PATIENT EDUCATION

carrier screening INFORMATION

GoodStart® Genetics

Helping to give the next generation a good start.℠

carrier screening AT A GLANCE

Why is carrier screening recommended?

Carrier screening is one of many tests that can help provide information to you and your doctor about your reproductive risks prior to or during pregnancy. And while most babies are born healthy, with each pregnancy, there is a small chance of having a baby with a severe genetic disorder. Carrier screening helps assess what the chance is for you.

How is carrier screening done?

The first step is to determine which disorders you should be screened for based on your ethnic background and family history. Then your doctor will draw one or two tubes of blood to send to the laboratory for testing. It typically takes two weeks for your doctor to get your results. While testing can never eliminate the risk of being a carrier, it does provide the opportunity to explore reproductive options if both partners are carriers for the same disorder.

Is carrier screening right for me?

Carrier screening can provide you with information that might be helpful if you:

- Are planning a pregnancy
- Have a family history of a genetic disorder
- Are at increased risk for a specific condition based on ethnicity
- Would like additional information about your reproductive risks

Discussing the benefits and limitations of carrier screening with a qualified health care professional, like a genetic counselor, may help you decide if carrier screening is right for you.

What is a carrier?

Being a carrier means that one of two copies of a gene has a change, called a mutation, that causes that copy of the gene to stop working properly. Carriers typically do not have any health problems associated with carrying a mutation and often do not know they are carriers until this screening is performed; however, as a carrier, there is a chance that your children could inherit that genetic disorder.

Could I be a carrier?

Anyone can be a carrier of a genetic disorder, even if no one in your family is affected. The chance of being a carrier is based on your ethnic background and family history; however, some disorders are fairly common regardless of these factors.

GoodStart Select™

The right choice for carrier screening.

Why GoodStart Select?

The information provided through carrier screening is often used to help guide reproductive decisions, so it is important that the information you and your doctor receive from these tests is not only accurate, but comprehensive as well.

Good Start Genetics is a company specializing in carrier screening. **GoodStart Select** is our customizable test menu, which includes tests clinicians routinely order for common genetic disorders.

Your doctor may have recommended **GoodStart Select** because Good Start Genetics uses a powerful technology called next-generation sequencing. By using this advanced technology, **GoodStart Select** detects more mutations than other routine carrier screens. This means a higher detection rate and more confidence in your carrier screening test results.

At Good Start Genetics, we believe everyone should have access to these important tests. Good Start Genetics has dedicated customer care and billing specialists who will work directly with you to help reduce out-of-pocket expenses for carrier screening, and in fact, this type of testing is frequently covered by insurance. Genetic counseling is also available upon request.

By using GoodStart Select, you benefit from results you can trust and dedicated customer care throughout your carrier screening experience.

For more information about carrier screening, genetic disorders tested and our customer care programs or to learn about Good Start Genetics, please visit **www.goodstartgenetics.com** or email us at **solutions@gsgenetics.com**.

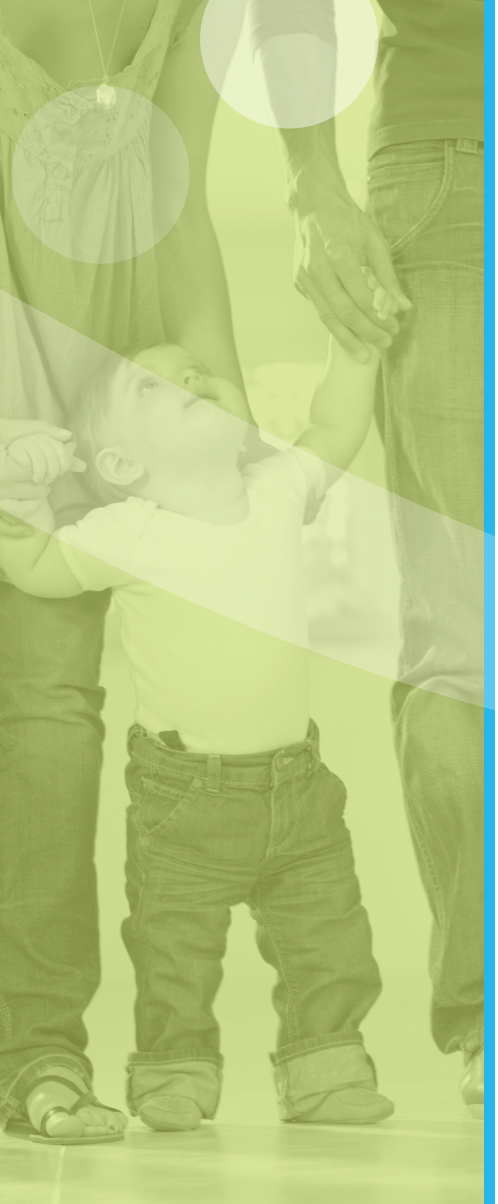
pan ethnic carrier screening

FOR COMMON DISORDERS

Some genetic disorders are relatively common and carrier screening may be offered to you, regardless of your ethnic background. These disorders are **cystic fibrosis (CF), spinal muscular atrophy (SMA)**, and **fragile X syndrome**. A brief description of each of these disorders is in the table below.

