PATIENT NAME_____ MPI#_____

Consent for Genetic Screening Tests

Genetic carrier screening is done to determine if one or both parents may have abnormal genes that may increase the chance that their child will have a specific genetic disease. For many genetic diseases, if someone has an abnormal gene, that person is considered a carrier for that genetic disease. If this abnormal gene is passed to the child, the child will usually not be affected with that genetic disease but will also be a carrier for that genetic disease. If both parents are carriers of the abnormal gene for the same genetic disease, there is a 25% chance that their child will inherit one abnormal gene from each parent and be affected with that genetic disease. Genetic screening is typically done on one parent first, and if the first parent tests positive, then the other parent is tested. The American College of Obstetricians and Gynecologists (**ACOG**) recommends screening for certain genetic diseases when indicated due to ethnicity, family history, or other known risk factors.

Cystic Fibrosis (CF) and **Spinal Muscular Atrophy** (SMA) are genetic diseases that may be screened for. It is estimated that 1 in 31 Americans are carriers of a mutation of the CF gene and 1 in 40 are carriers of a mutation in the SMA gene. Because so many people are carriers of the mutations for CF and SMA, across all races and ethnic groups, ***ACOG currently recommends that routine screening for Cystic Fibrosis be offered to all couples who are planning to have a baby and the American College of Medical Genetics recommends that all couples be offered carrier testing for Spinal Muscular Atrophy.***

ACOG currently recommends that carrier tests for the following genetic diseases be offered to ***all individuals of Ashkenazi Jewish descent*** who are having a baby, because of the high carrier frequency rate in this ethnic group: **Gaucher's Disease** (1 in 15), **Tay Sachs Disease** (1 in 30), **Familial Dysautonomia** (1 in 30) and **Canavan Disease** (1 in 40). ACOG currently recommends that carrier tests be offered only to individuals who have a ***family history*** of the following very rare genetic diseases due to the low carrier frequency rate: **Fanconi Anemia Group C** (1 in 89), **Niemann-Pick Disease Type A** (1 in 90), **Bloom Syndrome** (1 in 100) and **Mucolipidosis type IV** (1 in 127).

While **Tay Sachs Disease** is less common in Caucasians who are not of Ashkenazi Jewish descent, it has been observed to be more common in individuals of **French Canadian** or **Cajun** descent, so screening for Tay Sachs is also offered to individuals who have **French Canadian** or **Cajun** ancestry.

There are other genetic diseases which are transmitted directly from parent to child, so that if the parent is determined to have the gene causing that genetic disease, there is a 50% risk of the child being affected by the same genetic disease. Certain genetic diseases that are carried on the sex-determining (X & Y) chromosomes may cause disease primarily in male children but only rarely in female children. ACOG currently recommends that screening for **Fragile X Syndrome** be offered only to women with a ***family history*** of mental retardation, developmental delay of uncertain cause, autism or autistic-like behavior, as well as women with premature ovarian failure or elevated follicle stimulating hormone level before the age of 40 years, but not to the general population.

During the course of your evaluation and treatment at IVF New England, your physician may recommend screening for specific genetic disease(s) which may be indicated based on your medical history and/or family history, to determine whether or not you are a carrier for the specified genetic disease(s). There is no one genetic carrier test that detects all genetic diseases and therefore genetic carrier tests may only be done for specified genetic diseases and are usually performed on a blood sample.

PATIENT NAME_____ MPI#_____

Genetic testing is a valuable tool but there are limitations:

- *Negative result* – The genetic testing laboratory usually tests for the most common mutations (change in gene structure) and may not identify the less common mutations. So it is possible to have a negative test result but still have a genetic mutation that was not or could not be identified by the testing laboratory due to limitations of current technology.
- *Positive result* - A positive test result indicates that you are a carrier for a genetic mutation that can cause a specific genetic disease or can put you and/or your child at risk for developing a disease. If you are determined to be a carrier, your reproductive partner will then be advised to undergo genetic carrier testing.
- *Inconclusive result* – Sometimes it is not possible for the testing laboratory to determine genetic mutations. In this case, the genetic carrier test may need to be performed again at the same or different testing laboratory.

Current Federal & State laws prohibit discrimination by insurance carriers based on results of genetic tests.

My physician has recommended that I have a blood sample taken in order to conduct genetic testing for

_____

_____.

I have had the opportunity to meet with my physician and discuss the reason for the test and the reliability of a positive and negative test result. I acknowledge:

- This test is voluntary and is being performed to determine if I am a genetic carrier of the condition named.
- The importance of genetic counseling to further discuss this test and how it may affect me and my family. Information about genetic counseling has been provided.
- The cost of this test may not be covered by my insurance plan and I will be responsible for the costs associated with the genetic test.
- It may be possible that even with a negative result, I still could be a genetic carrier or predisposed to the above condition.
- The test results will be contained in my medical record and the Center will take all appropriate safeguards to protect the confidentiality of my personal health information. However, I hereby consent to the release of these results to my partner, the physicians and nursing staff directly involved in my care for the purpose of my treatment, my current insurance carrier in order to seek coverage or reimbursement, and others specifically authorized by law to gain access to my medical records
- No test other than the genetic test specifically authorized will be performed on my blood sample.

