

SPINAL MUSCULAR ATROPHY

*What happens if
both my partner and I
are SMA carriers?*



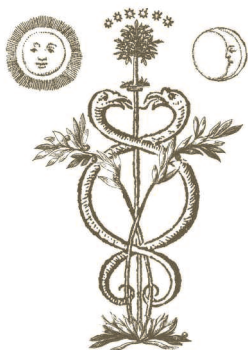
CLAIRE ALTMAN HEINE FOUNDATION, INC.
dedicated to identifying carriers of Spinal Muscular Atrophy

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.



The purpose of this brochure is to explain the meaning of your test results and what steps you can take next. You have been given this brochure because the results of your carrier test revealed that both you and your partner are carriers of the abnormal SMA gene. Along with reading this it is important and helpful for you to speak with your health care provider about the next steps you may want to take.



WHAT DO MY TEST RESULTS MEAN?

The test results mean that you and your partner are both carriers of the abnormal Spinal Muscular Atrophy (SMA) gene. This does not have any impact on your health. However, it means that you are at risk for having a baby with SMA.

It is important to remember that the result you received does *not* mean that your baby will definitely have SMA.

WHAT SHOULD I KNOW ABOUT SMA?

Spinal Muscular Atrophy (SMA) is a motor neuron disease. The motor neurons affect the voluntary muscles that are used for activities such as crawling, walking, head and neck control, and swallowing.

SMA affects muscles throughout the body, although the proximal muscles (those closest to the trunk of one's body—i.e. shoulders, hips, and back) are often most severely affected. Weakness in the legs is generally greater than in the arms. Sometimes feeding and swallowing can be affected. Involvement of respiratory muscles (muscles involved in breathing and coughing) can lead to an increased tendency for pneumonia and other lung problems.

Sensation and the ability to feel are not affected. Intellectual activity is normal. Patients are generally grouped into one of four categories, based on certain key motor function milestones and time of disease onset.

WHAT IS THE CHANCE THAT MY BABY WILL HAVE SMA?

There is a one-in-four (25%) chance that the baby will have SMA. Conversely, there is a three-in-four (75%) chance that your baby will *not* have SMA.

There is a one-in-two (50%) chance that your baby will be a carrier, like you and your partner. This will not have health consequences for the baby, but she or he could be at risk for having a child with SMA in the future.

There is also a one-in-four (25%) chance that the baby will not have the abnormal SMA gene, that is, neither has the disease nor is able to pass on the gene.



HOW CAN I FIND OUT IF MY BABY WILL HAVE SMA?

Through additional testing during pregnancy. Chorionic Villus Sampling (CVS), generally done around the 11th week of pregnancy, is done by

using a small instrument to remove a very small portion of the placenta. This portion is analyzed to determine if the baby has SMA. Alternatively amniocentesis, generally done around the 16th week of pregnancy, is performed by removing a small amount of fluid surrounding the baby using a thin needle inserted into a woman's uterus. The fluid is analyzed to determine if the baby has SMA.

If you decide not to have this prenatal testing, you can find out whether or not the baby is affected with SMA after its birth through a simple test done with a blood sample.

WHAT IF THE AMNIOCENTESIS OR CVS RESULTS SHOW THAT THE BABY DOES NOT HAVE SMA?

If the test results indicate the baby will not have SMA, you can go through the rest of your pregnancy feeling assured that the baby will not develop this condition. There is a three-in-four (75%) chance that this will be the result of your prenatal test.

ARE THE TEST RESULTS DEFINITE?

Yes. The results of these prenatal tests (that is amniocentesis or CVS) are nearly 100% accurate about whether or not the baby will have SMA. However, they usually cannot tell you how severe or mild the disease will be if the baby has SMA.

IF ALL TESTS SHOW THE BABY WILL HAVE SMA, WHAT ARE MY OPTIONS?

SMA is not a curable disease. There are no known treatments available. Therefore, there are only three decisions that are possible once it is found out that a fetus has SMA.

You can continue the pregnancy and prepare for the addition to your family of a child with SMA.

Couples can use this time to learn as much as possible about the disease by talking with care providers and the experiences of other families who have a child or children with SMA.

You may choose to terminate the pregnancy. If this is an option that you might consider, you should discuss with your doctor. This must usually be done before the end of the second trimester of pregnancy. Each state has somewhat different laws on pregnancy termination.

You may choose to continue the pregnancy and prepare to place the child up for adoption.



WHAT ARE THE HEALTH NEEDS OF CHILDREN WITH EACH TYPE OF SMA?

The four categories of SMA—Types I, II, III, IV are based upon the physical milestones achieved. It is important to note that the course of the disease may be different for each child and how severe or mild the disease will be is not usually known before birth.

