PATIENT INFORMATION

Inheritest® Core Panel

Prepare today for your family's tomorrow

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Carrier Screening for common genetic disorders

The Inheritest® Core Panel can help determine if you are at increased risk for having a baby with one of three common genetic disorders. Early insight can help you better prepare for your family's future.

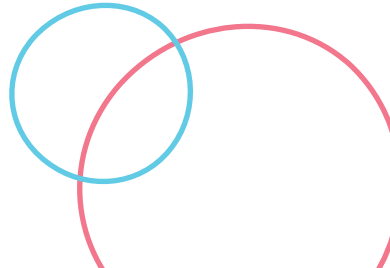
Inheritest Core Panel tests for three relatively common disorders which can be seen in families even when there are no known affected family members. Screening for these three disorders is available with a blood or saliva test, and the results are delivered to your doctor in approximately two to three weeks.

What are these common genetic disorders?

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

What is carrier screening?

Carrier screening is a genetic test that can help you learn if you (and if indicated, your partner) are a carrier of a genetic disorder. Examples of genetic disorders include CF and SMA, among others.



Anyone can carry a genetic disorder

You could be a carrier of a genetic disorder regardless of your ethnicity and even when there is no family history of the disorder. More than 80% of infants with CF are born to families with no prior family history.[1]

Being a carrier does not typically cause symptoms for that person, but it can mean that their future children are at risk of inheriting the disorder.

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

A negative result reduces, but does not completely eliminate, the risk of being a carrier of the genetic disorders included in a carrier screening panel. Screening cannot test for all mutations or all disorders.

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

If the test shows you are a carrier of CF or SMA, the next step is for your partner to have carrier screening performed. The chance of CF or SMA can then be assessed using both results.

If the test shows that you are a carrier of fragile X, your partner does not need testing. The chance of fragile X is calculated using only the biological mother's results.

Your doctor and our genetic counselors can help you better understand your test results and help you prepare for the future.

Disorder	Cystic Fibrosis (CF)	Spinal Muscular Atrophy (SMA)	Fragile X Syndrome
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 male infertility. Symptoms of the condition range from mild to severe, with an average life expectancy of 46 years.[2] Multiple therapies are currently available. CF does not affect intelligence.	**The most common inherited cause of early childhood death.** SMA affects nerve cells and impacts 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 in childhood and people can survive into adulthood. SMA does not affect intelligence. There is no cure for SMA, but there are several different treatments available which may delay symptoms or reduce their severity.[4]	**The most common inherited cause of intellectual disability.** Fragile X syndrome involves developmental delay, intellectual disability, autism and hyperactivity. It affects both boys and girls, although boys are usually more severely affected than girls. When the biological mother is a carrier for fragile X, a child is at risk to have intellectual disability.
Inheritance	If both biological parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.	If both biological parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.	If a biological mother is a carrier, there is up to a 50% chance to have a child with fragile X syndrome.
General population carrier frequency	As high as 1 in 24 (varies by ethnicity)[3]	As high as 1 in 47 (varies by ethnicity)[5]	~1 in 259 people assigned female at birth (all ethnicities)[6]

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, SMA and/or fragile X syndrome
- 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 me and my doctor, or to their 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 future offspring could have CF
- If one biological parent is a carrier and the other is not, it is still possible that future offspring will have CF, but the chance is less than 1%
- If both biological parents are carriers, prenatal testing is available to find out whether or not future offspring will have CF

For SMA

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

For Fragile X

- If I am a carrier, prenatal testing is available to find out whether or not future offspring will have fragile X syndrome

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: _____

☐ **I want fragile X carrier screening**
☐ **I do not want fragile X carrier screening**

Patient signature: _____

Date: _____

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