The American College of Obstetricians and Gynecologists

FREQUENTLY ASKED QUESTIONS
FAQ179
PREGNANCY

Preconception Carrier Screening

What is preconception carrier screening?

Preconception carrier screening is screening that you can have before becoming pregnant to help predict your chances of having a child with a genetic disorder (see the FAQ Genetic Disorders).

What is a carrier?

A ***carrier*** is a person who has no symptoms (or only mild symptoms) of a disorder but can pass on the ***gene*** for that disorder to his or her child.

How is carrier screening done?

Carrier screening involves testing a sample of blood or saliva. The sample is sent to a lab for testing. Typically, the partner who is most likely to have a defective gene 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.

When can carrier screening be done?

You can have carrier screening before pregnancy (preconception) or during pregnancy. If it is done before pregnancy, you have a broader range of options and more time to make decisions.

What carrier screening tests are available?

Carrier tests are available for a limited number of diseases, including ***cystic fibrosis***, ***fragile X syndrome***, ***sickle cell disease***, and ***Tay–Sachs disease***.

Who should consider preconception carrier screening?

A health care provider or ***genetic counselor*** can help find out if you are at increased risk of passing on a genetic disorder by obtaining a family health history. This involves asking certain questions about your health and your family's health. You are at increased risk if

- you have a genetic disorder

- you already have a child who has a genetic disorder
- there is a family history of a genetic disorder
- you belong to an ethnic group that has a high rate of carriers of certain genetic disorders

Which ethnic groups have an increased risk of genetic disorders and what carrier screening tests are offered to these groups?

People from certain ethnic groups have an increased risk of passing on certain genetic disorders. For this reason, carrier screening is offered to certain groups as follows:

- Non-Hispanic white individuals should be offered cystic fibrosis carrier screening.
- People of Eastern European Jewish descent (Ashkenazi Jews) should be offered screening for Tay–Sachs disease, Canavan disease, familial dysautonomia, and cystic fibrosis. Individuals can ask about screening for other disorders. Carrier screening is available for mucolipidosis IV, Niemann–Pick disease type A, Fanconi anemia group C, Bloom syndrome, and Gaucher disease.
- People of African, Mediterranean, and Southeast Asian heritage should be offered screening for thalassemias and sickle cell disease.

What can the results of a carrier screening test tell me?

A genetic counselor or your health care provider will use the results to calculate the chance of you having a child with a genetic disorder. For most of the disorders for which carrier screening is available, if both parents are carriers, there is a 25% chance that the child will get the abnormal gene from each parent and will have the disorder. There is a 50% chance that the child will be a carrier of the disorder—just like the carrier parent. If only one parent is a carrier, there is a 50% chance that the child will be a carrier of the disorder and a 0% chance that the child will have the disorder.

What is a false-positive test result? What is a false-negative test result?

A false-positive test result is when a person tests positive for being a carrier but does not actually the gene. A false-negative result is when a person tests negative for being a carrier but actually does have the gene. Because test results can be wrong, it is possible for you to have a child with a genetic disorder even if your and your partner's test results are negative.

What decisions do I need to make if I am a carrier?

If you and your partner learn that both of you are carriers of a genetic condition, you have several options. You may choose to proceed with becoming pregnant, with the option of considering prenatal diagnosis. You may choose to use ***in vitro fertilization*** with donor ***eggs*** or ***sperm*** to achieve pregnancy. ***Preimplantation genetic diagnosis*** can be used with this option. You also may choose not to become pregnant.

What is the Genetic Information Nondiscrimination Act (GINA)?

GINA is a law that makes it illegal for health insurers to require genetic testing results or use results to make decisions about coverage, rates, or preexisting conditions. Employers are prohibited from using genetic information for hiring, firing, or making any other decisions about a person's employment.

Glossary

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

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

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

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

Gene: A DNA "blueprint" that codes for specific traits, such as hair and eye color.

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

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

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

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: The male cells produced in the testes that can fertilize a female egg.

Tay–Sachs Disease: An inherited birth defect that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It occurs mostly in people of Eastern European Jewish (Ashkenazi Jews), Cajun, and French Canadian descent.

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.