

Carrier Screening for Hemoglobinopathies: Sickle Cell Disease and Thalassemia

Frequently Asked Questions

What is carrier screening?

Carrier screening is a type of test that can tell you whether you carry a **gene** for certain **genetic disorders**. When screening is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder.

Who should have carrier screening?

Anyone who is pregnant or thinking about getting pregnant should be offered carrier screening. This includes carrier screening for **hemoglobinopathies**. You can choose to have carrier screening, or you can choose not to. There is no right or wrong choice.

What are hemoglobinopathies?

Hemoglobinopathies are genetic disorders that affect red blood cells. Healthy red blood cells have a doughnut shape and carry oxygen to all parts of the body. The protein that lets red blood cells carry **oxygen** is called hemoglobin.

People with a hemoglobinopathy may have low levels of oxygen. This is because their red blood cells may be a different shape or there may be a shortage of red blood cells.

Either problem can make it harder for oxygen to travel through the body. [Sickle cell disease](#) and [thalassemia](#) are two different types of hemoglobinopathies.

What is sickle cell disease?

Sickle cell disease is a common genetic disorder. It causes red blood cells to have abnormal hemoglobin and a crescent or “sickle” shape, rather than the normal doughnut shape. These cells are called sickle cells. They can get caught in blood vessels and stop oxygen from getting to other parts of the body.

Sickle cell disease may cause pain, infection, stroke, and [anemia](#) (low levels of red blood cells). Anemia may make you feel tired and short of breath. Sickle cell disease can also cause problems with some organs, including the spleen, lungs, and kidneys. Treatment depends on the severity of the disease. Treatments for sickle cell disease may include medications, blood transfusions, and rarely, a bone-marrow transplant.

What are thalassemias?

Thalassemias are genetic blood disorders that cause anemia. These disorders are caused by a change in a part of hemoglobin, called “alpha” or “beta.” Some types of thalassemias are more severe than others. Mild forms of the disorder may not cause any symptoms. People with a severe form may have problems with some organs, including the bones, spleen, and heart. Treatment depends on the severity of the anemia and may include medications and blood transfusions.

What causes these disorders?

Sickle cell disease and thalassemias are genetic disorders, which means that they run in families. The disorders are caused by changes in the genes that help make hemoglobin. Genes are the instructions that control a function in the body or a physical trait, like eye color.

A person with sickle cell disease inherits two sickle cell genes, one from each parent. A person with thalassemia inherits changed hemoglobin or thalassemia genes.

What does it mean to be a carrier of sickle cell disease or thalassemia?

A [carrier](#) is a person who inherits one normal copy of a gene and one changed copy of a gene. Carriers of sickle cell disease are said to have “sickle cell trait.” People with

sickle cell trait do not show signs of the disorder, but they could pass the gene to their children. They often do not know that they have the disorder until they are tested.

People who inherit mild forms of thalassemia also may not know until they are tested. Those with severe thalassemia may find out early in life because they have symptoms of severe anemia. Many states screen all newborns for sickle cell disease and thalassemia.

Who is at highest risk of sickle cell disease?

Sickle cell disease occurs most often in people of African descent. About 1 in 10 African Americans has sickle cell trait.

Who is most commonly affected by genetic blood disorders?

- Sickle cell disease most often affects people of African, Mediterranean (especially Greek and Italian), Turkish, Arabian, Southern Iranian, and Asian Indian descent.
- Thalassemia most often affects people of Mediterranean (especially Greek or Italian), Middle Eastern, African, or Asian descent.

How is carrier screening for hemoglobinopathies done?

Carrier screening for hemoglobinopathies is done with a blood test. There are two options for how the blood sample is tested:

- Hemoglobin electrophoresis—This is a test that looks at the different types of hemoglobin in your blood. It can show if the blood is abnormal.
- Molecular genetic testing—This is a test of the genes in your blood. It checks for hemoglobinopathies and many other genetic disorders at the same time. This is also called expanded carrier screening.

One partner usually is tested first. If results show that the first partner is a carrier, the other partner is tested.

Read [Carrier Screening](#) to learn more.

