PATIENT EDUCATION

The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

Carrier Screening

*Carrier screening is a type of genetic test that can tell you whether you carry **genes** for certain **genetic disorders**. Some people decide to have carrier screening before having children. Doing so allows you to find out your chances of having a child with a genetic disorder. Although carrier screening can be done during pregnancy, getting tested before pregnancy gives you a greater range of options and more time to make decisions.*

This pamphlet explains

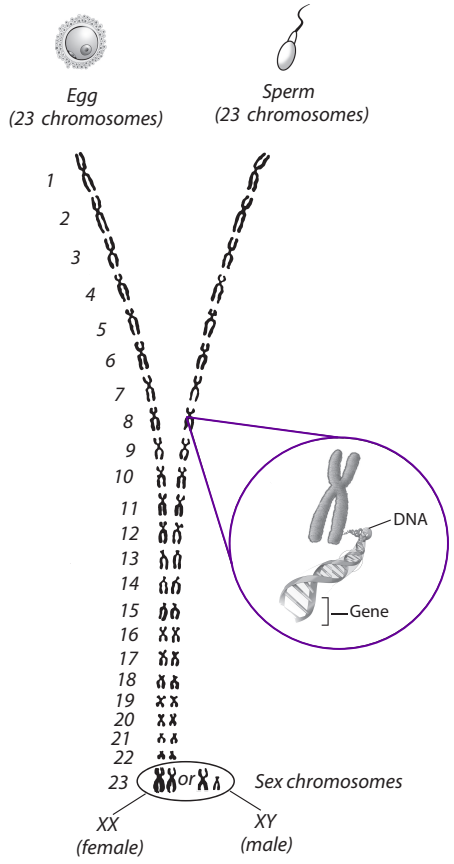
- *how genes work*
- *inherited genetic disorders*
- *how testing is done and what the results mean*
- *timing of carrier screening*
- *approaches to carrier screening*
- *deciding whether to have carrier screening*
- *what to do if you find out that you are a carrier of a genetic disorder*

How Genes Work

A gene is a short segment of a chemical called ***DNA***. Genes are the coded instructions for your physical makeup and every process in your body. Genes come in pairs and are located on ***chromosomes***. Chromosomes also come in pairs (one from each parent). Most ***cells*** have 23 pairs of chromosomes, making a total of 46 chromosomes. However, ***sperm*** and ***egg*** cells each have a single set of 23 chromosomes—half the number of other cells. During ***fertilization***, when the egg and sperm join, the two sets of chromosomes come together. In this way, one half of a baby's genes come from the mother and one half come from the father.

A baby's sex depends on the ***sex chromosomes*** it gets. Egg cells only contain an X chromosome. Sperm cells can carry an X or a Y. A combination of XX results in a girl, and XY results in a boy.

Chromosomes



Every cell in the body (except for eggs and sperm) has 46 chromosomes. One half comes from the mother and one half from the father. Chromosomes carry genes, which also come in pairs. The 23rd pair of chromosomes are the sex chromosomes.

Inherited Genetic Disorders

Some genetic disorders are caused by a change, or ***mutation***, in a gene. Most mutations are harmless. Other mutations cause diseases or affect a person's appearance or function. If a parent has a mutation, he or she can pass it down to a child. The chance of a child inheriting a mutation depends on whether the gene is dominant or recessive.

Recessive Disorders

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.

Some recessive disorders occur more often in certain races or ethnic groups. For example, ***sickle cell disease*** occurs most frequently in African Americans. ***Tay–Sachs disease*** typically occurs in people of Eastern or Central European Jewish, French Canadian, and Cajun descent, but anyone can have one of these disorders. Recessive disorders are not restricted to these groups.

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

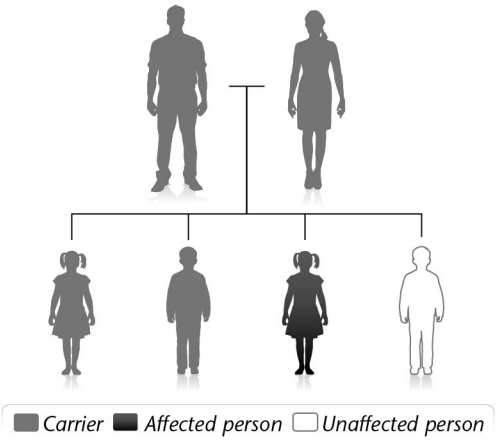
Dominant Disorders

In a ***dominant disorder***, only one gene from the mother or father is needed for a person to have the disorder. An example of a dominant disorder is neurofibromatosis, a group of disorders that causes the growth of tumors in the nervous system. Carriers of dominant disorders usually know that they have the disorder because of their symptoms. Carrier tests for dominant disorders usually are not needed.

Sex-Linked Disorders

Disorders in which the gene is carried on the sex chromosomes are called ***sex-linked disorders***. An example of a sex-linked disorder is ***hemophilia***. The gene for the most common type of hemophilia is on the X chromosome. A male child who gets the gene for this disorder will have the disorder. A female child who inherits the gene will be a carrier of the disorder.

