Informed Consent / Refusal for Genetic Testing

DNA Testing

1. The purpose of my DNA test is to look for mutation(s) known to be associated with the following genetic condition(s) or disease(s): *(check all that apply)*

☐ Bloom Syndrome	☐ Fanconi Anemia (Group C)	☐ Nemaline Myopathy
☐ Canavan Disease	☐ Fragile X Syndrome	☐ Niemann-Pick Disease (Type A & B)
☐ Cystic Fibrosis	☐ Gaucher Disease	☐ Prothrombin G20210A Mutation
☐ Dihydrolipoamide	☐ Glycogen Storage (Type 1A)	☐ SMA Spinal Muscular Atrophy
☐ Factor V Leiden Mutation	☐ Joubert Syndrome 2	☐ Tay-Sachs DNA (Reflex)
☐ Familial Dysautonomia	☐ Maple Syrup Urine Disease	☐ Usher Syndrome (Type 1F)
☐ Familial Hyperinsulinism	☐ MTHFR (C677T and A1298C) Mutation	☐ Usher Syndrome (Type 3)
	☐ Mucolipidosis (Type IV)	☐ Walker-Warburg

☐ Other _____

2. Mutations are often different in different populations. I understand that the laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results. Three mL (approximately two teaspoons) of blood is required for the test. DNA is then analyzed using well established methods, which include but are not limited to, PCR amplification, DNA sequencing and Invader methods.
3. When DNA testing shows a mutation, then the person is a carrier or is affected with that condition or disease. Consulting a doctor or genetic counselor is recommended to learn the full meaning of the results.
4. When the DNA testing does not show a known mutation, the chance that the person is a carrier or is affected is reduced. There is still a chance to be a carrier or to be affected because the current testing cannot find all the possible changes within a gene.
5. In some families DNA testing might discover non-paternity (someone who is not the biological father), or some other previously unknown information about family relationships, such as adoption.
6. The decision to consent to, or to refuse the above testing is entirely mine.
7. No test(s) will be performed and reported on my sample other than the one(s) authorized by my doctor, and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory.
8. NS-LIJ will disclose the test results ONLY to the doctor named below, or to his/her agent, unless otherwise authorized by the patient or required by law.
9. My signature below indicates that I have read, or had read to me, the above information and I understand it. I have had the opportunity to discuss it (including a general description of the disease and the purposes and possible risks of the test/s) with my doctor or someone my doctor has designated. I know that I may obtain professional genetic counseling if I wish, before signing this consent. I have all the information I want, and all my questions have been answered.

☐ **YES:** I REQUEST that Dr. _____ perform the genetic testing above. I understand and accept the consequences of this decision.

PATIENT SIGNATURE	DATE	WITNESSED BY

☐ **NO:** I DECLINE to have the genetic testing offered to me. I understand and accept the consequences of this decision.

PATIENT SIGNATURE	DATE	WITNESSED BY

New York has statutes requiring laboratories to send confidential results of certain genetic tests to state or federal health agencies for monitoring the detection of birth defects.

It is standard of care for physicians to obtain informed consent for genetic testing. This model consent form is provided by NS-LIJ as a courtesy to physicians and their patients. Relevant patient and/or physician educational materials are also available through NS-LIJ.

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