

In accordance with New York State law, an informed consent form must be completed prior to the performance of genetic testing. I understand that when signed and dated, this written consent is written authorization to participate in genetic testing. (The individual to be tested may wish to obtain professional genetic counseling prior to signing this informed consent.)

**In order to help me understand the implication of the test results, the results will be reported to me only through a physician or genetic counselor that I designate. The results are confidential.**

**Patient Name:**

**Designated Genetic Provider Name & Address:**

**Patient Address:**

▪ **Test Description (General):**

|                   |   |   |
|-------------------|---|---|
| DNA (Molecular)   | PCR Amplification and DNA analysis  | <b>Initials</b><br><input type="text"/> |
| Organic Acids     | Gas chromatographic mass spectrometric separation to quantify urine organic acids.  |   |
| Neuropharmacology | HPLC method for assessing vascular disease risk factors and neurological disorders.   |   |
| In-Vitro Studies  | Fibroblasts incubated with stable isotopes are used to detect blockages in the metabolic pathway of mitochondrial fatty acid oxidation using liquid secondary ion tandem mass spectrometry. |   |
| Acylcarnitine     | In-born errors of fatty acid and amino acid metabolism may be identified by tandem MS / MS.   |   |
| Carnitine         | Inborn errors in Carnitine metabolism may be identified by tandem MS / MS.  | <input type="text"/>                    |

▪ **Purpose of the test:** The potential benefit of this test is to obtain the possible diagnosis of an inherited metabolic disease. It may also determine which family members may be carriers of the disease. For prenatal diagnosis, it may determine the genetic status of the fetus.

▪ **Statement regarding test result:** A positive test result is an indication that the individual may have the specific disease tested for or be a carrier. That individual may wish to consider further independent testing, consult a personal physician, or pursue genetic counseling.

▪ **General description of diseases or conditions (symptoms may include):**

|                   |                          |                |                    |                      |
|-------------------|--------------------------|----------------|--------------------|----------------------|
| Reyes syndrome    | Developmental delay      | Seizures       | Metabolic acidosis | <input type="text"/> |
| Encephalopathy    | Failure to thrive        | Hypoglycemia   | Hyperammonemia     |                      |
| Skeletal myopathy | Sudden unexplained death | Cardiomyopathy | Ketosis            |                      |

▪ **Level of certainty:** A positive result for the DNA mutation is 100% certainty for that DNA mutation.

▪ **Person, or categories of persons, or organizations to which the test results may be released:**

▪ **Test Authorization:** No other tests shall be performed on this sample other than those authorized. Should the technology improve in the future and/or more mutations can be detected, I authorize the laboratory to re-analyze the remaining sample (which may be retained for up to 10 years for DNA samples) for the same disease testing that my physician ordered originally.

▪ **Release of sample for research:** After the testing is completed, an aliquot of my sample may be made anonymous and used for research purposes. Sample will be stored as long as deemed useful for research purposes. Since any results obtained could not be related to the original source, no results would be reported or available to me for that research testing.

|   |                    |
|---|--------------------|
| <b>Signature of Patient or person authorized to consent:</b> _____              | <b>Date:</b> _____ |
| <b>If person authorized to consent, describe relationship to patient:</b> _____ |                    |
| <b>Signature of Physician/Genetic Counselor:</b> _____                          | <b>Date:</b> _____ |