



## Carrier Screening

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### What is carrier screening?

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

### What is a carrier?

For some genetic disorders, it takes two genes for a person to have the disorder. A **carrier** is a person who has only one gene for a disorder. Carriers usually do not have symptoms or have only mild symptoms. Because they often do not know that they have a gene for a disorder, they can pass the gene on to their children.

### What are the chances of having a child with a genetic disorder?

If both parents are carriers of a recessive gene for a disorder, there is a 25% (1-in-4) chance that their children will get the gene from each parent and will have the disorder. There is a 50% (1-in-2) chance that the children will be carriers of the disorder—just like the carrier parents. If only one parent is a carrier, there is a 50% (1-in-2) chance that the child will be a carrier of the disorder.

### How is carrier screening done?

Carrier screening involves testing a sample of blood, saliva, or tissue from the inside of the cheek. Test results can be negative (you do not have the gene) or positive (you do have the gene). Typically, the partner who is most likely to be a carrier is tested first. If test results show that the first partner is not a carrier, then no additional testing is needed. If test results show that the first partner is a carrier, the other partner is tested. Once you have had a carrier screening test for a specific disorder, you do not need to be tested again for that disorder.

## When can carrier screening be done?

Some people decide to have carrier screening before having children. Carrier screening also can be done during pregnancy. Getting tested before pregnancy gives you a greater range of options and more time to make decisions.

## Do I have to have carrier screening?

Carrier screening is a voluntary decision. You can choose to have carrier screening, or you can choose not to. There is no right or wrong choice.

## What carrier screening tests are available?

Carrier screening is available for a limited number of diseases, including **cystic fibrosis**, **fragile X syndrome**, **sickle cell disease**, and **Tay–Sachs disease**. Some of these disorders occur more often in certain races or ethnic groups. For example, sickle cell disease occurs most frequently in African Americans. Tay–Sachs disease is most common in people of Eastern or Central European Jewish, French Canadian, and Cajun descent. But anyone can have one of these disorders. They are not restricted to these groups.

## Who should have carrier screening?

All women who are thinking about becoming pregnant or who are already pregnant are offered carrier screening for cystic fibrosis, **hemoglobinopathies**, and **spinal muscular atrophy**. You can have screening for additional disorders as well. There are two approaches to carrier screening for additional disorders: 1) targeted screening and 2) expanded carrier screening.

## What is targeted carrier screening?

In targeted carrier screening, you are tested for disorders based on your ethnicity or family history. If you belong to an ethnic group or race that has a high rate of carriers for a specific genetic disorder, carrier screening for these disorders may be recommended. This also is called ethnic-based carrier screening. If you have a family history of a specific disorder, screening for that disorder may be recommended, regardless of your race or ethnicity.

## What is expanded carrier screening?

In expanded carrier screening, many disorders are screened for using a single sample. This type of screening is done without regard to race or ethnicity. Companies that offer expanded carrier screening create their own lists of disorders that they test for. This list is called a screening panel. Some panels test for more than 100 different disorders. Screening panels usually focus on severe disorders that affect a person's quality of life from an early age.

## Is one approach better than the other?

Before testing, you and your **obstetrician–gynecologist (ob-gyn)** or other health care professional can discuss the benefits and limitations of each carrier screening approach and choose the one that is right for you. In some cases, both approaches can be used to tailor screening to your individual situation.

## What choices do I have if my partner and I are carriers of a genetic disorder?

If you have carrier screening before you become pregnant, you have several options. You may become pregnant and have prenatal **diagnostic tests** to see if the fetus has the disorder. You may choose to use **in vitro fertilization** with donor **eggs** or **sperm** to become pregnant. With this option, the fertilized egg can be tested before it is transferred to the uterus. You also may choose not to become pregnant. If you have carrier screening while you are already pregnant, your options are more limited. In either case, a **genetic counselor**, your ob-gyn, or other health care professional can explain your risks of having a child with the disorder.

## How accurate is carrier screening?

No test is perfect. In a small number of cases, test results can be wrong. A negative test result when you have a gene for the disorder tested is called a false-negative result. A positive test result when you do not have a gene for a disorder is called a false-positive result. Also, because carrier screening looks for only a limited number of genes, it is possible that you are a carrier of a genetic disorder even if your test results are negative.

## Are results of carrier screening confidential?

The Genetic Information Nondiscrimination Act (GINA) makes it illegal for most health insurers to require genetic testing results or use results to make decisions about coverage, rates, or preexisting conditions. Most employers are prohibited from using genetic information for hiring, firing, or making any other decisions about a person's employment.

If you find out that you are a carrier of a gene for a genetic disorder, you may want to tell other family members. They may be at risk of being carriers themselves. There is no law that states that you have to do so. If you choose to tell family members, your ob-gyn or genetic counselor can advise you about the best way to do this. It cannot be done without your consent.

## Glossary

**Carrier:** A person who shows no signs of a particular disorder but could pass the gene on 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.

**Cystic Fibrosis:** An inherited disorder that causes problems in digestion and breathing.

**Diagnostic Tests:** Tests that look for a disease or cause of a disease.

**Eggs:** The female reproductive cells produced in and released from the ovaries; also called the ova.

**Fragile X Syndrome:** A genetic disease of the X chromosome that is the most common inherited cause of intellectual disability.

**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. They are the basic units of heredity and can be passed down from parent to offspring.

**Genetic Counselor:** A health care professional with special training in genetics and counseling 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 caused by changes in the structure of hemoglobin, a substance found in red blood cells that carries oxygen. Examples include sickle cell anemia, sickle cell disease, and the different forms of thalassemia.

**In Vitro Fertilization:** 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 physician with special skills, training, and education in women's health.

**Sickle Cell Disease:** An inherited disorder in which red blood cells have a crescent shape, which causes chronic anemia and episodes of pain. It occurs most often in African Americans.

**Sperm:** A cell produced in the male testes that can fertilize a female egg.

**Spinal Muscular Atrophy:** An inherited disorder that causes wasting of the muscles and severe weakness.

**Tay–Sachs Disease:** An inherited disorder that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It most commonly affects people of Eastern and Central European Jewish, Cajun, and French Canadian descent, but it can occur in anyone.

### **If you have further questions, contact your obstetrician–gynecologist.**

**FAQ179:** Designed as an aid to patients, this document sets forth current information and opinions related to women's health. The information does not dictate an exclusive course of treatment or procedure to be followed and should not be construed as excluding other acceptable methods of practice. Variations, taking into account the needs of the individual patient, resources, and limitations unique to the institution or type of practice, may be appropriate.

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