CARRIER SCREENING FAQS

WHAT IS CARRIER SCREENING?

Carrier screens are optional tests that people can do before becoming pregnant or during pregnancy. Genetic carrier screening is done through a blood draw, cheek swab or a saliva sample and can tell you whether you are at a higher or lower risk to have a baby with certain genetic conditions, such as cystic fibrosis or spinal muscular atrophy. Most of the conditions that are screened for are autosomal recessive.

WHAT ARE AUTOSOMAL RECESSIVE GENETIC CONDITIONS?

- We each have two copies of most of our genes, one copy we get from our mom and one from our dad. If someone has an autosomal recessive genetic condition, they inherited a copy of a non-working gene from both parents. If someone has one copy of one non-working gene and one working copy, they are a carrier for that condition.
- Everybody is a carrier of at least 5-6 recessive genetic conditions, but that doesn't mean that disease will ever be seen in the family. People who are carriers for a recessive condition usually don't have any related symptoms or health problems. Therefore, we can't determine who is a carrier just by examining the person's personal or family history.
- If you and your partner are both carriers for the same genetic condition, there is a 1 in 4 (or 25%) chance that you will both pass down the non-working gene to the baby. If the baby gets a non-working copy of the gene from both parents, they will have the recessive genetic condition.

HOW COMMON ARE THESE CONDITIONS?

How common these conditions are depends on a few things. Certain genetic conditions are more common in some ethnic groups. For example, about 1 in 27 people who have Ashkenazi Jewish ancestry are carriers for Tay Sachs disease, which is very rare in other ethnic groups. If you have a family history of a genetic condition, you may have a higher chance to be a carrier for that condition.

WHAT TYPES OF CARRIER SCREENING ARE AVAILABLE?

Basic carrier screening is offered for conditions that are more common among some or multiple ethnicities (for example, cystic fibrosis). Carrier screening for other conditions is typically offered based on your ethnic background (for example, Tay Sachs for people of Jewish descent) or family history (for example, Fragile X for people with a family history of intellectual disability).

SO THIS TEST CAN TELL ME IF I CARRY THESE CONDITIONS FOR SURE, RIGHT?

No version of carrier screening can test every gene or look for every genetic change that causes the condition. Results of any carrier screen are intended to reduce, but not eliminate, the risk, and do not guarantee that you or your children are or will be healthy and disease free.

WHAT HAPPENS IF I FIND OUT I'M A CARRIER FOR ONE OF THE GENETIC CONDITION SCREENED FOR?

If you are found to be a carrier, the chance for your baby to have this genetic condition is small. With recessive conditions, in order for there to be a risk for a baby to have one of these genetic conditions, both parents must be a carrier for the same condition. Your healthcare provider can help you determine what the chance is to have a baby with a given genetic condition with information regarding your carrier screening. Your healthcare provider can also help you plan next steps such as offering carrier screening to your partner or considering referral to a genetic counselor.

HOW CAN I DECIDE IF CARRIER SCREENING IS RIGHT FOR ME?

Carrier screening allows you to determine whether you may be at increased risk of having a child with a particular genetic disease and learn more about the prenatal and preimplantation testing options available. The decision to have genetic carrier screening is completely yours.

ACOG/ACMG ECS CARRIER SCREENING PANEL

Includes	All conditions included in any American College of Obstetricians and Gynecologists/American College of Medical Genetics and Genomics carrier screen recommendation, regardless of ethnicity or family history
Who it's best for	Appropriate for all patients
Conditions	*21 conditions:* Bloom Syndrome, Canavan Disease, Cystic Fibrosis (CF), Familial Dysautonomia, Familial Hyperinsulinism, Fanconi Anemia Group C, Fragile X, Gaucher Disease, Galactosemia, Glycogen Storage Disease Type 1A, Hemoglobinopathies, Joubert Syndrome Type 2, Lipoamide Dehydrogenase Deficiency, Maple Syrup Urine Disease Type 1B, Mucolipidosis Type IV, Niemann-Pick Type A/B, Phenylketonuria, Spinal Muscular Atrophy (SMA), Tay Sachs, Usher Syndrome Type 1F, Usher Syndrome Type 3A

ABOUT PREMIER GENOMICS

Premier Genomics is committed to advancing the field of personalized genetic medicine by offering cutting-edge genetic screening services to help practitioners and their patients in pursuit of tailored treatment and optimized, personalized health care.

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