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Carrier Screening

Carrier screening is an optional test offered to all pregnant women or women considering pregnancy in order to find out if a they are at increased risk to have a child with certain genetic conditions. While carrier screening provides you with more information about your carrier status for many genetic conditions, it cannot screen you and your partner for all genetic conditions. It cannot guarantee the birth of a healthy baby, but can provide reassurance by reducing the likelihood that a child could have one of the conditions included on the testing.

In order to understand what carrier screening is, it is helpful to know a little about genes. Genes, which are made up of DNA, act as the instructions for our bodies to grow and develop. Every person has two copies of most genes, one inherited from each parent. Sometimes, a person carries only one working copy of a gene, while their other copy is non-working. A non-working gene is caused by a gene mutation. Individuals with one non-working copy are called carriers. It is common to be a carrier of at least one condition and carriers do not typically have any symptoms of the condition they carry. As such, most carriers do not have any previous family history of the condition. However, if an individual inherits two non-working copies (one from each parent), they can have a genetic condition. Genetic conditions inherited this way are called autosomal recessive conditions. Genetic conditions can cause a range of health complications; some have severe complications and may be life-threatening while others may be milder and/or treatable.

Carrier Screening Options

Ethnicity-Based Carrier Screening: This includes screening for the conditions for which you have the highest chance to be a carrier, usually determined by your reported ethnicity and family history as some conditions are more common in certain ethnic populations compared to others.

- All individuals should be offered cystic fibrosis (CF) and spinal muscular atrophy (SMA) carrier screening.
- People of Eastern European Jewish ancestry (also called Ashkenazi Jewish) should be offered screening for Tay–Sachs disease, Canavan disease, and familial dysautonomia in addition to CF and SMA. The American College of Medical Genetics also suggests screening for Gaucher disease type I, Bloom syndrome, Niemann-Pick type A, Mucolipidois IV, and Fanconi anemia group C.
- People of African, Mediterranean, and Southeast Asian ancestry should be offered screening for the thalassemias and sickle cell disease.
- Individuals with Cajun or French Canadian ancestry should be offered carrier screening for Tay-Sachs disease.

If you have a family member who is either affected or is a known carrier for a specific genetic condition, you may be offered carrier screening for that condition, if available.

Expanded Carrier Screening: Also called universal carrier screening, this type of screening allows for a large number of conditions to be screened at the same time, regardless of the patient's reported ethnicity or family history. Some of the included conditions may be rare, and therefore less information is available on them. Additionally, some diseases can have implications for individuals who are carriers of the disease. Typical expanded panel options include:

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- An expanded panel of approximately 60 conditions in which there is at least a 1% chance to be a carrier. The conditions on this panel are all autosomal recessive with a known impact early in life, including intellectual and physical disabilities.
- An expanded panel of approximately 175 or more conditions, many of which are autosomal recessive with impact early in life, while others may have milder impact or later onset. In addition, several X-linked conditions that may be carried by females and affect males are included.

Should I have carrier screening?

Carrier screening is always optional. Any person can elect to have carrier screening. In most cases, carrier screening is performed on a blood or saliva sample. You can have carrier screening done prior to becoming pregnant or during your pregnancy. If you decide to have carrier screening, you may be tested first and then your partner will be tested only if you are found to be a carrier. Other times, both partners are tested at the same time. Regardless of your decision to have carrier screening, newborn screening tests babies for some, but not all, of the conditions included on carrier screening panels.

What if we are carriers?

Most conditions included in carrier screening are inherited in an autosomal recessive manner. Meaning, if you and your partner are both carriers of the same genetic condition, there is a 25% chance with each pregnancy for the child to be affected and a 75% chance they will be not be affected. However, there are conditions on some panels that can result in a chance as high as 50% to have a child with a genetic condition. Additional testing by an invasive procedure (chorionic villus sampling or amniocentesis) would be offered to determine if a pregnancy is affected. Confirmatory testing can also be done after the baby is born.

Some couples who receive information on their carrier status before pregnancy may also consider options to minimize the chance of an affected pregnancy. These options can include in-vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD) performed on their embryos to determine which embryos have the genetic disorder and which do not, IVF with donor eggs and/or donor sperm, and adoption.

Genetic counseling is recommended to discuss all these testing options.

What if our results are negative?

Regardless of which carrier screening option you may choose, typically only the most common gene mutations are detectable through carrier screening. If your carrier screen is negative, it can reduce but not eliminate your risk of being a carrier. Therefore even with a negative carrier screen, there is still a chance to have an affected child. Your genetic counselor and/or health care provider can tell you more specific information about carrier testing and the remaining chance of having an affected child.