CARRIERS OF SPINAL MUSCULAR ATROPHY

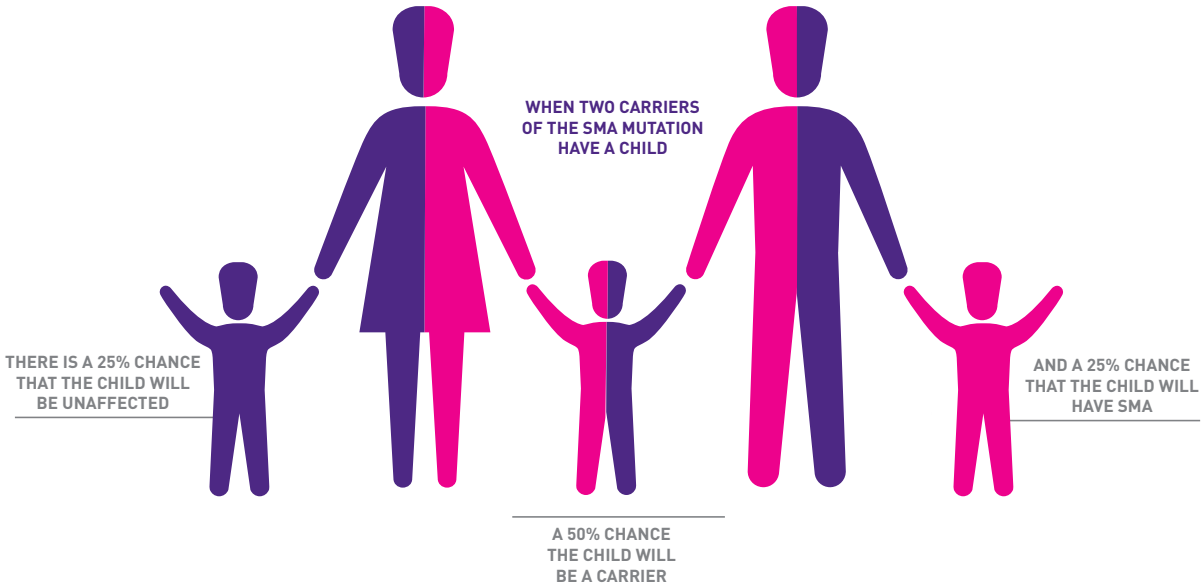
Most people have two functioning copies of the SMN1 gene. People with one non-working copy and one working copy of the gene are called "carriers."

Carriers generally do not show signs and symptoms of SMA, but could be at risk to have a child affected with the condition.

Approximately 1 in 50 people is a genetic carrier for SMA. Most carriers have no idea they are carriers until they have a child born with SMA.

How SMA is Inherited

SMA is an autosomal recessive genetic condition. This means that a child must inherit two non-working copies of the SMN1 gene, typically one from each parent, in order to have SMA.



cure SMA Make today a breakthrough.

If only one parent is a carrier, the child is usually not at risk for SMA (though the child does have a 50% chance of also being a carrier). In very rare cases, spontaneous genetic changes in the SMN1 gene can occur during egg or sperm production. In this situation, only one parent will be a carrier. In addition, a small percentage of carriers have genetic changes that cannot be identified through current testing technology. In this case, it will appear as though the disease has been caused by a single carrier.

Carrier Testing

A DNA test is the only way to know if a person is a carrier. The DNA test is a simple procedure, based on a blood test. In the general population, this test can detect about 95% of carriers. However, in African-American populations, detection is closer to 70%. This is because a difficult to detect mutation is seen more frequently in African-American populations than in other races.

The American College of Obstetricians and Gynecologists recommends that all women who are thinking about becoming pregnant or who are already pregnant be offered carrier screening for SMA and other genetic conditions. In addition, individuals with a family history of SMA are encouraged to have carrier screening. Deciding whether or not to undergo genetic testing is highly personal, and we strongly recommend discussing this with a physician or genetic counselor. Carrier screening via saliva testing is also available as an alternative to a blood test.

Reproductive Choices

For couples who are carriers, reproductive decisions can be sensitive. A number of options are available, such as no testing, prenatal testing, adoption, and pre-implantation genetic diagnosis (PGD). PGD screens embryos for genetic disorders and selects the unaffected embryos for implantation.

Currently, there are two FDA-approved treatments for individuals with SMA. These treatments have demonstrated beneficial results in many people with SMA. Additional treatments for SMA are being studied. Visit www.cureSMA.org/treatment

Cure SMA believes that your family has the right to choose whatever option is best for your own values. We help families understand their options and provide resources to support their decision-making process. We do not advocate any specific course of action, nor do we pressure families to choose one way or the other.

We encourage each family to discuss their situation with a physician, genetic counselor, and—if helpful—a therapist or a spiritual advisor.