

## Genetic Carrier Screening

**Patient Name:** \_\_\_\_\_ **DOB:** \_\_\_\_\_

There exist a class of genetic diseases, know as Autosomal Recessive (AR), in which individuals that do not suffer any symptoms of a disease may have a child with a significant genetic disease. There are hundreds of different AR conditions and it is common for normal individuals to be a carrier for at least one AR condition. In fact, more than 25% of the general population is a carrier for at least one AR condition. To have an AR disease, **BOTH** parents must be carriers for the disease. If both parents are carriers for AR Disease, the chances per birth are 25% for having a child affected with genetic disease, 50% for having a child who is normal but is a carrier for the genetic disease, and 25% for having a normal child that is not a carrier for the genetic disease. If one parent is a carrier, the child has a 50% chance of being a carrier as well and a much lower risk of actually having the condition or disease.

Universal genetic carrier screening allows couples to screen for a variety of genetic disorders prior to becoming pregnant. Genetic carrier screening is a simple blood or saliva test that evaluates an individual's DNA to determine if s/he is a carrier of any genetic abnormalities. Most individuals who are carriers of these genetic mutations do not have any symptoms. If both partners are carriers of the same genetic mutation, however, the couple is at risk of passing on the disease to their offspring. The most common genetic disorders that are tested for include: Cystic Fibrosis, Fragile X Syndrome, Spinal Muscular Atrophy, Tay-Sachs, and Sickle Cell Anemia. In addition to those conditions, genetic carrier screening can also identify other more rare diseases. A full list of diseases can be provided upon request. Some testing panels evaluate only the most common AR conditions with a relatively small number of tests while other panels are extensive. No panel, however, is capable of detecting all conditions in all people. Furthermore, all tests are associated with the possibility of error which means the chance of having a child with an affected genetic condition cannot be driven to zero even in the presence of a normal testing panel result.

Although universal genetic carrier screening is incredibly accurate, receiving a negative result cannot completely eliminate the chance that you will not have a child with a genetic disorder. More importantly, if a positive finding is reported in both you and your partner, we can offer specific treatment options to help reduce the chance of the disease being passed on to your offspring. **We recommend genetic carrier screening to all individuals of reproductive age.** The American College of Obstetrics and Gynecology (ACOG) in March of 2017 released a formal Committee Opinion (#691) recommending all individuals of reproductive age to undergo genetic carrier screening prior to attempting pregnancy. **We strongly recommend that all individuals discuss genetic testing and their family/personal medical history with a licensed genetic counselor; by signing this form I verify that I have been given this contact information: Natera Horizon Carrier Screening; Phone (650-249-9090) and [www.natera.com](http://www.natera.com)**

**There is a chance that your insurance company may not pay for some or all of the cost of the test.** If this is the case, you will be responsible for payment. If you do not have insurance, the out-of-pocket cash price depends on the lab utilized and is available upon request. Prior to beginning any fertility treatment we request that you review universal genetic carrier screening with your physician and as a couple you decide to decline or accept this testing.

**By signing this form, I verify that I understand the information explained in this form and will notify my physician should I choose to undergo this testing. If I choose to decline this testing, against the recommendations of Fertility Associates of Memphis, I understand that I am accepting the associated risks including having a child that may be handicapped or die from a genetic syndrome that may have been otherwise avoided.**

**Patient Signature** \_\_\_\_\_

**Date** \_\_\_\_\_