_____ _____

Signature of Clinician obtaining consent/Title **Date**

PATIENT NAME_____ MPI#_____

CONSENT FOR GENETIC TESTING

I acknowledge:

- This test is voluntary and is being performed to determine if I am a genetic carrier of the condition(s) named.
- The importance of genetic counseling to further discuss this test and how it may affect me and my family. Information about genetic counseling has been provided.
- The cost of this test may not be covered by my insurance plan and I will then be responsible for the costs associated with the genetic carrier test.
- It may be possible that even with a negative result, I still could be a genetic carrier or predisposed to the above condition.
- The test results will be contained in my medical record and the Center will take all appropriate safeguards to protect the confidentiality of my personal health information. However, I hereby consent to the release of these results to my partner, the physicians and nursing staff directly involved in my care for the purpose of my treatment, my current insurance carrier in order to seek coverage or reimbursement, and others specifically authorized by law to gain access to my medical records
- No test other than the genetic test(s) specifically authorized will be performed on the blood and/or saliva sample taken for this reason; however, additional blood samples may be taken at the same time for other ordered tests, such as infectious disease testing.
- Current Federal & State laws prohibit discrimination by insurance carriers based on results of genetic carrier tests.
- I will release my genetic carrier test results to my partner. I further acknowledge that it has been recommended that I discuss the results of my genetic carrier test results with family members.

I have had the opportunity to meet with my physician and discuss the reason for the genetic carrier screening test(s) and the reliability of a positive and negative test result. I have had the opportunity to ask questions and have had all my questions answered to my satisfaction.

Based on the information that has been provided to me and in discussion with my physician, I freely and without coercion **AGREE** PRIOR TO STARTING ANY FERTILITY THERAPY to have the recommended genetic carrier screening test(s) performed.

Name_____ **Signature**_____

Print **Date:**_____

OR

Based on the information that has been provided to me and in discussion with my physician, I freely and without coercion **WAIVE** the recommended genetic carrier screening test(s) performed. If we conceive, we recognize we could have a child with a genetic disease if we both happen to be carriers of a particular mutation. In that regard, we accept full responsibility for the transmission of any genetic disease to our child that may occur as a result of our treatment at IVFNE.

Name_____ **Signature**_____

Print **Date:**_____

Witness Name:_____ **Signature:**_____

Date:_____

PATIENT NAME_____ MPI#_____

PARTNER CONSENT TO GENETIC SCREENING

In the event that my partner screens positive for the specific genetic disease(s) described on Page 2 of this consent, I understand that screening will be recommended for me.

I acknowledge:

- This test is voluntary and is being performed to determine if I am a genetic carrier of the condition(s) named.
- The importance of genetic counseling to further discuss this test and how it may affect me and my family. Information about genetic counseling has been provided.
- The cost of this test may not be covered by my insurance plan and I will then be responsible for the costs associated with the genetic carrier test.
- It may be possible that even with a negative result, I still could be a genetic carrier or predisposed to the above condition.
- The test results will be contained in my medical record and the Center will take all appropriate safeguards to protect the confidentiality of my personal health information. However, I hereby consent to the release of these results to my partner, the physicians and nursing staff directly involved in my care for the purpose of my treatment, my current insurance carrier in order to seek coverage or reimbursement, and others specifically authorized by law to gain access to my medical records
- No test other than the genetic test(s) specifically authorized will be performed on the blood and/or saliva sample taken for this reason; however, additional blood samples may be taken at the same time for other ordered tests, such as infectious disease testing.
- Current Federal & State laws prohibit discrimination by insurance carriers based on results of genetic carrier tests.
- I will release my genetic carrier test results to my partner. I further acknowledge that it has been recommended that I discuss the results of my genetic carrier test results with family members.

I have had the opportunity to meet with my physician and discuss the reason for the genetic carrier screening test(s) and the reliability of a positive and negative test result. I have had the opportunity to ask questions and have had all my questions answered to my satisfaction.

Based on the information that has been provided to me and in discussion with my physician, I freely and without coercion **AGREE** PRIOR TO STARTING ANY FERTILITY THERAPY to have the recommended genetic carrier screening test(s) performed.

Name_____ **Signature**_____

Print **Date:**_____

Based on the information that has been provided to me and in discussion with my physician, I freely and without coercion **WAIVE** the recommended genetic carrier screening test(s) performed. If we conceive, we recognize we could have a child with a genetic disease if we both happen to be carriers of a particular mutation. In that regard, we accept full responsibility for the transmission of any genetic disease to our child that may occur as a result of our treatment at RSCNE.

Name_____ **Signature**_____

Print **Date:**_____

Witness Name:_____ **Signature:**_____

Date:_____