

Inheritest[®]

CARRIER SCREEN

An important step in
family planning



Inheritest[®] CF/SMA Panel – carrier screening
for common genetic disorders

CARRIER SCREENING FOR COMMON GENETIC DISORDERS

This brochure provides information to help you decide about having carrier screening for two common genetic disorders. If you have any additional questions please talk to your doctor or to a genetics professional.

There are two relatively common disorders which can be seen in families even when there are no known affected family members. Screening for these two disorders is available with a simple blood test, and can determine if you are a carrier for any of these disorders.

What are these common genetic disorders?

The chart on the inside provides a summary of information about cystic fibrosis (CF) and spinal muscular atrophy (SMA), and carrier screening for these disorders.

American College of Obstetricians and Gynecologists (ACOG) recommendation states that carrier screening for SMA and CF "should be offered to all women who are considering pregnancy or are currently pregnant."¹

Could you be a carrier of a genetic disorder?

You could be a carrier of a genetic disorder even if no one in your family is affected and even if you already have healthy children. Everyone has a chance to be a carrier, and, in general, carriers have no symptoms of the disorder.

If someone in your family or your partner's family has one of these disorders, or has been found to be a carrier, your risk to be a carrier is greater than someone without a family history. In these cases you should discuss the family history with your doctor or a genetics professional.



WHAT IS CARRIER SCREENING?

Carrier screening involves a blood test from one or both parents. The screening can be done either when you are planning a pregnancy or after you have become pregnant. All screening is optional and you can choose which tests are right for you.

If your test is negative, could you still be a carrier?

A negative test result significantly lowers, but does not completely eliminate the risk of being a carrier. Carrier screening is not able to detect all the genetic abnormalities that cause a particular disorder.

What if the test shows you are a carrier of CF or SMA?

If the test shows that you are a carrier of CF or SMA, the next step is for your partner to have carrier screening performed. Both parents must be carriers for the baby to be at risk for CF or SMA. If your partner has a negative test result and no family history of CF or SMA, the chance that your baby will have CF or SMA is less than 1%.

If both parents are shown to be carriers the next step is for you to consider prenatal testing by amniocentesis or chorionic villi sampling (CVS) to determine if your baby is affected.

CYSTIC FIBROSIS (CF)

Symptoms of disorder

The most common inherited disorder of children and young adults.

CF primarily involves the respiratory, digestive and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected, but individuals with severe symptoms may die in childhood. With treatments today, people with CF can live on average 40 years. CF does not affect intelligence. Multiple therapies are currently available for CF.²

Inheritance

If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.

General population carrier frequency

As high as 1 in 24³
Varies by ethnicity

SPINAL MUSCULAR ATROPHY (SMA)

Symptoms of disorder

The most common inherited cause of early childhood death.

SMA destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling or walking. The most common form of SMA affects infants in the first months of life and can cause death between 2-4 years of age. Less commonly the disorder starts later and people can survive into adulthood. SMA does not affect intelligence. There is no cure for SMA, but there is a new treatment available which may delay symptoms or reduce their severity.⁴

Inheritance

If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.

General population carrier frequency

As high as 1 in 47⁵
Varies by ethnicity



MODEL INFORMED CONSENT/DECLINE FOR CARRIER TESTING

You should be certain you understand the following points

- The purpose of these tests is to determine whether I am a carrier of one of the common genetic abnormalities that cause CF and/or SMA
- The tests do not detect all carriers of these disorders
- The laboratory needs accurate information about my family history for the most accurate interpretation of the test results
- The decision to have carrier testing is completely mine
- No other test will be performed and reported on my sample unless authorized by my doctor, and any unused portion of my original sample will be destroyed within two months of receipt of the sample by the laboratory
- The laboratory will disclose the test results ONLY to my doctor, or to his/her agent, unless otherwise authorized by me or required by law

For CF

- If I am a carrier, testing my partner will help me learn more about the chance that our baby could have CF
- If one parent is a carrier and the other is not, it is still possible that the baby will have CF, but the chance is less than 1%
- If both parents are carriers, prenatal testing is available to find out whether or not the baby will have CF

For SMA

- If I am a carrier, testing my partner will help me learn more about the chance that our baby could have SMA
- If one parent is a carrier and the other is not, it is still possible that the baby will have SMA, but the chance is less than 1%
- If both parents are carriers, prenatal testing is available to find out whether or not the baby will have SMA

I have read, or had read to me, the information in this brochure and I understand it. Before signing this form, I have had the opportunity to discuss carrier screening further with my doctor, someone my doctor has designated, or with a genetics professional. I have all the information I want, and all my questions have been answered. I have decided that:

- I want CF carrier screening
 I do not want CF carrier screening

Patient signature: _____

Date: _____

- I want SMA carrier screening
 I do not want SMA carrier screening

Patient signature: _____

Date: _____



Cystic fibrosis
Spinal muscular atrophy

Pioneering science, personalized service



Understand your cost options

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Convenient blood draws

Getting your blood drawn is easier than ever. As a LabCorp company, we have a nationwide network of patient service centers, allowing for convenient access to sample collection. Visit www.LabCorp.com to find your nearest location.



Genetic counseling

Patients with a positive test result may be offered counseling, and Integrated Genetics offers the largest national commercial network of genetic counselors to help inform and support patients.



Every Mom Pledge

We believe every mom should have access to the best possible care. That's why we work directly with you to make sure our testing services are accessible and out-of-pocket costs are transparent.

We are ready to help!

www.integratedgenetics.com

General questions?

Call toll-free within the US

800.848.4436



Billing and cost questions?

Call toll-free within the US

844.799.3243

Every Mom Pledge team is ready to answer questions about your insurance coverage and cost options
integratedgenetics.com/transparency



Watch a short video to learn about the test:
integratedgenetics.com/videos

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