OBSTETRICS & GYNECOLOGY
at The GW Medical Faculty Associates

2300 M Street, NW, Suite 110
Washington, DC 20037
Phone 202.677.6940

22nd and I Street, NW, 5th Floor
Washington, DC 20037
Phone 202.741.2500

Prenatal or Preconception Genetic Carrier Screening

Quick take: You can have a blood test to see if you carry things like sickle cell trait or cystic fibrosis. If you are a carrier you can have your partner screened to see if there is a risk to have a baby with that genetic disease or disorder.

Prenatal carrier screening involves a blood draw (or saliva sample) on one or both of the partners planning or during pregnancy to determine if they carry certain genetic conditions that could be passed on to their baby. With most carrier screening, the parent has no symptoms and the family history is normal. Most conditions are "recessive" or hidden and would only show up if a baby inherited two nonworking genes, one from each parent. Because results may be time sensitive, testing before pregnancy or soon after learning of the pregnancy is encouraged.

What is a carrier?

We all carry genetic conditions and experts estimate that each of us carries at least five conditions. For these recessive disorders, according to the Genetic Support Foundation, "…we have 2 copies of each gene, one from our mom and one from our dad. When we are a "carrier" of a genetic condition, this simply means that one of the copies out of the 2 genes has a change or mutation and is not working as usual. But, the other copy of the gene that is working compensates or makes up for the one that isn't working. So we don't have the genetic disorder or any symptoms; we are a "carrier" of the disease. Again, there are typically no health problems associated with being a carrier of a mutation for a recessive disease. However, as a carrier, there is a chance that your children could inherit that genetic disorder if your partner is also a carrier and you both pass the non-working gene on to your baby. If you AND your partner are carriers of the condition, there is a 25% chance or 1 in 4 chance for each of your children to inherit the condition". Examples of this would be cystic fibrosis and sickle cell disease.

Another pattern in genetics is called "X-linked" or sex-linked inheritance. These conditions are carried by the mother and typically would only be a risk to a son. Examples of this would be fragile X syndrome and Duchenne muscular dystrophy.

Carrier screening that is offered to everyone, regardless of their ancestry or background.

This carrier screening includes conditions that are relatively common in the population:

1) Cystic Fibrosis (CF): which is mainly a lung disorder but can also affect the digestion and pancreas function.

2) Hemoglobinopathies, including sickle cell, and beta thalassemia that affect how the blood functions and carries oxygen. These conditions are quite common in many areas of the world. They are especially important when one partner is of West African, African or Caribbean American, or Mediterranean (Greek). Individuals from southeast Asia who have a low blood count are offered screening for alpha thalassemia.

3) Spinal Muscular Atrophy (SMA) which is a neuromuscular disorder that affects how the nerves and muscles communicate. Affected individuals experience loss of muscle use over time.

For an affected individual (NOT for a carrier), all of these conditions can have a range of how early the symptoms occur (sometimes they may be seen before a baby is born and sometimes not until later in life), and how severe they can be (from mild to very severe). For some, treatment is available, or there may be significant research on treatment.

Carrier screening offered based on family history, background or ancestry:

Fragile X-Associated Disorders

These conditions are more commonly seen in males and may present with developmental delay, autism, and other symptoms. They may be offered when there is a family member with developmental delay, autism, and other cognitive disabilities. However, the American College of Medical Genetics and other professional societies do not suggest offering this to all patients unless there is a family history or some other indication for doing so.

Ashkenazi Jewish Heritage Carrier screening

There are several conditions that are more common in the Jewish population from Eastern Europe. Carrier screening for these conditions is offered, in addition to the common conditions noted above, to couples in which one or both members of a couple is of full or half Ashkenazi Jewish ancestry.

According to the Genetic Support Foundation, “There are a number of genetic disorders that are more common in people of Ashkenazi Jewish descent/ancestry; however, they may occur in people of other ethnic backgrounds as well. Many of these disorders can have a serious effect on the patient’s quality of life, some of which may result in shortened life expectancy. Some, like Gaucher, may have a milder effect. The following disorders are recommended (by the American College of Medical Genetics & Genomics) to be offered to couples who are planning a pregnancy or currently pregnant:

Familial dysautonomia, Tay-Sachs disease***,** Canavan disease, Fanconi anemia group C, Niemann-Pick type A, Bloom syndrome, Mucolipidosis IV, Gaucher disease Type I (There are additional conditions that are increased in risk in the Ashkenazi Jewish population for which you may be offered screening.)

