Genetic Carrier Screening – What is it and Do I Need it?

As with fetal screening for chromosomal abnormalities, genetic carrier screening of parents is optional but should be offered to all potential parents.

Carrier screening is testing that's done to see if you or your partner carry a genetic mutation that could cause a serious inherited disorder in your baby. Some of the more common disorders screened for include cystic fibrosis, sickle cell disease, thalassemia, and Tay-Sachs disease. If you have a family history of any of these diseases below, please let us know.

All of us may carry one or more genetic mutations that have no or minimal effects on our health. The more disorders you screen for, the more you are likely to find. Most disorders need both parents to carry the same gene mutation for there to be a risk to the baby. If you carry one or more of these mutations, your partner will need to be screened to determine if your fetus is at risk for the disorder.

How is the screening done?

Your practitioner will ask you a lot of questions about your risk factors for genetic disorders at your preconception appointment or first prenatal visit. If you or your partner is found to be at high-risk of being a carrier for a certain disorder, your practitioner should offer to screen you for it.

Risk factors include having a family member with the inherited disorder (or a family member who's a known carrier) or being part of an ethnic group at increased risk for the disease.

If you opt for this kind of screening, you'll probably be asked to give a blood sample first. Then if you're found to be a carrier, your partner will be screened as well. (Both partners may be screened at the same time to get the results faster.)

Expanded Carrier Screening of the Parents:

- Today most parents have some type of expanded carrier screening which includes a certain number of potential genetic mutations that could affect their children, including those genetic diseases recommended for carrier testing by the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG).

    - These panels have been expanded to include more advanced detection of Cystic fibrosis, Fragile X, Duchenne Muscular Dystrophy and Spinal Muscular Atrophy (SMA) carrier status.
    - With high accuracy, this panel offers multi-ethnic carrier screening. This helps doctors refine their understanding of the risk for each patient of passing on genetic diseases to the next generation.
    - For example, ***Horizon 27 Panel*** tests for: Alpha Thalassemia, Batten Disease (Neuronal Ceroid Lipofuscinosis CLN3-Related), Beta-Hemoglobinopathies, Bloom Syndrome, Canavan Disease, Citrullinemia Type 1, Cystic Fibrosis, Duchenne Muscular Distrophy, Familial Dysautonomia, Fanconi Anema Group C, Fragile X Syndrome, Galactosemia, Gaucher Disease, Glycogen Storage Disease Type la, Isovaleric Acidemia, Medium Chain Acyl-CoA Dehydrogenase Deficiency, Methylmalonic Aciduria and Homocystinurioa Type cbIC, Mucolipidosis Type IV, Mucopolysaccharidosis Type 1 (Hurler Syndrome), Niemann-Pick Disease Types A/B, Polycystic Kidney Disease Autosomal Recessive, Rhizomelic Chondrodysplasia Puncata Type 1, Smith-Lemli-Opitz Syndrome, Spinal Muscular Atrophy, Tay-Sachs Disease (DNA only); Tyrosinemia Type 1, Zellweger Spectrum Disorders PEX1-Related.
    - Other panels test for many more genetic carrier mutations.