ACOG
The American College of Obstetricians and Gynecologists

FAQs

Carrier Screening

Frequently Asked Questions

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 your chances of having a child with a genetic disorder.

What is a recessive disorder?

Most carrier screening is for recessive disorders. It takes two genes—one inherited from the mother and one inherited from the father—for a person to get a recessive disorder. If a person has only one gene for a disorder, he or she is known as a carrier. Carriers often do not know that they have a gene for a disorder. They usually do not have symptoms or have only mild symptoms.

What are the chances of having a child with a recessive disorder?

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

Some of these disorders occur more often in certain races or ethnic groups. For example, sickle cell disease often affects people of African descent. Tay–Sachs disease typically affects 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 getting pregnant or who are already pregnant are offered carrier screening for cystic fibrosis, hemoglobinopathies , and spinal muscular atrophy (SMA) . 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

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 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) can discuss the benefits and limitations of each carrier screening approach. You also can talk with a genetic counselor. In some cases, both targeted screening and expanded screening can be tailored 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 get pregnant, you have several options:

1. You can get pregnant and have prenatal diagnostic tests to see if the fetus has the disorder.
2. You can choose to use in vitro fertilization (IVF) with donor eggs or sperm to get pregnant. With this option, the embryo can be tested before it is transferred to the uterus.
3. You may choose not to get pregnant.
4. You may choose to adopt a child.

If you have carrier screening after you get pregnant, your options are more limited. In either case, your ob-gyn or a genetic counselor can explain your risks of having a child with the disorder.

How accurate is carrier screening?

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.

Are results of carrier screening private?

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

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 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.

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

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

Egg: The female reproductive cell made in and released from the ovaries. Also called the ovum.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

False-Negative: A test result that says you do not have a condition when you do.

False-Positive: A test result that says you have a condition when you do not.

Fetus: The stage of human development beyond 8 completed weeks after fertilization.

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

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.

Recessive Disorders: Genetic disorders caused by two genes, one inherited from each parent.

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

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

Spinal Muscular Atrophy (SMA): An inherited disorder that causes wasting of the muscles and severe weakness. SMA is the leading genetic cause of death in infants.

Tay–Sachs Disease: An inherited disorder that causes mental disability, blindness, seizures, and death, usually by age 5.

Uterus: A muscular organ in the female pelvis. During pregnancy, this organ holds and nourishes the fetus. Also called the womb.

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

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