***Tay-Sachs disease is also increased** in risk among individuals of French-Canadian, or Cajun ancestry and screening is typically also offered to individuals with these ethnic backgrounds.

Expanded Genetic Carrier Screening

Expanded genetic carrier screening panels were developed to offer carrier screening for multiple conditions at one time, as well as to cover many more conditions than those chosen based on ancestry or background. The panel may include all of the above conditions (often called a "Society-guided" panel). Larger panels include many more conditions, some even more than 100, for those couples who wish to get additional risk assessment for uncommon genetic conditions.

According to the Genetic Support Foundation, "Some of the conditions screened for in expanded carrier screening are more severe, some mild, some have treatment available and some don't. There is a very small chance that you could be found to have a genetic condition through this test as well (depending on what laboratory your doctor uses and what conditions are screened for). In general, this test often screens for most of the conditions commonly offered based on ethnic background (CF, common Ashkenazi Jewish diseases, and hemoglobinopathies). However, the labs offering expanded carrier screening may or may not be as comprehensive in their analysis as labs that focus on screening for individual conditions. For example, if you have a family history of a specific condition, expanded carrier screening may not be a good test for you.

As you can imagine, there are many viewpoints on the use of this test and many ethical issues involved. This test begs the question, "is bigger better?" Some would argue yes, some would argue no. We would say that like most things, there are pros and cons to this testing".

What do my results mean?

Normal/negative carrier screen results greatly reduce the chance that you are a carrier for the condition tested. If you have a positive result, you are a carrier. In this case the most important thing is to consider carrier screening for your partner. You may discuss your results with your provider or genetic counselor if you have questions.

What if both partners are carriers?

Even if both partners are carriers, there is a 75% chance that your baby will not inherit the condition. We provide genetic counseling for these couples to discuss their risks and options. Depending on the condition, some couples may do prenatal testing (amnio or CVS), others may wait to test the baby at birth. For couples doing IVF, the option of preimplantation genetic testing (PGT) may be considered.

Should I get carrier screening?

The decision to do carrier screening is personal. Many individuals choose to do carrier screening for the common conditions, conditions based on their ancestry or those related to a family history of a certain genetic disorder. Expanded carrier screening often appeals to individuals who feel more comfortable if they "know as much information as possible". Others find that "too much" screening makes them anxious.

The Genetic Support Foundation provides these helpful thoughts,

"This is an important question that we hope more people start asking themselves about this test and prenatal testing in general. Similar to other prenatal testing, there are some questions that can help you decide whether carrier screening is right for you.

- Am I the type of person who likes to know as much information as possible so I can plan accordingly and be prepared?
- Does more information increase my anxiety, and therefore, would I rather not find out more information regarding the conditions screened for by carrier screening?
- Do I want to minimize the possibility of intervention in my pregnancy?
- If carrier screening did indicate my baby was at an increased risk for one of the conditions, would I want to pursue further testing (possibly invasive testing that poses some risk to the baby)?
- If I knew that my baby had one of these conditions, would it affect my decision to continue the pregnancy and raise the child, or would I consider other options, such as, ending the pregnancy or placing the baby for adoption?

Ultimately, the decision to have carrier screening or any other prenatal screening is up to you. If you decide to do carrier screening you may ask for this at your regular gynecology visit, at your first obstetric visit, at your genetic counseling session, or you can contact your provider to order the test in advance".

For more information or to make an appointment for carrier screening and genetic counseling, please contact The GW Medical Faculty Associates:
202.741.2500
www.gwdocs.com/obgyn

Or go to:
https://geneticsupportfoundation.org/archive/genetics-and-you/pregnancy-and-genetics/pregnancy-and-genetics-tests/prenatal-carrier-screening

https://www.integratedgenetics.com/patients/pre-pregnancy