Carrier Screening for Genetic Diseases

Carrier screening is optional genetic (DNA) testing that can help you understand your risk of having a child with a genetic disease. Most people who have a child with a recessive genetic disease do not have a family history of the disease and were not previously aware that they are carriers of it. ORM therefore offers the Counsyl laboratory's Family Prep Screen to all patients. This test screens for over 175 recessive and X-linked genetic diseases such as cystic fibrosis, spinal muscular atrophy, Tay-Sachs disease, and sickle cell anemia.

> **Recessive genetic diseases:** We have two copies of most of our genes, one from our biological mother and one from our biological father. Being a carrier means that one of the two copies is not working correctly (has a mutation). If both an egg and sperm provider are carriers of the same disease, the child has a 25% chance to inherit two mutations, causing the genetic disease. Carriers are typically healthy; however, being a carrier of certain diseases can increase some specific health risks, such as cancer. Rarely, carrier screening will identify people who are actually affected with a recessive disease.
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> **X-linked genetic diseases:** X-linked diseases are caused by mutations on the X chromosome. Males (XY) are more likely to be affected by these mutations because they only have one copy of the X chromosome. Females (XX) have two copies of the X chromosome, and female carriers of X-linked diseases may have affected male children. Female carriers are often healthy; however, some may develop symptoms of the disease as well.

Being a carrier is common: about 60% of patients tested by the Counsyl Family Prep Screen are carriers of at least one of the diseases on the panel. The test focuses on genetic diseases that have moderate to profound severity; many are associated with learning problems, reduced life expectancy, or need for ongoing medical care or surgery. A list of the diseases included on this screen is available at www.counsyl.com.

In the unlikely event that you are found to be at high risk for having a child with a genetic disease, this information can help you to prepare for the possibility of an affected child. You will also have the chance to learn about options available to you to help reduce the chance of this happening.

As with any carrier screen, the test is not able to identify all carriers: a normal test result reduces, but does not completely eliminate, the chance you could be a carrier. If you have a family history of any genetic disease, we recommend that you talk with a genetic counselor prior to having this test and before conceiving. Genetic test results are confidential and are protected in health insurance and employment by the Genetic Information Non-Discrimination Act of 2008.14.

By signing below, I indicate my understanding of the above information and that genetic carrier screening will be arranged through Counsyl laboratory (an independent laboratory in San Francisco, CA). I have received all of the information I requested, and I have had all my questions answered in a satisfactory manner.

☐ **I consent to have carrier screening with the Counsyl Family Prep Screen. I understand that in order for my screening results to be applied to the goal of understanding the chance of and/or preventing a genetic disease from occurring in my offspring, I must not begin any fertility treatment until my results have been issued, I have received counseling about them, and, if desired, my reproductive partner has been screened.**

Counsyl phone genetic consultation date (to receive results): _____

Positive results must be reviewed with a genetic counselor at Counsyl before treatment can continue.

☐ **I decline to have carrier screening through ORM. This includes the Counsyl Family Prep Screen and other forms of carrier screening that may be offered based on my specific ethnic background.**

Patient Signature: _____ Date: _____

Patient Name (print): _____ DOB: _____