



## Informed Consent for Genetic Disease Carrier Screening

The goal of our practice is to make sure that you receive optimal care and attention to improve your chances of having a healthy pregnancy and a healthy child. As a part of your care, we offer screening for hereditary conditions that can be passed on to children from carrier parents. Carriers of hereditary conditions typically do not exhibit signs or symptoms of the disease so testing is necessary to detect carrier status. Most hereditary conditions can be inherited only if both parents are carriers of the gene that causes the condition; however, some can be passed on if only one parent carries the relevant gene. Genetic carrier screening can help you understand your risk of having a child with a genetic disease.

There is a wide range of genetic carrier screening options. Professional societies, including the American College of Obstetricians and Gynecologists ("ACOG") and the American College of Medical Geneticists ("ACMG"), recommend certain testing for prospective parents. For example, ACOG recommends that all women, regardless of their ethnicity, be screened for cystic fibrosis and it also recommends additional testing in persons who are members of certain ethnic groups or who have family histories that place them at increased risk of certain genetic diseases. ACMG recommends testing for additional disorders, such as spinal muscular atrophy. Expanded testing options are also available to you, which include testing for additional disorders not currently recommended by ACOG and ACMG. Your physician will discuss the available genetic carrier screening options with you, and he or she can help you to determine which tests are right for you based on your medical history and that of your family.

If both you and your partner are carriers for the same disease, your child has a 1 in 4 (25%) chance of having that disease. Certain disorders, such as Fragile X, can be inherited if just one parent is a carrier, and the risk could be as high as 1 in 2 (50%). Most autosomal recessive conditions do not impact the health of a carrier. Some carrier screening panels include conditions which may have medical consequences for carriers. Please notify your provider if you wish to opt out of testing for these conditions. If you are found to be at risk for having a child with a genetic disease, there may be reproductive treatment options available to you that could reduce your risk of having an affected child. In the majority of cases, pre-implantation genetic diagnosis ("PGD"), as well as prenatal diagnosis, are available to you. PGD can help reduce the risk of conceiving a child with a genetic disease and prenatal testing can identify if a pregnancy is affected. You will have the opportunity to speak with your physician and/or a genetic counselor about the options available to you.

You should be aware that because not every genetic mutation and disorder can be identified through genetic carrier screening, a negative genetic carrier screening means that you have a lower chance of having a child with a genetic disease, but it does not mean that you have no chance of having a child with a genetic disease. In addition, you may be a carrier of a genetic disease that was not tested through genetic carrier screening.

Regardless of your choice of testing or your test results, genetic counseling is available to you at any time. Genetic counseling itself may be an appropriate alternative to genetic carrier screening. If you would like to schedule an appointment with a genetic counselor, please let your physician or nurse know and they will assist you.

We will draw your blood in our lab and it may be sent out to an outside laboratory for processing/testing. You will receive a bill from Lucile Packard Children's Hospital at Stanford ("Stanford Children's") for the blood draw as well



Medical Record Number

Patient Name

**CONSENT FOR GENETIC DISEASE  
CARRIER SCREENING**

Page 2 of 2

Addressograph or Label - Patient Name, Medical Record Number

as the processing/testing if the tests are conducted at Stanford Children's. If the processing/testing is done by an outside laboratory, you may receive a separate bill for processing/testing fees from the outside laboratory. The genetic carrier screening we offer is covered by most insurance policies. If your insurance does not fully cover the testing, you may be responsible for paying the difference between the amount covered by your insurance company and the amount billed by the laboratory. You should check with your insurance carrier to see if the genetic carrier screening will be covered by your plan.

**ACKNOWLEDGEMENT:**

I have met with my physician and/or genetic counselor to discuss genetic carrier screening. I have been fully informed of the purpose of the genetic carrier screening, the reliability of the screening results, the risks and benefits of electing or declining genetic carrier screening and the available alternatives to genetic carrier screening, such as genetic counseling only.

I have been given the opportunity to ask questions and to discuss this testing with my physician and/or genetic counselor. My questions were answered to my satisfaction.

Please initial your selection:

\_\_\_\_\_ I would like to undergo genetic carrier screening and authorize the staff at Stanford Children's to perform a blood draw to perform genetic carrier screening.

\_\_\_\_\_ I decline genetic carrier screening.

\_\_\_\_\_ I wish to defer my decision regarding genetic carrier screening and will contact my health care team if and when I desire genetic carrier screening.

\_\_\_\_\_ I would like to schedule an appointment with a genetic counselor to review my family history and discuss carrier screening options

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**PATIENT'S SIGNATURE**

\_\_\_\_\_  
**PATIENT'S PRINT NAME**

**DATE** \_\_\_\_\_ **TIME** \_\_\_\_\_

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**PROVIDER'S SIGNATURE**

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**DATE** \_\_\_\_\_ **TIME** \_\_\_\_\_