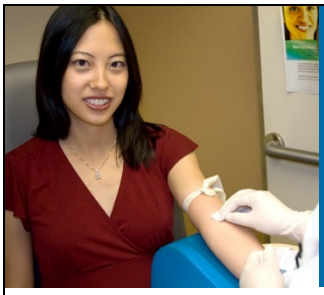

Genetic Carrier Screening

A test to see if you are a carrier for certain genetic diseases

Some genetic diseases happen when both parents silently carry a genetic change in the same gene and pass it to their child. Carrier screening checks to see if you carry one of these silent genetic changes.

What is a carrier?

A carrier is a healthy person who has a change in a gene that can cause genetic disease. A carrier does not have the genetic disease. You can be a carrier for a disease that has never happened in your family. You can also be a carrier for more than one disease. If two people are carriers for the same disease, their future children could inherit that condition.

Here's how it works: Most genes come in pairs; one inherited from each parent. The conditions included in carrier screening are called "recessive" diseases. A recessive disease happens when both copies of a gene are not working correctly. The chance for two carriers to have a child with a recessive disease is 1 in 4 (25%).

Which genetic diseases are included in carrier screening?

You will be offered carrier screening for the following conditions:

- Cystic fibrosis
- Spinal muscular atrophy
- Inherited anemias, like sickle cell and thalassemia
- Jewish carrier screening (when both parents have Jewish ancestry)
  Testing looks for 16 genetic diseases, such as Tay-Sachs. These diseases are very rare in people without Jewish (Ashkenazi) ancestry.

Testing does not screen for all genetic diseases. Many are so rare that they are not part of routine tests.

How is carrier screening done?

Genetic carrier screening is done by a blood test. Testing can be done in early pregnancy or before you get pregnant. Results take about 2 weeks.

TESTING IS OPTIONAL. You can decide if you want to have genetic carrier screening.
Learning about genetic risks during pregnancy helps some parents prepare for delivery and newborn care. It also lets parents consider testing the baby before birth to help make choices about how to manage the pregnancy.

How accurate is carrier screening?

Carrier screening will accurately identify most carriers. However, a small number of carriers will be missed by the screening test. The accuracy of the test partly depends on your ancestry or ethnic background.

Important to know: Genetic carrier screening only looks at select conditions. If you have concerns about specific genetic diseases in your family, talk with a genetic counselor. Other genetic testing may be needed based on your family history.

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What happens if you are a carrier?

If genetic carrier screening shows that you are a carrier, testing will be offered to your reproductive partner. A pregnancy is at risk for a **recessive** disease only when **BOTH parents are carriers** for the same disease. If your partner is not available for testing, you can talk with a genetic counselor about testing options.

Disease	Symptoms and Treatments	Ancestry	Carrier rate
Cystic fibrosis (CF)	Disease of the lungs and digestive system beginning in infancy. Thick mucus clogs the lungs, causing difficulty breathing and frequent lung infections. Lung disease worsens over time. Digestion problems cause slow growth. Average life span is 41 years. Treatment may include daily chest physical therapy, medications, and frequent hospital stays. Severity varies. Some live without serious illness. Currently cannot be cured.	African (Black) Ashkenazi Jewish Asian Caucasian Hispanic	1 in 61 1 in 24 1 in 94 1 in 25 1 in 58
Spinal muscular atrophy (SMA)	Disease of the nerves and muscles that usually starts in infancy or early childhood. Muscle weakness gets worse over time causing difficulty breathing, swallowing, and walking. Symptoms can start from before birth to adulthood. Severity is highly variable. The most common form of the disease starts in infancy and leads to death in early childhood. Medications can be used to treat SMA to a limited extent. Currently cannot be cured.	African (Black) Ashkenazi Jewish Asian Caucasian Hispanic	1 in 72 1 in 67 1 in 59 1 in 47 1 in 68
Hemoglobin E-beta thalassemia	Blood disease that causes moderate to severe anemia. Some live without any need for treatment. Others need lifelong blood transfusions. Generally, cannot be cured. **More common in people with Southeast Asian ancestry.**	Southeast Asian	As high as 1 in 2 is some areas
Sickle cell diseases	Blood diseases beginning in infancy or early childhood that cause anemia, bone pain, and frequent serious infections. Life span may be shortened. Treatment may include frequent hospital stays, medications, and blood transfusions. Severity varies. Some live without serious illness. Generally, cannot be cured. **More common in people with African (Black) ancestry.**	African (Black)	1 in 12
Ashkenazi Jewish diseases .	Genetic diseases that cause a wide range of health problems that can start in infancy or later. Many can be treated. Some have a shortened lifespan. Some are fatal in early childhood. **More common in people with Jewish (Ashkenazi/Eastern European) ancestry.** **16 diseases screened:** ABCC8-related hyperinsulinism, Bloom syndrome, Canavan disease, familial dysautonomia, Fanconi anemia group C, Gaucher disease, glycogen storage disease 1A, Joubert syndrome type 2, lipoamide dehydrogenase deficiency, maple syrup urine disease type 1B, mucolipidosis type IV, NEB-related nemaline myopathy, Niemann-Pick disease type A, Tay-Sachs disease, Usher syndrome type 1F and type 3.	Ashkenazi Jewish	1 in 15 to 1 in 143 Carrier rate depends on the disease

For additional information, contact your local Genetics Department:

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Modesto: 877-456-0004 Sacramento: 916-614-4075 San Jose: 408-972-3300