

# SPINAL MUSCULAR ATROPHY

*Carrier Testing:  
The Decision is Yours*



CLAIRE ALTMAN HEINE FOUNDATION, INC.  
*dedicated to identifying carriers 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 60% 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.



This brochure was prepared to give you information about Spinal Muscular Atrophy (SMA) and SMA carrier testing. Carrier testing is available to you on a voluntary basis. Whether or not you are tested is a personal decision. Before deciding, you should read this brochure so you understand what SMA is and what carrier testing is about.

If, after reading this brochure, you want to be tested, or simply want to know more about the test, you should tell your health care provider that you are interested in learning more about SMA carrier testing. You may also want to check to make certain your health insurance will cover the cost of SMA carrier testing.



## WHAT IS SMA?

Spinal Muscular Atrophy (SMA) is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement. Muscles that control breathing, swallowing, head and neck control, walking, and crawling are the most severely affected. There are four types of SMA—Types I, II, III, IV. The determination of the type of SMA is based upon the physical milestones achieved. Symptoms of the disease range from mild (symptoms that do not appear until adulthood and are not life threatening) to severe (symptoms that are present in early infancy and lead to a severely shortened lifespan). Over 60% of those who are diagnosed with SMA are severely affected. SMA does not affect intelligence. There is no cure or treatment for SMA. Research on effective treatments and cure are underway.

## WHAT CAUSES SMA?

SMA is a genetic disease. In order for a child to be affected by SMA, both parents must be carriers of the abnormal gene and both must pass this abnormal gene on to their child. If both parents are carriers the likelihood of a child inheriting the disorder is 25%, or one in four. If both parents are SMA carriers the likelihood of a child being an SMA carrier is 50%, or two in four. If both parents are SMA carriers the likelihood of their child being neither an SMA carrier nor affected with SMA is 25%, or one in four.

## WHAT IS THE PURPOSE OF SMA CARRIER TESTING?

The purpose of SMA carrier testing is to see if a couple is at increased risk for 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 high risk, additional testing can be done during pregnancy to see whether or not the baby will have SMA.

## COULD I BE A CARRIER OF SMA?

*Yes.* You could be a carrier of SMA even if no one in your family has SMA and even if you already have children without SMA. Carriers of the abnormal gene for SMA have no symptoms and are not sick. About one in every 40 people carries the gene.

If a relative of yours has SMA, or is known to be a carrier of SMA, your chance of being a carrier is greater based on your family history.

## IF MY TEST RESULT IS NORMAL COULD I STILL BE A CARRIER?

SMA carrier testing tells you if there is a greater chance that you are a carrier. A negative result significantly lowers, but does not completely eliminate the risk of being a SMA carrier. However, the likelihood that you are a carrier even though you had a normal result is very small.

## IF THE TEST SHOWS I AM A CARRIER WHAT SHOULD I DO?

If the test shows that you are a carrier, you may want to test the baby's father. Both parents must be carriers for the baby to have SMA.

If the father has a normal test result and no family history of SMA, the chance that your baby will have SMA is very, very small. Since this is a very rare occurrence, if you are a carrier but the father has a normal result, no further testing would be recommended.

## WHAT IF BOTH MY PARTNER AND I ARE SMA CARRIERS?

It is important to remember that if you and your partner have both been shown to be carriers of an abnormal SMA gene and have a child together, there is a one-in-four (25%) chance with each pregnancy that the child will have SMA. This is true even if you already have other children with or without SMA.

If SMA testing shows both parents are carriers, you might see a genetic counselor. There are several choices couples in your situation can make when thinking about possible future pregnancies. Some couples decide to:

- Have prenatal testing, such as amniocentesis or CVS;

- Accept this level of risk and have children without further testing;

- Go through in vitro fertilization and test the embryos using Preimplantation Genetic Diagnosis (PGD);

- Adopt;

- Use donor sperm or donor eggs or;

- Not have other children.

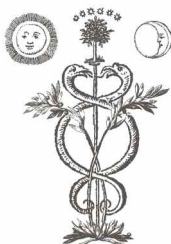
## SPINAL MUSCULAR ATROPHY RESOURCES

**Families of Spinal Muscular Atrophy**  
P.O. Box 196, Libertyville, IL 60048-0196  
Phone 800.886.1762    [www.fsma.org](http://www.fsma.org)

**National Society of Genetic Counselors**  
233 Canterbury Drive, Wallingford, PA 19086-6617  
Phone 312.321.6834    [www.nsgc.org](http://www.nsgc.org)

**Genetic Alliance**  
4301 Connecticut Avenue NW , Suite 404  
Washington, D.C. 20008-2304  
Phone 800.336.4363    [www.geneticalliance.org](http://www.geneticalliance.org)

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 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.



**CLAIRE ALTMAN HEINE FOUNDATION, INC.**  
*dedicated to identifying carriers of Spinal Muscular Atrophy*

1112 Montana Avenue . Suite 372 . Santa Monica . California . 90403  
Phone 310.260.3262    Fax 310.393.7154  
[www.clairealtmanheinefoundation.org](http://www.clairealtmanheinefoundation.org)