How Spinal Muscular Atrophy Happens

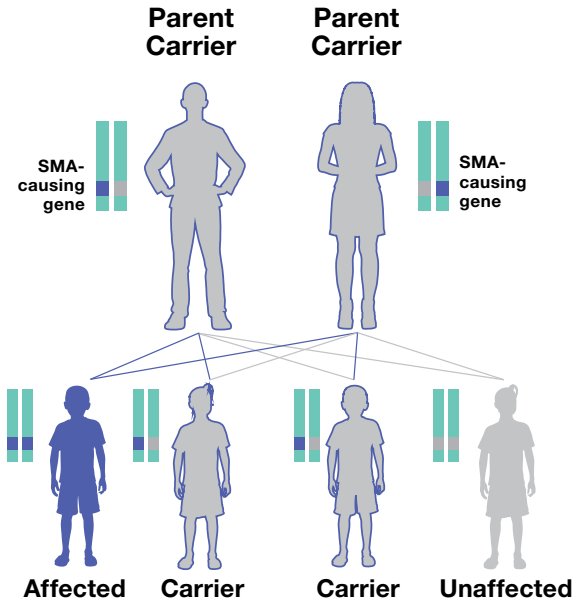
Inheritance patterns

Every person has two copies of each gene — one from their mother and one from their father. There are three ways an SMA-causing gene can be passed along in a family.

THE MOST COMMON CAUSE of spinal muscular atrophy (SMA) involves genetic changes or deletions of the *SMN1* gene. This gene is responsible for making the survival motor neuron (SMN) protein, which helps the body maintain nerve cells that control muscles. Some people inherit deleted or non-functional *SMN1* genes from one or both parents and do not produce functional SMN protein. Another gene, *SMN2*, also produces a lot of non-functional SMN protein. Together, *SMN1* gene defects cause disease, and the number of *SMN2* gene copies influences disease severity.

