- **Nutrition and feeding.**
- **Occupational therapy.**
- **Physical therapy.**
- **Speech and language therapy.**

Family Support

- **Child life specialists.**

- **Neuropsychologists.**
- **Psychologists.**
- **Social workers.**
- **Therapeutic recreation specialists.**

Integrated Care

If your child has SMA, you can rely on specialized treatment from Gillette, one of the region's top neuromuscular programs. The **Muscular Dystrophy Association** recognizes Gillette as a partner in providing multidisciplinary care for people who have SMA. Our combination of research and clinical services support your family's journey in living with and managing SMA.

Make An Appointment **651-290-8707**