

SPINAL MUSCULAR ATROPHY

*One person in 40
is a carrier
of SMA.*

Are you?



CLAIRE ALTMAN HEINE FOUNDATION, INC.
for the prevention of Spinal Muscular Atrophy

SPINAL MUSCULAR ATROPHY

(SMA) is the number ONE genetic killer of children under two. SMA is a devastating and relatively common children's genetic disease. One in every 6,000 babies is born with SMA. Infants diagnosed before six months of age (over 50% of those affected) have only a 5% chance of reaching their second birthday. While other organizations are working hard to find a treatment or cure, the Claire Altman Heine Foundation uses its funding to identify carriers of SMA, support population-based SMA carrier screening, raise awareness of SMA and educate the public and medical communities about SMA.



WHAT IS SMA?

Spinal Muscular Atrophy (SMA) is a motor neuron disease characterized by the progressive degeneration of nerve cells in the spinal cord and brainstem, leading to muscle weakness, respiratory complications and is often fatal in early childhood.



WHO IS AFFECTED BY SMA?

- SMA is the second most common diseased gene.
- Two children in the United States are born with SMA each day.
- SMA is an equal opportunity killer; SMA has *no* gender or racial preferences.
- One in every 40 people carries the mutated gene that causes SMA. That is *seven million* Americans.
- SMA is relatively common yet virtually unheard of in the general public. SMA is as common as Cystic Fibrosis in the entire population and Tay-Sachs in the Jewish population.



WHAT CAUSES SMA?

SMA is an autosomal recessive genetic disease. A child will be affected by SMA if both parents carry the mutated gene and pass this gene on to their child. If both parents are carriers of SMA a child has a 25% chance of being affected

with SMA. If both parents are carriers of SMA a child has a 50% chance of being a carrier of SMA.



THINKING ABOUT STARTING A FAMILY?

It is important to be informed about carrier screening and speak with your doctor or genetic counselor before initiating a pregnancy. You could be a carrier of SMA and not even know it. Carriers of the mutated gene for SMA have no symptoms and are not sick. Carriers of SMA typically have no traceable family history of the disease. Talk to your doctor or genetic counselor and get tested, it's a simple blood test.

WHAT IS THE PURPOSE OF SMA CARRIER TESTING?

The purpose of SMA carrier testing is to see if a couple is at an increased risk of giving birth to a child who will have SMA. SMA carrier testing is a laboratory test done on a sample of blood. If testing shows that a couple is at risk, additional testing can be done on the developing baby to determine if it has SMA.

**Carrier Testing is Available for
Spinal Muscular Atrophy**

www.preventsma.org

WHAT IS CARRIER TESTING?

Carrier testing is genetic testing to determine whether an individual carries one copy of an altered gene for a particular recessive condition. Genes are the basic units of heredity passed from parent to child. Genes occur in pairs and we inherit one member of each pair from our mother and the other from our father. A change in a gene, known as a mutation, can cause that gene not to work properly. This can lead to disease.

Certain diseases are caused when both genes of a pair have mutations. This is called a recessive disease. In order to have a child with a recessive disease, each parent has a gene that works properly and a gene that does not. The parents are known as carriers and do not have any health problems related to that gene. Genetic testing can identify carriers by a simple blood test to determine if a couple is at risk to have a child affected with a specific disease.

WHAT ARE THE BENEFITS OF CARRIER TESTING?

Carrier testing can help couples make decisions about whether and how to have children. Couples have many options. Some couples may choose not to risk having a child born with SMA and may adopt, use donated eggs or sperm or go through in vitro fertilization and test the embryos using Preimplantation Genetic Diagnosis (PGD). Other couples may choose to become pregnant and pursue the earliest available prenatal testing. Some couples may use carrier testing to learn about their risks before they become pregnant but not pursue prenatal testing.



ABOUT US

The Claire Altman Heine Foundation, Inc. is a 501(c)3 tax exempt organization and a publicly supported charity as determined by the Internal Revenue Service. The Foundation was created in memory of our daughter, Claire, whose life was claimed by Spinal Muscular Atrophy (SMA). While we wait for a cure the Claire Altman Heine Foundation's primary mission is to prevent SMA by raising awareness of the disease and implementing population based SMA carrier screening.

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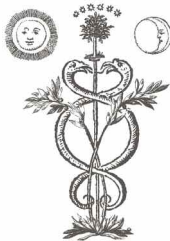
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