Sutter Health
Sutter East Bay
Medical Foundation

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.

continued on back

How is carrier screening performed at EBPMG/SEBMF?

We partner with an independent laboratory called Counsyl to provide genetic carrier screening. We have chosen to do this because the company provides a high-quality, cost-effective service.

Your provider will recommend which panel of tests is right for you and give you a form to take to a Sutter Health laboratory. The Counsyl test is covered by most insurance plans and will be billed to your insurance directly by Counsyl (not by Sutter Health). Within 48 hours of receiving your sample, Counsyl will send you a text or email message with information regarding your price estimate.

At this time, you can opt to run the test through your insurance at the cost provided in your estimate or choose the cash option ($349 in March 2017). If you have any questions about your estimate or insurance coverage, Counsyl can be reached at 888-COUNSYL.

For more information about Counsyl, please visit counsyl.com.

If you have been tested using Counsyl, genetic counseling services are available by calling 888-COUNSYL or by visiting counsyl.com/counseling. This service is free of charge.

Which carrier panels are offered by EBPMG/SEBMF?

FUNDAMENTAL PANEL

- Cystic Fibrosis
- Spinal Muscular Atrophy
- Fragile X Syndrome*

ASHKENAZI JEWISH PANEL

- Cystic Fibrosis
- Spinal Muscular Atrophy
- Fragile X Syndrome*
- ABCC8-Related Hyperinsulinism
- Bloom Syndrome
- Canavan Disease
- Familial Dysautonomia
- Fanconi Anemia Type C
- Glycogen Storage Disease type 1a
- Hexosaminidase A deficiency (including Tay-Sachs)
- Gaucher's Disease
- Joubert Syndrome
- Lipoamide Dehydrogenase Deficiency
- Maple Syrup Urine Disease Type 1B
- Mucolipidosis IV
- NEB-related Nemaline Myopathy
- Niemann-Pick Disease SMPD1-Associated
- Usher Syndrome Types 1F and 3

UNIVERSAL PANEL

For a complete list of diseases, go to counsyl.com.

* *Must be specifically requested to be in your panel*

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