DISORDER	CARRIER FREQUENCY	SYMPTOMS
Cystic Fibrosis (CF)	Ashkenazi Jewish: 1 in 23 Caucasian: 1 in 25 Hispanic: 1 in 58 African American: 1 in 61 Asian: 1 in 94	CF is characterized by chronic respiratory and digestive problems. Symptoms and severity of CF range from mild to severe. Intelligence and appearance are not affected. With treatment today, people with severe CF can live into their 30's.
Spinal Muscular Atrophy (SMA)	Caucasian: 1 in 47 Asian: 1 in 59 Ashkenazi Jewish: 1 in 67 Hispanic: 1 in 68 African American: 1 in 72	SMA is characterized by severe muscle weakness and progressive loss of voluntary muscle control. Symptoms often begin in early infancy and include difficulties breathing, swallowing, and crawling. Intelligence and appearance are not affected. In severe cases, SMA results in death before two years of age.
Fragile X Syndrome	All Ethnicities: 1 in 178 women	Fragile X syndrome is the most common cause of inherited intellectual disability and is characterized by developmental delay, autism, hyperactivity, and some characteristic physical features.

Source: Data on File



ashkenazi jewish
CARRIER SCREENING

Individuals of Ashkenazi Jewish descent typically have ancestors from Eastern Europe and there are a number of genetic disorders that have an increased frequency in this population.

These disorders can range in severity and symptoms; however, all of these disorders can have a serious impact on the affected individual's quality of life and often results in an early death. At this time, there are limited or no treatments available for most of these disorders.

While these disorders are most common in individuals of Ashkenazi Jewish descent, they may occur in people of other ethnic backgrounds.

A list of these disorders is provided here, along with the carrier frequency in individuals of Ashkenazi Jewish descent.

DISORDER	CARRIER FREQUENCY
Bloom's Syndrome	1 in 134
Canavan Disease	1 in 55
Cystic Fibrosis (CF)	1 in 23
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	1 in 107
Familial Dysautonomia	1 in 31
Familial Hyperinsulinism	1 in 68
Fanconi Anemia Group C	1 in 100
Gaucher Disease	1 in 15
Glycogen Storage Disease Type Ia	1 in 64
Joubert Syndrome 2	1 in 92
Maple Syrup Urine Disease Type A/B	1 in 97
Mucolipidosis Type IV	1 in 89
Nemaline Myopathy	1 in 168
Niemann-Pick Disease Type A/B	1 in 115
Tay-Sachs Disease*	1 in 27
Usher Syndrome Type IF	1 in 147
Usher Syndrome Type III	1 in 120
Walker-Warburg Syndrome	1 in 150

*Screening for Tay-Sachs Disease is also recommended for individuals of Cajun or French Canadian descent.

Source: Data on File

CARRIER SCREENING FOR hemoglobinopathies

Hemoglobinopathies are a diverse group of disorders characterized by abnormal or decreased production of hemoglobin, a component in blood that carries oxygen throughout your body. The severity of these conditions ranges from mild to severe, depending on the type of hemoglobin defect. The most common hemoglobinopathies (alpha-thalassemia, beta-thalassemia, and sickle cell disease) and the ethnicities in which they are most frequent are outlined in the table below.

ETHNICITY	ALPHA-THAL	BETA-THAL	SICKLE CELL DISEASE
African American	●	●	●
Asian	●	●	rare
Hispanic	●	●	●
Non-Hispanic Caribbean	●	●	●
Mediterranean	●	●	●
Middle Eastern	●	●	●
Southeast Asian	●	●	rare

● = increased carrier frequency

How is carrier screening for hemoglobinopathies done?

Two routine blood tests called a complete blood count (CBC) and a hemoglobin electrophoresis are the first steps to screening for most hemoglobinopathies. Based on these results, further testing may be needed to find the specific mutations involved.