Recessive Disorders



If both parents are carriers of a recessive gene for a disorder, there is a 50% chance (1-in-2) that a child they have will be a carrier, a 25% chance (1-in-4) that a child will have the disorder, and a 25% chance (1-in-4) that a child will neither be a carrier nor have the disorder.

How Testing Is Done and What the Results Mean

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).

Negative Test Results

If you are tested for a disorder and your test result is negative, no further testing is needed. But no test is perfect. In a small number of cases, test results can be wrong. A negative test result when you actually have a gene for the disorder tested is called a false-negative result. Because of the possibility of a false-negative result, a negative carrier screening test result does not completely rule out the risk that you are a carrier of a genetic disorder.

Positive Test Results

If your test result is positive for a disorder, the next step in carrier screening usually is to test your partner. If the results of both tests are positive, a ***genetic counselor***, ***obstetrician–gynecologist (ob-gyn)***, or other health care professional can explain your risks of having a child with the disorder. Like negative test results, there is a small chance that a positive test result is not accurate. A positive test result when you actually do not have a gene for a disorder is called a false-positive result.

Timing of Carrier Screening

Carrier screening can be done either before pregnancy or during pregnancy. If you have carrier screening before you become pregnant and you and your partner are carriers, you have the following four options:

1. You can become pregnant and then have prenatal ***diagnostic tests*** to see if the fetus has the disorder. Prenatal diagnostic testing involves having ***amniocentesis*** or ***chorionic villus sampling (CVS)***. Diagnostic testing is available for some, but not all, genetic disorders.
2. You can become pregnant using ***in vitro fertilization (IVF)***. You can use your own eggs or sperm or donor eggs or sperm. Tests can be done on the ***embryo*** before it is transferred to the uterus to see if the genetic disorder is present. This is called ***preimplantation genetic diagnosis***.
3. You may choose not to become pregnant.
4. You may choose to adopt a child.

If you have carrier screening after you become pregnant and you and your partner are carriers, your options are more limited. Pregnancy with prenatal diagnostic testing is an option. You also may decide not to have any testing.

Once you have had a carrier screening test for a specific disorder, you do not need to be tested again for the disorder. If new carrier screening tests become available for a disorder that you have not been tested for and for which you may be at risk, you may want to discuss carrier screening for these disorders with your ob-gyn or other health care professional.

Approaches to Carrier Screening

All women who are pregnant or thinking about becoming pregnant are offered carrier screening for certain disorders. These disorders include ***cystic fibrosis***, ***hemoglobinopathies***, and ***spinal muscular atrophy (SMA)***. Additional screening may be recommended based on certain factors.

There are two general approaches to carrier screening: 1) targeted carrier screening and 2) expanded carrier screening. Your ob-gyn, genetic counselor, or other health care professional can help you choose the approach that best addresses your concerns and also meets current recommendations for carrier screening (see Table 1).

Targeted Carrier Screening

Targeted carrier screening involves screening for certain disorders based on your ethnicity or family history. Traditionally, carrier screening has been recommended for people who belong to an ethnic group or race that has a high rate of carriers of a specific genetic disorder. Carrier screening for a specific disorder also may be recommended if you have a family history of that disorder, regardless of your race or ethnicity.

Screening based on your ethnic group or race is called ethnic-based screening. For example, ethnic-based carrier screening for Tay–Sachs disease has been recommended for people of Eastern or Central European Jewish, French Canadian, and Cajun descent.

An advantage of ethnic-based screening is that it is highly accurate. The accuracy of any laboratory test depends on how many people tested actually have the disorder. When you test only people who belong to a group at increased risk, test results are more likely to be accurate.

A main disadvantage of ethnic-based screening is that it has become harder to assign a person to a single race or ethnicity. Many people do not know their ancestry. As the population has become more diverse, carrier screening based on ethnicity or race may not be as accurate.

Expanded Carrier Screening

Advances in technology have resulted in another approach to carrier screening called expanded carrier screening (ECS). In ECS, many disorders are screened using a single sample. ECS is screening 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. Many of the disorders cause ***neurological*** problems, physical disability, or early death.

