

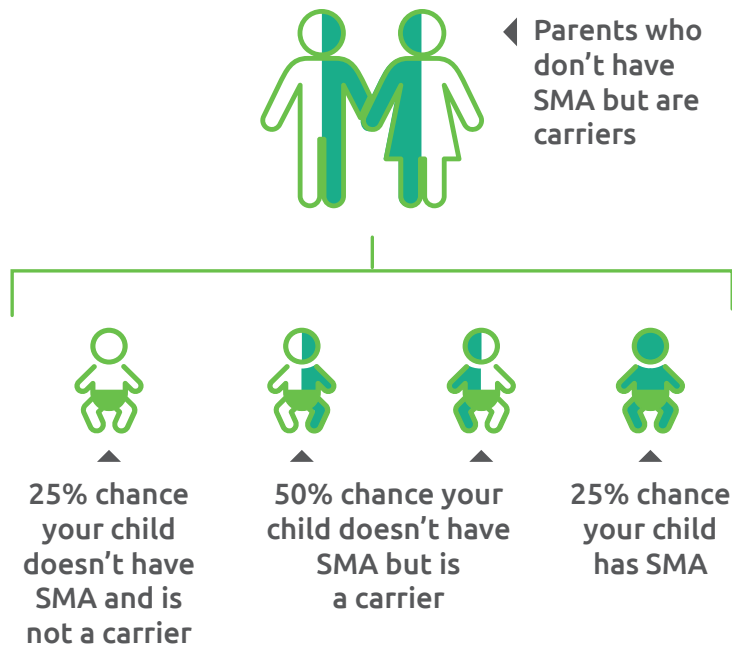
Facts About SMA



SMA is a progressive, rare genetic disease, yet it is the **number one genetic cause** of infant death.

How SMA is inherited

Spinal muscular atrophy (SMA) is an autosomal recessive disorder. This means a person must inherit one copy of a nonworking or missing gene from each parent to have the disease.



About 1 in



people in the United States (or 6 million* Americans) is a genetic carrier of SMA, and most don't know it.

*Calculations are based on an estimated US population of 300 million.



SMA affects about **1 in every 10,000** babies born each year.

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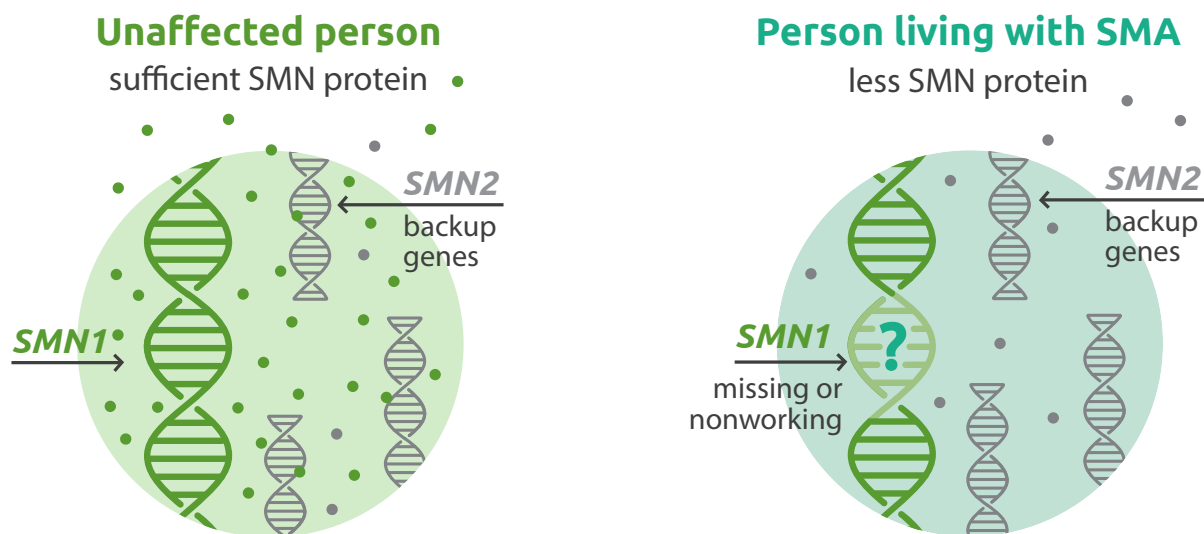
What causes SMA?

The genetic root cause of SMA is a survival motor neuron 1 (*SMN1*) gene that is missing or not working properly. When this main gene is missing or not working properly, the body can't make enough survival motor neuron (SMN) protein, which is needed for motor neuron cell survival. Motor neuron cells are responsible for communicating with the muscles and telling them to work properly. A person is born with a certain amount of these cells. Without enough SMN protein, motor neuron cells become weaker and weaker and eventually stop working, lose all function, and die. As a result, things many of us take for granted, like breathing, eating, speaking, and lifting the head, become difficult. **Once motor neuron cells die, they cannot be brought back.**

The role of a backup gene

There is a backup gene for the *SMN1* gene, called the *SMN2* gene. People can have one or more copies of this backup gene. This gene, like the *SMN1* gene, tells the body to make SMN protein. For people with SMA, the *SMN2* gene is the only source of SMN protein; however, it is unable to produce as much working protein as the *SMN1* gene. **In fact, the *SMN2* gene makes only about 10% of working protein compared to the protein produced by the *SMN1* gene.** Even people with several copies of the *SMN2* gene may not produce as much SMN protein as those with the working *SMN1* gene, and their motor neuron cells may not work as they should. Usually, the more copies of the *SMN2* gene a person has, the less severe his or her SMA is.