When can carrier screening be done?

Some people decide to have carrier screening before getting pregnant. Carrier screening can also be done during pregnancy. Getting tested before pregnancy gives you more choices and more time to make decisions.

What happens if I am a carrier?

If your test results show you are a carrier, the next step is to have your partner tested. If your partner is also a carrier, your risk of having a child with a hemoglobinopathy is 1 in 4. Your risk of having a child who is a carrier is 1 in 2. If your partner is not a carrier, your risk of having a child with the disorder is very low.

If you and your partner are both carriers, you should talk with a [genetic counselor](#) or a doctor who is an expert in genetics. They can give you more information and answer your questions, including options for having a healthy pregnancy.

I am a carrier and I am pregnant. What does this mean for my pregnancy?

Women with sickle cell trait or thalassemia trait can have healthy pregnancies. [Prenatal care](#) is important.

Talk with your [obstetrician–gynecologist \(ob-gyn\)](#) or other health care professional about how to find out if your [fetus](#) has a hemoglobinopathy. You can have tests such as an [amniocentesis](#) or [chorionic villus sampling \(CVS\)](#) . (Read [Prenatal Genetic Diagnostic Tests](#).)

My partner and I are carriers and want to get pregnant. What are our options?

If you find out that you and your partner are carriers before pregnancy, choices include the following:

- You can get pregnant and then have testing to see if the fetus has a hemoglobinopathy.
- You can get pregnant using [in vitro fertilization \(IVF\)](#) . You can use your own eggs or sperm. You can also use donor eggs or sperm. Tests can be done on the embryo before it is transferred to the uterus to see if it has a hemoglobinopathy. This is called [preimplantation genetic diagnosis](#) .

- You can get pregnant using [intrauterine insemination \(IUI\)](#) . With this option, sperm from a donor who is not a carrier would be used.
- You may choose not to get pregnant.
- You may choose to adopt a child.

Are my test results confidential?

Yes, results from any genetic test are confidential. But you may want to tell other family members if you find out that you are a carrier for a genetic condition. They may be at risk of being carriers themselves. There is no law that says you have to tell anyone. Your test results cannot be shared without your consent.

I am concerned about discrimination based on genetic testing results. What should I know?

The Genetic Information Nondiscrimination Act of 2008 (GINA) makes it illegal for

- health insurers to require genetic testing results
- health insurers to use results to make decisions about coverage, rates, or preexisting conditions
- employers to discriminate against employees or applicants because of genetic information.

GINA does not apply to life insurance, long-term care insurance, or disability insurance.

Glossary

Amniocentesis: A procedure in which amniotic fluid and cells are taken from the uterus for testing. The procedure uses a needle to withdraw fluid and cells from the sac that holds the fetus.

Anemia: Abnormally low levels of red blood cells in the bloodstream.

Carrier: A person who shows no signs of a disorder but could pass the gene to his or her children.

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks. **Fetus:** The stage of human development beyond 8 completed weeks after fertilization.

Gene: A segment of DNA that contains instructions for the development of a person's physical traits and control of the processes in the body. The gene is the basic unit of heredity and can be passed from parent to child.

Genetic Counselor: A health care professional with special training in genetics who can provide expert advice about genetic disorders and prenatal testing.

Genetic Disorders: Disorders caused by a change in genes or chromosomes.

Hemoglobinopathies: Any inherited disorder that affects the number or shape of red blood cells in the body. Examples include sickle cell disease and the different forms of thalassemia.

Intrauterine Insemination (IUI): A procedure in which a man's semen is placed in a woman's uterus.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Obstetrician–Gynecologist (Ob-Gyn): A doctor with special training and education in women's health.

Oxygen: An element that we breathe in to sustain life.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are done on the fertilized egg before it is transferred to the uterus.

Prenatal Care: A program of care for a pregnant woman before the birth of her baby.

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, which causes chronic anemia and episodes of pain.

Thalassemia: A group of inherited anemias.

If you have further questions, contact your ob-gyn.

Don't have an ob-gyn? [Learn how to find a doctor near you.](#)

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