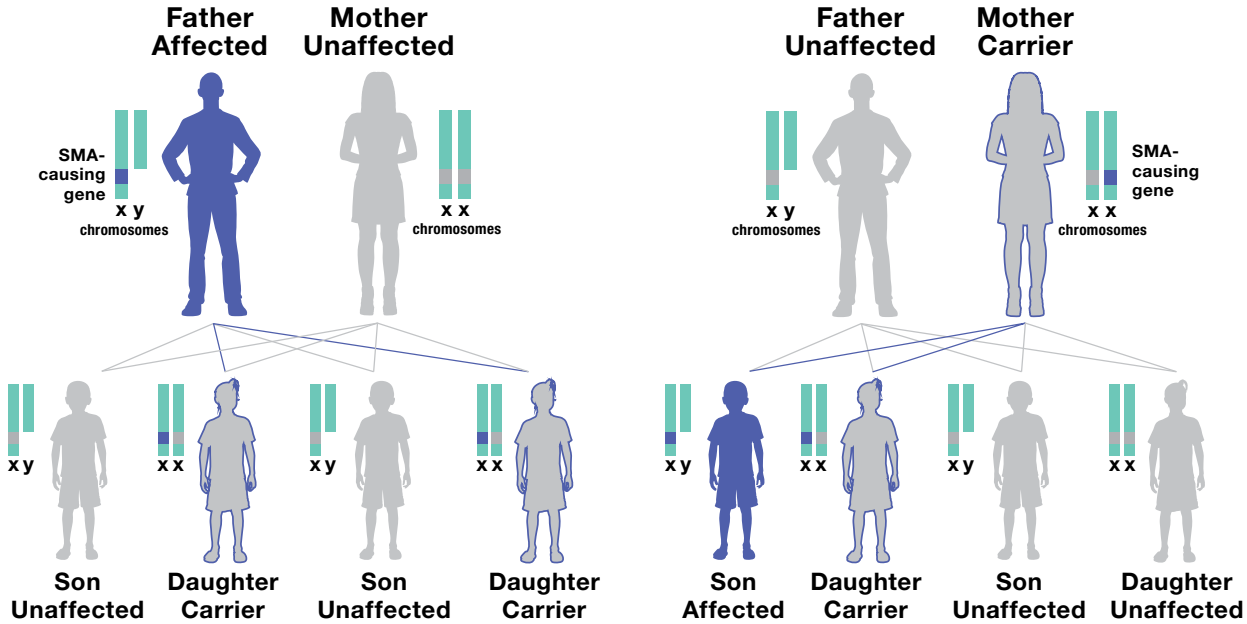
1. Autosomal recessive inheritance

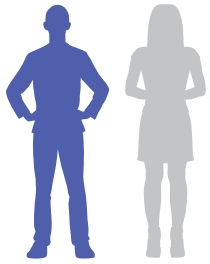
This is the most common inheritance pattern in SMA. A carrier has one working copy of the *SMN1* gene and does not have SMA symptoms.



2. X-linked recessive inheritance

In some cases, a change in a gene on the X chromosome will cause SMA. Because males have only one copy of the X chromosome, males who inherit the disease-causing gene will have SMA, while females who inherit the gene will be carriers.

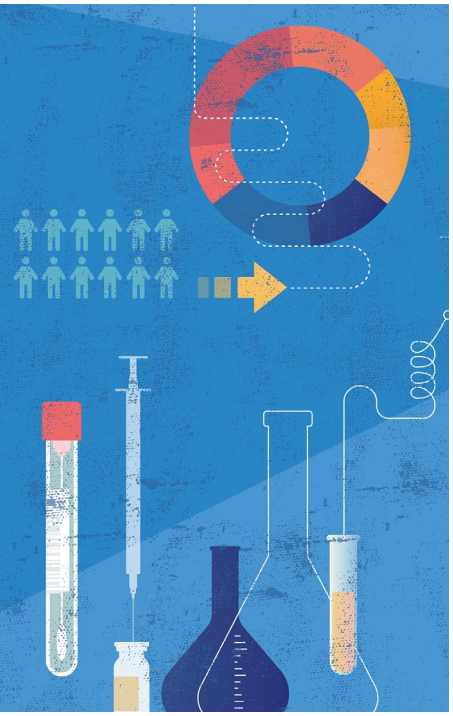




3. Autosomal dominant inheritance

When someone inherits the disease-causing gene from only one parent and displays symptoms of the condition, this is called autosomal dominant. This inheritance pattern is extremely rare in SMA.

Genetic testing for SMA



What does genetic testing do?

Many neuromuscular diseases have similar symptoms. Genetic testing can confirm a diagnosis of SMA and determine the exact type of SMA.

Knowing about your diagnosis is important. It allows you to understand potential health implications and to advocate for your healthcare needs. It also empowers you to connect with others who have the same condition through support networks and to seek out research and clinical trials that could be relevant for you.

More educational resources at mda.org/education:

- **How Genetic Neuromuscular Disease Happens**
- **Genetics and Neuromuscular Disease**
- **Basics of Genetic Testing**

Why do I need a genetic test?

A genetically confirmed diagnosis is important because:

- With several treatments for SMA now available and more on the horizon, a genetic diagnosis is often required to be eligible for certain treatments or clinical trials.
- Genetic testing can determine the type of SMA and provide other information that helps doctors understand what the course of the disease is likely to be.
- Genetic confirmation can help determine other family members' risk for SMA.

Genetic testing can be paid for in the following ways:

- Covered by insurance
- Covered by a company that supports treatment discovery for SMA
- Self-paid

Images: iStock.com/DrAfter123

MDA's Resource Center may be able to help you find resources for genetic testing. Call us at 833-ASK-MDA1.

This resource was developed with the expertise and knowledge of Laura Duncan, MS, CGC, from Monroe Carell Jr. Children's Hospital at Vanderbilt, and Krista Schatz, MS, CGC, from Johns Hopkins Hospital.

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