NOTE: Please obtain patient signature on consent form and provide a signed copy to EGL Genetics to permit testing and processing.

I, (name)________________________________________, voluntarily request of EGL Genetics to perform DNA-based testing for Spinal Muscular Atrophy on myself/my child (child’s name ____________________________), in an attempt to determine whether I/my child am a carrier of a disease gene or are at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

- Gaucher disease is an autosomal recessive lysosomal storage disorder caused by deficient activity of the enzyme beta-glucocerebrosidase. This deficiency leads to a progressive accumulation of glucocerebroside (glycosylceramide) in certain body tissues such as the spleen, liver, and bone marrow. Symptoms of Gaucher disease usually become apparent in childhood or early adulthood. Gaucher disease is classically categorized into 3 main subtypes: non-neuronopathic type I, acute neuronopathic type II, and subacute neuronopathic type III. Type I is the most common form of Gaucher disease and lacks primary central nervous system involvement (non-neuronopathic). Types II and III have central nervous system involvement (neuronopathic) and neurologic manifestations. There is overlap between the clinical presentation of both neuronopathic forms of Gaucher disease, and for this reason, it is thought that neuronopathic Gaucher disease represents a clinical spectrum ranging from mild symptoms to a perinatal-lethal form. All 3 forms of Gaucher disease are caused by mutation in the GBA gene. This test is indicated for individuals suspected to have Gaucher disease and individuals at risk to be carriers of Gaucher disease.

- DNA testing requires a blood sample, cheek or mouth swab, muscle or skin biopsy, all of which have risks associated with obtaining the sample. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted.

- DNA-based studies performed are specific to the condition indicated above. The accuracy of genetic testing is limited by the methods employed, the clinical diagnosis, and the nature of the specific condition for which testing is requested. In some cases, the test will detect an abnormality, called a mutation, in the gene. In other cases the test is unable to identify an abnormality although an abnormality may still exist. This event may be due to the current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (mutations) in a gene. These tests are currently available for clinical laboratory testing; however, improvements will be made as scientific knowledge advances. As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.

- Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis, and other sources. These analyses may not detect pathogenic variants in the promoter or other regulatory regions. Sequence analysis will not detect large deletions and duplications. This assay is not designed to detect all recombinant pathogenic alleles derived from recombination between the active gene (GBA) and pseudogene (PGBA).

- It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the testing ordered, and to educate the patient regarding these limitations. Additional information describing indications, methodology and detection can be found on the EGL website at: https://www.egl-eurofins.com/

- Accurate interpretation of test results is dependent upon the patient’s clinical diagnosis or family medical history and upon reported family relationships being true biological relationships. An erroneous clinical diagnosis in the patient or family member can lead to an incorrect interpretation in the laboratory result. Genetic testing in family members can sometimes reveal that true biological relationships are not consistent with the reported biological relationships. For example, non-paternity may be detected, which means that the stated or assumed father of an individual is not the true biological father.

- This analysis can have the following outcomes:
  a. Positive: A pathogenic variant (disease-causing) could be identified in one or more of the genes being tested for and the person is identified as being affected.
  b. Negative: No pathogenic variant is identified. This reduces the risk of being affected by the diseases specifically tested for, but does not eliminate it completely.
  c. Inconclusive: Due to technical issues the results were inconclusive and the test might need repeating. Results may also be inconclusive due to the identification of a variant of unknown significance.
NY Informed Consent – Gaucher Disease: GBA Gene Sequencing

- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient’s ordering provider, who will then review and discuss the test results with me. Patient-identifying results and information at EGL will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- Samples received from the State of New York for genetic testing will not be stored indefinitely or used for research purposes unless otherwise specified. Extracted DNA for NY samples and other clients with opt out policies will be discarded according to laboratory policy. Please contact EGL Genetics for details.
- I can request that remaining DNA be retained and used for research purposes by initialing here: ______
  a. EGL is not a DNA banking facility and does not guarantee the future availability of isolated DNA. Requests for additional studies must be ordered by the referring provider, and charges will be incurred. Once the test is complete, identifying information may be removed as permitted by the Health Insurance Portability and Accountability Act (“HIPAA”). Remaining DNA samples may also be used for EGL’s laboratory/internal purposes as permitted by HIPAA; these samples will not be available for future clinical studies.
  b. Scientific knowledge is continuing to advance in this area. I request and authorize EGL to notify the ordering provider if there is a research study or clinical trial studying the abovementioned condition or a drug, treatment or device which might assist in the patient’s care. I understand EGL may receive remuneration for those activities. The ordering provider will discuss the information that he/she receives with me, and it is my voluntary decision whether to participate. My decision will not affect the testing or results in any way.

My signature below acknowledges my voluntary participation in this test and I state that I have been appropriately counseled about the testing process and the different possible outcomes.

________________________________  ________________________________       _________________
Patient/Guardian Signature                 Printed Name                               Date

Physician/Counselor/Clinician Statement:
I have explained DNA testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient’s best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by EGL or others.

________________________________ _________________________________     _________________
Healthcare Provider Signature                Printed Name                             Date