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Carrier Screen Test Patient Brochure

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What is a carrier screen test?

A carrier is a person who has a disease-causing change (mutation) in a gene for a genetic disorder but does not have any symptoms. Usually, the healthy carriers do not have a family history of a genetic disorder. They simply “carry” the gene for the disease.

For some genetic disorders, two copies of a disease-causing change, one from each parent, must be passed on for a child to have the disorder. This means that if two carriers have a child, there is a 25 percent chance that their child will have the genetic disorder.

A carrier screen test checks for genetic mutations that may be passed on to a child. Carrier screening can determine if you carry mutations for specific genetic disorders.

What makes carrier screening different from other genetic tests?

The carrier screen test is an all-in-one test for many genetic disorders you may pass on to your child. The advantages are:

- It is noninvasive, so there is no risk to you or your baby.
- You can have it before or during pregnancy, so it is more flexible than other options.
- It is universal, offering an easy way to check for common genetic disorders, even if you are not certain about family history or ethnicity.
- It is affordable.
- Cleveland Clinic’s Maternal-Fetal Medicine team helped develop the test.

Who should have the carrier screen test?

The American College of Medical Genetics and Genomics (ACMG) and the American Congress of Obstetricians and Gynecologists (ACOG) recommend all women of childbearing age have carrier screening.

Some genetic disorders are more common among certain ethnic groups and races. However, in a diverse population, many couples do not know their complete family histories and ethnicities.

This is why the ACMG and ACOG recommend that all women have carrier screening, which covers multiple genetic disorders.

Both you and your partner can have the carrier screen test. Usually, healthcare providers will recommend that a woman get the test first. If her results are positive, then her partner will need to have the test. You may choose to have both partners screened at the same time.

When should you have the carrier screen test?

You may have carrier screening before or during pregnancy. Some women and their partners prefer to have the test before having children so that they can make better plans for their family's future.

What does the carrier screen test include?

The expanded carrier screen test includes two disease panels (see tables below):

- The standard panel, which targets disorders found in many ethnic groups.
- The Ashkenazi Jewish disease panel, which targets disorders more frequently found in this ethnic group.

You and your healthcare provider will choose the panel that is best for your situation.

Carrier Screen Panels

Standard Carrier Screen Panel	Ashkenazi Jewish Disease Carrier Screen Panel
Bloom Syndrome	Bloom Syndrome
Canavan Disease	Canavan Disease
Cystic Fibrosis	Familial Dysautonomia
Familial Dysautonomia	Familial Hyperinsulinism
Familial Hyperinsulinism	Fanconi Anemia Group C
Fanconi Anemia Group C	Gaucher Disease
Fragile X Syndrome	Glycogen Storage Disease Type 1a
Gaucher Disease	Joubert Syndrome Type 2
Glycogen Storage Disease Type 1a	Maple Syrup Urine Disease 1b
Hemoglobinopathies	Mucolipidosis IV
Maple Syrup Urine Disease 1b	Niemann-Pick Types A and B
Mucolipidosis IV	Tay-Sachs Disease
Niemann-Pick Types A and B	Usher Syndrome IF
Sickle Cell Anemia	Usher Syndrome III
Spinal Muscular Atrophy	

Tay-Sachs Disease Thalassemia Usher Syndrome IF Usher Syndrome III	

Descriptions of Disorders Included in the Carrier Screen Panels

Disorder Name	Description
Bloom Syndrome	Bloom syndrome is an inherited disorder that can cause skin rashes, sensitivity to the sun and short stature. It increases the risk of cancer, diabetes, recurring infections and learning disabilities.
Canavan Disease	Canavan disease is an inherited disorder that affects how the nerve cells in the brain send and get signals. It affects motor skills and development. In the severe form, infants have weak muscle tone, problems sitting up and motor skill delays.
Cystic Fibrosis	Cystic fibrosis is an inherited disorder that affects breathing and digestion because of mucus buildup. The respiratory and digestive systems are damaged over time. Life expectancy is about 40 years old with treatment.
Familial Dysautonomia	Familial dysautonomia is an inherited disorder that affects the development of nerve cells. It can affect many parts of the body including breathing, digestion, blood pressure and the nervous system.
Familial Hyperinsulinism	Familial hyperinsulinism is an inherited disorder that affects insulin and can lead to seizures and other health problems. Insulin levels are higher than normal, which causes low blood sugar levels. This disorder can cause brain damage and death.
Fanconi Anemia Group C	Fanconi anemia group C is an inherited disorder that affects different parts of the body including bone marrow and red blood cells. The body does not get enough oxygen, which can lead to development problems, infections and cancer.
Fragile X Syndrome	Fragile X syndrome is an inherited disorder that affects the X chromosome and causes intellectual disability. It can cause developmental delays, intellectual disabilities, attention deficit disorders and seizures.
Gaucher Disease	Gaucher disease is an inherited disorder that affects different parts of the body because it cannot break down a specific type of fat. It can affect the liver, spleen and nervous system. The disorder can increase the risk of bruising and bone disease.
Glycogen Storage Disease Type 1a	Glycogen storage disease type 1a is an inherited disorder that affects the liver and creates problems with glycogen, which is a storage sugar. Glycogen builds up in the body and affects many organs such as the kidneys and liver. It can lead to low blood sugar levels, seizures, diarrhea and bone problems.
Hemoglobinopathies	Hemoglobinopathies are inherited disorders that affect the structure of hemoglobin. They can vary in severity. Mild cases may have no symptoms. Severe cases may cause organ damage, anemia and death.
Joubert Syndrome Type 2	Joubert syndrome type 2 is an inherited disorder that affects brain development. It can cause developmental delays, intellectual disabilities, breathing problems and kidney issues.

