MAIN LINE FERTILITY

Carrier Screening: April 2017 American College of Obstetricians and Gynecologists

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 the chances of having a child with a genetic disorder.

What is a carrier?

For some genetic disorders, it takes two genes for a person to have the disorder. A carrier is a person who has only one gene for a disorder. Carriers usually do not have symptoms of have only mild symptoms. Because they often do not know that thy have a gene for a disorder, they can pass the gene on to their children.

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

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

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. Having testing done before pregnancy gives you a greater range of options and more time to make decisions.

Who should have carrier screening?

All women who are thinking about becoming pregnant or who are already pregnant are offered carrier screening.

What choices do I have if my partner and I are carriers of a genetic disorder?

If you have carrier screening before you become pregnant, you have several options. You may choose to use in vitro fertilization. With this option, the fertilized egg (embryo) can be tested before it is transferred to the uterus. Donor egg and donor sperm are also options. If you have carrier screening while you are already pregnant, your options are more limited. In either case, you will be referred to a genetic counselor who will explain the risks of having a child with the disorder.

Are results of carrier screening confidential?

The Genetic Information Nondiscrimination Act (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. Most employers are prohibited from using genetic information for hiring, firing, or making any other decisions about a person's employment.

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.

All Main Line Fertility physicians offer and recommend carrier screening for all patients.