Type I: Type I SMA (also called Werdnig-Hoffmann Disease) is the most severe form of SMA and the most common. Over 60% of children diagnosed with SMA are Type I.

Usually a child with Type I is never able to lift his/her head or accomplish the normal motor skills expected early on in infancy. They generally have poor head control, and may not kick their

legs as vigorously as they should, or bear weight on their legs. They do not achieve the ability to sit up unsupported. Swallowing and feeding may be difficult and are usually affected at some point, and the child may show difficulties managing his/her own secretions. The tongue may show atrophy, and rippling movements or fine tremors, also called fasciculations. There is weakness of the intercostal muscles (the muscles between the ribs) that help expand the chest, and the chest is often smaller than usual. The chest may appear concave (sunken in) due to the diaphragmatic (tummy) breathing. Due to this type of breathing, the lungs may not fully develop, the cough is very weak, and it may be difficult to take deep enough breaths while sleeping to maintain normal oxygen and carbon dioxide levels.

Type II: The Diagnosis of Type II SMA is almost always made before two years of age, with the majority of cases diagnosed by 15 months. Children with this type may sit unsupported when placed in a seated position, although they are often unable to come to a sitting position without assistance. At some point they may be able to stand. This is accomplished with the aid of assistance, bracing or standing frame. Swallowing problems are not usually characteristic of Type II, but vary from child to child. Some patients may have difficulty eating enough food by mouth to maintain their weight and grow, and a feeding tube may become necessary. Children with Type II SMA frequently have tongue fasciculations and manifest a fine tremor in the



outstretched fingers. Children with Type II also have weak intercostal muscles and are diaphragmatic breathers. They have difficulty coughing and may have difficulty taking deep enough breaths while they sleep to maintain normal oxygen levels and carbon dioxide levels. Scoliosis is almost uniformly present as these children grow, resulting in need for spinal surgery or bracing at some point in their clinical course. Decreased bone density can result in an increased susceptibility to fractures.

Type III: The diagnosis of Type III, often referred to as Kugelberg-Welander or Juvenile Spinal Muscular Atrophy, is much more variable in age of onset, and children can present from around one year of age or even as late as adolescence, although diagnosis prior to age three is typical. The patient with Type III can stand alone and walk, but may show difficulty with walking at some point in their clinical course. Early motor milestones are often normal. However, once they begin walking, they may fall more frequently, have difficulty in getting up from sitting on the floor or a bent over position, and may be unable to run. With Type III, a fine tremor can be seen in the outstretched fingers but tongue fasciculations are seldom seen. Feeding or swallowing difficulties in childhood are very uncommon. Type III individuals can sometimes lose the ability to walk later in childhood, adolescence, or even adulthood, often in association with growth spurts or illness.

Type IV (Adult Onset): In the adult form, symptoms typically begin after age 35. It is rare for Spinal Muscular Atrophy to begin between the ages of 18 and 30. Adult onset SMA is much less common than the other forms. It is defined as onset of weakness after 18 years of age, and most cases reported as Type IV have occurred after age 35. It is typically characterized by insidious onset and very slow progression. The bulbar muscles, those muscles used for

swallowing and respiratory function, are rarely affected in Type IV.

DO ALL PEOPLE WITH SMA HAVE THE SAME SYMPTOMS?

No. As described previously, some individuals have far milder symptoms than others. It is not always possible to tell from a prenatal test how mild or severe a child's symptoms will be. While in general, people with SMA have a severely shortened life span, many die in childhood, others live into their thirties or even longer. Although there is no cure for SMA, effective treatments are being researched.



MIGHT OTHERS IN MY FAMILY BE SMA CARRIERS?

Yes. Even if no one in your family has had SMA, other close relatives, such as brothers, sisters, aunts, uncles and cousins, may also be carriers. This information might be useful for them to know if they are planning pregnancies. This is something you might want to discuss further with your care provider.

WHAT ABOUT FUTURE PREGNANCIES?

It is important to remember that you and your partner have both been shown to be carriers of an abnormal SMA gene. This means that in each pregnancy the two of you have together, that baby will also have a one-in-four (25%) chance of having SMA. If you want to know whether or not that baby will develop SMA, you need to have amniocentesis or CVS in each pregnancy.

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.

Carrier Testing is Available for
Spinal Muscular Atrophy

www.clairealtmanheinefoundation.org

SPINAL MUSCULAR ATROPHY RESOURCES

Families of Spinal Muscular Atrophy
P.O. Box 196, Libertyville, IL 60048-0196
Phone 800.886.1762 www.fsma.org

National Society of Genetic Counselors
233 Canterbury Drive, Wallingford, PA 19086-6617
Phone 312.321.6834 www.nsgc.org

Genetic Alliance
4301 Connecticut Avenue NW , Suite 404
Washington, D.C. 20008-2304
Phone 800.336.4363 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.



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