UTHealth Houston | UT Physicians

WHAT IS CARRIER SCREENING?

Carrier screening is a noninvasive blood or saliva test available to anyone pregnant or considering pregnancy to find out if they have a higher risk of having a child with certain genetic conditions. Everyone "silently" carries genetic changes in their genes, even if they have a healthy family. These genetic changes can also be called mutations or variants. Since carriers usually don't have any symptoms, testing is the only way to find out if you are a carrier.

Most conditions on carrier screening are recessive disorders. This means that both biological parents must be carriers of the same condition to have a pregnancy or child at increased risk.

Carrier screening options

- Standard carrier screening – includes a few of the most common conditions, including cystic fibrosis (CF), spinal muscular atrophy (SMA), sickle cell anemia, and thalassemias. If you are of Eastern European Jewish ancestry (also called Ashkenazi Jewish), Cajun, or French Canadian, you may benefit from additional screening for conditions such as Tay-Sachs disease.
- Expanded carrier screening – includes hundreds of common and rare genetic disorders. Expanded screening is more likely to find carriers of genetic conditions than standard screening, which gives a better risk assessment. However, less information may be available about rare conditions or rare genetic changes.

What if we are both carriers for the same condition?

If both parents are carriers of the same recessive condition, there's a 25% (1 in 4) chance of having a pregnancy with that condition. Additional testing by an invasive procedure, chorionic villus sampling (CVS), or amniocentesis, would be offered to determine if a pregnancy is affected. Confirmatory testing can also be done after the baby is born.

Some families 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 testing (PGT), using donor eggs and/or donor sperm, and adoption.

What does a negative result mean?

A negative result significantly lowers, but does not eliminate, the risk of being a carrier. No test or combination of tests can screen for all genetic conditions and birth defects.

Is carrier screening required?

Carrier screening is always optional. Regardless of your decision, newborn screening tests babies for some, but not all, of the conditions included on carrier screening. Newborn screening is a blood test completed on the baby after delivery to check for a number of conditions. All babies born in Texas are required to have a newborn screening blood test soon after birth for certain disorders, since many babies with one of these conditions may not immediately present with noticeable symptoms. Abnormal newborn screens are reported to the pediatrician who helps coordinate confirmatory testing and treatment, if needed.

Where can I learn more?

Talking to a genetic counselor may be beneficial to learn more about these genetic testing options. **To schedule an appointment with a genetic counselor, please call our scheduling team at 713-486-9302.**

For more information, scan the QR code or visit **UTPhysicians.com/genetic-counseling.**