GENETIC CARRIER SCREENING INFORMATION & CONSENT

Genetic Carrier testing is a type of genetic testing that is used to determine if a person is a carrier for a specific disease. A carrier is a person who inherited a change or mutation in one gene copy of a pair of genes for a specific disease. Carriers often show no symptoms of the genetic disorder for which they carry an abnormal gene. For these kinds of genetic diseases to be present in a person, two copies of the abnormal gene are generally needed. This means that both parents must be a carrier for the child to inherit the disease. Usually, the only time a person finds out that they are a carrier for a specific genetic disorder is when they have an affected child. This kind of testing is used most often by couples who are considering becoming pregnant or who are already pregnant to determine the risks of their child inheriting one of these genetic disorders.

How is testing done

Carrier testing is most often done through a simple blood test. The results are available in approximately two weeks. Certain genetic disorders tend to be more common in people of a particular ethnicity. Genetic carrier testing generally is performed for the most common diseases within a population. As this testing involves genetics and genes do not change over time, testing is only required once in your lifetime.

Interpreting Results

A negative test result significantly lowers but does not eliminate the risk of being a carrier. Carrier testing is not able to detect all the genetic abnormalities that cause a particular disease. A positive test indicates that you are a **carrier** of the specific disease (this does not mean that you have the disease). For most diseases, the next step is for the father of the baby to have carrier testing performed. For most genetic conditions, both parents must be carriers for the baby to be at risk of having the genetic disease. If the father has a negative test, there is little/no chance that the baby will have that particular genetic disease. If both parents are carriers, the next step is for you to consider prenatal testing by amniocentesis or chorionic villi sampling (CVS) to determine if your baby is affected. For a few genetic conditions (X-linked conditions), only the mother has to be a carrier for a genetic abnormality. In those cases, prenatal testing would be offered following a positive test result of the mother. Genetic counselors are available.

Insurance Coverage

Many insurances cover genetic carrier testing. Testing may be subject to your deductible or a co-payment. It is your responsibility to confirm insurance coverage prior to testing or acknowledge your acceptance of financial responsibility.

Listed below are common diseases that can be screened for with genetic testing. Exact carrier screening is based on the panel that you are sent for or that your insurance covers.

Alpha-thalassemia	Beta-hemoglobinopathies (includes sickle cell disease)
Canavan disease	Cystic fibrosis
Familial dysautonomia	Duchenne/Becker muscular dystrophy
Fragile X syndrome	Galactosemia
Gaucher disease	Polycystic kidney disease
Spinal muscular atrophy	Smith-Lemli-Optiz syndrome
Tay-Sachs disease	Fanconi anemia
Niemann-Pick disease	Bloom disease
Medium-chain acyl-CoA dehydrogenase deficiency	

Based on the information provided by this handout and my provider, I choose the following regarding genetic carrier screening:

____ opt-in for genetic carrier screening

____ opt-out for genetic carrier screening

_____ _____

Patient Signature Date