PacificWomen's
OBSTETRICS & GYNECOLOGY

Carrier Screening For Genetic Diseases

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 of course, a healthy child. To achieve that goal, you can choose among a variety of testing options. One such option, parental genetic carrier screening, can help identify whether you are at risk for having a child with a genetically inheritable disease.

Although it is estimated that 1 in 4 healthy 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 0.6-0.8% range (6-8/1000)--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, most people do not know they are carriers until they have a child born with a disease.

The Universal Genetic Test screens for a variety of genetically inheritable diseases including Cystic Fibrosis, Tay-Sachs disease, and Sickle Cell disease. The Test screens for 118 mutations in total, so some genetically inheritable diseases are not 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 test return positive, you will have the opportunity to speak with a genetic counselor about the meaning of the test and, if you and your partner carry the same mutation, the medical options available to you.

The Universal Genetic Test is covered by most insurance policies. Results reach your physician about 2-3 weeks after you submit your sample. Once reviewed, your doctor will post your results along with extensive explanatory information at the secure Counsyl website, www.counsyl.com. We strongly recommend that you sign up on the site to track your results. We also recommend screening both biological parents simultaneously to eliminate the 2-3 week wait for definitive results should one parent carry a mutation.