What Is Genetic Carrier Screening?

Carrier screening is testing that is done to determine whether you or your partner carry a gene mutation that could cause serious disease in your baby.

You can have carrier screening before becoming pregnant or during your pregnancy. Ideally, having carrier screening before becoming pregnant allows you more time to consider your options and make decisions.

What Are Genes?

Genes are made up of DNA which contains the code that makes each of us unique. Genes are packaged into structures called chromosomes and are passed from parents to children. Traits such as eye color or risk for disease are passed to our children by our genes. Each of us received one chromosome from our mother and one chromosome from our father giving us our 23 pairs of chromosomes.

Dominant And Recessive Genes

Within each chromosome pair, one chromosome is dominant and the other chromosome is recessive. Usually the dominant chromosome masks the recessive chromosome and the traits from the dominant chromosome are expressed in our characteristics.

What Is A Carrier?

A carrier is a person who has a disease trait, but does not have any physical symptoms of the disease. A carrier has a gene mutation on the recessive gene. The dominant gene outweighs the recessive gene, so while a carrier does not develop the disease, a carrier can pass on the gene with a mutation to his or her child.

Because these disorders are recessive, this means that both you and your partner must have the same gene mutation in order for your children to inherit a given recessive disorder.

Who Should Have Carrier Screening?

DNA tests are available to help a couple decide if they are carriers for the same recessive disease and if they are at risk for having a baby with that recessive condition.

You are at increased risk if:

- You have a genetic disorder.
- You have a child who has a genetic disorder.
- Your family has a history of a genetic disorder.
- You are of ethnicity that has a high risk of certain carrier disorders.

Some Diseases Occur More Often In Certain Ethnic Groups

Here are the primary disorders related to ethnicity:

African, Asian, Mediterranean heritage: Thalassemia, Sickle Cell Anemia, and Cystic Fibrosis.

Ashkenazi Jewish heritage: Tay-Sachs, Canavan, Cystic Fibrosis, Familial Dysautonomia and other disorders.

European Caucasian and Hispanic heritage: Cystic Fibrosis.

French Canadian heritage: Tay-Sachs, and Cystic Fibrosis.

There are other disorders not listed here which can be screened for. Your doctor or healthcare provider will be able to provide you with more information.

Screening Methods for Carrier Disorders

There are two approaches to screening for carrier disorders: traditional carrier screening and expanded carrier screening. Both approaches share the same objective: - to inform couples of their risks so that they may consider their reproductive options.

Traditional Carrier Screening

Traditional Carrier Screening uses ethnicity and family history as a screen to identify individuals most likely to have a mutation. Traditional carrier tests re are designed to look for the most common mutations which occur in specific ethnic groups.

This approach yields high detection rates in specific populations, for example cystic fibrosis in Ashkenazi Jewish or sickle cell disease in African Americans. However, ethnic background is not always clear to define. Traditional carrier screening may not offer optimal detection rates in individuals who are not sure what ethnicity their ancestors were, or maybe be adopted or are mixed race.

Expanded Carrier Screening

Expanded Carrier Screening, ECS, sometimes referred to as pan-ethnic or universal screening allows testing of all individuals regardless of ethnicity. Expanded Carrier Screening utilizes new technology called Next Generation Sequencing and can screen for conditions across the spectrum from mild to severe. Otogenetics ECS panel screens for 168 diseases.

While ECS can provide better detection rates than traditional carrier screening in pan-ethnic groups, some of the conditions screened for are mild while others are quite severe.

Otogenetics Carrier Screening Test Panels

Single Gene Testing: CF, SMA, Tay-Sachs

Basic Panel screens for 5 diseases

ACOG/ACMG Panel screens for 13 diseases

Ashkenazi Jewish Panel screens for 39 diseases

Pan-Ethnic Panel screens for 168 diseases

Clinical Exome Sequencing

You will be able to decide with your doctor or healthcare provider which panel is right for you.

How is Genetic Carrier Screening Done?

If you choose to be screened, you'll be asked to give a blood, saliva or buccal sample which is sent to the lab where your DNA is examined for evidence of genetic mutations.

Accurate fetal risk assessment requires screening of the biological father of your baby. Therefore, if you're found to be a carrier, the father of your baby will be screened as well. Your doctor may also screen you and your partner at the same time.

What Do The Results Tell You?

If only one parent is a carrier, then the child has a 25% of being a carrier for that disorder and a 0% chance of having the disease.

If both you and your partner are carriers of the same gene mutation, then your child:

- Has a 25% chance of inheriting one affected gene from each of you and being born with the disease.
- Has a 50% chance of being a carrier just like the parent who is a carrier.
- Has a 25% chance of not being a carrier and not having the disorder.

This risk remains the same for each pregnancy.

Some conditions screened for in Expanded Carrier Screening can be semi-dominant, can have variable penetrance, or variable expressivity.

Semi-Dominant: Semi-dominance or incomplete dominance occurs when the physical characteristic of the child is distinct from both parents and is often intermediate to the physical characteristics of the parents. For example, in the snap dragon flower, the pink color of the flower is produced when true-bred parents of white and red flowers are crossed.

Variable Penetrance: For conditions that have variable penetrance, the age of onset of these disorders vary from person to person and in some cases an individual may not have any symptoms of the disorder.

Varying Expressivity: In genetic conditions with varying expressivity, the symptoms of the conditions are expressed differently among persons with the same genetic condition. This means that from your results of Expanded Carrier Screening you may learn about a previously unrecognized health risk which could be important to you and your family members. Some individuals may discover during the course of Expanded Carrier Screening that they are affected by one of the less severe or late-onset diseases.

A negative result suggests a low chance that you are carriers for the genetic conditions screened. However, a negative screening test does not eliminate the possibility that you are a carrier because there is always a residual risk.

The chance of having an affected child for the conditions screened depends on the carrier frequency in the population and the mutations tested. It may be difficult to calculate the residual risk for each condition screened for in Expanded Carrier Screening because the carrier frequency of certain conditions in the general population may be unknown.

What Are Your Options If You Find Out You Are Carriers?

If you find out that both you and your partner are carriers, you have several options:

You may choose to become pregnant and have prenatal diagnostic testing to confirm whether or not your developing baby has the disorder.

You may decide to try in vitro fertilization with donor eggs or sperm. Special testing of the embryo known as Preimplantation Genetic Diagnosis can be performed before it is implanted.

Some couples decide to adopt a child instead or not to have children at all.

Early Screening Offers Many Advantages

For some diseases, knowing your baby is affected before birth makes it more likely that your baby can be helped by early treatment.

It allows your doctor to line up the right medical specialists, who can be on hand to start treating your baby right after delivery.

For more information and patient advocacy groups, please visit our Patient Portal at www.otogenetics/patientportal.com

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Your Genetic Carrier Screening Options

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