

Newborn Screening FACT Sheet

Spinal Muscular Atrophy (SMA)

What is Spinal Muscular Atrophy (SMA)?

Spinal Muscular Atrophy (SMA) is an inherited condition that affects the cells in the spinal cord (motor neurons) that signal the muscles to work. Over time, when motor neurons don't function properly, the muscles get weaker and activities such as crawling, walking, sitting up, and controlling head movements become more difficult. Severe cases of SMA affect the muscles used for breathing and swallowing.

While children with SMA have limited physical function, learning and intellectual abilities are not affected. Children with SMA can think, learn, and build relationships with other people.

What Causes SMA?

SMA is caused by a partially missing or faulty gene known as the SMN1 gene. Babies receive two copies of this gene—one each from the mother and father. Because only one functioning SMN1 gene is needed to be healthy, parents may pass down a partially missing or faulty SMN1 copy (gene variant) without knowing it. A baby born with SMA received a SMN1 gene variant from both parents. When parents are carriers, meaning they have one functioning gene and one gene variant, they have a 25% chance (with each pregnancy) of having a child with SMA.

What are the Types and Symptoms of SMA?

The severity of SMA depends on changes to the SMN1 gene, and the presence of a closely related gene SMN-2. Generally, the more copies of SMN-2 an individual has, the milder the condition.

The disease is currently divided into five types based on the age of onset and severity of clinical symptoms. However, with treatment, individuals may gain more physical milestones than they would have otherwise.

As newborn screening for SMA becomes more common, infants can receive treatment even before symptoms begin. Because of these factors, doctors believe that we may soon stop describing SMA as specific “types” and instead focus on the highest motor milestone achieved: non-sitter, sitter, and walker. But even then, there will still be a wide range of severity associated with SMA.

TYPE 0 (PRENATAL ONSET SMA)

The most severe form of SMA and very rare. Symptoms begin before the baby is born. At birth, the newborn has severe weakness and difficulty feeding and breathing and requires respiratory and feeding support.

TYPE 1 (WERDNIG-HOFFMANN DISEASE OR INFANTILE-ONSET SMA)

The most common form of SMA. Symptoms begin at birth or within the first 6 months of life. Babies with Type 1 SMA have physical challenges such as muscle weakness, difficulty breathing, coughing and swallowing.

TYPE 2 (INTERMEDIATE OR CHRONIC INFANTILE SMA)

Symptoms generally start between 6 months and 18 months of age. Early symptoms may include muscle weakness and difficulty sitting-up without support.

TYPE 3 (KUGELBERG WELANDER SYNDROME)

Symptoms begin after 18 months of age and before 3 years of age, but Type 3 SMA can also be diagnosed into the teenage years. As a child grows, he/she may experience problems with balance, walking, and running.

TYPE 4 (ADULT-ONSET SMA)

Very rare form of SMA, less than 1% of all diagnosed cases. Individuals with Type 4 SMA are diagnosed later in life and usually after the age of 35. However, symptoms of mild muscle weakness in the legs and hips may begin as early as the late teens.

What is the Treatment for SMA?

There are multiple treatments for SMA approved by the U.S. Food and Drug Administration (FDA) and many other treatments being tested in clinical trials. This means that individuals with SMA and their families can choose the course of care and treatment.

EARLY TREATMENT

Regardless of what type of treatment is chosen, it is important that individuals with SMA begin therapy as soon after diagnosis as possible. This is especially important for SMN-enhancing therapies. When SMN levels are low, motor neuron cells shrink and eventually die. In infants with SMA Type 1, 90% of motor neurons have been lost by 6 months of age. And once these neurons are lost, they cannot be regenerated. Beginning therapy as early as possible is the only way to prevent this motor neuron loss.

MEDICATIONS

Early treatment with an FDA-approved medication can slow or stop the progression of SMA symptoms.

Spinraza (nusinersen) was the first FDA-approved therapy to treat SMA. It is an SMN-enhancing therapy that works by targeting the SMN2 gene, causing it to make more complete protein.

Evrysdi (risdiplam) is an FDA-approved therapy to treat SMA. It is an SMN-enhancing therapy that works by targeting the SMN2 gene. It is a small molecule that causes that gene to make more complete SMN protein.

GENE THERAPY

Zolgensma (onasemnogene abeparvovec-xioi) is an FDA-approved therapy to treat SMA. This type of treatment is referred to as gene therapy or gene replacement therapy. It is an SMN-enhancing therapy that works by replacing the missing or mutated SMN1 gene. A modified virus vector, AAV9, carries the replacement gene into the body but does not cause a viral infection. This virus vector with the SMN1 gene information is delivered to the cells with the new DNA.

More Information on SMA

Texas Department of State Health Services
1-800-252-8023 ext. 3957 or
dshs.texas.gov/newborn

SMA Education and Resources:

- Cure SMA: curesma.org
- National Institutes of Health: medlineplus.gov/genetics/condition/spinal-muscular-atrophy/
- Clinical trials for SMA: clinicaltrials.gov