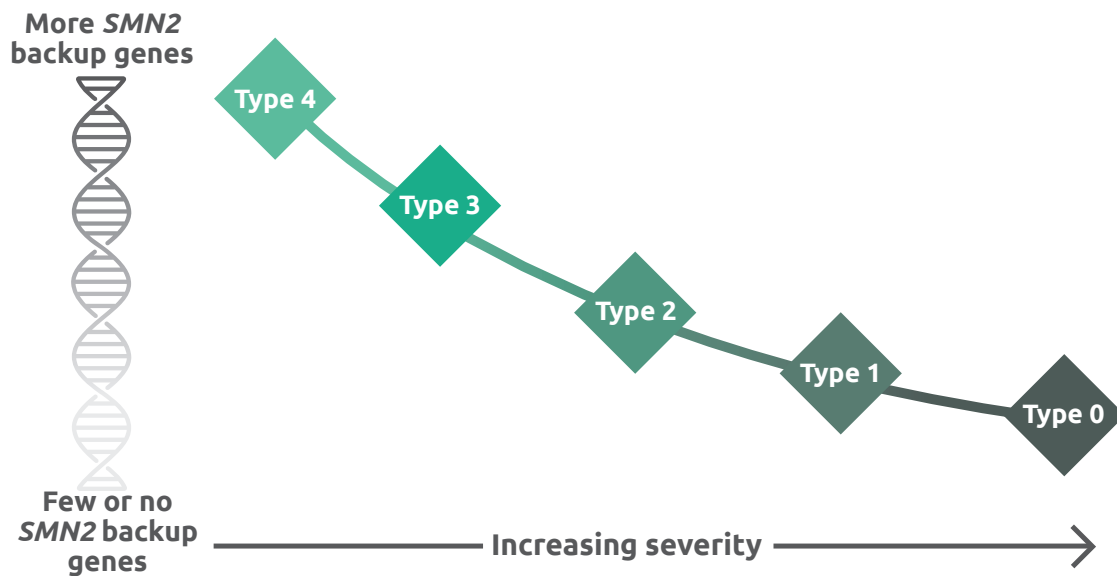
The *SMN1* and *SMN2* genes



Facts About SMA

SMA has a wide range of severity

Because some people can have more copies of the *SMN2* backup gene and others can have fewer, there is a wide range of severity in individuals affected by SMA. There are 4 main types of SMA (Types 1-4) that range in severity and when symptoms first appear. It can be fatal in some people and less severe in others. SMA Type 1 is the most common and is very serious. The fifth, and most severe, form of SMA is Type 0.



Learn more about advocacy groups that support and educate families living with SMA at [AveXis.com](https://www.avexis.com). In addition, search online for SMA communities that are dedicated to helping families and sharing experiences.

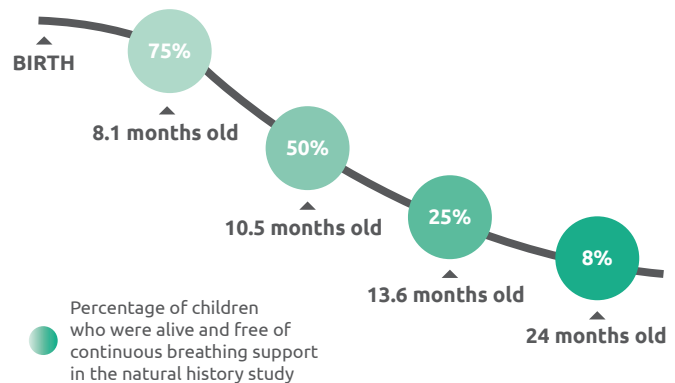
Facts About SMA

SMA Type 1

SMA Type 1 is the most common form of SMA and affects about **6 out of every 10** children with SMA. Most children with SMA Type 1 have 2 copies of the *SMN2* backup gene. In some cases, children may have more than 2 copies or fewer than 2 copies. SMA Type 1 is a life-threatening condition that needs immediate attention. SMA Type 1 can quickly lead to the need for breathing and eating support, and without treatment is likely to be fatal within the first 2 years of life. SMA Type 1 is very severe, but signs and symptoms may vary at first—which means caregivers may not see them right away. Knowing what signs and symptoms to look for is critical in getting a diagnosis and starting a treatment.

SMA Type 1 is life threatening

Only 8% of children in the natural history of SMA Type 1 were alive and free of continuous breathing support at 24 months old. Continuous breathing support means that children needed a machine to help them breathe for at least 16 hours per day for 2 weeks or more.



Signs and symptoms of SMA may include:

- A “floppy” baby or hypotonia
- Small or weak muscles
- Difficulty breathing/belly breathing
- Feeding issues, like choking or trouble swallowing
- Weak sucking and labored breathing during feeding
- Tongue fluttering
- Bell-shaped chest (a result of muscle weakness)
- Weak cough
- Lack of reflexes
- “Frog legs” or the inability to kick while lying on back
- Lack of motor development, like being unable to lift head or roll over
- Inability to sit
- Weak cry

Take action fast

Making a quick and informed decision is critical because SMA affects motor neuron cells that control muscles throughout the body, and those muscles weaken and waste away every day.