TEST RESULTS AND

reproductive options

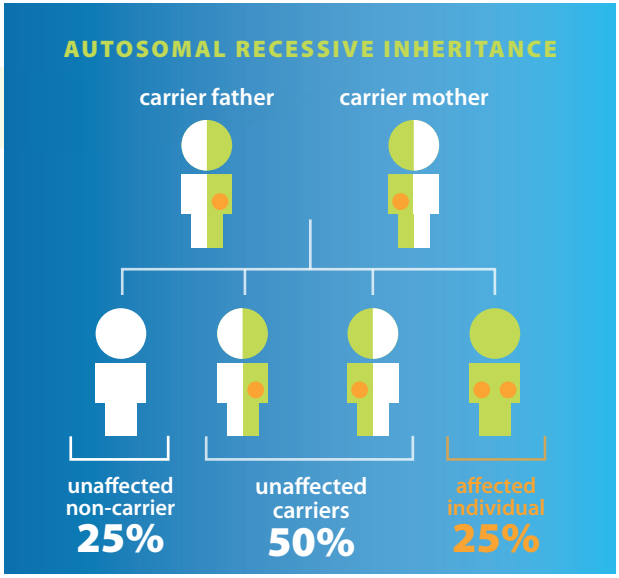
It typically takes one to two weeks to get your carrier screening test results.

What does a negative test mean?

A negative test means that no mutation was found for the disorder screened. This significantly reduces the chance that you are a carrier of this disorder. However, no carrier screen can detect all possible mutations that could cause disease, so there is always a small chance of being a carrier, even after a negative test; this chance is called **residual risk**.

What does a positive test mean?

A positive test means that a mutation was found for the disorder screened and there is an increased chance for you to have a child with this disorder. Even though you are a carrier, you typically will not have any symptoms associated with the disorder.



The next step is to test your reproductive partner. Most likely, they will not be a carrier of the same disorder, in which case the chance to have a child with this disorder is reduced. However, if both reproductive partners are carriers of the same disorder, there is a 1 in 4 (or 25%) chance to have a child with that disorder (see diagram of autosomal recessive inheritance).

Because the genetics of fragile X syndrome and alpha thalassemia are complex, carriers of these disorders have different reproductive risks than discussed here. If you are a carrier of either of these disorders, talk to your physician and/or a genetic counselor about the next steps for testing and chances to have an affected child.

What if my partner and I are both carriers for the same genetic disorder?

There are several reproductive options available for you and your partner.

- Continue with family planning and pregnancy without prenatal testing
- Prenatal testing by chorionic villus sampling (CVS) at 10-12 weeks of pregnancy or amniocentesis at 15-18 weeks of pregnancy
- In vitro fertilization (IVF) and preimplantation genetic diagnosis (PGD) prior to pregnancy
- Egg or sperm donation; screening the donor for the specific high-risk disorder is recommended
- Adoption is also available to couples who wish to have a child

GoodStartSelect™

The right choice for carrier screening.

DISORDER	RECOMMENDATIONS		
Pan-ethnic Tests	ACOG*	ACMG*	AJ*
Cystic Fibrosis (CF)	■	■	■
Spinal Muscular Atrophy (SMA)		■	■
Fragile X Syndrome**	■	■	
Ethnicity-specific Tests			
Alpha-Thalassemia	■		
Beta-Thalassemia	■		
Sickle Cell Disease	■		
Ashkenazi Jewish Tests			
Bloom's Syndrome		■	■
Canavan Disease	■	■	■
Familial Dysautonomia	■	■	■
Fanconi Anemia Group C		■	■
Gaucher Disease		■	■
Mucolipidosis Type IV		■	■
Niemann-Pick Disease Type A/B		■	■
Tay-Sachs Disease***	■	■	■
Dihydrolipoamide Dehydrogenase Deficiency			■
Familial Hyperinsulinism			■
Glycogen Storage Disease Type Ia			■
Joubert Syndrome 2			■
Maple Syrup Urine Disease Type A/B			■
Nemaline Myopathy			■
Usher Syndrome Type IF			■
Usher Syndrome Type III			■
Walker-Warburg Syndrome			■

*ACOG - American Congress of Obstetricians and Gynecologists; ACMG - American College of Medical Genetics & Genomics; AJ - Ashkenazi Jewish Panel Recommended by national Jewish advocacy societies

**Fragile X testing is recommended if indicated by a family history of fragile X syndrome, unexplained intellectual disability/autism, or premature ovarian failure.

*** Tay-Sachs Disease testing is also recommended for individuals of Cajun or French Canadian descent.

Carrier screening may also be appropriate for the partner of a known carrier or anyone with a family history of a genetic disorder.

GoodStart® Genetics

237 Putnam Avenue
Cambridge, MA 02139
855-765-0845
www.goodstartgenetics.com

Good Start Genetics Customer Care
855-765-0845 | solutions@gsgenetics.com