Reproductive Carrier Screening Consent Form

TEST INFORMATION
The purpose of this consent form is to provide you with information regarding the GeneAware test, however it should not take the place of discussing this information with your healthcare provider. The purpose of the GeneAware reproductive carrier screening test is to determine if you are a carrier of a pathogenic variant for a genetic disorder. Everyone is a carrier for several genetic disorders but may not know it because of lack of family history of any affected members. Disorders screened for by the GeneAware panel test focus on genetic conditions that present early in life, some may have treatment options available and others may not. If you are found to be a carrier, you can discuss with your healthcare provider next steps regarding additional testing and family planning options.

RESULTS
The categories of test results that may be reported include:

- Positive, pathogenic variant/s detected. This means you are a carrier of a pathogenic variant for a genetic disorder. You may be a carrier for more than one genetic disorder.
- Positive, two pathogenic variants detected in the same gene. Rarely a patient may be identified to have two pathogenic variants for one genetic disorder. In this circumstance, it is possible that the two pathogenic variants detected may be on the same chromosome, which would require additional family testing to further elucidate the risk of having a child with the genetic disorder. It is also possible this could indicate that the patient has the genetic disorder.
- Negative, no pathogenic variant detected. This means you are not a carrier for any of the pathogenic variants listed as part of the test. There is still a chance that you may carry a pathogenic variant not tested and still be a carrier for a genetic disorder included on the GeneAware panel.

POTENTIAL RISKS AND DISCOMFORTS
- You may still be a carrier for one of the genetic disorders included in the GeneAware panel.
- GeneAware does not rule out being a carrier for disorders that are NOT included on the GeneAware panel.
- Testing can reveal sensitive health information about you and/or your partner while helping to determine the likelihood that you and your partner will have a child affected with a genetic disorder. Testing results can have health implications for members of your immediate family as well, such as siblings, and their likelihood to be carriers of the same pathogenic variants. This information may also reveal unexpected information.
- This test is not for diagnostic purposes. If you suspect you are affected with a genetic disorder, full gene sequencing or other pathogenic variant panel tests are available upon request that are designed to detect pathogenic variants for diagnosis of genetic disorders.
- This test is not for prenatal diagnosis purposes. This test is only meant to determine pathogenic variant carrier status for you. If both you and your partner are carriers of pathogenic variants for the same disorder, prenatal diagnosis for all of the disorders on the GeneAware carrier screening is available through this laboratory.
- The scope of this test is limited to the pathogenic variants chosen by this laboratory to be the most common and most well-known and researched pathogenic variants associated with the genetic disorders listed. It is not designed to detect every pathogenic variant for each of the disorders, or to detect pathogenic variants for all known genetic disorders. If there is a known family history for a specific genetic disorder listed on this test and your result is negative, it does not necessarily mean that you are not a carrier of a pathogenic variant for that particular disorder. Testing of affected family members first to identify pathogenic variants carried in the family is ideal. If an affected family member is not available for testing, or in the event that information cannot be communicated, specific full gene sequencing may be recommended.
- This testing is complex and utilizes specialized materials so that there is always a very small possibility that the test will not work properly or that an error will occur.
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- Information obtained from the GeneAware carrier screening test may be used in scientific publications or presentations, but the identity of all persons studied will not be revealed in such publications/presentations.

FOR SAMPLES SUBMITTED FROM NEW YORK STATE
Specimen Retention: My sample shall be destroyed at the end of the testing process or not more than 60 days after completion of testing.

Due to the complex nature of this testing we recommend both pretest and post test genetic counseling. I have read the above consent form and give my permission for my sample to be tested for the GeneAware reproductive carrier screening.

Patient Name: ____________________________________________________________

Patient Signature: ___________________________ Date: _________________________