




SMA Carrier Testing is Easy.
And important.



LEARN MORE:
www.athenaSMAawareness.com



It's your future. SMA carrier testing puts you in control, helps you prepare, and gives you more options.



Deborah and Chris Heine did everything they could to ensure that their second child would be born as healthy as their first.

In addition to living a wholesome lifestyle, the California couple even got tested for genetic diseases like cystic fibrosis and Tay-Sachs disease before they conceived. In January 2004, the Heines joyfully welcomed Claire, a beautiful baby girl, into their lives.

Their joy was short-lived. When Claire was just nine weeks old, the couple received the heartbreaking news that their daughter had spinal muscular atrophy (SMA). Babies, like Claire, who have the most common form of SMA usually die from respiratory failure before they reach the age of two. Claire died when she was just nine months old.

What makes the Heines' experience especially tragic is that it might have been avoided. For years, there has been a blood test that will tell couples who are planning a pregnancy or are expecting whether they carry a genetic abnormality, or mutation, that causes SMA.

“Had we known how common this disease is, and that there was a test to screen for people who carry it, we certainly would have requested it.” — *Deborah Heine*

Athena Diagnostics sincerely thanks Deborah Heine for permission to share her story in this patient education brochure.



What is SMA?

- SMA (spinal muscular atrophy) is a severe, often fatal, genetic disorder in which muscles involved in many essential functions, such as breathing, eating, and movement, become progressively weaker and ultimately waste away (atrophy) and die.
- About one in every 40 people carries a mutated SMN1 gene and every year, about one in every 6,000 – 10,000 babies is born with SMA.
- There are three types of SMA. The most common form, type I, which affects about 70% of patients, is the most severe. Children with type 1 SMA usually die from respiratory failure before the age of two. In fact, SMA is the leading genetic cause of death in early childhood.
- Children with type II SMA may be able to sit unaided, but cannot stand or walk unaided. These children typically live past age four. Although they face many challenges, children with type III SMA are able to walk unaided and have a normal lifespan.
- A child can only have the disease if both parents carry the mutated SMN1 gene.
- When both partners are carriers, there is a 25% (one in four) chance with each pregnancy of having a child with SMA. (Of course, this means that there is a 75% chance that each pregnancy will not result in a child with SMA.)
- Fortunately for individuals planning a family, a simple blood test performed by Athena can determine with a very high degree of certainty whether you or your partner carries the mutation responsible for SMA.
- It is imperative that couples have access to genetic counseling and patient education materials. SMA carrier testing should be both voluntary and confidential.



SMA Carrier Testing Q&A

- Q:** If my doctor doesn't suggest SMA Carrier testing, does it mean I don't need to be tested?
- A:** While some doctors offer it to their patients already, you may have to ask your doctor about SMA Carrier testing as they may be unaware of SMA.
- Q:** My husband and I would never consider terminating a pregnancy. Why should we or couples who share our views consider SMA carrier testing?
- A:** While opposition to termination of a pregnancy is sometimes cited in arguments against genetic testing, it is important to realize that termination of a pregnancy is never the only option.
- Q:** Should I have SMA Carrier testing even if my other children are not affected?
- A:** This is a personal decision that only you can answer and that may be influenced by several factors such as insurance, familiarity with the disease, personal opinions and beliefs regarding termination or willingness to pursue other options if the test result is positive.
- Q:** Is SMA Carrier testing harmful to me or my child if I'm pregnant while getting tested?
- A:** No, it is a blood test on the mother and would have no effect on the child.

Because SMA is present in all populations it is now recommended by the American College of Medical Genetics that all couples, regardless of their race or ethnicity, be offered carrier testing for SMA.

Doctor's Discussion Guide

It is important to discuss SMA carrier testing with your doctor. To facilitate your conversation, here are some discussion points that may reflect your situation. It's your future. SMA carrier testing puts you in control, helps you prepare, and gives you more options.

I already have a normal, healthy child or children

You and your spouse may still be SMA carriers even if you already have a healthy child or children. If you and your spouse are both SMA carriers, there is a one in four chance with each pregnancy that you will have a baby with SMA.

I don't have a family history of SMA

Knowing that you have a family history of SMA (either a relative with SMA or a family member who is an SMA carrier) puts you at greater risk of being an SMA carrier. But even if you don't have a family history of SMA, you can still be an SMA carrier and pass along the abnormal gene that causes the disease to your baby.

I'm under 40 years of age

Unlike a genetic disease such as Down syndrome, age has nothing to do with whether you could have a baby with SMA. Couples of all ages can be SMA carriers and have a baby with SMA.

Carrier testing won't change the outcome of an affected baby

Knowing in advance that your baby has SMA can make a world of difference to you and your baby's quality of life. It gives you time to prepare emotionally and financially, learn as much as you can about the disease, make arrangements for healthcare services that can improve your baby's quality of life, and make important decisions that will maximize your family's time together.

I'm not sure whether my insurance covers SMA testing

Insurance plans differ in their coverage, so you should check with your insurance provider to find out if and to what extent they cover SMA carrier testing.

Similarities between CF and SMA

	CYSTIC FIBROSIS	SMA
Inheritance	Recessive	Recessive
Carrier frequency	1 in 27	1 in 34
Ethnicities	All	All
Prevalence in population	1 in 4,000	1 in 6,000 - 10,000
Impact on lifespan	Death at 38.5 yrs. age	Death <2 yrs. age*
Carrier test available	Yes	Yes

**With Type 1 SMA (70% of SMA children)*

Cystic Fibrosis and SMA share many similarities. CF is routinely tested for yet SMA is not. Athena Diagnostics can test for both diseases with a simple blood test. Why not test for both?

Additional Resources

Whether you want to learn more about the benefits of SMA carrier testing, have just found out that you and your partner are carriers, or are expecting or have a child with SMA, there are many resources available to help you.

athenaSMAawareness.com

This site was created to raise awareness of SMA and provide help to couples seeking reliable information, support, and access to a community of others affected by or focused on SMA.

www.preventsma.org

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.

www.nsgc.org

The National Society of Genetic Counselors promotes the genetic counselor profession as a recognized part of health care delivery, education, research, and public policy.

www.acmg.net

The American College of Medical Genetics provides education, resources, and a voice for the medical genetics profession. To make genetic services available to and improve the health of the public, the ACMG promotes the development and implementation of methods to diagnose, treat, and prevent genetic diseases.

www.fightsma.org

FightSMA is an international nonprofit organization dedicated to finding a treatment or cure for SMA by raising awareness of and funding for SMA research.

www.fsma.org

Families of Spinal Muscular Atrophy is dedicated to creating a treatment and cure for SMA by funding and advancing a comprehensive research program; supporting SMA families through networking, information, and services; improving care for all SMA patients; educating health professionals and the public about SMA; enlisting government support for SMA; and embracing all touched by SMA in a caring community.

www.smafoundation.org

The mission of the Spinal Muscular Atrophy Foundation is to accelerate the development of a treatment for SMA, the number one genetic killer of infants and toddlers. To help speed the realization of a treatment or cure, the SMA Foundation is committed to developing private sector involvement, increasing government funding, and raising awareness among government leaders and industry.





Testing that Makes a Difference.

Athena Diagnostics is a state-of-the-art laboratory dedicated to providing specialty diagnostic testing services that span neurology, endocrinology, nephrology and reproductive healthcare. Using innovative technologies, Athena provides physicians with diagnostic answers that help patients make informed decisions and improve the quality of health care.

www.athenaSMAawareness.com

Professionals please visit www.athenadiagnostics.com for test requirements.