Barton
Women's Health

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GENETIC CARRIER SCREENING FOR WOMEN CONSIDERING PREGNANCY

What is genetic carrier screening?

Genetic carrier screening is a test that checks to see if you (or your partner) are carriers of a recessive or X-linked genetic disorder. If you are a carrier of a recessive genetic disorder, you may have no symptoms or only mild symptoms. If both parents are carriers of the same condition, their children are at increased risk of developing that genetic disorder. A female carrier of an X-linked disorder has a risk of transmitting this disorder, particularly to male children. Some couples prefer to start by screening one partner. If the first partner tested is not a carrier, no additional testing may be needed. If test results show that the first partner is a carrier, the other partner should be screened as well. You can undergo carrier screening before you become pregnant or during pregnancy. If you are screened before pregnancy, you may have a broader range of options and more time to make decisions.

Who should be screened for genetic diseases?

Recommendations regarding who should receive genetic carrier screening and for which diseases continue to change. The American College of Obstetricians and Gynecologists (ACOG) as well as the American Council of Medical Geneticists and the California State Prenatal Screening Program all have slightly different recommendations. There is general agreement about testing in a few areas:

• All women should be offered screening for cystic fibrosis and spinal muscular atrophy.

• Ashkenazi Jewish women should be offered screening for Tay-Sachs, familial dysautonomia, cystic fibrosis and Canavan disease. Additional tests for this group may include mucolipoidosis IV, Niemann-Pick disease type A, Fanconi anemia, Bloom syndrome and Gaucher's disease.

• People of African, Mediterranean and Southeast Asian heritage should be offered screening for thalassemia and sickle cell diseases.

What does it mean if I test positive?

If you test positive, it means that you are a carrier for that specific disease. You may be unaffected, but depending on the carrier status of your partner, you may have an increased risk of having a child affected by the disease. If you test positive, you will have the opportunity to discuss the results and follow-up testing with a genetic counselor.

How is carrier screening performed?

We partner with an independent laboratory called Natera to provide genetic carrier screening. We have chosen to do this because the company provides a high quality, cost-effective service. The test is called Horizon and there are several different panels and your physician will help you choose which panel is right for you. Horizon is covered my most insurance providers and will be billed directly to your insurance by Natera. You will also be provided information to access the cost estimator which can be found at : http://services.natera.com/natera-calc/

For more information about Natera, please visit natera.com. If you have been tested using Natera, genetic counseling services are available by calling 650-249-9090 or by visiting naterasession.com This service is free of charge.

What is tested in each panel ?
Horizon 4 - Cystic Fibrosis - Spinal Muscular Atrophy - Fragile X Syndrome - Duchenne Muscular Dystrophy
Horizon 27 (Pan-ethnic standard) - All genes is Horizon 4 - Alpha- Thalassemia - Beta-Thalassemia - Bloom Syndrome - CLN3 Disease - Canavan Disease - Citrullinemia Type I - FANCC- Related Fanconi Anemia - Familial Dysautonomia - Galactosemia - Gaucher Disease - Glycogen Storage Disease Type Ia - Hemoglobin E, SC, SD, SS - Isovaleric Acidemia - MCAD deficiency - Methylmalonic Aciduria - Mucopolysaccharidosis Type I - Niemann Pick Disesase Type A and B - Polycystic Kidney Disease, autosomal recessive - Rhizomelic Chondrodyplasia Punctata Type 1 - Smith Lemli Opitz Syndrome - Tay Sachs Disease - Tyrosinemia Type 1 - Zellweger Syndrome Spectrum