



Carrier Screening For Genetic Diseases

The goal of our practice is to make sure that you receive optimal care and attention to improve your chance of having a healthy pregnancy, and of course, a healthy child. To help achieve that goal, you can choose among a variety of testing options. One such option, prenatal genetic carrier screening, can help identify whether you are at risk for having a child with a genetically inheritable disease.

Although many health individuals will be a carrier of some type of genetically inheritable disease, the likelihood of your child having such a disease remains small--in the 6-8/1000 range (0.6-0.8%)--because for the majority of diseases both parents must carry the same mutation, and your child must be the 1 in 4 (25%) who inherits the mutation from both parents. Because these circumstances are rare, and carriers are typically healthy, most people do not know they are carriers until they have a child born with a disease.

Carrier screening looks for a variety of genetically inheritable diseases including Cystic Fibrosis, Alpha Thalassemia, Tay-Sachs disease, and Sickle Cell disease, in addition to hearing disorders and metabolic problems. While the test screens for over 100 genetic conditions in total, some genetically inheritable diseases may not be detected. Testing therefore significantly reduces, but does not completely eliminate, the chance that a child would inherit a genetic disease. Some genetic diseases can significantly impair a child's normal development. For some of these conditions, early treatment can improve pregnancy outcomes. Should your carrier screen return positive, you will have the opportunity to speak with a genetic counselor about the meaning of your result and, if you and your partner carry the same mutation, the medical options available to you.

Carrier screening is covered by most insurance policies and results reach your physician about 2-3 weeks after you submit your sample.

Please sign this form and feel free to ask your doctor if you have any further questions.

My partner and I are declining any/all genetic carrier screening

My partner and I wish to proceed with Carrier screening

Name

Signature

Date

My partner and I will consider carrier screening

Name

Signature

Date