Table 1. Some Genetic Disorders for Which Carrier Screening Tests Are Available

Disorder	*What It Means*	*Who Is at Risk?*
Cystic fibrosis (CF)	CF affects the lungs, digestive system, and pancreas. Symptoms appear in childhood and include coughing, wheezing, loose stools, abdominal pain, and in men, infertility. Some people have milder symptoms than others. Over time the problems tend to become worse and harder to treat. The average life expectancy is 37 years.	Carrier screening should be offered to all women who are considering pregnancy or are currently pregnant.
Spinal muscular atrophy (SMA)	SMA causes muscles to break down (atrophy) and overall weakness. It is caused by a problem with the nerves that control movement. Of the three types, the most severe and most common (Type 1) causes death by age 2 years.	Carrier screening should be offered to all women who are considering pregnancy or are currently pregnant.
Fragile X syndrome	Fragile X syndrome is the most common inherited cause of intellectual disability. Disabilities range from mild (learning disabilities) to severe (autism). It affects both males and females, but some forms affect males more severely.	Carrier screening is recommended for women who have a family history of fragile X-related disorders, unexplained intellectual disability or developmental delay, autism with intellectual disability, or premature ovarian insufficiency.
Sickle cell disease	Sickle cell disease is a blood disorder that causes the red blood cells to have a crescent or "sickle" shape rather than the normal doughnut shape. The sickle cells can get caught in the blood vessels and prevent oxygen from reaching organs and tissues, which causes pain.	Carrier screening should be offered to women of African, Mediterranean, and Southeast Asian descent.
Thalassemias	Thalassemias are several types of blood disorders that cause anemia. Some types are more severe than others and can cause early death if not treated.	Carrier screening for the alpha-thalassemia trait should be offered to women of Southeast Asian, African, Mediterranean and West Indian descent. Carrier screening for beta-thalassemia mutations should be offered to women of Mediterranean, Asian, Middle Eastern, Hispanic, and West Indian descent.
Tay–Sachs disease	Tay–Sachs causes intellectual disability, blindness, and seizures. Symptoms first occur at about 6 months. Death usually occurs by age 5 years.	Carrier screening is recommended for women of Eastern or Central European Jewish, French Canadian, and Cajun descent.
Hemophilia	Hemophilia is a disorder caused by the lack of a substance in the blood that helps it clot. Affected people are treated with factors that help the blood clot and to help prevent excessive bleeding.	Women with a family history of hemophilia may request carrier screening.

The main advantage of ECS is that it allows you to be screened for a much greater number of disorders than ethnic-based screening. The cost of expanded carrier screening is about the same as that for single-disorder screening.

ECS also has many limitations. It is important to understand these limitations before choosing ECS:

- The tests on an ECS panel are not individually selected for you based on your personal and family history. For this reason, you may choose to be tested for fewer or additional conditions than those listed on an ECS panel based on your family health history or ethnicity. If you have a family history of a genetic disorder, you may benefit from having targeted genetic testing that looks specifically at your family mutation.
- ECS panels may include genes for disorders that in some people cause a disease but in others do not cause a disease. If a child inherits the gene for one of these disorders from both parents, it is not always certain whether the child will have the disorder.
- Because ECS tests for a large number of disorders, it is common to test positive as a carrier for one or more disorders. Most carriers of genetic disorders do not have any health problems. But in some instances, you may learn through carrier screening that you have the genes for a genetic condition that can affect your future health. If your partner is tested and he screens negative for the disorders you carry, your chance of having a child by the disorders is very low. If your partner screens positive for entirely different disorders than yours, it is unlikely that your child will be affected by any of the disorders.
- Many conditions that are screened are rare. You are probably at low risk of having the gene for many of the disorders that are tested. The likelihood

of having a false-positive result increases when you are at low risk. Also, not a lot of information may be known about rare disorders and how severely a child can be affected.

Deciding Whether 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.

If you do choose carrier screening, you should meet with your ob-gyn or genetic counselor before and after testing. Before testing, you should discuss the benefits and limitations of the screening approaches available. After testing, your health care professional can explain the results to you and help you make decisions if you have a positive result.

If you or your partner have ***consanguinity***, you also may be at an increased risk of certain conditions. In this case, talk with your health care professional about the benefits and limits of carrier screening.

What to Do If You Find Out That You Are a Carrier

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.

Many people are concerned about possible employment discrimination or denial of insurance coverage based on genetic testing results. 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.

Finally…

Carrier screening allows you to find out whether you carry genes for certain genetic disorders. There are many approaches to carrier screening. One approach is based on ethnicity or family history. Another is an expanded approach that screens for many disorders without regard to race or ethnicity. In some cases, both approaches can be used to tailor screening to your individual situation.

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.

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.

Cells: The smallest units of a structure in the body. Cells are the building blocks for all parts of the body.

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

Chromosomes: Structures that are located inside each cell in the body. They contain the genes that determine a person's physical makeup.

Consanguinity: A union between two people who are second cousins or closer in family relationship.

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.

DNA: The genetic material that is passed down from parent to child. DNA is packaged in structures called chromosomes.

Dominant Disorder: A genetic disorder caused by one gene.

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

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

Fertilization: A multistep process that joins the egg and the sperm.

Genes: Segments of DNA that contain 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.

Hemophilia: A disorder caused by a mutation on the X chromosome. Affected people are usually males who lack a substance in the blood that helps clotting. People with hemophilia are at risk of severe bleeding from even minor injuries.

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.

Mutation: A change in a gene that can be passed from parent to child.

Neurological: Related to the nervous system.

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

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.

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

Sex Chromosomes: The chromosomes that determine a person's sex. In humans, there are two sex chromosomes, X and Y. Females have two X chromosomes and males have an X and a Y chromosome.

Sex-Linked Disorders: Genetic disorders caused by a change in a gene located on the sex chromosomes.

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

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. It most commonly affects people of Eastern or Central European Jewish backgrounds, as well as people of French Canadian and Cajun backgrounds.

This is EP179 in ACOG's Patient Education Pamphlet Series.

ISSN 1074-8601

American College of Obstetricians and Gynecologists
409 12th Street, SW
PO Box 96920
Washington, DC 20090-6920