Maple Syrup Urine Disease 1b	Maple syrup urine disease 1b is an inherited disorder that affects the body's ability to process proteins. The name comes from the urine having a sweet odor. The disease varies in severity. It can be life threatening.
Mucolipidosis IV	Mucolipidosis type IV is an inherited disorder that affects development and vision, which get worse over time. It causes motor skill delays, intellectual disabilities, speech problems and weak muscles.
Niemann-Pick Types A and B	Niemann-Pick disease is an inherited disorder that affects lipid storage in the body. Symptoms include an enlarged liver, developmental regression, lung damage and eye problems. Those with type A do not survive past childhood. Those with type B usually survive until adulthood but have shorter life spans.
Sickle Cell Anemia	Sickle cell anemia is an inherited disorder that makes the red blood cells have a crescent shape. It can cause anemia and pain. Other problems include swelling, frequent infections, growth problems and eye problems.
Spinal Muscular Atrophy (SMA)	Spinal muscular atrophy is an inherited disorder that causes weakness and affects the muscles by making them waste away. It affects motor skills, eating and breathing. It is common for individuals to lose the ability to walk.
Tay-Sachs Disease	Tay-Sachs disease is an inherited disorder that causes seizures, intellectual disability and other health problems. The nerve cells in the brain and spinal cord are slowly destroyed. It can lead to death before the age of 5.
Thalassemia (Alpha-Thalassemia or α-Thal and Beta-Thalassemia or β-Thal)	Thalassemia is an inherited disorder that affects the blood and creates abnormal hemoglobin, which is a protein in red blood cells that carries oxygen. Symptoms can vary from mild to life-threatening. In general, there are fewer red blood cells in the body. Anemia is common.
Usher Syndrome IF	Usher syndrome type IF is an inherited disorder that causes hearing and vision loss. Hearing loss is usually present at birth. Vision loss can happen over time.
Usher Syndrome III	Usher syndrome type III is an inherited disorder that leads to the eventual loss of hearing and vision problems. Most individuals have problems later in life. Balance issues may also develop.

How do you prepare for the carrier screen test?

The test is noninvasive and requires no special preparation. However, you will need to have a blood draw. You may want to talk about any concerns you may have with your healthcare provider.

What are the limitations of the carrier screen test?

The carrier screen test only looks at the most common genetic mutations in the targeted (listed) disorders. It will not pick-up or detect carriers of uncommon or rare mutations for these disorders. Your chance of being a carrier of one of the tested disorders cannot be reduced to

zero or eliminated, but it can be greatly reduced. The carrier screen cannot guarantee that a child will not have one of these disorders due to a rare genetic mutation inherited from each of the parents. It also does not cover every possible disorder that a child may inherit.

It is not intended for patients who have symptoms of one of the disorders. It will not diagnose a genetic disorder affecting a baby during pregnancy. The carrier screen will provide a risk estimate for the parents, who can then consider further testing options.

If you or your partner have one of these genetic disorders or a family history of one of them, this may not be the best test for you. Your healthcare provider may have to order another type of test. Genetic counseling may be recommended based on your medical history or family history.

What do the results mean?

Test results are either positive or negative. A positive result means that you are a carrier for a genetic disorder. However, this does not mean that your child will definitely have this disease. It just means that you may be at a higher chance of having a child with this disease. Your healthcare provider will recommend that your partner have the test if they have not. Genetic counseling is available if you have any questions or concerns.

A negative result means that the screening did not find any of the genetic mutations included in the test. Based on a negative result, your chance of being a carrier for one of these disorders is greatly reduced. It cannot eliminate the possibility that you are a carrier because the screening does not test for every possible mutation. However, you do not need more testing if the results are negative.

Your healthcare provider will receive a PDF document with all of the results in one report. They can share the results with you and offer genetic counseling or additional testing.

How can you get more information?

Talk with your healthcare provider if you need more information about carrier screening or the results. Your doctor can also recommend genetic counseling and other resources.

To learn more about genetic disorders, you can visit:

- U.S. National Library of Medicine's Genetics Home Reference: https://ghr.nlm.nih.gov/
- National Human Genome Research Institute: https://www.genome.gov/10001204/specific-genetic-disorders/

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