Patient Name: __________________________________       MRN/DOB:____________

In accordance with New York State Law, the following has been discussed with the patient/legal guardian and informed consent obtained. The following was signed in my presence.

Please read the following carefully and discuss with your ordering physician/person obtaining consent before signing consent.

Pan-Ethnic Screen by Genotyping Array Informed Consent: ____________________(Initial if test is requested)

1. This is a test for carrier status using custom Affymetrix® GeneChip® resequencing microarrays (Affymetrix, Inc., Santa Clara, CA).
2. The purpose of this analysis is to test for carrier status of 466 mutations in over 80 different genetic disorders that are prevalent in many different populations.
2a. You (or the person for whom you are signing) may want genetic counseling before signing for consent.
3. This is a screen for carrier status (heterozygous carrier). If you or your partner is a carrier for a mutation in any disease gene tested you may wish to have further independent testing, consult your physician or have genetic counseling.
4. When two mutations are found, there is almost 100% likelihood of disease. When no mutation is found, in an asymptomatic person, there is almost 0% likelihood of disease in the tested person, but the risk of being a carrier depends upon race and family history. When one mutation is found, the person is a carrier. The risk of having an affected child depends upon mutation status of the partner. The conditions tested are inherited in an autosomal recessive manner. If both parents are carriers for the same genetic disease, then there is a 1 in 4, or 25 percent chance of the condition in each of their pregnancies.

Cystic Fibrosis Carrier Screen informed consent: ____________________(Initial if test is requested)

1. This is a test for an abnormality (mutation(s)) in the CFTR gene using PCR with a bead probes.
2. The purpose of this analysis is to test for a Cystic Fibrosis carrier status.
2a. You (or the person for whom you are signing) may want genetic counseling before signing consent.
3. This is a test for genetic susceptibility (“genetic predisposition”), the risk of having the disorder may be altered by family history and/or other factors. If the test is positive for the disorder or for an increased risk of the disorder, you may wish to have further independent testing, consult your physician or have genetic counseling.
4. The condition being tested is cystic fibrosis, which affects or leads to lung and digestive problems.
5. When two mutations are found, there is almost 100% likelihood of disease. When no mutation is found, in an asymptomatic person, there is almost 0% likelihood of disease in the tested person, but the risk of being a carrier depends upon race and family history. When one mutation is found, the person is a carrier. The risk of having an affected child depends upon mutation status of the partner.

FMR1 (Fragile X) Carrier Screen informed consent: ____________________(Initial if test is requested)

1. This is a genetic (DNA-based) test using PCR and If necessary, Southern Blot to test for expansion of the CGG repeat in the 5’ untranslated region of the Fragile X gene (FMR1).
2. The purpose of this analysis is to test for your carrier status for Fragile X-syndrome, which could result in a child with mental retardation.
2a. You (or the person for whom you are signing) may want genetic counseling before signing for consent.
3. This is a test for genetic susceptibility (“genetic predisposition”), the risk of having an affected child depends upon whether you are a carrier of an expanded FMR1 gene.
4. The condition being tested for is Fragile X syndrome, or its carrier state.
5. If you have a full mutation you may or may not have Fragile X syndrome; if you have a premutation, there is a 50% chance of passing on the abnormal premutation allele to your children with a 3% to 100% chance of expansion to a full mutation (depending upon the size of the premutation allele), which could result in an affected child. In addition, if you test positive for a premutation you may develop early menopause and in your later years, have a chance of developing a disorder called Fragile X tremor-ataxia syndrome. If you test negative, the chances of your child having Fragile X syndrome are very low, but not zero.
SMN1 (SMA) Carrier Screen informed consent: ______________________(Initial if test is requested)

1. This is a genetic (DNA-based) test for the detection of carrier state of autosomal recessive Spinal Muscular Atrophy (SMA) using Real-Time PCR.
2. The purpose of this analysis is to test for SMN1 gene deletion mutation.
2a. You (or the person for whom you are signing) may want genetic counseling before signing for consent.
3. This is a test for genetic susceptibility ("genetic predisposition"), the risk of having an affected child depends upon your SMN1 status.
4. The condition being tested for is Spinal Muscular Atrophy, or its carrier state, which affects muscle strength and can lead to moderate or severe weakness.
5. If you have fewer than 2 copies of the SMN1 gene, then you could be at risk of having a child with the disease SMA. The actual risk will depend upon your partner’s SMN1 status. If you have two copies of the SMN1 gene, you have a low chance of passing on an abnormal gene to your child. Some carriers cannot be detected by this test: those with two normal SMN1 on one chromosome and none on the other, and those with point mutations in the SMN1 gene. Therefore, with a negative result, your chances of having an affected child are lower than the general population, but not zero.

General considerations for all three above tests:
1. The results of the above test become a part of the patient’s medical record, and may be made available to individuals/organizations with legal access to the patient’s medical record, on a strict “need-to-know” basis, including, but not limited to the physicians and nursing staff directly involved in the patient’s care, the patient’s current and future insurance carriers, and others specifically authorized by the patient/authorized representative to gain access to the patient’s medical records.
2. No additional tests will be performed on this sample, without specific, signed authorization by the patient. After 60 days, unless consent is given the sample will be destroyed, or will be de-linked from all patient information and used for standard laboratory purposes.
3. Medicare/Insurance Carriers may not pay for the test, in which case, the patient/responsible party will be billed for the test.

Name of Person Obtaining Consent: ________________________                               Title:
Signature:  ________________________________                                                           Date:

I have read and fully understood the above, and give my consent for this testing.

Patient signature:___________________________________________________        Date:

If consent is given by parent or legal guardian:
Name:_________________________________________________________
Signature:______________________________________________________     Date:

Consent for sample retention:
I consent to the retention of this sample for: (check and sign on appropriate line)
☐ I do not consent to research. My sample may be used for routine laboratory use only._______________

☐ I consent to possible future genetic research on my sample if all identifying information is removed (name, address, date of birth, medical record number). The duration of the retention of my sample will depend on the individual research study. If the sample is not used in a study, it will be destroyed or anonymously used as described above. ____________